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Clinical Cases in Internal Medicine: Learning Through Practice
(9th McMaster International Review Course in Internal Medicine,
May 9–11, 2024, Kraków, Poland, hybrid course)

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ABSTRACT PROCEEDINGS OF THE BEST CASE REPORT CONTEST 2024

Clinical Cases in Internal Medicine: Learning Through Practice
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AWARD-WINNING CASES

1ST PLACE: BABETA ČÁPKOVÁ

Can zucchini kill you?

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INTRODUCTION The medical history–taking is a fundamental step in the diagnostic process. While paraclinical methods are essential aids, we cannot solely rely on them to establish a diagnosis. Nurses, who spend more time with the patient than physicians, often gather information that may be helpful in making a diagnosis.

CASE DESCRIPTION A 54-year-old healthy patient was admitted for sudden severe abdominal pain, vomiting, and hemorrhagic diarrhea. Upon admission, the patient was hypotensive. Laboratory tests revealed elevated levels of aminotransferases (alanine aminotransferase, 14.8 μkat/l; aspartate aminotransferase, 21.1 μkat/l) and leukocytosis (19.9 $\times$ 10^9/l). Transaminase elevation was attributed to hypotension. To rule out unspecified intra-abdominal pathology, an abdominal computed tomography scan was performed. Liver vessel occlusion was not confirmed. Other pathological findings complicated the differential diagnosis. Besides thickening of the colon wall, there was significant periportal "cuffing" and subhepatic region swelling. Acute gastroenteritis, probably of infectious origin, was suspected by the attending physician. After volume expansion, the blood pressure normalized; however, intense pain and diarrhea persisted. The disease course did not seem typical of infectious gastroenteritis. The etiology of the liver damage and intra-abdominal edema was unclear. "Cuffing" may accompany acute viral hepatitis, heart failure, or cholangitis; however, none of these conditions was subsequently not confirmed, nor does any of them involve hemorrhagic diarrhea. Therefore, the medical history was re-examined in detail. The symptoms started abruptly. Epidemiological history was negative. The patient had not consumed anything unusual except for baked zucchini with bread. She took no medications. The physicians contemplated what they might have overlooked. The nurse who was taking care of the patient wondered if the issue could be related to the zucchini. Having spent more time with the patient, she learned that the zucchini tasted unusually bitter but that the patient did not consider this fact important enough to report. The nurse added that bitter zucchini should not be consumed, although she did not know why. Based on information obtained from ChatGPT and online resources, a diagnosis of cucurbitacin poisoning was proposed. From the third day onward, the patient was practically symptom-free, and aminotransferase levels normalized within 5 days. In cooperation with the Institute of Forensic Medicine and Toxicology in Prague, we planned to confirm the presence of cucurbitacin in the blood by gas chromatography. Unfortunately, the sample could not be found as the cleaning staff mistakenly considered it waste.

DISCUSSION Cucurbitacins are found in edible plants from the *Cucurbita* group. Plants produce cucurbitacins in response to environmental stress during growth. Their content decreases as the plant matures. Cucurbitacins are intensely bitter. The symptoms of poisoning may be dramatic. However, a fatal course is rare. Predominant manifestation includes vomiting, abdominal cramps, and diarrhea, may be accompanied by the hematemesis. Hypotension is common. Our patient experienced a typical clinical presentation of poisoning. A nonspecific but frequent finding is the presence of edema in the intestinal wall and adjacent intra-abdominal organs. There is no specific antidote, and treatment is symptomatic.

CONCLUSIONS This case report highlights that a properly taken medical history is crucial in the diagnostic process, especially in unclear situations.

Key words
cucurbitacin, poisoning, zucchini

2ND PLACE: NISHA GEORGE

Down the rabbit hole: a rare case of pyrexia of unknown origin

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INTRODUCTION The definition of pyrexia of unknown origin (PUO) has evolved over the years but the diagnostic challenges remain the same. Broadly speaking, the etiologies fall under one of the categories encompassing infection, inflammation, neoplastic process, or miscellaneous.

CASE DESCRIPTION We present a case of PUO in a 68-year-old, fit, and healthy man, who underwent extensive diagnostic evaluation during 3 months of hospitalization. The workup encompassed 90 blood tests, 8 blood cultures, 45 tests for 30 different infections, 7 blood films, 5 sputum and urine cultures, 2 extensive autoimmune screens, 2 bone marrow biopsies, 6 computed tomography scans, 8 chest X-rays, 2 echocardiography examinations, 1 positron emission tomography / computed tomography scan, 1 muscle biopsy, 10 multidisciplinary team discussions, and 1 magnetic resonance imaging scan. Finally, a rare diagnosis of hemophagocytic lymphohistiocytosis (HLH) with underlying intravascular lymphoma was made. HLH and intravascular lymphoma are both rare conditions on their own. Diagnosing them together requires high clinical suspicion and a stroke of luck. The learning points for this case are the importance of history taking and physical examination, the understanding and high clinical suspicion of HLH, and perseverance.

Key words
intravascular lymphoma, hemophagocytic lymphohistiocytosis, pyrexia of unknown origin

3RD PLACE: STEPHANIE WANG

My worst nightmare: a case of granulomatous encephalitis

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INTRODUCTION Human free-living ameba (FLA) infections are rare and often go unrecognized.

CASE DESCRIPTION A 74-year-old Caucasian woman from Canada was admitted due to a 2-week history of a rapidly evolving cerebellar syndrome. She had no history of immunosuppression and no prior exposure to significant infections. The physical examination showed bilateral horizontal nystagmus and slight right upper limb dysmetria, and was otherwise unremarkable. Routine bloodwork was normal except for slightly elevated C-reactive protein levels (15.4 mg/l). Initial cerebral computed tomography scan and magnetic resonance imaging (MRI) showed nonspecific cystic lesions in the right cerebellar hemisphere and vermis with surrounding edema and leptomeningeal involvement. Primary extracerebral neoplasia workup was negative. Control MRI conducted 5 days later showed progression of the lesions into the left cerebellum.

Lumbar puncture was performed and was not contributory. Brain biopsy was performed 3 weeks after the onset of initial symptoms

and histopathology showed numerous granulomatous inflammatory infiltrates and foci of acute inflammation with focal necrosis. No microorganisms were initially identified with immunohistochemistry. Extensive serological and cerebrospinal fluid (CSF) workups for infectious (eg, *Mycobacterium tuberculosis*, *Toxoplasma gondii*), inflammatory, and IgG4 diseases were all negative.

In addition to dexamethasone, the patient was empirically started on rifampin, isoniazid, pyrazinamide, and ethambutol in for presumed central nervous system tuberculosis. The treatment was complicated by drug-induced hepatitis. Since all final tests for *M. tuberculosis* were negative, all drugs were discontinued after 10 weeks. Three months after initial presentation, the patient was asymptomatic and MRI showed marked regression of cerebral lesions.

The patient was readmitted 4 months after initial presentation for acute onset aphasia, dysarthria, ataxia, and right hemiparesis. Cerebral MRI demonstrated multiple new cerebral and brainstem masses associated with surrounding vasogenic edema. Second brain biopsy was performed, following a rapid deterioration of the patient's state of consciousness. This time amoebic trophozoites were identified. With the help of the Centers for Disease Control and Prevention, a diagnosis of granulomatous amoebic encephalitis (GAE) caused by *Acanthamoeba* spp. was confirmed. The patient was started on combination therapy of IV pentamidine, trimethoprim-sulfamethoxazole, flucytosine, fluconazole, miltefosine, and azithromycin. Unfortunately, the patient's clinical status deteriorated 4 days after starting therapy. Considering the poor prognosis, palliative care was initiated and the patient died shortly thereafter.

DISCUSSION Acanthamoeba spp. is one of the 3 FLA most known to cause human infections, the other two being Naegleria fowleri and Balamuthia mandrillaris. These amebas are ubiquitous. However, it is often difficult to identify the source of these infections and risk factors for exposure are not always evident. Specifically, Acanthamoeba spp. can cause subacute-to-chronic encephalitis, called GAE. Analysis of CSF and cerebral imaging are usually abnormal in GAE, but nonspecific. GAE has high mortality rate and no specific therapy is known. However, early treatment was associated with improved survival, hence the importance of early diagnosis.

LESSONS TO BE LEARNED Early recognition of an FLA infection depends on increased awareness among clinicians, collaboration between various specialists, and help from reference laboratories. These microorganisms should be suspected in cases of rapidly progressing meningitis or granulomatous encephalitis, even without specific risk factors or exposition.

Key words
Acanthamoeba, free-living amebas, granulomatous amebic encephalitis

ORAL PRESENATIONS

ANNA OUŘADOVÁ

Adrenocortical carcinoma with mineralocorticoid hypertension histologically mimicking pheochromocytoma

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INTRODUCTION Adrenocortical carcinomas (ACCs) are very rare tumors occurring in middle-aged adults, more often in women. The clinical manifestation depends on the usually present cortisol overproduction. ACC associated with 11-deoxycorticosterone (11-DOC) overproduction presenting with severe mineralocorticoid hypertension has been rarely described. . ACC clinically or histologically mimicking pheochromocytoma is also infrequent.

CASE DESCRIPTION A 34-year-old healthy woman presented to the emergency department with abdominal pain in December 2022. A computed tomography scan revealed a bulky right adrenal tumor with retroperitoneal hemorrhage, tumor thrombus in the inferior vena cava, left-sided pulmonary embolization, and multiple pulmonary metastases. Ongoing bleeding necessitated selective embolization of the right adrenal artery.

Hypertension was present from the beginning (treatment with the following combination of antihypertensive drugs was started: perindopril, spironolactone, amiloride, amlodipine, doxazosin, and metoprolol), along with severe hypokalemia (2.4 mmol/l) despite extensive substitution. A core cut biopsy of the subpleural lesion was interpreted as a metastasis of pheochromocytoma. However, plasma and urine metanephrines were repeatedly negative, chromogranin A was only slightly elevated, and central hypocortisolism (adrenocorticotropic hormone, 1.33 pmol/l; cortisol, 92.8 pmol/l), hypoaldosteronism (aldosterone, 0.078 nmol/l) with unelevated renin (4.2 ng/l) and extremely elevated 11-DOC (60.7 nmol/l) predominated. Scintigraphy (meta-iodobenzylguanidine, tectrotyd) showed a physiological distribution of the radiopharmaceuticals. The histopathological findings of the subsequently extirpated lung metastasis were identical with those obtained in the core cut biopsy (cytokeratin-negative, strongly positive for synaptophysin, and positive for chromogranin A). The germinal variant and the next generation sequencing variant were negative.

Initial treatment with sunitinib had no effect. Following a second opinion of an international team of experts, the histopathological findings were reclassified as ACC (high incidence of necrosis, SF1 positivity, and GATA3 negativity). Subsequently, treatment with combination chemotherapy (cisplatin, doxorubicin, etoposide) and mitotane was started, with a partial response (regression of lung metastases).

DISCUSSION The presented case documents a discrepancy between the clinical picture (severe mineralocorticoid hypertension with secondary hypocortisolism) and the initial histopathological evaluation (pheochromocytoma). However, this discrepancy led to a further discussion with a consensus in favor of ACC. At the same time, a new treatment strategy was initiated, with a partial response so far.

CONCLUSIONS The presented case highlights the fact that the final diagnosis and treatment strategy are always a synthesis of clinical, laboratory, histopathological, and complementary findings. It shows the importance of a multidisciplinary approach and the value of a second opinion, especially in rare diseases.

Key words
11-deoxycorticosterone, adrenocortical carcinoma, mineralocorticoid hypertension, pheochromocytoma, secondary hypocortisolism

ANRI GERBER

Takayasu arteritis: a clot or not?

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INTRODUCTION Takayasu arteritis is a large vessel vasculitis that affects the aorta and its main branches. Involvement of the pulmonary arteries is rare and leads to pulmonary hypertension, right ventricular failure, and early death. We present the case of pulmonary hypertension due to left main pulmonary artery stenosis in a female patient with Takayasu arteritis.

CASE DESCRIPTION A 34-year-old woman with a confirmed diagnosis of Takayasu arteritis presented to our facility with a 4-month history of dyspnea, orthopnea, paroxysmal nocturnal dyspnea, and lower limb swelling. At presentation, she was normotensive and tachypneic, with conjunctival pallor and lower limb oedema. She had an elevated jugular venous pulse, a left parasternal heave, a palpable P2, and an audible tricuspid regurgitation. The patient's lungs were clear and her abdominal examination was unremarkable. Her blood results showed iron deficiency anemia, normal renal function, normal C-reactive protein levels, and normal erythrocyte sedimentation rate. She had biochemical features in keeping with a congested hepatopathy with derangement of her hepatocellular enzymes. Echocardiogram demonstrated a right ventricular systolic pressure of 100 mm Hg, severe tricuspid regurgitation, and moderate pulmonary regurgitation with a left ventricular ejection fraction of 70%. The ventilation-perfusion study showed uniform radio aerosol distribution throughout both lung fields on the ventilation study with uniform distribution of the radiopharmaceutical in the right lung field in the perfusion study. However, a unilateral whole-lung ventilation-perfusion mismatch was noted in the left lung field. Single photon emission computed tomography/computed tomography reconstructed images showed no significant structural lung disease that could explain this finding. Computed tomography pulmonary angiography of the lungs demonstrated significant stenosis of the left common carotid artery and left main pulmonary artery with a dilated right heart with contrast reflux in the inferior vena cava. The pulmonary trunk, right main pulmonary artery, lobar, and segmental pulmonary arteries were adequately opacified with contrast without filling defects. Our workup concluded with a normal forced vital capacity and forced expiratory volume in the first second to forced vital capacity ratio. The patient was managed medically with furosemide, IV iron dextran, and enalapril since an endovascular intervention was not possible due to the extent of the stenosis.

CONCLUSIONS This case highlights the need for routine screening for pulmonary hypertension in patients with Takayasu arteritis. A complete workup must be done to determine the etiological cause of pulmonary hypertension. Single photon emission computed tomography ventilation–perfusion studies are helpful in patients with unexplained pulmonary hypertension, as they are highly sensitive and specific for detecting chronic thromboembolic pulmonary hypertension. However, as seen in our patient, an acquired unilateral whole-lung ventilation-perfusion mismatch rarely indicates acute or chronic pulmonary embolism. This finding is in keeping with external compression of the pulmonary artery, inflammatory or granulomatous diseases, or, even more rarely, from luminal narrowing of the pulmonary artery, as seen in our patient with Takayasu arteritis. Determining the etiology of pulmonary hypertension in patients with Takayasu arteritis is critical to more effective management and to preventing early death.

Key words
pulmonary hypertension, Takayasu arteritis

INÊS MIRANDA

A mixed heart: a dual type of cardiac amyloidosis

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INTRODUCTION Cardiac amyloidosis (CA) has emerged as a previously underestimated cause of heart failure and mortality. Its two most common forms are amyloid light-chain (AL) amyloidosis and transthyretin amyloidosis (ATTR). Due to the distinct treatments and different prognoses, amyloid typing is crucial. Case descriptions of dual pathology are extremely rare, which poses challenges to treatment strategies.

CASE DESCRIPTION We present a case of an 85-year-old man who presented with symptomatic hypertrophic cardiomyopathy (class 1 heart failure). The examination raised a suspicion of cardiac amyloidosis. Diagnostic workup was conducted, which demonstrated small monoclonal immunoglobulin G λ spike and a visual score of 2 at bone scintigraphy. The diagnosis of cardiac amyloidosis was confirmed by endomyocardial biopsy. Amyloid typing by immunohistochemistry and mass spectrometry demonstrated concomitant deposition of transthyretin and λ light chains, and the diagnosis of dual cardiac amyloidosis was made. The patient was referred for a hematologic consultation in order to define the therapeutic plan.

DISCUSSION CA is increasingly recognized as a cause of heart failure. There are cardiac "red flags" that should raise a suspicion of this diagnosis. Clinical presentation can be similar in AL and ATTR amyloidosis despite them being mutually exclusive. In some cases, if certain criteria are met, an ATTR diagnosis can be made by bone scintigraphy; however tissue biopsy is mandatory when plasma celldyscrasia is present or when nuclear medicine results are inconclusive in the face of high clinical suspicion. Amyloid typing is vital due to different treatments and prognoses. Immunohistochemistry is the most widely used technique for amyloid diagnosis and typing, but mass spectrometry has recently become the gold standard for amyloid typing, due to its higher diagnostic accuracy. The presence of a dual pathology, that is, coexistence of ATTR and AL amyloidosis in a single patient, as presented in this case report, has been demonstrated extremely rarely. However, the advancement in diagnostic tools will likely increase the detection rate of such cases, which will help in developing treatment strategies.

CONCLUSIONS Clinicians must be aware of cardiac "red flags" that raise a suspicion of CA, since with the emergence of new therapeutic targets this diagnosis is no longer an academic exercise. Amyloid typing is crucial for establishing the correct diagnosis and choosing the most appropriate treatment. The identification of dual pathology revolutionizes the way we look at this complex disease and provides new research options for finding more sophisticated targeting treatments that improve patient survival and quality of life.

Key words
amyloid typing, cardiac amyloidosis, heart failure, light-chain, transthyretin

JOANNA RYMUZA

Coexistence of central diabetes insipidus and cerebral salt wasting syndrome

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INTRODUCTION The simultaneous occurrence of central diabetes insipidus and cerebral salt wasting syndrome (CSWS) is an uncommon clinical phenomenon. When it does manifest, its identification and management pose significant challenges, demanding meticulous patient monitoring.

CASE DESCRIPTION We present the case of a 38-year-old woman who underwent multiple neurosurgical procedures for the management of multiple chordomas within the central nervous system. She developed postoperative panhypopituitarism and was admitted to the department of internal medicine and endocrinology due to increasing fatigue, cough, and progressing confusion over the preceding 2 weeks. Initially diagnosed with pneumonia, she received appropriate treatment. Subsequently, the emergence of polyuria, hypernatremia, and increased osmolality led to the recognition of central diabetes insipidus. Despite the initial positive response to desmopressin, polyuria and pronounced natriuresis with hyponatremia occurred. A subsequent diagnosis revealed the overlap of cerebral salt wasting syndrome with the existing central diabetes insipidus.

The patient underwent a comprehensive therapeutic management, including 0.9% sodium chloride (NaCl) fluid administration, additional sodium correction with 3% NaCl, and the administration of fludrocortisone and desmopressin. This combined therapy resulted in the stabilization of sodium and volume levels.

CONCLUSIONS In cases of sodium balance disorders in patients with a history of neurosurgical procedures, a thorough and comprehensive differential diagnosis is essential.

Key words
central diabetes insipidus, cerebral salt wasting syndrome, hyponatremia

JOHN MCCORMICK

Acquired Gerbode defect following high velocity motor vehicle accident

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INTRODUCTION A Gerbode defect is a rare form of atrioventricular septal defect, which creates direct communication between the left ventricle and right atrium. While most cases are congenital, and therefore diagnosed in childhood, in some rare cases Gerbode defect has been described in adults following a cardiac surgery, infective endocarditis or blunt chest trauma.

CASE DESCRIPTION A 48-year-old man was transported to the emergency department following a high-velocity, single-passenger traffic accident. Initial examination showed multiple long bone and rib fractures, bilateral pneumothorax, pulmonary contusions, pericardial effusion, and a complex fracture of the right acetabulum. The patient underwent emergency surgical fixation of his long bone fractures and was admitted to the intensive care unit with a plan for interval complex pelvic surgery. Despite intravenous diuretic therapy, over the following days he developed a severe acute kidney injury, markedly deranged liver function tests, and significant peripheral oedema. On the eighth day postadmission the patient became acutely unstable with clinical features of pericardial tamponade and underwent emergency pericardiocentesis. Despite optimal medical therapy, the patient's condition continued to deteriorate with clinical evidence of significant congestion, hepatopathy, coagulopathy, thrombocytopenia, respiratory failure, and oligo-anuric renal failure which required renal replacement therapy. Transesophageal echocardiography demonstrated a large Gerbode defect with

resultant massive dilatation of the right heart chambers. Following an extensive discussion, the heart team chose to close the defect with a percutaneous approach, since surgery was deemed to be high risk. A 14-mm membranous ventricular septal defect occlusion device was deployed with initial technical success. The patient's clinical and biochemical parameters improved dramatically over the following days. On the eighth day postprocedure, he became acutely unstable with recurrent signs of right-sided heart failure. Computed tomography confirmed embolization of the occlusion device to the left pulmonary artery. The device was retrieved percutaneously with a snare and removed from the right femoral vein by vascular cut down. As the patient's coagulopathy and multiorgan dysfunction had improved significantly over the period since the implantation of the device, surgical repair was now believed to be a viable option. This was achieved using Sauvage and pericardial patches, and a tricuspid annuloplasty ring. Following interval surgical fixation of pelvic fractures, the patient was discharged, fully independent.

DISCUSSION This case demonstrates some of the difficulties with identifying and treating rare pathologies amidst multiple distracting and confounding diagnoses. To our knowledge, this is the first attempted percutaneous closure of a traumatic Gerbode defect. The reason for delayed device embolization is uncertain, but may relate to progressive necrosis of the injured tissue at the margins of the defect.

CONCLUSIONS We report a rare case of acquired Gerbode defect associated with blunt chest trauma. We describe a percutaneous approach to the closure of the defect, which ultimately did not produce a durable result, despite initial technical success.

Key words
blunt trauma, Gerbode defect, embolization, percutaneous closure, septal defect

LIVIA LONTAI

Rare cause of acute pancreatitis and hepatitis syndrome

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INTRODUCTION Auto-brewery syndrome is a rare and globally underdiagnosed condition caused by the microbiome's endogenous alcohol production. The clinical presentation is characterized by the symptoms of alcohol poisoning.

CASE DESCRIPTION Our patient was a 40-year-old man. His medical history included multiple hospitalizations due to acute edematous pancreatitis, attributed to alcohol consumption and dyslipidemia. Fluctuating blood fat levels were observed despite the patient's diet and fenofibrate therapy. Single plasmapheresis was also performed. A few months later the patient was admitted to a hospital because of hepatitis syndrome with extremely high gamma-glutamyl transferase levels. Abdominal ultrasound showed a diffuse hepatic lesion.

We conducted hepatological examination due to elevated liver enzyme activity, diffuse liver lesions, and fluctuating malaise. Viral serology, immune markers, and tumor markers were negative, but liver elastography indicated F3 fibrosis without steatosis. Additionally, the patient experienced nausea, headaches, speech difficulties, and altered behavior. Blood fat levels began to rise again. Detailed medical history mentioned no contact with any toxic agents. Total abstinence was confirmed by heteroanamnesis. Further tests ruled out enzyme defects. Due to unknown cirrhosis, we performed a core biopsy, which showed histological signs of alcoholic liver disease. There were no signs of a storage disorder.

Looking at both the clinical and histological results, we diagnosed the patient with auto-brewery syndrome. Since then, he has been on a strict diet and his complaints have significantly decreased, his laboratory values have normalized, and the fibrosis has regressed to a mild F1 status. After months of dieting, provocation tests with per os glucose, protein, starch, and white flour were performed. Higher exhalation values were detected only during the white flour test, but blood alcohol levels remained negative. According to the patient's diary, higher breathalyzer values appear as a result of starvation and stress. To confirm the diagnosis, a stool microbiome analysis was performed, and it showed overgrowth of *Proteobacteria*, *Gammaproteobacteria*, *Enterobacterales*, and *Enterobacteriaceae* families, which are capable of alcoholic fermentation.

DISCUSSION Auto-brewery syndrome is a disease of ethanol intoxication caused by endogenous fungal or bacterial fermentation, which in turn is caused by a disruption of the normal micro- and mycobiome. The clinical picture is characterized by complaints of continuous drunkenness and hangovers, and long-term damage caused by endogenous alcohol. In our case, with the help of a strict diet and regularly measured alcohol values, both the laboratory findings, the fibrotic transformation of the liver, and the patient's complaints were significantly reduced.

CONCLUSIONS Auto-brewery syndrome is a rare, but globally underdiagnosed condition. Our case shows the diagnostic difficulties of this situation, which can lead to severe health and quality of life issues, especially without treatment.

Key words
alcoholic liver cirrhosis, auto-brewery syndrome, dyslipidemia, hepatitis, pancreatitis

MARIA JOSÉ ORREGO

Mycoplasma-induced rash and mucositis: a rare condition in a Colombian patient

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INTRODUCTION *Mycoplasma pneumoniae* infection typically involves the respiratory tract. *Mycoplasma*-induced rash and mucositis (MIRM) is an unusual extrapulmonary manifestation characterized by severe mucositis and several types of skin lesions. Here, we present a case of a Colombian patient with MIRM, emphasizing the diagnostic challenges, rarity of this condition, and atypical presentation of a relatively common infection.

CASE DESCRIPTION A 28-year-old woman with unremarkable medical history presented with upper respiratory symptoms, cough, fever, bilateral red eyes, photophobia, and painful oral and genital ulcers that at this point had been lasting for 15 days. On admission, bilateral red eyes with desquamative ulcers in both upper eyelids and signs of mucositis were present, with multiple ulcers of the lips and oral cavity, covered by a bloody crust. Lacerations and friability were observed in the vaginal mucosa. Laboratory workup showed leukocytosis and elevated inflammatory markers. Blood cultures were negative. A computed tomography (CT) scan of the chest showed ground glass opacities in the right middle lobe. A few days later, the patient developed a rash on her chest and back. The ophthalmologist diagnosed her with bilateral anterior uveitis, leading to an initial diagnosis of Behçet disease, due to mucosal ulcers and uveitis. She was started on prednisolone and azathioprine. Two weeks later, the patient presented to our hospital with productive cough, worsening of oral ulcers, paroxysmal nocturnal dyspnea, and orthopnea. She was found hypoxemic. New laboratory exams showed pancytopenia, and a follow-up chest CT showed left lower lobe consolidation. Due to unclear Behçet's disease diagnosis, azathioprine was stopped over hematological toxicity concerns. Oral

acyclovir was given empirically without improvement of the ulcers. Echocardiogram showed a new-onset biventricular heart failure with ejection fraction of 34% and global hypokinesia. Subsequently, acute myocarditis was confirmed on cardiac magnetic resonance imaging. Lip and tongue biopsies demonstrated inflammatory changes and tested negative for vasculitis, viral cytopathic changes, fungi, and herpes simplex virus. Given the mucosal and lung involvement, serological testing for *M. pneumoniae* was requested, which yielded positive (immunoglobulin [Ig] M >200 U/ml; IgG, 26.4 U/ml reactive). Simultaneously, a FilmArray Pneumonia panel test was performed on a sputum sample, detecting both *M. pneumoniae* and *Pseudomonas aeruginosa*. The cell count recovered after discontinuation of azathioprine. The patient was treated with azithromycin and piperacillin-tazobactam, resulting in complete resolution of the symptoms. She was discharged with optimal medical therapy for heart failure.

DISCUSSION MIRM is defined by prominent mucositis involving 2 or more sites, sparse vesiculobullous or targetoid cutaneous eruption (affecting less than 10% of the body surface area) and clinical/laboratory evidence of *M. pneumoniae* infection. On the other hand, heart involvement associated with *M. pneumoniae* is uncommon, but there are case reports of pericarditis, myocarditis, and conduction abnormalities.

CONCLUSIONS A severe mycoplasma infection explained the patient's multiple extrapulmonary manifestations including uveitis, myocarditis, and mucositis. Furthermore, pancytopenia caused by azathioprine toxicity predisposed her to bacterial superinfection with *P. aeruginosa*.

Key words
anterior uveitis, mucositis, myocarditis, *Mycoplasma pneumoniae*, oral ulcers

MICHAŁ OLEJARZ

Adrenal gland tumors: so common, yet still surprising. A rare case of alveolar echinococcosis of the adrenal gland

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INTRODUCTION An adrenal incidentaloma is a common endocrine condition. The majority of adrenal lesions are benign, non-secreting adenomas. However, the differential diagnosis is very broad. In this study, we present the case of a patient initially diagnosed with dopamine-secreting pheochromocytoma, which eventually turned out to be a parasitic infestation caused by *Echinococcus multilocularis* (alveolar echinococcosis [AE]). Tests for parasitic infestations are not routinely performed in patients with adrenal tumors, nor mentioned in any guidelines or recommendations.

CASE DESCRIPTION A 66-year-old woman with type 2 diabetes, angina pectoris, chronic gastritis, hypertension, Parkinson's disease, and iatrogenic hypothyroidism after thyroidectomy underwent imaging diagnostics because of an episode of hematuria. Computed tomography (CT) of the abdomen, followed by magnetic resonance imaging (MRI), revealed a tumor sized 64 mm × 74 mm × 70 mm with a density of over 25 Hounsfield units, likely arising from the right adrenal gland, infiltrating the hilum of the liver and the right crus of the diaphragm. Hormonal assessment showed elevated methoxytyramine excretion in 24-hour urine collection. The patient was diagnosed with dopamine-secreting pheochromocytoma and referred for surgery. Histopathological examination showed chronic inflammation with no signs of a neoplastic process. On follow-up MRI, tumor regrowth was observed, prompting a referral to our department for diagnostics.

