Net Letter

Chylomicronemia syndrome

Sir,

Chylomicronemia syndrome results from deficiency of lipoprotein lipase and Apo-C-2. Chylomicronemia may also occur in sporadic hypertriglyceridemia. Familial lipoprotein lipase deficiency is a rare autosomal recessive disorder that results from a deficiency of the extrahepatic lipoprotein lipase. Reported incidence is one in one million.[1] Lipoprotein lipase enzyme acts on chylomicrons to form free fatty acids and remnant particles. The enzyme is responsible for hydrolysis and removal of chylomicrons and very low density lipoprotein (VLDL) triglycerides from the circulation. Deficiency of lipoprotein lipase results in accumulation of chylomicrons in the plasma.[1] The condition presents in childhood, with recurrent abdominal pain or acute relapsing pancreatitis. Eruptive xanthomas, hepatosplenomegaly and retinopathy are frequent findings. The major threat to the health of these patients appears to be acute pancreatitis. Familial Apo-C-2 deficiency is a very rare autosomal recessive disease that results in an absence of normal Apo-C-2, an essential cofactor for lipoprotein lipase. As a consequence of this defect, there is a functional lipoprotein lipase deficiency because lipoprotein lipase cannot hydrolyze chylomicrons or VLDL in the absence of normal Apo-C-2. This defect in lipolysis causes a rise in both chylomicrons and VLDL. These patients usually present in adult life and do not show hepatosplenomegaly or eruptive xanthoma. Abdominal pain due to pancreatitis, however, remains a major threat to health. Plasma triglyceride levels are markedly elevated.

A 1-month-old female baby born to non-consanguineous parents was admitted to the pediatric ward of Gandhi Hospital for abdominal distension, refusal of feeds and decreased urinary output of 1-day duration. She was provisionally diagnosed as a case of sepsis with acute renal failure. She was referred to the dermatology department for evaluation of papular eruption involving the scalp, face and upper and lower limbs. There was no positive family history of similar eruption. General examination revealed that the neonate was febrile and dehydrated. Cutaneous examination revealed yellowish papules distributed on the scalp, face, upper limbs and thighs [Figures 1 and 2]. The papules were firm, discrete and non-tender. Some of the papules showed erythema around them.

Funduscopy revealed lipemia retinalis. She was provisionally diagnosed as a case of eruptive xanthomas and was investigated to establish the type of hyperlipidemia. The blood was found to be lipemic when it was drawn to carry out investigations. Serum

