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ABSTRACT PROCEEDINGS OF YOUNG TALENTS IN INTERNAL MEDICINE WORLD CONTEST 2025

(10th McMaster International Review Conference of Internal Medicine, May 8–10, 2025, Kraków, Poland; hybrid course)

TOP 20 SUBMISSIONS QUALIFIED FOR YOUNG TALENTS FINALS

1ST PLACE: PETER TODD

Kidney disease… Is it worth the weight?

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INTRODUCTION We describe a case of a patient who presented with rapidly deteriorating renal function. Renal biopsy demonstrated oxalate nephropathy: intratubular crystals of calcium oxalate. It was discovered that he had been self-medicating with orlistat and supratherapeutic doses of vitamins C and D_3. We describe how these nonprescription agents interacted with each other to cause the patient's condition, leaving him with chronic kidney disease and having narrowly avoided dialysis. This case demonstrates that over-the-counter medications may cause significant harm and that seeking information about nonprescription medications is an important aspect of medical assessment.

CASE DESCRIPTION A 60-year-old man with type 2 diabetes and obesity was referred to a nephrology department by his diabetes physician due to declining renal function in the absence of albuminuria. His estimated glomerular filtration rate (eGFR) had fallen from above 60 ml/min/1.73m^2 to 33 ml/min/1.73m^2 in 1 year. He reported intentional weight loss over the same time period and his glycemic control had improved. On initial nephrology clinic assessment, the patient's eGFR had fallen further to 22 ml/min/1.73m^2. He was also noted to have hypercalcemia. A renal biopsy showed oxalate nephropathy—calcium oxalate crystals deposited in the lumens of the kidney tubules. The patient subsequently reported having been buying orlistat, an intraluminal lipase inhibitor, over the internet as an aid in the weight loss process. He also declared the use of a large number of vitamin supplements—61 times and 33 times the UK recommended intake of vitamins C and D_3 respectively. The combination of these medications was the underlying cause of his oxalate nephropathy. The orlistat and vitamin supplements were stopped, eGFR reached a nadir of 8 ml/min/1.73m^2, and hemodialysis was provisionally planned. Thankfully, renal function then began to improve. After 3 months, eGFR reached 34 ml/min/1.73m^2.

DISCUSSION The association between orlistat use and secondary hyperoxaluria is well established. Increased presence of dietary fats in the intestinal lumen binds free calcium ions, reducing the availability of calcium ions to bind to dietary oxalate in the gut. Therefore, increased absorption of unbound oxalate increases urinary oxalate excretion. In this case, supratherapeutic vitamin C and D_3 intake played a synergistic role. Vitamin C is partially metabolized to oxalic acid, additionally increasing the urinary oxalate concentration. Hypervitaminosis D_3 causes hypercalcemia and subsequent hypercalciuria. In this case, high concentrations of both calcium and oxalate ions in the urine led to tubular supersaturation of calcium oxalate and subsequent pathological crystallization. The culprit over-the-counter medications in this case were bought on the internet and in a high street health shop. Despite this, they have caused this patient significant irreversible morbidity. It is important for healthcare professionals to be aware that nonprescription medications may cause severe harm and that patients may not offer information about them unless specifically asked.

CONCLUSIONS Orlistat is available without prescription and used as an aid in the weight loss process. Its potentially nephrotoxic interaction with vitamin C supplementation should be recognized. Over-the-counter medication use is prevalent and increasing, as they can be easily accessed on the internet. Information about nonprescription medication use should be actively sought from patients.

Key words
orlistat, over-the-counter medications, oxalate nephropathy, secondary hyperoxaluria, vitamin C

2ND PLACE: ARTHUR RENAUD

Refractory ascites and multinodular systemic involvement lead to the discovery of an exceptional multivisceral form of vascular epithelioid tumor associated with the *EWSR1::NFATC2* rearrangement

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INTRODUCTION Internists often investigate complex diagnostic cases that demand interdisciplinary collaboration. We present a rare case of a multivisceral epithelioid vascular tumor associated with an *EWSR1::NFATC2* gene fusion. The diagnostic process relied heavily on the integration of anatomo-clinical findings and molecular biology expertise, underscoring the value of collaborative approaches.

CASE DESCRIPTION A 39-year-old woman with a history of treated asthma and smoking cessation underwent cholecystectomy. Incidental findings included sclerotic and lytic skeletal lesions, vascular-like hepatic nodules, and peritoneal involvement. Initial histological analysis was nondiagnostic and the patient was lost to follow-up.

Two years later, she presented with recurrent exudative ascites requiring repeated paracenteses. Clinical evaluation showed no abnormalities and a good general condition. Routine laboratory tests, including hepatic function and albumin level, were unremarkable. Imaging studies (computed tomography, magnetic resonance imaging, positron emission tomography) showed persistent, mildly hypermetabolic nodular lesions with minimal progression, offering no definitive etiological clues. Transjugular liver biopsy showed normal hepatic architecture, encompassing sinusoidal dilatation, without neoplasia or granulomas. Hepatic catheterization confirmed portal hypertension. Multidisciplinary assessment concluded a diagnosis of noncirrhotic portal hypertension caused by extrinsic compression of hepatic veins, likely from hepatic nodules and peritoneal sheath encasement, mimicking Budd–Chiari syndrome. Comprehensive infectious and immunohematological workups were noncontributory. Laparoscopy showed an inflamed, multinodular peritoneum. Histopathological analysis of peritoneal biopsies showed fibrotic remodeling with minimal inflammation and capillary hyperplasia, which was deemed reactive following a consultation with a national expert.

Despite these conclusions, the clinical and radiological presentation suggested the *primum movens* of a multivisceral microvascular disorder. After multiple inconclusive computed tomography–guided bone biopsies, blind iliac crest bone marrow biopsy identified atypical vascular lesions with ossifying reactions. The case was discussed at a national bone tumor board. Histological findings suggested low grade vascular tumor, but clinical and radiological data were inconsistent. Suggested molecular analysis of a peritoneal biopsy ultimately identified an *EWSR1::NFATC2* gene fusion, confirming a rare vascular epithelioid tumor.

DISCUSSION This case involved extensive but indolent hepatic, peritoneal, and osseous involvement, which evaded diagnosis despite thorough investigations. Multidisciplinary collaboration and targeted biopsies directed the diagnostic process toward a vascular proliferation. Ultimately, the use of a national network and a specialized molecular biology laboratory led to the identification of a *EWSR1::NFATC2* gene fusion, a rare anomaly associated with a spectrum of vascular tumors ranging from malformations and hemangiomas to sarcomas. Molecular biology, a routine tool in

soft tissue and pediatric oncology, is transforming diagnostics in internal medicine, particularly for atypical proliferative disorders (eg, histiocytosis). High-throughput sequencing platforms can improve diagnostic accuracy for elusive cases and facilitate the discovery of novel entities, as exemplified by the discovery of VEXAS syndrome. **CONCLUSIONS** This case underscores the complexity of certain diagnostic scenarios and the critical role of multidisciplinary and molecular biology-based approaches. Broader access to molecular techniques may reduce diagnostic delays and improve outcomes for patients with unclassifiable cellular proliferations.

Key words
epithelioid vascular tumor, *EWSR1*, molecular biology, *NFATC2*

3RD PLACE: RADWA GENIDY

Pyrexia puzzle: take a closer look to solve the mystery

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INTRODUCTION Fever of unknown origin (FUO) is defined as a body core temperature above 38.3 °C for at least 3 weeks with no established diagnosis, despite intensive workup. Three large entities need to be explored, including infections, malignancies or autoimmune conditions. We present the case of a 64-year-old woman who presented with FUO and turned out to have an unusual diagnosis.
CASE DESCRIPTION A 64-year-old black woman, known to have irritable bowel syndrome, presented with fever, nausea, abdominal pain, and high inflammatory markers. Infectious workup was performed and antibiotics were initiated on admission. The fever did not resolve, despite the use of broad-spectrum antibiotics, and only a modest reduction in inflammatory markers was noted. Pancultures, viral serologies, and tuberculosis workup were all unremarkable. Malignancy workup including computed tomography (CT) of the chest, abdomen, and pelvis, bone marrow biopsy, and myeloma workup did not show any diagnostic clues. Autoimmune serologies were all negative. Three weeks of persistent fever necessitated investigations of rare causes of FUO. A revision of the CT of the chest, abdomen, and pelvis was requested. It reported circumferential symmetrical changes in the abdominal aorta, raising the possibility of an autoimmune inflammatory or infective etiology. Experts in the fields of rheumatology and infectious diseases recommended ruling out syphilis and other unusual bacterial causes of aortitis. All results came back negative. Further workup was initiated, looking into causes of autoimmune aortitis, including giant cell arteritis or Takayasu arteritis. The patient did not have any typical symptoms or signs of either condition. As part of the workup for autoimmune large vessel vasculitis, a magnetic resonance angiogram of the arch and thoracic aorta was performed, showing uniform thickening of the aortic wall in the arch and thoracic aorta, with extension into the root of the arch vessels. Following a multidisciplinary team discussion, including the internal medicine team, rheumatologist, infectious disease physician, and interventional radiologist, a diagnosis of extracranial giant cell arteritis was made. The patient was commenced on a 3-day course of methylprednisolone (1 g intravenously). Her response was remarkable, her symptoms resolved, including the fever, and the inflammatory markers were nearly normalized. She was discharged on a tapering dose of 50 mg prednisolone with a 2-week follow-up in the rheumatology clinic.
CONCLUSIONS FUO is a challenging clinical presentation necessitating a high index of suspicion. Often, the key to its management is the effort of a multidisciplinary team.

Key words
aortitis, fever of unknown origin, giant cell arteritis large vessel vasculitis

ALEKSANDRA RACZYŃSKA

Navigating complexities: a case of neuroinfection accompanied by thrombocytopenia

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CASE DESCRIPTION Immune thrombocytopenia is a hematological disorder caused by immune system dysfunction, resulting in low platelet count and bleeding into the skin and mucous membranes. Complications of the disease may endanger the patient's health and life. Therefore, it is important to properly identify any secondary causes of thrombocytopenia, which may have an infectious background. We presented the case of a 65-year-old woman admitted to a department of infectious diseases with fever of unknown origin and headache. The patient was diagnosed with aseptic meningitis. During the hospitalization, she developed hemorrhagic diathesis on the skin and mucous membranes, and her condition worsened. She required multiple blood component transfusions. Long diagnostic investigations of microbiological causes of the disease included blood polymerase chain reaction test for anaplasmosis. The result was positive, as were the results of immunoglobulin M and immunoglobulin G antibodies against anaplasmosis. As soon as we began considering anaplasmosis as the cause of the patient's condition, we started her on intravenous doxycycline treatment. The patient's state improved, as did the results of laboratory test. Human granulocytic anaplasmosis can be underestimated because of its non-characteristic symptoms. Health care specialists should take it into consideration as a rare causative infective factor for fever of unknown origin, meningitis, encephalitis, disseminated intravascular coagulation, and autoimmune thrombocytopenia.

Key words
anaplasmosis, meningitis, thrombocytopenia, tick-borne disease

ANNA ÜRGEOVÁ

Pulmonary edema: more than just a cardiological diagnosis

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INTRODUCTION Scleroderma renal crisis (SRC) is an acute vascular complication of systemic sclerosis (SSc) – predominantly in diffuse cutaneous SSc (dcSSc). It is characterized by malignant hypertension, acute kidney injury (AKI), and organ dysfunction (eg "flash" pulmonary edema). Clinical manifestation can be accompanied by laboratory findings of thrombotic microangiopathy, thrombocytopenia, and microangiopathic hemolytic anemia (MAHA).
CASE DESCRIPTION We present a case of a 28-year-old patient with no significant medical history. The patient was examined at the emergency department and newly diagnosed with arterial hypertension and dyspepsia. Laboratory findings confirmed AKI and elevated levels of N-terminal pro–B-type natriuretic peptide. Transthoracic echocardiography showed acute heart failure with malignant hypertension (blood pressure, 180/120 mm Hg). Concentric left ventricular hypertrophy, diffuse hypokinesis, left ventricular ejection fraction of 25%, grade 3 mitral regurgitation, and grade 2 aortic regurgitation were present. Clinical condition of the patient progressively deteriorated into pulmonary edema and hypoxemic respiratory insufficiency which required mechanical ventilation.

Thoracic drainage was indicated due to bilateral pleural effusion. After the patient's clinical condition was stabilized, differential diagnosis of the secondary hypertension was performed, with the focus on kidney disease.

Based on the clinical manifestation of dyspepsia with diarrhea and laboratory findings (MAHA), a diagnosis of TMA was suspected. Diagnosis of infection-related hemolytic uremic syndrome was not confirmed. Complement component 3 and complement component 4 activity was normal and signs of MAHA gradually disappeared. A disintegrin and metalloproteinase with a thrombospondin type 1 motif, member 13 activity was normal, which ruled out thrombotic thrombocytopenic purpura. At the time of the diagnosis, autoantibodies were negative, which ruled out autoimmune disease. The patient was started on renal replacement treatment and kidney biopsy was indicated. Histological findings confirmed the diagnosis of malignant nephrosclerosis and the patient continued hemodialysis with the diagnosis of chronic kidney disease.

After 4 months, the patient was hospitalized for the second time because of the progression of anemia. During the hospitalization, de novo findings of sclerodactylia, thickening of the skin, and Raynaud phenomenon were observed. Based on these observations, a diagnosis of dcSSc was suggested. Autoantibodies were tested again, and this time antinuclear antibodies (ANA) were positive, although the specific ANA type was not identified. Kidney biopsy was analyzed for the second time and the results supported the diagnosis of SRC. Based on these findings, the etiology of the hypertensive episode with AKI and signs of MAHA was reclassified as a manifestation of SRC. The patient continued hemodialysis for 17 months and underwent kidney transplantation at a later date.

DISCUSSION Scleroderma renal crisis is an acute manifestation of SSc with poor prognosis. This case is a severe manifestation of SRC. Multiple factors could contribute to difficult differential diagnosis. Negative autoantibody result received during the first hospitalization could be connected to the late diagnosis. Renal histopathology findings are not specific to SRC, which complicated kidney biopsy evaluation and a second analysis was necessary to establish that the changes were consistent with the SRC.

CONCLUSIONS Scleroderma renal crisis is an acute and potentially life-threatening vascular manifestation of SSc with unfavorable prognosis. In this case, it was the first manifestation of SSc, which had preceded skin or other organ manifestations. This case shows the challenges associated with the differential diagnosis of SSc and its management.

Key words
acute kidney injury, microangiopathic hemolytic anemia, pulmonary edema, scleroderma renal crisis, systemic sclerosis

AOIFE HARRISON

Recurrent VTE in a patient with multiple spontaneous intracerebral hemorrhages and a background of cerebral amyloid angiopathy

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INTRODUCTION Cerebral amyloid angiopathy (CAA) is relatively common in the aging population. To date, no cures or treatments halting the progression of the disease have been developed. Management involves preventing intracerebral hemorrhage through controlling hypertension and avoiding anticoagulation, if possible. Recurrent venous thromboembolism (VTE) can be potentially life-threatening. It is treated with lifelong anticoagulation, unless a provoking factor can be identified and treated. Our case presents the complexities of these 2 conditions coinciding in an acute setting.

CASE DESCRIPTION We present a case of a 59-year-old woman with a history of obesity and recurrent unprovoked VTE. She was taking anticoagulants (apixaban). The patient presented with a 1-day history of expressive dysphasia and no other neurological deficits. Initial computed tomography (CT) scan of the brain showed bilateral temporal lobe intraparenchymal hemorrhages. The patient was given andexanet alfa in order to reverse anticoagulation. The case was discussed with a neurosurgeon who saw no indications for a surgical intervention.

Unfortunately, further neurological deterioration was observed, including bilateral hearing loss and a decline on Glasgow Coma Scale (GCS). Repeat CT showed expansion of temporal hemorrhages, with new bleeds in the left parietal and right frontal regions. Again, no surgical intervention was warranted. CT imaging was repeated frequently with further evidence of bleeding. Magnetic resonance imaging (MRI) showed evidence of secondary infarction. The patient was managed in the intensive care unit, where her GCS score gradually improved over several days, without the need for intubation. Upon being transferred to the ward, expressive dysphasia and hearing improved, and further recovery was observed during the patient's rehabilitation.

Throughout this acute phase, anticoagulation was withheld and intermittent pneumatic compression was used for VTE prophylaxis. The hematology team was heavily involved in the case. Thorough review confirmed no underlying cause for the patient's recurrent VTE, with negative antiphospholipid screening.

Prior to her presentation to the emergency department, the patient had been experiencing frequent headaches. An MRI scan of the brain had been performed 1 month prior and it retrospectively showed evidence of chronic microhemorrhages in cortical areas, consistent with CAA. This provided us with a cause of her spontaneous intracerebral hemorrhage.

Three weeks after the initial presentation, the patient developed acute chest pain and syncope. CT pulmonary angiography showed bilateral pulmonary embolism and a right popliteal deep vein thrombosis. Brain CT showed no new intracerebral hemorrhages and anticoagulation was resumed with tinzaparin. Dosage was adjusted to the patient's heavy body mass according to the anti-Xa levels. An inferior vena cava filter was placed for long-term VTE management, and the patient was scheduled for a minimum of 6 weeks of anticoagulant therapy due to the high bleeding risk associated with CAA.

The patient made a full neurological recovery and was completely independent at discharge. She is reviewed regularly in the outpatient setting.

CONCLUSIONS This case presents the challenges and risks of managing anticoagulant therapy in patients with recurrent VTE who have evidence of intracerebral hemorrhage and a predisposition for further bleeding in the context of CAA. It also highlights the value of multidisciplinary collaboration in achieving the best possible outcome for the patient. Furthermore, it demonstrates the remarkable neuroplasticity of the brain, as evidenced by the patient's significant recovery, despite multifocal hemorrhages.

Key words
anticoagulation, bleeding risk, hemorrhage, recurrent VTE, stroke

BRIAN BEYERS

Neuropathy in a diabetic patient: think again

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CASE DESCRIPTION Distal acquired demyelinating symmetric (DADS) neuropathy, a rare form of chronic inflammatory demyelinating polyradiculoneuropathy, can closely resemble distal sensory

polyneuropathy (DSP)—the most common neuropathy in diabetes. We present a case of a 35-year-old man with diabetes mellitus who developed distal sensory and mild motor deficits, initially misattributed to DSP. A detailed clinical evaluation, along with nerve conduction studies and borderline positive anti-GM1 antibodies, ultimately confirmed the diagnosis of DADS neuropathy. This case highlights key features of DADS neuropathy and features differentiating it from DSP. It also serves as a timely reminder of the diverse neuropathic manifestations in patients with diabetes.

Key words
distal acquired demyelinating symmetric neuropathy, diabetic neuropathy

DANIEL-NICOLÁS MARCO PRATS

Human herpes virus-8 and AIDS: old friends and a new foe

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INTRODUCTION Human herpesvirus-8 (HHV-8) is a gamma herpesvirus implicated in malignancies and inflammatory syndromes, particularly in immunosuppressed individuals. One of its associated diseases is Kaposi sarcoma inflammatory cytokine syndrome (KICS)—a recently recognized but underdiagnosed condition characterized by severe systemic inflammation driven by excessive cytokine release. It shares overlapping features with other HHV-8–related conditions, including Kaposi sarcoma (KS), multicentric Castleman disease (MCD), and primary effusion lymphoma (PEL), which complicates the diagnosis and management. Here, we present a rare case of a patient with debuting HIV/AIDS in advanced state who simultaneously developed KS, KICS, and an extracavitary variant of PEL, highlighting the diagnostic and therapeutic challenges posed by HHV-8–driven pathologies.

CASE DESCRIPTION A 36-year-old Paraguayan man with no prior medical history was admitted with complaints of general malaise, fatigue, abdominal distension, and symmetric lower limb edema. He reported a history of unprotected sexual intercourse and recurrent herpes zoster over the past year. Physical examination showed oral candidiasis, generalized lymphadenopathy, hepatosplenomegaly, ascites, and violaceous macular skin lesions suggestive of KS. Pulmonary findings included bibasilar crackles. Laboratory findings demonstrated severe immunosuppression (CD4, 4 cells/μl), pancytopenia, marked elevation of inflammatory marker levels and severe hypoalbuminemia, polyclonal hypergammaglobulinemia, and positive plasmatic viral loads for HIV-1, HHV-8, and Epstein–Barr virus. Imaging showed hepatosplenomegaly, intra-abdominal lymphadenopathies, and ascitic fluid without pleural effusions.

Skin biopsy confirmed KS, and further investigations raised a suspicion of MCD or a lymphoproliferative disorder. Given the profound systemic inflammation, hemophagocytic lymphohistiocytosis and KICS were also considered. Empirical antimicrobial therapy and high-dose corticosteroids were initiated. However, the patient rapidly deteriorated, developing acute liver failure, distributive shock, and multiorgan dysfunction requiring intensive care unit admission. Liver biopsy showed no infectious or lymphomatous infiltration, whereas lymph node biopsy showed polymorphic lymphoid proliferation with areas of necrosis, suggesting an extracavitary variant of PEL.

The final diagnosis was the concurrence of KS, KICS, and extracavitary PEL. Targeted therapy with rituximab, cyclophosphamide, corticosteroids, and foscarnet was initiated. Despite the aggressive treatment, the patient's condition continued to decline, leading to refractory shock, multiorgan failure, and death.

DISCUSSION This case illustrates the complex interplay between HHV-8 and HIV/AIDS, with simultaneous presentation of KICS, KS, and extracavitary PEL—3 distinct but interrelated pathologies disproportionately affecting immunosuppressed patients. HHV-8–driven oncogenesis and inflammation involve viral cytokine mimesis, immune evasion, and Epstein–Barr virus coinfection, which exacerbates the disease severity. The overlapping features of KICS with hemophagocytic lymphohistiocytosis, MCD, and PEL make diagnosis challenging and often require an involvement of several medical specialties. Antiretroviral therapy remains the cornerstone of management, but optimal treatment strategies for KICS and extracavitary PEL remain undefined, requiring a multidisciplinary approach.

CONCLUSIONS This case underscores the importance of recognizing HHV-8–driven pathologies in AIDS patients presenting with systemic inflammation and hypoalbuminemia, even in the absence of typical "B-symptoms." Increased clinical awareness and early intervention are critical, as these pathologies carry a high mortality rate if left untreated. The distinction between their underlying mechanisms has important implications for adapting therapeutic approaches. Future research should focus on refining diagnostic criteria and exploring targeted therapies for HHV-8–associated diseases.

Key words
AIDS, human herpes virus-8, Kaposi sarcoma inflammatory cytokine syndrome, multicentric Castleman disease, primary effusion lymphoma

ELENE TKABLADZE

First documented use of the Merit WRAPSODY stent graft for the treatment of a nutcracker type of pelvic congestion syndrome

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INTRODUCTION Pelvic congestion syndrome (PCS) is a condition characterized by chronic pelvic pain, resulting from pelvic venous congestion. Possible causes of PCS are primary venous insufficiency of the ovarian veins, compression of the left iliac common vein (May–Thurner syndrome), and compression of the left renal vein (nutcracker syndrome).

CASE DESCRIPTION A 30-year-old woman reported recurrent pain in the left flank and lower abdomen, described as a convulsive spasm that had progressively worsened over the past several years. The pain began following her first childbirth in 2015. Initially mild, the discomfort increased in intensity after the birth of her second child in 2019. The pain is mainly localized to the left flank and does not radiate to the back. It worsened when the patient stood up after prolonged sitting, and occurred mostly in the evenings. The patient reported no gastrointestinal or systemic symptoms. Initially, a gynecological examination with negative results and urinary analysis with a finding of microhematuria were performed. Ultrasound examination showed findings consistent with nutcracker syndrome. Magnetic resonance angiography and computed tomography angiography were performed in order to evaluate the venous system, and confirmed the suspected left renal vein compression and pelvic venous congestion. Based on the combination of clinical and diagnostic findings, a decision was made to perform phlebography, and the left renal vein was treated with a Merit WRAPSODY™ stent graft (14 mm × 40 mm) —a unique endoprosthesis with maximal radial resistive force, typically used for dialysis shunts. Therapy with acetylsalicylic acid, 100 mg was initiated. The patient's symptoms resolved and the stent graft has been working well.

CONCLUSIONS In our experience, the endovenous technique appears to be the best initial treatment for nutcracker syndrome. This case

highlights the successful use of the Merit WRAPSODY stent graft in the treatment of nutcracker syndrome caused by the left renal vein compression. However, further research should be conducted to determine the long-term effectiveness of the treatment in relieving chronic pelvic pain and other complications associated with PCS.

Key words
nutcracker syndrome, pelvic congestion syndrome, Wrapsody stent graft

ERIK BÉNYEI

Peritoneal implantation of pheochromocytoma: pheochromocytomatosis

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INTRODUCTION Pheochromocytomas and paragangliomas are catecholamine-producing tumors originating from the adrenal medulla and sympathetic ganglia. The standard treatment of adrenal pheochromocytomas is surgical removal. Pheochromocytomatosis, defined as the implantation of pheochromocytoma cells limited to the space opened operatively during surgical manipulation, is an infrequent complication of surgical intervention. Only a handful of pheochromocytomatosis cases have been reported since the first case was described in 2001.

CASE DESCRIPTION In 2011, a 33-year-old man presented with episodic palpitations and hypertensive surges triggered by physical activity. Imaging showed a left adrenal tumor with intense radiopharmaceutical uptake on iodine-131 metaiodobenzyl guanidine (^{131}I-MIBG) scintigraphy. Urinary metanephrines confirmed pheochromocytoma and laparoscopic left-sided adrenalectomy was performed. Owing to its large size, intraoperative fragmentation was necessary for tumor removal. The patient remained asymptomatic for 5 years. In 2016, recurrent paroxysmal symptoms prompted imaging, which showed a lesion in the left renal hilum. In 2017, repeat surgery was performed, during which multiple peritoneal tumor deposits were observed and later confirmed histologically. Over the following years, the patient received conservative, symptomatic treatment with tolerable paroxysmal symptoms. In 2023, worsening symptoms led to the decision to commence ^{131}I-MIBG therapy. Despite treatment, biochemical and radiological tumor progression was observed; hence, a systematic tyrosine kinase inhibitor therapy with sunitinib was initiated, analogously to the treatment of advanced, metastatic pheochromocytomas.

DISCUSSION Intraoperative tumor capsule rupture and peritoneal seeding can lead to one of the rarest forms of pheochromocytoma tumor progression—pheochromocytomatosis. Case reports describe prolonged asymptomatic postsurgical intervals, emphasizing the need for long-term follow-up. Treatment strategies parallel those used for advanced pheochromocytomas, including cytoreductive surgery, peptide receptor radionuclide therapy, somatostatin analogues, and targeted systematic therapies, such as tyrosine kinase inhibitors. Future genetic studies may further improve management by identifying novel targets for systemic therapies.

CONCLUSIONS If capsule rupture is documented after surgical manipulation of pheochromocytoma, close biochemical and radiological surveillance is key for timely diagnosis and early treatment of pheochromocytomatosis. Ultimately, a multidisciplinary approach is required for patient management and disease control.

Key words
paraganglioma, pheochromocytoma, pheochromocytomatosis

JOANA NUNES

Aseptic abscess syndrome: unravelling a rare cause of fever and abscesses in inflammatory bowel disease

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CASE DESCRIPTION Aseptic abscess syndrome (AAS) is a rare condition characterized by the development of deep abscesses, usually in the internal organs, systemic symptoms, and elevated levels of inflammatory markers. In most cases, an associated condition is present, primarily inflammatory bowel disease. The diagnosis is challenging, based on evidence of deep abscesses, negative microbiological and serological tests, failure of antibiotic therapy, and good response to glucocorticoid (GC) therapy. Maintenance therapy should include a conventional or biological disease-modifying antirheumatic drugs. We present a case of a 37-year-old woman with ulcerative colitis on prednisolone. She presented with malaise, fever, diarrhea, and pleuritic left chest pain. *Clostridium difficile* colitis was diagnosed, and she further developed a severe, extensive pneumonia with no response to antibiotics or antifungals medication, and no microbiological identification, resulting in acute respiratory distress syndrome. The patient was started on GC therapy and antibiotic therapy was discontinued. Subsequently, respiratory and inflammatory markers improved remarkably, although low-grade fever persisted. Simultaneously, new radiological evidence of lung cavitations and multiple disseminated muscular abscesses was obtained. On multiple occasions, aspiration of the purulent fluid from those abscesses was sterile. The fluid was polymorphonuclear and neutrophil-rich, suggesting a hypothesis of aseptic abscess syndrome. The GC treatment was continued and a slow clinical and radiological improvement was seen. Three months later, the patient was started on adalimumab and almost complete resolution of abscesses was achieved. Although this is an atypical presentation (rare locations, late development of abscesses, and slow response to GC) of a rare condition, we believe there is no better alternative diagnosis. As a rare entity, AAS is often misdiagnosed as an infection and a diagnosis of exclusion is necessary. It is expected that new presentations and outcomes will come to light in the next few years; therefore, this condition must be considered even if not all criteria are met.

Key words
aseptic abscess syndrome, fever, inflammatory bowel disease

KLÁRA DOMBROVSKÁ

When the body rebels

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INTRODUCTION Atypical hemolytic uremic syndrome (aHUS) is a rare, potentially life-threatening disorder characterized by microangiopathic hemolytic anemia, thrombocytopenia, and renal dysfunction. Early recognition and appropriate treatment are crucial in order to prevent severe organ damage. We present a case of a young woman with rapidly progressing renal and respiratory failure, who was ultimately diagnosed with aHUS and treated with eculizumab.

CASE DESCRIPTION A 19-year-old woman with a history of nausea, vomiting, and weakness was admitted to the internal medicine department with acute kidney injury and respiratory insufficiency caused by diffuse alveolar hemorrhage. Shortly after admission, the patient developed respiratory failure requiring mechanical ventilation followed by extracorporeal membrane oxygenation and renal failure requiring renal replacement therapy. Due to the severity of the condition and the risk of treatment delay in the case of systemic diseases, such as vasculitis or lupus, exchange plasmapheresis was performed. Furthermore, rituximab and corticosteroids were administered. Over time, the patient's respiratory status improved, leading to extubation; however, renal dysfunction persisted and required intermittent hemodialysis. Kidney biopsy showed thrombotic microangiopathy. Laboratory findings indicated thrombocytopenia (which was not present on admission) and decreased complement component 3 (C3) levels, while immunological markers, such as antineutrophil cytoplasmic antibodies, anti-glomerular basement membrane antibody (anti-GBM), and antinuclear antibody (ANA) were negative. Based on the clinical picture, laboratory findings, and kidney biopsy results, the cause was determined to be aHUS. No secondary cause was found. Treatment with eculizumab was initiated, which led to further clinical improvement. Hematology markers of thrombotic microangiopathy normalized but renal failure has persisted, requiring dialysis treatment. Genetic testing confirmed a mutation in complement genes and long-term treatment with eculizumab was indicated.

DISCUSSION This case presents the diagnostic challenge of aHUS, especially in the case of incompletely expressed laboratory findings. Given the respiratory failure and diffuse alveolar hemorrhage, systemic vasculitis was initially considered, but the negative antibody results (anti-proteinase3, anti-myeloperoxidase, anti-GBM, ANA) and renal biopsy did not confirm this diagnosis. The presence of microangiopathic thrombosis on renal biopsy, thrombocytopenia, and low C3 levels made aHUS more likely than other causes of thrombotic microangiopathy (eg, thrombotic thrombocytopenic purpura or secondary hemolytic uremic syndrome). Rapid initiation of treatment with eculizumab, a C5 inhibitor, was essential in preventing further systemic complications.

CONCLUSIONS This case emphasizes the importance of early recognition of aHUS in critically ill patients with renal and respiratory failure, thrombocytopenia, and hemolytic anemia. Rapid intervention with targeted therapies, including eculizumab, may significantly improve patient outcomes.

Key words
acute renal failure, atypical hemolytic uremic syndrome, eculizumab

LESLIE NAESENS

From gut to gene: how colitis led to the diagnosis of a rare immunodeficiency

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INTRODUCTION Cytomegalovirus (CMV) colitis and Epstein–Barr virus (EBV) reactivation are typically associated with immunocompromised individuals. However, their occurrence in seemingly immunocompetent patients may indicate an undiagnosed inborn error of immunity (IEI). This case highlights the diagnostic challenges and clinical implications of severe viral infections in an adult who was considered to be immunocompetent.

CASE DESCRIPTION A 30-year-old woman presented with persistent fever, abdominal pain, weight loss, and diarrhea. After an extensive diagnostic workup, CMV colitis was confirmed via histopathology and CMV polymerase chain reaction in peripheral blood, necessitating intravenous ganciclovir administration. Despite clinical recovery, persistent lymphocytosis and concurrent high EBV viral loads in blood raised concerns about an underlying immune disorder. Positron emission tomography / computed tomography showed hepatosplenomegaly and hypermetabolic lymphadenopathy. Immunological assessment showed activated T-cell phenotypes, impaired lymphocyte proliferation, and absent polysaccharide antibody response, suggesting combined immunodeficiency. Whole exome sequencing identified pathogenic biallelic variants in the *SKIV2L* gene, associated with trichohepatoenteric syndrome, a rare syndromic autosomal recessive disorder linked to immune dysregulation. Retrospective assessment of the patient's childhood medical history showed subtle phenotypic features of trichohepatoenteric syndrome, including brittle hair and gastrointestinal malabsorption. Inflammatory cytokine profiling showed chronic T-cell activation despite impaired viral control. Due to persistent EBV viremia and the risk of lymphoproliferative disease, rituximab therapy was initiated, successfully leading to EBV control. Ongoing management includes infection surveillance, vaccination, and consideration of immunoglobulin replacement therapy. Given the chronic immune activation and inflammatory cytokine signature, the patient remains at a risk of autoinflammatory and autoimmune manifestations, requiring long-term immunological monitoring.

CONCLUSIONS This case underscores the importance of comprehensive immunologic and genetic evaluation in patients presenting with unusual severe viral infections. It highlights the role of whole exome sequencing in diagnosing IEIs, the clinical spectrum of *SKIV2L*-associated immunodeficiency, and the need for individualized management. Beyond susceptibility to infections, IEIs may drive chronic immune activation and autoinflammatory manifestations, complicating disease management. Early recognition and targeted interventions can improve patient outcomes by balancing immune suppression with infection control.

Key words
colitis, combined immunodeficiency, cytomegalovirus, inborn error of immunity, trichohepatoenteric syndrome

MAIA AGOSTINA LEPORE

A 57-year-old welder with altered liver biochemistry and Kaposi sarcoma: a rare case with a unifying hypothesis

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INTRODUCTION Hyperferritinemia, defined as serum ferritin levels above 200 µg/l in women and 300 µg/l in men, is often a nonspecific finding associated with acute phase reactions characterized by systemic inflammatory responses and elevated biomarker levels. In other instances, it can be caused by iron overload conditions, such as hemochromatosis, or by chronic liver diseases, including alcoholic liver disease and metabolic dysfunction–associated steatotic liver disease (MASLD). Occasionally, it is a finding observed in far rarer conditions that, nevertheless, may be suspected due to their syndromic presentation.

CASE DESCRIPTION A 57-year-old Caucasian lifelong welder presented to the liver clinic of an academic hospital with elevated levels of transaminases and pruritus. His medical history included asthma, interstitial lung disease related to welder's lung, past hepatitis C virus infection treated with interferon, evidence of previous hepatitis B virus contact, remote at-risk alcohol intake (always with normal liver biochemistry), and bilateral aseptic necrosis of the femoral head linked to prolonged steroid therapy for atopic dermatitis. One year before presentation, the patient had started taking upadacitinib, a Janus kinase inhibitor, for the latter condition. During upadacitinib treatment,he underwent regular laboratory monitoring that consistently showed elevated transaminase levels. Moreover, multiple, purple, papular lesions appeared on his limbs and later spread to other areas of the skin. These findings were investigated through liver biopsy, which showed micro- and macronodular cirrhosis with severe iron overload, and skin biopsy, which confirmed Kaposi sarcoma (KS) and human herpesvirus-8 (HHV-8) positivity. Serum ferritin level was 4971 µg/l. There was no history of exposure to blood products and testing for *HFE* gene mutations yielded negative results. The combination of 3 rare conditions in a relatively young person (primary, non–*HFE*-related iron overload, KS, and welders' lung) was puzzling and prompted us to search for a single explanation.
DISCUSSION Welder's lung (also known as pulmonary siderosis) is an occupational disease caused by long-term inhalation of welding fumes, leading to desquamative interstitial pneumonia and respiratory dysfunction. It has recently been proposed that welder's lung may be associated with systemic iron overload through mechanisms distinct from those observed in hemochromatosis. KS is an angioproliferative disorder that requires HHV-8 infection to develop. It is classified into 4 types: classic (typically affecting older individuals), endemic (in sub-Saharan Africa), iatrogenic (due to immunosuppressive therapies), and AIDS-associated. While HHV-8 is essential for the development of KS, other cofactors are also necessary. In this case, 2 factors may have triggered HHV-8 activation: Janus kinase inhibitor therapy and hyperferritinemia resulting from prolonged exposure to welding fumes. Both factors find support in the literature, which also contains a case report of KS in a 43-year-old welder.
CONCLUSIONS This case is notable because it highlights how rare combinations of diseases can sometimes point to a specific pathophysiological predisposition. Systemic iron overload due to occupational exposure, as evidenced by hyperferritinemia, contributed to advanced liver disease and served as a risk factor for the development of a rare cancer. It also underscores the importance of broadening our differential diagnosis of hyperferritinemia in complex clinical presentations.