We observed no abnormal hormonal activity. Routine laboratory testing revealed mild anemia and an increased eosinophil count. CT of the abdomen showed a tumor measuring 79 mm × 61 mm × 90 mm, localized in the adrenalectomy bed, expanding to the liver, and infiltrating the portal vein, hepatic veins, common bile duct, inferior vena cava, crus of the diaphragm, and upper pole of the right kidney. Reassessment of the surgical specimen was suggestive of a parasitic infestation. Consequently, we performed immunodiagnostic tests, which were unequivocally positive for *E. multilocularis*.

Increased 3-methoxytyramine urine excretion was a false positive result caused by levodopa treatment. Based on histopathology and immunodiagnostic test, the patient was diagnosed with alveococcosis in an advanced, unresectable stadium (P4N1M1) and started on chronic albendazole treatment.

DISCUSSION AE is a deadly and devastating disease that is characterized by a progressive, infiltrative growth of a tumor-like lesion, which mimics a malignant, metastasizing tumor. It mostly occurs in the northern hemisphere—in Asia (especially China) and America. AE is most commonly localized in the liver and can infiltrate the neighboring organs. However, AE presenting as a lesion primarily localized in the adrenal gland is exceptionally rare. The parasitic tumor in our patient had similar features to the ones described in 9 cases previously reported in the literature.

CONCLUSIONS A lack of awareness of AE can result in a significantly delayed diagnosis or misdiagnosis, which could have detrimental consequences for the patient's health. Thus, we suggest that screening for AE should be performed in a selected group of patients with hormonally inactive, indeterminate adrenal lesions of the right adrenal gland and suspicious imaging features that show infiltration of the liver, diaphragm, or vena cava and are associated with peripheral blood eosinophilia.

Key words
adrenal tumor, alveococcosis, differential diagnosis, *Echinococcus multilocularis*, incidentaloma

NICOLA WILLS

Candida pericarditis with tamponade: a case report of a complicated malignant esophago-pericardial fistula

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INTRODUCTION Isolation of *Candida* species from the pericardial space is a rare manifestation of invasive candidemia that has a high mortality rate. Here, we describe the case of a middle-aged man presenting in cardiac tamponade with paroxysmal atrial flutter and seeding of *C. glabrata* into the pericardial space secondary to a malignant esophago-pericardial fistula.

CASE DESCRIPTION A 61-year-old repairman and long-term cigarette smoker presented to our tertiary facility in cardiac tamponade, with hemodynamic instability aggravated by an atrial flutter, after being managed for heart failure at a local district hospital. After an immediate transfer to the cardiac unit, echocardiography showed a large, predominantly posterior and fibrinous pericardial effusion with right ventricular diastolic collapse. Following emergency pericardiocentesis, sinus rhythm and hemodynamic stability were restored. Given the raised adenosine deaminase activity (47 U/l) and exudative chemistry in the pericardial fluid, the patient was initially treated for *Mycobacterium tuberculosis* infection. However, pericardial fluid culture revealed a growth of *C. glabrata*. Sputum and pericardial fluid GeneXpert (polymerase chain reaction) testing

for *M. tuberculosis* was also negative. With this unexpected fungal growth, an interrogation for possible host immune defects or tissue barrier interruptions was conducted. HIV ELISA testing was negative and neutrophilia was demonstrated on blood testing. With weight loss and smoking history as red flags, computed tomography scan of the chest, abdomen, and pelvis was conducted, revealing circumferential thickening of the thoracic esophagus, measuring 110 mm in length, with no clear plane of separation from the pericardium. Low-attenuating mediastinal and epigastric necrotic lymph nodes were also seen. On revisiting the patient's history along with his wife, it came out that he had been suffering from dysphagia with solid foods for the last 6 months. Gastroscopy was conducted, revealing unilateral external midesophageal compression and a 5-mm defect with macroscopically friable and necrotic borders at 30 cm from the incisors. On biopsy, a moderately differentiated nonkeratinizing squamous cell carcinoma with features suspicious for lymphovascular invasion was confirmed. Esophageal biopsies also cultured *C. glabrata*. A stent was placed over the esophageal defect and the patient received 2 weeks of inpatient micafungin therapy. Connection to palliative and oncology care was initiated during his admission and continued after his hospital discharge.

DISCUSSION Invasive candidiasis is an increasing nosocomial concern that has a high mortality rate. Host immune defects and tissue barrier interruptions are major risk factors. In this case, pericardial inoculation with *C. glabrata* occurred secondary to a malignant esophago-pericardial fistula with invasive infection likely hastened by smoking and cancer-related immune aberrations. Reports of *C. glabrata* pericardial disease are scarce. In our patient, atrial irritation with a flutter worked in concert with tamponade physiology to cause the initial emergency clinical presentation.

CONCLUSIONS Cardiac tamponade is a clinical diagnosis whose recognition it is life-saving. Invasive candidemia is usually associated with indwelling catheters, prior antibiotic exposure, immunodeficiency, and surgery/tissue injury. Reports of *C. glabrata* pericardial disease are scarce. Isolating an atypical organism from an unusual space should prompt a search for how or why that organism got there.

Key words
atrial flutter, *Candida glabrata*, cardiac tamponade, esophago-pericardial fistula, pericarditis

PETER KROMKA

Myopericarditis in an ultra-endurance runner: a case of late-onset systemic lupus erythematosus

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INTRODUCTION Systemic lupus erythematosus (SLE) is a chronic, multisystem disease of autoimmune etiology with a greatly varying clinical course. A wide spectrum of clinical presentations constitutes a daunting diagnostic challenge. Although SLE is relatively rare in men, it tends to be more serious in those patients. Similarly, age has been found to modify disease trajectory and outcomes significantly. Late-onset SLE is uncommon and often associated with diagnostic delay leading to increased morbidity and mortality.

CASE DESCRIPTION A 51-year-old Caucasian man, an amateur long-distance runner, was admitted to the hospital because of fever, fatigue, and arthralgia. These symptoms were preceded by recurrent episodes of an itchy rash on his lower legs, that would appear shortly after physical activity, resolving spontaneously within a few days. Initial laboratory workup showed mild inflammatory syndrome. Intravenous fluids and antipyretics were given, but the fever persisted. A thorough investigation failed to demonstrate infectious, hematologic or neoplastic conditions. Imaging studies were negative except for a computed tomography scan of the chest, abdomen, and pelvis, which identified pleural effusion. We made a presumptive diagnosis of SLE. The autoantibody testing and the presence of antinuclear antibodies and anti-double strand DNA antibodies confirmed our suspicion. After a brief improvement following the treatment, the patient developed palpitations and chest pain. A diagnosis of myopericarditis complicating SLE was based on echocardiography findings and a moderate elevation of cardiac biomarkers. Positron emission tomography/computed tomography showed increased tracer uptake in the myocardium and ascending aorta. The patient received steroid pulse therapy, which led to a marked clinical improvement, and echocardiography confirmed a complete resolution of pericardial effusion with a reversal of left ventricular dilatation.

DISCUSSION SLE is an evolving disease and can affect nearly any organ system. The female preponderance has been long considered a hallmark of SLE. Consequently, less is known about the clinical features of SLE in male patients. Subtle skin or joint involvement among men may remain unnoticed. By the time of diagnosis, potentially life-threatening and disabling complications may be present. We assumed that the patient's cutaneous manifestation was consistent with a clinical picture of exercise-induced vasculitis (EIV), a small vessel vasculitis frequently unrecognized or mistaken for other disorders. EIV is widely regarded as a benign and self-limiting condition. In the present case, the patient returned to his previous sports activity 6 months after treatment. There has been no recurrence of this condition over the current follow-up period. This observation suggests that the occurrence of EIV could be related to the disease activity and may potentially carry useful information.

CONCLUSIONS Diagnosis of late-onset SLE is often delayed due to ambiguous clinical signs and symptoms. Exercise-induced vasculitis exhibits a characteristic clinical picture. Physicians need to be aware of this clinical condition and the possibility of underlying disease should lower the threshold for further investigation.

Key words
exercise-induced vasculitis, systemic lupus erythematosus

ROMAIL SHAHID

Mixed connective disease co-existence with refractory TTP: rare coexistence, diagnostic dilemma, and therapeutic delay lead to worse prognosis in resource-limited heath care areas

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CASE DESCRIPTION Thrombotic microangiopathy includes thrombotic thrombocytopenic purpura (TTP) and hemolytic uremic syndrome. TTP has rare coexistence with mixed connective tissue disease (MCTD) and is difficult to diagnose in a pre-existing flare of MCTD. Clinical pentad of TTP is not always fulfilled. We report a case of a 28-year-old woman with MCTD, who developed refractory secondary TTP and was admitted with flare of MCTD. Rare clinical scenarios, such as TTP-MCTD refractory to conventional first-line management, in resource-limited areas are not only difficult to diagnose, but also face delays in management. As with primary TTP, we emphasize that for secondary TTP the clinicians should have a lower suspicion threshold for therapeutic success after early diagnosis and management. In this case, the diagnostic challenge was differentiate microangiopathic hemolytic anemia (MAHA) from autoimmune Coombs-positive hemolysis. Positive Coombs test result directed attention towards autoimmune Coombs-positive hemolysis and delayed the diagnosis of TTP. Persistent thrombocytopenia, no response to treatment, development of neurological symptoms

over some time, and schistocytes on peripheral film predicted MAHA. Over the time, the pentad of TTP was fulfilled. The patient's fluctuating irritability posed a demanding question, leading us to consider if it was cause by hospital psychosis, steroid psychosis or central nervous system involvement due to TTP. This case report should encourage to keep in mind the following: 1) to prevent worst outcomes, a clinician should stick to the worst diagnosis; 2) since the clinical pentad of TTP fulfills over a period of time, it must be dealt with aggressively once TTP is diagnosed, even if the pentad is not complete at the time of suspicion.

Key words
mixed connective tissue disease, microangiopathic hemolytic anemia, thrombotic thrombocytopenic purpura, schistocytes

ROZAN YAHYA

Looks, sounds, and smells like lymphoproliferative disease, and potentially is just as lethal in the elderly

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INTRODUCTION We describe an elderly patient with chronic active Epstein–Barr virus (CAEBV) infection. It is a rare, potentially life-threatening disorder, which may evolve into a systemic organ disease and hyperinflammatory state, such as hemophagocytic lymphohistiocytosis (HLH) and liver failure. Lymphoproliferative disease (LPD) is one of its late complications. The etiology of the disease is not well understood, but it is associated with clonal proliferation of EBV-infected T or natural killer cells. Rapid diagnosis is crucial and poses a true challenge, especially in the elderly, as this condition is considered a childhood disease. Our patient may be the oldest patient diagnosed with CAEBV infection.

CASE DESCRIPTION An 82-year-old patient presented with recurrent fever, decreased appetite, weight loss, and splenomegaly that had been ongoing for the past 10 months, raising initial suspicion of LPD. Despite extensive investigations conducted during previous hospitalizations, including imaging studies and histological examinations of lymph nodes, definitive diagnosis was not obtained, highlighting the complexity of diagnosing CAEBV infection in older adults. The patient's medical history included idiopathic thrombocytopenic purpura and cyclic neutropenia. Laboratory tests showed leukocytosis and elevated C-reactive protein levels. Imaging studies showed lymphadenopathy, splenomegaly, and a new hepatic lesion. Differential diagnoses included LPD, solid cancer with paraneoplastic presentation, infectious diseases, autoimmune disorder, and CAEBV infection. Given the lack of evidence of solid malignancies, LPD, and other infectious or autoimmune diseases in previous imaging and histological examinations performed in recent months, we conducted serology testing for EBV and EBV polymerase chain reaction (PCR) in the blood. PCR for EBV DNA was elevated (15600 IU/ml). Results of the previously performed axillary lymph node biopsy were revised, and were found to be positive for EBER CISH (Epstein–Barr Early RNA) staining, supporting the diagnosis of CAEBV infection. Diagnostic criteria for CAEBV disease (based on recent guidelines) include symptoms resembling persistent or recurrent infectious mononucleosis that last for more than 3 months, detection of an increased number of EBV genomes in peripheral blood and/or affected tissues (≥10 000 IU/ml of EBV DNA using PCR; EBER in situ hybridization is performed for the detection of EBV-positive cells in tissue), detection of EBV-infected T or natural killer cells in peripheral blood and/or affected tissues, and chronic illness that cannot be explained by other known diseases. In this case, all criteria were met. Additionally, our patient exhibited features suggestive of HLH (ferritin, 654 ng/ml; triglycerides, 245 mg/dl; soluble interleukin 2 receptor, 5809 U/ml), emphasizing the systemic nature of CAEBV and its potential for life-threatening complications. The only curative therapy is hematopoietic stem cell transplantation. Considering the patient's age and performance status, alternative management strategies were pursued. A combination of dexamethasone and etoposide was initiated for the diagnosis of CABEV-related HLH, leading to a remarkable clinical improvement and resolution of symptoms. Follow-up assessment revealed undetectable EBV DNA levels and no evidence of recurrence.

CONCLUSIONS This case underscores the importance of considering CAEBV in the differential diagnosis of prolonged infectious mononucleosis symptoms, accompanied by evidence of lymphadenopathy and splenomegaly. While CAEBV primarily affects children, it should also be considered in the elderly population. Early recognition and appropriate management are paramount in mitigating the risk of severe complications and improving patient outcomes.

Key words
chronic active Epstein–Barr virus, elderly, Epstein–Barr virus, lymphoma

SARAH AL QASSIMI

A puzzling prelude: bowel perforation leading to a diagnosis of atypical familial Mediterranean fever in a young patient

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INTRODUCTION Familial Mediterranean fever (FMF) is the most common autoinflammatory syndrome. Historically, it affected populations in the Mediterranean region, but is now a borderless disease due to the frequency of global migration. The underlying pathophysiology of FMF is mediated by MEFV gene mutations. Manifestation of the disease is characterized by recurrent episodes of fever and systemic inflammation, that spontaneously resolve. Our patient illustrates a case of FMF that is atypical in presentation in comparison to known literature.

CASE DESCRIPTION A 31-year-old Emirati woman presented with recurrent fever associated with a sore throat. She was initially diagnosed with tonsilitis and treated with antibiotics. Two days later, she developed epigastric abdominal pain, which became diffuse and increased in severity. It was associated with recurrent fever. The patient had a history of episodes of recurrent tonsilitis that would spontaneously resolve since her late twenties. She had no previous surgical history, personal history of arterial/venous thromboses or miscarriages. On physical examination, the patient was febrile, but otherwise vitally stable. Her abdominal examination showed exquisite tenderness and rebound tenderness of the right lower quadrant, and a positive McBurney sign. Laboratory work up showed elevated white blood cell count and C-reactive protein levels , but normal complete blood count, lactic acid, lipase, and liver function tests. Computed tomography of the abdomen and pelvis showed no appendicitis, but was significant for free air, suggestive of perforated hollow viscus of the duodenum and/or right colon. The patient underwent emergent exploratory laparotomy with intraoperative findings of loculated gas in the mesentery around the second and third parts of the duodenum, and a sealed perforation of the proximal transverse colon at the hepatic flexure, due to the presence of duodenal adhesions. She underwent a right hemicolectomy with no complications. Tissue biopsy of the resected colon showed findings consistent with an ischemic bowel with no evidence

of granuloma, malignancy or amyloidosis. The patient underwent hypercoagulability and autoimmune workup that was positive only for a single positive lupus anticoagulant. Through the diagnosis of exclusion, a suspicion of FMF was raised and the patient underwent MEFV gene testing, which reported that "4 variants were identified and at least 3 were interpreted as heterozygous alterations with uncertain significance." The combination of the patient's ethnicity, clinical manifestations, and abnormal gene testing was deemed to be consistent with FMF and she was started on colchicine. She continues to be comply with the medical therapyand her FMF is clinically quiescent.

DISCUSSION In addition to the absence of the usual FMF symptoms, our patient demonstrated several features atypical for the disease. These include age (older than the usual FMF population), female sex (men are affected more often), absence of family history, and the lack of amyloid staining on biopsy. To our knowledge, our patient is the first reported case of FMF presenting with bowel perforation.

LESSONS TO BE LEARNED This case sheds light on the clinical diversity of FMF in non-Mediterranean populations and the adjunct role of genetic testing in atypical and silent cases. Vigilance in searching for these cases can potentially prevent long-term complications through initiating early therapy.

Key words
acute abdomen, bowel perforation, familial Mediterranean fever

SERGIO BUENO

Dasatinib-induced chylothorax in a patient with chronic myeloid leukemia

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INTRODUCTION Dasatinib is a tyrosine kinase inhibitor used to treat Philadelphia chromosome-positive chronic myeloid leukemia (CML). Some adverse effects of dasatinib use are skin rash, pancytopenia, and pleural effusion. Although the last is a common adverse effect, chylothorax is extremely rare. Its pathological mechanism and management are unknown.

CASE DESCRIPTION We present a case of a 49-year-old man with a 3-year history of CML. Presence of the *BCR/ABL* p210 transcript was detected on molecular testing and Philadelphia chromosome positivity was confirmed on flow cytometry. The patient was treated with dasatinib, which resulted in complete remission of CML. He also presented with mild hypercholesterolemia. After 2.5 years of dasatinib use, the patient developed a dry cough, weight loss, and exertional dyspnea. Computed tomography scan of the chest showed moderate bilateral pleural effusion. Echocardiography was normal. Two months later, progression of the effusion was observed, prompting drainage via a right pleural drainage tube, yielding 1200 ml of milky fluid. Pleural fluid analysis was compatible with chylothorax. The patient had no history of possible causes of chylothorax such as trauma, thoracic surgery or malignant thoracic lesions, leading to suspicion of dasatinib-induced chylothorax. Therefore, dasatinib was discontinued and imatinib 400 mg per day was initiated. During the 16 days of hospitalization, a diet based on light-chain fatty acids was prescribed and during that time pleural drainage gradually decreased without complications. The patient was discharged with pleural drainage still in place. In outpatient follow-ups, there was a gradual decrease in pleural drainage to values below 100 ml every 24 hours and after 4 months the pleural drainage tube was removed. Two months after pleural drainage removal, no recurrence was observed.

DISCUSSION Despite various heterogeneous etiologies of the development of chylothorax, dasatinib is the only drug known to be associated with this adverse effect. However, although some theories are recognized, the production mechanism is not well understood. Some observed risk factors for pleural effusion development include age, heart and/or autoimmune disease, advanced disease, hypertension, hypercholesterolemia, skin rash, and lymphocytosis. Dasatinib dosage should also be considered a risk factor for pleural effusion. However, while this explains pleural effusion production, it does not explain the presence of chylothorax without thoracic duct injury, hence the relationship remains unknown. This adverse effect of dasatinib use is extremely rare, with only 15 reported cases to date. Regarding treatment, there is insufficient literature to establish a therapeutic algorithm. In our case, drug discontinuation along with dietary changes led to a clinical response and resolution.

CONCLUSIONS Dasatinib-induced chylothorax is a very rare entity with limited literature. Therefore, case reports like this one set a precedent to support therapeutic management decisions in future cases and to stimulate diagnostic suspicion in patients with similar presentations.

Key words
adverse effect, chronic myeloid leukemia, chylothorax, dasatinib pleural effusion

SILE TOLAND

Recurrent hemoptysis and lung ground glass opacity in a 36-year-old woman

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CASE DESCRIPTION We describe a case of catamenial hemoptysis secondary to thoracic endometriosis syndrome in a 36-year-old woman who presented with moderate-volume hemoptysis and a right upper lobe (RUL) infiltrate visible on chest X-ray, which occurred on the first day of her menstrual cycle. She had been experiencing a metallic taste during her periods for the past 3 months. The following month, on the first day of her menses she again experienced significant hemoptysis and a computed tomography of the chest performed at this time showed dense area of ground glass change in her RUL, consistent with pulmonary hemorrhage. Bronchoscopy found mild clotting and old blood in the RUL, and endobronchial biopsy confirmed subepithelial hemorrhage. Blood tests showed normal coagulation, platelet count, and negative results for vasculitis and connective tissue diseases. This pattern of symptoms persisted for the following 6 months. The case was discussed at a multidisciplinary meeting, where it was decided that the clinical presentation, radiological findings, and timing of symptoms are consistent with catamenial hemoptysis, likely associated with endobronchial endometriosis. The patient's treatment has been complicated, as she wished to conceive; theferore, she was unwilling to commence hormonal treatment at this time. However, counselling has been provided regarding the potential dangers of more significant hemoptysis and the patient has been reviewed by a consultant obstetrician.

DISCUSSION Catamenial hemoptysis is a rare and intriguing condition associated with endometriosis, marked by recurrent lung or airway bleeding synchronized with the menstrual cycle. While infrequent, it poses a substantial diagnostic and therapeutic challenge. The precise mechanisms behind catamenial hemoptysis remain partially understood, with the leading theory involving ectopic endometrial tissue implanting in the thoracic cavity and bleeding under hormonal influences. Diagnosis requires comprehensive evaluation, including radiological imaging, bronchoscopy, and hormonal assessments to identify the source of the bleeding and confirm the diagnosis. In terms of management, hormonal therapy aimed at suppressing estrogen production may be considered. Other op-

tions include gonadotrophin releasing hormonal agonists or oral contraceptives. Surgical intervention, usually performed through video-assisted thoracoscopic surgery. allows for direct visualization and biopsy of the pleural cavity. It can be both diagnostic and therapeutic, and it enables the removal or ablation of ectopic endometrial tissue. In cases of significant lung involvement, lung resection may be considered. The positive outcomes observed in many cases underscore the significance of early recognition and intervention. However, due to the limited number of reported cases, further research is necessary to clarify the underlying pathophysiology, and refine diagnostic and therapeutic strategies. Developing diagnostic criteria and less invasive treatment options could enhance our ability to manage this rare condition effectively. Raising awareness and promoting collaboration among healthcare professionals are essential for improving patient outcomes and expanding our understanding of catamenial hemoptysis.

Key words
computed tomography, endometriosis, female, hemoptysis, lung

TARIQ RAMTOOLA

Plasmapheresis as a bridge to thyroidectomy in a patient with severe type 2 amiodarone-induced thyrotoxicosis with concurrent ischemic cardiomyopathy and refractory ventricular tachycardia

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INTRODUCTION Amiodarone is a widely used and highly effective antiarrhythmic drug. However, 5% of patients will develop amiodarone-induced thyrotoxicosis (AIT). This can either be self-limiting, following amiodarone cessation, or require treatment with antithyroid drugs. If thyrotoxicosis persists or in the presence of left ventricular dysfunction, salvage thyroidectomy must be considered. In patients with hemodynamic and cardiac instability, plasmapheresis is an effective way of rapidly removing excessive thyroid hormones from the circulation to stabilize the patient preoperatively. Our case also highlights the importance of recognizing AIT even after discontinuation of amiodarone.

CASE DESCRIPTION A 64-year-old man with ischemic cardiomyopathy and implantable cardiac defibrillator (ICD) presented with symptoms of thyrotoxicosis. He was found to be dysrhythmic with new atrial fibrillation (AF) and ventricular tachycardia (VT). Amiodarone therapy was stopped 3 months earlier. Thyroid profile was grossly deranged with negative thyroid autoantibodies. A diagnosis of type 2 AIT was made. Despite aggressive and maximal medical therapy, the patient remained thyrotoxic with persistent unstable cardiac arrhythmia. Multi-disciplinary evaluation favored emergency thyroidectomy. He underwent plasmapheresis with excellent reduction in T4 levels, followed by successful thyroidectomy. All antithyroid drugs were stopped and amiodarone was restarted. Pathology results confirmed AIT. The patient remains well and arrhythmia-free at 4 months follow-up.

DISCUSSION Amiodarone has a long half-life and patients can present with AIT several months after treatment discontinuation. Clinical assessment may reveal overt thyrotoxicosis, cardiac decompensation or new dysrhythmia. Diagnosis is based on abnormal thyroid profile and can be supported by radiology. Type 1 AIT occurs in patients with underlying thyroid disease because of excessive thyroid hormone production and release by autonomous areas of the gland. Thyroid autoantibodies may be positive and it responds well to antithyroid drugs. Type 2 AIT is an inflammatory and destructive process leading to excessive release of free thyroid hormones into the circulation. It can either be self-limiting or require steroid therapy. Decision to continue amiodarone must be made considering the patient's cardiac status and in conjunction with cardiology input. If medical therapy fails, thyroidectomy is a viable option and this can be supported by plasmapheresis for preoperative stabilization.

CONCLUSIONS This case highlights the importance of vigilance for AIT in patients with a history of amiodarone use, even after drug discontinuation. In patients with cardiac instability, prompt diagnosis and treatment should be initiated to restore euthyroidism. Multispecialty input is often required in complex cases. Thyroidectomy is recommended if medical therapy is ineffective. Plasmapheresis is a safe and effective bridge to surgery.

Key words
amiodarone, plasmapheresis, thyrotoxicosis

UGUR ARZU KULU

Case report of atypically presenting Lyme disease

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CASE DESCRIPTION Lyme disease is a zoonotic infectious disease caused by spirochetes called *Borrelia burgdorferi*. Diagnosis is based on history, symptoms, and a 2-step serologic test. Positive immune serology is confirmed by Western blot. Clinically, the disease usually affects the skin, nervous system, musculoskeletal system, and heart. It can be classified as stage 1 (early localized), stage 2 (early disseminated) or stage 3 (late persistent). Although antibiotics are used in the treatment, most patients progress to the chronic stage. Autoimmune hemolytic anemia is a disease characterized by the breakdown of erythrocytes and anemia resulting from the patient producing antibodies against their own erythrocytes. This condition, which is idiopathic in most patients, can be secondary to some autoimmune or infectious diseases, and hematologic/nonhematologic malignancies. There are no studies that clearly demonstrate the relationship between Lyme disease and secondary anemia.

In this case report, we describe a case of Lyme disease presenting with autoimmune hemolytic anemia, thrombocytopenia, and mononeuropathy.

Key words
autoimmune hemolytic anemia, *Borrelia burgdorferi*, Lyme disease

VAIBHAV AGARWAL

An unusual case of hypovolemic shock: *Strongyloides* hyperinfection syndrome

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INTRODUCTION Patients presenting to the emergency department with symptoms such as nausea, vomiting, diarrhea, and abdominal pain can rapidly escalate to a hypovolemic shock—a condition with high mortality, if not promptly diagnosed and treated. This case emphasizes the critical nature of undetected parasitic infections and the dangerous overuse of medications, such as nonsteroidal anti-inflammatory drugs (NSAIDs) and steroids, highlighting their significant impact in this particular case.

CASE DESCRIPTION A 55-year-old farmer presented to the emergency department suffering from nonbilious vomiting (10–12 episodes daily) for 7 days, intermittent loose stools (2–3 episodes), and occasional crampy abdominal pain. He was drowsy, pale, dehydrated, and had a toxic look. Examination showed cold peripheries, a feeble pulse, and a dry tongue. Blood pressure was 80/50 mm Hg, pulse rate was 124/min, and the respiratory rate was 20/min. His chest was clear and his abdomen was soft with normal peristaltic sounds. At the hospital, immediate treatment included intravenous fluids, proton pump inhibitors, and antiemetics. Arterial blood gas showed acute metabolic alkalosis with respiratory compensation, hyponatremia, and hypokalemia, likely from vomiting and diarrhea. Further examination revealed the patient's long-term use of NSAIDs and steroids for bilateral knee osteoarthritis. He exhibited bilateral pitting edema, moderate pallor, and lesions suggestive of tinea cruris and ungual onychomycosis. Additional findings ruled out cardiac issues, external blood loss, and significant liver or kidney problems, but indicated dimorphic anemia with iron and vitamin B_{12} deficiency, low albumin and globulin levels, and hypocalcemia. Normal 8 AM serum cortisol and adrenocorticotropic hormone levels ruled out adrenal insufficiency. Normal antitissue transglutaminase antibody levels excluded celiac disease. Urine examination showed no proteinuria. Initial stool examinations failed to identify pathogens, but a third test revealed *Strongyloides stercoralis* filariform larvae. Computed tomography enterography revealed skip lesions in the duodenum and jejunum. Colonoscopy found diffuse mucosal erosions in the caecum and colon. Respective biopsy showed villous atrophy, dense chronic inflammatory infiltrate, and parasitic larvae resembling *Strongyloides* in the crypts. A provisional diagnosis of chronic malabsorption syndrome and protein-losing enteropathy due to *Strongyloides* hyperinfection, exacerbated by chronic corticosteroid use for osteoarthritis, was made. The patient was treated with ivermectin (12 mg daily) for 14 days. Repeat stool cultures were negative for parasites. Symptoms, including nausea, vomiting, and abdominal pain, improved significantly. Anemia and anasarca resolved within 6 weeks of follow-up. The patient received terbinafine for onychomycosis and was referred for orthopedic management of knee osteoarthritis.

