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Successful treatment of pulmonary hypertension with macitentan in a patient with Hermansky-Pudlak syndrome

Short title: Macitentan treatment in Hermansky-Pudlak syndrome

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Hermansky-Pudlak syndrome (HPS) is a rare autosomal recessive genetic disorder which is characterized by oculocutaneous albinism, a bleeding diathesis, and, in some individuals, pulmonary fibrosis or immunodeficiency [1, 2]. In this report, we presented an adult patient who was diagnosed with HPS and pulmonary hypertension (PH). In addition, we emphasized the importance of case management in this patient who could not be easily classified into one of the PH groups.

A 54-year-old female patient was admitted to our clinic with progressive dyspnea. Her functional capacity was classified as New York Heart Association (NYHA) class 3. She had a history of albinism, easy bruisability, and near- sightedness. On physical examination, her skin was pale and her irises were pigmented as well as horizontal nystagmus was present. On echocardiography (ECHO), left ventricle ejection fraction was normal and the left atrium dilated. Additionally, pulmonary forward flow and systolic pulmonary artery pressure (sPAP) were measured as 1.3 ms and 65–70 mm Hg, respectively. There was a minimal pericardial effusion. Tests for lupus

anticoagulant and factor V Leiden mutation were negative, and there was no evidence of syphilis, hepatitis B or C virus, or human immunodeficiency virus infections. Pulmonary-function tests showed mild functional impairments in the forced vital capacity (FVC) and forced expiratory volume in 1 second (FEV_1) and her FEV_1/FVC was %89. Pulmonary computed tomography angiography was performed, revealing no filling defects in the pulmonary arteries and their branches. On cardiac catheterization, the pulmonary artery systolic mean pressure and pulmonary vascular resistance were 30 mm Hg and 5.3 Wood units. Adhesion tests to detect platelet adhesion defects in terms of HPS were studied, all of them were positive. Macitentan was considered in this patient because of its good safety profile and efficacy [3]. After the medical treatment, her 6-minute walking test, which was 175 meters at the beginning, improved to 210 meters at 1 month and 280 meters at 3 months. There was no syncope episode and her N-terminal pro-B-type natriuretic peptide (NT-proBNP) values were within the normal limits, and pericardial effusion regressed on the ECHO. On the third month's ECHO, sPAP was 35 mm Hg (Figure 1, Supplementary material, *Table S1*).

Diagnosing PH can be complex and difficult in some cases [4]. HPS, also known as an oculocutaneous albinism, can cause pulmonary fibrosis leading to PH. A question can come to mind in a such patient. Is this patient group 1, group 3 or group 5 PH [5]? What should be our treatment approach in such a patient? When we searched the current literature, we could not see a comprehensive study on this subject. In our case, macitentan was given based on clinical benefit and patient satisfaction. This brings the case closer to either type 1 or type 5. Initiating specific treatment may be the first step in patients with HPS as there is no reversible etiology. The clear benefit of macitentan therapy in this patient suggests that such therapy is an effective option. We consider that our case is remarkable because it reveals the new etiology of PH.

Supplementary material

Supplementary material is available at https://journals.viamedica.pl/kardiologia_polska

Article information

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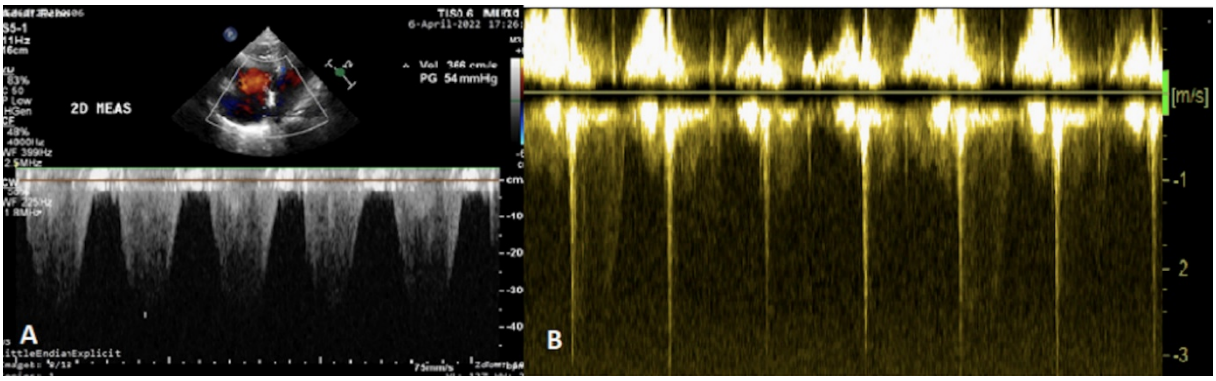

Figure 1. A. Systolic pulmonary artery pressure prior to macitentan therapy. **B.** Systolic pulmonary artery pressure at the 3rd month follow-up