Key words
hyperferritinemia, Kaposi sarcoma, welder's lung

MARIANA PEREIRA

When family history illuminates the path: an uncommon cause of esophageal stricture

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INTRODUCTION Multiple endocrine neoplasia type 1 (MEN1) is a rare autosomal dominant disorder caused by mutations in the *MEN1* tumor suppressor gene, leading to the development of tumors in multiple endocrine organs. It commonly affects the parathyroid glands and can lead to pancreatic neuroendocrine tumors and gastroduodenal tumors, such as gastrinomas. These gastrin-secreting tumors can cause Zollinger–Ellison syndrome (ZES), leading to severe gastric acid hypersecretion, peptic ulcers, and, in rare cases, esophageal stricture.

CASE DESCRIPTION We report a case of a 47-year-old woman with a long history of dyspepsia, epigastric pain, nausea, and vomiting. She was initially diagnosed with gastritis and Los Angeles grade D erosive esophagitis. Over time, she developed progressive dysphagia and severe weight loss (20 kg in 4 months). Investigations revealed hypercalcemia secondary to primary hyperparathyroidism and upper gastrointestinal endoscopy identified severe reflux esophagitis with a significant esophageal stricture. A detailed review of the patient's medical history uncovered a family history of brain tumors in her mother, and siblings with urolithiasis. Given the combination of primary hyperparathyroidism, chronic severe esophagitis, and a notable family history, MEN1 and gastrinoma were suspected. A positron emission tomography scan with octreotide showed an 11-mm hypermetabolic lesion in the duodenal bulb, consistent with a well-differentiated neuroendocrine tumor, along with a 5-mm pancreatic lesion. Duodenal biopsy confirmed the presence of a neuroendocrine tumor. Furthermore, pituitary magnetic resonance imaging detected a 6-mm microadenoma, classified as nonfunctioning based on endocrine studies. Genetic testing identified a pathogenic *MEN1* mutation, confirming the diagnosis. The patient subsequently underwent a subtotal parathyroidectomy. To manage the esophageal stricture, she received acid suppression therapy, an endoprosthesis was placed, and subsequent esophageal dilations were performed. Due to the small size of the gastrinoma (<2 cm), a conservative approach with close monitoring was chosen over immediate surgical resection.
DISCUSSION This case highlights an atypical presentation of MEN1 with severe esophageal stricture secondary to ZES, caused by a gastrinoma. Characterized by excessive gastrin secretion, ZES often remains undiagnosed due to its rarity and overlap with other gastrointestinal disorders. In this case, due to the severity of the stricture, hypergastrinemia could not be confirmed, but the clinical context strongly supported ZES. The diagnosis was suggested by the presence of parathyroid hormone–dependent hypercalcemia and a relevant family history, prompting suspicion of an underlying genetic condition. This highlights the essential role of a comprehensive medical history and a systematic approach in diagnosing rare and complex diseases.
CONCLUSIONS This case underscores the necessity of screening for gastrinomas in patients with severe gastroesophageal reflux or refractory symptoms. It highlights key clinical features of MEN1 and emphasizes the pivotal role of a detailed medical and family history in the diagnostic process.

Key words
esophageal stricture, gastrinoma, multiple endocrine neoplasia, Zollinger–Ellison syndrome

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Blood, brain, and a hidden battle: a case of eosinophilia and chronic strokes

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CASE DESCRIPTION There are many possible causes of stroke in young patients, including rare conditions such as hypereosinophilic syndrome, in which excessive eosinophil production can promote clotting and vascular damage. Schistosomiasis, a parasitic infection that primarily affects the gastrointestinal and genitourinary systems, is an uncommon but recognized cause of eosinophilia; however, its role in embolic stroke is rarely reported. This case report describes a 23-year-old man from Guinea who presented with

a seizure. Further evaluation showed multiple embolic infarcts and persistent eosinophilia, raising the suspicion of hypereosinophilic syndrome. The patient's history of travel from an endemic region, along with serological and parasitological testing, ultimately led to a diagnosis of *Schistosoma mansoni* infection as the underlying cause. Treatment with praziquantel initially triggered an expected temporary immune response, causing a surge in eosinophils before the condition resolved itself. This case highlights the diagnostic challenge of differentiating parasitic infections from other causes of hypereosinophilic syndrome in young patients with a stroke. Given the overlap in presentation, clinicians should consider parasitic infections as a potential etiology of embolic stroke and eosinophilia, particularly in individuals from endemic regions.

Key words
embolic stroke, eosinophilia, hypereosinophilic syndrome, *Schistosoma mansoni*, seizure

SHASHANK RAVI GANESH

Typhoid malady: a wolf in sheep's clothing

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INTRODUCTION Typhoid fever, a systemic infection caused by *Salmonella typhi*, manifests with fever, abdominal pain, and gastrointestinal symptoms. In some instances, particularly in individuals with undiagnosed immunodeficiency disorders, it can lead to severe complications, such as hemophagocytic lymphohistiocytosis (HLH) and meningitis, making management more challenging. This case report discusses a 28-year-old Indian man who presented with a 10-day history of fever and vomiting.

CASE DESCRIPTION A 28-year-old man from southern India presented with a 10-day history of high-grade fever, vomiting, and watery diarrhea. Initial improvement with oral cefixime and paracetamol was followed by symptom deterioration. His medical history was notable for recurrent gastrointestinal infections triggered by outside food consumption. On admission, he exhibited stable vitals, but physical examination showed pallor, mild icterus, hepatosplenomegaly, and epigastric tenderness. Laboratory investigations indicated mild anemia (hemoglobin, 12.4 g/dl), thrombocytopenia (76 000/mm^3), and elevated liver enzymes. Widal test was positive for *S. typhi* (typhi-H, 1:320) and urinalysis showed *Escherichia col*i growth sensitive to meropenem, with pyuria evident in urine cultures. Despite ceftriaxone therapy for typhoid fever and meropenem escalation for urinary tract infection, the patient's condition deteriorated. Further assessment uncovered hyperferritinemia (>35 000 ng/ml), elevated triglyceride levels, and an H-score of 244, indicating HLH. Comprehensive screening for infectious triggers (Epstein–Barr virus, cytomegalovirus, herpes simplex virus), autoimmune etiologies (antinuclear antibodies), and HIV infection was negative. Initial dexamethasone treatment led to temporary improvement; however, the patient relapsed after 8 days with recurrent fever, vomiting, and altered sensorium. Magnetic resonance imaging of the brain was unremarkable, but cerebrospinal fluid and paired blood cultures confirmed disseminated *S. typhi* infection. Suspecting an underlying immunodeficiency, clinical exome sequencing identified a hemizygous mutation in the *SH3KBP1* gene, linked to X-linked primary immunodeficiency. Bone marrow studies demonstrated hemophagocytosis, leading to a diagnosis of primary HLH triggered by typhoid fever. The patient was initiated on the HLH-94 protocol, coupled with a meningitic dose of ceftriaxone and azithromycin. Despite initial response, complications arose, including intracranial bleeding and brainstem dysfunction, culminating in mortality prior to the planned hematopoietic stem cell transplantation.

DISCUSSION Typhoid fever is a significant health concern that can lead to complications, such as HLH and, rarely, meningitis. Persistent infections despite appropriate treatment may indicate underlying immunodeficiency. In this case, the patient had a mutation in the *SH3KBP1* gene, resulting in X-linked immunodeficiency, recurrent infections, and primary HLH triggered by typhoid fever. Hemophagocytic lymphohistiocytosis is a serious immune dysregulation disorder often instigated by infections and associated with high mortality if untreated. Diagnosis relies on clinical assessment, laboratory markers, like elevated ferritin and cytopenia, and bone marrow findings. Early treatment with immunosuppressives and effective infection management is crucial for improving survival rates. For cases of primary HLH, hematopoietic stem cell transplantation may be necessary.

CONCLUSIONS Typhoid fever is still prevalent and can cause a myriad of complications including meningitis. Persistent symptoms warrant thorough investigation to look for other causes, such as HLH. Prompt initiation of treatment for the infection, timely recognition of complications, and comprehensive management with a multidisciplinary approach are crucial when addressing complex cases.

Key words
complicated typhoid fever, hemophagocytic lymphohistiocytosis, meningitis, primary immunodeficiency, *Salmonella*

VIRGINIA HUHN

Now you see me: Bazin disease

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INTRODUCTION Systemic autoimmune diseases are known to affect both humoral and cellular immunity, rendering patients with these conditions particularly vulnerable to infections, including tuberculosis. The latter is a re-emerging disease, often presenting with extrapulmonary manifestations.

Differentiating between infections and reactivation poses a significant challenge, particularly when confronted with atypical presentations of common diseases.

CASE DESCRIPTION The patient was a 28-year-old cisgender woman with a history of systemic lupus erythematosus (SLE) characterized by mucocutaneous involvement, arthritis, renal disease, positive antiphospholipid antibodies, nonspecific cholestatic hepatitis, and a positive result of the QuantiFERON-TB test, performed as part of clinical trial screening. She was admitted following a monthlong history of daily fever and night sweats, accompanied by generalized nodular skin lesions and ulcers in the lower limbs. The initial clinical impression suggested reactivation of SLE. Computed tomography scan of the chest showed micronodular lesions and the tree-in-bud pattern. Bronchoalveolar lavage was negative for acid-fast bacilli, while transbronchial biopsy showed interstitial granulomas. Skin biopsy findings included lobular panniculitis with vasculitis and granulomas at both septal and lobular levels. Ziehl–Neelsen and periodic acid–Schiff stains were negative. The patient developed lower back pain, leading to a diagnosis of tuberculous spondylitis (Pott disease) at D11 and L4 vertebral levels. The diagnosis was confirmed by positive GeneXpert polymerase chain reaction test result in a bone sample and biopsy findings consistent with granulomas containing necrotic caseous centers. First-line antitubercular therapy was initiated and the patient completed a 1-year treatment regimen. Due to poor control of the underlying disease,

mycophenolate was introduced alongside ongoing prednisone and hydroxychloroquine therapy.

One month after completing the antitubercular treatment, the patient experienced a significant worsening of her skin lesions, characterized by deep ulcerations and inadequate pain control, predominantly in the gluteal region. Repeat skin biopsy showed granulomatous dermatosis with caseous necrosis, interpreted as Bazin disease. Antitubercular therapy was reinitiated, resulting in improvement of the ulcerations in the following months.

As the managing team, we were faced with a very elusive diagnosis of disseminated tuberculosis.

Over the course of a year, extensive efforts were made to identify the bacillus, during which time the patient experienced significant impairments in her quality of life.

CONCLUSIONS Bazin disease is a rare form of necrotizing granulomatous dermatitis characterized by the appearance of deep ulcers. It is typically associated with tuberculosis infection and, more rarely, with SLE. In this case, we postulate that the presence of a refractory autoimmune disease, requiring multiple regimens of immunosuppressants, made our patient particularly susceptible to this condition.

In our clinical reasoning, we initially aligned with the principle of Occam's razor, where the simplest diagnosis initially pointed to lupus panniculitis, despite the inherent rarity of this condition. Alternatively applying Hickam's dictum, we considered that the patient might have multiple coexisting conditions, thereby broadening the diagnostic possibilities. By emphasizing the importance of clinical reasoning and a structured diagnostic method, we were ultimately able to guide our actions, arrive at an accurate diagnosis, and significantly improve the patient's quality of life.

Key words
Bazin disease, panniculitis, systemic lupus erythematosus, systemic tuberculosis

YIN NAN HUANG

The vessel of warning: a case of unexplained recurrent arterial thrombosis

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CASE DESCRIPTION Paraneoplastic vasculitis is an uncommon paraneoplastic syndrome in which the diagnosis is based on temporal correlation, rather than confirmatory tests, and requires high clinical suspicion.

A 49-year-old Caucasian man was referred to our tertiary hospital for the investigation of recurrent bilateral renal and splenic infarcts. Relevant medical history included myocardial infarction, hypertension, dyslipidemia, and a patent foramen ovale (PFO). Noteworthy clinical features included unintentional weight loss of 35 lb (15 kg) and hematuria. The patient denied other constitutional symptoms and the review of systems was otherwise negative. Initial angiographic imaging showed significant atherosclerosis of the transverse aorta, left subclavian artery, and bilateral iliac arteries. However, there were no signs of vascular dysplasia, vasculitis or other vascular anomalies. Doppler ultrasound of the lower limbs was performed and showed no thrombosis. The patient was started on therapeutic anticoagulation with dalteparin and was discharged with outpatient investigations. Despite therapeutic anticoagulation, he developed subsequent splenic and renal infarcts. An old cerebellar lesion was also detected on cerebral computed tomography.

The patient underwent a comprehensive diagnostic workup for unexplained arterial thrombosis, ruling out antiphospholipid syndrome, coagulation factor mutations, anticoagulant deficiencies, and hyperhomocysteinemia. Cardiac investigations showed a known small PFO, for which he underwent PFO closure. Subsequent computed tomography angiography showed parietal thickening of a segmental renal artery, raising suspicion of vasculitis without clinical correlation. The patient's inflammatory marker levels remained normal and additional tests—including rheumatologic panels, mutation screenings for myeloproliferative syndromes, flow cytometry for lymphoproliferative syndromes, and paroxysmal nocturnal hemoglobinuria—were negative.

Despite undergoing PFO closure, the patient developed another renal infarct and was readmitted after experiencing 3 episodes of transient dysarthria, right upper limb paresis, and paresthesia. His symptoms resolved upon admission and magnetic resonance imaging of the brain showed 2 new ischemic cortical infarcts. Further investigation with a whole-body positron emission tomography scan showed hypermetabolism of the aortic arch and its proximal branches, which was compatible with large vessel vasculitis. A comprehensive microbiology panel was conducted to rule out infectious aortitis. Negative findings for giant cell arteritis on temporal artery ultrasound led to a diagnosis of Takayasu-like vasculitis, despite its epidemiological improbability. However, the patient did not respond to immunosuppressive therapy. Two months later, he was readmitted again after experiencing yet another renal infarct. Follow-up positron emission tomography scan showed new hypermetabolic lesions affecting the adrenal glands, pancreas, bones, lungs, and urogenital region. Previous aortic hypermetabolism was still present and had even increased in some segments. These findings raised suspicion of a malignant process, leading to a shift in diagnosis from primary Takayasu-like vasculitis to malignancy with paraneoplastic large vessel vasculitis.

This is an interesting case of unexplained arterial thromboses, ultimately caused by a hypercoagulable state secondary to malignancy, with associated paraneoplastic vasculitis. This diagnosis should be suspected in cases of vasculitis with atypical clinical features and a lack of response to conventional treatment. It is also important to note that up to 20% of paraneoplastic vasculitis cases can manifest before the first signs of underlying malignancy. Therefore, appropriate cancer screening should be conducted in patients with unexplained arterial thromboses and atypical vasculitis.

Key words
arterial, paraneoplastic, thrombosis, vasculitis

YÜSRA AĞAOĞLU

Misdiagnosis risk in tuberculosis-endemic regions: a multidisciplinary case report

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CASE DESCRIPTION A 58-year-old woman from a tuberculosis-endemic region in Turkey, with a history of livestock farming presented to an external institution in 2019 due to complaints of chronic back pain. Imaging performed upon her presentation showed a large, calcified mass, invading multiple abdominal and spinal structures. Given her history and the imaging findings, tuberculosis was considered as a highly likely diagnosis and antitubercular therapy was initiated. In 2024, the patient presented to our internal medicine clinic at Cerrahpaşa Faculty of Medicine, with newly developed symptoms consistent with cauda equina syndrome, including urinary incontinence, balance disturbances, and walking impairment, which prompted further investigation.

The patient was evaluated by a multidisciplinary team and spinal decompression surgery was performed due to worsening neurological symptoms. Intraoperative biopsy showed lamellar structures positive for periodic acid–Schiff and Grocott staining, and negative for

Ehrlich–Ziehl–Neelsen staining, highly suggestive of a parasitic infection. The absence of bacterial growth and the infiltrative nature of the lesion raised suspicion for *Echinococcus multilocularis,* rather than tuberculosis. Serological tests confirmed echinococcosis, leading to the immediate discontinuation of antitubercular therapy. Given the extensive organ involvement and the infiltrative nature of the disease, a staged surgical approach was implemented, followed by lifelong antiparasitic therapy. The case underscores the need for improved differential diagnosis in tuberculosis-endemic areas.

Following the definitive diagnosis, the patient underwent hepatic segment 5–6 resection, right nephrectomy, right adrenalectomy, and inferior vena cava resection, followed by L3–L4 corpectomy and expandable cage placement to restore spinal stability. Postoperatively, she remained stable and continued albendazole therapy, with ongoing rehabilitation and radiological monitoring.

CONCLUSIONS This case highlights the diagnostic challenges of *E. multilocularis* in tuberculosis-endemic regions, where necrotizing granulomatous inflammation often leads to misdiagnosis. The initial spinal decompression not only prevented further neurological deterioration but also played a pivotal role in establishing the correct diagnosis. A stepwise, multidisciplinary approach is essential in such cases to prevent delays in appropriate treatment and improve outcomes.

Key words
Echinococcus multilocularis, granulomatous inflammation, parasitic infections, tuberculosis misdiagnosis, zoonotic diseases

POSTER PRESENTATIONS

BEST POSTER RECOGNITION: NATALIE GROSUP FRIEDOVA

Corn starch in the therapy of paraneoplastic hypoglycemia

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INTRODUCTION Paraneoplastic hypoglycemia is a rare but critical condition, typically observed in patients with various malignancies. This condition is frequently associated with tumors that secrete insulin-like growth factors such as insulin-like growth factor 2 (IGF-2). IGF-2 and its precursors interfere with glucose homeostasis leading to uncontrolled glucose consumption. An extensive range of tumors, such as vascular, epithelial, or mesynchemal-origin tumors, has been previously described as associated with paraneoplastic hypoglycemia. In the first place, various treatment strategies include surgical tumor resection and, alternatively, pharmacological interventions (such as corticosteroids and glucagon) or dietary precautions.

CASE DESCRIPTION A 75-year-old woman with a history of pleural and right lung fibrosarcoma (in progression at the time; indicated symptomatic treatment) and non-Hodgkin lymphoma (in remission) was transferred from the neurological ward, where she was hospitalized for confusion and recurrent falls. Intracranial pathology was ruled out as a contributing factor during the initial hospital assessment. Due to repeated episodes of severe hypoglycemia (1.2–3 mmol/l), the patient was subsequently transferred to an internal medicine ward for further diagnostic evaluation and management.

During hospitalization in our ward, the patient experienced recurring hypoglycemic episodes without loss of consciousness, despite the implementation of dietary modifications and parenteral glucose infusions. Laboratory evaluation showed markedly low C-peptide levels (72 pmol/l), while morning serum cortisol was normal (404 nmol/l). Clinical suspicions of insulinoma and adrenocortical insufficiency were ruled out.

As normoglycemia could not be maintained through specialized dietary modifications, parenteral nutrition was initiated. Corticosteroid therapy with prednisone (25 mg daily) and evening administration of corn starch (6 tablespoons dissolved in 200 ml of water) were introduced to stabilize blood glucose levels. Subsequently, glycemic control was satisfactory with this regimen, eliminating the need for ongoing parenteral nutrition. The patient was discharged with arrangements for home care.

DISCUSSION The diagnosis of paraneoplastic hypoglycemia can be challenging. It usually requires confirmation of symptomatic hypoglycemia and exclusion of other potential causes. Diagnostic evaluation often involves laboratory assessments (measuring IGF2 levels and the IGF-2:IGF-1 ratio) and imaging studies. Currently, IGF-2 levels quantification is not available in standard hospital laboratories in the Czech Republic. At present, we perform IGF-2 measurements in collaboration with Czech Centre for Phenogenomics.

CONCLUSIONS The patient's prognosis remains uncertain; however, the therapeutic intervention with glucocorticoids and corn starch resulted in a marked improvement in her quality of life, which had previously been significantly compromised by frequent hypoglycemic episodes, thereby facilitating discharge to home care. This case contributes valuable insights into the challenges of diagnosing and managing paraneoplastic hypoglycemia. Enhanced understanding of the differential diagnostics process will support more informed clinical decision-making for future patients presenting with suspected paraneoplastic hypoglycemia in the Czech Republic.

Key words
corticosteroid therapy, corn starch, fibrosarcoma, insulin-like growth factor 2, paraneoplastic hypoglycemia

BEST POSTER RECOGNITION: OLGIERD DRÓŻDŻ

De novo type 1 diabetes in a patient with lupus nephritis treated with steroids and a history of proximal venous thrombosis

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INTRODUCTION Chronic glucocorticoid (GC) therapy is commonly used across various specialties due to its broad therapeutic effects. However, prolonged use of these drugs is associated with 2 key diagnostic challenges: an increased risk of venous thromboembolism and difficulty in diagnosing diabetes. Steroid-induced diabetes can occur de novo or as an exacerbation of pre-existing hyperglycemia. Appropriate insulin therapy, often combined with metformin, and reduction in GC dosage are essential for effective glucose control. Although chronic GC use typically suggests steroid-induced diabetes, this case report demonstrates the need for a comprehensive diagnostic approach.

CASE DESCRIPTION A 28-year-old man with a 9-year history of class IV lupus nephropathy was admitted for evaluation of nephrotic syndrome treatment. Previously, he was treated with prednisone therapy (10–60 mg/day). A month prior to the hospitalization, the patient was taking 30 mg of prednisone, 1250 mg of mycophenolate mofetil, and chloroquine for skin symptoms related to systemic lupus erythematosus. He was treated for proximal deep vein thrombosis of the right lower limb, managed with stenting and thrombectomy. Upon admission, the patient presented with increased thirst, polyuria, and worsening visual acuity. Laboratory test results showed fasting hyperglycemia (438 mg/dl) and typical nephrotic syndrome markers, including hypercholesterolemia, hypoalbuminemia, and proteinuria.

A diabetic consultation was requested, during which the diagnostic workup was expanded to include the measurement of fasting C-peptide levels and testing for antiglutamic acid decarboxylase (anti-GAD) antibodies. The glycated hemoglobin (HbA_{1c}) level was 11.7%, C-peptide was at the lower end of the normal range, and anti-GAD antibody result was positive. Based on this, type 1 diabetes (T1D) was diagnosed and a basal-bolus insulin regimen was initiated (glargine and lispro). Continuous glucose monitoring was implemented and nephrotic syndrome treatment was adjusted. Tests for antiphospholipid syndrome and genetic thrombophilias were performed, with results pending.

One month later, the patient was admitted for insulin dose education, based on carbohydrate exchanges. The level of HbA_{1c} improved to 8.4%, with 90% time-in-range. The C-peptide level was slightly below normal.

DISCUSSION Patients with autoimmune diseases on immunosuppressants may develop hyperglycemia in various ways. Distinguishing between T1D and steroid-induced diabetes is crucial, as it guides treatment. Steroid-induced diabetes, in contrast to T1D, is not caused by an autoimmune process, but results from insulin resistance and altered glucose metabolism. Diagnosis is made using general diagnostic criteria for diabetes, but steroid-induced diabetes is often detected through routine glucose monitoring. Regular glucose monitoring is essential in patients on long-term GC therapy.

CONCLUSIONS Steroid-induced diabetes lacks clear diagnostic guidelines, requiring clinical evaluation based on patient history. Expanding diagnostic workup with C-peptide and anti-GAD testing can help avoid misdiagnosis. Early misdiagnosis and premature treatment can lead to serious complications. Genetic factors, such as antiphospholipid syndrome or prothrombin gene mutations, should also be considered in younger patients on steroid therapy, as these factors increase thrombosis risk. Early detection improves prevention and treatment.

Key words
C-peptide, diabetes mellitus, glucocorticoid therapy, insulin therapy, venous thromboembolism

BEST POSTER RECOGNITION: ROWAN NELSON

Successful rechallenge of rifamycin in rifampicin-induced thrombocytopenia

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CASE DESCRIPTION Drug-induced immune thrombocytopenia is an important, life-threatening and treatment-limiting adverse drug reaction (ADR) associated with first-line antitubercular (TB) drug regimens. Most frequently, the offending drug is rifampicin. Understanding the management of ADRs in TB treatment is important, especially in regions with a high TB burden, like sub-Saharan Africa. Drug-induced immune thrombocytopenia is a significant ADR associated with the use of rifampicin, a key drug in TB treatment. Despite its importance, data on the safe reintroduction of rifamycins following rifampicin induced thrombocytopenia is limited. However, safe reintroduction of rifabutin in cases of rifampicin-related ADRs appears feasible and generally well tolerated. This case report aims to contribute to the growing body of evidence supporting the likely safe use of alternative rifamycins, such as rifabutin, in patients with rifampicin-induced thrombocytopenia.

Key words
drug rechallenge, rifampicin-induced thrombocytopenia, tuberculosis

AGNIESZKA JAROSIŃSKA

1.5 in a million: acquired hemophilia A in an 85-year-old man

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CASE DESCRIPTION Acquired hemophilia A (AHA) is a rare, autoimmune bleeding disorder caused by autoantibodies against factor VIII, leading to impaired coagulation. It typically presents with spontaneous or prolonged bleeding episodes in patients with no prior history of coagulation disorders. This condition is most common in elderly individuals and its incidence increases with age. The mortality rate for AHA ranges from 8% to 48%, with higher rates observed in patients with comorbidities, such as cardiovascular disease. Although AHA can be associated with autoimmune disorders, infections, or malignancies, in approximately half of the cases it remains idiopathic.

Comprehensive diagnostic approach is necessary to identify AHA, particularly in patients presenting with unexplained bleeding.

This case describes an 85-year-old man with a history of hypertension and nephrolithiasis, who presented with high blood pressure and massive subconjunctival hemorrhage in the right eye. The patient also exhibited skin lesions, including ulcers on the right shin and exudative lesions between the toes. Laboratory findings showed prolonged activated partial thromboplastin time and elevated creatinine and C-reactive protein levels. Despite the absence of prior bleeding history, clinical and laboratory findings indicated the presence of a coagulation inhibitor. Tests confirmed a high titer of factor VIII inhibitor in the Bethesda assay and low factor VIII activity, which led to the diagnosis of AHA.

The patient was initially treated with desmopressin and corticosteroids. Subsequently, activated prothrombin complex concentrates (FEIBA) were administered to control the bleeding episodes. Additional tests showed a history of hepatitis C infection, but the patient refused further oncological workup. Given the patient's condition and the presence of a factor VIII inhibitor, the diagnosis of AHA was confirmed. Prophylactic treatment with FEIBA was recommended for continued management.

The patient developed a subcutaneous hematoma after administering insulin (as a result of developing poststeroid hyperglycemia). Due to a lack of improvement, he was transferred to the hematology ward for further treatment.

This case highlights the importance of a multidisciplinary approach, involving hematologists, specialists, and laboratory staff, for timely diagnosis and management of AHA. Early intervention is crucial in preventing severe complications and improving patient outcomes.

In summary, AHA should be considered in the differential diagnosis of unexplained bleeding, especially in patients with no prior history of coagulation disorders. Prompt diagnosis and appropriate treatment are crucial to minimizing mortality, highlighting the need for collaborative care in management of this rare and potentially fatal condition.

Key words
acquired hemophilia A, hemorrhagic conjunctival chemosis, hepatitis C, subconjunctival hemorrhage

AHMAD RAIYAN

Chronic diarrhea from a different point of view

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CASE DESCRIPTION Abdominal pain and diarrhea are common in primary care clinics. Workup and investigations are based on clinical decisions individualized for each patient. We describe a case of a 25-year-old man who presented with a 1-year history of watery diarrhea and abdominal pain. The symptoms followed a *Shigella* infection acquired on a trip to Bolivia. The patient's physical examination was unremarkable and blood test results were within the normal range. The fecal BioFire GI panel and parasitic polymerase chain reaction test were negative; so was the fecal calprotectin level. Gastroscopy and colonoscopy showed no marked pathological changes. Based on this, the patient was diagnosed with postinfectious irritable bowel syndrome and was treated symptomatically. Despite the prescribed treatment, his symptoms worsened. Further evaluation by magnetic resonance enterography showed a minimal and nonspecific inflammation in the distal ileum. The patient underwent video capsule endoscopy, which showed a foreign body in the mid- to distal ileum region (most probably a worm). A diagnosis of taeniasis was made, and the patient was treated with a single dose of praziquantel (10 mg/kg; orally) with marked improvement. Taeniasis is a cosmopolitan

helminthic disease caused by the *Taenia* species, which includes *T. solium*, *T. saginata*, and *T. asiatica*. These parasites typically infect the small intestine. Fecal polymerase chain reaction for *T. solium* or *T. saginatum* is required for final diagnosis. Though the infection is endemic to the tropics, nowadays the diagnosis is rare in developed countries. Due to how rare the disease is in Israel, reaching the diagnosis in this case required detailed and advanced auxiliary testing.

Key words
diarrhea, endoscopy, taeniasis

ANA GABRIELA MARQUEZ PEREZ

Beyond the bone marrow: bilateral orbital plasmacytoma as an atypical presentation of multiple myeloma

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INTRODUCTION Extramedullary plasmacytomas (EPs) are rare manifestations of multiple myeloma (MM), affecting soft tissues in approximately 5% of cases. They present predominantly in the head and neck area, but bilateral orbital involvement is extremely rare (<1% of orbital tumors). We describe a case of an uncommon presentation of relapsed MM, associated with leukemia and central nervous system (CNS) involvement, and highlight the diagnostic and therapeutic challenges presented by this condition.

CASE DESCRIPTION A 47-year-old man with a history of plasma cell leukemia and prior treatment with autologous bone marrow transplantation presented with progressive diplopia and right exophthalmos. Clinical examination showed a painless right orbital mass with associated splenomegaly. Laboratory findings showed pancytopenia and circulating plasma cells. Magnetic resonance imaging identified bilateral orbital masses and multiple lytic bone lesions. Flow cytometry confirmed leukemic plasma cells, while bone marrow biopsy demonstrated 54% plasma cell infiltration. Due to severe thrombocytopenia, initial orbital biopsy was postponed, prompting systemic chemotherapy with the PACE (cisplatin, cyclophosphamide, etoposide, doxorubicin) regimen. Three weeks later, biopsy became possible due to significant regression of the orbital lesions. It confirmed EP with monoclonal λ light chain expression. Cerebrospinal fluid analysis showed 20% plasma cells, confirming CNS involvement, which led to the decision to incorporate intrathecal chemotherapy into the treatment regimen.

DISCUSSION EPs remain challenging to diagnose and manage, as they always signify an aggressive and disseminated form of MM. Although rare, orbital involvement should prompt consideration of systemic MM relapse, as it is associated with a poor prognosis. Given its aggressive nature, early systemic therapy is essential for disease control and should not be delayed while awaiting histological confirmation in the cases of confirmed MM.

CONCLUSIONS This case highlights the unusual presentation of EP with bilateral orbital involvement. However, regardless of their location, EPs always represent an aggressive and disseminated form of MM. Recognizing this is crucial to prevent delays in treatment as, although uncommon, all manifestations of MM beyond the bone marrow—extramedullary plasmacytomas, plasma cell leukemia, and CNS involvement—are associated with worse prognosis.

Key words
extramedullary plasmacytomas, orbital tumors, plasma cell leukemia, relapsed multiple myeloma

ANITA WACH

A rheumatologist's gambit in treating ankylosing spondylitis and DRESS syndrome

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CASE DESCRIPTION Drug reaction with eosinophilia and systemic symptoms (DRESS) syndrome is a severe hypersensitive reaction. It can cause multiorgan involvement and requires timely diagnosis and treatment. Ankylosing spondylitis (AS), a chronic inflammatory disorder affecting the axial skeleton, presents challenges in managing patients with coexisting conditions or adverse drug reactions. In this case, we describe a 40-year-old patient with diagnosed AS who developed DRESS syndrome following treatment with sulfasalazine—a known trigger. The patient exhibited a delayed onset of rash, fever, liver dysfunction, and pericardial effusion. Diagnosis was confirmed through the RegiSCAR scoring system and lymph node biopsy. Initial treatment with high-dose prednisone showed partial improvement, but a relapse necessitated additional immunomodulatory therapy, including tofacitinib, a Janus kinase inhibitor, which resulted in resolution of DRESS symptoms and improved AS control. The case emphasizes the importance of early recognition, multidisciplinary management, and the potential role of tofacitinib as an alternative to long-term corticosteroid therapy in patients with DRESS syndrome and autoimmune comorbidities. Further research is needed to validate the efficacy and safety of Janus kinase inhibitors in the treatment of refractory DRESS syndrome.

Key words
ankylosing spondylitis, drug reaction with eosinophilia and systemic symptoms syndrome, Janus kinase inhibitors, sulfasalazine, tofacitinib

ANUSHA HEGDE

The Great Masquerader strikes again! Neurosyphilis presenting as general paresis of the insane and stroke-like syndrome

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INTRODUCTION Due to its frequent atypical presentation with clinical phenotypes ranging from meningitis, through meningovascular syphilis with strokes and, in advanced stages, "general paralysis of the insane," to simple chancres, syphilis is often considered to be "the Great Imitator." We present a case of a young man with neurosyphilis presenting with a stroke-like symptoms and dementia.

CASE DESCRIPTION A 32-year-old unmarried IT professional with diagnosed rheumatic heart disease and ankylosing spondylitis presented with fever, slurring of speech, limb stiffness, emotional lability, and memory impairment. Examination showed punched-out ulcers on the tongue and genital warts. The patient's attenders did not report any promiscuous sexual relations. Neurological examination showed bilateral upper and lower limb hypertonia with exaggerated deep tendon reflexes in the ankle and patellar clonus. Loud pulmonic valve closure and early diastolic murmur on auscultation were observed. Neuroimaging was suggestive of right midbrain infarct. Due to the presence of cutaneous lesions, the patient was tested for sexually transmitted infections. The results were positive for hepatitis B and *Treponema pallidum* in the serum. The Venereal Disease Research Laboratory test of the cerebrospinal fluid was positive for *T. pallidum*.

A diagnosis of tertiary syphilis with general paresis of the insane and stroke caused by vasculitis was made. The patient was treated with ceftriaxone injections for 14 days, oral glucocorticoids, single antiplatelet therapy, and statins, as well as antipsychotics for cognitive impairment and tenofovir alafenamide for hepatitis B. He was discharged on the 15th day with significant neurological improvement.

DISCUSSION Neurosyphilis remains diagnostically challenging due to its diverse clinical manifestations, which often mimic psychiatric and neurovascular disorders. General paresis of the insane is a rare but severe form of late neurosyphilis, often misdiagnosed due to its insidious onset of neuropsychiatric symptoms, cognitive decline, and motor dysfunction. This case highlights the importance of considering tertiary syphilis in patients with unexplained neurological and psychiatric symptoms, especially in the presence of vascular complications. Syphilitic vasculitis can lead to ischemic strokes, further complicating the course of the disease. In our patient, the presence of rheumatic heart disease and spondyloarthropathy added to the complexity, underscoring the need for a multidisciplinary approach in managing such cases. Diagnosis requires serological testing, neuroimaging, and cerebrospinal fluid analysis, with the Venereal Disease Research Laboratory test being a key diagnostic marker. Early recognition and treatment with high-dose intravenous antibiotics, as administered in this case, are essential to prevent irreversible neurological damage.

CONCLUSIONS This case underscores the importance of early neurosyphilis diagnosis and initiation of antibiotic therapy. It also emphasizes the importance of evaluating infective or treatable causes, especially in young-onset or rapidly progressive dementia.

Key words
general paresis of the insane, neuropsychiatric manifestations, neurosyphilis, rheumatic heart disease, vasculitic infarct

AZUL LANZILLOTTA

Beyond the usual suspects: unraveling a case of IgG4-related disease

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CASE DESCRIPTION Immunoglobulin G4–related disease (IgG4-RD) is an immune-mediated, fibrosing disease that presents as mass-forming lesions. It can mimic various neoplastic, inflammatory, and infectious conditions. If not properly diagnosed and treated, it may lead to end-organ failure.

We present a case of a 66-year-old man with a 30-year history of submandibular lymphadenopathy, previously diagnosed as follicular hyperplasia and chronic sclerosing sialadenitis, a biliary stent placed due to cholestatic syndrome, pancreatic calcifications, severe aortic stenosis, and chronic kidney disease requiring dialysis due to obstructive uropathy. Additionally, the patient had undergone a remote thyroidectomy for an unknown cause.

Given the fibrosing involvement of multiple organs, a re-evaluation of previous salivary gland biopsy was performed, showing lymphoid hyperplasia with storiform sclerohyaline fibrosis and 28.6 IgG4+ plasma cells per high-power field, with an IgG4+ to IgG+ ratio of above 40%. Renal biopsy showed diffuse glomerulosclerosis, severe chronic tubulointerstitial nephropathy with a diffuse lymphoplasmacytic infiltrate, and 10 IgG4+ cells per high-power field.

