

Case Report

Growing up with Idiopathic Pulmonary Arterial Hypertension: An Arduous Journey

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Abstract: Idiopathic pulmonary arterial hypertension (IPAH) is an uncommon and severe disease. We report the case of a 7-year-old boy investigated for cardiac murmur and exercise intolerance. Pulmonary hypertension (PH) was suspected at clinical examination and confirmed by echocardiography and cardiac catheterization. This case of pulmonary hypertension was classified as idiopathic given the negative etiological investigation. Vasoactive testing with oxygen and nitric oxide was negative. Therefore, treatment with sildenafil (1.4 mg/kg/d) and bosentan (3 mg/kg/d) was initiated. This allowed the stabilization of, but not a decrease in, pulmonary artery pressure for the next 5 years, during which the patient's quality of life was significantly reduced. At a later follow-up, the estimated pulmonary pressure was found to have increased and become supra-systemic, with a consequent deterioration in the child's condition. This led to the decision to enter him into a clinical trial that is still ongoing. Idiopathic pulmonary arterial hypertension is a severe disease that can present with non-specific symptoms, such as asthenia and exercise limitation, which are important not to trivialize. The disease is associated with significantly decreased quality of life in affected children and carries a high burden in terms of mortality and morbidity. The current knowledge about IPAH in children is reviewed, with a particular focus on the future prospects for its treatment and the related quality of life of patients.

Keywords: pulmonary arterial hypertension; idiopathic; children; vasoactivity; treatment; quality of life



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1. Introduction

Pulmonary hypertension is a rare but severe condition defined by a mean pulmonary arterial pressure (mPAP) > 20 mmHg at rest [1].

Depending on the etiology, five different groups of PH are described and listed in Table 1 [1,2]: pulmonary arterial hypertension (PAH) (group 1), PH associated with left heart disease (group 2), PH associated with lung disease and/or hypoxemia (group 3), PH associated with pulmonary artery obstruction (group 4), and PH with unclear and/or multifactorial mechanisms (group 5) [1–4].

PAH or pre-capillary pulmonary hypertension, which includes idiopathic pulmonary hypertension (IPAH), is defined as pulmonary hypertension with a pulmonary-artery-wedge pressure ≤ 15 mmHg with PVR ≥ 2 WU (2), excluding groups 4 and 5.

Idiopathic pulmonary arterial hypertension is a rare disease with an incidence ranging between 0.47 and 1–2 cases per million children, and with a prevalence of 2.1 to 4.4 cases per million children [2].

Although most children with PAH can now reach adulthood, thanks to recent therapeutic advances, the burden created by the disease in terms of morbidity and mortality remains high [5].

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