DISCUSSION Strongyloidiasis, a common but often overlooked parasitic infection, endemic to the Asian-African subcontinent, affects 100 million people globally. Diagnosis is frequently delayed due to low clinical suspicion or symptoms mimicking other gastrointestinal disorders. It can escalate to hyperinfection or disseminated disease with high mortality when the patient's immune status changes, typically due to steroid use (regardless of dose or duration).

CONCLUSIONS Despite advances in polymerase chain reaction testing, repeated stool examinations remain the definitive, cost-efficient, and gold standard method of diagnosing parasitic infections, requiring minimum 3 tests due to intermittent larval secretion. Misuse of corticosteroids significantly escalates the risk of hyperinfection syndromes alongside HIV, diabetes, and malignancies. Intriguingly, eosinophilia, commonly linked with parasitic infections, may be absent due to corticosteroid effects, underscoring the need for vigilant suspicion and prompt, precise diagnostics and treatment in immunosuppressed patients.

Key words

immunosuppressed condition, malabsorption syndrome, over the counter medication, stool examination, *Strongyloides* hyperinfection

POSTER PRESENTATIONS

KONRAD STĘPIEŃ

RECOGNIZED FOR THE BEST POSTER

Syndrome of a truly broken heart: left ventricular rupture in a patient with recurrent takotsubo syndrome

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INTRODUCTION Takotsubo syndrome (TTS) is an acute, potentially reversible clinical syndrome characterized by transient left ventricular (LV) dysfunction. It has been considered to be relatively benign and associated with favorable prognosis. However, recent studies have shown that the mortality rate in TTS patients is comparable to that observed in individuals with coronary syndrome. Moreover, it has been recognized that complications occur in half of the TTS patients.
CASE DESCRIPTION An 83-year-old woman was admitted to a hospital with ST-segment elevation myocardial infarction (STEMI). Coronary angiography was immediately performed and showed no significant narrowing in the coronary arteries. However, a slow-flow phenomenon was observed, mainly in the left coronary artery. Simultaneously, ventriculography presented a pattern of segmental contractility abnormalities typical of apical TTS. Echocardiography showed akinetic apex and periapical segments of the LV with hyperkinesis of the basal segments and LV ejection fraction (LVEF) of 35%. The patient denied the occurrence of emotional or physical triggers. The InterTAK Diagnostic Score was 37, with a 2.3% probability of TTS. The course of the hospitalization was uncomplicated. Follow-up echocardiography showed a gradual improvement in LV contractility with LVEF of 45%. The patient was discharged after a few days.

Four years later, she was readmitted due to similar stenocardial acute chest pain with STEMI. As before, coronary angiography showed no significant narrowing in the coronary arteries, with a clearly marked slow-flow phenomenon. Transthoracic echocardiography (TTE) demonstrated akinetic and ballooning apex and periapical segments of the LV with LVEF of 35%. No emotional or physical triggers were identified again during in-depth history-taking, and the same result was obtained in the InterTAK Diagnostic Score. TTS recurrence was diagnosed and conservative treatment was applied.

At night-time, the patient's condition deteriorated, as she developed hypotension and sudden cardiac arrest in pulseless electrical activity. Point-of-care TTE showed cardiac tamponade with features of LV free-wall rupture. Cardiopulmonary resuscitation was immediately initiated. Pericardiocentesis involving drainage of 500 ml of blood was performed by a cardiac surgeon but it was not completely effective due to active bleeding. Despite intensive treatment, the patient died due to a lack of hemodynamic response.
DISCUSSION Patients with TTS are at a risk of recurrence. As shown in the GEIST registry, the recurrence rate is 4%, and the individuals who suffer from recurrence more frequently develop pulmonary edema (13.3% vs 4.9%; $P = 0.04$). Moreover, most recurrences occur in the first 5 years after the index TTS episode. LV rupture is a rare and most serious complication of TTS, associated with an extremely high risk of death. In a recent systematic review, 35 such cases have been reported. To our best knowledge, this is the first description of LV rupture in a patient with recurrent TTS.
CONCLUSIONS Patients with recurrent TTS are at a risk of LV rupture, which is a rare complication with an extremely high risk of death.

Key words
cardiac tamponade, coronary slow-flow, myocardial rupture, takotsubo syndrome

LUCIA AVELLANEDA MOLINA

RECOGNIZED FOR THE BEST POSTER

Budd–Chiari syndrome secondary to antiphospholipid syndrome in a young patient

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INTRODUCTION Budd–Chiari syndrome (BCS) is a liver dysfunction caused by obstruction of hepatic venous outflow. The pathophysiology involves thrombosis of the hepatic veins owing to a state of hypercoagulability induced by certain oncological diseases, infections, autoimmune diseases, and some drugs. The association with antiphospholipid syndrome has been described before but requires high clinical suspicion, especially in the absence of previous thrombotic events and due to a wide spectrum of possible presentations, which may range from nearly asymptomatic to fulminant hepatic failure.
CASE DESCRIPTION A previously healthy 18-year-old man presented with a 1.5-year history of progressive ascites and collateral periumbilical venous circulation associated with the appearance of nonpainful erythematous papules on the fingers of both hands, palmoplantar erythema, and stasis dermatitis with symmetrical distribution on the lower extremities. He was admitted due to severe ascites, increased collateral venous circulation extending to the chest, imaging findings compatible with cirrhosis (without clinical signs of liver failure), and thrombocytopenia caused by hypersplenism secondary to portal hypertension. Storage diseases and various immune-mediated diseases were ruled out, which led to a diagnosis of primary antiphospholipid syndrome with triple-positive antiphospholipid antibodies (anticardiolipin immunoglobulin G, lupus inhibitor, and β2-antiglycoproteins). Systemic lupus erythematosus was excluded based on a negative result of antinuclear factor. Anticoagulant treatment with low-molecular-weight heparin was started. Splenoportal Doppler ultrasonography, computed tomography angiography, and magnetic resonance imaging of the abdomen were performed, which showed alteration of hepatic perfusion without portal thrombosis, and a hepatopetal flow. Transjugular hepatic venous pressure measurement confirmed an increased portosystemic gradient (10 mm Hg), and the finding of severe stenosis of the suprahepatic veins compatible with BCS. Video-assisted endoscopy of the upper digestive tract did not show esophageal varices, and transesophageal Doppler echocardiography revealed moderate mitral regurgitation linked to the antiphospholipid syndrome. Finally, the patient was selected as a candidate for liver transplant and referred for intrahepatic portosystemic shunt for management of portal hypertension due to ascites refractory to medical treatment, with periodic evacuative paracentesis.
CONCLUSIONS The value of this case focuses on the atypical presentation of primary antiphospholipid syndrome in a young patient with no history of thrombosis, who presented with slow-onset portal hypertension secondary to BCS, which ultimately led to liver cirrhosis due to a late diagnosis. BCS is a rare disorder that is difficult to diagnose, especially in a chronic course. Identifying the cause is always challenging, and timely diagnosis is crucial to prevent severe comorbidities.

Key words
antiphospholipid syndrome, Budd–Chiari syndrome, inferior vena cava syndrome, noncirrhotic portal hypertension

ZSUZSÁNNA FARKAS

RECOGNIZED FOR THE BEST POSTER

Dirofilariasis in Crohn's disease

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INTRODUCTION Erythema nodosum is considered to be a frequent extraintestinal manifestation of Crohn's disease, mainly presenting on the lower legs, thighs, and upper extremities. Recurrent and chronic forms are rare. Similar subcutaneous nodules can be seen in human *Dirofilaria repens* cases. Human-related infections are inflicted mainly by 2 species: *D. repens* causes subcutaneous manifestations, while *D. immitis* causes visceral manifestations.

CASE DESCRIPTION In our case, a 41-year-old woman was diagnosed with Crohn's disease and treated accordingly. After therapy was discontinued, a subcutaneous nodule appeared in a very atypical localization, in the right subclavicular region. Over just a month's period of time, the skin lesion migrated to the left side. Panniculitis or nodular skin manifestations are caused by a variety of diseases. In our case it was not typical for Crohn's disease, therefore further tests were performed. During ultrasound imaging of the subclavicular region, thread-like echogenicity was detected, which raised a suspicion of filariasis. After the skin lesion was surgically excised, an adult specimen of *D. repens* was identified. The patient had not traveled to the Mediterranean region in the previous years and had no history of contact with pets, but she lived near the Danube river and tended a garden.

DISCUSSION Humans are not suitable hosts for *D. repens* species and only become infected by these vector-borne nematodes by accident. The natural hosts are cats and dogs. The transmission of the disease occurs via mosquito bites (*Aedes* and *Anopheles* species). In human *D. repens* cases (except some rare reported incidents), only 1 nematode gets transmitted, which then settles in the subcutaneous adipose tissue (creating a subcutaneous nodule). There are no microfilaria in the peripheral blood, which makes the diagnosis relatively difficult.

CONCLUSIONS Previously, *D. repens* has been endemic only to the Mediterranean countries, but over these previous years multiple cases have been identified even as far as Hungary. The cause of this shift can be seen in human activity—the mosquito colonization, development, and activity are supported by the climate change, and through pet tourism infected hosts can easily spread the infection. Therefore, we can expect even more dirofilariasis cases, not only in Hungary, but also in the northern parts of Europe.

Key words

climate change, Crohn's disease, dirofilariasis, erythema nodosum

AHMET ALPARSLAN CERAN

The case diagnosed with newly defined detrimental disease: VEXAS syndrome

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CASE DESCRIPTION VEXAS (vacuoles, E1 enzyme, X-linked, autoinflammatory, somatic) syndrome, an emerging and uncommon autoinflammatory disorder, is distinguished by its distinctive clinical features. It presents with recurrent fevers, vacuole formation in myeloid cells, and somatic mutations in the *UBA1* gene, affecting primarily men. This X-linked condition manifests in various clinical aspects, including hematological abnormalities, pulmonary and cutaneous involvement, and musculoskeletal symptoms.

Genetic mutations in VEXAS disrupt ubiquitin homeostasis, leading to dysregulated inflammatory responses and contributing to the intricate pathophysiology of the disorder. Despite its recent recognition, VEXAS poses diagnostic challenges due to its diverse clinical presentation and the absence of specific biomarkers. Timely detection and accurate diagnosis are pivotal, given the elusive nature of the syndrome.

Treatment options for VEXAS are currently limited, emphasizing the need for an enhanced understanding of its pathophysiology in order to develop targeted therapeutic interventions. This necessitates ongoing research efforts and collaborative initiatives within the medical community, aiming to explore innovative treatment modalities and enhance patient outcomes.

Heightened awareness among healthcare professionals is crucial for effective management, emphasizing the importance of recognizing clinical patterns, understanding genetic markers, and staying informed about emerging research findings for timely intervention and personalized treatment strategies.

Key words

autoinflammation, *UBA1*, ubiquitination, VEXAS

ADELA SKOUMALOVA

Emberger syndrome: isolated positivity of rheumatoid factor and anticitrullinated protein antibodies in the IgA class as a sign leading to the diagnosis of a rare genetic disorder

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INTRODUCTION Emberger syndrome (primary lymphedema with myelodysplasia) is a rare autosomal dominantly inherited disease. It is caused by a mutation in the gene for the transcription factor GATA-binding protein 2 (*GATA2*). Deficiency of GATA2 leads to impaired genesis and function of hematopoietic progenitor cells. Its clinical manifestations are heterogeneous, mostly associated with primary lymphoedema, sensorineural deafness, hematological manifestations, immunodeficiency, and autoimmune manifestations.

CASE DESCRIPTION A 25-year-old woman of short stature has been followed for sensorineural deafness since the age of 4, as well as for moderate persistent bronchial asthma and recurrent infections (particularly extensive palmar and plantar warts, herpes labialis and nasalis, keratitis, scleritis, and upper respiratory tract infections). In childhood, immunological examination showed hyperimmunoglobulinemia A. This time, the patient was referred to our clinic by a rheumatologist for arthralgia and positivity of antinuclear antibody, rheumatoid factor, and anticitrullinated protein antibodies (the last 2 only in the IgA class). On initial examination, whole-body exanthema and extensive warts on the palms and soles, with no evidence of synovitis, were clinically dominant. Significant elevation of serum total protein was found and serum electrophoresis showed polyclonal hypergammaglobulinemia. The examination of immunoglobulin levels showed significantly elevated levels of immunoglobulin A. Other immunoglobulins levels were within normal range. The family history was remarkable for the presence of hearing loss in the patient's father and sister. The father also showed symptoms of warts. Based on clinical findings and results, the possibility of hereditary disease was considered. Suspicion was raised

for a primary immunodeficiency – a deficiency of the transcription factor GATA2. Genetic analysis confirmed mutation in the *GATA2* gene and Emberger syndrome was diagnosed.

DISCUSSION Emberger syndrome is a rare autosomal dominantly inherited disorder with a prevalence of less than 1 in 1 000000. It was first described in 1979, but its molecular basis was not discovered until 2011. Its clinical manifestations are heterogeneous, mostly associated with primary lymphoedema, sensorineural deafness, hematological manifestations, immunodeficiency, and autoimmune manifestations. The positivity of diagnostic antibodies for rheumatoid arthritis seems to be caused by overproduction of IgA. This condition is not a common finding in Emberger syndrome. We have found a case of hyperimmunoglobulinemia E in the literature but we have not found another described case of hyperimmunoglobulinemia A.

CONCLUSIONS This case report highlights the clinical presentation, diagnostic journey, and genetic confirmation of GATA2 deficiency syndrome in a 25-year-old woman. Consistent collection of anamnestic data and inclusion of rare causes to the differential diagnosis of complicated cases is essential to establish the correct diagnosis.

Key words
Emberger syndrome, *GATA2*, hypergammaglobulinemia, hyperimmunoglobulinemia A

AGNIESZKA JAROSIŃSKA

An unexpected diagnosis of suspected pancreatic cancer

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CASE DESCRIPTION A 57-year-old woman was admitted to the department of internal medicine for investigation of a hyperechogenic, irregular lesion, which measured 38 mm in diameter and was located in the head of the pancreas. The lesion was found during an outpatient abdominal ultrasound scan. The patient had a history of pain in the umbilical region and upper abdomen, unintentional weight loss (4 kg over 6 months), and skin changes, such as urticaria. Additionally, the patient was previously diagnosed with orbitopathy, asthma, and chronic sinusitis. On admission, physical examination showed pale, dry skin, numerous seborrheic keratoses, and dermographism. Additionally, bilateral proptosis (more prominent on the right side), eyelid edema (more prominent on the right side), and geographic tongue were observed. Laboratory tests showed increased activity of liver enzymes (alkaline phosphatase, 161 U/l; γ-glutamyl transferase, 142 U/l) and pancreatic enzymes (amylase, 115 U/l; lipase, 179 U/l). Antithyroid peroxidase antibodies (anti-TPO), antithyroglobulin antibodies (anti-Tg), and antithyrotropin receptor antibody (TRAb) tests were performed due to suspected thyroid orbitopathy; the results were negative. Determining the immunoglobulin concentration showed elevated IgE (1120 IU/ml) and IgG4 (11 000 g/l) titers. Medical imaging was extended to computed tomography scans of the chest, abdominal cavity, and pelvis. The results showed a discreetly hypodense area in the head of the pancreas. Additionally, the intrahepatic and extrahepatic tracts were widened, and the duct of Wirsung was severed at the location of the mentioned lesion. These results raised a suspicion of a cancerous or inflammatory lesion in the head of the pancreas. Endoscopic ultrasound (EUS) guided fine needle biopsy of the pancreas was carried out. Initial diagnosis of IgG4-related disease had been made. The histopathological result from the EUS-guided fine needle biopsy of the lesion in the head of the pancreas confirmed the diagnosis of IgG4-related disease. The patient had fulfilled the clinical and radiological criteria of typical organ involvement (changes in the pancreas, salivary glands, lacrimal glands, bile ducts, orbit, and lungs), the serological criteria (IgG4 titer), and the histopathological criteria. With the diagnosis of IgG4-related disease, the patient's immunosuppressive therapy was modified. Methylprednisolone boluses were administered during hospitalization (250 mg/day for 3 days), followed by prednisone with a reduced dose of 10 mg/day. After 2 weeks of steroid therapy, azathioprine was introduced. Clinical improvement was observed. In follow-up, the patient remains under care of outpatient rheumatology clinic, receiving 100 mg/day of azathioprine. The dose of prednisone has been reduced to 5 mg/day. This case shows that IgG4-related systemic disease is an insidious disease with a chronic course. In the light of current reports, the spectrum of symptoms presented by the patient over several years strongly suggests this disease entity. It is therefore important not to underestimate similar symptoms, especially if they occur at the same time, since the treatment is widely available, cheap, and effective. The presented case highlights the significance of a thorough medical interview and the importance of the diagnostic process that is based on the whole clinical picture, and not singular symptoms.

Key words
endoscopic ultrasound, IgG4-related disease, IgG4-syndrome, immunosuppressive therapy, suspicious lesion

ALEKSANDRA KUCHARSKA-LUSINA

Can an exacerbation of a rare disease be a symptom of another ultrarare disease? Challenges of iMCD-TAFRO syndrome diagnosis

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INTRODUCTION Castleman disease, described by Castleman in 1956, encompasses localized unicentric Castleman disease (UCD) and systemic multicentric Castleman disease (MCD). The latter includes various clinical subgroups, such as POEMS-associated iMCD and iMCD-TAFRO. The pathophysiology of iMCD involves hypercytokinemia, notably due to interleukin 6 (IL-6) levels. Diagnosis is challenging due to heterogeneous symptoms, but consensus criteria aid in the identification of the disease. Treatment typically involves anti-IL-6 therapies, siltuximab being the primary option.

CASE DESCRIPTION A 26-year-old woman with a history of dermatomyositis presented with severe upper abdominal pain, weakness, dry cough, and elevated inflammatory markers. She also experienced heart palpitations, intermittent fever, night sweats, unintentional weight loss, and a recent dermatomyositis relapse (despite treatment adjustments). Imaging showed pleural effusion, pericardial fluid, and perisplenic fluid. The patient developed hypoalbuminemia, acute kidney injury, and fluid-resistant hypotension, necessitating hydrocortisone pulses. Her condition stabilized and she was transferred to the rheumatology and immunology department. Laboratory findings included neutrophilic leukocytosis, anemia, thrombocytosis, elevated inflammatory markers, and mild thrombocytopenia (platelet count, 129 $\times$ 10³/μl). Superficial lymph nodes ultrasound showed numerous hypoechoic lymph nodes in the neck, without signs of pathological vascularization, and enlarged left axillary lymph node (11 mm $\times$ 16 mm) with a thickened cortical layer (up to 8 mm) to be verified by biopsy. A bone marrow biopsy showed hyperemic stroma with slight reticulin fibrosis. CD61+ megakaryocytes were moderately more numerous in relation to marrow cellularity, with slight dysmorphia, which meets the minor criteria of Castleman disease, but could be nonspecific. The histopathological findings suggested the multifocal Castleman disease, specifically the idio-

pathic (HHV8-negative) mixed type (iMCD). In correlation with clinical manifestation, we diagnosed multicentric Castleman disease (MCD) with a mixed histological type (iMCD-TAFRO; thrombocytopenia, anasarca, fever, renal dysfunction, and organomegaly). Clinical resolution: In the hematology department, the patient received 4 doses of siltuximab (11 mg/kg body weight). It was decided the first 4 doses would be administered at weekly intervals, and then every 21 days. The patient showed clinical improvement and was discharged with prescription for prednisone, spironolactone, allopurinol, proton pomp inhibitors, and antibiotic prophylaxis. However, 2 weeks later the patient was readmitted to hematology department due to generalized skin rash with itching and facial swelling (present despite the steroid therapy). The diagnosis of drug reaction with eosinophilia and systemic symptoms (DRESS) was made. The second line treatment for DRESS with oral cyclosporin was initiated, with clinical improvement. It was decided to switch the siltuximab therapy to tocilizumab. The patient received the first dose of IV tocilizumab (8 mg/kg body weight) and subsequent tocilizumab administrations had led to sustained improvement.

DISCUSSION Castleman disease, a rare lymphoproliferative disorder, encompasses unicentric or multicentric manifestations. This case focuses on the challenging multicentric iMCD-TAFRO subtype. The complexities of the diagnosis arise from the disease's rarity, heterogeneity, and nonspecific symptoms, compounded by the patient's history of dermatomyositis and steroid therapy. Treatment involved a multifaceted approach including siltuximab therapy, which was switched to tocilizumab due to DRESS syndrome. The dynamic nature of iMCD necessitates close monitoring and multidisciplinary care for successful outcomes.

CONCLUSIONS Vigilance is essential in patients with an exacerbation of autoimmune diseases and systemic symptoms such as fever, cough, weakness, lymphadenopathy, and weight loss. Continued diagnostic investigation should also be pursued for other rare diseases that may coexist with the primary diagnosis.

Key words
dermatomyositis, DRESS syndrome, idiopathic multicentric Castleman disease, TAFRO syndrome, siltuximab

ALICA MELLOVA

Staphylococcal endocarditis of the mitral valve complicated by MODS and septic embolisms in a young man with Marfanoid habitus: a case report

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INTRODUCTION Marfan syndrome is a genetic autosomal dominant connective tissue disorder caused by a mutation in the *FBN1* gene. Clinical symptoms affect multiple organ systems, including the skeleton, lungs, spine, and eyes. The most serious consequences arise from the involvement of the cardiovascular system, leading to aortic dilation, possible aortic dissection, and valve prolapse. Prolapse of the heart valve is a potential risk factor for the development of infective endocarditis. In this case, a young patient with marfanoid habitus and prolapse of the mitral valve was experiencing a severe course of staphylococcal infective endocarditis, complicated by multiorgan failure and development of septic pseudoaneurysm of the superior mesenteric artery.

CASE DESCRIPTION We describe a case of a 30-year-old patient with marfanoid habitus and a history of JJ stent implantation for nephrolithiasis, drainage of spontaneous pneumothorax, corrective chest surgery for pectus excavatum, myxomatous changes, and prolapse of the mitral valve. The patient was admitted to the hospital due to fever and a combined-type dyspeptic syndrome that had been present for 4 days. The patient was hypotensive. Electrocardiogram showed sinus tachycardia and an incomplete right bundle branch block pattern. The prominent finding on examination was a systolic murmur over the auscultation area of the mitral valve and subconjunctival bleeding on the right side. Transthoracic echocardiography showed mobile structures resembling vegetations on the mitral valve and moderate mitral regurgitation. Microbiological screening showed positive blood culture samples for *Staphylococcus aureus*. The patient was diagnosed with staphylococcal infective endocarditis. Despite targeted antibiotic therapy, the patient was experiencing numerous septic embolisms, not only in the peripheral but also in the central nervous system, with severe intracranial hemorrhages and subarachnoid bleeding. The course of the endocarditis was complicated by septic shock and multiorgan failure. Due to many serious complications, the timing of the cardiac surgery was difficult. The patient underwent mitral valve replacement with a mechanical prosthesis and tricuspid valve repair with an annuloplasty ring. Computed tomography scan and ultrasound performed after the cardiothoracic intervention showed rare septic pseudoaneurysm of the superior mesenteric artery, requiring surgical resection. Based on Ghent criteria, we assumed hereditary connective tissue disease and subsequent genetics tests confirmed Marfan syndrome.

CONCLUSIONS Approximately 57% of patients with Marfan syndrome present with mitral valve prolapse, and are therefore at a higher risk for the development of infective endocarditis. Infective endocarditis can be a life-threatening condition, often accompanied by multiorgan dysfunction and septic embolisms. It requires comprehensive intensive care treatment, targeted combined antibiotic therapy, and interdisciplinary collaboration. Investigating potential septic embolic complications is essential. Proper indication and timing, not only for cardiothoracic surgery but also for interventional management of septic complications, are crucial for the patient. It is important to substantiate clinical suspicions of Marfan syndrome through genetic testing. This enables patient monitoring and early management of complications, potentially documenting Marfan syndrome in close relatives. The time from the first manifestation of Marfan syndrome symptoms to its definitive genetic determination in Europe is 2 to 4.5 years. To shorten this interval, Ghent criteria can be used, allowing for rapid differentiation of patients indicated for genetic testing.

Key words
Ghent criteria, infective endocarditis, Marfan syndrome, mitral valve, septic pseudoaneurysm

ALYNA HAFEEZ

Hypercalcemia-induced respiratory failure as the primary presentation in chronic myelomonocytic leukemia

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INTRODUCTION Chronic myelomonocytic leukemia (CMML) is a rare clonal hematopoietic stem cell disorder, characterized by the presence of sustained peripheral blood monocytosis along with dysplastic features in the bone marrow. The disease shares overlapping features with both myelodysplastic syndromes and myeloproliferative neoplasms. Diagnosing hypercalcemia as the cause of acute respiratory failure in elderly population with no other comorbidities can be really challenging. This case report highlights a unique and clinically significant manifestation of CMML, emphasizing how important it is for a physician to consider hematological malignancy-induced hypercalcemia in cases of respiratory failure and drowsiness.

CASE DESCRIPTION A 69-year-old woman with no known comorbidities presented with complaints of an on-and-off fever that had begun 12 days prior, accompanied by a dry cough and worsening general weakness. Complete blood count showed leukocytosis along

with raised monocyte count. General physical examination was unremarkable. The patient was being managed along the lines of a lower respiratory tract infection with broad spectrum antibiotics. She gradually became afebrile, but her drowsiness increased. Arterial blood gas (ABG) analysis showed normal pH with raised pCO_2 levels. Noninvasive ventilatory (NIV) support was applied. Brain magnetic resonance imaging showed focal enhancement in the right temporal lobe. In order to rule out causes of drowsiness and neuromuscular weakness, neurological workup was performed, including cerebrospinal fluid testing, and electromyography and nerve conduction studies, the results of which were unremarkable. However, the patient's calcium levels were significantly elevated (15.2 mg/dl). Thus, she was started on IV fluids and given 1 dose of zoledronic acid. Gradually, as the underlying metabolic abnormalities were corrected, her consciousness improved and NIV support was spaced out with improvement in ABGs. Due to persistently raised monocyte count, along with hypercalcemia, hematological malignancy was suspected. Bone marrow aspirate and trephine were suggestive of CMML. The patient was then discharged, at her request, with advice to get further workup and management of CMML in the outpatient environment. After 4 weeks, the patient was readmitted with similar complaints of drowsiness and fever, along with hypercalcemia. Considering her clinical status, she was deemed unsuitable for administration of chemotherapeutic medications, such as hypomethylating agents. Thus, similar conservative management was undertaken. However, the patient's condition worsened and she died.

DISCUSSION Hypercalcemia of malignancy is the most common metabolic disorder in patients with advanced cancers. Moderate to severe acute hypercalcemia can present with hyporeflexia, lethargy, altered mental status, and musculoskeletal symptoms, such as muscle weakness. While hypercalcemia is a recognized complication in various malignancies, its association with respiratory failure, particularly in the context of CMML, remains rare and poorly understood. There are few case reports that describe the relationship between CMML and hypercalcemia. In our case, the patient presented with gradually increasing confusion, drowsiness, and generalized muscular weakness caused by hypercalcemia, which ultimately led to respiratory failure. Moreover, persistently elevated monocyte count should raise suspicion of a potential underlying neoplastic process.

CONCLUSIONS It is important to consider hematological malignancies and disorders, such as CMML, when met with an elderly patient with unexplained hypercalcemia and drowsiness accompanying a respiratory failure. Early identification and prompt intervention can improve the quality of life of these patients and allow for an early initiation of the treatment for their underlying malignancy.

Key words
chronic myelomonocytic leukemia, hypercalcemia, malignancy, monocytosis, respiratory failure

ANITA WACH

From systemic lupus erythematosus "rain" to a tsunami: how to stop it?

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INTRODUCTION Systemic lupus erythematosus (SLE) is an autoimmune disease affecting multiple organs. When SLE coexists with antineutrophil cytoplasmic antibody (ANCA)-associated vasculitis (AAV), it manifests with a severe disease course and heightened mortality. In cases of thromboembolic events, it is crucial to not only diagnose antiphospholipid syndrome (APS), but to also exclude inherited thrombophilia. Mortality in SLE stems primarily from high disease activity, infections, and cardiovascular complications. Our case exemplifies all the factors indicative of a severe SLE course and unfavorable prognosis.