Based on this, the patient was diagnosed with systemic IgG4-RD with glandular, pancreatobiliary, and renal involvement. He was initiated on glucocorticoids and rituximab, which led to significant clinical improvement, including better diabetes control, weight gain, regression of lymphadenopathy, and overall improved health status.

IgG4-RD predominantly affects men. In addition to pancreatic, salivary gland, and lymph node involvement, renal, aortic, retroperitoneal, and pulmonary involvement are frequently described. The initial presentation can be highly nonspecific or suggest more common diseases.

This case is of particular interest due to the severe multiorgan involvement, significant decline in the quality of life, and the absence of a prior diagnosis, partly attributable to the relatively recent recognition of IgG4-RD as a distinct clinical entity in the last 15 years.

CONCLUSIONS 1) IgG4-RD should be considered in the cases of multiorgan fibrotic disease with no clear etiology. 2) Diagnosis requires a combination of clinical, serological, and histopathological findings. 3) Timely initiation of glucocorticoids and rituximab can significantly improve outcomes. 4) Greater awareness and early recognition are essential to prevent irreversible organ damage.

Key words
chronic tubulointerstitial nephropathy, glucocorticoid therapy, lymphoplasmacytic infiltrate, multiorgan fibrosis, systemic IgG4-RD

BETUL YILMAZ

Aseptic abscess syndrome: a unique case of splenic involvement and systemic inflammation

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INTRODUCTION Aseptic abscess syndrome (AAS) is a rare inflammatory disorder characterized by sterile, encapsulated lesions filled with polymorphonuclear neutrophils, typically presenting with fever, abdominal pain, and weight loss. It is generally recognized as a diagnosis of exclusion when radiological evidence of abscesses lacks an infectious source, as antibiotics are ineffective while corticosteroids are often beneficial.

CASE DESCRIPTION We report a case of a 42-year-old woman who presented with prolonged fever, fatigue, weight loss, and elevated liver enzyme levels, alongside multiple hypodense splenic lesions. Despite comprehensive antibiotic therapy, her symptoms persisted and the levels of inflammatory, particularly C-reactive protein, remained elevated, prompting a shift in diagnosis toward AAS. Following the initiation of corticosteroid therapy, the patient demonstrated rapid clinical improvement. Her fever resolved and a C-reactive protein levels decreased significantly.

DISCUSSION This case highlights the diagnostic complexity and therapeutic considerations associated with AAS, including its potential overlap with other inflammatory conditions, such as inflammatory bowel disease and neutrophilic dermatoses. Given the frequent recurrence of AAS, long-term management may require maintenance therapies, potentially with anti–tumor necrosis factor agents or colchicine.

CONCLUSIONS This report underscores the importance of considering AAS in patients with sterile abscesses and persistent systemic inflammation, particularly when antibiotic resistance is observed.

Key words
aseptic abscess syndrome, infectious disease, splenic lesions, rheumatology

BRIGITTA SZOLGA

Post–gastric bypass hyperinsulinemic hypoglycemia

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INTRODUCTION Symptomatic hypoglycemia is a well-known complication of gastric bypass surgery. The underlying pathological features of post–gastric bypass hyperinsulinemic hypoglycemia may be similar to those of noninsulinoma pancreatogenous hypoglycemia syndrome. There remains a debate about whether it constitutes a distinct entity or is merely a specific form of noninsulinoma pancreatogenous hypoglycemia syndrome.

CASE DESCRIPTION We report a case of a 69-year-old man with a history of hypertension, ischemic cardiomyopathy, rheumatoid arthritis, osteoporosis, and hepatitis A infection.

At the age of 42, he underwent treatment to eradicate *Helicobacter pylori* infection caused by extensive gastric, duodenal, and jejunal ulcers. Following recurrent ulcer disease, the patient underwent gastric resection, truncal vagotomy, and ultimately, a complete gastrectomy with Roux-en-Y anastomosis as per the Billroth II procedure. Genetic analysis of the *MEN1* gene showed no pathogenic mutations. Subsequently, the patient developed intestinal adhesions that required a right-sided hemicolectomy and the creation of an ileo-sigmoidostomy. At the age of 49 years, 3 years after his Billroth II gastric resection, he experienced his first episodes of symptomatic postprandial hypoglycemia, accompanied by subtle neurological symptoms, such as speech disorder. By the age of 64, the patient had experienced spontaneous hypoglycemic episodes that necessitated hospitalization and further clinical investigations. Serum insulin and C-peptide levels were not suppressed, suggesting the possible presence of an insulinoma or other condition causing hyperinsulinemic hypoglycemia. Abdominal ultrasound identified a 5–6-mm hypoechoic area in the tail of the pancreas; however, subsequent examinations—including endoscopic ultrasound, abdominal and pelvic computed tomography scans, abdominal magnetic resonance imaging, somatostatin receptor scintigraphy, and fluorodopa (^{18}F) positron emission tomography computed tomography—did not show any morphological abnormalities in the pancreas. The selective arterial calcium stimulation test indicated that insulin secretion was stimulated in all 3 parts of the pancreas, with the most significant increase in insulin occurring from stimulation of the tail of the pancreas.

As part of the conservative treatment, oral diazoxide and acarbose were prescribed; however, hypoglycemic episodes did not resolve. Pancreatic tail resection was performed, based on the previous abdominal ultrasound and the functional test. Histological examination of the resected pancreas showed diffusely enlarged islets of Langerhans, where the islet cells showed a 100% positive reaction in an immunohistochemical test using an anti-insulin antibody. Histopathological morphology closely resembled that of noninsulinemic pancreatogenous hypoglycemia syndrome.

DISCUSSION The pathophysiological processes underlying post–gastric bypass hyperinsulinemic hypoglycemia remain unclear. A possible explanation is that food reaches the distal ileum prematurely, stimulating the secretion of glucagon-like peptide-1. This hormone promotes β-cell proliferation and increases insulin production in the pancreas.

CONCLUSIONS Insights gained from this case reinforce previous reports indicating that post–gastric bypass hyperinsulinemic hyperglycemia represents an acquired form of noninsulinoma pancreatogenous hypoglycemia syndrome. Distinguishing this clinical presentation from other types of hyperinsulinemic hypoglycemia and developing an effective treatment strategy remains a significant challenge.

Key words
hyperinsulinemic hypoglycemia, noninsulinoma pancreatogenous hypoglycemia syndrome, post–gastric bypass hyperinsulinemic hypoglycemia

CAMILA RODRIGUEZ

When the peritoneum surprises: peritoneal lymphomatosis in a patient recently diagnosed with HIV

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INTRODUCTION The term peritoneal lymphomatosis (PL) describes a rare involvement of the peritoneum as an extranodal manifestation of lymphoma. The only case reports in the literature are associated with highly aggressive lymphomas. PL can be easily confused with peritoneal carcinomatosis and is associated with poor prognosis. We present a case of a 37-year-old man recently diagnosed with HIV infection and PL secondary to Burkitt lymphoma.

CASE DESCRIPTION A 37-year-old man presented with night sweats, abdominal pain, and distension with intestinal rhythm disorders. Physical examination showed infraumbilical ascites and palpable tumors in the abdominal wall. Laboratory workup showed no significant alterations. The diagnosis of HIV was confirmed with CD4 levels of 157/mm^3 and a viral load of 100 000 copies/ml. Antiretroviral treatment and primary prophylaxis for *Pneumocystis jirovecii* were started. Serologies for hepatitis B virus, hepatitis C virus, toxoplasmosis and Chagas disease were negative. Carbohydrate antigen 19-9, carcinoembryonic antigen, and α-fetoprotein tumor markers were negative, while carbohydrate antigen was positive (1153 U/ml). Computed tomography of the abdomen and pelvis showed diffuse thickening of the greater omentum and mesenteric, retroperitoneal, and pelvic lymphadenopathy. Cytological study of ascitic fluid reported a presence of atypical lymphoid cells. Given the suspicion of lymphoproliferative syndrome, lymph node and peritoneal biopsy were performed, showing Burkitt lymphoma. The patient was started on R-CHOP (rituximab, cyclophosphamide, hydroxydaunorubicin, oncovin, prednisone) regimen, but it was not completed due to the development of septic shock and acute respiratory failure, caused by nosocomial pneumonia, leading to the patient's death.

DISCUSSION Peritoneal involvement secondary to lymphomas is extremely rare. Lymphomas usually do not affect the peritoneum, as it is made of fibrofatty tissue devoid of lymphoid cells. Reported cases are associated with high-grade non-Hodgkin lymphomas (mostly diffuse large B-cell lymphoma and Burkitt lymphoma in children). While these subtypes are most commonly associated with HIV infection, there is no increase in the incidence of this condition in seropositive patients in the literature. Clinical manifestations include abdominal pain and distension, fever, weight loss, palpable nodules in the abdominal wall, and mild-to-moderate ascites. On imaging, PL can easily be confused with peritoneal carcinomatosis and given the epidemiological context, especially in an immunosuppressed patient, tuberculous peritonitis should not be omitted as a differential diagnosis. The presence of an omental cake, with bulky masses and diffuse lymphadenopathy, could suggest lymphomatosis. The definitive diagnosis is confirmed by peritoneal biopsy. The discovery of this entity indicates poor prognosis, so early suspicion and initiation of chemotherapy are crucial.

CONCLUSIONS PL is a diagnosis that should be considered in patients with abdominal pain, ascites, and peritoneal thickening. Its main differential diagnosis is carcinomatosis and peritoneal tuberculosis. Its low frequency constitutes a diagnostic challenge.

Key words
Burkitt lymphoma, HIV, peritoneal lymphomatosis

NIKOLA COLOVIC

An unusual case of dyspnea in the sea of respiratory infections and acute chronic obstructive pulmonary disease exacerbations during the winter of 2025

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CASE DESCRIPTION Dyspnea is a subjective feeling of difficult breathing, most often related to respiratory or cardiovascular illness. However, dyspnea can also be the lungs' compensatory mechanism aiming to resolve metabolic acid-base disorders. In the case of our 67-year-old patient, dyspnea was a result of respiratory-compensated metabolic acidosis caused by urine retention, due to a penile urethra stricture. Based on his medical history, it would have been easy to explain dyspnea as a result of a simple acute exacerbation of chronic obstructive lung disease. However, point-of-care abdominal ultrasound results and correctly interpreted venous blood gas analysis quickly showed us the right direction.

Key words
dyspnea, metabolic acidosis, urinary tract obstruction

NOÉMI HAJDÚ

An octopus fisherman and endocrinology

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CASE DESCRIPTION A 50-year-old man with a history of hypertension and bronchial asthma was admitted to a cardiology ward due to cardiac decompensation, requiring temporary noninvasive ventilation. The tests confirmed non–ST-segment elevation myocardial infarction as the cause. Acute coronarogry angiography did not confirm coronary artery disease. During echocardiography, significantly reduced global left ventricular systolic function and mid-segment akinesia were observed. Cardiac magnetic resonance imaging showed a 6-cm tumor in the left adrenal region and was consistent with takotsubo cardiomyopathy. The patient was then referred to an endocrine ward. On admission, he reported recurrent episodes of hypertension (systolic blood pressure around 190 mm Hg) with intermittent agitation. Hormone test results showed elevated urinary catecholamines, which led to a diagnosis of pheochromocytoma. The patient underwent laparoscopic adrenalectomy, after which histopathological examination confirmed the diagnosis of pheochromocytoma. After the surgery, urinary catecholamine metabolites normalized and clinical symptoms resolved. Genetic testing confirmed an *SDHB* mutation. Additional imaging and hormonal studies of other localizations have not shown paraganglioma propagation, but genetic testing of the family confirmed the mutation in the patient's son (aged 25 years). Since then, the patient has been closely monitored and clinical manifestations of the disease have not yet been confirmed. Pheochromocytoma/paraganglioma is a rare hormone-producing tumor that can produce adrenaline, noradrenaline, and dopamine. These hormones can cause hypertension, even hypertensive crisis, and autonomic symptoms, which often lead to significant cardiovascular complications. The *SDHB* mutation is characteristic of familial paragangliomas, which can be associated with an aggressive clinical presentation and course, and, in some cases, lethality. Therefore, in the case of positivity, a family genetic testing is essential.

Key words
catecholamines, pheochromocytoma, *SDHB* mutation, takotsubo cardiomyopathy

NOEMI TARI

Food impaction and eosinophilic esophagitis

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INTRODUCTION Eosinophilic esophagitis is a localized, chronic, reversible inflammatory disease of the esophagus, which can manifest as dysphagia, food impaction, and chest pain. The number of newly diagnosed cases is approximately 7 per 100 000 population per year, with a prevalence of 113 per 100 000. It is more common in men, and mainly affects individuals younger than 40 years. Our paper aims to outline the diagnosis and therapy of eosinophilic esophagitis through an interesting and instructive case report, examine the factors that may play a role in recognizing the disease, and highlight some considerations that may help in making an early diagnosis.

CASE DESCRIPTION In 2023, we examined a 45-year-old man with no history of diseases affecting the internal organs. However, his gastroenterologic complaints had been recurrent since 2009. Several times, he had undergone urgent gastroscopy for frequent food impactions, but no clear cause was found. Present complaints started the evening before admission, after dinner. The patient experienced difficulties with swallowing, thoracic discomfort, and choking. He arrived to the emergency room the next morning with stable vital signs, but was unable to swallow. Electrocardiography showed no ischemic abnormalities. Arterial blood gas analysis showed a balanced metabolic state. We found a borderline eosinophil cell count (0.5 G/l; 5%). Otorhinolaryngology consultation identified no abnormalities up to the esophageal inlet. Contrast swallow X-ray showed partial obstruction in the upper third part of the esophagus. Urgent gastroscopy found a stricture, signs of food impaction, and features of eosinophilic esophagitis. The stuck bite was pressed into the stomach and multiple biopsy specimens were taken. During control swallow X-ray, the contrast material ran into the stomach with no evidence of perforation. Histopathological examination showed more than 15 eosinophilic granulocytes per high-power field between the superficial epithelial cells of the esophagus and confirmed the suspected diagnosis of eosinophilic esophagitis. The diagnosis is based on clinical signs, endoscopic abnormalities, and histopathological findings. Treatment of the disease involves proton pump inhibitors, oral steroid therapy, and empiric elimination diet. Severe strictures might also require endoscopic dilation. There is a new, additional therapeutic modality—a monoclonal antibody called dupilumab. The patient initially received proton pump inhibitor treatment, followed by oral steroid therapy (viscous budesonide) and dietary advice, and esophageal dilatation was planned. His complaints resolved, the esophagus lumen became passable, the endoscopic features improved significantly, and histological remission was confirmed. The patient is currently receiving budesonide maintenance treatment.

CONCLUSIONS The patients with eosinophilic esophagitis often do not seek medical care in time, and the diagnosis is usually further delayed by a general fear of endoscopic examinations and the lack of adequate histological specimens. Our team is collecting retrospective data, planning prospective research, and creating a registry of patients with eosinophilic esophagitis diagnosed in Hungary.

Key words
dysphagia, eosinophilic esophagitis, food impaction, gastroscopy, stricture

OKSANA FAIURA

Have you ever caught a chameleon?

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INTRODUCTION A differential diagnosis with connective tissue disorders might be needed due to a significant increase in the number of human parvovirus B19 (B19V) cases in adults, usually involving reticular rash and multiple joint pain.

CASE DESCRIPTION A 40-year-old woman complained of pain in the joints of hands, knees, fingers, and collarbone, morning stiffness, garland-shaped rash on the entire body, fever (up to 38 °C), palpitations, numbness in the arms, dyspnea, pain in the neck, and headache. The patient was diagnosed with systemic lupus erythematosus (SLE), but the symptoms did not improve with typical treatment. Objective examination of the osteoarticular system showed swelling and pain in the joints of hands, knees, fingers, and collarbone, decreased muscle tone, decreased hand strength, disfiguration and deformation of the small joints of the hands, decreased movements, and walking difficulty. Laboratory workup showed clinically significant abnormalities: estimated sedimentation rate, 15 mm/h; C-reactive protein (CRP), 8.6 mg/l (reference range [RR] <5 mg/l); antinuclear antibodies (ANA), immunoglobulin (IgG), 4.1 (index; RR <0.7). An autoimmune panel performed 3 days later was positive for class II double-stranded DNA (ds-DNA), class 1 liver cytosolic antigen type 1, SP100 antibody, gp210 antibody, and Ku antibody. The day after, IgG to ds-DNA was 76 IU/ml (RR <10 IU/ml). Polymerase chain reaction performed a few days later was negative for B19V. Immunoblot IgG was positive for VP1 protein, VLP protein, and VP2 protein; it was negative for NS1 protein. Immunoblot IgM was positive for VP1 protein, uncertain for VLP and NS1 protein, and negative for VP2 protein. The final diagnosis was SLE (acute course, active phase, activity stage II with immunological changes [ANA+, ds-DNA, LC1, SP100, gp210, Ku-positive] and injury of the skin ["butterfly" facial erythema in the past] and joints [arthralgia syndrome, functional joints insufficiency of second to third degree]), chronic tonsilitis, insomnia, tension headache, cervicalgia with myofascial syndrome, and parvovirus infection.

The patient's symptoms resolved within 1 month: ANA, 0.8 (RR ≤1.2); anti-dsDNA, 12.9 IU/ml (RR ≤46 IU/ml); alanine aminotransferase, 11 U/l (RR ≤33 U/l); aspartate aminotransferase, 12.4 U/l (RR ≤32 U/l); CRP, 0.6 mg/l. The patient was observed over the following year for possible symptoms of SLE, but none were found. ANA was negative (enzyme-linked immunosorbent assay), and absent (immunofluorescence test).

DISCUSSION Analyzing the patient's clinical symptoms, exposure history, and positive serologic test results showing B19V presence supported the diagnosis of acute parvovirus infection. However, the possibility of a first presentation of concomitant SLE could not be excluded because of other positive serologic test results (ANA, Ig to ds-DNA) and evidence of hypocomplementemia. The treatment of parvoviral infection is supportive. Rashes and arthralgia may be treated with anti-inflammatory drugs, as presented in our case.

CONCLUSIONS B19V infection can produce a transient autoantibody response and mimic the first presentation of a connective tissue disorder, especially SLE, thus underscoring the importance of obtaining an accurate history. To avoid the incorrect diagnosis of lupus, doctors should take these facts into account during the patient's examination and treatment.

Key words
differential diagnosis, erythema infectiosum, infection, parvovirus B19, systemic lupus erythematosus

ONDREJ SANTAVY

Of lumps and men

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INTRODUCTION Our case study describes unique manifestation of a rather frequent hereditary disease, which has not been described in Czech literature before.

CASE DESCRIPTION We describe a case of a 63-year-old woman with no prior medical history. During the summer of 2024, she started noticing formation of purple, painful nodules, which gradually transformed into large bullae. The patient also developed shortness of breath. During our clinical examination, 2 large bullae were found: one in the right axilla and the other on the outer labia. Anasarca was present. Laboratory tests showed elevated inflammatory markers (C-reactive protein level, 98 mg/l; leukocytosis with left shift). Total serum protein was 4.1 g/dl in the setting of severe hypalbuminemia. Computed tomography angiography of the pulmonary artery showed marked bronchiectasis and lung emphysema. Abdominal ultrasound yielded an image of hepatopathy. We admitted the patient and started decongestion therapy using loop diuretics and mineralocorticoid receptor antagonists. Gradually, we ruled out rheumatologic disorders, sexually transmitted diseases, and other infectious diseases. Finding of pneumopathy and hepatopathy led us to suspect α1-antitrypsin (A1AT) deficiency as the cause of the disease, and cutaneous lesions as its rare skin manifestation. We consulted our dermatology department and skin biopsy was performed, showing neutrophilic panniculitis. We started nonspecific therapy for panniculitis, treating the patient with dapsone. Her condition deteriorated severely due to septic shock resulting from nosocomial superinfection of open skin lesions. The patient was admitted to the intensive care unit and treated with combined therapy of intravenous antibiotics. Plasmapheresis was performed multiple times in order to substitute the lacking A1AT. After weeks of the diagnostic process, A1AT deficiency was genetically and histologically confirmed. Treatment with plasma-derived A1AT was started. Multiple plastic surgeries were performed, during which 8% of total the body surface was autotransplanted. After 3 months of hospitalization, the patient returned back to normal life and resumed her work as a teacher in a grammar school.

DISCUSSION A1AT deficiency is an autosomal-recessive hereditary disease. The A1AT protein is synthesized in the liver and released into circulation. It primarily protects the tissues from detrimental effects of neutrophil elastase. The liver and lungs are the most frequently affected organs. Chronic inflammation of the liver tissue leads to the development of liver cirrhosis. Loss of protective function in the lungs causes formation of emphysema and bronchiectasis, leading to the development of chronic obstructive pulmonary disease. Skin disease develops in less than 1% of patients. Formation of neutrophilic panniculitis is typical. There are no guidelines regarding the management of skin disease in A1AT-deficient patients. Treatment with immunosuppressants seems to be ineffective. First-line treatment is dapsone. In the acute setting, we use plasmapheresis in order to substitute A1AT. The most effective treatment is regular infusion with respreeza, an α1-protease inhibitor.

CONCLUSIONS The goal of our case study was to show the benefits of looking for the correct diagnosis. Thanks to the work of multiple specialists in various fields of medicine, our patient was able to return to normal life.

Key words
α1-antitrypsin deficiency, panniculitis

ROMAN DOHNAL

A complicated journey from the spine to the correct diagnosis

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INTRODUCTION We present a rare case of a patient with pathological findings in the lumbar spine found on magnetic resonance imaging (MRI), leading to an extensive diagnostic process. The case is notable for its multidisciplinary approach involving urology, radiology, and gastroenterology specialists, highlighting the complexity of diagnosing malignancy with initially nonspecific manifestations.
CASE DESCRIPTION A 70-year-old patient was referred to our department for further investigation of a pathological finding in the lumbar spine on MRI (heterogeneous bone marrow signal). Before admission, basic tumor markers were already taken (negative) and skeletal scintigraphy showed no pathological findings. Initial examination showed newly elevated creatinine levels (400 µmol/l), and ultrasound showed bilateral dilation of the renal pelvis and calyces. Native computed tomography (CT) of the kidneys did not show any intraluminal obstruction but confirmed dilation of the renal pelvis and ureters, leading to the placement of bilateral DJ stents by a urologist.

After conservative treatment was implemented and renal function improved, positron emission tomography (PET)/CT was performed, showing glucose hypermetabolism in the lumbar spine, stomach, thyroid gland, pancreas, seminal vesicles, and lymph nodes of the retroperitoneum and mediastinum. Subsequently, ultrasound examination of the thyroid gland with targeted biopsy was performed. Gastroscopy showed structurally altered mucosa with a 0–IIb lesion, 15–20 mm in diameter, at the transition between the antrum and body of the stomach. Samples were sent for histological examination.

The patient was discharged to outpatient care with planned readmission for endoscopic ultrasound with possible pancreatic biopsy. On the day of readmission, results of the stomach biopsy confirmed diffuse large B-cell lymphoma (DLBCL). Although the patient was asymptomatic, laboratory tests showed significantly elevated amylase and lipase levels, with differential diagnoses of acute pancreatitis or pancreatic infiltration by lymphoma. Conventional therapy for acute pancreatitis was initiated. Abdominal CT showed bile duct dilatation, for which endoscopic retrograde cholangiopancreatography with bile duct stenting was performed, but no signs of acute pancreatitis.

During hospitalization, the patient developed acute deep vein thrombosis in the lower extremity, managed with appropriate anticoagulant therapy. Once his condition stabilized, the patient was transferred to the Department of Haemato-Oncology of the University Hospital Olomouc for the initiation of immunochemotherapy.
CONCLUSIONS This case illustrates the complexity of the diagnostic process in a patient with initially suspected lumbar spine pathology, who was ultimately diagnosed with disseminated DLBCL. Of note, the results of skeletal oncologic examination and tumor marker tests were negative, even though PET/CT showed multiple metabolically active lesions. The key takeaway is the need for a comprehensive diagnostic approach in elderly patients with nonspecific radiological findings and the importance of interdisciplinary collaboration in the differential diagnosis of malignant diseases.

Key words
diffuse large B-cell lymphoma, lymphoma, positron emission tomography / computed tomography, spine, stomach

RUVEYDA SILAY

Rapid progression of amyloidosis with multiorgan involvement

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CASE DESCRIPTION A 57-year-old man was diagnosed with hypertension 10 years prior. He presented to the Istanbul Medical Faculty internal medicine polyclinic with complaints of weight loss (10 kg), nausea, vomiting, and abdominal swelling in the last year. He was subicteric; abdominal examination showed palpable liver 2 cm below the costal margin and open traube. On percussion, dullness from the umbilicus level was observed. The patient had bilateral 1+ pretibial edema. Laboratory test results were as follows: creatinine, 0.93 mg/dl; alkaline phosphatase (ALP), 574 I/U; γ-glutamyl transferase (GGT), 446 I/U; alanine aminotransferase (ALT), 31 I/U; aspartate aminotransferase (AST), 73 I/U; total bilirubin, 2.4 mg/dl; direct bilirubin, 2 mg/dl; albumin, 3 g/dl; low-density lipoprotein cholesterol (LDL-C), 210 mg/dl; gammaglobulinemia, 0.58 g/dl. Complete urinalysis showed 3+ proteinuria, and 3 g/day proteinuria was observed in spot urine sample. 24-hour urinalysis showed 5.9 g/day proteinuria and 3.9 g/day microalbuminuria. During follow-up, a rapid increase occurred in creatinine, ALP, GGT, and bilirubin levels (creatinine, 2.5 mg/dl; ALP, 850 U/l; GGT, 650 U/l; total bilirubin, 5 mg/dl; direct bilirubin, 4 mg/dl). Abdominal imaging showed that the liver craniocaudal dimension was 200 mm, with smooth contours, and a portal vein diameter of 17 mm. No splenomegaly was detected. Diffuse-free fluid was observed in all peritoneal compartments and no postrenal pathology was detected. The sampled fluid in paracentesis was clear and serous, serum ascites albumin gradient was 1.5 g/dl, leukocytes were 140×10^3/µl, and neutrophils were 10×10^3/µl. The patient's ascites was evaluated as portal type and poor in cells. No atypical cells were observed on cytological examination. The fibroscan test resulted in F 0–1 (5 kPa), S1 (CAP 250). Gastroscopy showed no esophageal varices. The patient had high cholestatic enzyme and direct bilirubin levels. Magnetic resonance cholangiopancreatography imaging showed no pathology in the bile ducts. Echocardiography performed to evaluate cardiac functions showed ejection fraction of 71%,interventricular septum thickness of 1.2 cm, and pulmonary artery pressure of 30 mm Hg. Valve functions were normal, and no pericardial effusion was detected. Protein electrophoresis showed serum monoclonal protein of 0.18 g/dl. The κ/λ ratio was 3.33, serum immunofixation electrophoresis was IgG κ, and urine immunofixation electrophoresis was κ. The differential diagnosis of nephrotic syndrome pointed toward primary amyloidosis and light chain disease. Renal biopsy showed κ monotype light chain–type amyloidosis and bone marrow biopsy showed a CD138(+) plasma cell level of 50%. Interstitial and patchy κ light chain monotype neoplastic plasma cell infiltration and amyloid-compatible eosinophilic material accumulation in the vessel wall were detected. The presence of ascites, cholestatic pattern enzyme, and bilirubin level elevation were evaluated as related to liver involvement in the infiltrative pattern of primary amyloidosis. The patient was treated with daratumumab, bortezomib, and dexamethasone with the diagnosis of primary amyloidosis and multiple myeloma. In our case, we aimed to present a multiple myeloma and AL amyloidosis association presenting with ascites and cholestatic pattern liver enzyme elevation.

Key words
hepatic failure, multiple myeloma, nephrotic syndrome, primary amylodiosis

SIMONA PAVUKOVA

From ibuprofen to external cardiostimulation

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CASE DESCRIPTION Nonsteroidal anti-inflammatory drugs (NSAIDs) have a wide range of beneficial effects on the body and play an indispensable role in the treatment of many diseases. However, it is also true that some of them, due to their dosage, are easily accessible over-the-counter drugs, which, when misused or interacting with other medications, can lead to life-threatening conditions. In addition to peptic ulcer disease with subsequent bleeding, one of these conditions is hyperkalemia, induced by aldosterone inhibition, leading to impaired ion excretion through renal tubules. Cardiomyocytes are particularly sensitive to changes and instability in serum potassium levels, which manifests as arrhythmias, usually bradycardia. Bradycardia is defined as a heart rate below the normal range (60 bpm), with variations depending on the literature. From a clinical perspective (especially in relation to the indication for pacemaker therapy), it is important to differentiate between reversible and irreversible causes of bradycardia. The most common reversible extrinsic cause (ie, without structural damage to the sinoatrial/atrioventricular node) is the effect of medications with negative chronotropic and dromotropic effects. In this case report, we present a case of a 78-year-old woman with an initially unclear history of chronic complaints and no specific information related to her current condition. She was brought in by the emergency medical team, intubated, sedated, and placed on external pacing with an initial heart rate of 27 bpm and a junctional rhythm on electrocardiography. The resolution of the case only occurred several hours after her arrival at the hospital.

Key words
bradycardia, nonsteroidal anti-inflammatory drugs, pacemaker therapy

SVEN L. VAN LAER

Steamy holiday: relaxed mind, but a nasty fever?

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INTRODUCTION Infective endocarditis, mostly caused by bacterial infection, is a well-known cause of fever of unknown origin. Despite advances in diagnosis and treatment, mortality remains high (15%–30%). The diagnosis depends on predisposing factors, clinical examination, blood cultures, and echocardiographic imaging. However, rarer causative species are often difficult to diagnose. We present a case of a 75-year-old man with a *Legionella pneumophila* (serogroup 2–7) endocarditis, probably caused by inhaling humidified air on holiday.

CASE DESCRIPTION A 75-year-old man presented to the emergency department with a 1-month history of intermittent fever, chills, night sweats, and significant weight loss. His medical history included type 2 diabetes, atrial fibrillation, myocardial infarction, a minor stroke, alcohol abuse, and a severe aortic valve stenosis with implantation of a Perceval bioprosthetic valve 2 years prior. The patient recounted a 3-month vacation throughout Thailand, where he suffered from a urinary tract infection and food poisoning. He mentioned a wall full of humidifiers next to his hotel room. His complaints started during his last week in Thailand. Physical examination was unremarkable. Blood tests showed a moderately elevated C-reactive protein level (42.7 mg/l) without leukocytosis. Blood cultures remained negative. Positron emission tomography/computed tomography showed no fluorodeoxyglucose-avid lesions. Transesophageal echocardiography showed normal function of the bioprosthetic aortic valve with pronounced sessile vegetations around the valvular edges. Culture-negative endocarditis was diagnosed; subsequently ceftriaxone and vancomycin were started. Extensive serological screening and polymerase chain reaction were positive for *L. pneumophila* (serogroup 1–7) immunoglobulin (Ig) M and IgG. Urinary antigen test was negative. Antibiotic treatment was switched to levofloxacin (500 mg twice a day). The patient underwent aortic root replacement with implantation of a Freestyle bioprosthetic valve. Microbiological examination of the resected bioprosthetic valve confirmed the diagnosis of *L. pneumophila* (serogroup 2–7) endocarditis. Levofloxacin was continued for 6 weeks and the patient made a rapid clinical recovery.

DISCUSSION Legionella is a rare cause of endocarditis, accounting for less than 1% of bacterial endocarditis cases. In particular, infection of the heart valves with nonserogroup 1 species is extremely rare. The occurrence of *Legionella* endocarditis is associated with prosthetic heart valves and alcohol abuse, both of which were present in our case. The prognosis is rather good, as the overall mortality is below 10%; however, surgical treatment is often necessary. Blood cultures remain negative and urinary antigen test is only reliable for *L. pneumophila* serogroup 1 infections. Diagnosis is based on vegetations or signs of valvular damage on echocardiography, and molecular techniques. There are no clear guidelines on how long antibiotic treatment should be continued. Due to positive cultures on the resected bioprosthetic valve, our patient was treated with levofloxacin for 6 weeks after the valve explant.

CONCLUSIONS This case describes a *L. pneumophila* (serogroup 2–7) endocarditis, which is a rare entity, often difficult to diagnose. We believe that the presence of risk factors (eg, prosthetic heart valve, alcohol abuse), especially in combination with triggering factors, such as exposure to potentially contaminated water (eg, hot tub, humidifiers), should raise awareness of possible *Legionella* endocarditis.

Key words
fever of unknown origin, infective endocarditis, *Legionella pneumophila*

TAMÁS ISTVÁN NAGY

End-organ damage reversed with complement inhibitor treatment

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CASE DESCRIPTION Thrombotic microangiopathy is a group of diseases with pathognomic triad of thrombocytopenia, microangiopathic hemolysis, and ischemic end-organ damage. Acute kidney failure requiring hemodialysis is common. One of its subtypes is atypical hemolytic uremic syndrome. Our 31-year-old patient was admitted to the infectiology ward with fever, respiratory complaints, and a positive SARS-CoV-2 antigen rapid test result. Laboratory workup showed acute kidney injury (creatinine, 616 µmol/l; estimated glomerular filtration rate, 9 ml/min/1.73 m^2), thrombocytopenia (42 G/l), and elevated lactate dehydrogenase (LDH) level (3227 U/l).

Abdominal ultrasonography showed bilateral kidney enlargement and hypervascularization. No signs of postrenal obstruction were observed. Respiratory symptoms showed rapid regression. There was no detectable pathogen other than SARS-CoV-2. Upper respiratory signs were followed by anemia, which raised a possibility of thrombotic microangiopathy. Diagnosis was confirmed by low levels of serum haptoglobin (0.19 g/l) and fragmentocytes visible on the blood smear. After hemodialysis was initiated, the patient was admitted to our nephrology ward for additional diagnostic tests and treatment due to suspected thrombotic microangiopathy. Previous microbiologic tests excluded the most common infections in the background of the syndrome (Shiga toxin–producing *Escherichia coli*, *Streptococcus pneumoniae*, influenza). Immunoserological tests were also negative. Complex complement diagnostic test was performed, showing overactivation of the terminal complement cascade with around 50% ADAMTS13 activity. This pointed toward the presence of atypical hemolytic uremic syndrome (aHUS) and complement inhibitor eculizumab therapy was started. As a result, renal function, anemia, and LDH levels showed rapid improvement and hemodialysis was stopped. Genetic analysis verified a *CD46* gene heterozygous mutation, confirming the diagnosis of aHUS. After a few months, eculizumab was switched to ravulizumab. The patient is in complete nephrological and hematological remission. In the case of coexisting thrombocytopenia, anemia, and target organ damage (acute kidney injury, encephalopathy, convulsion), LDH, serum bilirubin, and haptoglobin levels testing, as well as blood smear inspection, are necessary to identify fragmentocytes. Furthermore, quick exclusion of secondary etiologies and detailed complement diagnostics with the measurement of ADAMTS13 activity are crucial to establish the diagnosis. Overamplification of the alternative complement pathway can be identified in the background of aHUS. This syndrome is often associated with acute kidney injury, requiring dialysis, and milder cytopenias. It is important to timely begin adequate therapy, which may require transporting the patient to a center with apheresis accreditation and nephrologic, hematologic, and an intensive care unit background. Complement C5 inhibitors, eculizumab and ravulizumab, play an increasingly important role in the therapy of aHUS. If the treatment is started timely, the target organ damage is largely reversible.

Key words
atypical hemolytic uremic syndrome, eculizumab, thrombotic microangiopathy

TINA HERLJEVIĆ

Sometimes things are not as they might initially seem

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INTRODUCTION In clinical practice, patients commonly present with nonspecific symptoms that might obscure a more complex clinical picture. It is important to be careful when making a clinical judgement and take into consideration a broader spectrum of differential diagnoses, so that potentially more serious causes are not missed.
CASE DESCRIPTION A 67-year-old man presented to the emergency department due to vomiting of black-and-green content. Vomiting lasted for about 2 days, and usually occurred after a meal. On the day he presented, the patient had vomited about 20 times. Initial laboratory workup showed normocytic anemia, white blood cell count of 11.3×10^9/l, and a C-reactive protein level of 90.2 mg/l. Abdominal X-ray showed a large shade in the central portion. There were no signs of pneumoperitoneum or ileus. Distended stomach filled with content suspected to be blood was visible on ultrasound. There was a 20-g/l decrease in hemoglobin level, compared with the results from 2 weeks prior, and gastric ulcer bleeding was suspected. The patient's vital signs were stable; coagulation and hepatic enzyme levels were normal. Because of this, a gastroenterologist was consulted. A decision was made to observe the patient overnight and re-evaluate in the morning. A nasogastric tube was placed and about 2 liters of black-and-green gastric content were evacuated during the night. In the morning, no reduction in the hemoglobin level was observed, as compared with the night before. Since a large amount of gastric content was evacuated and it did not resemble hematemesis, the differential diagnosis was re-evaluated, and high gastrointestinal obstruction was suspected. A computed tomography (CT) scan of the abdomen showed a large retroperitoneal hematoma (146 mm × 54 mm × 133 mm [laterolateral × anteroposterior × craniocaudal]), causing obstruction by compressing duodenum and, consequently, gastric distension. A second CT scan, performed using a multiphase postcontrast protocol, showed no active bleeding in or around the hematoma. Due to the patient's Waldenström macroglobulinemia, a hematologist was consulted and an extensive evaluation was conducted (coagulation factors, platelet function, protein analysis), but no signs of coagulopathy were found. A successful surgical removal of the hematoma was performed and the patient was discharged a few days later.
DISCUSSION In this case, a decrease in hemoglobin levels and black color of the gastric content pointed to a differential diagnosis of hematemesis, when the correct diagnosis was high gastrointestinal obstruction. The CT scan made it clear that retroperitoneal hematoma was the reason for the hemoglobin level decrease.
CONCLUSIONS This case report is an example of a missed initial diagnosis, which inevitably will happen in the busy emergency department. The presentation of the patient often is not clear, as symptoms are either nonspecific or overlapping. Because of that, it is extremely important to re-evaluate the patient during the observation and take into consideration another differential diagnosis.

