

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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Table of Contents A Journey With Pulmonary Arterial Hypertension

1. Understanding the eBook A Journey With Pulmonary Arterial Hypertension
 - The Rise of Digital Reading A Journey With Pulmonary Arterial Hypertension
 - Advantages of eBooks Over Traditional Books
2. Identifying A Journey With Pulmonary Arterial Hypertension
 - Exploring Different Genres
 - Considering Fiction vs. Non-Fiction
 - Determining Your Reading Goals
3. Choosing the Right eBook Platform
 - Popular eBook Platforms
 - Features to Look for in an A Journey With Pulmonary Arterial Hypertension
 - User-Friendly Interface
4. Exploring eBook Recommendations from A Journey With Pulmonary Arterial Hypertension
 - Personalized Recommendations
 - A Journey With Pulmonary Arterial Hypertension User Reviews and Ratings
 - A Journey With Pulmonary Arterial Hypertension and Bestseller Lists

5. Accessing A Journey With Pulmonary Arterial Hypertension Free and Paid eBooks
 - A Journey With Pulmonary Arterial Hypertension Public Domain eBooks
 - A Journey With Pulmonary Arterial Hypertension eBook Subscription Services
 - A Journey With Pulmonary Arterial Hypertension Budget-Friendly Options
6. Navigating A Journey With Pulmonary Arterial Hypertension eBook Formats
 - ePub, PDF, MOBI, and More
 - A Journey With Pulmonary Arterial Hypertension Compatibility with Devices
 - A Journey With Pulmonary Arterial Hypertension Enhanced eBook Features
7. Enhancing Your Reading Experience
 - Adjustable Fonts and Text Sizes of A Journey With Pulmonary Arterial Hypertension
 - Highlighting and Note-Taking A Journey With Pulmonary Arterial Hypertension
 - Interactive Elements A Journey With Pulmonary Arterial Hypertension
8. Staying Engaged with A Journey With Pulmonary Arterial Hypertension
 - Joining Online Reading Communities
 - Participating in Virtual Book Clubs
 - Following Authors and Publishers A Journey With Pulmonary Arterial Hypertension
9. Balancing eBooks and Physical Books A Journey With Pulmonary Arterial Hypertension
 - Benefits of a Digital Library
 - Creating a Diverse Reading Collection A Journey With Pulmonary Arterial Hypertension
10. Overcoming Reading Challenges
 - Dealing with Digital Eye Strain
 - Minimizing Distractions
 - Managing Screen Time
11. Cultivating a Reading Routine A Journey With Pulmonary Arterial Hypertension
 - Setting Reading Goals A Journey With Pulmonary Arterial Hypertension
 - Carving Out Dedicated Reading Time
12. Sourcing Reliable Information of A Journey With Pulmonary Arterial Hypertension
 - Fact-Checking eBook Content of A Journey With Pulmonary Arterial Hypertension
 - Distinguishing Credible Sources
13. Promoting Lifelong Learning

- Utilizing eBooks for Skill Development
- Exploring Educational eBooks

14. Embracing eBook Trends

- Integration of Multimedia Elements
- Interactive and Gamified eBooks

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