CASE DESCRIPTION A 31-year-old man, diagnosed with SLE and APS, presented with a medical history of fever, malar rash, arthralgia, arthritis, pulmonary embolism, deep vein thrombosis, elevated antinuclear antibodies (ANA) titer, and positive anticardiolipin antibodies in IgG and IgM classes, along with anti-β2-glucoprotein I antibodies in IgM class antibodies. Treatment with systemic glucocorticoids (SGCs) resulted in symptom improvement; however, subsequent SLE exacerbations occurred with concomitant AAV. Laboratory investigations showed a high ANA titer (1:20 480; reference range, 1:160), positive anti-double stranded DNA (anti-dsDNA) antibodies (titer, 1:5120; reference range, 1:10), leucopenia, decreased C3 and C4 complement levels, proteinuria, and granular casts in the urine sediment. Furthermore, a high titer of ANCA directed against proteinase-3 (PR3 >200.0 RU/ml; reference range >20 RU/ml) was detected. Kidney biopsy showed active diffuse global proliferative lupus nephritis. Additional tests showed reduced activity of total protein S and heterozygous factor V Leiden gene mutation. The patient underwent treatment with SGCs, cyclophosphamide, and rituximab. Subsequently, the patient developed infected ulcers with necrosis on the right lower limb, necessitating limb amputation. Additionally, he experienced a sacral pressure ulcer, septic shocks, acute kidney failure, multiple cardiac arrests, and infective endocarditis. During a 9-year follow-up, the patient experienced 2 more SLE exacerbations. Currently, the patient is in a stable condition, being treated with SGCs and methotrexate.

DISCUSSION SLE and AAV are severe autoimmune diseases that share some clinical features. Using enzyme-linked immunosorbent assay, 9.3% of SLE cases were found to have antimyeloperoxidase (anti-MPO) positive AAV, whereas only up to 1.7% had anti-PR3 positive AAV. SLE–AAV overlap syndrome patients are predominantly women, suffering from hematological abnormalities, arthritis, and renal involvement, who have positive ANA and anti-MPO antibodies. The occurrence of thromboembolic events in our patient underscores the importance of diagnosing APS and the need to include testing for antithrombin III deficiency, protein C and S deficiency, factor V Leiden, and prothrombin G20210A gene mutations. Mortality in SLE patients is higher compared to healthy controls and is primarily a consequence of high SLE activity, infections, and cardiovascular complications, all of which were present in our patient. Additionally, an intrahospital cardiac arrest with concomitant sepsis is associated with an approximately 85% mortality rate.

CONCLUSIONS It is imperative to broaden the diagnostic assessment in SLE patients who also display symptoms indicative of AAV, by conducting tests for anti-MPO and anti-PR3 antibodies. Additionally, for thrombotic events, aside from APS, inherited thrombophilia should always be investigated. Recognizing the primary causes of mortality in SLE—namely, high disease activity, infections, and cardiovascular complications—is crucial.

Key words
endocarditis, sepsis, systemic lupus erythematosus, thrombophilia, vasculitis

ANNA NAGÓRSKA

Pseudo-Cushing syndrome associated with pheochromocytoma

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INTRODUCTION The biochemical picture of pseudo-Cushing syndrome (PCS) may present as adrenocorticotropic hormone (ACTH)--dependent hypercortisolemia. The differentiation between these diagnoses may be a diagnostic challenge.

CASE DESCRIPTION A 66-year-old woman in a severe general condition was placed under observation due to weakness, loss of appetite, apathy, weight loss (5 kg within a month), and newly diagnosed diabetes mellitus and hypertension. In addition, the patient had a history of fainting spells. Physical findings showed tachycardia and plethora. Laboratory evaluation showed ACTH--dependent hypercortisolemia, severe hypokalemia, metabolic alkalosis, hyperglycemia, and markedly elevated metanephrines. Abdominal computed tomography with inhomogeneous contrast enhancement showed a heterogeneous, well-demarcated lithocystic lesion (with dimensions 32 mm × 25 mm) in the right adrenal gland. Gallium-68 DOTA-0-Tyr3-Octreotate positron emission tomography showed heterogeneous, but high, somatostatin receptor expression within the lesion. Hypercortisolemia caused by ectopic ACTH-producing pheochromocytoma was suspected. The patient was qualified for right adrenalectomy and prepared with increasing doses of doxazosin and a low-dose of bisoprolol. Due to significant hypercortisolemia, metyrapone was applied. A rapid decrease in plasma cortisol levels was observed, associated with hypotension and weakness. The patient was given hydrocortisone substitution because of a suspicion of developing adrenal insufficiency. The patient required constant potassium supplementation. The hydration, salting of meals, enteral feeding by nasogastric tube, protein supplementation, insulin therapy, and thromboprophylaxis were implemented. A gradual, spontaneous decrease in ACTH levels was observed although no "anti-ACTH" treatment was introduced. After surgery, no features of adrenal insufficiency were observed. The histopathological examination showed pheochromocytoma. The tumor staining for ACTH was negative. Two months after surgery, laboratory tests showed normalization of cortisol and metanephrines levels. Very high plasma ACTH and cortisol levels, and a loss of diurnal rhythm were observed in the patient, so ectopic ACTH secretion (EAS) was suspected. High levels of metanephrines and right adrenal non-adenoma tumor with high somatostatin receptors expression suggested pheochromocytoma as a source of EAS. However, considering the immediate suppression of plasma cortisol levels after metyrapone administration, spontaneous decrease in ACTH levels before surgery, no secondary adrenal insufficiency after surgery, and negative ACTH-staining in histopathological examination, pseudo-Cushing syndrome was diagnosed.

DISCUSSION ACTH-dependent hypercortisolemia can be caused by an ACTH-secreting pituitary adenoma or ectopic ACTH secretion, and it may present in form of PCS. EAS is a paraneoplastic syndrome common in patients with neuroendocrine tumors (some cases of EAS caused by pheochromocytoma were described), while PCS is associated with physiological overactivation of the hypothalamic-pituitary-adrenal axis in various conditions. One of them is pheochromocytoma, in which the hypermetabolic state caused chronic overproduction of catecholamines can be observed. Catecholamines exert their metabolic effects directly (by adrenoceptors in metabolically active organs and tissues) and indirectly (by the overproduction of proinflammatory cytokines, which activates hypercatabolism). Moreover, they induce a glucocorticoid-dependent positive-feedback loop through glucocorticoid receptor expression, which accelerates both ACTH and catecholamine secretion. That was the probable mechanism of developing PCS in our patient.

CONCLUSIONS In the differentiation of hypercortisolemia associated with pheochromocytoma, ectopic production of ACTH and PCS induced by hypermetabolic state should be considered.

Key words
ectopic adrenocorticotropic hormone secretion, hypercortisolemia, pheochromocytoma, pseudo-Cushing syndrome

BALÁZS LEVENTE LENGYEL

An unusual pituitary-hypothalamic mass lesion

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INTRODUCTION Sarcoidosis is a disease of unknown etiology, characterized by noncaseating granulomas that can affect any organ. The most common manifestations occur in the lymph nodes and lungs. In approximately 14%–50% of cases neurosarcoidosis is confirmed through postmortem examination, but clinical symptoms manifest in only 5% to 13% of patients. Lesions involving the pituitary and hypothalamus are rare, and as of the time this case report was written, no standardized recommendations have been established for their management. These lesions can lead to hypopituitarism, diabetes insipidus, and optic neuropathy. In cases involving the pituitary and hypothalamus, the primary therapeutic goals include hormone replacement and reduction of visual field deficits.

CASE DESCRIPTION A 41-year-old woman with a history of sarcoidosis and secondary pulmonary hypertension caused by a pulmonary embolism presented with headaches and menstrual disturbances. Initial workup showed elevated prolactin levels and hypophyseal macroadenoma. Thus, dopamine agonist therapy was initiated, resulting in the normalization of prolactin levels. In April, the patient was admitted to the Korányi National Pulmonology Institute's intensive care unit due to hypotension, severe weakness, polyuria, and polydipsia, prompting endocrine consultation. During our consultation, we diagnosed central hypoadrenalism and central diabetes insipidus, prompting glucocorticoid supplementation with hydrocortisone and nasal desmopressin therapy. Central hypothyroidism was ruled out. After initiating therapy, the symptoms resolved rapidly. However, during follow-up, the patient complained of headaches and recurrent fever. Bronchoscopy was performed, confirming the recurrence of pulmonary sarcoidosis, and reintroduction of high-dose methylprednisolone therapy was warranted.

Even though the pituitary lesion decreased in size by September, on follow-up magnetic resonance imaging (MRI) it no longer resembled a macroadenoma, but rather a granulomatous lesion. Considering the patient's symptoms, examinations, laboratory results, and MRI findings, we established a diagnosis of neurosarcoidosis localized in the pituitary and hypothalamic region. Treatment with methylprednisolone was continued in collaboration with a pulmonologist.

CONCLUSIONS If dysfunction of the hypothalamic-pituitary axis is suspected in patients undergoing treatment for sarcoidosis, the possibility of neurosarcoidosis should be considered, prompting urgent investigation and treatment.

Key words
endocrinology, hypopituitarism, pituitary lesions, pulmonology, sacroidosis

BARBORA POSPISILOVA

Adverse effects of medications or systemic disease? Systemic scleroderma with gastrointestinal involvement

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INTRODUCTION We report a case of a woman who was treated for pulmonary hypertension in a specialized center since 2017. She

suffered from nausea and diarrhea, and the symptoms were believed to be adverse effects of her ongoing medication. However, after 5 years she was referred to a gastroenterological center and diagnostic studies showed that systemic scleroderma was the cause.

CASE DESCRIPTION A 76-year-old woman had been treated for pulmonary hypertension in a specialized cardiological center for 5 years. During the treatment, she experienced nausea, vomiting, and diarrhea, which were considered to be adverse effects of her ongoing medication—treprostinil and riociguat. The patient was referred to a gastroenterologist, as she started to lose weight more progressively, and vomiting and diarrhea limited the quality of her daily life. Stool culture was negative, but she had medical history of diverticulosis of the colon, and so she was prescribed rifaximin. The clinical effect was only mildly positive. Gastroscopy showed severe esophageal candidiasis and grade B erosive esophagitis (Los Angeles classification). Control gastroscopy performed after treatment with antimycotics and proton pump inhibitors showed dilatation of the esophagus and food leftovers in the stomach. Since several submucosal prominences were also found in the dilatated esophagus, computed tomography (CT) scan of the chest and abdomen was performed, ruling out malignancy as a cause. Esophageal manometry showed type 1 achalasia, so the patient underwent endoscopic dilatation with a clinical effect. Total colonoscopy was not feasible due to diverticulosis. However, sigmoidoscopy with biopsies excluded other mucosal pathologies, including microscopic colitis. CT colonography showed not only severe colonic diverticulosis, but also duodenum and small bowel dilatation. Suspicion of systemic disease led to laboratory screening, which showed positive antinuclear and anticentromere antibodies, and normal κ/λ free light chain ratio. The patient admitted she had been experiencing Raynaud phenomenon for years and rheumatological examination confirmed a limited systemic sclerosis with the finding of cutaneous lesions. Considering the patient's preferences and multimorbidity, she was not given any immunosuppressants. Although diarrhea persists and weight loss is mildly progressive, the patient is refusing parenteral nutrition. Cyclic rifaximin is also required, because of small intestinal bacterial overgrowth.

DISCUSSION Systemic scleroderma is an autoimmune disease of the connective tissue with chronic progressive character. Pulmonary hypertension and gastrointestinal involvement are frequent finds. Typically, esophageal manometry shows aperistalsis and hypotonic lower esophageal sphincter; however, in our patient's case the results mimicked type 1 achalasia with higher integrated relaxation pressure. The profit from endoscopic dilatation was confirmed, as the vomiting stopped after this procedure.

CONCLUSIONS Systemic scleroderma is a rare disease with varying symptoms. In this case report, we want to point out that despite the long-term course of the disease, not enough attention was paid to the patient's complaints. Pulmonary hypertension, especially if accompanied by gastrointestinal or cutaneous findings, should lead to a suspicion of a systemic disease. Thus, blood tests or rheumatological examination should be considered

Key words
achalasia, esophagus, pulmonary hypertension, systemic scleroderma

BRADLEY WOOLFENDEN

A good history should not be sneezed at

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INTRODUCTION Patent foramen ovale (PFO) is relatively common in the general population and can be a potential cause of a paradoxical embolism. Temporal association between Valsalva maneuvers and stroke onset should raise concern for this potential mechanism. This case is an important example of how a working knowledge of physiology in the right clinical context can expedite an important diagnosis.

CASE DESCRIPTION A 54-year-old, left-handed, man presented to our emergency department with a 30-minute long episode of left arm and leg weakness, and associated persisting sensory disturbance. A sudden onset of these symptoms was heralded by a sneeze, that occurred while the patient was bending to put on his socks. Past medical history was notable for hypertension and allergic rhinitis. Social history was notable for recreational scuba diving. His National Institutes of Health Stroke Scale score on arrival was 2. Initial computed tomography scan of the brain and intracranial angiogram were normal. Laboratory investigations, electrocardiogram, and cardiac monitoring were all unremarkable. Magnetic resonance imaging (MRI)of the brain showed a small region of acute ischemia in the right frontal cortex and 2 old lacunar infarcts within the left corona radiata. Transesophageal echocardiogram was performed with bubble study, which demonstrated a clear right-to-left shunt of a patent foramen ovale on Valsalva maneuver. The patient was started on Aspirin and was referred for consideration of PFO closure. He was advised not to scuba dive and was started on a nasal spray and antihistamines for his allergic rhinitis.

DISCUSSION On initial presentation, our main differential diagnosis was a transient ischemic attack, due to the, now resolved, neurological features of unilateral weakness and paresthesia. Following the positive findings of multiple infarctions on the brain MRI, we explored the relationship between the sneeze and the onset of symptoms. Combined with the patient's past medical history of allergic rhinitis and his recreational scuba diving, we considered raised intrathoracic pressure as a mechanism leading to his presentation. He scored 6 points on paradoxical embolism risk score, which meant there was a 62% chance that this infarct was caused by a PFO. Taking into account all of the above, our proposed etiology for this presentation was a paradoxical embolism.

CONCLUSIONS Sneezing precipitating a paradoxical embolism via PFO is rarely described in the literature. This case highlights the importance of taking a clear history, which later guides stroke mechanic workup and treatment considerations. In this case, discovering the correct etiology may prevent further strokes in a young patient through a PFO closure device and relevant lifestyle modifications.

Key words
patent foramen ovale, stroke, transient ischaemic attack, Valsalva maneouver

CATERINA MARCOCCIA

Kidney disease and vasculitis: atypical presentation of a case of microscopic polyangiitis

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CASE DESCRIPTION We describe a case of a 65-year-old woman with a history of right lung mucinous adenocarcinoma (treated with surgery, chemotherapy and radiotherapy in 2010), who is now free of the disease. The patient reported a month-long history of asthenia, adynamia, myalgias, and deterioration of her general condition. She had upper respiratory tract catarrh and received amoxicillin and clavulanic acid treatment for 7 days. Systemic symptoms persisted, and they were accompanied by intolerance to food intake, nausea, gastric reflux, and vomiting, which prompted the patient to reach out for a consult. Blood testing showed mild leukocytosis and urine testing showed urea levels of 73 mg/dl, creatinine levels of 2.44 mg/dl, and urine sediment of 5–7 leukocytes and 0–1 red blood

cells. Computed tomography scan of the chest showed pulmonary infiltrate in the right apex. Treatment with levofloxacin for 7 days was prescribed. General deterioration persisted, with marked weight loss and nondysenteric diarrheal stools. The patient returned with new laboratory test results (urea, 139 mg/dl; creatinine, 4.5 mg/dl; urine sediment, few cells, isolated leukocytes and 6–8 red blood cells; leukocytosis with neutrophilia), and she was admitted to the hospital. The results were interpreted as a probable prerenal failure. Efforts were made to restore the fluid balance, unfortunately without success. Tubulointerstitial nephritis was suspected. Due to increasing acute kidney injury with acidosis, deterioration of urea and creatinine levels, erythrocyte sedimentation rate of 112 and C-reactive protein level of 177 mg/l, rapidly progressive glomerulonephritis was suspected. Methylprednisolone pulses and hemodialysis were started due to fluid overload and decreased diuresis. The patient presented with conjunctival injection and chemosis, pericatheter bleeding and dysfunction, neck hematoma with pharyngeal edema, sialorrhea, and dysphagia, and was admitted to intermediate therapy for monitoring and further diagnostic studies. Renal biopsy was deferred due to the patient's initial clinical condition and was performed 15 days later. Complementary studies were performed: FR, FAN and anti-DNA Ac were negative, while C3 and C4 were normal. Proteinuria of 0.26 g/24 hours and 0.30 g/l was noted. Uroproteinogram showed slight increase in albumin excretion. Results of monoclonal κ free light chains were very weakly positive. Diuresis of 860 ml/24 hours was noted. Plasma κ and λ light chains were normal. Immunoelectrophoresis showed no homogeneous bands. Renal ultrasound showed preserved cortical thickness. Renal biopsy showed 2/15 glomeruli with glomerulosclerosis, 1/3 with mild mesangial proliferation and matrix expansion, tubules with variable epithelial damage, lymphocytic and leukocytic exocytosis, tubular atrophy, hyaline, and granular casts. Interstitium with mixed mononuclear inflammatory infiltrate, with polymorphonuclear leukocytes, and isolated eosinophils with lymphoid aggregates and periglomerulitis with focal interstitial fibrosis were observed. Interlobular vessels with parietal thickening due to myointimal hyperplasia indicated tubulointerstitial nephritis. Negative IgA, G, M, c3 and C1q were found on microscopy.

Key words
acute kidney injury, microscopic polyangiitis, vasculitis

CÉSAR JAVIER MARTÍNEZ

Acute myeloid leukemia in a patient with a diagnosis of Behçet disease

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INTRODUCTION Behçet disease (BD) is a multisystemic vasculitis, involving both arteries and veins, characterized by recurrent oral and genital ulcers with typical cutaneous and ocular features. There are a few case reports describing the association of malignancies with BD. Among hematological malignances, reports of acute myeloid leukemia (AML) are rare. The authors report a case study involving a patient afflicted by Behçet disease and recently diagnosed with AML.

CASE DESCRIPTION A 17-year-old woman was admitted to a public hospital in Catamarca (Argentina) in July 2023, due to persistent abdominal pain, vomiting, fever, and pain in the lateral-right region of her neck with presence of cutaneous tumors. Laboratory analysis showed anemia (7.9 g/dl), leukocytosis (19 630 /mm^3) and thrombocytopenia (76 000/mm^3). The patient underwent multiple thromboses, involving several veins in upper limbs, neck, right subclavian region, and right transverse and sigmoid sinuses. She was prescribed anticoagulant and gamma globulin treatment. The patient suffered an ischemic stroke with no possibility of thrombolysis. She developed permanent neurological sequelae: bilateral hearing and vision loss, and right-sided brachio-crural hemiparesis. Hematological and rheumatological analysis came back negative for potential causes of thrombosis.

The patient had skin tumors on upper limbs (pyoderma gangrenosum). Skin biopsies showed small vessel neutrophilic vasculitis and neutrophilic dermatitis (intermittent flare-ups and remissions). Pathergy test was negative. Bicytopenia, increased acute phase reactants (elevated inflammatory markers, ferritin and C-reactive protein), and skin findings were interpreted as a reactivation of BD. Pulses of methylprednisolone and adalimumab were administered successfully. The patient had multiple convulsive episodes, which were treated with levetiracetam and phenytoin. Electroencephalogram showed low amplitude without epileptiform discharges. Imaging showed left frontoparietal ischemic hypodensity, petechial hemorrhages, intracranial vasculitis, and right temporal arteritis. Meningitis was ruled out. The patient was anemic and thrombocytopenia was found. Based on the following findings, a diagnosis of intermediate risk AML was established. Flow cytometry showed 30% myeloid blasts– and peripheral blood smear showed 15% immature cells. The patient started induction chemotherapy (7+3 regimen) and consolidation chemotherapy (rHDRAC and idarubicin regimen).

DISCUSSION BD is a multi-systemic vasculitis. Neutropenia may be caused by BD itself or by associated myelodysplastic syndrome (which may cause bone marrow failure). Our patient did not suffer from leucopenia, but she had anemia and thrombocytopenia. Treatment needs to be tailored to the clinical manifestations of BD. Systemic glucocorticosteroids, azathioprine, colchicine, and tumor necrosis factor inhibitors are effective. Allogenic hematopoietic stem cell transplant (AHSCT) is considered curative for AML and effective in inducing remission in BD. In our patient, the prospect of AHSCT is currently under consideration.

CONCLUSIONS In the presence of clinical features such as recurrent oral and genital ulceration, and deep vein thrombosis, BD diagnosis is not difficult. However, our patient's marked cytopenia drew attention to a second diagnosis: AML. Published literature has evidence of both BD and AML coexistence, and leukemia masquerading as BD. This clinical scenario remains a challenge and perhaps the degree of response to antileukemic therapy in this patient will help resolve this dilemma.

Key words
acute myeloid leukemia, allogenic hematopoietic stem cell transplant, Behçet disease, neutropenia, pathergy reaction

CHOWDHURY ADNAN SAMI

Unmasking disseminated histoplasmosis: 18-month battle of a non-HIV young febrile man

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INTRODUCTION Disseminated histoplasmosis (DH) is a rare systemic fungal infection caused by Histoplasma capsulatum. This infection occurs through high inoculum of microconidia inhaled through alveoli, direct invasion or in individuals with a disrupted immune system. In a tuberculosis (TB) endemic zone like Bangladesh, disseminated histoplasmosis is not fully appreciated and is hardly reported, which can attributed to its similarities to TB.

CASE DESCRIPTION A 19-year-old febrile man presented with weight loss, skin rash, and lymphadenopathy. He was treated with antitubercular medications for more than 12 months in multiple insti-

tutions, based on repeated reports of lymph node biopsies showing granulomatous lesions suggestive of tuberculosis. Before he was finally diagnosed with DH at Bangabandhu Sheikh Mujib Medical University, 18 months after the onset of his disease, he had already lost 20 kg of weight and developed multiple inflammatory small joint pains together with back pain, on top of his previously mentioned symptoms. Blood tests performed at Bangabandhu Sheikh Mujib Medical University showed hemoglobin levels of 9.0 gm/dl, white blood cell count of 1.87 $\times$ 10^9/l and platelets count of 22 $\times$ 10^9/l. Peripheral blood film was consistent with pancytopenia. Bone marrow study showed normoblastic erythroid hyperplasia. The patient's adrenal reserve, serum angiotensin-converting enzyme, antinuclear antibodies, rheumatoid factor, immunoglobulin (Ig) G, IgM, IgA, and serum uric acid were normal. Interferon-gamma release assay, anti-HIV 1/2, and enzyme-linked immunosorbent assay were negative. X-ray of the right hand showed multiple periarticular lytic lesions. Magnetic resonance imaging of the spine showed spondylodiscitis of the third lumbar (L) vertebrae and resolving abscesses at L3–L4 level. Skin, bone, and lymph nodes biopsies showed noncaseating granulomas with periodic acid-Schiff stain positive for budding oval yeast cells. Fungal culture revealed growth of a dimorphic fungus in Sabouraud's dextrose agar, suggestive of Histoplasma. After 2 weeks of treatment with intravenous liposomal amphotericin B and continued itraconazole, the patient's fever subsided, his joint pain reduced, and his well-being improved. He started to gain weight and skin lesions started to heal.

DISCUSSION Histoplasma is endemic in many parts of the world, especially the United States and Latin America. Disseminated histoplasmosis is rarely reported in Bangladesh, where its first and second cases were reported in 1982 and 2005, respectively. Although Bangladesh is thought to be an endemic risk zone for histoplasmosis, only 26 cases were reported in a systematic review conducted in 2020. It might be due to the symptoms mimicking a granulomatous disease (tuberculosis), a lack of suspicion from clinicians, or scarcity of laboratory support in many places. Disseminated histoplasmosis is a disease of immunocompromised patients, but it's very rare in immunocompetent patients, especially when involving the skin, joints, vertebrae, and bone marrow.

CONCLUSIONS This case serves as a significant reminder to consider an alternative diagnosis of histoplasmosis in febrile patients who fail to show improvement with antitubercular treatment for granulomatous lesions in a tuberculosis-endemic zone, even if the patient is immunocompetent.

Key words
disseminated histoplasmosis, fungal infection, pyrexia of unknown origin, prolonged pyrexia

DARIA KELLER

Diagnosing anemia: can a rare disease complicate the diagnostic path?

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INTRODUCTION As the global population ages, the prevalence of anemia in the elderly becomes a critical health concern. Recent evidence highlights that anemia in this demographic signals poor health and increased vulnerability. This case report details the diagnostic journey of an 84-year-old woman, emphasizing the dual etiologies of anemia resulting from heart failure caused by cardiac transthyretin amyloidosis (ATTR-CM) and coexisting colon cancer.

CASE DESCRIPTION An 84-year-old woman presented to a cardiology clinic with heart failure and suspected cardiac amyloidosis. Concurrent diagnoses included chronic kidney disease, anemia, hypertension, and persistent atrial fibrillation. The final diagnosis made in May 2023 showed ATTR-CM and treatment with tafamidis commenced in June. Despite transient relief from heart failure symptoms, the patient reported weakness and limited exercise tolerance in July. Laboratory results indicated signs of anemia and iron deficiency.

In September 2023, the patient reported weight loss, potentially due to progressive cachexia from advanced heart failure. Laboratory tests still indicated iron deficiency, leading to a daylong hospitalization for intravenous iron administration. However, in November, the patient's hemoglobin dropped significantly, necessitating transfer to the department of internal diseases for blood transfusion. Tumor marker assessment showed elevated carcinoembryonic antigen (CEA) levels. Imaging identified irregular thickening in the ascending colon, prompting a colonoscopy that showed adenocarcinoma G2.

In December, the patient underwent right hemicolectomy with complications (renal failure requiring hemodiafiltration among others). Despite the challenges, the patient was discharged from the hospital in a stable condition in January 2024.

DISCUSSION A dual etiology for iron deficiency anemia was identified, involving heart failure caused by ATTR-CM and concurrent colon cancer. The intricate interplay between anemia, heart failure, and advanced age exacerbates health challenges. Anemia further strains the cardiovascular system, leading to such symptoms as fatigue and dyspnea. Timely screening becomes vital to uncover potential malignancies, obscured by the complex interplay of conditions.

LESSONS TO BE LEARNED This case emphasizes the critical need to diligently explore the hidden agenda in medical diagnoses. The fact that colon cancer was discovered while addressing the primary condition (ATTR-CM) underscores the risk of overlooking a secondary, life-threatening diseases. Recognizing overlapping symptoms requires a comprehensive and collaborative approach, ensuring a thorough consideration of all aspects of a patient's health.

In conclusion, this case highlights the intricate nature of medical challenges in the elderly, where anemia serves as a clinical indicator of underlying health issues. The importance of a comprehensive diagnostic approach, especially in the presence of rare diseases, cannot be overstated. The lessons drawn from this case underscores the need for a holistic perspective to optimize outcomes and enhance overall care quality.

Key words
anemia, cardiac transthyretin amyloidosis, colon cancer, heart failure, iron deficiency

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More than just renal impairment

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INTRODUCTION Thrombotic microscopic angiopathy (TMA) presents with thrombocytopenia, macroangiopathic hemolytic anemia (MAHA), and end organ damage, including acute kidney injury. TMA can be classified into primary (a disorder of complement regulation) and secondary (a condition resulting from another disease process). Waldenström macroglobulinemia (WM) is a lymphoplasmacytic lymphoma that results in the presence of an IgM monoclonal protein in the blood and lymphoplasmacytic lymphoma in the bone marrow.

CASE DESCRIPTION A 50-year-old man presented to the emergency department with shortness of breath that had been lasting for 2 weeks. He reported significant fatigue and anorexia. At baseline,

the patient was in good health and moderately active. Prior to the onset of the symptoms, he had 3 episodes of watery diarrhea. On review, the patient was in renal failure with fluid overload and hemodialysis was commenced. Renal failure persisted after admission. Autoimmune and viral screens were negative. His hemoglobin and platelet count, low on presentation, continued to fall with schistocytes on blood film.

Renal biopsy was performed, showing thrombi within the capillaries of the glomerulus on light microscopy. Immunofluorescence was negative for κ light chains or complement deposition. Findings were consistent with TMA. ADAM metallopeptidase with thrombospondin type 1 motif 13 was negative. Eculizumab, a complement 5 inhibitor, was initiated for primary complement disorder. The patient's renal function did not recover and he remained dependent on hemodialysis.

After 2 weeks, electron microscopy showed short fibrils within the thrombi, suggesting lymphoplasmacytic invasion of the kidney. Bone marrow biopsy confirmed a diagnosis of WM and combination therapy of rituximab, dexamethasone, and cyclophosphamide was commenced. Only partial remission was achieved with combination therapy. The patient was started on ibrutinib, a tyrosine kinase inhibitor. Currently, light chains are undetectable and the patient is under consideration for a renal transplant. Ibrutinib would be continued following transplantation to prevent relapse. However, an adverse effect of ibrutinib is platelet dysregulation, which is a concern when undergoing surgery.