Key words
high gastrointestinal obstruction, retroperitoneal hematoma

VÍCTOR ROLANDO ROCHA

Thrombotic microangiopathy and atypical uremic syndrome secondary to dengue infection

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INTRODUCTION Dengue, the most globally prevalent arbovirus infection, necessitates a high degree of clinical suspicion. Severe forms of dengue can manifest with various complications, including hematological issues, such as thrombocytopenia, leukopenia, and hemoconcentration. Thrombotic microangiopathy (TMA) is a rare but severe complication that can be triggered by dengue, characterized by microangiopathic hemolytic anemia, schistocytes, increased lactate dehydrogenase and reticulocytes, low or absent haptoglobin, negative direct Coombs test, and thrombocytopenia. This case report describes a patient presenting with dengue-induced TMA.
CASE DESCRIPTION A 39-year-old man with a history of alcoholism and smoking cessation was diagnosed with classic dengue infection. Initially, he presented with fever, myalgias, asthenia, and adynamia, and on the fourth day, he developed mild thrombocytopenia. Subsequently, he was admitted to the emergency department with macroscopic hematuria, fever, myalgias, and retroocular headache. Laboratory findings showed macrocytic anemia, thrombocytopenia, renal dysfunction, hyperbilirubinemia, and elevated levels of transaminases. Within 12 hours of admission, the patient developed altered mental status, confusion, lethargy,

and generalized petechiae, along with a drop in hematocrit and hemoglobin, and worsening renal function. Peripheral blood smear showed 5% schistocytes and hemolysis parameters.

Investigations ruled out other causes of hemolysis, including vasculitis, rheumatological diseases, paroxysmal nocturnal hemoglobinuria, and thrombotic thrombocytopenic purpura (TTP). Serologies for hepatitis B and C, HIV, Chagas disease, and syphilis were negative, as were immunological studies. Direct antiglobulin test was negative, and computed tomography showed no abnormalities. Dengue infection was confirmed by the NS1 antigen test. Flow cytometry ruled out paroxysmal nocturnal hemoglobinuria. ADAMTS13 enzyme activity was normal, and anti-ADAMTS13 antibodies were negative, which ruled out TTP. The patient was ultimately diagnosed with atypical hemolytic uremic syndrome (aHUS).

The patient received emergency plasma exchange, methylprednisolone pulses, and, later, prednisone. He developed acute renal failure requiring hemodialysis, but subsequently recovered full renal function. By the sixth day, schistocytes disappeared from the peripheral blood smear. Anti–factor H antibodies were negative. The patient was discharged but was lost to follow-up; a 4-month checkup showed normal blood counts, renal function, and transaminases, but elevated indirect bilirubin levels.

DISCUSSION Thrombotic microangiopathies, including TTP and aHUS, are rare entities with high morbidity and mortality. Prompt diagnosis and treatment, primarily with plasma exchange and corticosteroids (for suspected TTP), are essential. aHUS is a rare disease, and genetic mutations and anti–factor H antibodies should be tested to offer targeted treatment with eculizumab.

CONCLUSIONS Although arbovirus-associated TMAs are uncommon, it is essential to consider this complication, particularly during outbreaks, such as the 2024 dengue surge in Buenos Aires. Dengue can trigger TMA, manifesting as either TTP or aHUS. This case highlights the importance of recognizing and promptly managing these complications to improve patient outcomes.

Key words
dengue, hemolytic uremic syndrome, thrombotic microangiopathies

WIEM HELALI

Scurvy: age-old disease, new understandings

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CASE DESCRIPTION Scurvy, a disease caused by vitamin C deficiency, is considered rare, but remains relevant in certain at-risk populations, including the elderly, those with malnutrition, and individuals undergoing bariatric surgery. We report a case of a 75-year-old woman with primary Sjögren syndrome, who presented with atypical symptoms, including bruising, hyperpigmentation, autoimmune hemolytic anemia, and other systemic manifestations. Despite initial treatment with corticosteroids, her condition persisted and further investigation pointed at scurvy caused by severely low vitamin C levels. Vitamin C supplementation led to a rapid improvement in the patient's skin lesions and anemia. This case highlights the importance of considering scurvy in elderly patients with multisystem symptoms and emphasizes the need for early diagnosis and treatment to prevent complications. Scurvy, although rare, should remain in the differential diagnosis when faced with unusual clinical presentations, particularly in populations at risk for malnutrition.

Key words
autoimmune anemia, elderly, sjogren syndrome, scurvy, vitamin C deficiency

ZHI TIAN CHEN

Acute kidney injury: a challenging case during on-calls

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INTRODUCTION Acute kidney injury is a common medical condition in our daily practice. However, sometimes the diagnosis can be challenging, especially during on-call periods. We present a challenging case of acute kidney injury in an elderly man.

CASE DESCRIPTION A 79-year-old man was admitted to our hospital following a blocked catheter. A week prior to this event, he experienced numerous episodes of diarrhea after eating street-food during his trip to Norfolk. He was subsequently admitted to our ward, following his deranged renal profile.

The patient had a past medical history of hypertension, type 2 diabetes, and known prostate carcinoma, because of which he was placed on long-term catheter. He was medicated with indapamide, lisinopril, pravastatin, alogliptin, and leuprorelin injection.

The initial differential aimed to rule out an obstructive cause of acute kidney injury. Numerous bladder scans and formal abdomen ultrasound showed no evidence of obstruction. In the context of his diarrhea and high inflammatory markers prior to admission, we believed that the culprit was most likely a prerenal cause of acute kidney injury; hydration therapy was continued.

On the third day of admission, the patient's renal profile and metabolic acidosis continued to deteriorate at an alarming rate, despite aggressive hydration. Newly developed anemia, thrombocytopenia, and mild elevation of bilirubin levels were observed. This prompted us to think of a rare cause, such as hemolytic uremic syndrome or thrombotic thrombocytopenic purpura. Physical examination showed distended abdomen and hypotension, but otherwise no focal neurological deficit or obvious rashes.

Urgent full blood picture, hemolytic profile, and autoimmune screen were performed and came back unremarkable. Urgent computed tomography of the kidney, ureter, and bladder was performed in view of the diagnostic dilemma and showed severe hydronephrosis with bilateral moderate hydroureter. A urologist was urgently consulted, and a Tiemann Tip silicone catheter was inserted.

Afterwards, the patient's renal profile and blood parameters drastically improved. Direct antiglobulin test was positive and other renal autoimmune profiles were unremarkable. Stool sample showed no *Echerichia coli* or Shiga toxin. The patient was discharged on medical advice after 3 days.

DISCUSSION This case serves as a reminder that even seemingly routine medical conditions can evolve unpredictably. While hydronephrosis is often identified on ultrasound, its sensitivity may decrease in certain clinical scenarios, such as operator dependency or suboptimal visualization. Therefore, in the cases of diagnostic uncertainty or clinical deterioration, a computed tomography scan should be promptly considered in order to rule out obstruction.

Furthermore, the development of anemia, thrombocytopenia, and biochemical abnormalities warrants consideration of rare but critical conditions, like hemolytic uremic syndrome or thrombotic thrombocytopenic purpura, as these require distinct and timely interventions.

Key words
acute kidney injury, hemolytic uremic syndrome, hydronephrosis

ZUZANA MIERTOVÁ

Semaglutide in the treatment of obesity in a nondiabetic patient after kidney transplantation

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INTRODUCTION Glucagon-like petide-1 receptor agonists, primarily semaglutide, have been identified as one of the most promising medical interventions in treating obesity. In addition, the recently published FLOW study proved the nephroprotective effect of semaglutide in patients with chronic kidney disease and type 2 diabetes mellitus. A specific group is represented by patients after kidney transplant. Studies published so far with a smaller sample of patients (<50) with diabetes mellitus indicate that the use of semaglutide is safe, even in the mentioned group of patients, and at the same time leads to a significant reduction in weight. However, there are no studies investigating the use of semaglutide in post–kidney transplant patients without diabetes.

CASE DESCRIPTION We present a case of a 21-year-old woman who in 2021 underwent primary kidney transplant from a postmortem donor, due to end stage kidney disease caused by chronic tubulointerstitial nephritis. She was hospitalized at the transplantation and nephrology ward for suspected acute antibody-mediated rejection. The patient suffered from secondary arterial hypertension and secondary hyperlipoproteinemia, but not diabetes mellitus or prediabetes. After initial obesity examination, the patient was diagnosed with central obesity class III (according to the World Health Organization classification) of combined etiology, mostly due to exogenous factors interacting with immunosuppressive treatment. Our patient was instructed on a complex lifestyle adjustment; she was recommended a reduced-calorie diet (500 kcal/day deficit relative to estimated energy expenditure) and increased physical activity (at least 150 min of aerobic activity/week and 2–3 strength trainings per week). She was administered semaglutide at an initial dose of 0.25 mg subcutaneously per week with monthly check-ups. During the 6-month follow-up, treatment with semaglutide was uptitrated to 0.5 mg subcutaneously per. week. The treatment led to a weight loss of 19.6 kg, which represents 14.67% of the starting weight, and was well tolerated by the patient.

DISCUSSION Weight gain is a major issue after kidney transplant and the development of post-transplant diabetes or metabolic syndrome are poor prognostic factors after renal grafts. In younger patients, possible need for a secondary kidney transplant must be taken into account, and obesity may present a contraindication for transplant. Therefore, the treatment of the patient after kidney transplant should include the prevention and early treatment of the mentioned comorbidities. Our case reports the first use of semaglutide in the complex management of obesity in a patient after kidney transplant without diabetes. To the date, no such case report has been published worldwide.

CONCLUSIONS Based on currently available studies with limited sample size, the use of semaglutide seems to be safe in post–kidney transplant patients, but studies and case reports in patients without diabetes are lacking. The treatment of obesity in such patients should be individualized, taking into account comorbidities and the risk/benefit ratio.

Key words
cardio-renal-metabolic syndrome, obesity, semaglutide

VIRTUAL POSTER PRESENTATIONS

BEST VIRTUAL POSTER RECOGNITION: OUMAYA BEN HAMDA

When clots meet cells: a rare case of acute lower limb ischemia pointing at acute promyelocytic leukemia: the unseen link

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INTRODUCTION Acute lower limb ischemia is a life-threatening emergency with various potential causes. One rare etiology is acute promyelocytic leukemia (APL). In this case, we present a patient in whom acute lower limb ischemia was the first manifestation of APL, emphasizing the importance of early recognition and prompt management.

CASE DESCRIPTION A 24-year-old patient with no significant medical history presented to our department with acute limb ischemia, characterized by pain, pallor, and absence of pedal and posterior tibial pulses in the left lower limb. On physical examination, the patient was afebrile and hemodynamically stable with normal electrocardiogram. No signs of purpura or livedo were observed. Biological tests showed no thrombocytopenia, hemostasis disorders or thrombophilia, and peripheral blood smear was normal. There was no indication of inflammatory biological syndrome. Autoimmune screening, including tests for antinuclear antibodies, antineutrophil cytoplasmic antibodies, cryoglobulins, and antiphospholipid antibodies, was negative. Transthoracic echocardiography showed no evidence of vegetations, valvular disease or heart failure. Doppler ultrasound and computed tomography angiography of the left lower limb identified a staged occlusion of the left common and external iliac arteries, along with an infrapopliteal artery blockage. The patient was initially diagnosed with thromboangiitis obliterans. Despite treatment with curative anticoagulants, platelet aggregation inhibitors, statins, and courses of ilosprost, the patient underwent left leg amputation due to progressive ischemia, followed by recurrent arterial thromboses, multiple amputations, and visceral infarctions. During follow-up, the patient developed thrombocytopenia (16 000/µl), anemia (hemoglobin, 9.4 g/dl), and leukocytosis (19 700/µl) with a predominance of neutrophils. Subsequent blood smear showed blasts and bone marrow biopsy confirmed the diagnosis of acute promyelocytic leukemia. Treatment with all-trans retinoic acid was promptly initiated.

DISCUSSION Thrombotic manifestations in APL occur in approximately 2%–15% of cases. The mechanisms underlying thrombosis in APL are multifactorial, including disseminated intravascular coagulation, leukostasis, and high procoagulant potential of leukemic promyelocytic cells. Thromboses primarily affect deep veins, coronary arteries, and cerebral arteries. Arterial involvement of the limbs is rare, with only 6 reported cases in the literature, 4 of which occurred before treatment initiation.

CONCLUSIONS This case is unique for the unusual location of the thromboses at the disease's onset, as well as the absence of hyperleukocytosis and blasts in the initial laboratory tests, which underscores the importance of conducting an exhaustive etiological investigation of distal ischemia, even when classic biological signs of leukemia are absent.

Key words
acute limb ischemia, acute promyelocytic leukemia, blast, thrombosis

BEST VIRTUAL POSTER RECOGNITION: SARA ALKHEMEIRI

Unrecognized transfusion-transmissible malaria in a nonendemic setting

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INTRODUCTION In 2007, the World Health Organization declared the United Arab Emirates free of local malaria transmission. Although blood donors who visited or lived in malaria-endemic countries are deferred or screened, malaria remains a concern as a transfusion-transmitted infection in nonendemic settings due to the exponential rise in international travel and migration.

CASE DESCRIPTION A woman in her 30s with transfusion-dependent thalassemia major presented with fever and chills lasting for 7 days. She had no history of foreign travel. She had persistent high-grade fever with no localizing symptoms or signs. Extensive diagnostic workup was unremarkable. On the twelfth day of hospitalization, scanty ring stages of *Plasmodium falciparum* were incidentally detected during routine peripheral blood film examination, triggered by profound anemia. The patient recovered after receiving antimalarial treatment. A look-back exercise was carried out to trace her previous blood donors. High-throughput molecular malaria screening was subsequently introduced by the blood bank service to augment the pre-existing repertoire of antigen testing and blood smear examination.

CONCLUSIONS Given the potentially disastrous consequences of unrecognized transfusion-transmitted malaria in nonendemic settings, additional diagnostic procedures should be considered to enhance the safety of blood transfusion and strike a reasonable cost-benefit balance.

Key words
blood donors, malaria, transfusion-transmissible infections

BEST VIRTUAL POSTER RECOGNITION: SEBASTIÁN CHÁVEZ

Hypercalcemia, a diagnostic puzzle: the role and potential biases of AI in a challenging case

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CASE DESCRIPTION Hypercalcemia is an uncommon but significant metabolic disturbance in patients with chronic liver disease, which often poses a diagnostic challenge. This case report describes a 70-year-old woman with a history of cirrhosis caused by overlap syndrome between autoimmune hepatitis and primary biliary cholangitis, who developed persistent hypercalcemia despite appropriate medical management. The case was further complicated by the complexity of differential diagnosis and the use of artificial intelligence (AI)-based large language models for clinical reasoning, which yielded divergent responses depending on the language of the query.

On admission, the patient exhibited progressive cognitive impairment, anorexia, and gastrointestinal symptoms. Extensive laboratory evaluations showed persistent hypercalcemia with low parathyroid hormone and normal vitamin D levels. Comprehensive hematological workup was negative for multiple myeloma or other hematologic malignancies. Imaging studies, including abdominal Doppler ultrasound and contrast-enhanced computed tomography (CT) scans of the abdomen and pelvis, showed cirrhotic liver changes, hepatofugal portal flow, and splenomegaly, with no evidence of

malignancy. Contrast-enhanced CT showed a small left hepatic nodule, slightly increased in size, compared with prior imaging. It appeared isodense to the surrounding parenchyma in contrast sequences and was not visible on Doppler ultrasound. Given the persisting diagnostic uncertainty, further assessment with positron emission tomography/CT was pursued, showing no hypermetabolic lesions suggestive of malignancy or granulomatous disease.

Faced with a complex clinical scenario with no clear etiology, an AI-based diagnostic aid was utilized, based on a recently published article in the *Journal of the American Medical Association* that employed a structured prompt for ChatGPT. This prompt employed a structured 3-step analysis to enhance diagnostic accuracy. In the first step, 3 possible diagnoses were selected, and supporting and opposing arguments were presented for each. The second step involved determining the final diagnosis, while the third step suggested next clinical steps. The same ChatGPT model was used to translate the published prompt into Spanish and the clinical case from Spanish to English. Both translations were reviewed to ensure coherence. ChatGPT's responses in English and Spanish were analyzed to identify discrepancies in diagnostic outputs. Notably, AI-generated conclusions varied by language: the Spanish query suggested hepatocellular carcinoma with low fluorodeoxyglucose uptake, while the English query proposed liver disease–related hypercalcemia. Importantly, the AI-generated suggestions differed considerably, raising concerns about the potential clinical impact, if taken literally without further expert evaluation. This discrepancy highlights potential biases in AI-assisted diagnostics, particularly in non-English medical contexts.

The patient was managed by endocrinology and hepatology teams. Magnetic resonance imaging of the liver was performed, which ruled out hepatocellular carcinoma and the condition was ultimately interpreted as hypercalcemia secondary to advanced cirrhosis.

These findings emphasize the need for further research into AI's role in clinical decision-making across different linguistic and cultural contexts. To our knowledge, this is one of the first reports highlighting language biases in ChatGPT when applied to real clinical cases. The discrepancies observed in AI-generated responses based on language input underscore the necessity for further evaluation. Furthermore, hypercalcemia remains a complex metabolic disorder with a broad differential, warranting careful clinical assessment to avoid misdiagnosis and ensure appropriate management

Key words
artificial intelligence, cirrhosis, hepatocellular carcinoma, hypercalcemia, large language models

AIMAL KHAN AFRIDI

Beyond the gut: enteric fever leading to acute myocarditis

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CASE DESCRIPTION Enteric fever continues to be a serious public health risk, especially in areas with poor sanitation and hygiene standards. Typically presenting with fever, abdominal pain, and gastrointestinal symptoms, enteric fever can also affect other systems including cardiovascular system. Myocarditis, an inflammation of the heart muscle, is a rare but severe complication of enteric fever, that can lead to heart failure and myocardial dysfunction. In this report, we present a case of a 20-year-old man diagnosed with myocarditis, following an episode of enteric fever, and discuss the clinical course, diagnostic challenges, therapeutic interventions, and outcome. Myocarditis associated with enteric fever is rare and often presents with nonspecific symptoms. In this case, the diagnosis was supported by clinical presentation, lab results, and echocardiographic findings. We aim to contribute to the existing literature, shedding light on this rare but critical clinical entity, and emphasizing the importance of early recognition and appropriate intervention in improving patient outcomes.

Key words
cardiomyopathy, echocardiography, enteric fever, myocarditis, NSTEMI

AKASH PARUTHI

Extensive metameric involvement in a case of Cobb syndrome presenting as compressive myelopathy

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INTRODUCTION Cobb syndrome, or cutaneomeningospinal angiomatosis, is a rare neurocutaneous disorder characterized by vascular malformations affecting the skin, spinal cord, and vertebrae within the same metameric segment. Most reported cases involve localized spinal lesions with early-onset neurological symptoms. However, extensive multisegmental involvement and late-onset presentation are exceedingly rare. Early recognition is crucial to prevent irreversible deficits. This case highlights an unusual presentation of Cobb syndrome with diffuse vertebral hemangiomas and epidural arteriovenous malformation, leading to progressive compressive myelopathy.

CASE DESCRIPTION A 28-year-old man presented with insidious-onset lower back pain, radiating to the left lower limb, progressing over 2.5 months to complete paraplegia. He developed bowel and bladder dysfunction. Examination showed widespread port-wine nevi and hemangiomas across multiple dermatomes. Neurological evaluation showed spastic paraplegia, sensory deficits below the T6 dermatome, and autonomic dysfunction. Spinal magnetic resonance imaging showed diffuse vertebral hemangiomas involving all vertebrae and compressive lesions from D3–D7 and D10–D12, leading to myelopathy. Biopsy confirmed cavernous hemangioma. Given the progressive neurological decline, the patient underwent D3–D7 laminectomy and decompression, leading to significant neurological improvement.

DISCUSSION Cobb syndrome results from aberrant vasculogenesis, affecting mesodermal and neural crest-derived structures. Spinal arteriovenous malformations, vertebral hemangiomas, and cutaneous vascular anomalies within the same metamere suggest an early embryological vascular defect. Unlike typical cases, presenting in childhood with localized involvement, this patient exhibited widespread vertebral hemangiomas and a late-onset neurological decline, making it an extraordinary presentation. Diagnosis relies on magnetic resonance imaging and digital subtraction angiography to define spinal and soft tissue involvement. Treatment depends on neurological severity and includes embolization, decompression or radiation therapy. In this case, surgical decompression led to significant recovery, underscoring its role in preventing permanent disability.

CONCLUSIONS This case highlights the importance of recognizing Cobb syndrome as a rare but significant cause of compressive myelopathy. Extensive multisegmental vascular involvement is unusual, and late-onset neurological deterioration further adds to the rarity of this presentation. Early diagnosis and timely intervention are crucial in preventing permanent deficits. This case contributes to the expanding literature on Cobb syndrome, highlighting the need for a multidisciplinary approach in managing complex spinal vascular disorders.

Key words
Cobb syndrome, compressive myelopathy, cutaneomeningospinal angiomatosis, metameric vascular malformation, spinal arteriovenous malformation

ALEKSANDRA PILŚNIAK

When first impressions lead in a wrong direction: a spectacular presentation of facial erysipelas

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CASE DESCRIPTION Our study presents a case of a 35-year-old woman who was admitted to a department of internal medicine because of massive edema of the face and neck with elevated inflammatory markers. Before admission, the patient was treated for herpes zoster and angioedema, but the treatment seemed to exacerbate the patient's symptoms. Further investigation showed positive blood and conjunctival swab cultures, identifying *Streptococcus pyogenes*. The patient was treated with targeted antibiotic therapy. Clinical and laboratory improvement was observed. The microbiological findings, clinical image, and positive response to the chosen treatment method confirmed, beyond a shadow of a doubt, that the patient presented a spectacular case of facial erysipelas.

Key words
blood culture, facial erysipelas, sepsis, *Streptococcus pyogenes*

ALOJZIJE LACKOVIĆ

Portal vein thrombosis with cavernous transformation in a patient with secondary myelofibrosis: a complex case of portal hypertension and hematological complications

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INTRODUCTION Portal vein thrombosis (PVT) with cavernous transformation is a rare and potentially life-threatening condition that can develop in the context of hematologic disorders, particularly myeloproliferative diseases, such as essential thrombocythemia and secondary myelofibrosis. These conditions predispose patients to thrombotic events, leading to complications, including portal hypertension, splenomegaly, and variceal bleeding.

CASE DESCRIPTION This case report highlights the challenges of managing a 41-year-old man with a history of PVT with cavernous transformation, secondary myelofibrosis, and allogeneic hematopoietic stem cell transplantation. The patient was diagnosed with PVT with cavernous transformation in 2018, during a routine check-up. The underlying etiology, secondary myelofibrosis (post-essential thrombocytopenia, JAK2-positive), was confirmed after further diagnostic workup. The patient underwent allogeneic hematopoietic stem cell transplantation, achieving 100% chimerism and an uneventful post-transplant course. He was readmitted to the emergency department due to hematemesis several hours after experiencing nausea, sweating, and dizziness. On examination, he was hemodynamically stable with mild pancytopenia and laboratory tests showed anemia, thrombocytopenia, leukopenia, and mild cholestatic liver injury. Upper gastrointestinal endoscopy showed actively bleeding esophageal varices, which were controlled by placing 6 "gummy band" ligatures. Ultrasound imaging showed enlarged spleen and known PVT with cavernous transformation. This transformation compressed the common bile duct, leading to mild intrahepatic bile duct dilatation. The diagnosis of obstructive jaundice was confirmed, prompting further imaging with magnetic resonance cholangiopancreatography, followed by endoscopic retrograde cholangiopancreatography, which resulted in the placement of a metal stent. During the latter procedure, significant hemorrhage from the common bile duct occurred, likely due to the patient's pronounced thrombocytopenia, but the bleeding was successfully controlled after stent placement. Given concerns of potential relapse of the underlying hematological condition due to pancytopenia and hypersplenism, bone marrow biopsy was performed. It confirmed remission of the hematological disorder, with 100% chimerism and preserved marrow cellularity. Throughout the hospitalization, the patient required transfusion support due to melena. Repeated gastroscopies showed no further bleeding, except for variceal bleeding. Considering the complications of portal hypertension, hypersplenism, and variceal bleeding, the patient was referred for potential liver transplantation or shunt placement.

DISCUSSION This case underscores the complexity of managing PVT with cavernous transformation in the context of secondary myelofibrosis. Despite preserved liver function, the presence of portal hypertension, hypersplenism, and variceal bleeding posed significant management challenges. The patient's pancytopenia, likely exacerbated by hypersplenism, complicated the clinical picture. Liver transplant or shunt placement may be required for severe portal hypertension, and careful consideration of the risks and benefits of such interventions is essential for optimal patient outcomes.

CONCLUSIONS This case emphasizes the importance of a multidisciplinary approach in managing patients with these complex hematologic and vascular conditions.

Key words
cavernoma, myelofibrosis, portal hypertension

ALON PORAT

An unusual late presentation of systemic Bacille Calmette–Guérin infection

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INTRODUCTION Bacille Calmette–Guérin (BCG) washes are a commonly used adjunctive therapy for the treatment of nonmuscle invasive bladder cancer. Infectious complications of intravesical BCG washes are relatively rare with a wide clinical presentation, early or delayed, local to the genitourinary tract or systemic with multiple organ involvement.

CASE DESCRIPTION This is a case of a 67-year-old man with a history of BCG washes performed in 2019 due to transitional cell carcinoma. In 2022, he was hospitalized for a mycotic aneurysm that was surgically repaired. In 2023, he was readmitted due to ongoing complaints of fever, weight loss, night sweats, and dyspnea. Positron emission tomography/computed tomography showed a strong accumulation of fluorodeoxyglucose in the lungs in "crazy paving" pattern, with no accumulation in the endovascular graft. Laboratory blood test results showed elevated C-reactive protein level and elevated creatinine levels of up to 2.5 mg/dl. Extensive serologic tests for infectious and inflammatory diseases did not yield any positive results. Lung biopsy demonstrated interstitial granulomatous inflammation without necrosis, alongside elevated angiotensin-converting enzyme levels. Because of a presumed diagnosis of sarcoidosis, treatment with glucocorticoids was started. The treatment resulted in resolution of the fever and night sweats, but respiratory symptoms and laboratory tests did not improve. The insufficient improvement under treatment lead to an additional polymerase chain reaction examination of lung tissue, showing *Mycobacterium tuberculosis* complex. Treatment with glucocorticoids was stopped and a regimen of isoniazid, rifampin and ethambutol was started.

DISCUSSION BCG infection is rare and thus difficult to study. Most of our current knowledge is derived from retrospective studies and clinical experience. Systemic infection accounts for approximately 25% of cases and usually presents within 3 months of treatment, making a delayed systemic infection occurring years after treatment a rare presentation and a diagnostic challenge.

This case demonstrates a rare presentation of delayed BCG infection occurring years after installation, which poses a major diagnostic challenge with a long diagnosis time.

The elusive and insidious nature of systemic BCG infections and the rising number of patients treated with BCG washes support the need for clinical suspicion among patients with a history of transitional cell carcinoma, who underwent treatments with BCG, that present with systemic complaint alongside the need for further studies and case series regarding infectious complications of BCG.

CONCLUSIONS High clinical suspicion in patients with a history of transitional cell carcinoma who underwent treatments with BCG, presenting with unexplained symptoms involving multiple systems, even years after treatment, should raise a suspicion of BCG infection. It might contribute to shorten diagnosis time and improve outcomes.

Key words
Bacille Calmette–Guérin, infectious complications, mycotic aneurysm

ASHRIT CHOHAN

Smith-Kingsmore syndrome: an unusual complication

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CASE DESCRIPTION Smith–Kingsmore syndrome (SKS) is an extremely rare genetic condition caused by gain-of-function mutations in the *MTOR* gene. Known for its neurodevelopmental impacts, including global developmental delay, intellectual disability, hypotonia, and epilepsy, SKS also has less understood systemic effects. One area of uncertainty are its potential gastrointestinal manifestations. While gastrointestinal symptoms are not a well-documented feature of SKS, this case explores the intersection between the neurodevelopmental abnormalities of SKS and recurrent sigmoid volvulus.

We report a case of an 18-year-old man diagnosed with SKS, who experienced recurrent sigmoid volvulus over a 3-month period, necessitating repeated endoscopic decompression and, eventually, surgical intervention. The patient underwent an elective robotic-assisted high anterior resection with stoma formation after conservative measures failed to prevent recurrence. The case highlights the challenges in recognizing and managing gastrointestinal complications in syndromic patients and raises questions about the potential role of hypotonia and connective tissue abnormalities associated with *MTOR* mutations in predisposing patients to colonic volvulus.

This report aims to explore the plausible link between SKS and sigmoid volvulus, with a focus on the implications for clinical management and future research. It emphasizes the importance of early recognition and intervention, particularly surgical, in cases of recurrent volvulus, where conservative management proves inadequate. The case also highlights the need for a multidisciplinary approach to improve patient outcomes and provides a basis for further investigation into the gastrointestinal phenotype of SKS and the molecular mechanisms underlying these associations.

Key words
complication, *MTOR*, sigmoid, Smith–Kingsmore, volvulus

CECILIA GALLO

Myelin oligodendrocyte glycoprotein antibody–associated disease with acute disseminated encephalomyelitis phenotype following a primary Epstein–Barr virus infection

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CASE DESCRIPTION Myelin oligodendrocyte glycoprotein antibody–associated disease (MOGAD) is an inflammatory demyelinating disease of the central nervous system, different from multiple sclerosis and neuromyelitis optica spectrum disorder, identified by anti-MOG antibodies. The diagnosis requires MOG immunoglobulin G tests and may appear with acute disseminated encephalomyelitis (ADEM). A case of a teenager with ADEM following an EBV infection was reported, questioning previous evidence of an association between Epstein–Barr virus (EBV) and MOGAD.

This is a case of a 16-year-old woman with a medical history of celiac disease and substance use, who sought consultation for gait disturbance, instability, tremor at rest, and difficulties in coordinating fine and gross motor skills movements.

At first, the patient presented with headache, malaise, and weakness, followed by urinary and rectal symptoms. Acute EBV infection was detected with negative results for other infections and autoimmune diseases. She was discharged after symptomatic treatment, but was readmitted 2 days later with transient diplopia. Imaging studies highlighted ground-glass opacities in the right lung. The patient received antibiotic treatment for suspected respiratory infection. Weeks later, she developed progressive neurological symptoms, including ataxia and pyramidal signs. Magnetic resonance imaging (MRI) showed hyperintense injuries in the brain and spinal cord, and studies were positive for anti-MOG antibodies.

The diagnosis of ADEM was made, with features of MOGAD possibly triggered by an EBV infection. Treatment with methylprednisolone was started, with a significant clinical improvement. The patient evolved positively, completely recovering her neurological function and returning to her usual sporting activity without sequelae. The relationship between MOGAD and EBV is barely reported on in the literature, with only 1 similar case identified. MOGAD is an autoimmune demyelinating disease with such main phenotypes as AEDM, optic neuritis, and myelitis. ADEM is common in children and manifests on MRI with encephalopathy and extensive injuries. It has been suggested that infections, such as EBV, could trigger MOGAD by disrupting the hematoencephalic barrier and activate T and B cells. The persistence of MOG immunoglobulin G is associated with greater risk of relapse, especially after ADEM.

Long-term immunotherapy, including corticosteroids and immunosuppressants, is the key to reducing recurrences and improving the long-term prognosis. Serological monitoring is essential

Key words
acute disseminated encephalomyelitis, Epstein–Barr virus, myelin oligodendrocyte glycoprotein antibody–associated disease

CHOWDHURY ADNAN SAMI

A misleading urinary tract infection: when culture-negative pyuria hides something more

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INTRODUCTION Sterile pyuria is a diagnostic dilemma, often attributed to genitourinary tuberculosis. Nontuberculous mycobacteria (NTM) are infrequent entities known to present with mimics of genitourinary tuberculosis, particularly in immunosuppressed patients. NTM infective endocarditis is a rare but lethal opportunistic infection, which can manifest as native or prosthetic valve endocarditis that accompanies cardiac intervention.

CASE DESCRIPTION A 63-year-old man, diabetic and hypertensive, with a history of coronary artery stenting, presented with fever, burning micturition, and weight loss lasting for 2 months. Initial evaluation showed sterile pyuria, but later the urine was positive for acid-fast bacilli, which was corroborated by a positive interferon-γ

release assay of a blood sample. This led to a diagnosis of genitourinary tuberculosis, and the patient was started on antitubercular therapy. Despite treatment, the fever continued and later the patient developed dyspnea and palpitations. This time, on re-evaluation, echocardiography showed multiple vegetations on the aortic valve and new blood and urine cultures were persistently positive for Mycobacterium abscessus subspecies abscessus, consistent with disseminated NTM infection with infective endocarditis. The patient was then placed on injectable amikacin, linezolid, and clarithromycin, with transient improvement. However, after 11 weeks he experienced a myocardial infarction and died 13 hours later.

DISCUSSION NTM infective endocarditis is a rare but serious complication, especially after stenting, generally characterized by subacute symptoms and negative cultures, especially in the early phase. Endothelial dysfunction secondary to chronic infection may have contributed to the fatal cardiac complication.

CONCLUSIONS This case illustrates the common diagnostic challenge encountered in distinguishing NTM from genitourinary tuberculosis, particularly in the context of culture-negative pyuria with persistent fever. Clinicians need to consider NTM as a cause for culture-negative pyuria, the need for mycobacterial cultures for diagnostic purposes, and the importance of screening for infective endocarditis.

Key words
disseminated nontuberculous mycobacteria , NTM infective endocarditis, NTM mortality, NTM urinary tract infection

DOMINGA GARCÍA

Testicular mass with a twist: from malignancy to polyarteritis nodosa

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INTRODUCTION The primary concern in young patients with a solid testicular mass, is malignancy (>90%). However, benign causes (<8%), such as testicular vasculitis (<1%), should also be considered. Polyarteritis nodosa (PAN) is the most common vasculitis affecting the testes (70%), though testicular involvement occurs only in 2%–17% of PAN cases, typically presenting as testicular pain without a palpable mass. Presentation as an isolated testicular mass is rare (<1%) and poses a diagnostic challenge, as illustrated in this case.

CASE DESCRIPTION A 28-year-old man presented with acute left inguinal and lumbar pain. Imaging showed a 2.5-cm left testicular tumor with uncertain inflammatory/hemorrhagic features on ultrasound. Given suspected malignancy, radical orchiectomy was performed. Histopathology showed necrotizing vasculitis of medium-sized arteries without neoplastic cells. Comprehensive rheumatologic evaluation ruled out common primary and secondary vasculitides and systemic involvement, leading to the diagnosis of localized PAN.

DISCUSSION Localized PAN lacks specific diagnostic criteria, making histopathology the gold standard. Less invasive biopsy techniques and advanced imaging have been proposed for presurgical identification, but their effectiveness remains uncertain. Establishing management protocols for atypical testicular lesions is crucial to optimize diagnosis and minimize unnecessary invasive surgeries.

CONCLUSIONS Isolated testicular PAN remains a diagnostic challenge. Improved pre-surgical strategies are needed to distinguish inflammatory from neoplastic testicular masses, reducing unnecessary morbidity from invasive procedures.

Key words
polyarteritis nodosa, testicular neoplasms, vasculitis

EMA SOMEN

Acute pancreatitis caused by hemobilia, following liver trauma

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INTRODUCTION Gastrointestinal bleeding is a common condition associated with high morbidity and mortality. Hemobilia, though rare, is a potential cause of bleeding into the biliary ducts. In Western countries liver trauma is the leading cause of hemobilia, with other causes including neoplasms, abscesses, and hepatic artery aneurysm rupture.

