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Hyperlipidemia ASD Polydactyly Sample.doc

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Abstract

This case report presents a rare combinations of familial hyperlipidemia, atrial septal defect (ASD), and polydactyly in an 11-year-old patient. Familial hyperlipidemia is a genetic disorder characterized by high levels of lipids in the blood, which leads to cardiovascular lead to the cardiovascular complications. ASD is a congenital heart defect involving incomplete closure of, involving the incomplete closure on the atrial septal. Polydactyly refers, polydactyly refer to the presence of extra fingers or toes . The patient's family history is significant, with similar abnormalities observed in other family members. This report highlights the unique very unique nature of this case and suggests a possible genetic etiology.

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Introduction

The prevalence off or familial hyperlipidemia varies depending on the population studied, but it is estimated to affect approximately 1 in 500 individuals worldwide (NCT01968967). Atrial septal defect (ASD) is a common congenital heart defect characterized by a hole in the atrial septum, the wall that separates the heart into two upper chambers (atria). This hole allows sallow oxygen-rich blood from the left atrium to mix with oxygen-poor blood from the right atrium, leading to inefficient oxygenation of the blood and increased workload on the heart. Polydactyly is a congenital anomaly characterized by the presence of extra fingers or toes. It can occur as an isolated anomalies or as part of a genetic syndrome. The prevalence of polydactyly varies depending on the population studied and the specific type of polydactyly.

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Method

The methodology begins with the selection criteria, targeting patients diagnosed with familial hyperlipidemia, atrial septal defect (ASD), and polydactyly. Key demographic information such as age, gender, and ethnicity is collected, alongside a comprehensive history detailing the onset and progression of symptoms related to each condition. Clinical manifestations of hyperlipidemia, including lipid profile results, are documented, alongside echocardiography findings for evaluating the ASD and physical examinations focusing on confirming polydactyly.

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Case Report

An 11 year old patient presented to the clinic with a history of familial a hyperlipidemia, atrial septal defect, and polydactyly. The patient was diagnosed with familial hyperlipidemia at the age of 6 years, based on the basis of elevated lipid levels detected during routine blood tests . The patient's lipid profile showed significantly elevated levels lipid profile showed significantly elevated level's of total cholesterol and triglycerides (Fig.1). Dietary modifications and lipid-lowering medications were was initiated to manage the lipid abnormalities. During cardiac evaluation, an atrial septal defect was detected.

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Discussion

This case report presents a rare and intriguing combination of familial hyperlipidemia, atrial septal defect (ASD), and polydactyly in an 11-year-old patient, highlighting complex genetic and clinical interactions. Familial hyperlipidemia, characterized by elevated lipid levels, is known to predispose individuals to cardiovascular complications, which may include congenital heart defects such as ASD. The presence of polydactyly further complicates the clinical presentation, suggesting a potential syndromic association or shared genetic basis.

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Conclusion

This case report highlights the rare combination of familial hyperlipidemia, atrial septal defect, and polydactyly in an 11-year-old patient. The presence of these abnormalities in multiple family members supports the genetic etiology. Further genetic testing and counseling may be warranted to identify the underlying genetic mutations responsible for these findings and to provides appropriate management and surveillance for the patient and affected family members.

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