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A RARE CASE OF DYSLIPIDEMIA DUE TO LIPOPROTEINASE DEFICIENCY

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Lipoprotein lipase is a key enzyme needed for the hydrolysis of triacylglycerol in chylomicrons and very low density lipoproteins.(1)LPL deficiency is a rare autosomal recessive disorder with a prevalence of 1 in 1 000 000 in the United States and higher in other regions of the world.(1) LPL deficiency is characterised biochemically by severe hypertriglyceridemia more than 2000 and clinically by features including failure to thrive, eruptive xanthomas, hepatosplenomegaly, recurrent pancreatitis and lipemia retinalis.(1)

Here we describe a case of 5-year-old female presented to our pediatric service for evaluation and treatment of pain in abdomen with excessive crying and irritability. The birth and neonatal course were unremarkable. No H/O DM, Renal, Hepatic or thyroid disorder but H/O pancreatitis Y present.

On clinical examination mild hepatomegaly was present and blood samples were noted to be grossly lipemic. A lipid profile revealed a triglyceride concentration of 6043mg% with a total cholesterol of 565 mg%. Further evaluation was done and the management of such case is always remains a challenge.

References

(1) Dinesha Maduri Vidanapathirana, Thushara Rodrigo*., Lipoprotein Lipase Deficiency in an infant with Chylomicronemia,Hepatomegaly , and Lipemia Retinalis*(2017)*Global Paediatric Health Volume 4.,* Page No: *1-4*

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