CASE DESCRIPTION This report describes a case of a 29-year-old man who presented with hematemesis caused by hemobilia following liver trauma, which also led to acute pancreatitis. The patient, who had sustained severe injuries in a traffic accident, including a liver laceration, underwent emergency surgery. He was discharged the following day in a stable condition, but was readmitted to the emergency department with hematemesis. Laboratory workup showed mild anemia (hemoglobin, 105 g/l), stable renal function (urea, 7.6 mmol/l; creatinine, 46 µmol/l), and normal prothrombin time. Physical examination was unremarkable and the patient's condition remained stable. Upper gastrointestinal endoscopy showed moderate erosive mucosal changes in the antrum, but no active bleeding or mucosal defects were identified and no additional treatment was required. The patient was admitted to the intensive care unit for further monitoring. Several hours later, he developed chest pain and difficulty breathing. Normal electrocardiogram and D-dimer levels ruled out pulmonary embolism and pulmonary computed tomography angiography was performed for further evaluation. Follow-up laboratory tests showed no abnormalities that could explain the symptoms. The patient then developed abdominal pain, initially in the epigastric region and later radiating to the right upper quadrant. Abdominal ultrasound showed moderate intrahepatic bile duct dilatation and a common bile duct measuring 6 mm. Laboratory test results showed elevated liver enzymes (aspartate aminotransferase, 481 U/l; alanine transaminase, 483 U/l; γ-glutamyl transferase, 557 U/l; alkaline phosphatase, 1212 U/l), serum amylase (346 U/l), and lipase (457 U/l) levels, leading to the diagnosis of acute pancreatitis. Despite conservative management, the patient continued to experience hematemesis, prompting repeat upper gastrointestinal endoscopy. A large clot was found in the gastric fundus and irrigation showed no active bleeding or mucosal defects. Clots were also noted in the duodenal bulb extending into the descending duodenum, with fresh bleeding observed at the papillary orifice, likely the cause of acute pancreatitis. After irrigation, the bleeding was controlled, likely due to tamponade of the common bile duct. Computed tomography aortography confirmed hemobilia caused by a ruptured pseudoaneurysm of the right hepatic artery. Emergency embolization of the right hepatic artery was performed and the patient remained stable. Follow-up abdominal magnetic resonance imaging showed no pathological findings. The patient was discharged on the 10th day of hospitalization in good general condition, with no further complications.

DISCUSSION This case highlights a rare occurrence of hemobilia following liver trauma and the subsequent development of acute pancreatitis. Hemobilia should be considered in patients with gastrointestinal bleeding following liver trauma, as it can lead to severe complications, such as biliary obstruction, cholestasis, and pancreatitis.

CONCLUSIONS Early diagnosis and intervention, including embolization, are essential to managing such cases effectively.

Key words
acute pancreatitis, hemobilia, liver trauma

ENFAL SILINI

Beyond the obvious: atypical immune-mediated syndrome in an elderly patient

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INTRODUCTION Late-onset systemic lupus erythematosus (LOLE) is an uncommon entity that often deviates from classical lupus presentations, making diagnosis challenging. In elderly patients, systemic inflammation with multisystem involvement can mimic malignancy, chronic infection or inflammatory myopathies, leading to unnecessary investigations and delayed treatment.

CASE DESCRIPTION A 74-year-old woman with a 6-month history of progressive asthenia, fever, weight loss, and lower limb weakness was referred for further evaluation. She developed nonpitting edema, ulcerative cutaneous lesions, and progressive functional decline, ultimately becoming nonambulatory.

Initial workup suggested paraneoplastic syndrome or inflammatory myopathy due to persistent systemic inflammation (C-reactive protein, 200 mg/l), anemia (hemoglobin, 8.6 g/dl), hypoalbuminemia (22 g/l), and renal impairment (creatinine, 8 mg/dl; proteinuria, 0.5 g/24h). Imaging showed right-sided pleural effusion without malignancy and bone marrow examination was unremarkable.

Further immunologic testing showed high-titer antinuclear antibody (1/1000) and elevated anti–double-stranded DNA (1/640), prompting reconsideration of an immune-mediated etiology. Retrospective review uncovered a history of postpartum inflammatory arthritis, reinforcing the suspicion of autoimmune disease. The patient was diagnosed with LOLE presenting as an inflammatory-edematous syndrome, a phenotype often misclassified due to its overlap with other systemic conditions.

Treatment with high-dose corticosteroids (0.7 mg/kg/day) led to a rapid resolution of edema, improvement in functional status, and normalization of inflammatory markers, confirming the autoimmune origin of the disease.

CONCLUSIONS This case highlights the atypical presentations of LOLE in elderly patients, particularly when classical lupus features are absent. The presence of nonpitting edema in systemic inflammation should prompt consideration of capillary hyperpermeability rather than nephrotic proteinuria. Additionally, historical clinical events, such as past episodes of inflammatory arthritis, may serve as crucial retrospective diagnostic clues. A high index of suspicion and timely immunosuppressive therapy are essential to prevent misdiagnosis and unnecessary oncologic investigations.

Key words

autoimmune disease, geriatric lupus, late-onset systemic lupus erythematosus, nonpitting edema, systemic inflammation

FLORENCIA LILIANA GARCIA

À propos of dementia

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CASE DESCRIPTION Cryptococcosis is a fungal infection caused by the *Cryptococcus* complex, including *C. neoformans*, which primarily affects immunocompromised individuals, and *C. gattii*, which commonly affects immunocompetent individuals. The infection typically begins in the lungs and can spread to other organs, particularly the central nervous system, where it presents as meningoencephalitis and, less frequently, as localized granulomatous lesions known as cryptococcomas. On this occasion, in 2022, at San Bernardo Hospital, a 76-year-old woman from the province of Salta, with a history of high blood pressure, insulin-dependency, hypothyroidism, diabetes, and occasional alcohol consumption diabetes (according to a companion relative) began her consultations for acute confusional state. Neurologist evaluations, laboratory testing, and imaging had been carried out on multiple occasions, without reaching an accurate diagnosis. The symptoms coincided with multiple urinary tract infections. The response to antibiotic treatment was good. Multiple cerebrospinal fluid tests showed hypoglycorrhachia. Due to a suspected diagnosis of tuberculosis, a sample was sent for culture, and it was decided to initiate antitubercular and infectology treatment. The latter was suspended due to elevated transaminases and negative results of sample test. Up until now, the patient had been neurologically evaluated and managed in outpatient follow-up with multiple hospitalizations due to confusional states at times associated with seizures. In January of 2025, emergency services transferred the patient to our hospital due to altered mental status. Emergency computed tomography of the brain was performed, showing hypodense lesion with perilesional edema, which was evaluated by a neurologist. Biopsy of the lesion was performed and it was positive for *Cryptococcus*. Antibiotics and amphotericin were started.

Cryptococcus infection is an opportunistic and rare disease in immunocompetent individuals. Therefore, this entity should be considered as a differential diagnosis in patients with neurological symptoms, as major complications are involved. It is vital to emphasize the care and handling of bird droppings and to avoid exposure to them.

Key words

central nervous system, cerebral abscess, cryptococcosis, immunocompetent

ISAAC NG

Does this ring a bell? An unusual case of idiopathic peripheral facial nerve palsy with cerebrospinal fluid varicella-zoster virus positivity

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CASE DESCRIPTION Idiopathic peripheral facial nerve palsy (also known as "Bell palsy") is a common clinical diagnosis given in the presence of characteristic sudden-onset, unilateral, isolated facial nerve palsy without other concerning symptoms or signs. However, presence of atypical features, such as gradual symptom onset, bilateral or recurrent facial nerve palsies, polycranial neuropathies, focal neurological deficits or other systemic manifestations, warrants further investigations, such as laboratory tests, neuroimaging, and lumbar puncture. Herein, we describe a unique case of an elderly Chinese woman, with left-sided peripheral facial nerve palsy associated with subjective dysphagia or dysarthria, and a 3-week long prodromal history of left-sided headache with sensory loss over left trigeminal nerve territory on crude touch. As such, magnetic resonance imaging of the brain and internal acoustic meatus was performed, showing no cavernous sinus or cerebellopontine angle mass lesions on leptomeningeal enhancement. Facial nerve conduction study showed evidence of left facial neuropathy, while blink reflex did not detect trigeminal nerve involvement. Cerebrospinal fluid analyses showed normal white cell counts and biochemistry (glucose/protein levels), but varicella-zoster virus DNA polymerase chain reaction was positive. Given the absence of other clinical,

radiological, or cerebrospinal fluid findings suggestive of meningoencephalitis, the patient was treated for peripheral facial nerve palsy probably triggered by varicella-zoster virus reactivation with 7-day regimen of prednisolone and 10-day regimen of antiviral (acyclovir/valacyclovir) treatment, and she made a good clinical recovery at 1-month follow-up.

Key words
facial nerve palsy

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Headache secondary to neurosyphilis

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INTRODUCTION Neurosyphilis is a central nervous system infection caused by *Treponema pallidum*. It can occur at any stage of syphilis and presents diverse neurological symptoms, often mimicking other conditions. While uncommon in the general population, neurosyphilis remains a significant concern in immunocompromised patients, particularly those with HIV infection.

CASE DESCRIPTION We report a case of a 37-year-old man from Ciudad Juárez with a history of idiopathic thrombocytopenic purpura and anxiety disorder. He presented with a 3-week history of vertigo, progressive headache, nausea, and neurological deterioration, including mood changes, bradyphasia, bradypsychia, and left-sided weakness. Initial neurological examination showed mild left-sided motor weakness without sensory deficits or meningeal signs. Laboratory test results showed mild anemia and thrombocytopenia, with elevated inflammatory markers. The patient subsequently developed worsening left-sided hemiplegia, seizures, and signs of meningeal irritation. Follow-up imaging showed right frontoparietal ischemia. Serologic testing confirmed HIV infection with a cluster of differentiation 4 count of 26 cells/µl. Cerebrospinal fluid analysis was negative for bacterial pathogens, but positive for *T. pallidum* (fluorescent treponemal antibody absorption test), with a reactive serum venereal disease research laboratory test confirming neurosyphilis. The patient was started on intravenous penicillin G. Despite treatment, he exhibited a poor neurological response and progressive deterioration, ultimately resulting in a fatal outcome.

DISCUSSION This case highlights the importance of considering neurosyphilis in patients presenting with unexplained neurological symptoms, especially those with HIV or other immunosuppressive conditions. Neurosyphilis can present variably from asymptomatic forms to stroke-like syndromes, making diagnosis challenging. In this case, initial symptoms were nonspecific, and the delayed recognition of neurosyphilis contributed to a poor prognosis. The presence of vascular complications, as evidenced by cerebral ischemia, underscores the potential severity of untreated neurosyphilis.

CONCLUSIONS Neurosyphilis remains a diagnostic challenge due to its diverse presentations and overlapping with other neurological disorders. Early recognition and treatment are crucial, particularly in high-risk populations, such as HIV-positive individuals. This case underscores the need for routine syphilis screening in patients with neurological symptoms of unclear etiology, especially those with risk factors for sexually transmitted infections.

Key words
central nervous system infection, headache, neurosyphilis

JOSÉ LUIS CAMBRON JIMENEZ

From silent clots to neuropathy: a delayed diagnosis of pernicious anemia

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INTRODUCTION Vitamin B_{12} is essential for neurological function and hematopoiesis. Its deficiency can cause anemia, neuropathy, and increased thrombotic risk due to hyperhomocysteinemia. Pernicious anemia is a frequent but underdiagnosed cause, often detected after neurological complications. We present a case of pernicious anemia associated with recurrent deep vein thrombosis (DVT) and neurological impairment.

CASE DESCRIPTION In 2018, a 53-year-old man with a history of tobacco use presented with an unprovoked right-sided DVT. He was asymptomatic and achieved recanalization after rivaroxaban treatment. In 2019, due to recurrent DVT and no clear risk factors, he was referred for thrombophilia screening but was not evaluated. In 2020, a contralateral DVT developed, leading to reinitiation of anticoagulation.

By 2021, the patient developed progressive lower limb weakness, proprioceptive deficits, and plantar dysesthesia. Neurological examination showed a positive Romberg sign, bilateral DVT with hypotrophy, and reduced anterior hair growth.

Laboratory results showed macrocytosis (mean corpuscular volume, 120 fl) without anemia, severe hyperhomocysteinemia (>50 µmol/l), elevated methylmalonic acid levels, and vitamin B_{12} level of under 150 pg/ml. A diagnosis of vitamin B_{12} deficiency-related neuropathy was made and parenteral vitamin B_{12} was initiated. Weakness persisted despite treatment. Electromyography confirmed peripheral neuropathy and magnetic resonance imaging showed L4–L5 spondylolisthesis. The patient improved from a walker to a cane. Further studies showed macroovalocytes, dacryocytes, hypersegmented neutrophils, normal lactate dehydrogenase, and negative autoimmune markers. By mid-2021, the patient showed partial recovery with persistent Romberg positivity. In 2022, antiparietal cell antibodies confirmed pernicious anemia. Homocysteine normalized (7.42 µmol/l). In 2023, macrocytosis resolved (mean corpuscular volume, 90.3 fL), and endoscopic biopsy showed *Helicobacter pylori*–associated gastritis.

By 2024, the patient remained stable under hematologic surveillance. Doppler ultrasound confirmed DVT recanalization, but untreated *H. pylori* infection raised concerns about gastric atrophy and B_{12} malabsorption.

DISCUSSION Vitamin B_{12} deficiency can manifest as macrocytosis without anemia, delaying diagnosis. Neurological complications include proprioceptive loss, paresthesia, and ataxia due to spinal cord degeneration. Hyperhomocysteinemia increases thrombotic risk, in this case likely contributing to recurrent DVT. Delayed diagnosis led to significant morbidity. Vitamin B_{12} deficiency was overlooked despite macrocytosis, pernicious anemia was identified only after severe symptoms showed, and untreated *H. pylori* infection may have worsened malabsorption.

CONCLUSIONS Macrocytosis warrants vitamin B_{12} level assessment, even without anemia. Hyperhomocysteinemia in unprovoked DVTs should prompt B_{12} level evaluation. Early diagnosis can prevent irreversible neurological damage. Pernicious anemia should be suspected in chronic atrophic gastritis cases. *H. pylori* eradication is essential in pernicious anemia to prevent malabsorption. This case underscores the importance of early detection and management of vitamin B_{12} deficiency to prevent severe complications.

Key words
deep vein thrombosis, hyperhomocysteinemia, neuropathy, pernicious anemia, vitamin B12 deficiency

JUAN DAVID SALAZAR OSPINA

Facing Koch's shadow: a battle against drug-resistant tuberculosis

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INTRODUCTION Tuberculosis (TB) remains a global threat, particularly in low- and middle-income countries, where diagnostic and therapeutic limitations persist. We present a remarkable case of gastrointestinal multidrug-resistant TB (MDR-TB) in an immunocompetent young woman, highlighting diagnostic challenges, therapeutic constraints, and drug-related complications.

CASE DESCRIPTION A 21-year-old woman presented with a 2-month history of bloody diarrhea, abdominal pain, weight loss, and constitutional symptoms. Initial evaluation showed moderate anemia and inflammatory markers elevation; HIV testing was negative. Abdominal computed tomography showed retroperitoneal lymphadenopathy and cecal thickening, prompting colonoscopy, which showed ulcerative lesions with histologic evidence of granulomatous inflammation. Thoracic computed tomography demonstrated endobronchial spread with cavitary lesions, despite the absence of respiratory symptoms. Polymerase chain reaction assays on samples obtained through bronchoscopy and colonoscopy confirmed *Mycobacterium tuberculosis* with rifampicin resistance. The patient was diagnosed with gastrointestinal and pulmonary TB with rifampicin resistance. Due to limited local availability of bedaquiline and pretomanid, a modified regimen including levofloxacin, linezolid, clofazimine, and isoniazid was initiated. The course was complicated by ileal perforation, requiring surgical resection, and later by irreversible peripheral neuropathy attributed to prolonged linezolid use. Despite these setbacks, the patient completed 12 months of therapy, achieving clinical remission with residual neurological deficits.

DISCUSSION This case exposes the devastating course that MDR-TB can take even in HIV-negative patients. It challenges the traditional perception of TB as primarily a pulmonary disease, revealing how extrapulmonary involvement can delay diagnosis, complicate management, and worsen prognosis. Despite the World Health Organization's endorsement of novel, highly effective regimens, such as BPaLM, many high-burden countries still lack access to essential drugs, such as bedaquiline and pretomanid, forcing clinicians into piecing together suboptimal and toxic combinations.

Beyond individual outcomes, this case illustrates a broader systemic failure—the persistent inequity in access to modern TB care. In settings where TB is most prevalent, patients paradoxically face the greatest barriers to effective treatment. This widening gap undermines global eradication efforts and perpetuates preventable suffering. A comprehensive response must not only involve expanding drug availability, but also equipping clinicians with the expertise to tailor regimens safely and detect early signs of toxicity. The war against MDR-TB will not be won with outdated arsenals; it demands bold policy changes, global solidarity, and relentless innovation at the bedside.

CONCLUSIONS The management of MDR-TB demands more than medical expertise; it calls for strategic decision-making under resource limitations, adaptability to therapeutic constraints, and close monitoring for adverse effects. This case highlights how the lack of access to optimal antibiotics prolongs diagnostic uncertainty, complicates treatment, and exposes patients to preventable morbidity. Strengthening global drug availability, fostering clinical expertise in complex TB management, and promoting early recognition of extrapulmonary forms are crucial steps to improving outcomes in high-burden settings.

Key words
gastrointestinal tuberculosis, global health, linezolid toxicity, multidrug-resistant tuberculosis, tuberculosis

CHANDANA KUMARA SAI JANNU

Unmasking antisynthetase syndrome: when interstitial lung disease takes the lead

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INTRODUCTION Antisynthetase syndrome (ASyS), an autoimmune disease that affects multiple organs, was first described in 1990. The global estimated incidence of ASyS is 0.56 cases per 100 000, and an estimated prevalence is 9.2 cases per 100 000.

CASE DESCRIPTION A 44-year-old woman without diabetes or hypertension presented with progressive dyspnea lasting 3 months, without any associated cough, fever, facial swelling, or pedal edema. There was no significant history of past, family, drug, or environmental exposures. Bilateral basal inspiratory crackles were heard on auscultation. High-resolution computed tomography showed features of interstitial lung disease (ILD). The results of extensive serologic workup were strongly positive for anti-Jo1 antibodies. Later in the course of the disease, the patient developed mechanic's hands.

With the picture of ILD, anti-Jo1 antibody positivity, and mechanic's hands, the patient was diagnosed with ASyS. Then, the patient was started on rituximab along with steroid therapy. After 2 months, the patient developed steroid-related adverse effects and, as a result, steroid-sparing mycophenolate mofetil therapy was initiated. Later, in 6 months, clinical and radiological improvement was seen. At present, the patient is on maintenance steroid-sparing mycophenolate mofetil therapy and receives rituximab every 6 months.

DISCUSSION The most common causes of ILD in India are hypersensitivity pneumonitis, sarcoidosis, connective tissue diseases, and idiopathic pulmonary fibrosis. Connective tissue diseases most commonly associated with ILD include rheumatoid arthritis, systemic sclerosis, and idiopathic inflammatory myopathies.

ASyS is a rare autoimmune, inflammatory myopathic disorder with limited evidence-based guidelines for its diagnosis and management. Common presentations include myositis, polyarthritis, ILD, mechanic's hands, and Raynaud phenomenon. The affected individuals do not usually have all of the symptoms. The autoantibodies that have been identified in this disorder include anti-Jo1, anti-EJ, anti-OJ, anti-PL7, anti-PL12, anti-SC, anti-KS, anti-JS, anti-HA, anti-YRS, anti-tryptophanyl, and anti-Zo antibodies, and each of these target a different aminoacyl-tRNA synthetase. Anti-Jo1 is the most common autoantibody in individuals with ASyS. Mechanic's hands and anti-Ro-52 antibody are most commonly associated with the development of ILD in patients with ASyS. Several case reports and case series have been published where rituximab and glucocorticoids were successfully used as the first-line treatment for rapidly progressive ILD. Based on those data, rituximab was initiated in our patient and led to clinical improvement.

CONCLUSIONS Only 2% to 11% patients with ASyS present with ILD in the absence of myositis, where the diagnosis may be critically delayed, hindering the management of this rapidly progressive

disease. Early rituximab therapy have shown positive outcomes in these patients.

Key words
antisynthetase syndrome, interstitial lung disease, rituximab

CHRISTIAN SZABÓ

A headache that made us think twice

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INTRODUCTION Headache is a common symptom encountered in emergency medicine, often benign but sometimes indicative of an underlying pathology. This case highlights an elderly patient with post-traumatic headache, which later revealed an underlying electrolyte disturbance requiring targeted management.

CASE DESCRIPTION A 71-year-old woman called emergency services on January 24, 2025, complaining of persistent headache. She had fallen the previous morning following orthostatic dizziness, possibly experiencing transient loss of consciousness. Since then, she reported a constant headache in the occipital and left parietal region, along with nausea but no vomiting or vertigo. Upon arrival, she was alert, fully oriented, and without neurologic deficits. No external signs of trauma were observed. Vital signs were stable (blood pressure, 150/94 mm Hg; body temperature, 36.4 °C; Glasgow Coma Scale, 15). She was transported to the hospital for further evaluation.

At the hospital, her past medical history revealed hypertension, rheumatoid arthritis, incompetence of the gastric cardia, and treated breast carcinoma. Her medications included vitamin D, calcium, prednisone, antihypertensive drugs, and proton pump inhibitors. The physical examination showed pain on palpation of the occipital and left parietal skull. Computed tomography of the head showed a paramedial parietal skull fracture without intracranial hemorrhage. Laboratory findings included hyponatremia (132 mmol/l) and mild hyperglycemia (6.25 mmol/l). She was diagnosed with orthostatic syncope with fall, mild traumatic brain injury, and skull fracture. Management included analgesia, hydration, vital sign monitoring, and slow mobilization. The results of the neurologic examination remained normal, and she was discharged on January 27, 2025, with follow-up recommendations. On February 2, 2025, she returned due to worsening headache and new-onset diplopia. The pain had progressively intensified, unrelieved by analgesics, and was exacerbated by mild activity. The neurologic examination showed paresis of central facial nerve and left abducens nerve. No abnormalities were found on repeat head computed tomography. Laboratory tests showed severe hypotonic hyponatremia and hypochloremia. Further endocrinologic workup ruled out hypothyroidism and hypocortisolism. The diagnosis of post-concussion syndrome of inappropriate antidiuretic hormone secretion (SIADH) was established. Treatment included intravenous hypertonic saline, fluid restriction, dietary salt supplementation, and a temporary increase in corticosteroid dosage.

With treatment, electrolyte levels improved, and symptoms resolved. The patient was discharged in a stable condition with strict fluid management and scheduled follow-ups.

DISCUSSION This case highlights the need for thorough evaluation in elderly patients after trauma. Delayed neurologic symptoms prompted further investigation, revealing SIADH-related hyponatremia. Post-traumatic SIADH is an underrecognized condition that can lead to severe complications if not identified and managed appropriately. This case highlights the necessity of ongoing monitoring and electrolyte surveillance in high-risk patients.

CONCLUSIONS Persistent headache in elderly patients after trauma warrants close follow-up. Post-traumatic SIADH should be considered in cases of worsening headache with electrolyte imbalance. Neurologic symptoms, such as diplopia, may indicate metabolic or endocrine disturbances rather than direct trauma. The early recognition and management of SIADH can prevent serious complications. Multidisciplinary follow-up is crucial to ensure long-term stability.

Key words
electrolyte imbalance in traumatic brain injury, head trauma complications, management of SIADH in head injury, post-traumatic syndrome of inappropriate antidiuretic hormone secretion

CLARA CHEUNG

The great glucose heist: hypoglycemia beyond the pancreas

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CASE DESCRIPTION Hypoglycemia in a young patient with no prior remarkable medical history is a rare and potentially serious clinical situation. The main causes to consider include endocrine and metabolic disorders, drug intoxications, and paraneoplastic syndromes. A rigorous diagnostic approach is essential to identify the underlying cause and guide therapeutic management.

A 38-year-old man with no medical history was found unconscious. Upon arrival of emergency services, the patient had decreased alertness but was calm, with a normal blood pressure of 134/74 mm Hg, a heart rate of 34 bpm, oxygen saturation of 98% on 9 liters of oxygen via mask, a blood glucose level of 0.7 mmol/l, and a potassium level of 2.3 mmol/l, without electrocardiographic abnormalities. He was immediately supplemented with 2 ampoules of intravenous 30% glucose and intravenous potassium in a dose of 2 g. The patient's condition improved rapidly, with resolution of symptoms and normalization of capillary blood glucose and electrolyte imbalances. Recurrent hypoglycemia upon discontinuation of glucose supplementation motivated his admission to the intensive care unit, where glucose supplementation was continued with up to 2 liters of 10% glucose in addition to oral intake, alongside potassium supplementation. A decline in general health was reported (fatigue, weight loss with loss of appetite, abdominal discomfort). No other noticeable clinical findings were seen. Laboratory test results revealed hypercalcemia associated with hypophosphatemia and anicteric hepatic cholestasis accompanied by mild hepatic cytolysis predominantly affecting aspartate aminotransferase. Standard laboratory tests were otherwise normal. Insulin and C-peptide levels during hypoglycemia were low, with negative anti-insulin receptor and anti-insulin antibodies. Further calcium-phosphorus workup showed notably elevated parathyroid hormone–related protein levels. The CA19-9 level was moderately elevated, in contrast to normal levels of other oncologic biomarkers. A thoraco-abdomino-pelvic computed tomography scan showed suspicious pulmonary and hepatic lesions along with peritoneal carcinomatosis, but no other secondary lesions or pancreatic involvement (confirmed by dedicated magnetic resonance imaging).

The patient was started on diazoxide and a course of zoledronic acid. Following clinical stabilization, the patient was transferred to the internal medicine department. A liver biopsy showed a CK7+/CK20– adenocarcinoma of undetermined primary origin, microsatellite stability, HER2 negativity (score 0), no identified mutations, no gene fusion, and no PD-L1 overexpression. Considering the histologic findings, disease extent, predominance of lesions in the biliary tract, and the patient's young age, a diagnosis of metastatic cholangiocarcinoma was established.

The diazoxide dose was increased later and combined with systemic corticosteroids and a second course of zoledronic acid. Ultimately, insulin-like growth factor 1 (IGF-1) and IGF-2 levels were measured, with an IGF-2/IGF-1 ratio greater than 15, which supported IGF-2 hypersecretion of paraneoplastic origin. The patient began chemotherapy with cisplatin-gemcitabine, later combined with durvalumab. Early follow-up showed favorable outcomes, including normalization of calcium levels, weaning of glucose supplementation, and discontinuation of diazoxide and hydrocortisone.

Non–islet-cell tumor hypoglycemia is a phenomenon characterized by the production of a partially processed form of pro-IGF-2, responsible for insulin-like biological activity and increased IGF bioavailability. IGF-2 interacts with IGF-1 receptors and insulin receptors on target tissues, leading to hypoglycemia. It is a rare but important differential diagnosis to consider in cases of refractory hypoglycemia with a paraneoplastic presentation.

Key words
hypoglycemia, non–islet-cell tumor hypoglycemia, paraneoplastic syndrome

CLARE O'BRIEN

When all is not what it seems

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CASE DESCRIPTION A 63-year-old woman presented to the Galway University Hospital with a 2-week history of vomiting and diarrhea on a background of poorly controlled type 1 diabetes. She reported being generally unwell for the preceding 6 months and had associated unintentional weight loss. Four weeks prior to her presentation, she was treated in another hospital for cellulitis. She was hypotensive and tachycardic on examination, with a Glasgow Coma Scale score of 13/15 and generalized abdominal discomfort. Investigations showed hyperglycemia and ketonemia with high anion gap metabolic acidosis along with acute kidney injury, hyperkaliemia, and hypoalbuminemia.

The working diagnosis was diabetic ketoacidosis secondary to sepsis of unknown origin. A diabetic ketoacidosis protocol was commenced with intravenous antibiotics and steroids, and the patient was transferred to the high-dependency unit for inotropic support. Of note, her list of regular medications was not available at presentation. Chest X-ray showed bilateral pleural effusion, and computed tomography of the thorax, abdomen, and pelvis—a likely thrombus within the left common femoral vein. Therapeutic anticoagulation was initiated. Despite resolution of the patient's hyperglycemia and ketonemia, her metabolic acidosis persisted, and on day 5 of her admission she developed acute transaminitis with synthetic liver dysfunction requiring vitamin K and fibrinogen replacement. The differential diagnosis was vitamin K deficiency secondary to malnutrition. She was commenced on enteral feeding, pancrelipase and intravenous vitamin B and C. Considering her bleeding risk, therapeutic anticoagulation was held. A subsequent Doppler ultrasound was normal. Gastroscopy showed flattening of the duodenum, and colonoscopy was normal, with no evidence of microscopic colitis on histology.

An initial drug reconciliation identified that during her last hospital admission amlodipine was discontinued due to ankle swelling and olmesartan was started. Further reconciliation with her pharmacy showed that she was commenced on amlodipine/olmesartan 20 mg/5 mg in May 2019 by her registered general practitioner. After this initial prescription, all subsequent prescriptions were issued by another general practitioner without clinical review, and the dose was noted to increase in September 2022 to 40 mg/5 mg. Considering this, the differential diagnosis was now angiotensin II receptor–induced enteropathy. The patient was commenced on rifaximin and budesonide. Her symptoms gradually improved and she was discharged home on day 33 of admission. She has since been reviewed in the gastroenterology outpatient clinic. She has completed treatment with budesonide and remains well.

Sprue-like enteropathy associated with angiotensin II receptor blockers was first described in 2012. Although the incidence is not entirely clear, it is thought to be rare. The mechanism of injury is not well established; however, an immune-mediated disorder is thought to occur in susceptible individuals. Cessation of the angiotensin II receptor blocker results in complete resolution of both clinical and histologic features. This case highlights the importance of vigorous medication reconciliation and the risks associated with prescribing for patients without clinical review.

Key words
angiotensin II receptor blocker–induced enteropathy, olmesartan-induced enteropathy

DIEU MI BUI

A severe case of diarrhea: Pharaoh's revenge

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INTRODUCTION Nowadays, the sodium-glucose cotransporter 2 inhibitors (SGLT-2i) represent the first-line therapy for heart failure (HF), diabetes, and chronic kidney disease. Based on the mechanism of action of SGLT-2i, we can predict the drug's main adverse effects: dehydration, hypotension, euglycemic ketoacidosis, genital mycotic and urinary infections.

CASE DESCRIPTION A 58-year-old woman was admitted to the intensive care unit due to hypotension, with a history of severe diarrhea, vomiting, and dizziness after returning from Egypt. The patient had a history of HF with reduced ejection fraction and was using the recommended medication for this condition all the time (beta-blockers, angiotensin receptor–neprilysin inhibitor, SGLT-2i, spironolactone)—those medications worsened her hypotension even more. On admission, blood tests showed severe acute kidney failure (the level of creatinine was 1721 μmol/l) caused by excessive volume depletion due to diarrhea in combination with osmotic diuresis and natriuresis induced by SGLT-2i. Other findings included hyponatremia and hyperkalemia. The kidney ultrasound did not show any significant abnormalities. The patient initially required catecholamine support and aggressive parenteral hydration to keep her stable. After a few days of intensive care (and discontinuation of dapagliflozin and other medication for HF), the patient's condition became stable. After that, polyuric phase of renal insufficiency occurred. The renal parameters normalized and the patient's condition got better. She was discharged with proper education about the adverse effects of gliflozins and was recommended an early check-up in an outpatient clinic.

DISCUSSION AND CONCLUSIONS In this case, the patient was not properly informed about temporarily stopping gliflozins if unwell, especially if experiencing vomiting, diarrhea, or fever. This lack of knowledge led to the situation when the patient got even more dehydrated by mistake, which could have been fatal. On the other hand, gliflozins are important in HF treatment, and this patient would absolutely benefit from these medications after complete recovery.

Key words
adverse effects of sodium-glucose cotransporter 2 inhibitors

EZEQUIEL PABLO LIBOVICH

Primary hyperparathyroidism and thromboembolic disease: an uncommon association

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INTRODUCTION Primary hyperparathyroidism (PHPT) is the third most common endocrine disorder, with an increasing prevalence ranging from 0.4% to 11% worldwide. It is characterized by the overproduction of parathyroid hormone (PTH), resulting in elevated serum calcium levels. On the other hand, acute venous thromboembolic disease, which includes venous thromboembolism and deep vein thrombosis (DVT), is a significant cause of global morbidity and mortality. While the musculoskeletal and renal manifestations of PHPT have been extensively documented, the association between hypercalcemia secondary to PHPT and hematologic alterations, particularly coagulopathy, remains poorly studied.

CASE DESCRIPTION A 49-year-old man with a history of recurrent nephrolithiasis and DVT presented with left lower limb edema and recent-onset dyspnea. Doppler ultrasound showed DVT, and chest computed tomography angiography— intermediate-risk bilateral thromboembolism. Laboratory findings included ionized calcium of 1.75 mg/dl, normal alkaline phosphatase and phosphate levels, and parathyroid hormone of 528 pg/l. Parathyroid ultrasound and scintigraphy showed left inferior parathyroid adenoma. Anticoagulation with low-molecular-weight heparin (1 mg/kg of body weight every 12 h) was started, along with bisphosphonates and parenteral hydration. Discharge was planned with temporary cinacalcet use for hypercalcemia associated with primary hyperparathyroidism, with a recommendation to defer surgery due to recent venous thromboembolism.

DISCUSSION Ionized calcium, better known as coagulation factor IV, plays a role in the intricate interactions that form the Virchow triad, tipping the hemostatic balance towards a prothrombotic state. Furthermore, patients with PHPT exhibit elevated levels of plasminogen activator inhibitor 1 and decreased levels of tissue factor pathway inhibitor, while PTH itself stimulates the production of interleukin 6, which promotes various procoagulant processes.

CONCLUSIONS Hyperparathyroidism and hypercalcemia may not be considered traditional risk factors for thrombotic events, but they can contribute as facilitating factors for thrombotic vascular occlusion. Special consideration is needed for thrombosis prophylaxis and strict monitoring of acute venous thromboembolic disease in patients with PHPT.

Key words

acute venous thromboembolic disease, hypercalcemia, parathyroid hormone, primary hyperparathyroidism, venous thromboembolism

FERHAT CAN

Sarcoidosis, tuberculosis, or IgG4-related disease? A diagnostic dilemma in the overlapping spectrum of granulomatous disorders

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INTRODUCTION Sarcoidosis is a chronic, multisystem granulomatous disease characterized by non-necrotizing granulomas, primarily affecting the lungs and other organs. It varies in severity from asymptomatic cases to fibrosis-related complications, with a higher prevalence in Northern Europe and African Americans. Immunoglobulin G4–related disease (IgG4-RD) is a rare fibroinflammatory disorder affecting multiple organs, characterized by storiform fibrosis and IgG4-positive plasma cell infiltration. It primarily affects middle-aged men. Due to overlapping clinical, radiologic, and histopathologic features, differentiating sarcoidosis from IgG4-RD is challenging. This report underscores the diagnostic complexity associated with distinguishing sarcoidosis from IgG4-RD.

CASE DESCRIPTION A 53-year-old woman, a primary school teacher, presented with a 1-year history of dry cough, fever, weight loss (10 kg in 4 months), night sweats, abdominal distension, and dyspnea. Physical examination showed generalized ascites, diminished breath sounds, xanthomas, and facial hyperpigmentation. Vital signs were stable.

The patient had a history of cholecystectomy (December 2022) for symptomatic gallstones. Histopathology suggested caseating granulomatous cholecystitis, leading to an initial diagnosis of intra-abdominal tuberculosis and a 9-month antituberculosis therapy (isoniazid, rifampicin, pyrazinamide, and ethambutol followed by isoniazid and rifampicin). However, systemic symptoms persisted. In February 2023, positron emission tomography / computed tomography showed widespread lymphadenopathy, hepatosplenomegaly, and bone marrow involvement. Biopsies showed necrotizing granulomatous lymphadenitis with IgG4-positive plasma cells and multifocal granulomas in bone marrow, requiring differentiation from granulomatous infections, sarcoidosis, and IgG4-RD. Re-evaluation of gallbladder biopsy at another center showed noncaseating granulomas, shifting the suspicion toward sarcoidosis. Nine months later, the patient was admitted to our hospital with persistently high erythrocyte sedimentation rate (>140 mm/h), C-reactive protein (60 mg/dl), elevated alkaline phosphatase (253 U/l), γ-glutamyl-transferase (86 U/l), and hypergammaglobulinemia (IgG, 32 g/l; IgG4 >1.52 g/l). Angiotensin-converting enzyme levels were elevated (79 U/l), but autoimmune and infectious serology results were negative. GP210 and SP100 antibody test results were also negative, excluding primary biliary cholangitis. Imaging showed heterogeneous liver parenchyma, pancreatic cystic lesions, and grade 1 esophageal varices, which suggested chronic liver disease. Hematologic studies showed polyclonal gammopathy but no clonal plasma cell proliferation.

