CASE REPORT

A Case of Massive Hypertriglyceridemia in 6 Year Old Boy

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Abstract Type V hypertriglyceridemia in children is a rare condition since it has often been associated with obesity, type II diabetes, metabolic syndrome and hormone therapy. We encountered a case of massive hypertriglyceridemia (1900 mg/dl) in a 6 years old boy with complains of acute pancreatitis but no physical manifestations. There was no family history of sudden cardiac death, father and younger male sibling were found to be normal. The mother however had hypertriglyceridemia. The child was managed by dietary changes, omega 3 capsules and low dose fenofibrate. On follow up showed there was reduction in lipid profile and lipoprotein electrophoretic pattern showed bands for VLDL and chylomicron indicating type V hyperlipidemia. Early diagnosis via screening for lipid profile of such patients and their family can improve prognosis and quality of life of these children.

Keywords Hyperlipidemia · Chylomicronemia · Pancreatitis · Dyslipoproteinemia

Background

There have been multiple studies revealing that the early stages of atherosclerosis begin in childhood [1] and premature anticipation of cardiovascular disease can help manage and prevent atherosclerosis. [2]. Hypertriglyceridemia refers to a fasting plasma triglyceride measurement that is increased, typically above the 95th percentile

for age and sex [3]. Hyperlipidemia in children is usually secondary to obesity, hypothyroidism, diabetes and nephrotic syndrome. Primary causes of hypertriglyceridemia (familial hypertriglyceridemia) is a rare condition observed in Fredrickson's hyperlipidemia type I and type V. It is a rare disorder in childhood. According to the National Cholesterol Education Program (NCEP), normal triglyceride level is less than 150 mg/dl (less than 1.7 mmol/L). Primary hypertriglyceridemia is a heritable defect in lipoprotein metabolism leading to defective clearance of TG rich lipoproteins. Familial chylomicronemia syndrome (FCS) is mostly seen in infants and children under the age of 10 years. This is a disorder due to familial lipoprotein lipase (LPL) or apolipoprotein C-II deficiency (Apo C-II) or the presence of inhibitors to lipoprotein lipase. It is a very rare autosomal recessive disease with prevalence of approximately 1 in 1 million for homozygotes. Familial type V hypertriglyceridemia is also known as mixed hyperlipoproteinemia familial or mixed hyperlipidemia. It is similar to type I, but with high VLDL in addition to chylomicrons. It is manifested in adulthood (1 in 600) usually associated with diabetes, metabolic syndrome, alcoholism, hormone therapy etc. This disorder is not usually expressed in children, but there are few reports of affected pre-adolescents [4].

Case

A 6 years old male child came to the outpatient clinic with complaints of recurrent bouts of abdominal pain, followed by vomiting which was non-projectile, non-bilious, containing food particles. The episodes were mostly afebrile, with no altered sensorium or jaundice or dysuria.

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History of Present Illness

The child presented with severe, recurrent bouts of abdominal pain for the first time at the age of 4 years and was diagnosed as pancreatitis due to hyperlipidemia. However, type of hyperlipidemia was not confirmed.

Past Medical History

The child is a product of second degree consanguinity with a younger male sibling. There is no family history of sudden death, premature cardiovascular disease, or recurrent pancreatitis in the family. The parents and sibling were analysed for serum lipid profile which was normal for father and the sibling but the mother was hypertriglyceridemic with triglyceride levels 1178.2 mg/dl. There was no history of drug intake.

Physical and Clinical Examination

On examination the weight was 15 kg (−2 to −3 z score), height- 103 cms (−2 to −3 z score), vitals were normal, there was mild pallor, no icterus, cyanosis, clubbing, lymphadenopathy, edema, or acanthosis nigricans. Per abdomen examination showed no hepatosplenomegaly, rest of the systemic examinations, examination of the cardiovascular system (including blood pressure) and ultrasound abdomen were normal. The blood sample of the child was collected in a clot activator gel vacutainer and was found to be milky/extremely lipemic (Fig. 1). The sample was centrifuged and kept at 2 °C overnight and it showed a creamy

supernatant and a turbid infranatant (Fig. 1). The biochemical investigations were performed (Table 1).

