A 38-year-old man with an unremarkable medical history was referred to the National Center of Familial Hypercholesterolemia in Gdańsk, Poland, because of a significantly increased LDL cholesterol level of 8.24 mmol/l. He presented with arcus cornealis, which is typical of FH, and had a family history of high LDL cholesterol levels (mother, 9.61 mmol/l; sister, 7.25 mmol/l). Importantly, he had no history of any hypolipidemic therapy. Further genetic testing identified the presence of an LDL-receptor gene mutation (p.Glu140Asp). Transthoracic echocardiography and the carotid intima-media thickness measurements revealed no abnormalities. Therefore, we decided to extend the standard diagnostic protocol and assess microvascular endothelial function.

Here, we present the results of microcirculation measurements assessed simultaneously on the forearm during and following brachial artery occlusion by LSCI (PeriCam PSI System, Perimed, Järfälla, Sweden) and FMSF (Angionica, Ltd., Łódź, Poland). Speckle contrast analysis with colors ranging from blue (low perfusion) to red (high perfusion) showed relatively low basal microvascular perfusion (FIGURE 1A). We also observed a very weak postocclusive reactive hyperemic response (FIGURE 1B), suggesting a low vaso-dilatation potential, which is mainly endothelium-dependent. Similarly, FMSF revealed a poor endothelium-dependent hyperemic response, while ischemic response, which may reflect tissue sensitivity to hypoxia, remained within normal limits (FIGURE 1C).

To the best of our knowledge, this is the first report on a complex noninvasive assessment of microcirculation in a patient with FH. Although less extensively studied, microvascular disorders may precede endothelial dysfunction in large arteries and the subsequent clinical symptoms.

REFERENCES

CONFLICT OF INTEREST None declared.

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FIGURE 1 Imaging of a patient with familial hypercholesterolemia: A – basal microvascular perfusion on laser-speckle contrast imaging with colors ranging from blue (low perfusion) to red (high perfusion); B – weak postocclusive reactive hyperemic response with more intense perfusion; C – flow-mediated skin fluorescence showing a weak hyperemic response and normal ischemic response.