A multidisciplinary team (rheumatology, hematology, gastroenterology, infectious diseases, pathology, radiology) determined sarcoidosis as the most likely diagnosis. The patient responded well to corticosteroid therapy, with resolution of systemic symptoms and normalization of inflammatory markers.

DISCUSSION Distinguishing systemic granulomatous diseases like sarcoidosis and IgG4-RD is challenging due to overlapping clinical, radiologic, and histopathologic features. Our patient was initially misdiagnosed with tuberculosis and received prolonged treatment, but persistent symptoms led to further evaluation. The discovery of noncaseating granulomas and elevated angiotensin-converting enzyme levels shifted the suspicion toward sarcoidosis, while increased IgG4-positive plasma cells raised concern for IgG4-RD. Given the absence of storiform fibrosis and the marked response to corticosteroids, sarcoidosis was favored as the primary diagnosis. However, long-term follow-up is essential to monitor for possible IgG4-RD manifestations.

CONCLUSIONS This case highlights the complexities of differentiating sarcoidosis from IgG4-RD due to overlapping features. A thorough histopathological review, integration of clinical and laboratory findings, and a multidisciplinary approach are essential for accurate diagnosis and effective management. Early recognition and appropriate immunosuppressive treatment are crucial to prevent disease progression and improve patient outcomes.

Key words

granulomatous inflammation, immunoglobulin G4–related disease, sarcoidosis, tuberculosis

GONZALO HERAS

A wolf in sheep's clothing: genital tuberculosis simulating obstetric sepsis

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INTRODUCTION *Mycobacterium tuberculosis* infection continues to be a major cause of death from infectious/contagious disease in low-income countries. The most frequent clinical presentation is pulmonary in 80% of cases and disseminated in 20% to 25%. According to different reports, genital localization occurs in fewer than 1% of cases.
CASE DESCRIPTION A 20-year-old woman with no known medical history, 15.4 weeks pregnant, was admitted with a fever of 38 °C lasting 3 days and pain in the hypogastrium, which was interpreted as acute pyelonephritis, and started treatment with IV ceftriaxone 1 g every 12 hours. After 72 hours, she began to have little blood loss through the genitals associated with mild pain on deep palpation of the hypogastrium and fever; the absence of fetal heartbeats was also noted. Due to suspicion of septic abortion, antibiotic therapy was changed to ampicillin-sulbactam plus clindamycin, and instrumental uterine curettage was performed. Due to persistent fever, chest computed tomography was performed and showed bilateral interstitial pulmonary infiltrates. Abdominopelvic ultrasonography showed free fluid in the peritoneum and enlarged right adnexa. It was interpreted as nosocomial sepsis, so the patient was switched to meropenem, colistin, and vancomycin. Blood culture was positive for *Burkholderia cepacia*, and colistin and vancomycin were withheld. As the patient remained febrile, tachycardic, with desaturation and progressive increase in abdominal distention, exploratory laparotomy was performed and the postoperative diagnosis was pelviperitonitis requiring total hysterectomy. The patient was transferred to the intensive care unit immediately after the procedure, where she remained for 5 days requiring MRA. Her condition was clinically stable, so she was moved to the common ward where due to continued febrile condition she was switched to meropenem for 12 days and vancomycin for 6 days. Fever did not improve, so it was decided to drain the surgical wound, leaving it exposed for the second closure. The pathology report from the surgical procedure demonstrated a chronic, granulomatous, caseating, tuberculous giant cell inflammatory process in the uterine body, cervix, right and left adnexa, and omentum, therefore treatment was started with rifampicin, isoniazid, pyrazinamide, and ethambutol. After 72 hours of starting the therapy, the fever curve decreased and the patient's general condition improved, so she was discharged from the hospital.
CONCLUSIONS It is important to have a high clinical suspicion of gynecological tuberculosis because if it is not suspected at the early stages, the condition can have catastrophic results. Diagnostic methods pose a special problem, since if they are not requested in time and the sample is not obtained correctly, they make the diagnosis of this disease even more difficult.

Key words
sepsis, septic abortion, tuberculosis

HANAË DE VECCHI

One purpura may hide another

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INTRODUCTION The causes of febrile purpura are diverse and sometimes life-threatening. Management depends on the underlying cause, and an often urgent treatment may negatively impact prognosis if improperly chosen.
CASE DESCRIPTION We report a case of a 55-year-old man, followed up due to well-controlled HIV-1 infection, who experienced recurrent episodes of purpura of the lower limbs since 1995. Over time, the patient developed some myalgias, diffuse arthralgias, urticarial lesions, and a facial edema episode.

Biological workup revealed hypocomplementemia, positive rheumatoid factor, and type II cryoglobulinemia. Skin biopsy confirmed leukocytoclastic vasculitis with C1q and C3c deposits, leading to the diagnosis of hypocomplementemic urticarial vasculitis.

We started to treat the patient with hydroxychloroquine but no improvement was observed. Febrile purpuric flare-ups became more frequent until the patient developed persistent febrile asthenia, purpura with violaceous and erythematous lesions of the lower limbs, reticulated purpuric lesions on the trunk and back, myalgias, arthralgias, and an elevated C-reactive protein level (100 mg/l).

Finally, an aerobic blood culture yielded positive results in 100 hours and gram-negative bacilli were identified, later confirmed as *Helicobacter cinaedi*. Rituximab therapy was stopped and the patient was treated with amoxicillin 1 g 3 times a day for 8 weeks with significant improvement. However, the patient experienced a relapse 1 year later, necessitating prolonged antibiotic therapy by amoxicillin and then doxycycline to minimize the risk of relapse.

Despite this treatment, the symptoms returned few years later. The patient was treated again for bacteremia and we decided to eradicate the gastrointestinal carriage of the bacteria with oral gentamicin for 3 months. Afterward, the patient developed new recurrent pseudo-urticarial lesions associated with edema and vascular purpura but he never had fever again. We concluded these were hypocomplementemic urticarial vasculitis flares and we initiated treatment with anakinra.
DISCUSSION *H. cinaedi* is a rare cause of infection and is difficult to diagnose because of the prolonged culture time (4–10 days). The number of cases is increasing not only among immunocompromised patients but also among those immunocompetent. The symptoms are nonspecific. Fever is the most common symptom. Frequent manifestations include gastrointestinal (proctitis, abdominal pain, gastroenteritis), cutaneous (dermohypodermitis, cellulitis, pyoderma gangrenosum, purpura), and articular (arthritis) involvement. Bacteremia is also common. More rarely, infected aortic aneurysms, abscesses, endocarditis, osteomyelitis, myopericarditis, and meningitis have been reported. The optimal treatment for *H. cinaedi* infections remains unknown. It is susceptible to carbapenems (meropenem, imipenem), amikacin, gentamicin, cefepime, and minocycline. It is recommended to use prolonged antibiotic therapy but the relapses are frequent.
CONCLUSIONS *H. cinaedi* is a rarely encountered pathogen in clinical practice, likely underdiagnosed due to prolonged culture times. Frequent relapses and the need for prolonged antibiotic therapy create a risk of antimicrobial resistance, representing a public health concern, especially in immunocompromised patients. In our case, distinction between hypocomplementemic vasculitis flare-ups and H. cinaedi bacteremia recurrence remains a clinical challenge that complicates therapeutic management.

Key words
bacteremia, febrile purpura, *Helicobacter cinaedi*, hypocomplementemic vasculitis

JANA MACÁKOVÁ

Various symptoms of peripheral arterial disease in a single patient: when the blood pressure increases and the weight decreases

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INTRODUCTION Peripheral arterial disease (PAD) is a broader term that involves multiple arterial systems, aside from the coronary arteries and aorta. It can have various manifestations and forms depending on the location and severity of symptoms.

CASE DESCRIPTION In this case report, we present a 72-year-old woman, in whom PAD was present in various arterial systems with different symptom severities, ranging from severe to asymptomatic states, and required intervention within a short period of time.

The first contact with the patient occurred through a consultation angiologic examination during her hospitalization in the internal medicine department for decompensated arterial hypertension (with a maximum blood pressure of 235/110 mm Hg). In our angiology outpatient clinic, renal artery examination was performed and showed significant to severe stenosis of the right renal artery.

The angiographic findings confirmed severe stenosis of the right renal artery and occlusion of the left renal artery and showed other forms of PAD affecting the lower limbs and aorta, including occlusion of the distal aorta (Leriche syndrome). Based on that, an endovascular procedure within the right renal artery was indicated and performed.

After hospitalization, the patient was once again examined during her visit in our clinic, this time for dyspeptic syndrome. Ultrasonography suggested severe stenosis of the superior mesenteric artery and celiac trunk. During the subsequent hospitalization and selective catheterization of the visceral arteries, we identified an occlusion of the superior mesenteric artery and confirmed severe stenosis of the celiac trunk, which were also indications for endovascular treatment.

Given the aforementioned findings, further examinations of other arterial systems was conducted and confirmed various forms of PAD. Symptoms were evaluated and the management strategy was determined.

CONCLUSIONS Due to complex polyvascular PAD and aorta involvement in this patient, several invasive treatments were necessary. She is currently asymptomatic and regularly monitored in our outpatient clinic to prevent or intervene in any severe progression of PAD and aortic involvement.

Key words
chronic mesenteric ischemia, Leriche syndrome, peripheral arterial disease, polyvascular arterial disease, renal artery stenosis

JOANNA KULA

Villous adenoma of the bile ducts: a long and winding road to make the diagnosis

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CASE DESCRIPTION Our study presents 3 different cases of patients who were admitted to the hospital between 2019 and 2025 to investigate biliary duct dilations. After several hospitalizations and biopsies during endoscopic retrograde cholangiopancreatography, the diagnosis of a rare condition, villous adenoma of the bile ducts with potential malignant transformation, was established.

Key words
biliary duct dilations, biliary stenting, endoscopic retrograde cholangiopancreatography, surgical treatment, villous adenoma of the bile ducts

JUHI DATWANI

Disseminated yet elusive: *Cryptococcus neoformans* with a negative antigen puzzle

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INTRODUCTION Cryptococcosis, caused by *Cryptococcus neoformans*, is a common opportunistic fungal infection in individuals with severe immunosuppression, particularly those with advanced HIV infection and CD4+ T lymphocyte count lower than 100 cells/μl. While cryptococcal meningitis is the most common manifestation, dissemination to less frequently involved sites, such as bone marrow and ocular structures, is rare. Cryptococcal antigen (CrAg) testing plays a pivotal role in diagnosis, but false-negative results due to the postzone phenomenon may lead to delays in treatment and subsequently, poor outcomes.

CASE DESCRIPTION A 26-year-old Venezuelan man, recently diagnosed with HIV (CD4+ T lymphocyte count, 21 cells/mm^3; viral load, 39 700 copies/ml), presented with a 4-month history of weight loss, night sweats, fever, asthenia, headaches, and lower limb weakness. Physical examination revealed cachexia, oral candidiasis, lymphadenopathies, and decreased muscle strength in lower limbs. Laboratory findings indicated pancytopenia and elevated inflammatory markers. Imaging studies showed lymphadenopathies, hepatosplenomegaly, splenic lesions, and spinal magnetic resonance imaging findings suggestive of transverse myelitis. A broad diagnostic workup for opportunistic infections was made, showing a negative CrAg test result. Due to pancytopenia, a bone marrow aspirate was performed, showing yeast-like organisms. Subsequently, fundoscopy identified bilateral papilledema, cerebrospinal fluid analysis confirmed cryptococcal meningitis with a positive CrAg test result, and encapsulated yeasts were seen on India ink stain. Finally, cerebrospinal fluid, blood, and bone marrow cultures grew *C. neoformans*. Due to the high degree of fungal burden, serum CrAg testing was repeated with positive findings upon serum dilution. Induction therapy with liposomal amphotericin B and fluconazole was initiated and extended for 4 weeks, achieving progressive clinical improvements.

DISCUSSION Disseminated cryptococcosis involving bone marrow and ocular structures is rare and is associated with poor prognosis. Ocular involvement, particularly choroiditis, can mimic other opportunistic infections such as tuberculosis, histoplasmosis, or toxoplasmosis, delaying diagnosis. Pancytopenia can be attributed to direct bone marrow infiltration by *C. neoformans*, which is reported in up to 42% of disseminated cases. Our patient also presented transverse myelitis, probably secondary to HIV infection; however, we cannot rule out cryptococcosis as a contributing factor, especially since the patient showed clinical improvement following antifungal therapy.

Regarding the diagnosis of meningitis and disseminated cryptococcosis in patients with HIV, CrAg testing exhibits excellent diagnostic performance. Given the patient's CD4+ T lymphocyte count, routine screening with serum CrAg was performed, initially yielding a negative result. This discrepant finding was attributed to the postzone phenomenon, in which an excess of antigen paradoxically prevents antibody-antigen crosslinking, leading to the absence of precipitation and a false-negative result. Given the pivotal role of CrAg testing in the diagnosis and early management of cryptococcosis, clinicians must be aware of potential false-negative results and recognize when further investigation is warranted.

CONCLUSIONS This case highlights that cryptococcosis can affect multiple organ systems. It also emphasizes that, while CrAg testing demonstrates excellent diagnostic performance, it is not without limitations. Recognizing such diagnostic pitfalls is essential for ensuring timely diagnosis, initiating appropriate antifungal treatment,

preventing complications, and optimizing patient outcomes and long-term recovery.

Key words
choroiditis, cryptococcosis, HIV, pancytopenia, postzone phenomenon

KARLIS KLEIMANIS

Uncharted waters: parathyroid hormone as a lighthouse in the diagnosis of dual primary tumors

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CASE DESCRIPTION The significance of this case lies in the rarity of synchronous dual malignancies. It sheds light on the importance of rapid decision-making to ensure appropriate treatment is started.

A 41-year-old man was hospitalized in the neurology department due to complaints of headaches and dizziness, instability, and double vision for 2 weeks.

While in hospital, the patient underwent brain, abdomen, and lung computed tomography (CT) as well as a brain magnetic resonance imaging (MRI). In a CT scan, a large suprarenal lesion was discovered, which was described as nonadenoma. The MRI findings showed multiple pathologically appearing nodules (along the lateral ventricles, ependymal and subependymal regions, in the third and fourth ventricles, as well as perineurally along the VII and VIII nerves and along the tentorium) described as possible primary central nervous system lymphoma or dissemination from a lesion in the adrenal gland, or granulomatous disease such as neurosarcoidosis. Additionally, a wide set of laboratory tests were done for infections, tumor markers, and antibodies. The patient underwent an endocrinology consultation, followed by comprehensive blood tests, including parathyroid hormone (PTH), cortisol, renin, and aldosterone, as well as calcium levels in both urine and plasma. Despite the extensive workup, results were largely unremarkable, providing no clear endocrine explanation for the findings and suggesting a lymphoproliferative origin of the disease due to mild PTH suppression. Laboratory test results showed mild leukocytosis, a small increase in the C-reactive protein level, and a high IgG level in cerebrospinal fluid. Cerebrospinal fluid contained white blood cells as well.

The patient was started on glucocorticoids, after which complaints of headaches decreased, and the patient was discharged in a stable general condition with a recommendation to consult an endocrinologist for additional blood tests. On the third day after being discharged, the patient returned to the hospital with neurologic deterioration, complaining of sleepiness, nausea, vomiting, and cognitive impairment. Upon neurological examination, the patient had worsening of eye movement, nystagmus, abducens nerve paresis, and right-sided hemiparesis. The patient complained of generalized fatigue. MRI was repeated and a significant disease progression was observed. For further strategy of treatment, the neuro-oncology multidisciplinary board decided to perform biopsy to identify a pathologic process.

Despite receiving symptomatic treatment, the general condition of the patient worsened and he died before the planned biopsy. Autopsy was performed, where the process of the brain was identified as diffuse large B-cell lymphoma, and the adrenal lesion was identified as pheochromocytoma without endocrine activity.

Symptoms of neurologic deficit led to expect a neurologic pathology, but following visual diagnostic scans showed an additional lesion in the adrenal gland that raised questions about a possible endocrine malignancy. Additional imaging and laboratory test results shifted the final diagnosis towards primary central nervous system malignancy and an adrenal lesion as a coexisting diagnosis. The initial treatment with corticosteroids had likely masked the rapid growth of the malignancy.

This case sheds light on important and time-sensitive actions to ensure a definite diagnosis and specific therapy. It highlights the importance of multidisciplinary team influence on the decision-making process.

Key words
pheochromocytoma, primary central nervous system tumor

KHADIDJA ABDELHAMID

Paraneoplastic lupus syndrome

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INTRODUCTION Associations between inflammatory diseases, systemic autoimmune diseases, and myelodysplastic syndromes have been reported for at least 25 years, both through isolated clinical cases and larger case series, suggesting a possible pathogenic relationship.

CASE DESCRIPTION We report a case of a 62-year-old woman followed for atypical chronic myelomonocytic leukemia diagnosed in August 2022 based on persistent monocytosis, thrombocytopenia, polyclonal hypergammaglobulinemia, molecular biology data (*NRAS* and *SCMA1* mutations), and flow cytometry data (46 chromosomes with deletion of the long arm of 16).

In August 2024, the patient was hospitalized for deterioration of general condition and dyspnea. Examination showed hyperleukocytosis at 206 G/l, progression of monocytosis, myelemia, and bilateral pericardial and pleural effusions with infracentimetric mediastinal-hilar lymph nodes on computed tomography. Positron emission tomography showed osteomedullary hypermetabolism and splenomegaly. A first course of azacitidine and venetoclax treatment was indicated.

A few weeks later, the patient developed ST-elevation myocardial infarction, ischemic stroke, and deep vein thrombosis, requiring hospitalization at the intensive care unit and treatment with dual antiplatelet therapy and anticoagulation. Immunological assessment showed the presence of anti-nuclear antibodies at 1/1280, anti-Ro52 anti-Sm/RNP complex, and anti-U1 RNP antibodies. Conventional antiphospholipid antibodies were negative.

Given this presentation, the diagnosis of systemic lupus according to ACR/EULAR 2019 criteria was made, and hydroxychloroquine treatment was initiated. In the following days, hyperleukocytosis worsened to 100 G/l, necessitating initiation of hydroxyurea treatment, followed by azacitidine and ruxolitinib treatment by hematologists, which led to clinical and biological improvement. The patient is awaiting allograft in mid-March 2025.

DISCUSSION AND CONCLUSIONS Autoimmune or inflammatory manifestations are observed in 10% to 30% of patients with myelodysplastic syndromes or chronic myelomonocytic leukemia with a broad clinical spectrum. In the case of our patient, the clinical picture associated with immunologic abnormalities and atypical myelodysplastic/myeloproliferative syndrome suggests a lupus-like syndrome rather than true lupus.

Key words
lupus syndrome, paraneoplastic

KRISZTIÁN BIRTALAN

What kind of celiac disease is this? Refractory celiac disease

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INTRODUCTION Celiac disease affects approximately 1% of the Hungarian population. An immune response to gluten leads to villous atrophy in the small intestine, which typically resolves with

a strict gluten-free diet. In refractory celiac disease, intraepithelial lymphocytes (IELs) remain active despite adherence to a gluten-free diet, necessitating immunosuppressive therapy.

CASE DESCRIPTION A 30-year-old woman was diagnosed with celiac disease in 2020 (MARSH 3B villous atrophy and positive serology results). Her condition improved with a gluten-free diet. In December 2023, she was hospitalized due to abdominal pain, significant weight loss, and persistent diarrhea. Colonoscopy and stool cultures did not reveal any etiology, and celiac serology yielded negative results. Laboratory tests showed elevated amylase, lipase, and suppressed thyroid-stimulating hormone levels, leading to diagnoses of pancreatitis and hyperthyroidism. Treatment was initiated and the patient was discharged.

However, her diarrhea persisted and her weight continued to drop. She was admitted to our department with a body weight of approximately 37 kg (height, 170 cm; body mass index, 12.8 kg/m^2). Laboratory test results showed normocytic anemia, elevated liver enzymes, hypoproteinemia, and nephrotic-range proteinuria (ca, 3 g/d). Duodenal biopsy demonstrated total villous atrophy (MARSH 3C), and renal biopsy confirmed IgA nephropathy. Repeat celiac serology remained negative. Suspecting refractory celiac disease, further analysis of the intestinal biopsy specimen for IELs showed no abnormal phenotype or *TCR* gene rearrangement, leading to a diagnosis of type 1 refractory celiac disease. Treatment with budesonide resulted in the disappearance of IELs, restoration of villous structure, weight gain, and resolution of proteinuria.

CONCLUSIONS In type 1 refractory celiac disease, IELs remain active without gluten exposure but do not display abnormal immunophenotypes or *TCR* gene rearrangements. In type 2 refractory celiac disease, IELs exhibit abnormal phenotypes and/or monoclonal *TCR* gene rearrangements, requiring more intensive immunosuppression. The most severe form of the disease occurs when IELs become malignant, leading to enteropathy-associated T-cell lymphoma, which necessitates hematological/oncological treatment. Extraintestinal manifestations of celiac disease include hepatitis, pancreatitis, and IgA nephropathy, all of which were present in this case. In patients with a confirmed history of celiac disease and negative serology results, the reappearance of villous atrophy with IEL infiltration should prompt consideration of refractory celiac disease.

Key words
celiac disease, malabsorption, refractory celiac disease

KRZYSZTOF PROC

Eosinophilic granulomatosis with polyangiitis with involvement of the lungs and heart

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CASE DESCRIPTION A 19-year-old woman with Marfan syndrome, after surgical treatment of chest and spinal deformities, was admitted to the internal medicine ward due to general signs of infection. Pneumonia was diagnosed due to high levels of inflammatory markers: leukocytosis of 24.16 × 10^3/µl (reference range [RR], 4–10 × 10^3/µl) with eosinophilia of 0.93 × 10^3/µl (RR, 0.04–0.4 × 10^3/µl), an erythrocyte sedimentation rate of 69 mm/h (RR <12 mm/h), and a C-reactive protein level of 79.34 mg/l (RR <5 mg/l), along with inflammatory changes on chest X-ray. Due to a lack of clinical improvement after the administration of 2 antibiotics, a computed tomography scan of the chest was performed, which showed lesions consistent with eosinophilic granulomatosis with polyangiitis (EGPA).

The patient was transferred to the rheumatology department for further diagnostic workup. Attention was drawn to worsening exercise tolerance, persistent tachycardia of about 110 beats per minute, and high N-terminal pro–B-type natriuretic peptide (NT-proBNP) levels of 11 600 pg/ml (RR, 0–125 pg/ml). A transthoracic echocardiogram showed impaired left ventricular function down to 29%. Cardiovascular magnetic resonance showed signs of cardiomyopathy and features of nonischemic myocardial damage that may correspond with changes occurring in EGPA. Based on the conducted diagnostic workup, it was established that the patient met the criteria for EGPA diagnosis according to ACR/EULAR 2022. In line with treatment recommendations, rituximab and high doses of intravenous steroids were administered.

After 3 months of treatment, the previously reported symptoms resolved. Follow-up tests showed no abnormalities. There was a significant decrease in NT-proBNP levels to 332.7 pg/ml. Follow-up computed tomography of the lungs showed a significant regression of the previously described lesions. Cardiovascular magnetic resonance showed a reduction in ventricles' volumes with an improvement in left ventricular ejection fraction.

DISCUSSION In the course of EGPA, the involvement of the respiratory system, heart, gastrointestinal tract, kidneys, and nervous system may occur. In young patients, due to high compensatory capabilities, progressive heart failure may present asymptomatically or with few symptoms. Therefore, it is advisable to conduct screening imaging and laboratory testing to assess the cardiovascular system.

CONCLUSIONS In patients with symptoms of respiratory infection unresponsive to antibacterial treatment, it is necessary to broaden the diagnostic approach to include vasculitis. Patients diagnosed with vasculitis require extensive organ evaluation, even in the absence of characteristic symptoms. Diagnostic workup and treatment should be conducted in experienced rheumatology centers.

Key words
eosinophilic granulomatosis with polyangiitis, heart failure, heart failure with reduced ejection fraction

LOUISE WARD

A patient with acute ischemic stroke due to carotid web presenting to a Model 3 hospital

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INTRODUCTION Stroke is an emergency with increasing prevalence across Ireland. FAST calls are offered as part of hyperacute stroke care in 27 hospitals across Ireland. Acute assessment is mostly completed by nonconsultant hospital doctors, and acute management is led by a general physician without specific stroke training in 37% of cases. Carotid web is a nonatheromatous and nondissecting membrane strand protruding into the lumen of the carotid artery. They are an under-recognized cause of recurrent acute ischemic stroke (AIS), particularly in younger women. This case describes a young patient presenting with a major stroke to a Model 3 hospital; outlining the importance of acute stroke management for all clinicians on a medical roster and an under-recognized and treatable cause of stroke.

CASE DESCRIPTION An emergency department in a Model 3 hospital received a pre-alert about a 49-year-old woman with wake-up symptoms of left hemiparesis. On arrival at 10 AM, her National Institutes of Health Stroke Scale (NIHSS) score was 14 points—left hemiplegia, facial droop, and neglect. She was last seen well at 11.30 PM the previous night. She had no medical history/medications apart from

smoking. Brain computed tomography and angiography showed right M1 occlusion. As she had wake-up symptoms, she was not thrombolyzed. She was transferred to a thrombectomy center. On arrival, her NIHSS score was 18, with profound visual and sensory deficits. She had a successful thrombectomy with an immediate improvement to a NIHSS score of 6. During the thrombectomy, a significant carotid web was noted at the bifurcation of the right internal carotid artery. On day 1 post endovascular thrombectomy, she had a brain computed tomography scan showing an acute right middle cerebral artery territory infarct. She had ongoing speech and sensory deficits with neglect. On day 3 post endovascular thrombectomy, her NIHSS score improved to 2, with mild motor weakness on the left side and she was transferred back to the Model 3 referring hospital. While the carotid web was recognized as being the likely etiology of her stroke, a full stroke workup was performed. She was discharged home 1 week post stroke with a NIHSS score of 0. She subsequently received definitive treatment with internal carotid artery stenting.

DISCUSSION This case highlights the under-recognized cause of AIS secondary to carotid web, which has a high risk of stroke recurrence. It also highlights the importance of acute stroke care provided by nonstroke physicians—particularly in Model 3 hospitals. In this case, the stroke was recognized, assessed, and treated quickly. This patient had a remarkable recovery and was home within a week of a potentially devastating and life-changing stroke without any deficits. Treatment options for carotid webs are being debated in the literature but it is important for general physicians to be aware of conditions such as carotid webs and to refer patients to appropriate centers for definitive management.

CONCLUSIONS Carotid webs are an under-recognized cause of AIS in young female patients, who need quick assessment and has the potential for complete full recovery.

Key words
carotid web, Model 3 hospital, stroke

LUCIA MIHAĽOVOVÁ

What was hidden behind severe hypercalcemia?

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INTRODUCTION Sarcoidosis is a chronic disease leading to an abundant immune response forming noncaseating nodules known as granulomas. Approximately 90% of primary cases of sarcoidosis involve the pulmonary system, with only 10% of cases presenting with primary extrapulmonary involvement. The condition predominantly affects women around 40 years of age. The goal of this case report is to bring a closer look on the possible correlation between severe hypercalcemia and the manifestation of extrapulmonary sarcoidosis.

CASE DESCRIPTION In this study, we report a case of a 53-year-old woman with type 2 diabetes, celiac disease, and hypertension. The patient presented with nonspecific symptoms, such as general weakness, fatigue, vomiting, constipation, and a significant weight loss of 21 kg in 5 months. The patient was referred to our clinic by a general practitioner for uncontrolled severe hypercalcemia. The patient's history regarding potential adverse effects of hypercalcemia was negative. Upon the patient's admission to our clinic, we noted massive splenomegaly extending into the left iliac crest area, as well as anemic complexion.

Basic laboratory test results showed severe hypercalcemia with suppressed parathyroid hormone, as well as elevation of renal markers, hypokalemia, and mild microcytic anemia. In further laboratory tests, a low rate of angiotensin-converting enzyme along with positivity of light chains (κ, λ) were identified. Computed tomography (CT) of the abdomen and pelvis, positron emission tomography/CT, and CT-guided splenic biopsy performed in October 2024 prior to hospitalization showed multifocal pathological noncaseating granulomas in the liver and spleen, indicating nearly complete infiltration of the spleen parenchyma, suggestive of tumor-related hepatosplenomegaly, with retroperitoneal and partially intraperitoneal lymphadenopathy. Initially, the clinical presentation and examination results suggested a lymphoproliferative disorder. However, the results of immunophenotyping of peripheral blood, genetic testing, microscopic examination of the bone marrow, and trephine biopsy were negative for hematologic malignancy. Serum immunofixation demonstrated a reactive pattern of oligoclonal immunoglobulin and free light chains. In light of the hypercalcemia and noncaseating granulomas, we conducted an extensive differential diagnosis to identify a potential underlying disease and organ involvement including microbiologic, immunologic, and infectious causes. Mycobacterial infection was also considered. We performed pulmonary, gynecological, ophthalmological, endocrinological, and endoscopic examinations. Findings included nephrolithiasis, chorioretinal lesions in the left eye and an eufunctional, bilateral, nodular goiter with suspected papillary thyroid carcinoma (Bethesda V). Hyperparathyroidism was excluded on endocrinological evaluation.

Given the above, we veered toward a diagnosis of an extrapulmonary, atypical form of sarcoidosis involving the liver, spleen, and abdominal lymph nodes. After initiating corticosteroid therapy, follow-up abdominal CT performed 6 weeks later demonstrated reduction in the spleen and lymph node size, stabilization of granulomatous lesions, and stabilization of calcium levels in laboratory findings.

CONCLUSIONS Extrapulmonary sarcoidosis presents with a diverse array of symptoms. Serum angiotensin-converting enzyme levels are typically elevated due to granulomatous inflammation. We interpret its decreased level as indicative of the extrapulmonary form of sarcoidosis. An early recognition and appropriate management of sarcoidosis are crucial to prevent complications and improve outcomes. In our case report, a reduction in the size of granulomatous lesions, lymph nodes, along with stabilization of calcium levels, indicates an effective management of the condition

Key words
hypercalcemia, lymph nodes, noncaseating granulomas, sarcoidosis, splenomegaly

MAGDALENA PŁONKA-STĘPIEŃ

Differential diagnosis in diabetes can be difficult: the first case of digenic maturity-onset diabetes of the young with compound heterozygosity of severe mutations in the *PDX1* and *HNF1B* genes

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INTRODUCTION Maturity-onset diabetes of the young (MODY) is the most common form of monogenic diabetes. The most frequent types of MODY are the *HNF1A*-MODY, *GCK*-MODY, and *HNF4A*-MODY. In some extremely rare cases, genetic testing reveals more than 1 mutation. So far, only a few cases of digenic MODY have been reported.

CASE DESCRIPTION A 23-year-old woman with an 8-year history of diabetes initially diagnosed as type 1 was referred to the University Hospital in Kraków for differential diagnosis. Soon after diagnosis, as

type 1 diabetes–related autoantibodies were negative, the patient underwent genetic testing of the most frequent MODY genes by Sanger sequencing. It showed a sequence difference in the *HNF1A* gene: c.293C >T, p.Ala98Val, a nonpathogenic variant. In family history, the patient's father had prediabetes. Her paternal grandfather was also diagnosed with diabetes. Her normoglycemic mother had been diagnosed with renal cysts. We proposed supplementary genetic testing using next-generation sequencing for the patient. It demonstrated 3 heterozygous variants in the MODY genes. Two of them were pathogenic, resulting in an early termination codon: 1) c.697G >T, p.Glu233Ter in the PDX1 gene, 2) c.827_828insT, p.Gly277ArgfsTer17 in the *HNF1B* gene. The patient was initially treated with a sulfonylurea compound and DPP-4 inhibitor. However, she was not able to achieve satisfactory postprandial glucose control. She was switched back to intensive insulin therapy, on which she remains normoglycemic. The genetic testing of the family was implemented. The patient's father was a heterozygous carrier of the variant c.697G >T in the *PDX1* gene, while the pathogenic mutation in the *HNF1B* gene was not found. The mother and brother of the patient had a heterozygous variant c.827_828insT in the *HNF1B* gene but no variants in the *PDX1* gene.

DISCUSSION We present the first case of digenic MODY due to severe pathogenic variants in the *PDX1* and *HNF1B* genes, which are transcription factors responsible for pancreas development and insulin secretion. The c.697G >T, p.Glu233Ter *PDX1* mutation is of a nonsense type; the pathogenicity of this variation is associated with protein shortening. The c.694G >A, p.Gly232Ser *PDX1* mutation is of a missense type of uncertain significance. Shortening a protein caused by the c.697G >T, p.Glu233Ter mutation will likely severely impact its function. The c.827_828insT (Gly277ArgfsTer17) *HNF1B* mutation is associated with a shift in the reading frame and the formation of a precession translation termination codon. The transcript is most likely targeted for degradation in the nonsense-mediated mRNA decay pathway and the protein is not formed. Developmental renal disease, called renal cysts and diabetes, is characteristic of patients with *HNF1B* mutations. Despite the severity of both *PDX1* and *HNF1B* mutations, neither the mother nor the father has developed diabetes so far. This points to the nonpenetrance of some heterozygotic MODY-related mutations in both genes.

CONCLUSIONS Differential diagnosis in patients with diabetes due to double MODY mutations seems to be particularly difficult. Oral hypoglycemic treatment may not be effective in these patients. Next-generation sequencing may identify more such patients in the future.

Key words
HNF1B-MODY, digenic MODY, next-generation sequencing, MODY, *PDX1*-MODY

MARIA JOÃO A. BARBOSA

Connecting the dots: a case of scleroderma-polymyositis overlap syndrome

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INTRODUCTION Systemic sclerosis (SSc) is a heterogeneous condition that can coexist with other autoimmune disorders, such as polymyositis/dermatomyositis (PM/DM). We present a case of SSc–PM/DM overlap syndrome.

CASE DESCRIPTION We present a case of a 75-year-old man, who reported fatigue, weakness of the shoulder and pelvic girdle, dysphagia to liquids and solids, weight loss of 18 kg, bilateral gonalgia and elbow pain, hair loss, and Raynaud phenomenon in the hands. Prior to admission, the patient presented to the emergency room after an episode of syncope. On physical examination, he had hoarseness, sclerodactyly, thickening of the skin on the face and forearms, telangiectasis and redness of the face, desquamative lesions of the scalp, outer ear, shoulders, elbows, buttocks, and anterior face of the legs, and proximal tetraparesis with muscle atrophy. Laboratory workup showed an elevated erythrocyte sedimentation rate, elevated lactate dehydrogenase and creatine kinase levels, with positive antinuclear antibody, anti-PM/Scl100, anti-PM/Scl75, and Ro52KD and borderline Th/To antibodies. Electroneuromyography confirmed myopathy of the proximal muscles of the arm and leg. Cardiac magnetic resonance imaging showed no signs of myocardial inflammation or fibrosis, and the transthoracic echocardiogram was normal. The muscle biopsy was still underway. Computed tomography of the thorax, abdomen, and pelvis showed an adrenal mass with borderline washout indeterminate for adenoma/nonadenoma. As the clinical presentation and results of complementary examinations fulfilled the criteria for SSc–PM/DM overlap syndrome with visceral involvement, adequate treatment was initiated and clinical improvement was observed. The patient was discharged and remained in close follow-up.

DISCUSSION The classification criteria for SSc are well defined in the ACR/EULAR guidelines, and 1 criterion alone is sufficient for diagnosis (skin thickening of the fingers extending to the metacarpophalangeal joints), which was observed in our patient. Given his clinical features, as well as the antibodies found and the electroneuromyography results, our patient best fitted the diagnosis of SSc–PM/DM with multiple organ involvement. There are no specific guidelines for the treatment strategy. Still, early diagnosis and treatment should be our primary concern. These can alter the prognosis, as these patients have a worse prognosis, specifically when visceral involvement is identified.

CONCLUSIONS SSc–PM/DM overlap syndrome has a different presentation and prognosis, and this case shows the importance of recognizing patterns of disease, leading to workup, diagnosis, and starting therapy as soon as possible.

Key words
dysphagia, muscle weakness sclerosis, myositis, weight loss

MARIA KOMISARZ-CALIK

Severe thromboembolic sequelae after an effective treatment of hypercortisolemia in a patient with ACTH-independent Cushing syndrome

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INTRODUCTION Cushing syndrome (CS) is a rare disease but it is associated with an 18-fold increased risk of venous thromboembolism (VTE). This case report describes a patient with bilateral adrenal tumors and adrenocorticotropic hormone (ACTH)-independent CS, who experienced ischemic stroke in the left brain hemisphere after adrenalectomy.

CASE DESCRIPTION A 54-year-old woman with severe osteoporosis and arterial hypertension, who had previously undergone endovascular treatment of a giant left internal carotid artery aneurysm complicated by ischemic stroke in the left brain hemisphere, was admitted to the endocrinology department due to suspected CS. She presented with a typical cushingoid silhouette, plethora, and multiple ecchymoses. The rigid circadian cortisol rhythm, lack of suppression on the 1-mg dexamethasone test, low ACTH concentration, and high late-night saliva cortisol confirmed the diagnosis of CS. On abdominal computed tomography, numerous nodular lesions with a high absolute washout coefficient suggestive of benign adenomas were found. Metyrapone was implemented, and later the patient was qualified for adrenalectomy, despite her

multimorbidity and high cardiovascular risk. Due to the increased thromboembolic risk (after the re-embolization of the left internal carotid artery aneurysm), the patient received acetylsalicylic acid (ASA) at a dose of 150 mg/day and a prophylactic dose of low-molecular-weight heparin (LMWH).