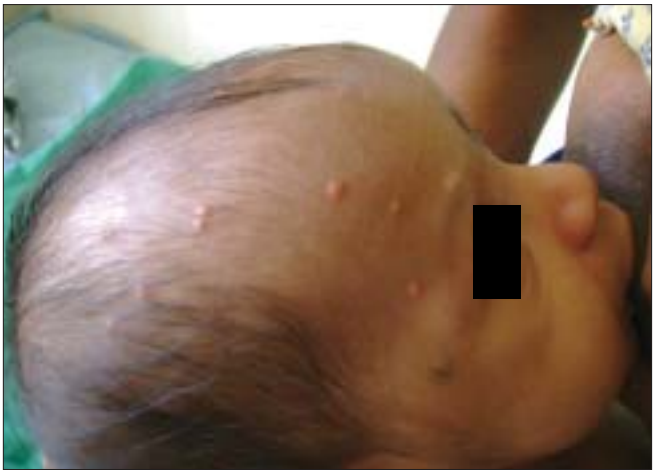

Figure 1: Scalp and forehead showing yellowish papules

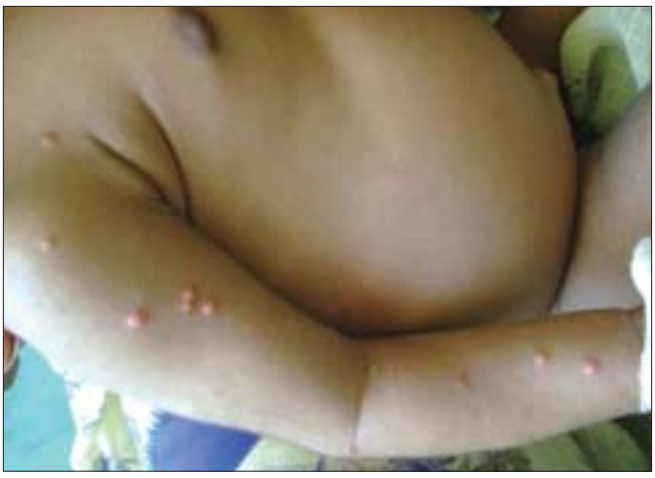

Figure 2: Right upper limb showing yellowish papules

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lipid profile revealed increased serum triglycerides 2050 mg% (normal, up to 160 mg%), serum cholesterol 828 mg% (normal, 150–250 mg%), blood sugar 70 mg/dL, blood urea 211 mg/dL and serum creatinine 2 mg/dL. Liver function tests were normal. Human immunodeficiency virus I and II were non-reactive. T_3, T_4 and thyroid stimulating hormone (TSH) were normal. Parent's serum lipid profile were within normal limits. Skin biopsy showed hyperkeratotic, acanthotic epidermis with follicular plugging. Dermis showed loose edematous fibrocollagen tissue along with lymphocytes, polymorphs and histiocytes with scattered lymphocytic infiltrate. Microscopy using polarized lens at 40X showed refractile needle-like cholesterol granules. Serum lipoprotein electrophoresis showed increased chylomicrons, reduced alpha bands and normal beta and pre-beta bands, suggestive of Fredrickson's type 5. Because of the lack of facility, chylomicron levels and post-heparin lipoprotein lipase and Apo-C-2 estimation could not be carried out. These can be performed only in specialized lipid centers.

Lipoprotein lipase enzyme acts on chylomicrons to form free fatty acids and remnant particles. The gene for lipoprotein lipase is located on chromosome 8. More than 30 structural defects in the gene have been reported to result in lipoprotein lipase deficiency. Kavarzakis *et al*, while conducting genetic study in Chylomicronemia syndrome, found compound heterogenesity for common lipoprotein lipase gene mutation (G188E) and novel missense mutation (M301R).[2] The enzyme is responsible for hydrolysis and removal of chylomicrons and VLDL triglycerides from the circulation. Deficiency of lipoprotein lipase results in accumulation of chylomicrons in the plasma.[1] The severity of the symptoms is proportional to the degree of chylomicronemia. Rarely, neurological abnormalities like memory loss, dementia and peripheral neuropathy have been described. Lipoprotein lipase deficiency does not seem to be associated with atherosclerosis and premature heart disease. The prevailing view has been that the chylomicrons are too large to filter into the arterial wall to initiate atherogenesis.[3] Circulating chylomicrons occurring in this condition are acted upon by pancreatic lipase. The resultant partially hydrolyzed products produce inflammation of the pancreas with further release of pancreatic lipase, thereby producing a vicious cycle.[4] The diagnosis of lipoprotein lipase deficiency can be made based on the finding of absent or low lipoprotein lipase activity in the serum after heparin administration or directly from analysis of the biopsy specimen of adipose tissue. It may also be confirmed by demonstration of structural defects of the lipoprotein lipase gene.[4] The reported our patient was a 30-day-old neonate. Mohandas *et al*,[4] Siafakas *et al*,[5] and Sushamabai *et al*,[6] have all reported similar cases presenting in the age group between 20 and 60 days. Sepsis was the presenting feature in our case. Mohandas *et al*,[4] and Sushamabai *et al*,[6] have also reported a similar presentation in their cases. Our patient developed systemic complication and acute renal failure, from which she recovered completely.

A Gnaneshwar Rao, Chandana Konda, Kamal K. Jhamnani

Department of Dermatology, Gandhi Medical College and Hospital, Musheerabad, Hyderabad, India

Address for correspondence:
Dr. A. Gnaneshwar Rao, Flat 12, Block-8, HIG 2, APHB, Bagh Lingampally, Hyderabad - 500 044, India.
E-mail: dr_angoori@yahoo.co.in

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REFERENCES

1. Sarkany RP, Breathnach SM, Seymour CA, Weismann K, Burns DA. Metabolic and nutritional disorders. In: Burns T, Breathnach S, Cox N, Griffiths C, editors. Rook's textbook of dermatology. 7th ed. Blackwell Science; 2004. p. 57.74.
2. Kavarzakis E, Stabouli S, Gourgiotis D, Roumeliotou C, Traeger-Synidinos J, Bossios A, *et al*. Severe hypertriglyceridemia in a Greek infant: A clinical biochemical and genetic study. Eur J Pediatr 2004;163:462-6.
3. Grundy SM. Xanthomatoses and lipoprotein disorders. In: Freedberg IM, Eisen AZ, Wolff K, Austen FK, Goldsmith LA, Katz SI, *et al*, editors, Fitzpatrick's dermatology in general medicine. 5th ed. McGraw Hill; 1999. p 1808.
4. Mohandas MK, Jemila J, Krishnan AS, Thomas GT. Familial chylomicronemia syndrome. Indian J Pediatr 2005;72:181.
5. Siafakas CG, Brown MR, Miller TL. Neonatal pancreatitis associated with familial lipoprotein lipase deficiency. J Pediatr Gastroenterol Nutr 1999;29:95-8.
6. Sushamabai S, Subrahmanian D. Lipoprotein lipase deficiency. Indian Pediatr 1997;34:1047.