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Case Reports

Recurrent Acute Pancreatitis Secondary to Lipoprotein Lipase Deficiency

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Familial lipoprotein lipase (LPL) deficiency is acquired by autosomal recessive inheritance and the prevalence is 1 per 100000 population. It usually presents in childhood and is characterized by severe hypertriglyceridemia with episodes of abdominal pain, recurrent acute pancreatitis, eruptive cutaneous xanthomata, and hepatosplenomegaly. These patients have defective clearance of chylomicrons from the plasma and their plasma tends to be lipemic. LPL deficiency has no sex predilection and one-fourth of the affected children develop symptoms before one year of age and the majority develop symptoms before 10 years of age. The severity of symptoms correlates with the degree of chylomicronemia.[1] We present an interesting case of this rare genetic disorder.

Case Report

A five-year-old female child, firstborn of a non-consanguineous marriage, presented with severe epigastric pain, radiating to the back for 2 days. This pain increased on the consumption of food and liquids and was associated with repeated episodes of non-bilious, non-projectile vomiting. She had not passed flatus or motions for 2 days. The parents gave a history of three similar episodes in the past. In each episode, she had improved after intravenous medications and taking clear liquids for 2-3 days. On examination, the child was malnourished. Height was between 25-50 centile for age and weight was below 5 centiles for age. There was epigastric tenderness and sluggish bowel sounds were noted.

Baseline laboratory investigations showed haemoglobin 9.4 gm/dl, total leucocyte count 9600/ cu.mm and platelet count of 3.5 lacs/ cu.mm. Serum amylase levels were 235 IU. Liver biochemical tests and serum creatinine were normal. Ultrasound abdomen showed bulky tail of pancreas and normal gall bladder. A contrast-enhanced computed tomography scan of the abdomen showed features of acute edematous pancreatitis with a CT severity index of 4. Thus, a diagnosis of acute pancreatitis was made. Etiological work up for aetiology of acute pancreatitis was done – serum calcium was normal and lipid profile showed highly increased levels of triglycerides (6235 mg/dl) with normal levels of other fractions (LDL, VLDL, HDL and total cholesterol). Both parents had normal lipid profiles, fasting blood sugar levels and ultrasound of the abdomen. Genetic workup of the child detected a proband mutation at chromosome 8p21. Both parents were detected to be obligate heterozygotes and thus, autosomal recessive inheritance was confirmed.

The child was started on atorvastatin and fenofibrate. A low fat (10 gm/day), high protein diet was recommended. On follow up, the triglyceride level reduced to 1765 mg/dl. Later, the child had two further episodes of pancreatitis within 12 months of the first documented episode, which were associated with the stoppage of drugs and dietary restrictions. During both episodes, the triglyceride levels were detected to be 4345 mg/ dl and 2348 mg/dl respectively. After the management of each episode, the need for continuous medication use and proper diet was reemphasized to the parents. Currently, the child is symptom-free for the last 12 months.

Discussion

Patients with LPL deficiency can lead a fairly normal life on a diet very low in total fat content. The secondary complications of pancreatitis – diabetes mellitus, steatorrhea, and pancreatic calcification – are unusual in these individuals and rarely occur before middle age.[2] Lipemia retinalis and reversible neuropsychiatric findings like mild dementia, depression, and memory loss, have also been reported with chylomicronemia.[3,4] The diagnosis is established bythe identification of proband on molecular genetic testing. Management is based on medical nutrition therapy to maintain plasma triglyceride concentration below 1000 mg/dL. Maintenance of triglyceride levels below 2000 mg/dL prevents recurrent abdominal pain. Restriction of dietary fat to ≤20 g/day or 15% of a total energy intake is usually sufficient to reduce plasma triglyceride concentration.

On follow up, regular monitoring of triglyceride levels is required. Agents known to increase endogenous triglyceride concentration such as alcohol, oral estrogens, diuretics, isotretinoin, glucocorticoids, selective serotonin reuptake inhibitors, fish oil supplements and beta-adrenergic blocking agents need to be avoided. Secondary causes of hypertriglyceridemia include diabetes mellitus; paraproteinemia and lymphoproliferative disorders; use of alcohol; and therapy with estrogen, glucocorticoids, selective serotonin reuptake inhibitors, atypical antipsychotic agents, isotretinoin, or certain antihypertensive agents. Other than LPL deficiency, the chylomicronemia syndrome may be caused by biallelic pathogenic variants in apolipoprotein C-II (APOC2), apolipoprotein A-V (APOA5), lipase maturation factor 1 (LMF1) or GPIHBP1. Gene therapy is available in the west[5,6,7,8]. Other treatment modalities like plasmapheresis and antioxidant therapy have been studied but do not appear to be needed for either acute therapy or long-term care.

Conclusion

Hypertriglyceridemia is a rare cause of pancreatitis in children and lipoprotein lipase deficiency should be considered in any child with severely raised triglyceride levels. Management with drugs can result in lowering triglyceride levels and sustained symptomatic relief.

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