After adrenalectomy, the patient experienced weakening of the muscle strength of the right upper limb, dropping objects from the hand, and dysarthria. A computed tomography scan of the head showed an area of complete ischemia of the frontal-parietal border of the left middle cerebral artery and a suspected aneurysm of the division of the right middle cerebral artery. Ischemic stroke in the left hemisphere of the brain was diagnosed. ASA treatment was commenced as secondary stroke prevention. Moreover, LMWH was continued for 3 months due to the elevated thromboembolic risk associated with CS.

Three months after surgery, recurrence of the hypercortisolemia symptoms was noted and osilodrostat was started. The patients received biochemical control and the CS symptoms were alleviated.

DISCUSSION Thromboprophylaxis may decrease the incidence of postoperative VTE in patients with CS. Literature data show that the risk of VTE associated with surgery in patients with CS is lower than that associated with hip surgery. However, ACTH-dependent and ACTH-independent CS similarly elevate the incidence of VTE during the preoperative period.

Furthermore, a sudden decrease in cortisol levels following the administration of cortisol-lowering medications or a successful surgical intervention may heighten the risk of VTE. LMWH seems equally effective in patients with CS and the general population and remains the base of thromboprophylaxis in this patient population. On the other hand, there have been reported cases of VTE episodes in patients with CS who received LMWH.

CONCLUSIONS CS is associated with a significantly increased VTE risk. An effective treatment of hypercortisolemia is associated with an increased risk of VTE episodes. However, clear guidelines regarding VTE treatment and prophylaxis in patients with CS are still missing.

Key words
adrenalectomy, Cushing syndrome, thromboembolic risk

MARIA KRÓLIKOWSKA

Can car keys break your heart?

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INTRODUCTION It is rumored that stress can kill you, but we used to think about it as a truism and triviality. However, this case report shows us that it is deeply true and more frequent than we can imagine.

CASE DESCRIPTION This case report refers to a 66-year-old man, smoker, with arterial hypertension and no other diseases. That morning, he had a heated argument with his wife, later he was visiting his dying friend in a hospital, and on top of all above, after coming back to his car in the rain, he discovered that car keys are locked inside. He was found after syncope by a passer-by in the hospital parking lot, in bad condition, presenting with sudden chest pain and shortness of breath. The patient was immediately transported to the emergency department.

Physical examination showed bradycardia with a heart rate of 30 bpm and hypotension with blood pressure of 90/60 mm Hg. Several minutes later, electrocardiography demonstrated a sinus rhythm of 80 bpm with left bundle branch block. Transthoracic echocardiography showed an extensive akinesis of the left ventricle (LV) with apical "ballooning" and systolic anterior motion of the mitral leaflet. This induced severe left ventricle outflow tract obstruction (LVOTO) with a peak gradient of 128 mm Hg and LV ejection fraction of about 30%. Coronary angiography demonstrated no hemodynamically relevant lesions of coronary arteries; however, during the procedure, a third-degree atrioventricular block requiring ventricular pacing occurred. Levels of creatine kinase and troponin were slightly elevated.

Taking all above clinical data into consideration, takotsubo syndrome (TTS) with LVOTO appeared to be the most plausible diagnosis. Intravenous fluids and metoprolol have been administered. After 7 days, LVOTO was resolving, with a peak gradient decreasing to 10 mm Hg and complete regression of apical "ballooning." The preliminary diagnosis of TTS was confirmed. Today, 5 years after the incident, the patient is in good general condition and receives treatment in a cardiology clinic.

DISCUSSION Takotsubo syndrome, also known as "broken heart syndrome," is a type of acute, reversible myocardial dysfunction first described in Japanese women in 1990s. It is characterized by sudden, transient LV dysfunction, and its clinical presentation resembles typical acute coronary syndrome with chest pain, electrocardiogram abnormalities, and increased levels of cardiac injury biomarkers. However, angiography shows no relevant coronary artery obstruction. The LV contraction abnormality extends beyond the typical territory supplied by a single epicardial artery. The syndrome predominantly affects postmenopausal women and is usually preceded by a strong physical or emotional stress. LVOTO may be an additional and life-threatening complication leading to cardiogenic shock. Early identification of this complication indicates appropriate therapy with β-blockers and fluids, avoiding adrenergic drugs.

CONCLUSIONS TTS has been long regarded as a benign condition; however, recent research showed the rate of complications and mortality among these patients similar to that of patients with coronary artery obstruction. The onset of LVOTO and cardiogenic shock is a challenging complication of an acute TTS phase with prevalence between 7% and 25%. It is crucial and very challenging to distinguish between ischemia and TTS.

Key words
broken heart syndrome, cardiogenic shock, left ventricular outflow tract obstruction, systolic anterior motion, takotsubo syndrome

MARIA MOSQUEIRA

A portal to the unknown: a unique case of portal hypertension without cirrhosis

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INTRODUCTION Noncirrhotic portal hypertension (NCPH) is a rare condition characterized by portal hypertension in the absence of cirrhosis. It is often associated with systemic diseases, hematologic disorders, immune-related causes, or exposure to drugs and toxins.

CASE DESCRIPTION We present a case of a 74-year-old woman with gastrointestinal bleeding due to large esophageal varices. Imaging showed no signs of cirrhosis or vascular thrombosis, and liver biopsy ruled out the presence of cirrhosis as well as porto-sinusoidal vascular disease–specific histologic patterns. She was diagnosed with non-NCPH, prompting a comprehensive etiological workup, which identified 2 monoclonal components: immunoglobulin (Ig)A-λ and IgG-λ. Bone marrow biopsy demonstrated plasma cell dyscrasia with 24% abnormal plasma cells. However, the patient did not meet the International Myeloma Working Group criteria for multiple myeloma, and further studies ruled out associated amyloidosis. Consequently, a diagnosis of smoldering myeloma with biclonal gammopathy, associated with NCPH, was established.

DISCUSSION NCPH is a complex and often misdiagnosed condition due to its low prevalence, diverse etiologies, and clinical overlap with cirrhosis. The Vascular Liver Disorders Interest Group recently defined porto-sinusoidal vascular disease as a subset of NCPH with distinct histopathological features, none of which were present in our patient. The extensive workup excluded common causes, suggesting a potential link between NCPH and plasma cell dyscrasia. While prior reports have associated monoclonal gammopathy with NCPH, cases involving multiple myeloma typically exhibit hepatic plasma cell infiltration, which was absent in this case. The presence of a biclonal component further distinguishes this case. This emphasizes the importance of a comprehensive diagnostic approach and interdisciplinary collaboration in managing NCPH and recognizing hematologic disorders as potential contributors.

CONCLUSIONS This case highlights a rare association between biclonal smoldering myeloma and NCPH, expanding the spectrum of plasma cell disorders. It emphasizes the need for a systematic, multidisciplinary approach to enhance diagnosis, clarify pathogenesis, and develop personalized treatment strategies to improve patient outcomes.

Key words
monoclonal gammopathy, noncirrhotic portal hypertension, smoldering myeloma

MARTA PERSANA

When a family argument goes wrong

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INTRODUCTION McCune–Albright syndrome (MAS) is a rare genetic disorder resulting from somatic variants of the *GNAS* gene. MAS is most commonly diagnosed at a young age and may present with varying clinical manifestations. This case report presents a patient with a characteristic finding of MAS first diagnosed at an atypical age, which posed challenges for diagnosis and treatment strategies.

CASE DESCRIPTION A 71-year-old man was admitted after sustaining head concussion during a domestic dispute. Upon admission, general examination showed that he was short-statured and underweight and exhibited skeletal deformities and facial bone asymmetry. A head computed tomography scan indicated "lytic" and "sclerotic" bone changes, while a chest radiograph showed a deformed thorax, raising an initial suspicion for Paget disease. Abdominal computed tomography demonstrated a 2-cm intraductal papillary mucinous neoplasm in the head of the pancreas. Additionally, bone scintigraphy identified multiple pathologic areas with high metabolic activity. Paget disease was ruled out, as serum alkaline phosphatase levels were found repeatedly normal. Bone fibrous dysplasia was suspected based on chest computed tomography. Blood testing demonstrated subclinical hyperthyroidism and a multinodular goiter was found on ultrasonography of the thyroid gland. Based on the patient's physical characteristics, investigation results, and diagnostic criteria from the literature, he was diagnosed with MAS. Following the diagnosis, the patient began antithyroid treatment.

DISCUSSION MAS is a rare genetic disorder resulting from somatic variants of the *GNAS* gene, specifically in the cAMP-regulating protein, Gs alpha. MAS is typically diagnosed clinically. The clinical presentation of MAS may differ among patients. Fibrous dysplasia (FD) of bones is the most prevalent feature of MAS. MAS can be diagnosed based on the characteristic presence of FD in bones along with 1 or more characteristics of hyperfunctioning endocrinopathy (eg, precocious puberty, hyperthyroidism, growth hormone excess, or endogenous Cushing syndrome) and/or café-au-lait macules. Physical examination can show distinct skeletal deformities. The skull bones are the most commonly and earliest affected sites in FD. Most skeletal lesions in patients with MAS occur during the first 10 years of life. Patients with MAS often have concurrent intraductal papillary mucinous neoplasm and an increased risk of malignant transformation. Treatment is based on the MAS features of each patient. In most cases, the primary approach is to provide symptomatic management and correct endocrine disorders.

CONCLUSIONS The clinical presentation and treatment of MAS vary widely due to genetic mosaicism. While MAS typically begins in early childhood, it can also be diagnosed later in life. A timely diagnosis and ongoing monitoring are essential for effective management and to prevent complications associated with the disease. Further research is needed to determine the most effective treatment for patients with MAS.

Key words
fibrous dysplasia, hyperthyroidism, McCune–Albright syndrome

MARTA SKOCZYŃSKA

TB or not TB: the clinical context must give us pause

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CASE DESCRIPTION We present a nearly fatal case of systemic mycobacteriosis in a 73-year-old woman treated with a tumor necrosis factor-α inhibitor (adalimumab) for rheumatoid arthritis, initially misdiagnosed as metastatic lung cancer.

The patient was admitted for a routine visit after 3 months of therapy with adalimumab. On admission, she reported a 10-kg weight loss and general weakness following initial improvement in arthritis while on adalimumab treatment. On physical examination, general cachexia was noted. Laboratory workup showed markedly elevated inflammatory parameters. Over the course of hospitalization, the patient's general condition gradually deteriorated. A slight improvement was observed after introducing levofloxacin for presumed pneumonia. Contrast-enhanced computed tomography of the chest, abdomen, and pelvis demonstrated numerous enlarged pathological mediastinal lymph nodes with signs of disintegration, forming conglomerates, and features of lymphangitis carcinomatosa in the lungs, as well as pathological portocaval lymph nodes and uncountable small metastatic outbreaks in the liver and spleen. The radiological diagnosis was metastatic cancer, possibly originating in the lung. However, due to positive QuantiFERONTB Gold Plus test results and a temporal relationship between the onset of symptoms and adalimumab therapy initiation, tuberculosis was considered as an alternative diagnosis. Blood and urine polymerase chain reaction tests for *Mycobacterium tuberculosis* were negative. Due to the patient's generally poor condition, she was disqualified from bronchoscopy and other invasive tests for lung cancer. She was discharged on antimycobacterial treatment, and atypical mycobacteriosis was confirmed *ex juvantibus*, following the patient's recovery and regression of disseminated changes on follow-up computed tomography. The patient's rheumatoid arthritis has currently been satisfactorily managed with low-dose prednisone and leflunomide.

This case report emphasizes that mycobacteriosis is a great mimicker and can present both clinically and radiologically very similarly to metastatic cancer. Taking a pause to consider mycobacterial infection probability in the clinical context is crucial in the evaluation of disseminated lesions in immunocompromised patients.

Key words
adalimumab, lung neoplasms, mycobacteriosis

MARTÍN LEONARDO GIL FOLGAR

A tanned vampire

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INTRODUCTION Porphyria cutanea tarda (PCT) is recognized as the most prevalent form of porphyria. The relationship between PCT and HIV infection remains inadequately defined in the literature. In this report, we present a case of a patient in whom PCT was diagnosed concurrently with HIV infection.

CASE DESCRIPTION A 43-year-old woman with a smoking history and untreated atrial fibrillation presented with a 6-month history of asthenia, significant weight loss, abdominal pain, and generalized pruritus. On examination, the patient was in poor general condition and had facial and upper trunk hyperpigmentation.

Laboratory tests showed normocytic, normochromic anemia (hematocrit, 30.9%; hemoglobin, 9.9 g/dl), elevated liver enzymes (aspartate aminotransferase, 91 UI/l; alanine aminotransferase, 76 UI/l) amylase level of 50 UI/l, a normal ionized calcium level of 1.16 mmol/l, C-reactive protein of 6.6 mg/l, and erythrocyte sedimentation rate of 78 mm/h. Ferritin levels were 1247 ng/ml. A rapid HIV test was reactive, showing a CD4 count of 38 cells/mm^3 and a viral load of 157 000 copies/ml. A fasting plasma cortisol level was measured at 12.4 µg/dl, with an adrenocorticotropic hormone level of 14 pg/ml, effectively ruling out adrenal insufficiency. Computed tomography of the chest, abdomen, and pelvis was conducted and yielded normal results. Serial sputum tests for acid-fast bacilli were performed with negative results.

A more detailed history revealed a family history of porphyria. PCT was suspected, although it was recognized that the patient's abdominal pain was not typical of this type of porphyria. The dermatology department performed a Wood lamp urine test, which yielded negative results. Blood and urine samples were forwarded to another center. A total porphyrin study indicated 3165 µg/24 h (reference range, 20–250 µg/24 h), and porphobilinogen in urine testing was negative. Chromatography of esterified porphyrins demonstrated the following composition: copro, 10%; penta, 5%; hexa, 5%; fyria, 40%; and uro, 40%. The fluorescence spectrum plasma porphyrin test showed a value of 3.88 (wavelength, 619 nm; reference range $<$1.3 nm). Diagnosis of PCT was established, and antiretroviral treatment was initiated with good tolerance.

DISCUSSION The relationship between HIV infection and PCT remains inadequately established in the medical literature. Notably, the average level of coproporphyrin has been shown to be elevated in patients who are positive for hepatitis C virus, with even higher levels observed in individuals with coinfections involving both HIV and hepatitis C virus infections. In the majority of cases, the onset of PCT occurs in the advanced stages of HIV infection, particularly in patients with CD4 counts below 200 cells/mm^3 and viral loads greater than 30 000 copies/ml.

CONCLUSIONS Hyperpigmentation can often be the initial manifestation of an underlying systemic condition. Porphyrias themselves are rare, which presents a notable challenge in terms of diagnosis; indeed, the suspicion of these disorders is crucial for their timely identification. It is well recognized that there exists a correlation between porphyria and European folklore surrounding vampirism. The patient in this case was a member of night security personnel at an airport.

Key words

constitutional syndrome, HIV, hyperpigmentation, porphyria cutanea tarda

MARTIN PEHR

Osteopenia and hypercalcemia as a presentation of ectopic parathyroid adenoma

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INTRODUCTION Osteoporosis is highly prevalent among postmenopausal women, so underlying conditions may sometimes remain undiagnosed for an extended period of time. Even upon confirmation of hypercalcemia and elevated parathyroid hormone (PTH) levels, the path to correct diagnosis and localization of the parathyroid body might be challenging.

CASE DESCRIPTION A 57-year-old woman was referred to the endocrinology center with a 3-year history of intermittent hypercalcemia (peak calcium level, 2.97 mmol/l), cramps in lower extremities, osteopenia (T-score, –1.1), and elevated osteoresorption markers. The patient had a slight but persistent elevation of the PTH level at 7.46 pmol/l, low 25-hydroxyvitamin D level (25.9 ng/l), a hypoechogenic nodule in the left lobe of the thyroid gland, and a negative MIBI scan (sestamibi nuclear imaging of parathyroid tissue).

An expert ultrasonography of the neck identified a suspicious thyroid cystoid, but aspiration biopsy was negative for parathyroid tissue or PTH. Positron emission tomography / computed tomography showed glucose avid tissue 24 mm $\times$ 12 mm $\times$ 60 mm in the mediastinum adjacent to the esophageal wall, and repeated MIBI scans showed positivity within the same spot. Esophageal endosonography was ordered for biopsy verification, but it demonstrated no lesion in that place. Multidisciplinary gastroenterology and pneumology consultations recommended transbronchial ultrasonography for repeated biopsy attempts, which were successfully taken. A histologic examination confirmed parathyroid tissue, but immunohistochemistry was negative for PTH.

The patient was referred to a parathyroid surgery center for elective thoracoscopy. Perioperative biopsy confirmed parathyroid adenoma and the whole ectopic gland was removed. After surgery, both PTH and plasma calcium levels normalized.

DISCUSSION Hypercalcemia has a broad differential diagnosis, and while primary hyperparathyroidism (PHPT) is a relatively common etiology, especially in the presence of hypovitaminosis D, an elevated PTH level is an expected physiological response. Despite initial workup, no abnormalities were found in the parathyroid gland; adenomas in the ectopic localization account for only about 10% of PHPT cases. The mediastinal localization makes diagnostic verification challenging, as the MIBI scan has a sensitivity and specificity of 73% and 80%, respectively. Therefore, a confirmatory imaging method is needed for verification. Endosonography was initially assessed as the best option due to the proximity of the lesion to the esophagus, but it could not visualize the lesion, which might have been attributed to the rather difficult localization, a sonographer's experience, and the patient's cooperation. On the other hand, bronchoscopy, even though at first seen as inferior, was performed by an expert bronchoscopist and resulted in a successful biopsy. The negative PTH result from the biopsy was misleading, but according to the literature, this could be due to assay limitation and possible different forms of PTH being secreted, which the assay cannot detect.

CONCLUSIONS Osteopenia should be thoroughly investigated, even in the cases that appear benign. Once hypercalcemia is detected, and malignancy and other diagnoses are ruled out, an elevated PTH level is suggestive of PHPT and the main effort should be focused on localizing parathyroid adenomas. Diagnostic approaches should be adapted to local expertise, and alternative methods may yield better results than rigidly following guidelines. This case shows

the importance of specialized, multidisciplinary care in achieving optimal outcomes.

Key words
ectopic parathyroid adenoma, hypercalcemia, osteopenia, primary hyperparathyroidism

MARTYNA KURCZ

In medicine, there are no shortcuts: infective endocarditis

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CASE DESCRIPTION Our study presents a case of a 58-year-old patient who was hospitalized in the department of internal medicine for further investigation of progressive anemia, which during the diagnostic workup was recognized as one of the symptoms of infective endocarditis of the mitral valve, complicated by extracardiac manifestations.

Key words
infective endocarditis, spleen infarction, *Staphylococcus lugdunensis*

MICHAL SVOBODA

Cardiac arrest caused by pulmonary embolism: catheter-directed treatment as a lifesaving procedure?

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CASE DECRIPTION Pulmonary embolism is a potentially reversible cause of cardiac arrest. Standard treatment methods include advanced life support, heparin, and systemic thrombolysis. The aim of this case report is to describe possibilities of catheter-directed treatment as *ultimum refugium*.

I present a case of a 55-year-old man admitted to an intensive care unit for shock. He presented with 3-day dyspnea and unilateral lower limb edema. He developed cardiac arrest immediately after admission to the intensive care unit. Even though he received advanced life support, including echocardiography and administration of systemic thrombolysis, asystole maintained during almost the whole procedure. Autopsy proved an embolus of the pulmonary artery as the cause of death.

What else could possibly have been done after systemic thrombolysis was ineffective? My aim is to describe catheter-related treatment as a possibly lifesaving treatment method of cardiac arrest caused by pulmonary embolism.

Key words
cardiac arrest, catheter-related thrombectomy, high-risk pulmonary embolism

MIŁOSZ KNURA

Electrolyte enigma and the mystery of hyperaldosteronism: a case of self-induced dysregulation

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CASE DESCRIPTION Our study presents a case of a 25-year-old patient who was hospitalized in the department of internal medicine for further investigation of hyperaldosteronism, severe recurrent electrolyte disturbances, and secondary amenorrhea. The broader clinical context, including inconsistencies in medical history and nocturnal symptom patterns, pointed toward a iatrogenic underlying cause. Toxicology testing confirmed furosemide abuse, which explained the biochemical abnormalities.

Key words
furosemide abuse, hypokalemia, secondary hyperaldosteronism

MONIKA BUDZICH-NAPIWODZKA

Mixed neuroendocrine non-neuroendocrine neoplasms of the pancreas: a dangerous adversary with two faces. A case report and review of clinical and pathological characteristics

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INTRODUCTION Mixed neuroendocrine non-neuroendocrine neoplasms (MiNEN) of the pancreas are a rare and heterogeneous type of tumor containing both neuroendocrine and non-neuroendocrine components, each occupying 30% or more of the tumor mass. The most common form is mixed ductal adenocarcinoma and neuroendocrine carcinoma, localizing mainly in the head of the pancreas in men around the age of 60. Diagnosis is usually possible only after surgical resection, due to diagnostic difficulties associated with tumor heterogeneity.

CASE DESCRIPTION We report a case of a 78-year-old woman with advanced pancreatic adeno-MiNEN, consisting of ductal adenocarcinoma and a calcitonin-secreting neuroendocrine component. The patient presented with abdominal pain, watery diarrhea, weight loss, and general fatigue. Laboratory results showed elevated levels of procalcitonin, tumor markers (CA 19-9 and carcino-embryonic antigen), as well as calcitonin and neuron-specific enolase. Imaging studies showed a tumor in the head of the pancreas with multiple liver metastases, and histopathological analysis confirmed the diagnosis of MiNEN with a mixed adeno-neuroendocrine structure.

This case highlights the importance of exploring further the diagnosis of high procalcitonin and calcitonin values after excluding infection and medullary thyroid carcinoma. In addition, it points to the key role of comprehensive histopathological and immunohistochemical evaluation in establishing the diagnosis of MiNEN.

CONCLUSIONS MiNEN remains a diagnostic and therapeutic challenge, requiring a multidisciplinary approach that takes into account the histologic structure of the tumor and the clinical aggressiveness of the disease. This case report also emphasizes the need for increased awareness, interdisciplinary collaboration, and the creation of case registries.

Key words
malignant tumor, mixed neuroendocrine non-neuroendocrine neoplasms, pancreas

MONIKA TOMAKA-BURDZIAK

The invisible cage: a story of cardiac constriction

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INTRODUCTION The pericardium is the heart's defensive wall that protects the heart and holds it in place in the chest cavity. Pericardial diseases have a broad spectrum of presentations, with

constrictive pericarditis (CP) remaining particularly challenging in terms of diagnosis. This case report aims to demonstrate the diagnostic process for CP.

CASE DESCRIPTION A 28-year-old HIV-positive man on antiretroviral therapy, with a history of substance abuse and childhood polytrauma, was admitted to the emergency department. On admission, he presented with progressive dyspnea, chest discomfort, and reduced exercise tolerance that had lasted for over 2 weeks. Results of diagnostic workup, including magnetic resonance imaging and echocardiography, showed pleural effusion, thickened pericardium, impaired systolic function of the right ventricle, and signs of mechanical restriction such as paradoxical septal motion and annulus reversus. An invasive hemodynamic assessment confirmed balanced diastolic pressures with a characteristic dip and plateau pattern, and the diagnosis of constrictive pericarditis was established. Hence, pericardiectomy was performed, which alleviated the patient's symptoms.

DISCUSSION Differentiating CP from restrictive cardiomyopathy is critical, given the therapeutic implications, particularly the role of pericardiectomy as a definitive intervention. The contemporary management of pericardial disease involves multimodal imaging, including echocardiography, cardiac computed tomography, and magnetic resonance imaging. Cardiac catheterization is reserved for patients in whom noninvasive modalities are insufficient for definitive diagnosis. In this case, a hemodynamic assessment provided additional diagnostic confirmation, complementing prior findings.

CONCLUSIONS This case emphasizes the importance of a multimodal diagnostic approach and interdisciplinary collaboration in managing constrictive pericarditis, particularly in high-risk populations such as patients with HIV infection. Advanced imaging and hemodynamic studies are critical for differentiating CP from other causes of heart failure, thereby guiding timely and effective surgical intervention.

Key words
constrictive pericarditis, echocardiography, HIV, multimodal imaging, pericardiectomy

NAYLA LÉVEILLÉ

An inflammatory puzzle: how to link scleroderma, antimyeloperoxidase antibodies, and Q fever

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INTRODUCTION Antineutrophil cytoplasm antibody (ANCA)–associated vasculitis (AAV) is a multisystemic autoimmune disease primarily affecting small blood vessels, often involving the kidneys and lungs. While cardiac manifestations are rare, myocarditis has been described in 1% to 2% of cases. Furthermore, AAV has been reported in association with other inflammatory syndromes, including scleroderma. Infectious agents, such as *Coxiella burnetii* have also been described as potential triggers for AAV.

CASE DESCRIPTION We report a case of a 58-year-old man with a history of limited scleroderma, pulmonary hypertension, and chronic kidney disease who presented with a 2-week history of cough and dyspnea. Initial laboratory findings showed elevated inflammatory markers, acute kidney injury, and mild leukocytosis. Thoracic imaging demonstrated bilateral pulmonary opacities, prompting initiation of broad-spectrum antibiotics. However, the patient's condition deteriorated with worsening renal function, neurologic symptoms, and signs of cardiac involvement with elevated troponins. Echocardiography showed reduced ejection fraction with diffuse hypokinesia and mild pericardial effusion, raising suspicion for myocarditis.

Further investigations demonstrated positive myeloperoxidase ANCA antibodies. Myocardial biopsy showed perivascular inflammation and fibrinoid necrosis. Skin biopsy from newly developed bullous lesions demonstrated vascular thrombosis. These findings led to a diagnosis of new-onset AAV. Additionally, serology for *C. burnetii* indicated a recent resolving infection, arousing suspicion of a potential causal induction of AAV by Q fever. The patient was treated for AAV with high-dose intravenous methylprednisolone, followed by cyclophosphamide. Following treatment, he showed a significant clinical improvement, with normalization of inflammatory markers, recovery of cardiac function, and resolution of pulmonary opacities and proteinuria. He completed induction therapy and transitioned to rituximab and avacopan for long-term maintenance.

DISCUSSION This case highlights an atypical presentation of AAV with predominant cardiac involvement in a patient with scleroderma. The presence of myocarditis as the primary manifestation of AAV is rare, but has been described in the literature, among other possible cardiac manifestations. This case emphasizes the importance of considering AAV in patients with multisystemic inflammatory syndromes, particularly in those with predisposing autoimmune conditions.

Indeed, an association between scleroderma and AAV has been described in the literature, with a higher prevalence of AAV among patients with scleroderma. Shared HLA haplotypes have been observed between patients who develop both syndromes. This association is stronger for patients with limited scleroderma, and the most frequent presentation of AAV described in these cases is renal and pulmonary involvement.

The association between infections and the onset of autoimmune conditions has been widely described. In the case presented, the patient had serology positive for *C. burnetti* due to recent resolving infection, with possible induction of AAV by Q fever. This potential induction is described in multiple case reports, and reviews of the literature show that 13% of patients with Q fever also have positive ANCA autoantibodies.

CONCLUSIONS Clinicians should maintain a high index of suspicion for AAV in patients with systemic inflammatory syndromes, especially if they are known for having scleroderma, even if their presentation is atypical. The potential role of *C. burnetii* in triggering autoimmunity, and more specifically vasculitis, is an area that requires further investigation.

Key words
ANCA-associated vasculitis, myocarditis, Q fever, scleroderma

NAZLI PELIN KIRKAYAK

A rare case of multicentric Castleman disease of hyaline vascular type presenting with hepatobiliary obstruction

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İNTRODUCTION Castleman disease is a rare lymphoproliferative disorder. The multicentric form is usually characterized by systemic inflammation and lymphadenopathy, while hepatobiliary obstruction is an uncommon presentation. This report describes a case of multicentric Castleman disease of hyaline vascular type causing bile duct compression and jaundice.

CASE DESCRIPTION A 62-year-old man presented with progressive jaundice and pruritus persisting for 3 weeks. He had no known comorbidities. The physical examination showed scleral and cutaneous icterus with normal vital signs. No hepatosplenomegaly or palpable lymphadenopathy were detected. Laboratory tests showed an elevated bilirubin level (total, 10.9 mg/dl; direct, 8.1 mg/dl) and mildly elevated liver enzymes. Abdominal ultrasonography showed bile duct dilation, and computed tomography (CT)—a 34 mm × 21 mm

necrotic lymph node in the portal hilum compressing the common bile duct, along with a 120-mm irregular mass invading the spleen and pancreatic tail. Positron emission tomography/CT showed a maximum standardized uptake value (SUVmax) of 17.09 in the splenic mass and 31.6 in the portal lymph node. Enlarged paraaortic and hepatogastric lymph nodes were also detected. Endoscopic retrograde cholangiopancreatography failed due to an inaccessible papilla, necessitating percutaneous transhepatic cholangiography and biliary drainage. During the second attempt, a biliary stent was successfully placed but brush biopsy was inconclusive. Laparotomy was performed and periportal lymph node excision indicated hyaline vascular Castleman disease. Hematology consultation confirmed multicentric Castleman disease and rituximab / steroid therapy was initiated. After the first treatment cycle, liver function improved and the patient was discharged.

CONCLUSIONS Multicentric Castleman disease of hyaline vascular type rarely causes hepatobiliary obstruction. This case highlights the need to consider Castleman disease in the differential diagnosis of obstructive jaundice with lymphadenopathy, particularly when malignancy is suspected.

Key words
Castleman disease, hepatobiliary obstruction, lymphoid hyperplasia

KRISTINA ZIUTELIENE

Nodules, nodules on an arm, which one's the most abominable among them all? A case report of a local dirofilariasis in Lithuania

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INTRODUCTION Dirofilariasis is a vector-borne parasitic disease caused by *Dirofilaria* species that affects primarily domestic and wild canines and felines, with *D. immitis* and *D. repens* types posing the most significant risk for humans. Historically considered an exotic disease, dirofilariasis has recently expanded to Europe, likely due to the relocation of infected animals and climate change-induced increases in mosquito activity. This article presents a case of subcutaneous dirofilariasis managed by a family physician in Lithuania that demonstrates the importance of early diagnosis and interdisciplinary management in emerging infectious diseases.

CASE DESCRIPTION A 28-year-old man, with no previous illnesses in his medical history, sought medical attention due to the presence of a well-defined, painless nodule on his right forearm, which developed after a mosquito bite approximately 3–4 months prior. Initial examination revealed a subcutaneous nodule, and imaging tests, including ultrasound and magnetic resonance imaging, raised suspicion of a soft tissue neoplasm. Following surgical excision, the pathology report identified an encapsulated nematode, confirming the diagnosis of human subcutaneous dirofilariasis. Subsequent laboratory tests did not show eosinophilia or microfilariae in the blood, and the patient fully recovered after the surgical procedure.

DISCUSSION Over the past 2 decades, dirofilariasis has become an emerging disease in Europe, with *D. repens* being the primary cause of human infections. The disease manifests as slow-growing, painless subcutaneous nodules, often accompanied by local itching with no systemic symptoms. The clinical presentation is nonspecific and can mimic other pathologies, complicating diagnosis. Diagnostic steps include obtaining medical history, conducting imaging (ultrasound and color Doppler), and definitive confirmation via surgical excision and histopathological analysis. The management of dirofilariasis typically involves surgical removal of the nodule, while chemotherapy is not routinely recommended. Canines, as the primary reservoir, contribute to the spread of *D. repens*, with increased tourism and pet travel facilitating its geographic expansion. In Lithuania, the prevalence of dirofilariasis has risen significantly, with positive cases identified in both domestic canines and the human population. Despite the observed increase, the disease remains rare. It is imperative for clinicians, especially those in primary care, to possess clinical awareness to ensure timely diagnosis and management of the disease.

CONCLUSIONS Dirofilariasis, a disease once considered limited to warmer climates, is spreading across Europe and further up north, with an increasing incidence in both animals and humans. Timely and accurate diagnosis can be challenging, especially in early stages, but interdisciplinary cooperation among family physicians, radiologists, surgeons, pathologists, and infectious disease specialists is crucial for effective management. The case presented in the article highlights the importance of a coordinated approach to diagnose and treat emerging infectious diseases in primary healthcare settings, facilitating optimal clinical outcomes and contributing to broader awareness among primary care physicians.

Key words
dirofilaria, nematode, nodule, primary care, zoonosis

MAHRA ALMHEIRI

Meningococcal bacteremia presenting as acute pericarditis: a rare clinical challenge

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CASE DESCRIPTION *Neisseria meningitidis* is a well-known pathogen responsible for meningitis and septicemia However, its association with pericarditis is exceedingly rare, with only approximately 25 documented cases in the literature. Meningococcal pericarditis accounts for less than 1% of bacterial pericarditis cases and represents less than 0.1% of all meningococcal infections. This case report highlights a rare presentation of meningococcal bacteremia with pericardial involvement, emphasizing diagnostic challenges, treatment considerations, and the need for a structured management approach.

A 44-year-old man with multiple cardiovascular risk factors presented with acute, severe pleuritic chest pain radiating to the neck and shoulders. Initial investigations suggested acute coronary syndrome, but coronary angiography ruled out obstructive coronary artery disease. Further evaluation revealed a subsegmental pulmonary embolism, and despite anticoagulation, the patient developed hemodynamic instability requiring intra-aortic balloon pump support. Laboratory results demonstrated leukocytosis, elevated inflammatory markers, and blood cultures confirming *N. meningitidis* bacteremia. On day 3, the patient developed pericardial tamponade requiring emergent pericardiocentesis, which revealed purulent pericardial fluid positive for *N. meningitidis*. The patient was administered a 6-week course of intravenous ceftriaxone, resulting in the resolution of pericardial effusion and clinical stabilization. The diagnosis and management of meningococcal pericarditis pose significant challenges due to the rarity of the condition and the absence of standardized treatment guidelines. In contrast to viral pericarditis, which is often self-limiting and managed with nonsteroidal anti-inflammatory drugs and colchicine, bacterial pericarditis requires prolonged intravenous antibiotic therapy and, in some cases, surgical intervention. The duration

of therapy varies significantly across reported cases, ranging from 2 to 6 weeks. Monitoring treatment response in bacterial pericarditis remains complex, as there are no established markers predicting resolution or recurrence. Serial echocardiography and inflammatory markers, such as C-reactive protein and procalcitonin, are utilized to monitor disease progression.

A number of studies have indicated that colchicine may have a role in the management of pericardial inflammation, but its efficacy in cases of infectious pericarditis remains uncertain. Additionally, pericardiocentesis is often necessary to relieve tamponade, but the decision for repeated drainage or surgical pericardial window placement remains controversial due to a lack of consensus. Case reports have described aortitis and systemic inflammation in association with meningococcal pericarditis, further complicating management and requiring multidisciplinary involvement. Given the absence of clear treatment guidelines, some authors propose a structured approach to bacterial pericarditis, emphasizing early microbiological confirmation, aggressive antimicrobial therapy, serial imaging, and consideration of anti-inflammatory adjuncts. Further studies are needed to establish standardized protocols for meningococcal pericarditis and to determine the role of corticosteroids or prolonged anti-inflammatory therapy in improving clinical outcomes.

This case underscores the importance of clinical vigilance, early recognition, and a multidisciplinary approach in managing rare presentations of meningococcal pericarditis. The case also highlights the necessity for ongoing research to refine treatment guidelines and optimize patient outcomes.

Key words
acute pericarditis, diagnostic challenges, meningococcal bacteremia

MÁRTON KALABAY

An interesting case of HIV-associated infectious disease

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CASE DESCRIPTION HIV-associated infections are more severe in individuals with compromised immune systems due to HIV's destruction of CD4+ T cells. As the number of CD4+ cells decreases, the body becomes more susceptible to opportunistic infections, which increases the risk of developing AIDS. Early treatment with antiretroviral therapy and prophylaxis helps prevent disease progression. A 38-year-old HIV-positive man residing in the United Kingdom presented with fever, night sweats, hip pain, and swollen lymph nodes. He had a history of gastroenteritis with blood in stool that had persisted over 6 months and was found to have leukocytopenia and lymphocytopenia. Despite extensive testing, no clear infectious cause was initially identified, though imaging did show enlarged lymph nodes. Bone marrow aspiration revealed a *Salmonella enteritidis* infection, which was treated with targeted antibiotics. The patient's extremely high HIV copy number indicated a potential lack of adherence to treatment or viral resistance. Diarrhea is common in untreated HIV patients, affecting 40%–80% of cases. Diagnostic methods include stool microbiology, molecular techniques, and endoscopy, with flexible sigmoidoscopy being the most effective initial test for cytomegalovirus colitis. Advanced diagnostic tools are helpful in identifying underlying infections and guiding the appropriate treatment.

