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October 2008

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## Recommended Citation

Lone, S., Imdad, A., Billoo, A. (2008). Familial chylomicronemia in a nine months old infant. *Journal of the College of Physicians and Surgeons Pakistan*, 18(10), 655-6.

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# Familial Chylomicronemia in a Nine Months Old Infant

Saira Waqar Lone, Aamer Imdad and Abdul Gaffar Billoo

## ABSTRACT

Familial chylomicronemia syndrome is a rare disorder of lipoprotein metabolism due to familial lipoprotein lipase or apolipoprotein C-II deficiency or the presence of inhibitors to lipoprotein lipase. It manifests as eruptive xanthomas, acute pancreatitis, and lipaemic plasma due to marked elevation of triglyceride and chylomicrons levels. We report a rare case of familial chylomicronemia in a 9-month-old infant, who was diagnosed after his plasma was incidentally found to be milky. Lipid profile showed familial chylomicronemia (Type 1 Hyperlipidemia). The infant was started on a low fat diet and advised a regular follow-up.

**Key words:** *Familial chylomicronemia. Eruptive xanthomas. Acute pancreatitis.*

## INTRODUCTION

Dyslipidemia refers to the elevation of plasma cholesterol and/or triglycerides or a low HDL level that contributes to the development of atherosclerosis and they can be primary or secondary.<sup>1</sup> Primary disorders are genetically transmitted and are the common cause in children.<sup>2</sup> Secondary causes contribute to most cases of dyslipidemia in adults and are due to sedentary life-style with excessive intake of saturated fats, cholesterol and trans fatty acids. Dyslipidemia itself causes no symptoms but can lead to coronary artery disease and peripheral arterial disease. Familial chylomicronemia is a rare genetic disorder of lipoprotein metabolism with an incidence of one per million in the general population.<sup>3</sup> Clinically, this condition can be silent and discovered incidentally owing to the lipemic appearance of the blood.

Very scarce literature and data is available on hypertriglyceridemia in children internationally. We report a rare case of familial chylomicronemia in a 9-month-old infant.

## CASE REPORT

A 9-month-old male child immunized, weighing 7.5 kg, came to the Outpatient Department with complaints of abdominal distention and constipation off and on since the past 7 months. The boy was the product of a consanguineous marriage, born to a primigravida mother. Birth history was unremarkable and he had been breast fed after birth. At 2 months of age, the baby

developed fever, abdominal pain and constipation. For these complaints, the baby was shown to a private hospital and was advised some laboratory tests. When blood was drawn for laboratory tests, it appeared milky in color. The 8 hours fasting lipid profile of the baby was done, which showed a total lipids of 640 mg/dl (normal range= 500– 1000 mg/dl), triglycerides at 430 mg/dl (normal range= 46–236 mg/dl), cholesterol level of 43 mg/dl (normal range= 160-200 mg/dl) and HDL level of 7 mg/dl (normal range= 35 mg/dl). The baby was put on skimmed milk. He remained well except for off and on complaints of constipation and abdominal distension, for which he presented to this OPD.

On examination, he was found to be an active boy and his general physical and systemic examination including fundoscopy was unremarkable. The lipid profile and electrophoresis were repeated, the triglycerides and VLDL were elevated, cholesterol at 128 mg/dl, triglycerides at 692 mg/dl, HDL of 20 mg/dl, LDL of 40 mg/dl and VLDL of 138 mg/dl (normal < 30 mg/dl). The lipoprotein electrophoresis showed elevated chylomicrons and pre-beta lipoprotein. The serum chylomicrons level was 9.8 REL % (normal range= 0-2.0), beta lipoprotein of 41.7 REL % (normal range= 39.2-65.2), pre-beta lipoprotein of 36.7 REL % (normal range= 3-30.2) and alpha lipoprotein level of 11.9 REL % (normal range = 16.7-45.7). On the basis of those results, the diagnosis of familial chylomicronemia (Type 1 hyperlipidemia) was made; the infant was started on a low fat diet and advised a regular follow-up. The lipid profile of the father and mother was also done, which were normal.

On his last follow-up, he was one and-a-half years old; complaints of abdominal distention and constipation had improved and triglycerides were also decreasing.

## DISCUSSION

Familial chylomicronemia is an autosomal recessive disorder. Familial lipoprotein lipase deficiency usually

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Received May 5, 2008; accepted September 9, 2008.

manifests by 10 years of age, and in 25% occurs during infancy. Often, this condition is silent and the initial clue to the diagnosis is the presence of lipemic plasma. It manifests during childhood with lipemia retinalis, hepatosplenomegaly, irritability and recurrent epigastric pain with increased risk of pancreatitis. It is characterized by the pathologic presence of chylomicrons after a 12-14 hours period of fasting.<sup>4</sup> Acute relapsing pancreatitis is the most significant and often life-threatening complication.

Eruptive xanthomas present as asymptomatic, evanescent, yellowish papules over buttocks, shoulders and extensors of limbs when the serum triglycerides exceed 2000 mg/dl.<sup>5</sup> In the present case, the triglyceride level was initially 432 mg/dL but had risen to 692 mg/dL. There was no family history of hypertriglyceridemia and the lipid profile of parents were normal. Lipoprotein lipase or Apo C-II deficiency could not be performed as these tests are done at specialized lipid centers, not available in Pakistan.

Dietary modification plays a key role in the management of this disease. Lowering elevated triglyceride level is beneficial and levels less than 150 mg/dl are desirable. Dietary fat restriction supplemented by fat soluble vitamins and including fish oils may also be beneficial. The intake of saturated fats and trans fatty acids should be reduced and replaced by polyunsaturated and mono-unsaturated fats.<sup>6</sup> Dietary fat should be restricted to less than 20 g/day and 15% of the total caloric intake so that triglyceride levels are maintained below 1500 mg/dL. Medium-chain triglycerides are the preferred source of dietary fat.

Intake of fish high in omega-3 fatty acids may be effective and vegetables can replace animal proteins.<sup>7</sup> The complications in these are due to an increase intake

in the amount of fat, which causes recurrent bouts of illness leading to formation of cysts, hemorrhage and death. In follow-up of these children, lipid levels should be monitored periodically after starting treatment. Dietary counselling and genetic counselling of the family was advised.<sup>8,9</sup>

Literature and case reports on hypertriglyceridemia in pediatric age group is very scarce and there are no specific treatment guidelines in children. Therefore, there is a need for consensus suggestions on the management of these children.

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