CLINICAL IMAGE

Familial chronic thromboembolic pulmonary hypertension in a mother and a son: successful treatment with refined balloon pulmonary angioplasty

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Chronic thromboembolic pulmonary hypertension (CTEPH) is an uncommon sequela of pulmonary embolism (PE), characterized by pulmonary hypertension resulting from unresolved thrombotic lesions in the pulmonary bed and pathological remodelling of small distal pulmonary arteries that are either affected by or spread from thrombotic lesions.[1,2] Recent studies have identified factors associated with an increased risk for CTEPH; however, no specific significant genetic factor mutations or familial history were reported among them.[1] Refined balloon pulmonary angioplasty (BPA) is an emerging therapeutic option for patients with CTEPH either with contraindications to pulmonary endarterectomy (PEA) or with unacceptably high perioperative risk.[3] Optical coherence tomography (OCT) is a novel, promising tool that has been reported to be useful in the diagnosis, management, and treatment of CTEPH.[4] To our knowledge, only 2 case reports of familial CTEPH have been published to date.[5,6] In these reports, familial CTEPH was defined as the occurrence of CTEPH in 2 cases in a family.[5,6] Thus, we present a report and OCT images of familial CTEPH in a mother and a son successfully treated with refined BPA.

The mother was diagnosed with deep vein thrombosis and unprovoked PE at the age of 80 years. CTEPH was diagnosed according to the European Society of Cardiology criteria at the age of 83 years, with a mean pulmonary artery pressure (PAP) of 36 mmHg and a pulmonary vascular resistance (PVR) of 6.5 wood units.[1] Laboratory test results for genetic or acquired thrombophilia were negative. Clinical characteristics and hemodynamic data are presented in supplementary material online. Due to advanced age, she was not referred for PEA. She underwent 2 successful sessions of BPA performed according to the previously described protocol.[2] In total, 5 segmental and subsegmental arteries were dilated with a significant decrease in the mean PAP to 21 mmHg and in PVR to 2 wood units (**FIGURE 1A–1D**; Supplementary material online, *Table S1*).

The son was diagnosed with acute unprovoked PE at the age of 55 years, without documented deep vein thrombosis. CTEPH was diagnosed at the age of 56 years, with a mean PAP of 42 mmHg and a PVR of 10.6 wood units. The test results for thrombophilia were negative, antinuclear antibodies were not detected, and other hematological parameters were negative (clinical characteristics and hemodynamic data are presented in Supplementary material online, *Table S1*). Despite potential surgical accessibility of intrapulmonary lesions, the patient was not referred for PEA owing to relapsing episodes of mental disease. He underwent 7 successful BPA sessions (14 arteries were dilated), with a significant decrease in the mean PAP to 23 mmHg and in PVR to 4 wood units (**FIGURE 1E–1H**; Supplementary material online, *Table S1*).

We described a case of familial CTEPH in a mother and a son. In both patients, refined BPA resulted in the normalization of mean PAP and significant functional improvement. Recently, Desmarais and Elliott[5] published the first description of familial CTEPH in a 54-year-old woman and her maternal aunt. Moreover, in a group of 160 Japanese patients with CTEPH, one pair of brothers had a documented familial pattern, which constituted 1.25% of all cases.[6] All these observations suggest an inherited predisposition to develop familial CTEPH.

