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A TYPICAL CASE OF LIPOPROTEIN LIPASE (LPL) DEFICIENCY: A CASE REPORT

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ABSTRACT
Lipoprotein lipase deficiency is a genetic disorder with an autosomal recessive pattern of inheritance. It mostly presents in childhood and is characterized by severe chylomicronemia and hypertriglyceridemia. Lipoprotein lipase deficiency is a rare disorder. Its prevalence is approximately 1 in 1,000,000 in the general population. Lipoprotein lipase deficiency usually presents with Abdominal pain, xanthomas, loss of appetite, nausea, vomiting, arthralgia, and myalgia. Here we present a case of a 39-year-old man, who presented with a slightly different presentation of Lipoprotein lipase deficiency.

KEYWORDS: Lipoprotein Lipase deficiency, LPL deficiency, pancreatitis, Abdominal pain.

INTRODUCTION
Lipoprotein lipase deficiency is a genetic disorder with an autosomal recessive pattern of inheritance. It mostly presents in childhood and is characterized by severe chylomicronemia and hypertriglyceridemia.[1] Lipoprotein lipase deficiency occurs due to the presence of the defective gene for lipoprotein lipase that leads to the reduction or complete absence of lipoprotein lipase enzyme activity. Nonsense mutations, pathogenic deletions, and splice-site variants lead to the formation of an abnormal Lipoprotein Lipase gene product that leads to absent or curtailed enzyme with a reduced or absent catalytic activity.[2]

Lipoprotein lipase deficiency is a rare disorder. Its prevalence is approximately 1 in 1,000,000 in the general population. A reduction or elimination of lipoprotein lipase enzyme activity prevents the breakdown of triglycerides. Therefore, there is an accumulation of triglycerides in the blood and tissues, leading to the clinical manifestations of lipoprotein lipase deficiency.[1] In homozygous individuals, the serum triglyceride levels may reach 10,000 mg/dL or higher. In heterozygous individuals, the serum triglyceride levels may be up to 750 mg/dL.[2] Here we present a case of a 39-year-old man, who presented with a slightly different presentation of Lipoprotein lipase deficiency.

Case presentation
Here we present a case of a 39-year-old male patient, Married with one child. He presented with complaints of multiple episodes of pancreatitis and lipoprotein lipase deficiency and has now developed complications. Birth history was insignificant, and he was born at full term without complications and his family history is unremarkable with no previous or present diagnosed cases of lipoprotein deficiency. There was also no family history of hypertriglyceridemia, pancreatitis, or consanguinity. The patient sometimes complained of abdominal pain in the past but was treated symptomatically. There was no known drug allergy or history of smoking. The history of previous surgeries was insignificant.

He was diagnosed with lipoprotein deficiency at the age of 27 years in 2009. He was diagnosed at that time due to a severe episode of pancreatitis. Since then he has had recurrent episodes of pancreatitis with an interval of 2-3 months. He had five episodes of pancreatitis alone in 2016. After four of these acute pancreatitis episodes, he developed pseudocyst as well. His TAG and LDL levels are high despite mediation compliance and proper diet according to the chart. He was treated with statins, niacin, eicosapentaenoic acid and docosahexaenoic acid, and nizatidine. He was also instructed to adhere to a low-fat diet with 30 g of fat daily and subsequently to take

only 20 g of dietary fat daily because he continued to develop acute pancreatitis. both of which did not appear to affect his clinical or biochemical course

Now presented with central chest pain which was sudden in onset, severe in intensity, non-radiating, and relieved by medication. The pain was also associated with sweating and palpitation. There was no history of dyspnea, RUQ pain, constipation, diarrhea, melena. The Department of medicine ruled out any cardiac pathology and was referred to the gastroenterology department. Biochemical tests confirmed acute on chronic pancreatitis. His amylase and lipase were elevated. His TAG and cholesterol levels were 3950 and 444. These were sky-rocket high. In this admission, he was also found to be diabetic and insulin was started according to the sliding scale. On examination, he did not have hepatosplenomegaly, eruptive xanthomas, or lipemia retinalis. No clinical features of lipodystrophy were noted.

CT scan confirmed acute on chronic pancreatitis. CT Abdomen showed areas in pancreases with hypoechoic parenchyma with fuzzy margins and surrounding echogenic fat. This time his acute pancreatitis presented differently with no epigastric pain. Cardiac causes were ruled out. During this admission, he was diagnosed with diabetes, chronic pancreatitis, and lipoprotein lipase deficiency that was resistant to a strict diet and medical compliance.

DISCUSSION

The majority of cases of lipoprotein lipase deficiency are diagnosed in childhood, usually, before ten years of age, and one-quarter of the individuals are diagnosed in the first year of life. However, some individuals may not develop symptoms until adulthood, like women may present for the first time during pregnancy.[3] In our case patient was diagnosed for the first time at the age of 29 years, which is pretty late for lipoprotein lipase deficiency.

Lipoprotein lipase deficiency usually presents with Abdominal pain, xanthomas, loss of appetite, nausea, vomiting, arthralgia, and myalgia.

Abdominal pain is the most common presentation and occurs due to acute pancreatitis. The attacks of acute pancreatitis are recurrent and usually culminate in chronic pancreatitis.[4] Xanthomas occur in about 50 % of individuals with lipoprotein lipase deficiency. Eruptive xanthomas are yellow papules of around 1 mm in size, appear mostly on the trunk, knees, buttocks, and extensor surfaces of arms. These may coalesce to form larger patches. Our patient did not have xanthomas even though the level of TAG was very high.[1]

The patient presentation in our case report is slightly different. Where, patients with LPL deficiency present with a wide variety of symptoms, this patient just had abdominal pain and on presentation to our facility that too with chest pain. Our patient also didn't complain of Loss of appetite, Nausea, vomiting, Arthralgia, Myalgia.

Hepatomegaly and splenomegaly occur due to highly elevated levels of plasma triglyceride. The excessive chylomicrons in the bloodstream are ingested by macrophages(phagocytes), which then travel to the liver and spleen. The fatty cells accumulate in the liver and spleen, leading to an increase in their size but our patient didn't have any hepato-splenomegaly.[2]

Retinalis lipemia is when the retinal arterioles, venules, and fundus appear pale pink as a result of scattering of light by the large chylomicrons. Retinalis lipemia is seen when the plasma triglyceride levels rise above 2500 mg/dL. These findings were not appreciated in our case.

In an individual with lipoprotein lipase deficiency, recurrent attacks of acute pancreatitis lead to chronic pancreatitis. The secondary complications of chronic pancreatitis are diabetes mellitus, Steatorrhea, and pancreatic calcifications. These complications are rare in an individual with lipoprotein lipase deficiency. Even if these complications occur, they are rare before mid-age.[2] However, these findings were appreciated in our patient.

CONCLUSION

Lipoprotein lipase deficiency is a severe problem that mostly present in childhood. But atypical presentations do exist as in this case presentations. It is very important to test for LPL deficiency even if the patient present in later years. Another important thing is that acute pancreatitis can present as chest pain and it's very important to consider acute pancreatitis if cardiac pathology is ruled out in chest pain.

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