DISCUSSION The results of light microscope showed thrombi within the vasculature supplying the glomerulus, which is in keeping with TMA. Immunofluorescence did not show any immune depositions within the kidney, suggesting that paraprotein deposition was not the primary cause of the renal impairment. WM affects 3 in 1 000 000 people per year. A *MYD88* genetic mutation is found in 90% of patients, just like in this case. A broad spectrum of the disease exists with more severe forms presenting with hyperviscosity syndrome. Treatment is based on disease severity. This case is unusual, as renal impairment at presentation is not common in WM and only occurs in 3% of cases. At presentation, this patient was in end-stage renal failure with secondary TMA.

CONCLUSIONS WM is a rare condition. Rarer still is for it to present with renal implications. This case highlights the importance of a comprehensive assessment of a patient presenting with renal impairment. Plasma cell disorders and TMA are important considerations for patients with acute kidney injury. A multidisciplinary approach is essential in the diagnosis and management of this condition.

Key words
acute kidney injury, plasma cell disorder, renal failure, thrombotic microscopic angiopathy, Waldenström macroglobulinemia

DAVID KISS

Cardioneuroablation? Investigate first!

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CASE DESCRIPTION We present an interdisciplinary case report of a 40-year-old man with newly developed recurrent syncope and presyncope states. Our case shows a rare cause of bradycardia with a difficult differential diagnosis, including internal and neurological assessment. Limbic encephalitis is a rare disease with many symptoms, including autonomic system dysfunction and syncope. Arrhythmias are most often described in limbic anti-LGI1 encephalitis. Cardioneuroablation is an alternative therapeutic option which makes it possible to avoid a, potentially futile, pacemaker implantation, mainly in young adults with proven increased vagal tone. A 40-year-old man was admitted to the district hospital's department of neurology for further investigation of recurrent disorders of consciousness. Neither the initial laboratory examination nor the electrocardiogram showed any significant pathologies. Essential biochemical examination, thyroid hormones, and cardioenzymes were within the normal range. Initially, after comprehensive cardiological and neurological assessments, a primary rhythm disorder was thought to be the cause of the patient's symptoms. The initial electroencephalogram (EEG) examination was unremarkable for syncope with asystole during the examination. On telemetry, sinus arrests for up to 40 seconds occurred. The clinical manifestations included loss of consciousness, hypotonia, and presyncope states. A temporary pacemaker was introduced and during the procedure, a paroxysm of atrial fibrillation was also induced. Subsequently, the patient was transferred to our clinic's intensive care unit for further investigation. Within the comprehensive internal investigation, we performed computed tomography of the thorax and cardiac magnetic resonance imaging (MRI), which also did not show significant pathologies. Lyme disease was also excluded. Due to persistent symptomatic bradycardias, implantation of a permanent pacemaker or cardioneuroablation was considered. After a positive atropine test, cardioneuroablation and isolation of the pulmonary veins were successfully performed. Following the procedure, the patient was telemetrically monitored and showed no recurrence of bradyarrhythmia. However, the symptoms reoccurred a month after discharge, although the clinical manifestations were different this time. The patient was admitted to the district hospital's department of neurology, where a comprehensive neurological assessment was repeated. EEG showed abnormal findings, and there were signs of lymphocytic pleocytosis with a suspicion of encephalitis in the cerebrospinal fluid. Brain MRI confirmed unilateral limbic encephalitis. LGI1 antibodies were present in our patient's cerebrospinal fluid. The patient was then transferred to a specialized center for specific treatment based on immunosuppression. This case report represents a rare secondary rhythm disorder in a primary neurological disease. It shows the difficulty of the differential diagnostic process of syncope and presyncope. Even if cardioneuroablation is a viable therapeutic option for the treatment of bradycardia in selected patients, secondary causes should also be excluded. As part of the diagnostic process in young adults with a rapid onset of symptoms usually unrelated to their age, we should also consider rare diseases, such as encephalitis. Brain MRI, EEG, and cerebrospinal fluid analyses may also be beneficial.

Key words
bradycardia, cardioneuroablation, limbic encephalitis, vasovagal syncope

DEBI MOAGI

Unveiling the rarity: bilateral embryonal rhabdomyosarcoma causing kidney failure in a young adult

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INTRODUCTION Bilateral multifocal kidney masses on imaging present diagnostic and therapeutic challenges, often warranting consideration of rare aetiologias, such as renal malignancies. Rhabdomyosarcoma (RMS), an aggressive soft tissue tumor, primarily observed in pediatric populations, is an exceptionally uncommon finding in adults, often presenting late and with metastasis. Renal RMS can mimic genitourinary tuberculosis (TB), renal cell carcinoma,

and nephroblastoma, both clinically and on imaging, making its diagnosis challenging. Herein, we present a rare case of bilateral, adult embryonal rhabdomyosarcoma of the kidneys, which was diagnosed following radical nephrectomy for persistent macrohematuria. This case report highlights the complexity of diagnosing and managing an adult patient with bilateral multifocal kidney masses, emphasizing the importance of considering alternative diagnoses.

CASE DESCRIPTION A 32-year-old woman with dialysis-requiring kidney failure presented with constitutional symptoms and bilateral kidney masses. Laboratory findings showed anemia and raised inflammatory markers. Kidney ultrasound demonstrated enlarged kidneys with bilateral dilated renal calyces with soft tissue masses and associated parenchymal distortion. Initial suspicion of renal TB led to the initiation of empirical standard TB treatment. Mycobacterium cultures were reported to be negative after 42 days of incubation. Despite negative microbiological evidence, she was treated as possible culture-negative disseminated TB. Subsequent clinical deterioration and computed tomography scan findings suggestive of hemorrhage in the right kidney prompted right nephrectomy. Histopathological examination of the right kidney was in keeping with embryonal RMS (ERMS). Left nephrectomy is planned for the near future.

DISCUSSION Bilateral kidney masses pose diagnostic and therapeutic dilemmas, encompassing various differential diagnoses, including rare malignancies, such as RMS, inherited conditions, such as autosomal dominant polycystic kidney disease (ADPKD), and infectious conditions, such as TB. ERMS, exceedingly rare in adults, mimics other renal pathologies, necessitating histopathological confirmation for accurate diagnosis. Radical nephrectomy remains the mainstay of therapy, although adjuvant therapies may play a role. Urine culture is the gold standard of genitourinary TB diagnosis. The treatment of drug-sensitive genitourinary TB is the same as standard treatment for pulmonary TB (antituberculosis medication continued for 6 months).

LESSONS TO BE LEARNED This case underscores the importance of considering rare malignancies, such as ERMS, in the differential diagnosis of bilateral kidney masses. Prompt histopathological evaluation is crucial for definitive diagnosis, especially in cases refractory to empiric treatment to a presumed diagnosis and those with a complex clinical picture that does not fit a specific disease process. Vigilance for extra-pulmonary manifestations of TB is vital, particularly in TB endemic areas. The complex clinical course underscores the challenges of distinguishing between malignancy and infectious etiologies in renal masses. Imaging alone is insufficient to distinguish between malignant and benign causes of renal masses, and microbiological evidence of TB should be sought in suspected cases. Delays in diagnosis could lead to potentially fatal outcomes in patients who have rarer causes of bilateral kidney masses, which are often malignant.

Key words
bilateral, kidney, masses, rhabdomyosarcoma, tuberculosis

DELISA HLAWE

A case of mitral stenosis presenting with life-threatening hemoptysis

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INTRODUCTION Mitral stenosis is a rare cause of life-threatening hemoptysis. However, in low-income countries rheumatic heart disease is still prevalent. Various mechanisms have been described in the pathogenesis of hemoptysis in mitral stenosis. Pulmonary apoplexy is the most likely cause in cases presenting with large volume hemoptysis.

CASE DESCRIPTION We report a case of a 49-year-old man without any known comorbidities and no prior lung disease, presenting with hemoptysis, pulmonary hypertension, and pneumonia. Subsequent clinical decompensation required the institution of full organ support. A diagnosis of previously undiagnosed mitral valve stenosis was made. His acute deterioration was likely precipitated by an infectious process complicated by pulmonary apoplexy.

CONCLUSIONS Clinicians should have a high index of suspicion for alternative causes of life-threatening hemoptysis in patients without apparent risk factors and no prior history of lung disease.

Key words
hemoptysis, mitral stenosis

DHINAGAREN NARAYANAN

An extremely rare case of locally recurrent adenocarcinoma of the spleen with unknown primary

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CASE DESCRIPTION The incidence of splenic tumor is relatively low compared to other organs. Sometimes, they are incidentally discovered on imaging. The majority of primary splenic tumors are diagnosed as benign. Primary malignant tumor of the spleen most commonly involves lymphosarcoma, reticulosarcoma, angiosarcoma, and fibrosarcoma. The diagnostic algorithm to determine the nature of the splenic tumor should include series of laboratory tests, imaging studies, and positron emission tomography (PET) scans. Sometimes it can warrant the need for splenectomy followed by immunohistochemistry profiling. While the occurrence of primary splenic adenocarcinoma is rare, this case study reports recurrent primary splenic tumor with poorly differentiated adenocarcinoma in a patient, who initially presented with (and was investigated for) anemia of unknown cause. To the best of our knowledge, this case has not been reported before, hence this study provides insight into the response to standard treatment for locally recurrent adenocarcinoma of the spleen with unknown primary.

Key words
cystokeratin AE1/AE3, cystokeratin 5D3, cystokeratin 7, epithelial membrane antigen, paired-box gene 8

DUŠAN POLÁK

Use of lumen-apposing metal stent in a patient with advanced pancreatic cancer

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INTRODUCTION The use of lumen-apposing metal stents is becoming increasingly common in gastroenterology. The most common indications are pancreatic fluid collections after acute pancreatitis with the formation of a pseudocyst or collections filled with fluid and necrotic tissue of the pancreas. These stents can also be used as endoscopic biliodigestive anastomoses, connecting the digestive tract with the biliary tract to overcome malignant biliary obstruction.

CASE DESCRIPTION We present the case of a 73-year-old patient with locally advanced, histologically confirmed adenocarcinoma of the pancreas. The patient was hospitalized for loss of weight, weak-

ness, and the development of jaundice and hepatopathy. A tumor of the head of pancreas was diagnosed on computed tomography. Due to the progression of jaundice and hepatopathy, we performed endoscopic retrograde cholangiopancreatography. However, it was no possible to overcome the stenosis in the intrapancreatic part of the bile duct. Histological confirmation of advanced pancreatic adenocarcinoma was obtained from a peripancreatic lymph node. The opinion of our pancreatic-biliary surgical team was that at this stage the patient was inoperable. The tumor had grown into major vessels and malignant lymphadenopathy was present. Since jaundice was progressing, we needed to bridge the malignant obstruction. We decided to create a biliodigestive endoscopic anastomosis. We could endoscopically connect the duodenum with the bile duct or gallbladder, or the stomach with the gallbladder, possibly with the bile duct. Everything depended on the anatomical conditions in the tumorous terrain. Anatomically, the connection of the stomach with the gallbladder seemed to be the best option. We created a nonsurgical anastomosis between the digestive tract and the biliary tract using the lumen-apposing metal stent with endoscopic ultrasonography guidance. We achieved adequate biliary drainage without surgery or X-ray intervention, without extracorporeal drainage of bile.
CONCLUSIONS These mini-invasive endoscopic methods are constantly being developed and improved in gastroenterology. There are always new indications and possibilities of their use in clinical practice, which is very beneficial for our patients, which our case report exemplifies.

Key words
biliodigestive anastomosis, case report, lumen-apposing metal stent, pancreatic cancer

ESTER KURAŠOVÁ

Deterioration of transplanted kidney function due to BK virus infection: what are the therapeutic options?

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INTRODUCTION BK virus nephropathy (BKVN) is an important complication that causes allograft dysfunction, particularly during the first year after transplantation. Our case exemplifies the complexity of diagnosing and treating patients after kidney transplantation, with acute kidney injury, in the setting of BKVN.
CASE DESCRIPTION We report a case of a 65-year-old patient with end-stage renal disease due to IgA nephropathy, who developed a slightly progressive renal dysfunction at a creatinine level of 191 µmol/l, 3 months after a second kidney transplantation from a deceased donor. Significant finding of BK viremia was detected in the blood through polymerase chain reaction (PCR). The patient underwent protocol biopsy that confirmed advanced BKVN. Within the primary therapy, immunosuppressive therapy was reduced, aiming for lower levels of immunosuppressants. Subsequently, administration of high doses (2 g/kg) of intravenous immunoglobulins (IVIGs) was started for persistent BK virus (BKV) replication, with a significant effect. BKV levels decreased (from 50 000 to 30 000 copies in serum), which led to the stabilization and repair of renal function. Subsequent rebiopsy was performed after the end of treatment, which detected histological regression of the lymphocytic infiltrate and repair of the renal parenchyma with minimal positivity of the SV40 BKV surface antigen in immunohistochemical examination. Two years later, worsening of renal function was seen (creatininemia, 190–200 µmol/l) and PCR detected new BKV positivity in the blood. Uncomplicated protocol biopsy was performed and late BKVN was confirmed. The histological sample was sent for molecular microscope examination (MMDx) in IKEM (Institute for Clinical and Experimental Medicine in Prague). It proved a coexistence of early humoral rejection, which was not found in the classical histological examination. Combined therapy with immunoadsorption and IVIGs was administered. The treatment ended with rituximab. BKV count in the blood was under the qualifying limit and renal function remains stable at a creatinine level of 174 µmol/l.
CONCLUSIONS BKVN presents a challenge for renal transplant outcomes. Early routine screening for BKV allows for an intervention to prevent the development of BKVN and permanent allograft damage. While immunosuppression reduction is the first-line therapy, second-line therapy is not well established. IVIGs seems to be an effective treatment for persistent BKV and can be considered as a therapeutic option in these patients. The therapeutic dilemma represents a coincidence of BKVN with acute rejection. A new MMDx technology represents the future of diagnostics.

Key words
acute rejection, BK virus nephropathy, intravenous immunoglobulins, kidney transplantation, molecular microscope examination

ESZTER SÁRA KORMÁNYOS

Rare causes of common symptoms: chronic diarrhea and primary Addison disease as a complication of multiple myeloma

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INTRODUCTION Chronic diarrhea is defined as diarrhea lasting for over 4 weeks. It is complicated by fluid and electrolyte loss, and consequential hypotension. Because of the wide range of causes, the history, symptoms, and their development over time need to be considered in a complex approach to establish the correct diagnosis.
CASE DESCRIPTION A 62-year-old man with multiple myeloma was transferred from the infectious diseases department for further examination and treatment of chronic diarrhea and hypotension lasting for 2 months.

The patient had been diagnosed with multiple myeloma 2 years prior. By then, cardiac and renal complications of the disease had already developed. When the patient was admitted to the infectious diseases ward, the diarrhea had already been present for a month. He had more than 20 stools a day, was dehydrated, and had low blood pressure. It was assumed that the symptoms were caused by a gastrointestinal infection. The stool culture test initially showed toxin-producing *Clostridium difficile*, for which the patient received targeted oral vancomycin therapy, but the frequency and amount of stools were not significantly reduced. Furthermore, no significant abnormalities were seen during the abdominal ultrasound examination. The patient was treated with cholestyramine, diosmectite, and protein supplementation, but diarrhea did not resolve. Even with parenteral fluid replacement, persistently low blood pressure was recorded. After ruling out the infectious etiology, the patient was referred to the gastroenterology department. The investigation was continued with endoscopy. No abnormalities, other than hiatal hernia, were detected on gastroscopy. Gastric histological test was negative for *Helicobacter pylori* and the duodenal sample showed no sign of coeliac disease. No macroscopic disorders were seen on colonoscopy. Microscopic examination confirmed amyloidosis.

The patient's initial high stool count gradually decreased after administration of loperamide and budesonide. Fever did not occur. However, contrary to expectations, the patient's blood pressure did not rise when diarrhea had ended. He was unable to get out of bed due to hypotension. At this time, it occurred to us that perhaps our approach was wrong and diarrhea was not the cause of the patient's low blood pressure. Therefore, further laboratory tests were performed. Hypotension, previously thought to be a consequence of diarrhea, was finally confirmed to be caused by severely low (3 nmol/l) cortisol levels. Given the underlying illness (multiple myeloma), this proved to be Addison disease—an adrenal manifestation of amyloidosis. The patient's blood pressure normalized after a single dose (100 mg) of hydrocortisone. He was discharged with orders to take 9 mg of budesonide daily.

DISCUSSION Chronic diarrhea is not always caused by a gastroenterological disease. Simultaneous diarrhea and low blood pressure may not be causally related. Persistent hypotension may indicate other, less common etiologies.

CONCLUSIONS When faced with seemingly unexplained, persistently low blood pressure, the possibility of adrenocortical insufficiency should be considered.

Key words
Addison disease, chronic diarrhea, multiple myeloma

GISEL BEATRIZ VERA RODRIGUEZ

Hidden behind dermatomyositis

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INTRODUCTION Dermatomyositis is an idiopathic inflammatory myopathy (IIM), a heterogeneous group of chronic systemic autoimmune myopathies associated with high morbidity and functional disability. It is a rare condition, which epidemiologically occurs more frequently in adults between the ages of 40 and 60, predominantly in women. Strong association between dermatomyositis and its coexistence with malignant neoplasms has been described, but association with primary central nervous system lymphoma is extremely rare. Herein, we present a case of dermatomyositis as a paraneoplastic syndrome of B-cell non-Hodgkin lymphoma, primary to the central nervous system and localized in the infratentorial region.

CASE DESCRIPTION A 29-year-old man from Bolivia, with no significant past medical history, was referred to our unit for pruritic erythematous lesions with well-defined borders on the face, elbows, wrists, and knees, which presented 3 months prior to the consultation in association with dysphagia (initially to solids, progressing to liquids), feverish sensation, and ascending muscle weakness of the lower limbs (later involving the pelvic girdle and forearms). Upon admission, elevated creatine phosphokinase, lactate dehydrogenase, liver transaminases, and acute phase reactants were noted, with negative serology. Lumbar puncture was unremarkable and electromyography indicated inflammatory myopathy. Suspecting dermatomyositis, corticosteroid pulse therapy was initiated with a good clinical response, followed by cyclophosphamide. Due to the strong association of this pathology with adjacent neoplasia, computed tomography (CT) of the neck, chest, abdomen, and pelvis, and testicular ultrasound were performed. They showed no associated lesions, and subsequently the patient was discharged from the hospital. Five months later, he was readmitted due to a 10-day history of holocranial headache of an 8 out of 10 intensity, accompanied by vomiting. CT scan of the brain showed a heterogeneous hyperdense image in the left cerebellar hemisphere with a mass effect. Brain magnetic resonance imaging showed hypointense lesions with hyperintense edges in T1- and T2-FLAIR sequences, with peripheral contrast enhancement. Tumoral excision was performed at the neurosurgery department. A sample taken for pathological anatomy showed findings compatible with diffuse B-cell non-Hodgkin lymphoma of germinal center phenotype. The patient decided to proceed with the chemotherapy treatment at the local hospital.

CONCLUSIONS According to the literature, neurological paraneoplastic syndromes occur in less than 1% of solid tumors and are uncommon in lymphomas, with the most frequent location being supratentorial. The latest updates on dermatomyositis recommend performing CT scans of the chest, abdomen, and pelvis in search of occult tumors. Brain CT scans are not part of the initial screening. However, after the presentation of this case, we emphasize the importance of considering it when approaching the follow-up of these patients, as an early diagnosis of associated neoplasms allow for targeted and timely treatment.

Key words
dermatomyositis, hidden, lymphoma, syndrome

JÁN KILÍK

The patient's hidden enemy in a long-term hemodialysis program

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INTRODUCTION Calciphylaxis, or calcifying uremic arteriolopathy, is a rare but serious condition, especially in patients with end-stage chronic kidney disease. Calciphylaxis significantly increases patient morbidity due to nonhealing wounds and severe pain, resulting frequent hospitalizations. The annual mortality of patients with calciphylaxis is greater than 50%, mainly due to sepsis. However, an early diagnosis with following treatment significantly improves the quality of life and patient prognosis. The goal of this case report is to draw attention to this rare condition and to clarify the pathophysiological, diagnostic, and therapeutic possibilities.

CASE DESCRIPTION We present a case of a 34-year-old polymorbid patient with type 2 diabetes mellitus and multiple organ complications, who has been participating in the long-term dialysis program since 2022. The patient was referred to our department from a hemodialysis center due to a chronic infection. He presented with numerous dry necroses of the lower limbs and penis that occurred without an obvious traumatic cause, and nonhealing wounds after amputations on the lower extremities. Laboratory testing showed high inflammatory activity indicative of sepsis, with secondary hypocoagulation, hyperphosphatemia, and secondary hyperparathyroidism. Angiography confirmed presumed ischemic disease of the lower limbs. Since angiographic findings did not explain the presence of pathological lesions outside of the predilection areas, and due to the fact that the patient had been participating in a long-term dialysis program, calciphylaxis was suggested as the diagnosis. The diagnosis was confirmed through skin biopsy. At that time, the patient was undergoing hemodialysis 3 times a week for an average time of 3–4 hours, by his own decision and tolerance. After the diagnosis, the frequency and duration of hemodialysis sessions was increased in an effort to achieve maximum dialysis flow rates. Complex antibiotic therapy was applied, and phosphate binders were added. A discussion about using thiosulfate sodium was started. However, within a short period of time after the hospitalization, the patient died.

CONCLUSIONS Calciphylaxis, or calcifying uremic arteriolopathy, is a rare and severe complication, especially in patients with chronic renal failure. The prevalence of calciphylaxis reaches approximately 0.04% to 4%. Pathophysiologically, calcium deposition gathers in the tunica media of small arteries and arterioles with subsequent secondary thrombosis, resulting in obliteration of the tissue due

to ischemia. Predilection areas are located in the subcutaneous fat tissue, especially in the thighs, buttocks, and finger joints. An extensive multidisciplinary therapeutic approach is required, with the cooperation of internal and surgical departments. Despite intensive medication, the annual mortality of the disease is at the level of 50%.

Key words
calciphylaxis, chronic kidney disease, hemodialysis, necrosis, sodium thiosulfate

JAROSLAV ZEMBJAK

The smart watch: not just for detection of atrial fibrillation

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CASE DESCRIPTION Despite the advanced technology and Holter electrocardiogram recorders that patients can wear for days or weeks at a time, it is still difficult to detect atrial fibrillation. Implantable loop recorders or less invasive smartwatches are also used for this purpose. In those cases, detection of other heart rhythm disorders, especially supraventricular tachycardias, is even more difficult. We present a case of a 34-year-old woman with a history of palpitations (complication of COVID-19) and confirmed tetany with no other significant internal underlying diseases, whose issues could not be documented for a long time. She repeatedly experienced atrial fibrillation, although it did not cause her significant problems. As her condition recurred several times a month, she decided to buy an Apple watch. After she had begun wearing the smartwatch, she experienced palpitations with a frequency of 210 bpm. The device detected supraventricular tachycardia with subsequent transition to atrial fibrillation, with rapid transfer to the ventricles. Because of the finding, we performed an electrophysiological examination, where a jump with retrograde echo and repeated inductions of typical atrioventricular nodal reentry tachycardia occurred during the atrial protocol, followed by a successful ablation of the slow path of the atrioventricular node. With the progress of science and technology, we can help patients more quickly and clearly. Smartwatches may not only be useful in detecting atrial fibrillation, but also in other rhythm disorders. It is necessary and appropriate to use the given knowledge to improve the quality of differential diagnosis in ambiguous cases.

Key words
atrial fibrillation, atrioventricular nodal reentrant tachycardia, smartwatch, supraventricular tachycardia

KAROL GRAŇÁK

Rapidly-progressing glomerulonephritis as an unusual complication of sarcoidosis

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INTRODUCTION Sarcoidosis is a multisystemic inflammatory disease of unknown etiology, characterized by formation of noncaseating epithelioid granulomas. Clinically, significant renal involvement is rare in sarcoidosis. It most commonly manifests as chronic tubulointerstitial nephritis and nephrocalcinosis with nephrolithiasis. On the other hand, glomerular involvement is observed sporadically, mainly membranous glomerulopathy or focal segmental glomerulosclerosis. Rapidly progressive glomerulonephritis (RPGN) represents one of the most serious conditions in clinical nephrology. It is characterized by presence of crescents in histological examination of a kidney biopsy and rapid deterioration of renal function with active urinary sediment. Without prompt initiation of treatment, it can lead to end-stage renal damage.

CASE DESCRIPTION We describe a clinical case of a 49-year-old patient with a history of sarcoidosis of the lungs and intrathoracic lymph nodes, who was hospitalized for acute kidney injury, hypercalcemia, hypoxemic respiratory insufficiency, general weakness, weight loss, and fever. Conservative treatment was not successful, therefore early initiation of renal function replacement in the form of intermittent hemodialysis was necessary. During the differential diagnosis process, we found nephrotic range proteinuria with microscopic hematuria. Complement and autoantibody levels were normal. Histologically, a unique constellation of renal lesions in the form of severe chronic tubulointerstitial nephritis combined with diffuse sclerosing crescentic glomerulonephritis was confirmed. Electron microscopy showed no immune complex deposits. Computed tomography scan of the lungs showed recurrence of sarcoidosis and subpleurally localized primary calcified tuberculous infection. Due to the extent of the irreversible changes in the kidneys and the patient's risk profile (latent tuberculosis), we decided to keep the patient on prednisone treatment and not to add other immunosuppressants or cytotoxic agents. Treatment with corticosteroids was initiated in collaboration with a pulmonologist, with rapid improvement in the patient's extrarenal clinical condition.

DISCUSSION Sarcoidosis in not routinely considered to be a part of the differential diagnosis of glomerular involvement, especially RPGN. Instead, it is diagnosed based on the exclusion of other possible conditions. The fundamental problem is that there are no clinical or histopathological characteristics that distinguish glomerulopathy in sarcoidosis from the primary forms. Considerable literature is available to guide the management of RPGN not related to sarcoidosis (initial use of cytotoxic or antilymphocyte agents, followed by the use of maintenance combined immunosuppressants), but it is unclear whether this treatment can be applied to RPGN related to sarcoidosis.

CONCLUSIONS Glomerular involvement of sarcoidosis is a rare cause of RPGN and it can even precede the diagnosis of systemic sarcoidosis. It is important to think about this case as well, as delayed diagnosis and treatment significantly reduce the probability of renal function restoration. The management of sarcoidosis-related RPGN remains a clinical challenge and requires multidisciplinary approach. Histological examination of the kidneys can help us choose the right strategy and aggressiveness of treatment.

Key words
rapidly-progressive glomerulonephritis, sarcoidosis

KAROLINA SCHNABEL

Heart transplantation in a patient with pathogenic sarcomere and α-Galactosidase A mutation

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INTRODUCTION Titin is a key component of the sarcomere. Mutations in the titin-encoding *TTN* gene are the most common genetic cause of dilated cardiomyopathy. Fabry disease is a rare X-linked lysosomal storage disease caused by mutations in the α-Galactosidase A (AGAL), that encodes the *GLA* gene. Reduced enzymatic activity of AGAL results in accumulation of glycosphingolipids in multiple cell types. Cardiac involvement may present with left ventricular hypertrophy.

CASE DESCRIPTION We report a case of an 18-year-old man who was referred to our center with Fabry disease. He presented with signs of heart failure at the age of 16 years. Cardiac magnetic resonance imaging showed severe dilated cardiomyopathy. Due to rapid progression of severe heart failure, the patient underwent successful heart transplantation. Genetic panel for dilated cardiomyopathy was performed. Mutations of *TTN* (c. 7399_7390) and *GLA* (c.773G>A) genes were identified. The AGAL activity was decreased (0.7 mmol/l/h) and lyso Gb3 levels were elevated (6.7 ng/ml). Histological analysis of the explanted heart did not show any signs of storage disease. Clinical signs of other possible manifestations of Fabry disease were absent. Segregation analysis showed the same *GLA* gene mutation in the proband's mother, who has been treated for dilated cardiomyopathy. The patient attends regular cardiology appointments and is asymptomatic.

DISCUSSION Genetic counseling categorized both the *TTN* and *GLA* gene mutations as pathogenic. The *GLA* variant is described in the literature as a "late-onset" variant. In a clinical geneticist's opinion, both mutations may have contributed to the clinical picture, therefore the cause of the severe heart failure may have been the *TTN* mutation, but we should look out for later onset cardiac involvement of Fabry disease as well. Taking all the information into the account, we diagnosed the patient with Fabry disease and we conduct yearly follow-up appointments in our center. We have not found any other reported cases of a combination of *TTN* and *GLA* gene mutations in the literature.