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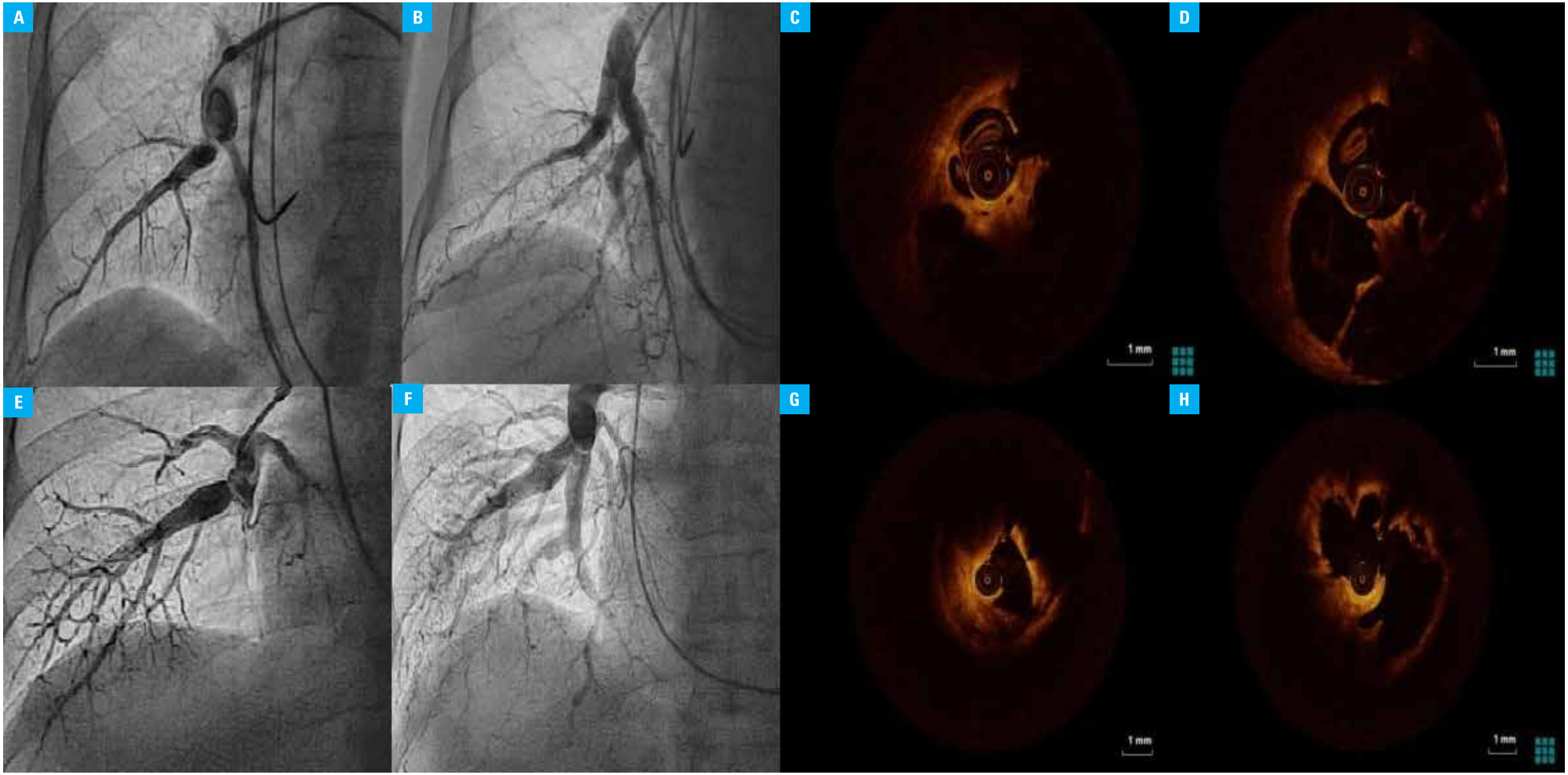

FIGURE 1 Angiographic and optical coherence tomography (OCT) images in a mother (**A–D**) and her son (**E–H**) with familial chronic thromboembolic pulmonary hypertension;
A – an angiographic view of the right lower pulmonary artery in the mother before balloon pulmonary angioplasty (BPA; web and total occlusions);
B – an angiographic view of treated segmental artery after successful BPA;
C, **D** – OCT images of thromboembolic lesions in the right lower pulmonary artery before BPA;
E – an angiographic view of the right lower pulmonary artery in the son before BPA (web and total occlusions);
F – an angiographic view of treated segmental artery after successful BPA;
G, **H** – OCT images of thromboembolic lesions in the right lower pulmonary artery before BPA.

Supplementary material online Supplementary material is available with the online version of the article at www.pamw.pl.

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