CASE REPORT

AN UNUSUAL CASE OF FAMILIAL HYPERLIPIDAEMIA

Renu Nagar and Uma Arora*

Department of Biochemistry, Geetanjali Medical College, Udaipur and *Christian Medical College, Ludhiana, India

ABSTRACT

A 40 days old male baby born to a consanguineous couple was found to have highly viscous and milky serum with caking of chylomicrons on refrigeration of serum. Cholesterol was 889.5 mg/dl (23.04mmol/L) and Triglycerides 12881mg/dl (141.69mmol/L). He was active and did not have any hepatosplenomegaly, xanthomas or dysmorphic features. Thyroid functions were normal. Lipid electrophoresis showed thick chylomicron band. There was positive family history of hypertriglyceridemia in a first cousin. Both siblings and both parents of the index case had normal lipid profiles. This child was referred to higher centre where he was put on Lipid lowering drugs (Gemfibrozil), Iron drops and special formula for feeding containing medium chain fatty acids.

KEY WORDS

Hypertriglyceridemia, Xanthomas, Lipid lowering drugs.

CASE REPORT

A 40 days old male baby born to a consanguineous couple was admitted in NICU of a Children Hospital in Saudi Arabia with mild febrile illness to rule out sepsis.

The baby was youngest of 3 siblings, was born at term by normal vaginal delivery with a birth weight of 3.3 kg and was being exclusively fed breast milk. During phlebotomy, his blood was found to be highly viscous and milky. Serum was thick and milky with caking of chylomicrons on refrigeration. Serum was analyzed using serial on board dilutions on Cobas Integra 400 and following results were obtained

A second sample was received for the same baby after keeping him on intravenous fluids and no oral intake for 5 days.

Cholesterol 406.8 mg/dl(10.54mmol/L)

Triglycerides 6191 mg/dl(68.10mmol/L)

Lipid electrophoresis showed a band of chylomicrons.

Address for Correspondence :

Dr. Renu Nagar,

Department of Biochemistry,

Geetanjali Medical College,

Geetanjali Medicity, Udaipur, India

E-mail: dr_renu_bhardwaj@rediffmail.com

Baby was active, pink and afebrile from 2nd day of admission and did not have any hepatosplenomegaly, xanthomas or dysmorphic features. His blood, urine and stool cultures showed no growth and CSF examination was normal and Thyroid functions were as follows:

TSH 7.8 mIU/ml (Normal range 1-30)

T₄ 21 pmol/L (Normal range 11-17)

There was positive family history of hypertriglyceridemia in a first cousin who presented with hypertriglyceridemia and multiple renal cysts at the age of 1 month. He was found to have normal renal functions and renal cysts which responded to lipid lowering drugs (Gemfibrozil), Niacin and Aspirin.

His cholesterol levels were within normal range but, triglycerides remained high 663.6mg/dl (7.3 mmol/L) even 3 years after treatment.

Both siblings and both parents of the index case had normal lipid profiles. There was no family history of premature myocardial infarction or stroke.

This child was referred to higher centre in Riyadh for further investigations and treatment, where he was put on Lipid lowering drugs (Gemfibrozil), Iron drops and special formula for feeding containing medium chain fatty acids. Gemfibrozil

Table 1 : Patient's Blood Picture on Admission

Parameter	Result	Normal Range	Result in SI Unit	Normal Range
Cholesterol	889.5 mg/dl	115-203mg/dl	23.04mmol/L	2.95-5.25 mmol/L
Triglycerides	12881mg/dl	40-200mg/dl	141.69mmol/L	0.452-2.26 mmol/L
Blood glucose	99.8 mg/dl	60- 100 mg/dl	6.07mmol/L	3.3-5.6mmol/L
Uric acid	2.2 mg/dl	2.2-6.5 mg/dl	0.129mmol/L	0.13-0.39mmol/L
Magnesium	1.7 mg/dl	1.5-2.2 mg/dl	0.699mmol/L	0.62-0.91mmol/L
Calcium	7.97 mg/dl	9.0-11.0 mg/dl	1.99mmol/L	2.25-2.75mmol/L
Phosphate	5.16 mg/dl	4.5-6.7 mg/dl	1.67mmol/L	1.45-2.16mmol/L
Albumin	2.9 gm/dl	3.4-5.0 gm/dl	29 gm/L	34-50 gm/L
Urea	20.11 mg/dl	10.0-40.0mg/dl		
Creatinine	0.7 mg/dl	0.6-1.2mg/dl	61.88umol/L	53-106umol/L
Sodium			139.8mmol/L	135-145 mmol/L
Potassium			4.48mmol/L	3.4-5.5 mmol/L
Chloride			109.4mmol/L	92.0-107.0 mmol/L
TSH	7.8 mU/L	1-30 mU/L		
T ₄	21 pmol/L	11-17 pmol/L		

inhibits lipolysis and reduces hepatic fatty acid uptake. It also inhibits secretion of VLDL from liver.

DISCUSSION

Familial hyperchylomicronemia is a rare autosomal recessive trait. Some cases of familial chylomicronemia result from mutations in the gene encoding lipoprotein lipase or its cofactor apoC-II. Some cases of familial combined hyperlipoproteinemia result from mutations in the upstream transcription factor 1 (*USF1*) gene encoding a transcription factor. Dysbetalipoproteinemia (type III hyperlipoproteinemia) requires 2 copies of the binding defective *APO E E2* allele. Secondary factors that worsen hypertriglyceridemia include poor diet, obesity, alcohol use, diabetes, renal disease, pregnancy, nonalcoholic fatty liver disorder, paraproteinemias, autoimmune disorders and certain medications, such as steroids (1). Presenting symptoms of Lipoprotein lipase deficiency are episodes of abdominal pain, recurrent pancreatitis, xanthomas, hepatosplenomegaly and lipaemic retinalis (2). Most of the patients become symptomatic within first decade of life but patient might be asymptomatic and may be accidentally discovered due to the milky appearance of serum (3). Diagnosis is based on lipaemic appearance of serum, caking of chylomicrons on serum on refrigeration, serum triglycerides more than 11.3mmol/L, raised serum cholesterol, markedly raised chylomicrons(4) and decreased lipoprotein lipase activity (5). Incidence of Familial hyperchylomicronemia is one in a million (6). Lipoprotein electrophoresis is considered superior to overnight standing

of serum to detect chylomicronemia which might be missed if triglycerides are not so high (7). Although in index case that was not a problem.

This baby was accidentally discovered to have hyperchylomicronemia. He was asymptomatic and did not have xanthomas or hepatosplenomegaly; had caking of chylomicrons on refrigeration of serum overnight, raised cholesterol(23.04mmol/L) and alarmingly raised triglycerides(141.69mmol/L).Lipid electrophoresis showed thick chylomicron band. Low fat formula and lipid lowering drugs can dramatically improve the lipid parameters and prevent premature myocardial infarction and death. Family should be given genetic counselling and should undergo prenatal screening of any future baby that they might conceive, as the risk of having an affected baby is 25%.

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