MILKY SERUM: A COINCIDENTAL FINDING

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ABSTRACT

BACKGROUND

Hypertriglyceridemia characterized by elevated serum levels of triglycerides which can be primary (Genetic/familial) or secondary. Primary hypertriglyceridemia includes type 1, 2b, 3, 4 and 5 according to Fredrickson's classification (WHO). We present a case of a 2 yrs. boy with milky serum detected coincidentally. The serum triglycerides were raised and standing plasma test was positive. However, the child did not have the typical signs and symptoms of hypertriglyceridemia. When a patient presents at such an age familial hyperlipidemia should be considered. A provisional diagnosis of type 1 or type 5 hypolipoproteinaemia was given with a need to further evaluate the child.

KEYWORDS

Hypertriglyceridemia, Chylomicrons, Standing Plasma Test, Lipemic, Fredrickson's Classification.

INTRODUCTION

Hypertriglyceridemia is defined as fasting plasma triglyceride levels that is, typically above the 95th percentile for age and sex.¹ ² Higher levels of plasma triglyceride concentrations is associated with increased risk of cardiovascular disease, both directly and indirectly.² Plasma triglycerides have two sources one is exogenous which is carried in chylomicrons and other is endogenous carried in very-low-density lipoprotein (VLDL) particles.³ These lipoproteins and chylomicrons are hydrolysed by lipoprotein lipase into free fatty acids in capillaries of adipose and muscle tissues.³ Hypolipoproteinaemia is classified into five types by WHO (Fredrickson's classification) based on serum lipid concentration, appearance of serum after centrifugation and lipoprotein electrophoresis. According to this classification Type I is characterised by hyperchylomicronaemia and hypertriglyceridemia, confirmed by lipoprotein lipase deficiency; and Type V by higher levels of VLDL, chylomicrons, cholesterol and triglycerides.¹ ² In children, high levels of triglyceride are usually secondary to conditions such as obesity and type 2 diabetes mellitus. However, familial hyperlipidemia should be considered when a patient presents at such a young age.⁶ We present a case of a 2 yr. boy with milky serum which we had come across in our laboratory.

CASE REPORT

A 2 yr. male child was brought by his mother to the orthopaedics outpatient department with c/o instability while walking. The following investigations X-ray of both legs and blood tests namely serum calcium, phosphorous and alkaline phosphatase were ordered. X ray report was normal.

The serum sample sent to the biochemistry laboratory for analysis was found to be lipemic (Fig.1). Lipid profile was done as the sample was lipemic, which showed elevated levels of serum triglycerides (Table 1). Standing plasma test was positive thus, showing the presence of chylomicrons (Fig.2).

On further evaluation of the child it was found that, he is the only child of his parents born out of non-consanguineous marriage, belonging to lower socioeconomic status. The father gave a history that at the time of birth the doctor had informed him that his child's serum was lipemic. There were no signs of hyperlipidemia like xanthoma etc. Serum triglycerides of both parents were within reference range (Table 1). Dietary restriction of fats was advised and patient was asked to review again for further workup.

<table>
<thead>
<tr>
<th>Lipid Profile</th>
<th>Father</th>
<th>Mother</th>
<th>Child</th>
</tr>
</thead>
<tbody>
<tr>
<td>Serum Total Cholesterol (mg/dl)</td>
<td>136</td>
<td>206</td>
<td>124</td>
</tr>
<tr>
<td>Serum HDL-Cholesterol (mg/dl)</td>
<td>22</td>
<td>48</td>
<td>33</td>
</tr>
<tr>
<td>Serum Triglycerides (mg/dl)</td>
<td>129</td>
<td>83</td>
<td>370</td>
</tr>
<tr>
<td>Serum VLDL-Cholesterol (mg/dl)</td>
<td>26</td>
<td>17</td>
<td>74</td>
</tr>
<tr>
<td>Serum LDL-Cholesterol (mg/dl)</td>
<td>88</td>
<td>141</td>
<td>17</td>
</tr>
</tbody>
</table>

Table 1: Fasting lipid profile of the child and his parents.

Fig. 1: Patient's lipemic serum sample
DISCUSSION
Primary hypertriglyceridemia includes familial chylomicronemia (Type 1), familial combined hyperlipoproteinaemia (Type 2b) familial dysbetalipoproteinaemia (Type 3), familial hypertriglyceridaemia (Type 4) and primary mixed hyperlipidemia (Type 5). In the present case a provisional diagnosis of either type 1 or type 5 hyperlipidemia was given as, the serum was lipemic, development of a creamy supernatant layer when sample was refrigerated overnight (presence of chylomicrons) and elevated levels of serum triglycerides.

Both type 1 and 5 have common clinical features of eruptive xanthomata, lipemia retinalis, enlargement of liver and spleen, focal neurological deficits and recurrent pain abdomen with risk of pancreatitis. Although, in this case the child needs to be evaluated further. Standing plasma test positive and elevation of serum triglycerides are also common to both types.

While the differences between type 1 and 5 include presentation during childhood in type 1 and in adulthood in type 5. There is a deficiency of lipoprotein lipase, apo CII activity or homozgous gene mutations in type 1 while a less severe functional deficiency in type 5. The prevalence of type 1 (1:10^9) being much lower than type 5 (1:10^9). Secondary factors are associated with type 5. Total cholesterol is elevated to a greater extent in type 5 relative to that in type 1. The presence of markedly increased triglyceride concentrations should prompt consideration of an inherited condition either detected by inspection of milky appearing serum or during routine screening. Basic treatment is dietary restriction of fats to 20g/day or less or to 15% or less of the total energy intake. Medical treatment with drugs like fibrates niacin is rarely indicated.

Lipemic serum was a purely coincidental finding in this case. The serum triglycerides were elevated but, not to such an extent so as to cause the appearance of clinical manifestations. Dietary restriction of fats will prevent elevation of serum triglycerides further, thereby preventing complications of hypertriglyceridaemia like pancreatitis, cardiovascular disease. Thus, genetic diagnosis of hyperlipidaemias and gene based therapies should be made easily available so that, these patients can be managed more effectively.

REFERENCES