Creamy clues to monogenic hypertriglyceridaemia

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A previously healthy 21-month-old Pakistani girl presented to our institution in June 2025 with a 1-year history of a pruritic papulovesicular rash affecting her limbs and groin. Apart from intermittent viral illnesses, there were no reported episodes of abdominal pain, vomiting or other symptoms suggestive of acute pancreatitis. She had been thriving well with normal intake, urine output and bowel habit. She was born at term by vaginal delivery with an uneventful perinatal history. Her parents were healthy second cousins.

Physical examination of the patient revealed multiple discrete yellow papules over all four limbs and the groin, sparing the face, trunk and back. Otherwise, she appeared well with age-appropriate growth parameters.

Two months prior to the current presentation, the patient had been admitted with an upper respiratory tract infection. Laboratory investigations showed mild microcytic anaemia, but routine biochemistry was unremarkable. The chemical pathology report noted serum turbidity requiring clearance by high-speed centrifugation prior to non-lipid analysis; however, this critical finding was overlooked, and the patient was discharged home.

FIG 2. The creamy appearance of a blood sample due to the presence of excess chylomicrons

FIG 1. Eruptive xanthomata over the extensor surfaces of the patient's limbs

At the latest clinic visit, the skin lesions were recognised as eruptive xanthomas, appearing as small yellow papules with a creamy centre (Fig 1). A grossly lipaemic blood sample showed a thick creamy layer (Fig 2), raising clinical suspicion of severe hypertriglyceridaemia. Laboratory testing confirmed this, with triglyceride level measuring 100.6 mmol/L.

The patient was admitted to the paediatric intensive care unit, kept nil by mouth and commenced on intravenous Actrapid (Bagsværd, Denmark) 0.1 unit/kg/hr, with a dextrose infusion to maintain euglycaemia. After 43.5 hours of infusion therapy, triglyceride level had reduced to 1.3 mmol/L (Fig 3). Following intravenous insulin therapy, she was managed with dietary restriction and essential fatty acid supplementation. Throughout the course, serial amylase and lipase levels remained normal. Computed tomography excluded acute pancreatitis. A family history revealed a nephew with monogenic

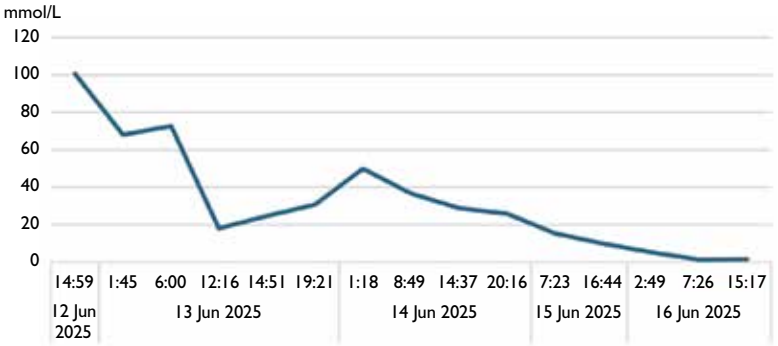

FIG 3. Trend in triglyceride levels over time following commencement of intravenous insulin infusion

hypertriglyceridaemia due to a *GPIHBP1* mutation, which was subsequently confirmed in our patient.

Monogenic hypertriglyceridaemia, also known as type I hyperlipidaemia, is an uncommon autosomal recessive condition most often resulting from *LPL* gene mutations that impair lipoprotein lipase function.[1] The disorder usually manifests in childhood with recurrent abdominal pain, pancreatitis, or characteristic features such as eruptive xanthomas, lipaemia retinalis, and hepatosplenomegaly. Without appropriate management, recurrent pancreatitis may progress to chronic disease with subsequent exocrine and endocrine insufficiency. Clinical manifestations typically emerge before 10 years of age, and approximately one quarter of cases present within the first year of life.[2]

Acute pancreatitis typically arises when serum triglyceride concentrations exceed 11.3 mmol/L.[3,4] Management involves fasting, intravenous fluid support, and continuous insulin infusion. This activates lipoprotein lipase, accelerating triglyceride clearance—reducing levels within 24 hours by approximately 40% with insulin alone and up to 80% when combined with fasting.[5] Despite this, some individuals, as in our case, remain asymptomatic even with extreme hypertriglyceridaemia.[6] In retrospect, earlier recognition of these 'creamy' clues—eruptive xanthomas and lipaemic serum—could have enabled a timelier diagnosis of this rare lipid disorder.

Author contributions

Concept or design: A Fu.
Acquisition of data: Both authors.
Analysis or interpretation of data: Both authors.
Drafting of the manuscript: A Fu.
Critical revision of the manuscript for important intellectual content: A Fu.

Both authors had full access to the data, contributed to the study, approved the final version for publication, and take responsibility for its accuracy and integrity.

Conflicts of interest

Both authors have disclosed no conflicts of interest.

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Ethics approval

The patient was treated in accordance with the Declaration of Helsinki. The parents of the patient provided written consent for all treatments and procedures, and verbal consent for publication, including the publication of the accompanying clinical images.

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