The child was treated for hyperlipidemia and was already on fenofibrate (100 mg BD) when he reported to our outpatient clinic. We ruled out secondary causes of hypertriglyceridemia and advised a low fat diet (<15% of total caloric intake) and Omega 3 fatty acid (500 mg) BD [5]. Dose of fenofibrate was reduced to 1 tab OD. The child was called for a follow up after 2 months and an extended lipid profile done using serum electrophoresis (Fig. 2).

On follow up the child showed a reduction in the serum lipid profile, with the triglyceride levels dropping markedly (Table 1). CK-Nac dropped owing to the reduction in the fenofibrate dosage. The electrophoresis pattern of lipoproteins indicated a type V hyperlipidemia with high VLDL percentage and presence of chylomicrons in the fasting sample.

Discussion

A serum triglyceride (TG) level of more than 1000 mg/dl in patients with type I or V hyperlipidemia (Fredrickson's classification) is an identifiable risk factor for acute pancreatitis [6]. Type V can rarely occur in children [7]. Since the prevalence of type V hyperlipidemia is greater than type I, thus clinically encountered chylomicronemia is more often of this type. Chylomicrons are usually present in the plasma of individuals with TG >1000 mg/dl and hyperchylomicronemia term is used when the TG levels exceed 2000 mg/dl. Physical symptoms appear usually beyond 2000 mg/dl [6].

The child has type V hyperlipidemia—a rare condition in childhood. The mother too was hypertriglyceridemic

Fig. 1 Blood sample pre (a) and post (b) refrigeration

Table 1 Biochemical investigations for the Case X

Analyte	Ist visit	Follow up visit
Random sugar (in mg/dl)	118.00	–
Total cholesterol (150–200 mg/dl)	198.00	133.00
Triglyceride (<150 mg/dl)	1900.00	1337.00
HDL (>40 mg/dl)	21.00	12.00
LDL (<100 mg/dl)	65.00	–146.40
Uric acid (3.5–7.2 mg/dl)	2.20	–
Amylase (28–100 U/L)	123.00	57.00
Lipase (<67 U/L)	341.00	100.00
Calcium (8.8–10.8 mg/dl)	9.58	9.72
Alkaline phosphatase (93–109 U/L)	150.00	180.00
Direct bilirubin (<0.2 mg/dl)	0.10	–
Indirect bilirubin (0.3–1.2 mg/dl)	0.61	–
Asparate transaminase (<50 U/L)	276.00	70.00
Alanine transaminase (<50 U/L)	105.00	20.00
CK-Nac (<145 IU/ml)	5085.00	66.00
FT3 (80–200 ng/dl)	98.15	–
FT4 (5.10–14.10 µg/dl)	7.79	–
TSH (0.60–4.84 µIU/ml)	1.36	–
Apo B (60–140 mg/dl)	–	36.00
Apo A1 (105–175 mg/dl)	–	26.00
Lp(a) (<30 mg/dl)	–	<10.20
Serum electrophoresis		
HDL (15–39%)	–	9.50%
LDL (42.3–69.5%)	–	51%
VLDL (2–31.2%)	–	39.5%
Chylomicron (nil)	–	Present

Fig. 2 Serum lipoprotein electrophoresis showing type V hyperlipidemia

indicating an autosomal dominant inheritance. The hypertriglyceridemia associated acute pancreatitis is a significant manifestation observed in almost 7% of acute pancreatitis

cases. The current recommendations by the Expert Panel on Integrated Guidelines for Cardiovascular Health and Risk Reduction in Children and Adolescents are universal screening in late childhood, selective screening in other age groups with cardiovascular risk factors and additional universal screening between ages 17–21 years [5]. Early diagnosis via screening of such patients and their family can improve prognosis and quality of life of these children.

Conclusion

We conclude that chylomicronemia/type V hyperlipidemia is an important etiological factor of acute pancreatitis in children and Paediatricians should be alert for the possibility of chylomicronemia as a cause of acute pancreatitis.

Compliance with Ethical Standards

Conflict of interest Author Purvi Purohit declares that she has no conflict of interest. Author Daisy Khara declares that she has no conflict of interest. Author Praveen Sharma declares that he has no conflict of interest. Author Kuldeep Singh declares that he has no conflict of interest.

Ethical Approval All procedures performed in studies involving human participants were in accordance with the ethical standards of the institutional and/or national research committee and with the 1964 Helsinki declaration and its later amendments or comparable ethical standards.

Informed Consent Informed consent was obtained from the parents of the participant included in the study.

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