Letters to the Editor

Lipoprotein-Lipase Deficiency

Lipoprotein lipase (LPL) deficiency is a rare disease,, so far not been reported in infants from India. A two months old boy was brought to our hospital for upper respiratory infection. He was the first and only child of non-consanguineous parents. There was no history suggestive of pancreatitis or ischemic heart disease in the parents or their relatives or of unknown childhood death in the family. Physical examination revealed left axillary BCG adenitis, firm hepatosplenomegaly, and optic fundi showed pale vessels. Other systems, growth and development were normal. Routine investigations were normal. As plasma was milky, serum lipid profile was done. It revealed hyperlipidemia with total serum cholesterol level of 982 mg/dL and triglyceride level of 1100 mg/dL. HDL and LDL levels were 12 mg/dl and 75 mcg/dl, respectively and triglyceride: HDL ratio was 82. Bone marrow examination did not reveal foam cell infiltration. Serum lipoprotein electrophoresis showed reduced alpha bands, normal beta and prebeta bands and increased amounts of chylomicrons, a pattern suggestive of Fredricksons type I lipoprotein deficiency. With this clinical and laboratory picture, a diagnosis of LPL deficiency was made. Due to the lack of facility, chylomicron level and post heparin LPL activity could not be assessed. Within 3 months of diagnosis, the child had two episodes of colicky abdominal pain suggestive of pancreatitis which was treated on conservative lines. Plasma of his parents was screened by electophoresis and was found to be normal.

LPL deficiency is inherited as an autosomal recessive disorder and an incidence rate of 1 in 100,000 is reported(l). It is usually due to mutation at the LPL gene(2-4). The LPL enzyme acts on chylomicrons to form free fatty acids (FFA) and remnant particles. Its deficiency produces lipemic plasma which could be the only manifestation requiring investigative work-up, as in our case. It is also associated with lipemia retinalis, as revealed by the pale vessels in optic fundus as in this baby, hepatosplenomegaly due to foam cell infiltration, chylomicronemia and intermittent attacks of eruptive xanthoma. Pancreatitis is the most dreaded complication. Circulating chylomicrons occurring in this condition are acted upon by pancreatic lipase. The resultant partially hydrolyzed products produce inflammation of pancreas with further release of pancreatic lipase thereby producing a vicious cycle. The LPL deficiency is diagnosed on the basis of chylomicronemia, Fredrickson's type I pattern on electrophoresis and demonstration of reduced activity of LPL in heparinized blood. LPL deficiency is managed by supplementing medium chain triglycerides as they are not incorporated into chylomicrons. Episodes of pancreatitis are treated conservatively. Unlike other dyslipidemias this condition dose not produce accelerated atherosclerosis.

S. Sushamabai,
Dipti Subrahmanian,
Department of Child Health,
Medical College,
Kottayam-686 036,
Kerala.

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Pulmonary Agenesis

I read with interest the case report on this subject(1) and wish to share more information on this anomaly. The incidence of pulmonary agenesis is one in 10,000-15,000(2). Parental consanguinity has been found by some workers(3). Associated congenital abnormalities and the involvement of the normal lung is directly proportional to the overall prognosis. In one series 14% of infants with pulmonary agenesis were still born, and 50% of the remainder died by the age of 5 years(4). These patients should be given antibiotics for pulmonary infection and influenza vaccine.

Lung transplantation in pediatric patients is being done with success. This operation was recently performed on a two week old baby, who is the youngest child in the world to have a lung transplant. Dr. Starnes, a cardiothoracic surgeon in the USA, performed lung transplant operations, both on young and older children. The long term prognosis following transplantation is better in older children. It is too early to predict the prognosis in smaller children. The oldest child surviving after lung transplantation has survived for 10 years(5).

Ghulam Nabi,
Neonatologist,
Maternity and Children Hospital,
Madina Al Munawarah,
Saudi Arabia.

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