Key words
HIV, HIV-associated diarrhea, *Salmonella enteritidis*

MOHAMED REDHA BERRIM

When incense use becomes fatal: the first reported case of DRESS syndrome triggered by incense, successfully treated with cyclosporine after relapse under corticosteroids

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INTRODUCTION Drug reaction with eosinophilia and systemic symptoms (DRESS) syndrome is a severe hypersensitivity reaction characterized by fever, rash, eosinophilia, lymphadenopathy, and multiorgan involvement, with a mortality rate of approximately 10%. While medications are the primary triggers, the underlying mechanisms of non–drug-related causes remain poorly understood. To our knowledge, this is the first reported case of DRESS syndrome induced by incense use.

CASE DESCRIPTION A 39-year-old woman with a medical history of tuberculous lymphadenitis (treated 15 years ago) and 3 early miscarriages presented with generalized pruritic, dry, desquamative erythroderma, initially affecting the oral mucosa and progressively extending to the face and body. The patient exhibited fever (39 °C), severe asthenia, superficial lymphadenopathy (cervical, axillary, inguinal), abdominal pain, and dyspnea. No recent medication intake was reported, except for incense consumption 7 days prior to symptom onset, which she used for cognitive enhancement.

Laboratory investigations revealed hyperleukocytosis (13 100 cells/mm^3) with neutrophilia and eosinophilia (2600 cells/mm^3), elevated C-reactive protein level at 48 mg/l, hepatic cytolysis (3 times the normal levels), and a markedly elevated creatine phosphokinase (40 times the normal levels). The results of the viral serologies, autoimmune markers, and peripheral blood smears were all negative. Imaging revealed deep mediastinal, hilar, intra- and retroperitoneal lymphadenopathy, minimal hydropneumothorax, and chronic splenic vein thrombosis. Skin biopsy confirmed DRESS syndrome, with a RegiSCAR score of 6.

Initial treatment included topical corticosteroids, systemic corticosteroids (1 mg/kg/day for 4 weeks, then tapered), anticoagulation, and antibiotics, leading to rapid clinical improvement. However, at 0.75 mg/kg/day of corticosteroids, the patient experienced a severe relapse with recurrent erythroderma, facial edema, palmoplantar pustulosis, worsening dyspnea, myalgias, and systemic lymphadenopathy. Laboratory findings confirmed persistent hyperleukocytosis and eosinophilia. High-dose corticosteroids and intravenous immunoglobulins (2 g/kg over 2 days) were reintroduced, followed by cyclosporine (5 mg/kg/day). This led to rapid and sustained clinical improvement, with no recurrence after 3 years of follow-up.

DISCUSSION This case marks the first documented instance of incense-induced DRESS syndrome, thereby expanding the spectrum of potential triggers for this life-threatening condition. The absence of conventional drug exposure highlights the importance of considering environmental and herbal exposures in unexplained hypersensitivity reactions. The case also underscores the role of cyclosporine in corticosteroid-dependent or refractory DRESS, supporting its use as a second-line immunosuppressant in severe cases.

CONCLUSIONS 1) Nonpharmaceutical triggers, such as incense, should be considered in unexplained hypersensitivity reactions. 2) The RegiSCAR score remains essential for diagnosing DRESS syndrome. 3) Cyclosporine is an effective therapeutic option in corticosteroid-dependent or refractory cases. 4) Early recognition

and appropriate immunosuppressive therapy are crucial to prevent life-threatening complications.

Key words
cyclosporine, DRESS syndrome, first case, incense

NAZAR NEGRYCH

Guillain–Barré syndrome in a patient with multiple sclerosis on the background of immunosuppressive therapy

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INTRODUCTION Multiple sclerosis (MS) and Guillain–Barré syndrome (GBS) are autoimmune demyelinating diseases that affect the central and peripheral nervous systems, respectively. The co-occurrence of these 2 diseases in a single individual is extremely rare.
CASE DESCRIPTION A 43-year-old patient with a history of remitting-relapsing MS since 2016, treated for 4 years with a selective immunosuppressant (anti-CD20 therapy), was admitted to the neurology department on September 4, 2024, with complaints of dysphagia and speech impairment, accompanied by severe limb weakness.

According to the patient's medical history, the most recent administration of a monoclonal antibody occurred in early June 2024. In July 2024, the patient experienced an enterovirus infection, which was accompanied by a rash and fever, and after symptomatic therapy, her condition returned to normal. On August 20, 2024, the patient experienced a recurrent viral infection with fever reaching 39 °C, nausea and vomiting. Two days later, the patient exhibited additional symptoms, including a severe headache and generalized tremor. She was transported to the hospital by ambulance. Examination revealed meningeal symptoms, high tendon and periosteal reflexes, and a positive pathological Babinski reflex on both sides. Lumbar puncture was performed, with a significant cytosis of 320 cells/μl (95% neutrophils and 5% lymphocytes). The patiend was referred to the intensive care unit with a diagnosis of meningoencephalitis. The patient's motor function gradually declined, with weakness in her legs and arms, as well as difficulty swallowing and speaking. On August 27, 2024, magnetic resonance imaging (MRI) of the brain was performed, with cervical and thoracic spine: MRI signs of demyelinization process without signs of activity were discovered. On September 24, electroneuromyography (ENMG) was done, showing signs of axonal peroneal nerve damage on the left with function distal to 47% of the lower limit of normal, and on the right distal to the lower limit of normal were observed.

Objective condition of the patient: consciousness is clear, physical examination without pathology, neurologically: nystagmus, difficulty swallowing and speech; no active movements in the right leg, sharply limited in the left leg and right arm (only fingers can move); tendon reflexes are depressed; there are no pathological signs.

After a thorough review of the patient's medical history, neurological examination, cerebrospinal fluid analysis, MRI and ENMG results, a diagnosis of GBS with bulbar disorders and severe peripheral tetraparesis was made, along with MS, remitting-relapsing type, remission phase (without process activity).

The patient received intensive therapy to correct the immune response. Due to the patient's swallowing disorder, a nasogastric tube was used to ensure adequate nutrition. The associated physical rehabilitation was instrumental in restoring the patient's limb function.
DISCUSSION The patient's medical history, including anti-CD20 therapy and a recent viral infection, suggests the potential presence of triggering factors that may have contributed to the development of autoimmune peripheral nerve damage.

The combination of 2 autoimmune diseases complicates the prognosis, but in the remission phase of MS, the chances of partial recovery of motor function are high.
CONCLUSIONS The clinical case presented in the article emphasizes the importance of an interdisciplinary approach in the diagnosis and treatment of patients with comorbidities.

Key words
Guillain–Barré syndrome, multiple sclerosis

NAZLI ELAYADI

Autoimmune myelofibrosis: a rare but serious condition

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INTRODUCTION Autoimmune myelofibrosis (AIMF) is a rare but potentially life-threatening condition characterized by nonmalignant bone marrow fibrosis occurring in the context of autoimmune diseases. In contrast to primary myelofibrosis (PMF), which is a clonal myeloproliferative disorder associated with *JAK2*, *CALR*, or *MPL* mutations, AIMF is characterized by immune-mediated stromal damage and cytokine-driven fibroblast activation. It has been reported in association with systemic lupus erythematosus (SLE), Sjögren syndrome, rheumatoid arthritis, systemic sclerosis, and primary biliary cholangitis (PBC). Due to its rarity and overlapping clinical features with hematologic malignancies, AIMF remains a diagnostic and therapeutic challenge. We present 2 cases of secondary AIMF, which are associated with different autoimmune diseases. The cases highlight the importance of early recognition and timely intervention.
CASE DESCRIPTION Case 1 refers to AIMF associated with Sjögren syndrome and PBC. A 61-year-old hypertensive woman with a history of overlap syndrome combining Gougerot-Sjögren's disease and primary biliary cholangitis (PBC) presented with deep tumor syndrome (marked lymphadenopathy and hepatosplenomegaly), bicytopenia (anemia and leukopenia), and polyclonal hypergammaglobulinemia. Investigations showed *JAK2* mutation and negative bone marrow biopsy (BMB; grade 2 myelofibrosis without blast excess). Immunologic workup showed positive anti-SSA/Ro, anti-mitochondrial antibodies, and elevated IgG4 levels. The patient was started on corticosteroids (1 mg/kg/day) and hydroxychloroquine, leading to significant clinical and hematologic improvement.

Case 2 refers to AIMF associated with SLE. A 35-year-old woman with no prior medical history presented with right hypochondrial heaviness and mild inflammatory anemia (hemoglobin, 10.4 g/dl, C-reactive protein, 18 mg/l). Abdominal ultrasound showed hepatosplenomegaly, BMB revealed prefibrotic myelofibrosis with an inflammatory T- and B-cell infiltrate. Autoimmune panel showed positive ANA (1:1280), anti-dsDNA, and low complement levels (C3, C4). The SLE diagnosis was confirmed based on the 2019 ACR/EULAR criteria. The patient was treated with corticosteroids (1 mg/kg/day) and immunomodulatory therapy (mycophenolate mofetil, 2 g/day), leading to rapid clinical and hematologic remission.
CONCLUSIONS AIMF should be considered in the differential diagnosis of bone marrow fibrosis in autoimmune diseases. BMB is essential to confirm nonclonal fibrosis. Corticosteroids remain the cornerstone of treatment, with immunosuppressive agents as steroid-sparing options. Prompt recognition and early intervention are crucial to prevent progressive bone marrow failure.

Key words
autoimmune myelofibrosis, bone marrow fibrosis, corticosteroid therapy, hematologic disorders, systemic autoimmune diseases

NUR BEYZA TUKEK KILICASLAN

Unmasking mucormycosis: a deadly cause of vision loss in uncontrolled diabetes

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INTRODUCTION Mucormycosis is a rare but life-threatening fungal infection commonly affecting immunocompromised individuals, including those with uncontrolled diabetes mellitus (DM). This case report discusses a 75-year-old woman with poorly controlled type 2 DM who presented with diabetic ketoacidosis (DKA) and later developed rhinocerebral mucormycosis, a severe complication of her underlying condition. The patient had discontinued both insulin therapy and oral antidiabetic medications several months prior, resulting in hyperglycemia and ketoacidosis.

CASE DESCRIPTION Upon admission, the patient presented with headache, fatigue, and significantly elevated blood glucose levels (790 mg/dl), along with metabolic acidosis and ketonuria, consistent with DKA. Despite initiating insulin infusion, fluid resuscitation, and broad-spectrum antibiotic therapy, the patient's condition worsened rapidly. She developed facial numbness, ptosis, and bilateral vision loss within hours. Ophthalmic examination revealed anterior optic neuropathy, and imaging showed diffusion restrictions in the bilateral frontal lobes, suggesting central nervous system involvement. Further investigation, including orbital magnetic resonance imaging and paranasal sinus computed tomography, revealed signs of fungal sinusitis, including contrast-enhanced densities in the right orbit and extensive mucosal thickening in the maxillary and ethmoid sinuses. Endoscopic examination revealed black necrotic crusts in the nasal cavity, raising suspicion of mucormycosis. Two aspiration samples from the nasal cavity confirmed mucor growth. Liposomal amphotericin B was initiated, but the infection had already spread to the orbit and brain, preventing surgical intervention. Despite undergoing aggressive antifungal treatment, the patient ultimately succumbed to the infection, emphasizing the high mortality rate associated with delayed diagnosis and extensive involvement.

DISCUSSION This case highlights the critical role of uncontrolled diabetes, particularly in the setting of DKA, as a major risk factor for mucormycosis. The acidic environment in DKA, with elevated serum iron levels, facilitates fungal growth. Timely diagnosis and treatment are essential to improve outcomes, as mucormycosis can progress rapidly, particularly in immunocompromised patients. The clinical features of rhinocerebral mucormycosis, including facial pain, nasal congestion, and vision loss, require high clinical suspicion in diabetic patients. Early intervention with antifungal therapy, such as liposomal amphotericin B, and surgical debridement when feasible, are critical in reducing mortality.

CONCLUSIONS This case underscores the need for better patient education on diabetes management to prevent severe complications such as mucormycosis.

Key words
blindness, diabetic ketoasidosis, mucormycosis

PAULA RAMIREZ

Caged heart: a case of tuberculous constrictive pericarditis

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CASE DESCRIPTION Constrictive pericarditis is a rare condition of the pericardium where inflammation and fibrosis cause stiffness and severe diastolic dysfunction, leading to right-sided heart failure. Its incidence is estimated at 1.8% after episodes of acute pericarditis. The main symptoms are edema, ascites and hepatosplenomegaly. Diagnosis can be complex, as it depends mainly on differentiation from other causes of diastolic heart failure. The initial evaluation includes an electrocardiogram, chest X-ray and echocardiogram, the latter being the most useful. There are several causes, but idiopathic cases constitute a high percentage. Treatment depends on the stage of the disease; nonsteroidal anti-inflammatory drugs and colchicine are usually used, and if there is no response, pericardiectomy may be necessary.

The purpose of this report is to present a case of tuberculous constrictive pericarditis in a 21-year-old man with a history of incarceration, HIV-negative. The patient presented with typical symptoms and signs of right heart failure. Blood work and imaging studies are needed due to the complexity of the diagnosis. Finally, the cause was established through the study of pericardial tissue obtained on pericardiectomy. Therapeutics were performed following the suggested treatment guidelines and according to the availability of drugs in our country.

This case emphasizes the importance of considering tuberculous pericarditis regarding patients with a history of persistent pericarditis, especially considering high-risk population such as those with incarceration history. Timely diagnosis and a multidisciplinary approach are critical to prevent disease progression and improve clinical outcomes. The rising incidence of tuberculosis highlights the need for heightened clinical suspicion and the development and implementation of early detection strategies.

Key words
constrictive pericarditis, heart failure, tuberculosis, Uruguay

PETER KROMKA

A slimmer waist but a hazy mind: a case of Wernicke encephalopathy following bariatric surgery

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INTRODUCTION Wernicke encephalopathy (WE) is an acute, potentially life-threatening neurological disorder resulting from severe thiamine deficiency. The clinical presentation primarily involves both the central and peripheral nervous systems. Individuals with severe alcohol abuse are notoriously at risk; however, any condition leading to malnutrition predisposes to the development of WE.

CASE DESCRIPTION A 26-year-old woman was admitted to the emergency department after being referred by her psychiatrist. Two months prior, she had undergone bariatric gastric surgery, which was complicated by a postoperative course requiring rehospitalization due to profuse vomiting. She remained stable until several days before the current admission, when she developed weakness and malaise following recurrent episodes of vomiting. On admission, her main complaint was an altered mental status, which progressively worsened, manifesting as confusion and disorientation. She also reported generalized weakness and mobility impairment. Initial laboratory evaluation showed no significant abnormalities. The infectious disease and toxicology work-up yielded negative results. Chest X-ray and brain computed tomogrpahy were unremarkable. During hospitalization, oculomotor dysfunction manifested, with the initial symptoms persisting. The clinical picture was highly suggestive of WE, suspected to be causally related to the recent bariatric procedure. High-dose parenteral thiamine supplementation was initiated, leading to a rapid improvement in both cognitive and

motor functions. Brain magnetic resonance imaging (MRI) findings supported our diagnosis. She was discharged in satisfactory condition after seven days of hospitalization.

DISCUSSION WE is a well-recognized neurologic complication of thiamine (vitamin B_1) deficiency. If left untreated or treated inappropriately, WE may progress to Korsakoff syndrome, a permanent form of brain impairment. Although WE is primarily linked to severe alcohol use disorder, it can also occur in other settings. Some degree of thiamine deficiency is frequently observed after bariatric surgery. The estimated prevalence of WE in this population is minimal; however, it is likely underdiagnosed. Our patient underwent laparoscopic sleeve gastrectomy, with symptom onset occurring eight weeks postoperatively. Within hours, she developed the full triad of symptoms. Given the clinical context, protracted vomiting was considered the primary precipitant of WE development in our patient. Our diagnosis of WE was purely clinical, although MRI findings later supported the diagnosis. Neither imaging nor laboratory studies should delay treatment; it must be initiated even on suspicion. Is it well established that medical consideration of WE in nonalcoholic individuals is often neglected. Patients after bariatric surgery represent a high-risk group, and the threshold for diagnosing WE must be low. Due to the excellent safety profile of thiamine, even an approach of overdiagnosis and overtreatment may be preferable.

CONCLUSIONS WE is a serious yet frequently unrecognized condition in clinical practice. The most common barrier to an accurate diagnosis is insufficient clinical suspicion, particularly in nonalcoholic individuals and in the absence of the classic symptom triad. Physicians involved in the care of post-bariatric surgery patients should maintain a high index of suspicion for this condition. The clinical significance of this issue is underscored by the rising frequency of bariatric procedures.

Key words
bariatric surgery, Wernicke encephalopathy

PETER TURAI

Normouricemic gout: the role of lubricin

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CASE DESCRIPTION The pathomechanism of gout is typically associated with hyperuricemia, which leads to the deposition of urate crystals and inflammatory reactions in the joints and other tissues. However, gout symptoms can also develop with normal serum uric acid levels, a phenomenon known as normouricemic gout. This condition has been increasingly recognized in recent literature, with growing evidence suggesting that alternative mechanisms, including genetic predisposition and inflammatory pathways, may contribute to its pathogenesis.

In our case study, we were the first in Hungary to identify the alternative transcription of lubricin (PRG4) in a patient treated for steroid-dependent gout despite normal serum uric acid levels. Lubricin, a glycoprotein encoded by the *PRG4* gene, is essential for joint lubrication and cartilage homeostasis. Recent studies suggest that PRG4 dysfunction may contribute to inflammatory joint diseases, including gout.

Our 58-year-old patient with smoldering (IgG κ) myeloma presented with progressively worsening hand and foot joint complaints starting in November 2022. Given his underlying disease, a positron emission tomography / computed tomography scan was performed, which revealed mixed degenerative and inflammatory joint alterations. Laboratory findings showed mildly elevated inflammatory markers and total protein levels, with no other significant abnormalities; the uric acid levels consistently remained within the normal range. Based on imaging, immunoserological tests, and clinical findings, systemic autoimmune diseases and spondyloarthropathies were excluded. However, a DECT scan of the ankle and foot confirmed the presence of intraarticular and periarticular urate crystals.

Normouricemic gout has been documented in the literature, with studies linking impaired urate excretion, fluctuating uric acid levels, or tissue-specific factors to crystal deposition. Genetic research identifies susceptibility loci, including genes regulating urate transport and inflammasome activation, potentially promoting crystal formation at lower concentrations.

Based on prior literature, we performed germline genotyping of the *PRG4*, *NLRP3*, *THBS1*, and *SERPINB3* genes associated with gout flares occurring at normal uric acid levels. Additionally, RNA-level analysis was conducted on synovial fluid, bone marrow, and peripheral blood samples collected during the patient's gout flare. The patient's treatment was supplemented with allopurinol.

With allopurinol treatment, the patient's joint symptoms improved significantly, enabling the discontinuation of steroid therapy. Exome sequencing did not reveal any pathogenic germline mutations; however, RNA transcript analysis of the synovial sample identified an alternative splicing mechanism of the lubricin transcript. This finding aligns with recent studies that suggest lubricin dysfunction may play a role in inflammatory arthritis and cartilage degradation, potentially facilitating urate crystal deposition even in normouricemic conditions.

This case underscores that gouty arthritis can occur even with normal serum uric acid levels. In our study, no germline mutation involving the lubricin gene (*PRG4*) was detected; however, synovial RNA transcript analysis revealed an alternative splicing mechanism of lubricin (PRG4), which may play a role in gout development in the context of normal uric acid levels. The significance of lubricin alterations in gout pathogenesis warrants further investigation, as it may represent a novel target for diagnostic and therapeutic strategies. Our findings contribute to the growing body of evidence that gout is a heterogeneous disease with diverse molecular mechanisms beyond hyperuricemia.

Key words
inflammatory arthritis, lubricin, *PRG4*, inflammatory arthritis, normuricemic gout, urate crystals

PREM THILAK PALANI

Heart-breaking: a devastating manifestation of an overlap syndrome

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INTRODUCTION Dermatomyositis (DM) is an autoimmune disorder characterized by muscle weakness and distinctive skin manifestations. In some cases, it overlaps with other connective tissue diseases, such as systemic lupus erythematosus (SLE). Although cardiac involvement in DM is well-documented, its association with acute coronary syndromes remains unclear, especially in the context of myocardial infarction with nonobstructive coronary arteries (MINOCA).

CASE DESCRIPTION We report a case of a 56-year-old female presenting with a six-month history of fever, proximal limb and neck muscle weakness, weight loss, and skin rashes. Laboratory investigations revealed pancytopenia, Coombs-positive hemolytic anemia and significant proteinuria. She tested positive for Mi-2a, Mi-2b and NXP-2 antibodies on an autoimmune myositis panel. An anti-nuclear antibody test was positive with a speckled pattern and a titer of 1:100, and also tested positive for anti-double stranded DNA (anti-dsDNA), anti-

Smith (SmD1), anti U1-Ribonucleoprotein (anti-U1snRNP), anti-SSA and anti-nucleosome antibodies on an extractable nuclear antigen profile; this confirmed our suspicion of dermatomyositis-SLE overlap syndrome. The patient began the prescribed course of immunosuppressive therapy with intravenous methylprednisolone. Despite timely intervention, she developed sudden-onset chest pain, dyspnea, and hemodynamic instability 3 days after completion of 3-day course of induction immunosuppression. Electrocardiography revealed ST-elevation myocardial infarction in the chest leads V2–V4, and echocardiography demonstrated anterior and lateral regional wall motion abnormalities. The patient also had serially rising troponin levels. She was transferred to the critical care unit and intubated; however, she had persistent shock not responding to inotropes and a repeat echocardiogram revealed pericardial fluid. She was emergently taken for surgical intervention, which revealed nonobstructive coronary arteries but no evidence of hemopericardium, raising the possibility of a contained rupture of the left ventricle. Despite aggressive management, the patient died from cardiogenic shock.

DISCUSSION This case highlights the potential for catastrophic cardiovascular events in autoimmune diseases, even in the absence of traditional atherosclerotic risk factors. Autoimmune inflammation may contribute to MINOCA through endothelial dysfunction, microvascular inflammation, or myocarditis. Additionally, the presence of anti-NXP-2 antibodies led to the suspicion of a paraneoplastic process, further complicating the disease course.

CONCLUSIONS Patients with dermatomyositis-SLE overlap syndrome are at increased risk of severe cardiovascular complications, including MINOCA and structural myocardial injury. Early recognition, aggressive immunosuppression, and routine cardiovascular screening are essential in these patients. Further studies are needed to establish guidelines for risk stratification and prevention strategies in autoimmune-related cardiac events.

Key words
dermatomyositis, myocardial infarction, overlap, systemic lupus erythematosus

RAMSHA ABID

Beyond the rash: case of varicella induced kidney injury

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INTRODUCTION Varicella-zoster virus (VZV) causes varicella (chickenpox) as a primary infection and remains latent in the ganglia of different dermatomes until it becomes reactivated causing herpes zoster (shingles) as a secondary infection. This case report aims to highlight the potential for kidney involvement in chickenpox and emphasize the importance of early recognition and management of such complication in an otherwise young, healthy adult.

CASE DESCRIPTION A 29-year-old immunocompetent man who developed acute kidney injury (AKI) following VZV infection presenting with high-grade fever, epigastric pain, vomiting, decreased urine output, and a vesicular rash. Initial management with intravenous hydration and acyclovir was insufficient to stabilize renal function, leading to worsening AKI and necessitating hemodialysis. A renal biopsy revealed chronic tubulointerstitial nephritis. The patient's renal function began to recover, and he was discharged with follow-up in clinic.

DISCUSSION The case highlights the rare but serious complication of AKI in VZV infection, emphasizing the importance of early recognition and prompt intervention (including renal replacement therapy and adjusted antiviral treatment), which ultimately led to complete recovery of kidney function.

Key words
acute kidney failure, acyclovir, varicella-zoster virus

RAMZI BRIK

Autoimmune encephalitis: a diagnostic challenge in the setting of multisystem autoimmunity

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CASE DESCRIPTION Autoimmune encephalitis (AE) is a rare central nervous system disorder, often associated with systemic autoimmune diseases. We report a case of a 47-year-old woman with a history of antisynthetase syndrome, Hashimoto thyroiditis, and autoimmune hepatitis, admitted for subacute febrile headaches preceded by visual hallucinations and confusion. Clinical examination revealed altered consciousness, right-sided flaccid hemiplegia with abolished deep tendon reflexes, a unilateral Babinski sign, and signs of dysautonomia. Investigations showed an inflammatory syndrome, cerebrospinal fluid lymphocytic pleocytosis, and magnetic resonance imaging findings of frontal and basal ganglia lesions suggestive of AE. The patient showed a favorable response to corticosteroids, immunoglobulins, and cyclophosphamide, highlighting the importance of early diagnosis and prompt management to improve outcomes.

Key words
autoimmune encephalitis, immunoglobulins, magnetic resonance imaging findings

ROMAIL SHAHID

Thyroidectomy's ripple effect: the Fahr's fallout

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CASE DESCRIPTION Fahr syndrome is a rare disorder characterized by neurological abnormalities, such as tetany, seizures, spasticity, speech disturbances, parkinsonism and involuntary movements. The prevalence of Fahr syndrome is very low, it could be primary idiopathic deposition of calcium in bilateral basal ganglia thus causing Fahr disease, which is preferably labelled by the area of calcification, or it could be due to an endocrine disorder such as hypoparathyroidism or secondary hypoparathyroidism. If calcifications are due to secondary hypoparathyroidism, it is referred to as Fahr syndrome. Thyroidectomy is a commonly performed procedure nowadays, but post-thyroidectomy hypoparathyroidism is relatively rare and permanent hypoparathyroidism leading to calcium deposition in the brain, resulting in secondary parkinsonism, is again a rare or under reported condition. In this case report, we describe a patient who developed secondary parkinsonism 30 years after thyroidectomy, and also developed temporomandibular joint displacement (TMJ) due to spasm of facial muscles. He underwent thyroidectomy due to multinodular goiter, and for 30 years had been using thyroxine 150 µg with poor compliance, but never had symptoms of hypocalcemia or hypoparathyroidism. Suddenly, he developed tetanic spasms and TMJ displacement for which when he sought medical attention and was then treated for hypocalcemia; he was having low intact PTH (i-PTH), and during his hospitalization we noticed an episode of psychosis. On detailed examination, he had generalized rigidity and pill-rolling tremors which he said he never noticed to be bothersome. For this cause, we investigated this patient by neuroimaging (brain computed tomography), revealed bilateral calcifications. On this basis, the patient was diagnosed with Fahr syndrome.

Key words
Chvostek sign, Fahr syndrome, intact PTH, Parkinson disease, temporomandibular dislocation

SIRINE SAADAOUI

Diagnosis of Erdheim–Chester disease following a traumatic injury: atypical mode of onset

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CASE DESCRIPTION We describe a case of a young man with a diagnosis of Erdheim–Chester disease (ECD) following a traumatic injury. A 24-year-old patient presented to our department for inflammatory polyarthralgia, reporting a recent fall on his left knee. Imaging revealed a medullary reconversion with a thin layer of effusion and diffuse osteosclerosis. Bone biopsy confirmed the suspicion of ECD. It is a rare systemic non-Langerhans cell histiocytosis of the middle-aged adult. The most common characteristic is bilateral and symmetric osteosclerotic lesions in long bones. Pathological examination provides the definitive diagnosis; ruling out differential diagnosis. Imaging and clinical biology assess the extent of the disease. Treatment has to be decided on a case-to-case basis depending on the patient's symptoms.

Key words
Erdheim–Chester disease, histiocytosis

VAIBHAV AGARWAL

Connecting the dots

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INTRODUCTION Pregnancy and the postpartum period are prothrombotic states, increasing the risk of venous thrombosis. Scrub typhus may exacerbate coagulation disturbances. Paroxysmal nocturnal hemoglobinuria (PNH) is a rare acquired hematologic disorder characterized by complement-mediated hemolysis, thrombosis, and bone marrow failure. We report a case of PNH unmasked by scrub typhus in a postpartum female, leading to cortical venous thrombosis and deep vein thrombosis (DVT).

CASE DESCRIPTION An 18-year-old postpartum woman from Kolkata, India, with no known comorbidities, presented with high-grade fever with severe headache and projectile vomiting for 3 days, bilateral dimness of vision for 2 days, and altered sensorium following multiple seizures a day prior to admission.

She had delivered a full-term baby via normal vaginal delivery 6 days before admission and had been discharged in a stable condition 3 days postpartum. There was no history of cough, dysuria, diarrhea, external blood loss (hemoptysis, hematemesis, melena), joint pain, oral ulcers, or alopecia. She denied using oral contraceptive pills and had no history of prior pregnancy loss or thrombotic events. On admission, she was comatose (Glasgow Coma Scale, E2V2M3) and febrile. Vital signs were: blood pressure, 140/90 mm Hg; pulse rate, 110 bpm; respiratory rate, 20 breaths per minute, oxygen saturation: 96% (on 4 l O_2/min).

Neurological examination showed bilaterally dilated pupils with sluggish reaction to light and a left-sided extensor plantar response. General examination revealed marked pallor without icterus, cyanosis, clubbing, edema, or hepatosplenomegaly. Multiple necrotic black ulcers were noted over the dorsal aspects of both upper and lower limbs.

Investigations and findings comprised severe anemia (hemoglobin, 7 g/dl), elevated inflammatory marker levels (C-reactive protein, 76 mg/l; erythrocyte sedimentation rate, 120 mm/h), and scrub typhus IgM positivity in the enzyme-linked immunosorbent assay. Magnetic resonance imaging of the brain showed right parieto-occipital venous infarct, magnetic resonance angiography revealed extensive cerebral venous sinus thrombosis, and Doppler ultrasound yielded an image of DVT in bilateral axillary, saphenofemoral, and popliteal veins

The patient was managed with intracenous doxycycline for scrub typhus, levetiracetam for seizures, and heparin overlapped with warfarin for thrombosis. Thrombophilia and autoimmune workup (ANA, ds-DNA, C3, C4, APLA, ANCA, factor V Leiden, protein C, protein S, and antithrombin III) were negative. Despite treatment, persistent hemolytic anemia and thrombosis in atypical sites (cortical veins, limb ulcers) raised suspicion of PNH. Flow cytometry (FLAER test) confirmed deficiency of CD55 and CD59, leading to a PNH diagnosis. The patient was started on oral prednisolone (1 mg/kg) along with continued anticoagulation (heparin and warfarin). Follow-up showed progressive improvement in muscle power, vision, and limb ulcers, with resolution of thrombi and stable hemoglobin levels.

DISCUSSION This case illustrates how pregnancy, scrub typhus, and PNH can synergistically heighten thrombotic risk. Pregnancy induces a hypercoagulable state, while scrub typhus triggers endothelial dysfunction, which likely exacerbated PNH-related hemolysis and thrombosis. PNH-associated thrombosis commonly involves atypical sites, such as cortical and deep veins, making it a crucial differential in unexplained thrombosis with hemolytic anemia, prompting the need for urgent anticoagulation and immunosuppressive therapy initiation to prevent mortality and morbidity.

Key words
atypical site thrombosis, hemolytic anemia, paroxysmal nocturnal hemoglobinuria, scrub infection, vasculitic ulcer

WARDAH MOHSIN

Discerning etiology of refractory hypertension in a young woman

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INTRODUCTION Hypertension is a leading global risk factor for cardiovascular morbidity and mortality. Approximately 10% of cases result from secondary hypertension, defined as elevated blood pressure due to an identifiable cause. Early diagnosis and treatment of secondary hypertension are crucial, as addressing the primary pathology can alter patient outcomes and potentially eliminate the need for long-term antihypertensive therapy. Paragangliomas are rare neuroendocrine tumors of neural crest origin and account for only 0.1% of cases of hypertension. These tumors secrete catecholamines, resulting in a wide array of symptoms and posing diagnostic challenges.

CASE DESCRIPTION A 17-year-old woman presented with a 6-month history of episodic palpitations, anxiety, and headaches, which had increased in frequency over the last 4 weeks. She also reported an episode of transient loss of consciousness preceded by lightheadedness and diaphoresis 6 weeks ago. She had no significant past medical history aside from poorly controlled hypertension for 6 months, despite optimal doses of irbesartan, nifedipine, and hydrochlorothiazide. Her family history was unremarkable for cardiovascular, renal, or endocrine disorders.

At presentation, her pulse was 96 bpm, blood pressure was elevated at 166/102 mm Hg, with no postural changes with normal blood glucose and oxygen saturation. Fundoscopy revealed grade 2 hypertensive retinopathy. Systemic examination yielded no substantial findings. Preliminary investigations indicated the presence of normocytic normochromic anemia, an elevated white

blood cell count, and a mildly elevated HbA_{1c} level of 6%, which is indicative of prediabetes.

Rapid troponin levels were within normal ranges. The results of remaining laboratory tests, including bicarbonate, serum creatinine, thyroid function tests, lipid profile, urea, and electrolytes, were unremarkable. Urine examination was conducted, yielding negative results for hematuria and proteinuria. ECG results demonstrated sinus tachycardia, left axis deviation, and left ventricular hypertrophy (LVH), findings that were corroborated by echocardiography, which showed moderate concentric LVH.

Renal artery stenosis was ruled out via Doppler ultrasound and renal angiography. Abdominal ultrasound detected a well-defined echogenic mass in the aortocaval region, prompting further evaluation. Given the patient's fluctuating blood pressure and the presence of an abdominal mass, a paraganglioma was suspected. Plasma-free metanephrine levels were elevated (845 pg/ml; reference range <57 pg/ml), confirming catecholamine excess. Magnetic resonance imaging of the abdomen was performed that identified a 4 cm × 2 cm well-defined mass in the aortocaval region, displacing the inferior vena cava laterally. After a multidisciplinary discussion involving internal medicine, endocrinology, and surgery specialists, the patient was initiated on α-blockade with doxazosin and a high-sodium diet with fluid intake prior to undergoing surgical resection. A subsequent exploratory laparotomy resulted in the successful removal of a well-circumscribed 4 cm × 4 cm tumor. Postoperative recovery was uneventful with no episode of hypotension or hypoglycemia. Histopathology confirmed the diagnosis of paraganglioma with a characteristic Zellballen pattern. At the 6-month follow-up, the patient remained normotensive without antihypertensive therapy, with normal ambulatory blood pressure monitoring and HbA_{1c} levels.

DISCUSSION AND CONCLUSIONS Paragangliomas can present with a variety of symptoms, including refractory hypertension, episodic palpitations, headaches, and diaphoresis. A high index of suspicion is paramount for diagnosis, and surgical excision remains the definitive treatment. It is essential for young patients with hypertension to undergo a comprehensive workup to rule out any secondary causes. This case demonstrates the importance of thorough clinical evaluation and collaborative care in diagnosing and managing rare, yet manageable, conditions.

Key words
metanephrines, paraganglioma, secondary hypertension

ZSÓFIA TURGONYI

Thymoma-associated paraneoplastic gastroparesis

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INTRODUCTION Although gastroparesis of unknown origin is a rare condition, it can sometimes be an early symptom of certain types of cancer. In our case, we present a paraneoplastic gastroparesis associated with thymoma.

CASE DESCRIPTION A 55-year-old woman was admitted to our department with a 2-week history of obstipation. Abdominal imaging revealed gastric retention, and a nasogastric tube drained a large volume of atonic gastric content. Gastroscopy confirmed gastric and duodenal paresis. With the use of prokinetic agents and laxatives, bowel movements resumed, and she was discharged. The symptoms were initially attributed to a recent respiratory infection and antibiotic use. However, her symptoms recurred shortly thereafter, presenting with obstipation, vomiting, and new-onset diplopia, which led to admission to our Neurology Department. Her neurological examination revealed internuclear ophthalmoparesis without nystagmus. Cranial computed tomography (CT) and magnetic resonance imaging showed no significant abnormalities, and routine cerebrospinal fluid analysis, immunological panel, antiganglioside antibodies, tumor markers, and serological tests for viruses and *Borrelia* were all negative. Paraneoplastic antibodies were not detected in either the serum or cerebrospinal fluid.

Nerve conduction studies showed no evidence of myopathy or myasthenia. The Tensilon test was negative, as were adult and fetal acetylcholine receptor antibodies and anti-MuSK antibodies. Pyridostigmine treatment did not improve her symptoms, thus ruling out a definitive diagnosis of myasthenia gravis.

Given the suspicion of a paraneoplastic process, a chest CT was performed, revealing a thymoma. The patient was referred to the Thoracic Surgery Department of the National Institute of Oncology, where she underwent radical thymectomy followed by postoperative radiotherapy. After the treatment, her diplopia resolved, and her gastrointestinal symptoms improved gradually.

CONCLUSIONS Although autonomic dysfunction is rare in myasthenia gravis, and paraneoplastic gastroparesis can occur in other malignancies, the coexistence of suspected myasthenia gravis and gastrointestinal symptoms should prompt an evaluation for thymoma.

Key words
gastroparesis, myasthenia gravis, thymoma