Key words
dilated cardiomyopathy, Fabry disease, heart transplantation, lysosomal storage disease, *TTN* gene

KATEŘINA KOUDELKOVÁ

Unravelling a rare cause of osteomalacia: a clinical encounter with phosphaturic mesenchymal tumor

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INTRODUCTION Osteomalacia, often associated with lifestyle factors and vitamin D deficiency, can also be caused by some rare conditions, such as phosphaturic mesenchymal tumor. This case study highlights the importance of a broad differential diagnosis of osteomalacia and emphasizes the role of a multidisciplinary team in its management. By exploring this rare cause of osteomalacia, we aim to improve clinicians' understanding and management of similar cases, ultimately improving patient outcomes.

CASE DESCRIPTION A 75-year-old patient presented with severe pain in the long bones and ribs. Computed tomography and magnetic resonance imaging showed multiple lesions and fractures. The patient's medical history included type 2 diabetes, hyperuricemia, tracheoesophageal puncture of the left knee, and *Borrelia burgdorferi* infection. Previous hematological investigations, including trepanobiopsy and bone biopsy, showed no evidence of a malignant process.

Skeletal scintigraphy showed evidence of osteoporosis and lower bone remodeling. Laboratory results showed hypophosphatemia (0.4 mmol/l), normal vitamin D levels and low serum parathyroid hormone. The fibroblast growth factor-23 (FGF23) levels were significantly above the laboratory norm of 85 (320 pg/ml). A positron emission tomography / computed tomography scan showed a tumor in the right maxillary region and 2 other intracranial lesions. Based on these findings, a preliminary diagnosis of a mesenchymal phosphaturic tumor was made.

The multidisciplinary team opted for primary extirpation of the maxillary tumor. Partial right maxillectomy was performed. One of the postoperative complications was a development of oroantral communication, which required a percutaneous endoscopic gastrostomy to be placed for oral healing. The patient's control FGF23 level decreased to 23.8 pg/ml and phosphate levels normalized. Muscle tissue samples from the maxillary region taken at the time of surgery confirmed the diagnosis of a phosphaturic mesenchymal tumor. However, the patient was readmitted to hospital due to bleeding from the percutaneous endoscopic gastrostomy. Gastroscopy showed grade 3 esophagitis and circular erosion in the terminal esophagus with confluent bleeding (Forrest Ib), which was managed by the gastroesophageal team. On ENT control, oroantral communication persisted. The patient was given an obturator and is attending a nutrition clinic to ensure adequate nutrition.

DISCUSSION This case represents a rare cause of osteomalacia induced by a mesenchymal tumor. Despite the patient's advanced age and comorbidities, the severity of osteomalacia and uncorrectable hypophosphatemia led the experts to perform a therapeutic trial and remove the primary tumor. This resulted in the normalization of bone turnover parameters. However, it also resulted in complications, requiring nutritional support, and resolution of the resulting oroantral communication.

CONCLUSIONS This case demonstrates the need for a multidisciplinary approach to the management of complex cases. Despite the potential complications that arose during the treatment, the efficacy of the procedure prolonged the patient's life. It highlights the importance of careful follow-up to ensure the best possible outcome for the patient.

Key words
DOTA-based imaging, fibroblast growth factor, fractures, osteomalacia, phosphaturic mesenchymal tumors

KRYSTIAN MRÓZ

Primary brain lymphoma masked as periprocedural delirium in acute coronary syndrome: a case of a 66-year-old man

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INTRODUCTION Delirium is an acute fluctuation in cognitive functions, affecting up to 50% of patients undergoing invasive procedures. Sometimes postoperative delirium may be associated with organic causes. Central nervous system (CNS) tumors are relatively rare. Although their symptoms involve neurologic impairment related to their location, sometimes psychiatric disorders may be their initial and sole presentation.

CASE DESCRIPTION A 66-year-old Caucasian man with a history of ST-segment elevation myocardial infarction treated with primary percutaneous coronary intervention was admitted to the interventional cardiology unit due to a suspicion of acute coronary syndrome (ACS). On admission, the patient was in an overall moderate condition. He was fully alert and well-oriented. He reported chest pain with very little physical exertion and short episodes of resting angina. Laboratory tests showed normal levels of high-sensitivity cardiac troponin and mild anemia. Unstable angina was diagnosed and the patient underwent coronary angiography, which showed multivessel coronary artery disease. A decision to refer the patient for coronary artery bypass grafting was made. The following day, the patient presented with delirium, short-term memory impairment, recurrent disorientation, and emotional lability, with no neurological deficit symptoms. Computed tomography (CT) was indicative of a hemorrhagic transformation of an ischemic stroke in the left hemisphere. Repeat CT scan showed

no evolution of the previously visible abnormalities and the image was reassessed as suspicious of a cerebral lymphoma. Magnetic resonance imaging of the brain proved a working diagnosis of primary CNS lymphoma. Additionally, a gradual decrease in hemoglobin value was observed with no apparent source of bleeding. The patient was qualified for further optimal medical therapy of coronary artery disease. Brain biopsy was performed. Histopathology confirmed the diagnosis of diffuse large B-cell lymphoma. Due to the comorbidities, the patient was disqualified from chemoimmunotherapy and underwent radiotherapy of the brain. The patient remained under home hospice care and died a year later.

DISCUSSION In the presented case, a patient with ACS and several comorbidities developed delirium after coronary angiography. Delirium is a common disorder complicating ACS and coronary interventions, which in rare cases may be mimicked by organic brain diseases. Up to 78% of patients with CNS tumors present with psychiatric symptoms, but none of them are specific to a certain brain tumor. In the presented case, additional symptoms (beyond disorientation and aggressive behavior) arose a suspicion of periprocedural stroke and led to additional imaging. Another atypical abnormality was anemia with no apparent cause of bleeding, which was related to the lymphoma. Due to the comorbidities, the patient was disqualified from the standard treatment. Brain radiotherapy was ineffective and the patient died within a year of the initial diagnosis.

CONCLUSIONS Delirium and other psychiatric symptoms are relatively common findings among hospitalized patients. In some cases, they may be the initial symptoms of an underlying CNS pathology. Their atypical presentation and co-occurrence with neurological disorders should be of particular concern. In such situations, it is advisable to extend the diagnostics to perform brain imaging.

Key words
acute coronary syndrome primary, delirium, central nervous system lymphoma

LASZLO HANKO

Esophagus mass and acute myeloid leukemia

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CASE DESCRIPTION Acute myeloid leukemia (AML) is an uncontrolled proliferation of immature progenitor stem cells in the medulla and/or outside of it. The prevalence of extramedullary infiltration (EMI) can reach 19%. Extramedullar manifestation is called myeloid sarcoma (MS) and it can be located at several anatomic sites. Most commonly, it occurs in the skin, central nervous system (CNS), and in the lymph nodes. MS can occur in AML at the time of diagnosis, during relapse or even primely. In the last case, transformation to AML is frequent. In the gastrointestinal tract, MS usually occurs in the stomach, ileum, and proximal colon (10%–11%), in the form of nodules, diffuse infiltrates or polyp formation. Clinical signs include abdominal pain caused by intussusception, total/subtotal ileus, and bleeding or, in some cases, perforation. Esophageal MS is rare. Only a few case studies can be found in the literature. We present a case of a 77-year-old man. He presented in our institute with odynophagia and weight loss (3 kg). His total blood count showed elevated white blood cell count (WBC, 44.1 g/l), slight anemia (hemoglobin, 94 g/l), and thrombocytopenia (70 g/l). Acute leukemia was suspected. Barium swallow X-ray examination and computed tomography (CT) scan of the chest showed a mass (2 cm long) in the proximal part of the esophagus. Further manifestations in other localizations were not described. Gastroscopy showed prominent, edged, protrusion-like mass in the proximal third of the esophagus, which resembled a malignancy. However, histology did not confirm our diagnosis. It only showed inflammatory cells and regenerative tissue. Bone marrow biopsy verified acute myelomonocytic leukemia. Given the patient's old age, palliative cytoreductive therapy was introduced (venetoclax, 100 mg/day). Due to the negative histology, second gastroscopy was performed. It showed an utterly different picture. The abnormality grew in size, but the picture was still characteristic for a malignancy. This time, with a correct histologic diagnosis of AML and confirmed of the CD117 positivity, we were able to diagnose MS. Unfortunately, the disease progressed quickly and we lost our patient due to septic complications. In conclusion, MS in AML is a relatively common complication. On the other hand, esophageal location is rare. MS is a bad prognostic factor, but it reacts to adequate therapy and the size of the lesions will decrease rapidly, like in our case.

Key words
acute myeloid leukemia, esophagus, myeloid sarcoma

LAURA KOROSKENYI

Vasculitis mimicking dysthyroid optic neuropathy

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INTRODUCTION Thyroid eye disease (TED) is an autoimmune condition and an extrathyroidal complication of Graves disease. Its severe form, called dysthyroid optic neuropathy, can lead to vision loss through compression of the optic nerve. Wegener granulomatosis is a necrotizing inflammation involving small and medium-sized vessels, characterized by granuloma formation. It often causes purpura, skin ulcers, and upper respiratory symptoms. Based on literary data, orbital involvement ranges between 15%–60%.

CASE DESCRIPTION A 27-year-old man had no significant previous medical history. His symptoms began in November 2021 with hearing loss, leading to repeated examinations at otorhinolaryngology clinics. Chronic sinusitis and otitis were diagnosed, and a hearing aid was suggested due to hypoacusis. The patient underwent functional endoscopic sinus surgery and bilateral paracentesis. In August 2022, neurological examination showed double vision, vertigo, and left-sided oculomotor paresis. Lumbar puncture ruled out neuroinfection. Six months later, rapid loss of vision prompted emergency hospitalization. On ophthalmic examination, bilateral exophthalmos was found. Best-corrected visual acuity were light perception with projection on the right eye and 20/25 (Snellen equivalent) on the left eye. Based on these findings, sight-threatening TED with optic nerve compression was suspected, and the patient was referred to the endocrine department. Urgent orbital computed tomography performed on the day of admission supported this diagnosis. Despite significant weight loss, laboratory tests confirmed TSH receptor antibody positivity, and normal TSH and thyroid hormone levels. Following the treatment protocol for dysthyroid optic neuropathy, high-dose parenteral corticosteroid treatment was initiated, leading to significant improvement in ophthalmic status. Meanwhile, orbital magnetic resonance imaging (MRI) failed to show characteristic features of TED and raised a possibility of lymphoma or other systemic disease. Biopsy performed from the right retrobulbar region was suggestive of vasculitis. Antineutrophilic cytoplasmic antibody positivity was confirmed. Investigations showed involvement in the dura, orbit, paranasal sinuses, middle ear, and central nervous system, consistent with the systemic type of Wegener granulomatosis. The patient underwent rituximab induction therapy, resulting in further improvement in both his vision and hearing.

CONCLUSIONS Orbital signs of systemic diseases may resemble the sight-threatening form of TED. In equivocal cases, MRI of the orbits should be among the early imaging modalities. The involve-

ment of the different disciplines is essential to establish an accurate diagnosis and targeted therapy.

Key words
thyroid eye disease, vasculitis

LILI VARGA

The very last diagnosis

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INTRODUCTION This case presentation aims to raise awareness of rare diseases causing abdominal pain.

CASE DESCRIPTION We present a case of a 27-year-old woman. Her medical history included an appendectomy. In the spring of 2023, she was admitted to the hospital with abdominal pain, wherefor cholecystectomy was performed. After being discharged from the hospital, she presented to the neurologic department with tetraparesis and was diagnosed with Guillain–Barré syndrome. Her improved condition enabled rehabilitation, during which she again complained of abdominal pain. Despite further diagnostic steps (abdominal ultrasound, laboratory tests, urinalysis, surgical, and urological consultation) and symptomatic treatment, the patient's condition progressed. Abdominal pain became uncontrollable and psychotic symptoms appeared, therefore the patient was transferred from the department of rehabilitation to our department of internal medicine.

Worsening pain, unusual course, and the development of a new onset of neuropsychiatric symptoms, despite previous therapy, guided us towards an unusual diagnosis. Finally, during a morning visit, we noticed a discoloration of the urine in the sunlight, which raised the possibility of an acute porphyria attack as a definitive diagnosis.

The therapeutic patient management was immediately revised. With adequate therapy, the patient's symptoms rapidly disappeared, although her general condition was still worrying. After contacting the National Porphyria Center, the required samples were taken, which proved the diagnosis. The patient was transferred to the Porphyria Center for further specialized treatment.

CONCLUSIONS Based on our case, it can be concluded that rare diseases should also be considered in the differential diagnosis of abdominal pathologies, especially if there is no definitive diagnosis that corresponds to the symptoms and results in clinical improvement. Excellent cooperation between various healthcare providers (laboratory, patient transport service, ambulance service, center hospital) is also necessary for the patient to receive appropriate special treatment in time.

Key words
abdominal pain, porphyria, rare diseases, urine

MAGDALENA PŁONKA-STĘPIEŃ

"I suffer from several serious diseases". A case report of the Dunnigan type of familial partial lipodystrophy: a multiple-symptom disorder caused by genetic mutation

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INTRODUCTION Monogenic diabetes represents a wide range of clinical phenotypes. This group includes diabetes connected with lipodystrophic syndromes, which are characterized by either complete or partial lack of adipose tissue (lipoatrophy). There is also an apparent accumulation of fat in other regions of the body (lipodystrophy). The loss of fat is associated with the severity of metabolic abnormalities, such as insulin resistance, hyperlipidemia or fatty liver disease.

CASE DESCRIPTION A 32-year-old woman was admitted to the outpatient metabolic clinic at the University Hospital. Her major concerns were 2 not-fully explained episodes of acute pancreatitis. The most likely cause of acute pancreatitis was hypertriglyceridemia. The woman was also diagnosed with type 2 diabetes. Her last therapy included oral hypoglycemic drugs, which did not provide satisfactory glycemic control. At history-taking, the patient also stated that her muscular physique had attracted the attention of other people since she was a child. On physical examination, fat accumulation around the face, neck, and shoulders, accompanied by thin limbs with clearly visible muscles due to atrophy of subcutaneous adipose tissue were described. Additionally, loss of fat tissue in the region of the buttocks, acanthosis nigricans in the cervical region, and hirsutism were noticed. The patient informed us that her 9-year-old daughter had a very similar body composition, but no documented metabolic abnormalities. Her father was diagnosed with type 2 diabetes and his body composition also seemed to show some similarities to the proband. Based on medical history that included hypertriglyceridemia, pancreatitis, diabetes, abnormal body composition of the proband, and possible hereditary nature of the disease, familial partial lipodystrophy was hypothesized. Genetic testing was performed. The result of next generation sequencing showed pathogenic variant in *LMNA* gene, reported in earlier publications as a cause of type 2 familial partial lipodystrophy (FPLD 2), known as Dunnigan lipodystrophy. Currently, the daughter of the patient has been referred for genetic testing. The 32-year-old proband is planned to be included in the clinical trial with metreleptin, a synthetic analogue of human leptin.

DISCUSSION Our patient presented with body composition typical for FPLD 2, which, with other clinical features, was an argument for genetic testing. The transmission of this syndrome is autosomal dominant. Risk of disease occurrence in a first-degree relative is 50%, therefore genetic testing of family members is indicated. As a woman of reproductive age, our patient could make a conscious decision regarding other children.

The diagnosis of genetically confirmed Dunnigan lipodystrophy allows for a consideration of new treatment options. Metreleptin is an analog of human leptin, which improves insulin sensitivity and decreases both hepatic glucose output contributing to hyperglycemia and hepatic steatosis. Our patient expressed willingness to participate in a clinical trial with metreleptin.

LESSONS TO BE LEARNED In diseases frequently involving genetic testing, careful medical history and physical examinations are a basis for further proper diagnostics. Genetic testing in monogenic forms of metabolic diseases can be applied in clinical practice to establish diagnosis, define prognosis in family members, and propose treatment options.

Key words
Dunnigan, lipodystrophy, lipodystrophy, *LMNA* gene, metreleptin

MALEK OTHMAN

Takotsubo cardiomyopathy complicating diabetic ketoacidosis. A challenging diagnosis and complicated course: a case report with literature review

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INTRODUCTION Takotsubo cardiomyopathy (TCM) is a transient wall motion abnormality of the left ventricle with apical balloon-like dilation, typically associated with emotional or physical stress. While the pathophysiology of TCM remains unknown, various hypotheses have been discussed, including myocardial ischemia, left ventricular outlet tract obstruction, and neurohormonal release of catecholamines. Diabetic ketoacidosis (DKA)-associated TCM is rarely reported in the literature, with atypical and wide range of presentations. With the high prevalence of diabetes in the Middle East, it is vital to examine the pathophysiology and symptomatology of TCM, as it represents a rare, but potentially fatal, complication of DKA.
CASE DESCRIPTION A 73-year-old bedridden Middle Eastern woman was brought to the emergency department with a 3-day history of progressive fatigue, poor appetite, and nausea. She developed drowsiness on the day she presented. Past medical history was notable for hypertension hypercholesterolemia and poorly controlled type 2 diabetes mellitus. The patient was hypothermic (34.7 °C) but otherwise vitally stable. On initial evaluation, she was comatose with a score of 6/15 on Glasgow Coma Scale. Pupils were equal and reactive to light. Laboratory investigations identified severe DKA (random glucose levels, 450 mg/dl; serum bicarbonate levels, 2 mmol/l; pH, 6) and severe hypernatremia (170 mmol/l). She was started on IV fluid, insulin infusion, and antibiotics, followed by subcutaneous insulin and free water flushes though nasogastric tube. Incidentally, the patient was found to have ST elevations of the inferolateral leads (troponin I, 10.61 ng/ml). The patient was therefore taken to the cath lab for coronary angiography, which showed no significant obstructive coronary artery disease. Echocardiogram showed features suggestive of TCM (ejection fraction, 35%–40%). The patient was then started on Aspirin, β-blockers, and high-dose statins. Her troponin levels started to trend downward. Following her hospital stay, prolonged due to nosocomial infections (eg, aspiration pneumonia and wound infection), the patient was discharged after 37 days, with optimized home medications.
DISCUSSION DKA-associated TCM was examined in 15 published cases. Average age was 53.5 years with male percentage of 18.75%. Symptomatology was divided almost equally into DKA symptoms, TCM symptoms, and mixed symptoms groups. Our patient was treated for TCM with aspirin, β-blockers, and high-dose statins, with no long-term TCM-specific treatment. Treatment approaches to DKA-associated TCM in the literature included: supportive treatment, antiplatelets, anticoagulants, diuretics, nitrates, β-blockers, and inotropes. Almost all of the reviewed cases had good outcomes, except for one case in which acute respiratory distress syndrome developed, resulting in death. Several mechanisms were suggested in the literature, including catecholamine surge compounded with severe acidosis, which can lead to myocardial calcium channels dysfunction. This may result in myocardial stunning and, later, TCM.
CONCLUSIONS DKA-associated TCM can be challenging to diagnose with atypical presentations and possible complicated courses. In the literature, the association between TCM and DKA has been rarely reported. This study highlights the need for continuous monitoring and early recognition of TCM, especially in patients undergoing physical and psychological stress, such as DKA, since it can lead to significant morbidity and mortality.

Key words
diabetic ketoacidosis, stress cardiomyopathy, Takotsubo cardiomyopathy

MANUELA DELGADO

Massive digestive hemorrhage as a manifestation of cytomegalovirus colitis in an immunocompromised patient

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INTRODUCTION Cytomegalovirus (CMV) is a Betaherpesvirinae belonging to the Herpesviridae family, that can cause primary, latent, chronic, and persistent infections. In immunocompetent individuals, CMV infection is usually asymptomatic or presents mildly. In immunocompromised individuals, it behaves as an opportunistic pathogen, capable of causing severe disease and even death. In the case of patients with HIV, it is more common in individuals with CD4 counts less than 100 cells/μl. Digestive involvement is the second most frequent after retinitis, with colitis being the most described form, and massive digestive hemorrhage being a severe variant of presentation. For the definitive diagnosis of CMV gastrointestinal disease in acquired immunodeficiency syndrome (AIDS) patients, histological study is necessary, where pathognomonic "owl's eye" cytomegalic inclusions are observed. A patient's clinical presentation and virus demonstration through culture, polymerase chain reaction (PCR), and antigenemia support the diagnosis. The treatment consists of endoscopic and surgical resolution associated with the use of antiviral drugs and highly active antiretroviral therapy (HAART).
CASE REPORT A 47-year-old woman with unknown medical history was admitted to our hospital with fever and proctorrhagia. Diagnosis of severe anemia (hemoglobin, 6.1 g/dl; hematocrit, 19%) and HIV infection was made, with a CD4 count of 24 cells/μl and a viral load of 586 000 copies. Computed tomography of the chest showed diffuse miliary interstitial infiltrate, "tree-in-bud" images, and focal consolidation with air bronchogram in the middle lobe. Sputum for acid-fast bacilli (AFB) was requested and ruling out tuberculosis, it was interpreted as *Pneumocystis* pneumonia in an immunocompromised patient, initiating empirical antibiotic treatment. The patient experienced massive lower digestive track hemorrhage and hypovolemic shock. Urgent upper and lower gastrointestinal endoscopy was performed, both reporting erosive gastroduodenitis and active bleeding with inability to detect its origin. Intraoperatively, blood was found in the sigmoid colon and the last 50 cm of the small intestine through transillumination. Due to suspicion of CMV colitis, PCR was requested and new videocolonoscopy was performed, showing an ulcerated lesion in the rectum, from which a biopsy was taken. Empirical treatment with intravenous ganciclovir was initiated. Subsequently, positive CMV PCR and positive pathological anatomy of the rectal ulcer biopsy for the same virus were received. Additionally, fundoscopic exam showed retinitis in the right eye, compatible with CMV. The patient became hemodynamically stable and afebrile, so she was discharged from the hospital.
DISCUSSION Cases of AIDS patients with CMV colitis described in the literature have fatal outcomes due to complications of this disease. It was reported, that the median survival time after the diagnosis of CMV colitis in HIV patients was 4 months, even with ganciclovir treatment.
CONCLUSIONS The importance of this case is highlighted by its good evolution, as the patient improved significantly once antiviral therapy directed at CMV and HAART was initiated. It is a good outcome, considering that in the vast majority of reported cases the patient dies due to either complications or a lack of treatment.

Key words
cytomegalovirus, HIV, massive digestive hemorrhage

MARCELA MARTÍNEZ

Deafening pneumonia as an unusual presentation of *Legionella* infection

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CASE DESCRIPTION A 50-year-old man with a history of active smoking and obesity presented for a consult due to sudden bilateral hearing loss without other associated symptoms. Examination was

performed in the emergency department, showing the following findings: C-reactive protein (CRP) levels of 662 mg/dl, white blood cell count of 15.68 × 10^3/mm^3, sodium levels of 127 mmol/l, and lactate dehydrogenase levels of 639 IU/l. Angiography of the brain and neck vessels showed no abnormalities. Computed tomography of the chest without contrast showed extensive multifocal pneumonia. In the emergency department, the patient experienced episodes of desaturation which required support with supplemental oxygen through the nose, and was transferred to the critical treatment unit of our campus, where empiric antibiotic treatment with ampicillin/sulbactam was initiated and microbiological study was ordered with human immunodeficiency virus testing (nonreactive). From the respiratory point of view, the presentation began with a progressive increase in oxygen requirements; therefore, noninvasive mechanical ventilation was initiated. The patient did not show any major improvement, so advanced airway management was performed with orotracheal intubation. Microbiological study results showed urinary antigenemia for *Legionella pneumophila* positivity. The antibiotic treatment was switched to azithromycin. Due to the hearing loss associated with Legionella pneumonia, prednisone was added at a dose of 1 mg/kg/day for 10 days. The patient progressed favorably, and as the disease resolved, he was discharged in good general condition.

DISCUSSION *L. pneumophila* is a strict anaerobic gram-negative bacillus, and it is transmitted through fresh water (eg, lakes, rivers), through which it can access various types of equipment running on hot water. It can infect humans via inhalation of microaerosols. Typical clinical presentation of *L. pneumophila* infection is pneumonia, known as Legionnaires disease, which is characterized by fever, cough, dyspnea, fatigue, and myalgia. In addition, cases involving altered mental status (up to 25% of cases) and diarrhea (19% of cases) are reported. The associated laboratory alteration include hyponatremia, hypophosphatemia, and significant elevation of CRP levels. The mortality of patients who require admission to the intensive care unit due to *L. pneumophila* infection varies from 9.1% to 41.4%. Factors that influence susceptibility to infection include immunosuppression, chronic obstructive pulmonary disease, smoking, and age over 50 years. To date, only 2 cases of *Legionella* pneumonia involving sudden and severe bilateral hearing loss have been described. The pathogenesis is unclear, but an exotoxin-mediated or immuno-mediated effect is hypothesized, as the bacteria rarely invade the central nervous system. However, they are capable of producing endotoxin-like substances. There are limited data on the treatment of *L. pneumophila* infection in the case of sudden deafness. The use of prednisone at a dose of 1 mg/kg/day for 10 days is endorsed. Suggested treatment regimens for the Legionella infection itself are based on quinolones or macrolides.

CONCLUSIONS *L. pneumophila* infection requires a high index of suspicion in patients with risk factors and pneumonia with atypical features. There are very few case reports of the infection being accompanied by hearing loss; however, the use of systemic corticosteroids seems to be effective.

Key words
corticosteroids, deafness, *Legionella*, pneumonia

MARIA IGNACIA ALVAREZ ARGALUZA

When fungus takes the blame: disseminated histoplasmosis as a cause of hemophagocytic syndrome

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CASE DESCRIPTION Hemophagocytic lymphohistiocytosis (HLH) is a syndrome characterized by aggressive immune dysregulation, often life-threatening due to an uncontrolled hyperinflammatory response. It can be inherited or secondary to various causes, including infections, autoimmune disorders, immunosuppression, and malignancy. In cases of human immunodeficiency virus (HIV) infection , patients with advanced immunosuppression associated with opportunistic infections are at increased risk of developing HLH. Histoplasmosis, an endemic fungal disease, presents with diverse clinical manifestations, typically resolving in immunocompetent individuals but posing significant risks, including secondary HLH, in immunocompromised patients, particularly those with acquired immunodeficiency syndrome . Here, we report the case of a 36-year-old Venezuelan man with HIV with severe immunosuppression, who presented with fever of unknown origin. Laboratory and imaging studies were compatible with HLH, and further studies confirmed disseminated histoplasmosis as the trigger of HLH. Antifungal therapy with amphotericin was initiated, followed by transition to itraconazole. The patient also began antiretroviral therapy for HIV. Prompt diagnosis and management were crucial in this case, emphasizing the critical role of early recognition and treatment initiation in improving outcomes. This case report highlights the importance of considering histoplasmosis as a cause of HLH in HIV patients, particularly in highly endemic regions, to facilitate early initiation of treatment when there is a high clinical suspicion.

Key words
hemophagocytic lymphohistiocytosis, histoplasmosis, HIV

MARIA KOMISARZ-CALIK

How to proceed in giant prolactinoma? Long-term follow-up of giant prolactinoma partially refractory to dopamine agonists in young men

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INTRODUCTION Giant prolactinomas (GP) are a rare subtype of a lactotroph pituitary neuroendocrine tumor, which constitutes about 3% of prolactinomas, measuring over 40 mm and occurring predominantly in young men. The management of GP is a diagnostic and therapeutic challenge for clinicians, while literature data regarding this condition are scarce.

CASE DESCRIPTION We present the case of a young 23-year-old man referred to the department of endocrinology with suspicion of macroprolactinoma. He reported acute headaches and visual field deficits lasting for 6 months, and symptoms of hypogonadism that were present since puberty. Magnetic resonance imaging (MRI; 2018) showed a polycystic mass (52 × mm 52 × mm 41 mm) with a suprasellar extension and infiltration of the optic chiasm. Investigation showed very high serum concentration of prolactin (PRL; 21525 µIU/ml (reference range, 86–342 µIU/ml) with suppressed luteinizing hormone and follicle-stimulating hormone, and low levels of testosterone. Serum concentration of free T4 and thyroid stimulating hormone were indicative of central hypothyroidism. The tests did not show insufficiency of the adrenal axis. Additionally, dual-energy X-ray absorptiometry (DXA) showed decreased bone density. The ophthalmologic examination showed a loss of visual field in the upper temporal quadrant. The patient was presented during Pituitary Tumor Board, but there were no indications for surgery. Pharmacological treatment with a dopamine agonist was continued with bromocriptine, after which a decrease of PRL (63%) and slight improvement in headaches were noted. Levothyroxine in a dose of 75 µg daily, testosterone injection (once for 2 weeks),

and cholecalciferol in a dose of 4000 IU daily was prescribed. Follow-up MRI (November 2018) showed the shrinkage of the tumor (40 mm × 40 mm × 35 mm). Bromocriptine was replaced with cabergoline at a maximal dose of 4 mg/week with a spectacular improvement in headaches. On follow-up MRI (May 2019), further tumor shrinkage was observed (32 mm × 25 mm × 31 mm). PRL concentrations decreased to 7519 µIU/ml (35% of the baseline concentration). Genetic test for *AIP* and *MEN1* gene mutations was performed with a negative result. Due to cabergoline therapy at a dose higher than 2 mg, echocardiography was performed and no abnormalities of heart valves were found. The tumor's dimensions have been stable since December 2019 (25 mm × 13 mm × 23 mm), with an increase of the cystic vs solid structure of the tumor. PRL concentrations remain increased with average levels of 4500 µIU/ml. The cabergoline dose was decreased to 1.5 mg/week.

