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A RARE CASE REPORT OF FAMILIAL COMBINED HYPERLIPIDEMIA AND LIPOPROTEIN LIPASE DEFICIENCY PRESENTING AS ACUTE PANCREATITIS DURING INFANCY

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ABSTRACT Familial combined hyperlipidemia(FCHL) is an autosomal dominant genetic condition characterized by elevation in plasma low density lipoprotein(LDL) cholesterol and triglycerides and reduced plasma high density lipoprotein(HDL) cholesterol. Lipoprotein lipase (LPL), a member of triglyceride lipase gene family, it is synthesised by the parenchymal cells of the heart, skeletal muscle and adipose tissues and its main function is to hydrolyse plasma lipoproteins. LPL deficiency is a rare autosomal recessive disorder, often resulting in severe hypertriglyceridaemia from birth. We present a case of a infant with familial combined hyperlipidemia and LPL deficiency with elevated plasma triglyceride levels and LDL cholesterol, presenting with acute pancreatitis . This case report reviews the evidence for potential adverse effects of hypertriglyceridemia on health of the child.

KEYWORDS : low density lipoprotein(LDL), Lipoprotein lipase (LPL), high density lipoprotein(HDL),Autosomal dominant/recessive

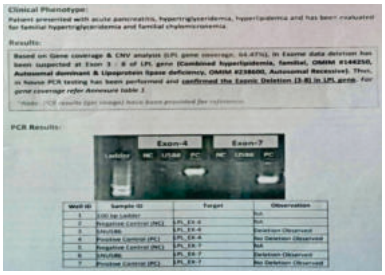
INTRODUCTION

FCHL is one of the common genetic hyper-lipidemias in the general population with prevalence estimated: 0.5%–2.0%.1 Familial combined hyperlipidaemia (FCHL) is a distinct entity, where the exact genetic and metabolic basis of the disease is not well understood.2 FCHL is often associated with early myocardial infraction, hypertension. 3 Lipoprotein lipase is a key enzyme for catalyzing the breakdown of triglycerides to lipoproteins.4 LPL deficiency is often characterized marked elevation of triglycerides.5

Hypertriglyceridemia often presents with acute pancreatitis.6 We present a rare case of genetically confirmed FCHL and LPL deficiency presenting as Acute pancreatitis during infancy.

Case Report

A 4 month old female child, first born for second degree consanguineously married couple with normal birth and developmental history ,with no family history of congenital hyperlipidemia syndrome, presented with history of excessive crying ,vomiting and abdominal distension for 2 days. At admission, child was febrile and hypotensive. On examination ,abdominal distension and hepatomegaly were noted. Blood investigations showed high triglycerides (24832 mg/dl), high cholesterol (562 mg/dl), Elevated LDL(213 mg/dl) ,Low HDL (11 mg/dl) ,elevated serum lipae (1294U/L). USG abdomen showed features suggestive pancreatitis. Child was treated symptomatically for acute pancreatitis. Child was started on fenofibrate and omega 3 fatty acid. Genetic workup(whole exome sequencing) results confirmed the EXONIC DELETION (3-8) in LPL gene and the diagnosis of FCHL and LPL deficiency was made. Child gradually improved and lipid profile showed decreasing trend of triglycerides(424mg/dl), cholesterol(255mg/dl) and currently child is on fenofibrate and omega 3 fatty acids and fat-free formula to provide daily energy requirements through the provision of carbohydrates and protein, resulting in a gradual fall in plasma lipid levels over the next few months with normal growth and development.



Genetic Test Report

Gross Lipaemia In A Blood Sample Collected Prior To Diagnosis

DISCUSSION

Primary disorders of lipid metabolism causing hypertriglyceridemia result from genetic defects in triglyceride synthesis and metabolism. With the exception of lipoprotein lipase deficiency, these primary disorders of hypertriglyceridemia usually present in adulthood. Each of the causes of hypertriglyceridemia is associated with an increased risk to develop recurrent pancreatitis and some may increase the risk of premature cardiovascular disease. 7 Most individuals with LPL deficiency present in the first decade of life.8 Only 25% of individuals with LPL deficiency present in the first year of life, and neonatal presentation a rare event.8

CONCLUSION

Our case illustrates a rare disorder that was timely diagnosed and managed that saved the life of a child. Due to the rarity of the disease, there is insufficient data regarding treatment protocol for familial hyperlipidemia syndrome which can present with acute pancreatitis. Hence if a patient presents with acute abdomen and hypertriglyceridemia , congenital hyperlipidemia syndromes must be ruled out as it is a reversible and manageable condition with dietary restrictions, special formula diet, lipid-lowering agents, and plasmapheresis.

Competing Interests: None declared.
Patient Consent: Obtained.

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