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Infant with milky blood: A rare case of familial chylomicronemia presenting with infantile eruptive xanthoma

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Hypertriglyceridemia with significant elevation in triglycerides are becoming increasingly common in American children who are obese and have sedentary lifestyle. But genetic disorders, which cause hypertriglyceridemia in infants, are very rarely reported. The latter defect can be induced by an abnormality either in the lipoprotein itself, Lipoprotein lipase deficiency or lipoprotein receptor defect. Here we report a 6 weeks old female infant of Middle Eastern decent who presented with fever, cough and nasal congestion for 1 week. Physical examination was significant only for eruptive Xanthoma present around the right eyes. During blood draw for sepsis screen, patient noted to be having pink milky blood. Baby was evaluated for sepsis and acquired causes for hyperlipidemia as the baby had Eruptive Xanthomas. Though the sepsis screen was not significant, the lipid profile was alarming with very high cholesterol of 975 mg/dl (normal value: 120-200 mg/dl) and triglycerides 1580mg/dl (normal value: 40-150 mg/dl), HDL 6 mg/dl (normal value 60-80 mg/dl). Ophthalmology examination was significant for Lipemia Retinalis. Ultrasound abdomen done in view of pancreatitis due to hypertriglyceridemia was reported normal. EKG and Echo were also normal. Further evaluation of the parents and siblings revealed that the father and the older sister who is 6 years old also had pink milky blood with increase in cholesterol and triglycerides, which was undiagnosed. Due to the presence of Infantile Xanthomas, very high cholesterol, hypertriglyceridemia and familial presentation, further evaluation of genetic causes for hyperlipidemia was done. Genetic analysis showed a T108R mutation in GPIHBPI gene suggestive of Chylomicronemia due to LPL deficiency. Initially mother was advised exclusive breastfeeding with modification in her diet. This induced as sharp increase in triglycerides. Thus breastfeeding was discontinued. Baby was started on special formula with medium chain triglycerides oil. After 8 months, there has been a substantial decrease in the cholesterol to 145 mg/dl and triglyceride to 812 mg/dl without administering any lipid lowering medications. The Eruptive Xanthoma spontaneously resolved as the cholesterol and triglycerides showed decreasing trend. The child continues to closely follow with cardiologist, gastroenterologist and the lipid clinic as Chylomicronemia is associated with increased incidence of premature coronary vascular disease, pancreatitis and death. This case is being presented because of its uncommon presentation in infant. The use of lipid lowering medications in infants has not been studied because of rarity and hence much data is not available about treating familial hypercholesterolemia and hypertriglyceridemia in infants. The photos of the patient cannot be published, as parents did not consent for the photo due to religious reasons. Biopsy of the Cutaneous Xanthoma could not be performed due to cosmetic reasons as lesions were very near to the eyes. The photo of the milky blood is attached.

Biography

Soumya Nagaraja is a board certified Dermatologist with MD (Dermatology, Venereology, Leprosy) from India. Her area of interest is Pediatric Dermatology. Her research has been in the area of psychoneuroimmunology in chronic dermatological disease like psoriasis. She was working as Dermatologist and Cosmetologist in Bangalore, India before moving to USA. Currently she is pursuing MD (pediatrics) at Brookdale University Hospital and Medical Center, New York. Her current area of research has been on "Evolution of clinical and lab parameters before and after IV immunoglobulin therapy" in Kawasaki disease patients. She has also been doing her electives in Allergy and Immunology as she plans to pursue her career also as Researcher with special interest in Immune Mediated Dermatological Disorders.

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