Approach to Hereditary Storage Diseases in Patients with Hepatosplenomegaly

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Maryam Razzaghy Azar, MD

Liver involvement of some kind is a presenting feature of a number of inherited metabolic diseases. One approach to the diagnosis of inherited metabolic diseases presenting as hepatic syndrome is to consider four possible presentations, recognizing that there is considerable overlap between them. They are:

- Jaundice
- Hepatomegaly
- Hypoglycemia
- Hepatocellular dysfunction

Hyperbilirubinemia in infants with classical galactosemia is often initially unconjugated, converting only after a period of some days to the conjugated hyperbilirubinemia that is widely regarded as characteristic of the disease. Conjugated hyperbilirubinemia as a manifestation of inherited metabolic disease is more common than unconjugated hyperbilirubinemia because it includes those diseases, like galactosemia, hepatorenal tyrosinemia, and hereditary fructose intolerance, in which hepatocellular dysfunction is prominent. The hepatomegaly associated with inherited metabolic diseases is generally persistent and non-tender. If the liver is so soft that the edge is difficult to palpate, enlargement is likely to be due to accumulation of triglyceride, a typical feature of GSD (glycogen storage disease) type I. At the other extreme, a hard and irregular liver edge, often associated with only modest enlargement of the organ, is characteristic of cirrhosis. When it is enlarged as a result of lysosomal storage, the liver is usually firm, but not hard. Is the spleen also enlarged? A history of hematemesis or the presence of ascites or abdominal venous dilatation, would suggest that splenomegaly is caused by portal hypertension resulting from cirrhosis. However, the spleen may be enlarged by infiltration or accumulation of the same cells or metabolites causing enlargement of the liver. Besides sharing the portal circulation, the liver and spleen both contain components of the reticuloendothelial system (RES). Conditions causing expansion of the RES, either as a result of cellular proliferation or storage within RES cells (i.e., macrophages) commonly present with clinical enlargement of both organs. This is characteristic of hepatorenal tyrosinemia (hereditary tyrosinemia, type I). Hypoglycemia is caused by the deficiency of enzymes which are involved in glycogenolysis, gluconeogenesis and beta oxidation of fatty acids and etc. The presentation of inherited metabolic diseases with onset in the newborn period or early infancy as acute hepatocellular disease is characterized in most cases by some combination of failure to thrive, mild to severe hyperbilirubinemia, hypoglycemia, hyperammonemia, elevated aminotransferases, bleeding diathesis, edema, and ascites. Approaching to inherited metabolic diseases is by

Department of Pediatric Endocrinology and Metabolism, H. Aliasghar Hospital, Iran University of Medical Sciences, Tehran, Iran

Corresponding Author:
Razzaghy Azar M.MD
H. Aliasghar Hospital, Tehran, Iran
razavizahra@yahoo.com.au

ruling out infectious diseases and developmental defects and doing special laboratory tests for inherited metabolic disorders. Management is by drug therapy, bone marrow transplantation, liver transplantation and gene therapy.

Keywords: Approach; Storage Disease; Hepatosplenomegaly