DISCUSSION Despite their usually benign character, GPs can extend the suprasellar region and involve the optic chiasm or cavernous sinuses. They are associated with high PRL concentrations and often respond to dopamine agonists (cabergoline). In cases of pituitary apoplexy, cerebrospinal fluid rhinorrhea, and progressive mass growth (despite optimal treatment), surgical treatment is indicated. However, the surgical procedure is associated with significant morbidity. In aggressive prolactinomas with persistent growth, temozolomide or pasireotide could be a part of the treatment. GPs often require a multimodal approach and regular follow-up visits, including echocardiography and DXA.

CONCLUSIONS A conservative pharmacological approach to GP could be a choice of treatment, even in case of partial responsiveness to cabergoline. Partial decrease of serum prolactin, tumor shrinkage and headache remission are considered as a successful treatment.

Key words
dopamine agonists, giant prolactinoma, prolactin, prolactinoma

MARTA OREL

Ectopic thyroid

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INTRODUCTION Thyroid gland is a small, butterfly-shaped endocrine gland that consists of 2 lobes, connected by the thin isthmus. It is normally located in the anterior neck, posterior to the sternothyroid and sternohyoid muscles, wrapping around the cricoid cartilage and tracheal rings, which typically correspond to the vertebral levels C5–T1. Ectopic thyroid can be described as functional thyroid tissue that is located anywhere other than its anatomic position. Its prevalence is about 1 in 100 000–300 000 population and increases to 1 in 4000–8000 patients with thyroid pathology.

CASE DESCRIPTION A 16-year-old woman applied to the facial surgery department for medical treatment, with complaint on painless swelling of the submandibular area. The patient's past medical history showed thyroid stimulating hormone (TSH) levels of 3.36 mIU/l (normal values, 0.27–4.2 mIU/l) and free T4 (fT4) levels of 1.34 ng/dl (normal values, 1.1–1.7 ng/dl). Ultrasound examination showed thyroid gland hypoplasia. The swelling occurred long time prior to the patient's visit and slightly increased in size. Objective examination confirmed the presence of a mobile, painless circumscribed mass of a dense-elastic consistency (1.5 cm in diameter), located in the submandibular area, which raised above the skin. The skin above it was pale pink and not changed. Results of the histological examination showed the tissue of ectopic thyroid gland with areas of transformation into colloid nodular goiter. The diagnosis made was colloid nodular goiter. Excisional biopsy was performed. There were no complications in the postoperative period. Control supervision of a surgeon and endocrinologist in 1 and 6 months were recommended. However, 6 months after the surgery, the patient developed hypothyroidism with laboratory tests results of TSH levels of 20.88 mIU/ml and fT4 levels of 1.02 ng/dl.

DISCUSSION The thyroid gland is the first endocrine gland that develops during fetal embryology. Ectopic thyroid tissue occur due to an aberrant development of the thyroid gland during its migration to the pretracheal region and is typically located along the path of the thyroid gland's descent, at any position from the foramen cecum at the base of the tongue to the mediastinum. Approximately 90% of ectopic thyroid cases are lingual thyroid. Both men and women are equally affected. Ectopic thyroid can present at any age, with most cases identified through newborn screening, but some cases are delayed up to the adolescence, when they present with symptomatic manifestations. The studies suggest that hypothyroidism occurs in about 33% of such individuals, thus, all the patients suspected for thyroid ectopy should undergo a biochemical thyroid profile. Although malignant transformation is relatively uncommon (1%), it can appear with different histotypes: papillary, follicular, mixed follicular and papillary, Hurthle cell, and medullary. High-resolution ultrasound scanning can help as an initial assessment; magnetic resonance imaging, computed tomography, positron emission tomography–computed tomography, and chest X-ray can also be used. If the case is highly suspicious for malignancy, tissue biopsy for histology or fine needle aspiration cytology should be performed.

CONCLUSIONS The doctors, especially pediatricians, family doctors, endocrinologists, and surgeons, should be aware about such medical problem and consider it in differential diagnosis of the tumors of that area, as often it is initially considered as malignancy. Lifelong thyroxin replacement therapy and regular follow-up are an optimal choice for such patients.

Key words
ectopic thyroid, hypothyroidism, levothyroxine, tumors

MÁRTON KALABAY

A rare form of hypothalamic diabetes mellitus and hyperlipidemia after surgery for craniopharyngioma

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INTRODUCTION Hypothalamic diabetes mellitus is a rare form of diabetes that develops when the delicate balance mechanisms in the hypothalamus that regulate food intake are compromised. Among the signaling pathways in the nucleus arcuatus, nucleus paraventricularis, and nucleus ventromedialis of the hypothalamus, pro-opio-melanocortin (POMC) neurons, which are the only neurons that inhibit food uptake, and Agouti-related peptide (AgRP) and neuropeptide Y (NPY) neurons, which stimulate food uptake and increase blood glucose levels, play a prominent role. In addition to regulating sugar metabolism, the hypothalamus is also involved in fat metabolism, as AgRP and NPY neurons are able to mobilize lipids from the adipose tissue in the periphery through activation of the sympathetic nervous system.

CASE DESCRIPTION A 33-year-old man was admitted to our clinic 3 times between August 2022 and February 2023. In July 2022, he underwent a resection of craniopharyngioma with supra- and parasellar spread to the cerebral ventricle III. The patient was first admitted due to postoperative diabetes insipidus and panhypopituitarism, accompanied by short-term memory impairment. "Incidental" dyspnea due to bilateral massive pulmonary embolism was confirmed. He was discharged home after adjustment of anticoagulation and hormone substitution. His second admission was in November 2022 due to

a sudden onset of significant muscle weakness and pain, polyuria, hyperosmolar hyperglycemia (34 mmol/l; negative acetone), exsiccosis, and left hemiparesis. The most routine laboratory parameters of blood samples taken at admission were indeterminate, due to extreme lipemia (triglycerides, 55 mmol/l). In the presence of a negative cranial magnetic resonance imaging (MRI), neurological abnormalities could be caused partly by hyperosmolarity and partly by hyperviscosity syndrome associated with hyperlipidemia. The patient had gained 10 kg since surgery. In addition to fluid balance, low molecular weight heparin therapy, intravenous insulin therapy, and high-dose fibrate were started. Treatment resulted in a gradual decrease in triglyceride levels. Laboratory tests showed elevated cholesterol levels and combined hyperlipidemia, so when triglyceride levels allowed for it, the fibrate treatment was supplemented with statins. In addition, hemoglobin A_{1c} level was 12.2% with preserved C-peptide production. Insulin therapy was changed to a glucagon-like peptide 1 (GLP-1) receptor agonist, which brought the patient's blood glucose into the desired range. His third admission was for hyponatremia. His ion balance was settled by reducing the dose of desmopressin. The metabolic parameters were within normal range with the treatment regimen.

CONCLUSIONS Postoperative diabetes insipidus and panhypopituitarism are common complications after a craniopharyngioma surgery. Severe abnormalities of fat and glucose metabolism observed in our patient raised a suspicion of hypothalamic injury, which was further strengthened by the fact that since his surgery, the patient also suffered from short-term memory impairment. Available MRI images also showed a hypothalamic lesion. Further MRI examination was performed to determine if nuclear localization was ongoing. The recent onset of diabetes mellitus and combined hyperlipidemia might have been caused by POMC-AgRP imbalance, which may have led to metabolic derailment through hyperphagia and consequent obesity, increased hypothalamic-pituitary axis activity, and hypothalamic energy sensor dysregulation. Our case demonstrates that in partial hypothalamic injury, GLP-1 receptor agonists may provide effective glycemic control and the presence of hypothalamic GLP-1 receptors may contribute to this.

Key words
glucagon-like peptide 1, hyperlipidemia, hypothalamic diabetes mellitus

MAXIMO ROUSSEAU PORTALIS

Neurosarcoidosis as a rapidly progressive dementia associated with normal pressure hydrocephalus

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INTRODUCTION Neurosarcoidosis (NS) is a rare neurologic manifestation of sarcoidosis. It often poses a diagnostic and therapeutic challenge for physicians, as it has diverse clinical presentations and is associated with poor prognosis.

CASE DESCRIPTION We describe a case of a 53-year-old man who was diagnosed with lingual sarcoidosis 2 years earlier and developed ataxia, and rapidly progressive cognitive impairment. On examination, he was disoriented with marked gait imbalance. He had no fever and meningeal signs were absent. Lumbar puncture showed hypoglycorrhachia, hyperproteinorrachia, and lymphocytic pleocytosis. Blood and cerebrospinal fluid (CFS) cultures were negative. Brain magnetic resonance imaging showed hyperintense periventricular and subcortical lesions on T2-weighted fluid-attenuated inversion recovery sequence without contrast enhancement, resembling progressive multifocal leukoencephalopathy (PML), but polymerase chain reaction (PCR) negative for JC virus ruled out the diagnosis. CSF had elevated interleukin (IL)-6 levels (600 pg/ml) and flow cytometry showed elevated CD4+ lymphocyte concentration and a CD4+/CD8+ ratio of 3.91, indicative of NS. Following a favorable evolutionary course with corticosteroid pulses, the patient was discharged with a maintenance regimen of deltisone and azathioprine. However, the patient relapsed 8 months later with ataxia, ventriculomegaly, and lumbar puncture with low opening pressure and low inflammatory markers. As the initial approach with immunosuppressants was ineffective, the patient underwent ventriculoperitoneal shunting with a good response to date.

DISCUSSION NS often develops within 2 years of systemic sarcoidosis and although uncommon, rapidly progressive dementia may be its initial symptom. The clinical presentation and neuroimages of NS are often unspecific, but high levels of IL-6 and a CD4+/CD8+ ratio higher than 3.5 have been recently found to be useful for differential with demyelinating and other inflammatory diseases. In this case, bilateral leukoencephalopathy resembled PML, but PCR negative for JC virus and CSF biomarkers were essential for the diagnosis. Our patient relapsed months later with communicating hydrocephalus. This complication is unusual in NS, observed in roughly 1 in 10 affected individuals, with obstructive hydrocephalus being the most common presentation. When hydrocephalus is present, both immunosuppressive therapy and neurosurgical interventions are imperative.

LESSONS TO BE LEARNED This case underscores the importance of maintaining a high index of suspicion for NS in individuals with sarcoidosis and neurologic symptoms. In these cases, CSF biomarkers such as IL-6 and CD4+/CD8+ ratio are essential to arrive at the diagnosis. Furthermore, this case highlights that hydrocephalus is a rare complication and requires a multidisciplinary approach, including medical and neurosurgical treatment.

Key words
neurosarcoidosis, normal pressure hydrocephalus, progressive multifocal leukoencephalopathy

MICAELA GOMEZ GIGLIO

An uncommon case of nonsurgical acute abdomen

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INTRODUCTION Mesenteric panniculitis, characterized by inflammation of the mesentery, is a rare condition with elusive etiology. While its origins are unclear, it is commonly considered to be idiopathic or associated with such factors as abdominal surgeries, trauma, malignancies (including lymphoma, colorectal cancer, and lower urinary tract malignancies), and, less frequently, rheumatic conditions. Recent studies showed that almost 40% of the patients with mesenteric panniculitis had a history of metabolic syndrome and 22% had coronary artery disease.

CASE DESCRIPTION Here, we present the case of a 75-year-old woman who presented to the emergency department with 5 days of intense acute abdominal pain, severe distension, nocturnal fever, and anorexia. The patient had a medical history significant for prior abdominal surgeries, arthritis with sicca symptoms, cutaneous vasculitis, and Berger nephropathy. Physical examination and complementary studies showed severe abdominal distention and diffuse peritoneal reaction, along with elevated inflammatory markers and computed tomography scan demonstrating severe mesenteric panniculitis without other pathological findings. Malignancy and other rheumatoid entities were excluded, and medical treatment with steroids led to significant clinical and radiological improvement.

DISCUSSION Diagnosis of mesenteric panniculitis primarily relies on imaging, with inflammatory nodules found in approximately 70%

of cases, although diffuse inflammation of variable size may also occur. Given its potential association with malignancy in rare cases, it is advisable to consider positron emission tomography-computed tomography scanning or other relevant screening studies based on the patient's age and epidemiology. Additionally, there have been reported associations with rheumatic diseases in certain instances. Management of affected patients involves prioritizing pain relief with a steroid as a first drug of choice. When necessary, other immunosuppressive drugs can de added (eg, azathioprine). Recent studies showed clinical improvement with colchicine.

Key words
acute abdomen, mesenteric panniculitis, steroids

MONIKA BELIANČINOVÁ

Hereditary autoinflammatory diseases associated with AA amyloidosis

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INTRODUCTION Amyloidosis represents a spectrum of serious diseases characterized by the deposition of an excessive amount of incorrectly conformed proteins (amyloid structures) in extracellular spaces. Currently, the most widely used diagnostic method for diagnosing amyloidosis is biopsy of the damaged organ, light microscopy with the help of special Congo red staining, and mass spectrophotometry and proteomic analysis, which can be used to identify the type of amyloid.
CASE DESCRIPTION In the presented case report, we introduce a complicated differential diagnostic process in a patient treated at the transplantation-nephrology department at the Martin University Hospital. In the clinical findings, the image of a fully developed nephrotic syndrome with an estimated glomerular filtration rate (eGFR) of 93 ml/min/1.73 m^2 prevailed. As part of the differential diagnostic procedure, we performed kidney biopsy, which confirmed AA amyloidosis with early renal and vascular involvement. For the finding of nodal syndrome, we consulted a hematologist, who, even in the context of the biopsy result, indicated trepanobiopsy of the bone marrow with the finding of B-non-Hodgkin lymphoma (multiple myeloma type), treated with bortezomib. We also completed genetic examination of the exome to detect the presence of mutations in the genes responsible for the development of SAA amyloidosis. We confirmed the finding of a *MEFV* gene mutation in a heterozygous state, which is causal for the development of familial Mediterranean fever and a second VUS gene with an unclear clinical significance. Therefore, in cooperation with an immunologist we started an optimized treatment in the direction of familial Mediterranean fever (colchicine, canakinumab). The patient was stabilized, without the presence of nephrotic syndrome, with an eGFR of 85 ml/min/1.73 m^2.
CONCLUSIONS Serum amyloid A (SAA) consists of 104 amino acids. It is among the reactants of the acute phase of inflammation and is produced by the liver during inflammation and infection. At least 5 allelic variants that encode SAA have been identified and several hereditary autoinflammatory conditions closely related to the development of SAA amyloidosis have been described. SAA amyloidosis remains a life-threatening complication of hereditary autoinflammatory diseases; therefore, interdisciplinary cooperation is an integral part of its diagnosis.

Key words
amyloidosis, autoinflammatory diseases, familial Mediterranean fever, serum amyloid A

NADIA HUSSEY

Ocular flutter in an immunocompromised patient with anti-GQ1b antibody syndrome and pulmonary tuberculosis

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CASE DESCRIPTION This case report explores a complex presentation in a 24-year-old woman with HIV infection, anti-GQ1b antibody syndrome, and pulmonary tuberculosis (TB). The patient exhibited neurological symptoms, including ocular flutter, ataxia, and incontinence. Ocular flutter is an unusual eye manifestation of the anti-GQ1b antibody syndrome, which is not well described. The coexistence of HIV, TB, and Miller–Fisher syndrome variant poses challenges in diagnosis and treatment, specifically with timing of antiretroviral treatment. This report contributes to the understanding of anti-GQ1b antibody syndromes in the context of HIV infection, particularly in regions with limited data and high prevalence rates of HIV and TB coinfection, and highlights a need for further research.

Key words
anti-GQ1b antibody syndrome, HIV, Miller–Fisher, ocular flutter, syndrome variant, tuberculosis

NÓRA GARAM

The aspects of the diagnosis of testosterone-producing tumors

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INTRODUCTION Numerous endocrine diseases could cause elevated testosterone levels in women. During the differential diagnosis, polycystic ovarian syndrome, congenital adrenal hyperplasia, Cushing syndrome, exogenous testosterone intake or the testosterone-producing tumors of the ovary, or the adrenal gland have to be mentioned. Hormone levels check and various imaging methods are useful for the diagnosis.
CASE DESCRIPTION A 29-year-old woman was diagnosed with late-onset congenital adrenal hyperplasia. Checkup hormone test showed regular supplementation of glucocorticoid and elevated testosterone level. High testosterone levels present after the administration of dexamethasone therapy raised a possibility of autonomous androgen production. Computed tomography (CT) scan of the adrenal gland was negative. Transvaginal ultrasound of the ovaries detected amixed echogenic solid tumor (3 cm) of the left ovary. Pelvic magnetic resonance imaging (MRI) showed a 3.3 cm × 2.6 cm mass which corresponded to the left ovary. Left-sided adnexectomy was performed. Histology showed Leydig-cell tumor. A 65-year-old with hypothyroidism came to the clinic because of the presence of hirsutism. Testosterone levels were elevated and lutenizing hormone (LH) and follicle-stimulating hormone (FSH) levels were suppressed. CT did not show any pathology in the adrenal gland. Pelvic MRI showed 2 cysts in the right ovary, whereas the left ovary could not be located. After a long discussion, explorative laparoscopy was made and bilateral adnexectomy was completed. Based on the pathology report, a Leydig-cell tumor was found in the right ovary.

A 50-year-old woman was examined because of virilism. Her testosterone levels were 524 ng/dl and LH and FSH levels were suppressed. Based on the hormone levels, Cushing syndrome, polycystic ovarian syndrome, and congenital adrenal hyperplasia were not likely. CT scan of the adrenal gland raised the possibility

of right-sided adenoma of the adrenal gland. MRI did not indicate an obvious tumor in the ovary. Transvaginal ultrasound did not show any abnormalities. For accurate assessment, we performed selective vein canulation from both the ovarian and adrenal vein. Cannulation was made and the results showed us that the testosterone came from the right ovary. Adnexectomy was done and a Leydig-cell tumor was described in the right ovary.

A 50-year-old woman with hirsutism, androgenic alopecia, and central obesity was diagnosed with hyperandrogenism. Abdominal ultrasound and CT scan did not describe any pathologies. Transvaginal ultrasound could not visualize the left ovary. On MRI, a 6.3 cm × 4.8 cm × 6.1 cm heterogenic, contrast-enhancing tumor was seen. Bilateral adnexectomy was done. The histology report described a Sertoli-cell tumor.

DISCUSSION Clarifying the etiology of hyperandrogenic state is not easy. Hormone levels are crucial, but if there is a possibility of testosterone-producing tumor, imaging techniques are useful. Both testosterone-producing tumors are extremely rare (0.02%–0.1% of all ovarian tumors). The prognosis after surgical removal was good.

CONCLUSIONS According to our case report, there is no exact algorithm to make the diagnosis of testosterone-producing tumors. Close cooperation with radiologists and gynecologists is needed and each case should be evaluated individually.

Key words
endocrinology, hirsutism, hyperandrogenism, testosterone-producing tumor

OKSANA FAIURA

It's never too late to diagnose celiac disease

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INTRODUCTION Although celiac disease usually manifests in childhood, it can manifest itself at different ages. Its onset in adults can be accompanied by various atypical symptoms, becoming an increasingly common phenomenon every year.

CASE DESCRIPTION A 31-year-old biologist presented with complaints of abdominal pain (umbilical and left subcostal areas), abdomen enlargement, leg edema, nausea, diarrhea, bloating, fatigue, dry skin, and dizziness. Objective examination showed moderate general condition, pale, dry, and elastic skin, erythematous rash that did not rise above the skin on the face, pale, moist, mucous membranes, moist tongue coated with white layering, enlarged abdomen, dull percussion sounds, soft palpation that was painful in the periumbilical and left hypocostal areas, irregular defecation (brown, irregular, iquid stool), and leg swelling up to the level of the knees. The patient underwent laboratory and instrumental examination. Complete blood count showed hemoglobin levels of 102.0 g/l, eosinophils at 6, and hypochromia. Biochemistry results showed Fe2+ of 5.1 (normal range [N], 10.7–21.5 mmol/l), antistreptolysine "O" of 500 U (N, 250 U), rheumatoid factor of 13.07 (N, negative), total protein of 68.3 g/l (N, 65–85 g/l), albumins of 49% (N, 52%–65%), α1-globulins of 5.2% (N, 2.5%–5%), α2-globulins of 10.8% (N, 7%–13%), β-globulins of 14.2% (8%–14%), γ-globulins of 20.8% (N, 12%–22%). Ultrasonography of the internal organs showed free fluid in the abdominal cavity (approximately 2 l). Fibrogastroscopy showed pyloritis, gastroptosis, gastroduodenal reflux, and a suspicion of celiac disease. Fibrocolonoscopy showed dolichocolon and enteritis. Biopsy of the descending part of the duodenum showed chronic enteritis with significant atrophic-inflammatory changes. Biopsy of the intestine sample taken during colonoscopy showed chronic enteritis with significant atrophic-inflammatory changes. Serological testing was done to confirm celiac disease, showing that IgA to deamidated gliadin was higher than 9 (N <1), IgG to deamidated gliadin was 0.22 (N, 1), IgA to tissue transglutaminase 2 was 2.88 (N, 1), IgG to tissue transglutaminase 2 was 0.06 (N, 1). Taking into account the results of all provided examinations, the diagnosis of celiac disease of adults was confirmed. After 10 days of treatment, including a gluten-free diet, the patient was discharged in an improved condition.

DISCUSSION In recent years, celiac disease has witnessed a growing recognition in adults. The symptoms of celiac disease can widely vary and they include abdominal pain of different localization, infectious-like diarrhea, and general or localized edema, making diagnosis even more elusive. Balancing serological tests with intestinal biopsies remains a crucial aspect of celiac disease diagnostics in adults. This dual approach helps in achieving a more accurate diagnosis and ensures that the patient with celiac disease was not overlooked due to potential false negatives from serological tests alone.

CONCLUSIONS Currently, celiac disease is being diagnosed also in older patients. In presentation, it can be very similar to, or accompanied by, chronic pancreatitis, gastritis, or irritable bowel syndrome. The combination of the characteristic symptoms should lead to a thorough patient examination, including fibroscopy with biopsy and, if any suspicion exists, serological testing.

Key words
celiac disease, gluten-free diet, gluten intolerance, malnutrition small bowel disease

PABLO IGLESIAS

Secondary hypokalemic paralysis as a presentation of primary aldosteronism: a case report

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CASE DESCRIPTION Secondary hypokalemic paralysis can be a rare manifestation of primary aldosteronism. The case involves a 74-year-old, severely hypertensive, patient with difficult blood pressure control, who developed acute symmetrical proximal weakness, preventing him from getting up. On admission, potassium levels were 2.68 mEq/l and creatine kinase levels were 286 UI/ml (1.5 times the upper normal limit). Treatment was initiated and symptoms resolved. With persistent hypokalemia and metabolic alkalosis, a study of primary aldosteronism was initiated. Aldosterone levels were at 28.3 and plasma renin activity at 0.3. Computed tomography of the adrenal glands showed left adrenal adenoma and early renal cell carcinoma. Acute hypokalemic weakness can occur without reaching very low levels of potassium, which is a common complication of primary aldosteronism (due to very high aldosterone levels). At the same time, according to recent evidence, elevated levels of aldosterone could lead to, and promote, the development of renal cell carcinoma.

Key words
primary aldosteronism, renal cell carcinoma, secondary hypokalemic paralysis

PAUL BESTER

Protein-losing enteropathy in a young patient with chorea

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INTRODUCTION Protein-losing enteropathy is an umbrella term for a diverse group of disorders causing uncompensated plasma protein loss in the gastrointestinal tract. The clinical presentation is highly variable and depends on the underlying cause, and the subsequent complications that may have developed. The diagnosis of protein-losing enteropathy should be suspected if no other cause for low plasma proteins is found, such as inadequate intake, inadequate synthesis or excessive renal loss. The preferred method of confirming the diagnosis is α1-antitrypsin clearance; however, scintigraphy can also be used. After protein-losing enteropathy has been confirmed, further investigation should focus on the cause, guided by the history and physical exam. This may include further imaging, endoscopy or specific laboratory testing.

CASE DESCRIPTION A 16-year-old woman presented with a 4-day history of involuntary choreiform movement of all 4 of her limbs. She also reported chronic diarrhea. Hospital records showed she was previously diagnosed with rheumatic heart disease and protein-losing enteropathy. The cause of the protein-losing enteropathy was attributed to lymphatic obstruction from disseminated tuberculosis when she was 4 years old; however, it was never confirmed by α1-antitrypsin clearance or scintigraphy. Initially, a working diagnosis of acute rheumatic fever was made. Using the Jones criteria, the 1 major criterion met was Sydenham chorea and the 2 minor criteria met were raised inflammatory markers and a history of fever. On further testing, she still had low levels of total protein (37 g/l), albumin (8 g/l), IgG (6.82 g/l), and vitamin D (27.6 nmol/l). Kidney and liver functions were normal. The patient also had hypocalcemia with ionized calcium of 0.77 mmol/l and normal parathyroid and thyroid levels. Her anti-streptolysin-O-titer proved negative. The protein-losing enteropathy was confirmed by using both α1-antitrypsin clearance and scintigraphy. Using the tracer Technetium 99m-methylene diphosphonate (99mTc-MDP), scintigraphy showed radiopharmaceutical accumulation in the bowel, originating in the terminal ileum region. The α1-antitrypsin clearance was 81 ml/day, in keeping with protein-losing enteropathy. In further investigation for the cause, bidirectional endoscopy was performed. The histology showed xanthomas of the duodenum and chronic active colitis of the colon. Serological screening for celiac disease proved negative with normal IgA levels. The brain magnetic resonance imaging findings were normal. Chorea improved following neuropathic therapy and calcium / vitamin D supplementation. A working diagnosis of inflammatory bowel disease was made and the patient was started on sulfasalazine. At follow-up, the patient reported improvement of the diarrhea and her albumin level had increased.

CONCLUSIONS Protein-losing enteropathy has a wide range of clinical presentations, depending on its underlying cause and complications. In our case, it appears that the malabsorption of calcium and vitamin D led to chorea. The recommendation is to confirm protein-losing enteropathy with an α1-antitrypsin clearance test, but we found scintigraphy, more specifically the use of 99mTc-MDP (a tracer usually used for bone scans), to be valuable. In the pursuit of the underlying cause, our case affirmed that bidirectional endoscopy is mandatory in the investigation of protein-losing enteropathy.

Key words
chorea, vitamin D, malabsorption, protein-losing enteropathy, technetium 99m-methylene diphosphonate

PETR DRENKO

Hepatic hemangioma as a cause of consumption coagulopathy

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INTRODUCTION Disseminated intravascular coagulopathy (DIC) is a generalized, dynamically evolving, and ultimately life-threatening condition reflecting a complex process of dysregulation of the coagulation system. DIC is determined by abnormal, and often massive, systemic activation of coagulation and fibrinolysis by various mechanisms interfering with the physiological regulation of hemostasis, such as excessive exposure to procoagulant factors (eg, tissue factor expression by cancer cells), formation of circulating neutrophil extracellular traps (NETs) or morphological abnormalities of the endothelium with platelet uptake and activation in the case of large vascular malformations and tumors. Uncontrolled, ongoing activation of the coagulation process leads to extensive microthrombus formation with subsequent consumption of endogenous coagulation factors and platelets. The clinical condition, framed by the risk of dysfunction of any target organ related to hypoperfusion, thrombotic or bleeding complications, correlates with the severity, process rate, and nature of the underlying disease.

