cephalic tetanus [4]. Cephalic tetanus usually follows middle ear infections like suppurative otitis media, as in our case or craniofacial injuries [1]. Such otogenic tetanus are common in the pediatric age group which may be explained by the immune status and frequency of middle ear infections. This case was rare in its type as it presented with isolated ptosis without any other cranial nerve involvement, unlike the cephalic tetanus reported earlier with trismus, ptosis and facial palsy mimicking Bell's palsy. Around 2/3rd patients with cephalic tetanus progress to generalized tetanus, which could be a possible reason fo the generalized spasms in this child.

The mechanism of cranial nerve palsies is not fully understood but few studies have given explanations like swelling of facial nerve under the influence of the toxin leading to strangulation in the stylo-mastoid canal, third-nerve lesions due to intense absorption of toxin from the orbicularis and ciliary regions, which are supplied by this nerve.

Survival rates in children receiving tetanus immunoglobulins *via* the dual route were significantly higher compared with children who received the intramuscular immunoglobulin only [5-7] and hence we preferred dual route for TIG administration.

High index of suspicion for tetanus should be considered in an unimmunized child presenting with ptosis without apparent trismus or facial palsy.

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An Infant with Milky Serum and a Rare Mutation

A 40-day-infant having milky serum, eruptive xanthomas, hepatosplenomegaly, lipemia retinalis, high cholesterol and triglyceride, was found to have lipoprotein lipase (LPL) deficiency on genetic workup. Triglyceride decreased with dietary fat restriction, medium chain triglyceride and fibrates. LPL deficiency in early infancy can be treated with pharmacological and dietary interventions.

Keywords: *Hypertriglyceridemia, Lipoprotein lipase, Outcome*

Familial chylomicronemia syndrome (FCS) is an autosomal recessive disorder of lipoprotein metabolism due to deficiency of lipoprotein lipase or Apo C II deficiency or presence of inhibitor to lipoprotein lipase. About 25% of FCS cases usually diagnosed during infancy [1]. We present a 40-day-old child with milky serum diagnosed to have lipoprotein lipase deficiency.

A 40-day-old female infant presented with 1 day history of poor feeding and lethargy. The baby was a product of non consanguineous marriage, born at term by vaginal delivery with birth weight of 2.8 kg. Antenatal and post natal period was uneventful. Child was exclusively breastfed since birth. Child has a healthy elder sister. On blood sampling for possible sepsis, child's blood was found to be pinkish white and viscous, which gradually turned into milky white after some time. She had hepatosplenomegaly and eruptive xanthomas over knee, face and buttocks. Liver and renal functions were normal. Sepsis markers were negative. Blood and urine cultures were sterile. Ultrasound abdomen revealed hepato-

splenomegaly with normal hepatic echotexture. Electrocardiography and echocardiography revealed no abnormality. On fundus examination, lipemia retinalis was found. Serum lipid profile revealed cholesterol 1467 mg/dL (Normal <200), triglyceride (TG) 5997 mg/dL (Normal <150), VLDL 1199 mg/dL (Normal <30), LDL 310 mg/dL (Normal <110), and HDL is 133 mg/dL (Normal 40-60**)**. Thyroid function, serum amylase and lipase were normal. Hemoglobin was low (7 gm/dL). Both parents had normal lipid profile, but her elder sister had moderately elevated triglycerides. No history of convulsion, jaundice, bleeding manifestation, skin rash. There was no known case of hyperlipidemia or premature sudden death in family.

Provisional diagnosis of familial chylomicronemia syndrome was considered because of extremely high triglyceride with moderately raised cholesterol with hepatosplenomegaly, eruptive xanthoma and lipemia retinalis. Blood was sent for genetic analysis and child was started on Fenofibrate, medium chain triglyceride containing oil and low fat diet (skimmed milk). Lipid profile repeated after 15 days revealed Cholesterol decreased to 297 mg/dL, triglyceride- 1793 mg/dL, VLDL -358 mg/dl and LDL-106 mg/dL. Fundus was normal on re-evaluation.

Genetic analysis revealed homozygous nonsense mutation in exon 5 of *LPL* gene in chromosome 8 resulting in premature truncation of the protein at codon 191 (p.Tyr191Ter) a rare mutation not reported previously in literature**.**

Hypertriglyceridemia is defined as having plasma triglyceride above the 95th percentile for age and sex [2]. Diseases having hypertriglyceridemia have a high risk metabolic dysfunction and cardiovascular diseases. Lipoprotein lipase is a key enzyme needed for hydrolysis of triacylglycerol in chylomicrons and LDL. Worldwide incidence is 1 in 1 million for LPL deficiency [3]. LPL deficiency in children usually has varied presentation. When TG >2000 mg/dL with eruptive Xanthoma mostly appear on shoulder, buttocks and extensor surface of limbs, lipemia retinalis is manifested when TG level surpasses 2500 mg/dl [4]. They are at increased risk of recurrent pancreatitis leading to pancreatic insufficiency which is the major morbidity of this disease and risk increases with level >1000 mg/dl [2]. Genetic analysis is the preferred and most readily applied method for diagnosis, as LPL mass assay is not easily available everywhere. Common *LPL* gene mutations reported in literature are Asp9Asn, Gly188Glu, Pro207Leu, Asp250Asn, Asn291Ser, Ser447X, Pro214Ser etc [5]

Differential diagnosis are Familial dysbetalipoproteinemia in which cholesterol and TG are elevated to a similar degree and presentation is in adulthood with tuberoeruptive xanthoma, Familial hypertriglyceridemia which presents without xanthoma with increased TG but normal cholesterol and Familial combined hyperlipidemia where cholesterol is more raised than TG without xanthomic eruptions.

Mainstay of treatment is severe dietary fat restriction for the lifetime [4]. MCT oil is recommended in chylomicronemia as it gets absorbed directly to portal circulation. The drugs studied and recommended for hypertriglyceridemia are fibric acid derivatives. These have the effect of both raising HDL and lowering triglycerides. Various case reports are there favouring use of fibrates without many side effects [6]. In our case we used fenofibrate which child was tolerating well till 3 months of follow-up.

Finding of lipemic serum during routine investigations should always be evaluated in detail. Early diagnosis, medical intervention by lipid-lowering agents and dietary modification can improve the prognosis and maintain a near normal lifestyle as the risk of pancreatitis and frequency of hospital admissions is significantly reduced.

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