Pulmonary Vascular Disease Award funded in part by Janssen Pharmaceutical Companies of Johnson & Johnson

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Title: ***Investigating the Genes Involved in Pulmonary Arterial Hypertension in Congenital Heart Disease***

Heart defects are the most common type of birth defect, affecting up to 1% of the population worldwide. Congenital heart disease (CHD) can manifest in a variety of ways, ranging from simple lesions to complex defects that require major surgeries. Advances in pediatric cardiology and cardiac surgery have provided extraordinary opportunities for children with severe CHD, with the majority now surviving into adulthood. However, a major long-term complication of CHD that is still prevalent today is pulmonary arterial hypertension (PAH).

PAH is a progressive disease characterized by chronically elevated blood pressure in the pulmonary vasculature leading to right ventricular hypertrophy, heart failure, and premature death. PAH can occur in patients with repaired or unrepaired CHD, and studies have shown that as much as 10% of adult congenital heart disease patients are at risk. PAH usually results from increased blood volume or high-pressure blood flow in the lungs most commonly due to ventricular septal defects (VSD), atrial septal defects (ASD), or patent ductus arteriosus (PDA). Over time, this leads to changes in the vessel walls that result in PAH. PAH is a significant burden for patients, with common symptoms being shortness of breath, chest pain, and exercise intolerance. PAH can be a debilitating and life-shortening condition, which highlights the importance of further research to improve patient outcomes.

In recent years, there have been great strides in the understanding of the genetic basis of PAH. However, the genetic basis of PAH in patients with CHD is not well understood. Our research project aims to identify and characterize genetic mutations in patients with CHD and PAH, which could have a significant impact on predicting the long-term prognosis of patients with CHD by guiding diagnostic testing and treatment. This could improve the quality of life of patients with CHD and potentially extend their lifespan.

In conclusion, PAH is a severe complication associated with CHD that is a significant source of morbidity and mortality. The genetic basis of PAH in patients with CHD has not been comprehensively studied to date, and it remains unclear if specific genetic mutations contribute to the pathogenesis of this disease. This research project aims to uncover the genetic basis of PAH in patients with CHD and provide valuable information for predicting the long-term prognosis of patients with CHD, guide further diagnostic testing, and inform therapeutic decisions. The results of this research could be a game-changer, improving the quality of life and lifespan of patients with CHD and PAH.