CASE DESCRIPTION A previously healthy 60-year-old patient presented for the first medical evaluation with an approximately 4-month history of progressive increase in abdominal girth, dyspeptic syndrome, and weight loss. Computed tomography showed multiple solid cystic expansions of the liver, occupying most of the abdominal cavity, that were not characteristic of hepatocellular carcinoma or metastatic infiltration. The initial complement of laboratory, imaging, and endoscopic examinations did not narrow the differential diagnosis of the liver masses. Therefore, puncture biopsy of the liver was performed with collection of 3 samples, which correlated in their histopathological morphology with numerous large vascular lesions with features of cavernous hemangioma. The patient subsequently required hospitalization for prolonged bleeding after dental extraction, anemia that required blood transfusions, and, especially, spontaneous coagulopathy with prolonged coagulation tests (prothrombin time, thrombin time, and activated partial thromboplastin time), and mild thrombocytopenia. Detailed examination of coagulation parameters found slightly reduced plasma factor VII and plasma factor XI activity, moderate fibrinogen deficiency, and high D-dimer levels. After correction of the examined plasma with the reference plasma, prothrombin time and activated partial thromboplastin time were normalized in vitro. Normocytic anemia of combined cause was accompanied by microangiopathic changes in the peripheral blood smear. The patient was hemodynamically stable throughout the hospitalization, no other organ dysfunction was present, and inflammatory markers were negative. After substitution with coagulation factors and repeated dental treatment, the bleeding stopped. Since one of the cornerstones of the therapeutic approach to a patient with DIC is the elimination of the process generating dysregulation in the coagulation system, cadaveric liver transplantation was successfully performed for our patient in category 2 for hospitalized patients.

CONCLUSIONS Early identification of the coagulation disorder, its stage, and recognition of the underlying, generating disease are crucial for the proper management and patient outcome. Here, we present a rare case of progressive hepatic cavernous hemangioma underlying hemorrhagic symptomatology through consumption coagulopathy with features of decompensated chronic course of DIC. In this patient, the causative treatment of consumption coagulopathy was cadaveric liver transplantation.

Key words
bleeding disorder, consumption coagulopathy, disseminated intravascular coagulopathy, hepatic hemangioma, liver transplantation

PETR URBAN

Beads on a string: a hidden etiology of secondary hypertension

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INTRODUCTION Secondary hypertension accounts for up to 10% of hypertensive patients, with a significant proportion of younger adults affected. Timely and precise identification of the underlying cause is essential for successful management, which frequently also means a marked improvement in a patient's quality of life. In this case report, we present the diagnostic and therapeutic approach to a 29-year-old patient with clear secondary hypertension, in whom the etiology has been much more covert.

CASE DESCRIPTION We describe the case of a 29-year-old woman, who had been diagnosed with severe systolic-diastolic hypertension during a routine examination by a general practitioner. Until then, all her blood pressure readings had been normal. Subsequently, the hypertension was not sufficiently controlled even with a combination of 3 antihypertensives, and given its severe grade and the patient's age, it exhibited typical signs of secondary hypertension. Therefore, the diagnostic workup was initiated. The patient had a prior history of chronic kidney disease and had been under the care of a nephrologist since early childhood due to chronic hydronephrosis of the right kidney. It could have been easily assumed that the etiology of hypertension was renal parenchymal, which is one of the most common ones. However, further examinations were also performed to rule out other causes. Eventually, sonographic examination showed significant stenosis of the left renal artery. This had been the true cause of the abrupt onset of hypertension, which had been renovascular. The patient underwent angiographic examination and subsequent balloon angioplasty. The patient's young age and the angiographic image, representing the unifocal type of fibromuscular dysplasia (FD) with one isolated stenosis made FD the most likely cause of the stenosis. Because the patient subsequently developed a restenosis, she had to undergo a second intervention with drug-coated ballon angioplasty. Since then, the patient has had her hypertension under control, even without any antihypertensive medication, and she has been feeling well.

DISCUSSION Renovascular hypertension is one of the rarer types of secondary hypertension and represents impaired renal perfusion. Main differential diagnosis includes atherosclerotic stenotic lesions of renal arteries and fibromuscular dysplasia. The latter is much more common in younger patients and women, and exhibits 2 distinct angiographic images. The multifocal type displays the typical beads-on-a-string pattern, with multiple stenoses and interposed aneurysms, whereas the unifocal type presents with an isolated stenosis. The cause of the disease is unknown, therefore, patient management is focused on mechanical dilation of the affected arteries. Due to the overall younger age of the patients, stenting is generally avoided.

CONCLUSIONS This case highlights the significance of a methodical diagnostic approach to secondary hypertension. Although in our patient the explanation of the hypertension could have been seen at first glance, thorough examination showed the true etiology and enabled efficient treatment. Had the renal artery stenosis not been identified, the patient would have progressively lost function of the healthy left kidney and her quality of life would arguably have been significantly reduced.

Key words
fibromuscular dysplasia, renal angioplasty, renovascular hypertension, secondary hypertension

RADKA HREŠKOVÁ

Pseudoaneurysm of the mitral-aortic intervalvular fibrosa: serious complication or benign finding?

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CASE DESCRIPTION Pseudoaneurysm of the mitral-aortic intervalvular fibrosa (P-MAIF) is an uncommon, but potentially life-threatening, consequence of infective endocarditis or surgical valve operation. It can cause symptoms and complications requiring cardiac surgical intervention. The natural course of asymptomatic P-MAIF is largely unknown, since surgery is offered to most patients. In the following case report, we discuss the clinical decision-making in a patient with P-MAIF after aortic valve replacement (AVR) surgery with a clinical history of infective endocarditis (IE) and perivalvular abscess formation. Early follow-up after AVR surgery was complicated by the presence of P-MAIF arising from the left ventricular outflow tract (LVOT) with severe functional mitral regurgitation (MR). Because of acute heart failure, due to hemodynamically significant P-MAIF, the heart team decided to perform urgent reoperation with suture repair for aortic bioprosthesis paravalvular leak. Mitral and tricuspid valve repair was performed with annuloplasty ring. The next echocardiographic follow-up found persistent P-MAIF below aortic bioprosthesis. There was no evidence of possible fatal complications. Because of asymptomatic, uncomplicated P-MAIF the patient was managed conservatively with close clinical and echocardiographic follow-up. Up to this day, mild symptoms of heart failure remain (New York Heart Association class II). During 9-year echocardiographic follow-up no significant changes in the size of the P-MAIF or its flow pattern were seen. There was a mild progression of left ventricular end-diastolic diameter. No significant valve dysfunction occurred. Direct oral anticoagulant treatment for preventing thromboembolic events in atrial fibrillation was used. In some clinical situations, P-MAIF did not seem to be a fatal condition that would require prompt surgical intervention. In some asymptomatic patients with uncomplicated P-MAIF, conservative management with close clinical and echocardiographic monitoring of this rare complication seems to be a possible way of treatment.

Key words
echocardiography, infective endocarditis, mitral-aortic intervalvular fibrosa, pseudoaneurysm

RADKA KOUBKOVÁ

Where does a pneumococcus hide?

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INTRODUCTION General weakness, spinal pain, productive cough, and breathlessness. These symptoms are very common in oncologically treated patients with the primary diagnosis of generalized respiratory tumor. Could there be something more behind it all? And how helpful can sonography be in clinical practice?

CASE DESCRIPTION Our patient was an oncologically treated woman with the primary diagnosis of generalized bronchogenic carcinoma (now in local progression), who finished the second series of chemotherapy shortly before presenting. She was admitted for progressive weakness, shortness of breath, and pain in the cervical spine. Basic biochemical blood tests showed a marked elevation of inflammatory parameters. We took local cultures, including

hemocultures, although the patient was completely afebrile from the onset of her symptoms and during the hospitalization. Chest scan ruled out any inflammatory infiltration. Only a large tumorous lesion in the right upper lung field was present, which has already been already described. We started empiric antimicrobial therapy. The next day, hemoculture came back positive with *Streptococcus pneumoniae*. At first, we thought it might be pneumonia, hiding behind the large tumor. Therefore, we continued the established antibiotic therapy. Then, control blood samples showed a decrease in inflammatory parameters. Subjectively, the patient complained of shortness of breath and development of symmetrical swelling of the upper and lower extremities was observed. As a part of the sonography training at our department, the patient underwent a complete sonographic examination. After applying the probe to the heart area, vegetation on the mitral valve was visible. Transthoracic echocardiography also showed grade III mitral regurgitation with preserved left ventricular ejection fraction. We performed a whole-body computed tomography scan to rule out systemic embolism. It showed an advanced lung tumor with significant right-sided fluidothorax (signs of anasarca), but an embolism was ruled out. We performed diagnostic thoracocentesis of the right pleura, which yielded macroscopically yellow fluid. Samples were sent for further examination. Biochemical analysis showed that the fluid was transudative and cytology came back negative. Therefore, the patient was referred to the cardiac surgery department, where, taking into account the underlying disease, she was indicated for conservative treatment of infective endocarditis.

CONCLUSIONS Infective endocarditis is a rare disease and without timely diagnosis and treatment, its consequences are fatal. It can be all the more insidious, if the causative pathogen is uncommon (eg, *S. pneumoniae*). Although this disease is rare, it should be considered, especially in patients who do not respond typically to antibiotic therapy. Immunocompromised patients represent a special group, where it is necessary to search for atypical localizations of the pathogen, in case of an infection. In this case, the main factor contributing to the early diagnosis was the regular sonography training at our department. Sonography is often overshadowed by other, sometimes more burdensome imaging methods, although it is a very quick and easy examination. This inexpensive, non-invasive examination method is very helpful and can guide the physician in the right direction, leading to early diagnosis.

Key words
immunosuppression, infective endocarditis, invasive pneumococcal disease, *Pneumococcus*, sonography

ROCÍO GARCÍA GARCÍA

Acute aortic thrombosis in a young patient

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CASE DESCRIPTION Despite being commonly associated with such conditions as aneurysm, aortitis, atherosclerosis, aortic dissection, and aortic graft material, aortic thrombus is rarely seen in patients with a healthy aortic wall. Aortic thrombus is most often asymptomatic, but may present with limb ischemia, visceral ischemia or stroke from embolization.

A 36-year-old woman was admitted to our hospital for sudden pain in the right lower limb. She rated her pain as 8 out of 10, and described it as radiating from the knee to the foot. She felt tickling in the external part of the foot and fingers and had some difficulty moving them.

Her past medical history was relevant for: restless leg syndrome, hypothyroidism, and hypercholesterolemia. She also smoked 10 cigarettes per day. She was medicated with oral contraceptives (estrogen/progesterone), levothyroxine, and pramipexole.

The patient's physical examination showed paleness and lower temperature of the right limb, and allodynia. The posterior tibial, peroneal, and popliteal pulses of the right leg could not be palpated. The pulses of her left leg were present. The rest of her cardiovascular exam was normal.

The patient was considered to have a stage IIb acute limb ischemia, as per the Rutherford classification. Doppler ultrasonography showed a marked decrease in flow velocities and no flow in the pedal artery.

The investigation was completed with computed tomography angiography, which showed mural thrombus in the right lateral face from the distal aorta, occluding 30% of its lumen and extending into the right primitive iliac artery, occluding it almost entirely. Anticoagulation with intravenous heparin was started and the patient was taken to the operating room, where embolectomy was performed with a Fogarty catheter. Deep femoral embolic and thrombotic material was extracted and abundant reflux was restored. Thrombi were also removed from the aorta and the common iliac artery. The patient's postsurgical period was unremarkable and she was discharged within 6 days. Embolic source was ruled out with Doppler and Holter echocardiography. Thrombophilia study showed a slight decrease in protein C levels, without evidence of antiphospholipid antibody syndrome. At the time of diagnosis, her low-density lipoprotein cholesterol levels were 165 mg/dl and high-density lipoprotein cholesterol levels were 38 mg/dl. The patient was counselled on smoking cessation. She was discharged on anticoagulation, clopidogrel, and high doses of statins .

Primary aortic abdominal thrombus occurs in a normal, or minimally atherosclerotic, aorta and is often pedunculated and protrudes into the aortic lumen. Most often, no cause can be identified, but some cases have been associated with hypercoagulable states (eg, antiphospholipid syndrome). They are rarely reported in the literature. The diagnosis is typically confirmed by either computed tomography angiography or transesophageal echocardiography. Treatment requires anticoagulation along with an endovascular intervention or open surgical therapy. Long term management of these patients remains a subject for debate.

Key words
aortic thrombosis, rare presentation, vascular diseases

SANTIAGO GOMEZ JORDAN

Tuberculous abscess of the prostate: molecular diagnostic testing

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CASE DESCRIPTION Urogenital tuberculosis (UGTB) poses significant diagnostic and therapeutic challenges, particularly in patients coinfected with HIV. We describe a case of a 57-year-old man from northern Colombia who attended the emergency department with a month-long history of progressive hypogastric pain, dysuria, and urgency. He also reported consumption of alcohol, tobacco, and drugs. Apart from that, his medical history was unremarkable. Physical examination showed low body mass index, bilateral pulmonary crackles, and generalized abdominal pain. The prostate that was tender and nodular on digital rectal examination. Rapid HIV test was positive with a low CD4 count of 111/mm^3 and high viral load (1 715 225 copies/mm^3). Imaging studies showed miliary pattern on computed tomography (CT) of the lung, suggestive of tuberculous involvement. Pulmonary tuberculosis was confirmed with positive real-time polymerase chain reaction (rt-PCR) test in bronchoalveolar

lavage. Regarding abdominal pain and urinary symptoms, an 18-cc prostatic abscess was identified on the abdominal CT scan. After transurethral prostatic drainage, reverse transcription polymerase chain reaction (rt-PCR) confirmed *Mycobacterium tuberculosis*. Culture for common pathogens and rt-PCR test for *Neisseria gonorrhoeae* and *Chlamydia trachomatis* from the drainage, were negative. After the diagnosis, the patient received antituberculosis treatment and 14 days later, antiretroviral therapy was also started. The patient demonstrated clinical improvement and was discharged with follow-up recommendations. This case underscores the importance of early recognition of UGTB, particularly in HIV-positive individuals, and the critical role of molecular testing in the accurate diagnosis of tuberculosis.

Key words
diagnosis, *Mycobacterium tuberculosis*, polymerase chain reaction, prostate abscess, urogenital tuberculosis

SHAHROZ AZHAR

Multiple endocrine neoplasia: a nightmare to diagnose in resource-limited settings

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INTRODUCTION Multiple endocrine neoplasia type 2A (MEN2A) is a rare autosomal dominant inherited cancer syndrome. It makes up approximately 70%–80% of cases of MEN2. It is characterized by occurrence of distinct proliferative disorders of endocrine tissue. The incidence of MEN2A is 1 in 200 000 live births. MEN2 is clinically characterized by MTC, pheochromocytoma, and hyperparathyroidism. We present a case of a family with MEN2A syndrome.

CASE DESCRIPTION A 27-year-old woman presented to a secondary care hospital with palpitations, intermittent headache, and hypertensive urgency that have been going on for a year and had been managed as per guidelines in cases of emergency. Investigations were requested to rule out causes of secondary hypertension. Doppler abdominal ultrasound showed normal flow indices and raised 24 hour urinary free metanephrines (1076.92 µg/day). The patient's maternal uncle also underwent bilateral adrenalectomies for pheochromocytoma (15 years earlier). Ultrasound of the neck showed normal thyroid gland with TI-RADS, a spongiform nodule in the right thyroid lobe with normal thyroid function. Calcium (12.5 mg/dl), PTH (318.3 pg/ml), and calcitonin (37.5 pg/ml) levels were raised. Prolactin (6.2 ng/ml) and normal serum 25-0H vitamin D (45.8 ng/ml) were normal. Parathyroid planner imaging with single photon emission computed tomography showed scintigraphic evidence of hyperfunctioning parathyroid tissue at the inferior pole of the right lobe of the thyroid gland. Computed tomography with adrenal protocol showed bilateral adrenal nodules. Positron emission tomography-computed tomography DOTA scan was not done due to financial constraints and technical difficulties, but it was discussed with a radiologist, who was certain that it was bilateral pheochromocytoma.

Since the patient was symptomatic with biochemical and radiological evidence of pheochromocytoma, before surgery, she was started on α- and β-blockers. Histopathology report showed encapsulated neoplasm arising in the adrenal gland, composed of cells with basophilic cytoplasm and variable salt and pepper nuclei. The cells were polygonal with numerous areas of spindled appearance. Some cells had markedly pleomorphic nuclei and were arranged in a Zell Bellen pattern.

The patient underwent hemithyroidectomy and selective parathyroidectomy. Histopathology showed medullary thyroid carcinoma and parathyroid adenoma, now booked for total thyroidectomy and cervical lymph node dissection.

DISCUSSION In MEN2A, approximately 70%–95% of individuals develop MTC, 50% develop pheochromocytoma, and 15%–30% develop hyperparathyroidism. Early diagnosis established through screening of family members is essential in MEN2, because of medullary thyroid cancer associated with its morbidity and mortality. MTC can be cured or prevented by early thyroidectomy. On further investigations, raised calcitonin levels were found in our patient, despite of no suspicious intrathyroidal nodules on ultrasound. In the presented case, the patient's maternal uncle, who previously underwent bilateral pheochromocytoma, was the first family member to come to medical attention with a known manifestation of MEN2. This phenomenon, known in genetics as "anticipation" (ie, manifestations of a genetic disorder occurring earlier in the succeeding generations), occurring in MEN2A, has only been described in the literature once. This case was a real challenge for us to diagnose in the presence of financial constraints and unavailability of mutational analysis.

CONCLUSIONS Logical explanation of symptoms and signs helps a clinician to diagnose rare syndromes and manage them.

Key words
medullary carcinoma of thyroid, multiple endocrine neoplasia type 2A, phaeochromocytoma, parathyroid adenoma, *RET* proto-oncogene

SIMON HOYOS PATINO

Acute interstitial nephritis secondary to atheroembolic renal disease, mimicking ANCA vasculitis

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INTRODUCTION Atheroembolic renal disease is a rare cause of acute kidney injury (AKI) resulting from the obstruction of the renal arteries by cholesterol crystals. It is a multisystemic disease that affects the kidneys, skin, gastrointestinal system, and brain, presenting with livedo reticularis, purpura, nodules, blue toe syndrome, ischemia, uncontrolled hypertension, anemia, thrombocytopenia, eosinophilia, elevation of inflammatory markers, proteinuria, and eosinophiluria. In some cases, the presence of antineutrophil cytoplasmic antibodies (ANCA) have been reported, causing it to mimic ANCA-associated vasculitis.

CASE DESCRIPTION A 66-year-old man with a history of smoking, hypertension, prediabetes, obesity, knee osteoarthritis, dyslipidemia, nephrolithiasis, and chronic kidney disease, experienced a decline in renal function a month after the correction of an abdominal aortic aneurysm with a stent graft. Complex atheromatosis of the aorta was observed during the procedure and the patient developed blue toe syndrome postoperatively. Upon admission, the patient was hypertensive and had unintentionally lost 10 kg in the last month. Physical examination showed purplish discoloration in several toes and livedo reticularis. The diagnostic approach for acute kidney disease was initiated; renal ultrasound showed no abnormalities and urine cytology showed proteinuria. Ancillary tests were positive for ANCA, as measured by enzyme-linked immunosorbent assay and peripheral eosinophilia. Renal biopsy reported acute interstitial nephritis, interpreted in the clinical context as secondary to atheroembolic renal disease. This condition explained ANCA positivity, peripheral

eosinophilia, skin findings, and histological features. Ultimately, the patient received immunosuppressive treatment, leading to improved renal function. To date, the patient has had a satisfactory outcome.

DISCUSSION Atheroembolic renal disease, occurring in patients with systemic atherosclerosis, can affect renal function acutely, subacutely or chronically. This disease can mimic small vessel vasculitis, presenting with similar clinical manifestations and laboratory findings. Several cases in the literature describe patients developing cholesterol embolism syndrome, secondary to arterial procedures or spontaneously, with ANCA positivity and systemic clinical manifestations. These cases highlight the complexity of diagnosing and managing atheroembolic renal disease, emphasizing the need to consider it in the differential diagnosis of small vessel vasculitis.

CONCLUSIONS In this case, clinical manifestations, laboratory findings, and ANCA positivity suggested vasculitis. However, renal biopsy showed absence of typical vasculitis findings and signs of acute interstitial nephritis. Furthermore, clinical presentation and history of repairing an abdominal aortic aneurysm supported the diagnosis of atheroembolic renal disease. This disease can mimic ANCA-associated vasculitis, underscoring the importance of a careful differential diagnosis.

Key words
antineutrophil cytoplasmic antibodies, atheroembolic renal disease, vasculitis mimics

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From sport to thromboembolism

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CASE DESCRIPTION Paget-von Schrötter syndrome is a primary thrombosis of the subclavian vein in the area of the costoclavicular junction, that results from repeated overload and manifests as thoracic outlet syndrome (TOS). Anatomical abnormalities and repeated injuries to the vascular endothelium are key factors in its initiation and progression. Due to the threat of pulmonary embolism, possible development of post-thrombotic syndrome of the upper limb, and recurrence of thrombosis, early diagnosis and proper treatment are important. We present the case of a young woman, a professional volleyball player, with confirmed thrombosis of the left upper limb as a result of compression within the venous TOS. As a case, we point out the variability of the cause of thromboembolic disease in young individuals and the need to know its rare causes (strait syndrome).

Key words
deep vein thrombosis, exertional thrombosis, upper thoracic aperture syndrome

SOLOMIIA GUTA

Acute myocardial infarction in a woman

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INTRODUCTION Differential diagnosis of patients with angina can be challenging. It is difficult to ascertain the presence of angina pectoris in a middle-aged woman, and a primary diagnosis of coronary heart disease is rarely considered. The purpose of the work is to discuss another cause of myocardial infarction in a patient without atherosclerosis of the coronary arteries.

CASE DESCRIPTION We describe a case of a 54-year-old woman who was admitted to the cardiology department with complaints of intense pressing pain radiating to the interscapular area and left arm, accompanied by shortness of breath, pronounced general weakness, and increased blood pressure.

Pain of a similar nature would occur in the patient over the past 5 years during physical activity with a change in body position. The positive effect of nitrates was not pronounced.

The pain began to bother the patient when she changed her job to a one requiring more physical effort. The patient did not smoke and was not genetically burdened. On admission, her breathing was vesicular, her cardiac activity was rhythmic, and her tones were muffled. Blood pressure was 150/90 mm Hg, and heart rate was 80 bpm. Echocardiography showed normal heart chambers, and total contractility was satisfactory. The patient was prescribed medications (nitrates, β-blockers, anticoagulants, and disaggregants), which resulted in pain resolution.

Coronary angiography visualized balanced blood circulation, with the middle segment of the anterior interventricular artery showing a muscular bridge with up to 50% stenosis in systole, and atherosclerotic changes without hemodynamically significant stenoses in the right coronary artery. The patient remained stable after discharge. She was prescribed Coplavix (acetylsalicylic acid with clopidogrel), atorvastatin, diltiazem, nebivolol, and ramipril, and her tolerance to physical exertion increased.

DISCUSSION Myocardial muscle bridge (MM) is a cardiac muscle fiber that passes over a segment of the epicardial coronary artery that narrows the lumen of the vessel only during myocardial systole. Its localized almost exclusively over the anterior interventricular branch. Unfortunately, invasive treatment is rarely justified. In most cases, the presence of MM is not accompanied by clinical symptoms. In this case, it was reflected by the occurrence of systolic compression of the coronary artery during exercise.

CONCLUSIONS The peculiarity of the clinical course of a myocardial bridge in a patient with myocardial infarction was considered. In the case of typical angina pectoris in an atypical young patient, it is necessary to carry out additional invasive examinations to identify possible organic changes and congenital anomalies of the coronary vessels, and further prescribe appropriate treatment methods, in order to prolong the patient's life and improve its quality.

Key words
myocardial infarction, myocardial muscle bridge

TEODOR TAKAC

Anemia in a patientwho underwent liver transplantation, due to primary sclerosing cholangitis: a rare cause, a rare treatment, and an uncertain prognosis

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INTRODUCTION Primary sclerosing cholangitis (PSC) represents a chronic autoimmune liver disease, in which inflammation and fibrosis lead to multifocal bile duct strictures. The disease is of progressive nature and in most cases, leads to the development of liver fibrosis. PSC is often associated with inflammatory bowel disease (IBD) and has a significant malignant potential. The only prognosis-modifying treatment is liver transplantation (LT). The risk of recurrence of PSC after LT is approximately 30%, and various early and late complications can occur in the post-transplantation period

CASE DESCRIPTION We present a case of a 38-year-old woman who underwent LT due to PSC, in whom the underlying disease

recurred and progressed rapidly to advanced chronic liver disease. The patient was qualified for liver retransplantation placed on the waiting list. The reason for her last hospitalization was severe normocytic normochromic anemia with pronounced anemic syndrome. Blood loss due to portal hypertension was at the top of the list in our differential diagnosis. We performed an upper endoscopy with the finding of small esophageal varices, but without signs of active bleeding. Contrast-enhanced computed tomography scan of the abdomen did not show signs of active bleeding either. Within a few days, the nature of the anemia changed to macrocytic hyperchromic and severe thrombocytopenia developed. Therefore, we investigated the parameters of hemolysis and made the diagnosis of Evans syndrome, which is characterized as autoimmune bicytopenia or pancytopenia. Most often, it is warm autoimmune hemolytic anemia in combination with immune thrombocytopenic purpura (ITP) or, rarely, with autoimmune neutropenia. The immunological condition of our patient did not allow for blood transfusion. This led to contraindication of LT. We initiated corticosteroid treatment, which did not lead to remission of the disease. In an almost hopeless situation, we decided to realize 5 immunopheresis with the subsequential administration of a monoclonal antibody against CD20. With this treatment, we managed to achieve the disappearance of autoantibodies and the negation of cross-match tests. Thus, we created a short window of time, in which it would be possible to perform liver transplantation. We struggled with various complications, such as refeeding syndrome, hepatorenal syndrome causing acute kidney injury, upper gastrointestinal bleeding, fluidothorax, and recurrent infections. Therefore, several times we had to temporarily remove the patient from the waiting list and put her back on, when she was in a better condition. After 82 days of hospitalization, a suitable liver donor appeared. From the beginning, the operation was complicated by significant bleeding, severe coagulopathy, and hemodynamical instability. After the procedure, the patient was in critical condition, ventilated and transferred to the intensive care unit, where acidosis progressed and the patient died from septic shock.

CONCLUSIONS Primary sclerosing cholangitis represents a rare disease that can affect the graft, even after liver transplantation. Autoimmune hemolytic anemia and thrombocytopenia are rare complications associated with PSC, IBD, and, occasionally, with the transplantation itself. Autoimmune hepatitis and ITP represent contraindications to liver transplantation in the active stage. Immunoadsorption and anti-CD20 therapy are effective treatment methods, which can lead to the induction of remission in patients with end-stage liver disease, thus enabling liver transplantation even in seemingly hopeless situations.

Key words
Evans syndrome, liver transplantation, primary sclerosing cholangitis

UPAMANYU BAG

TB or not TB, that is the question!

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INTRODUCTION Prolonged undiagnosed fever in an immunocompromised patient can be challenging for a physician, because a multitude of infectious and noninfectious etiologies can complicate the clinical picture.

CASE DESCRIPTION A 38-year-old man with no known comorbidities was admitted to a hospital with constitutional symptoms, cough, and generalized lymphadenopathy. He was promptly diagnosed to be in an immunocompromised state, with chest X-ray showing miliary mottling. At presentation, he was greatly emaciated, highly dehydrated, and tachypneic. On examination, he was pale, icteric, tachycardiac, and hypoxic. A provisional diagnosis of HIV infection with AIDS-defining disseminated tuberculosis (TB) was made, as this was the most common opportunistic infection in our country with such a presentation. Laboratory workup showed pancytopenia and elevated levels of liver enzymes. The patient was started on a modified antitubercular drug regimen. Due to a continuous decrease in erythrocyte sedimentation rate over the next 2 days, persisting pancytopenia, and clinical deterioration, secondary hemophagocytic lymphohistiocytosis (HLH) was suspected. Serum ferritin and triglyceride levels were also raised. Bone marrow examination showed histoplasma with hemophagocytes. Finally, a diagnosis of disseminated histoplasmosis with secondary HLH in a HIV-infected patient was made, and treatment with liposomal-amphotericin B and dexamethasone was started.

DISCUSSION Miliary shadows are micronodules resembling millet seeds, visible on chest imaging. The presentation of our patient mimicked a case of disseminated TB associated with an immunocompromised state, both clinically and radiologically. The prevalence of a disease in a geographical region molds the physician's thinking process to and influences their treatment decisions. In the presented case, TB was diagnosed based on the patient's clinical presentation, without bacteriological confirmation, and antitubercular treatment was started accordingly. Continuous assessment is necessary to rule out other associated contributors to the disease process. Bone marrow examination was performed in our patient with the idea of demonstrating hemophagocytes and looking for evidence of bone marrow infiltration by infectious agents. A delay in the diagnosis of histoplasmosis led to a delay in treatment.

CONCLUSIONS This case highlights the importance of microbiological confirmation and etiological diagnosis of an infectious agent in any immunocompromised patient admitted with fever. A differential diagnosis of histoplasmosis should always be kept in mind in the cases presenting with miliary shadowing on chest radiography images.

Key words
histoplasmosis, hemophagocytic lymphohistiocytosis