Case Report

Galactosaemia presenting as poor feeding in infancy

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Introduction

Main source of dietary galactose is lactose from milk, which is converted by galactokinase to galactose-1-phosphate (Gal-1-P) which, in turn, is converted to uridine diphosphate galactose by galactose-1-phosphate uridyl transferase. This metabolism occurs in the liver and red blood cells. Galactosaemia is due to a defect in Gal-1-P uridyl transferase. This causes the accumulation of Gal-1-P in the tissues resulting in injury to parenchymal cells of the kidney, liver and brain[1]. In the liver it causes cholestasis.

Case report

A ten week old baby boy from Kalutara presented with poor feeding and inadequate weight gain from birth. He was the product of a non-consanguineous marriage, delivered at term without complications.

On admission, baby was pale and mildly icteric. His length and weight were below the 3rd percentile. There was no hepatomegaly. In the eyes, bilateral cataracts were found. Pale stools were noted which, according to the mother, had been present for more than three weeks.

His haemoglobin was 6.6 g%. Serum bilirubin showed an increase in the direct fraction. Prothrombin time was prolonged. Urine ward test for sugar was yellow to orange on two occasions. Baby was transfused with packed red cells, 15 ml/kg and was transferred to Lady Ridgeway Hospital for a HIDA scan and liver biopsy. The HIDA scan was normal. The liver biopsy showed fatty changes with mild to moderate lymphohistiocytic infiltrates in the portal tract. Focal interface hepatitis, mild lobular inflammation and evidence of increased glycogen within the hepatocytes were also noted. The above features were in favour of galactosaemia.

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Following investigation, breast feeding was stopped and the child was fed with O–Lac which is a lactose free formula. After starting lactose free formula, rapid weight gain was noted, urine sugar became bluish green and cataract disappeared within one month.

Discussion

Galactosaemia is a rare inborn error of metabolism with an autosomal recessive inheritance but mutations also occur[2]. Severity depends on the quantity of milk ingested and the presence of residual transferase activity in the liver. The accumulation of Gal-1-P in various organs causes poor feeding, vomiting, jaundice, hepatomegaly, liver dysfunction, cataract, anaemia, hypoglycaemia, and mental retardation. Cataract can be present from birth or appear later[3].

To diagnose galactosaemia, the simplest screening test is to check urine for reducing substances[3]. Confirmatory diagnosis is by assay of red blood cell (RBC) Gal-I-P uridyl transferase activity. However, any suspicious results are an indication for initiating treatment[2].

Patients can present with shock, haemorrhage, septicaemia and disseminated intravascular coagulation within two days of birth. At the end of the first week, poor feeding, jaundice, vomiting, hepatomegaly and diarrhoea can develop. At two to six months of age, failure to thrive, cataract, hypoglycaemia, anaemia and liver dysfunction can be a presentation. Damage to kidney can cause renal Fanconi syndrome. The most common presentation is prolonged jaundice with failure to thrive. Liver cirrhosis develops if galactose ingestion continues.

Early diagnosis and treatment of galactosaemia will improve the prognosis. Early treatment will halt the progress of liver involvement and can even make it disappear. However, the brain damage is irreversible. Thus, any suspicious results are an indication for initiating treatment. If any patient presents with failure to thrive and obstructive jaundice, immediate investigations

should include full blood count, blood group, blood sugar, urine for reducing substances, liver function tests and TORCH screening. If the clinical features and basic investigations are in favour of galactosaemia, RBC Gal-I-P uridyl transferase enzyme assay should be done to confirm the diagnosis but this facility is not available in Sri Lanka.

Therapy involves elimination of galactose containing foods like breast milk and milk products. Elimination of galactose from the diet causes reversal of growth failure, improvement in renal and hepatic dysfunction and the disappearance of cataract. Patient can be fed with a soya milk formula like O-Lac and they should avoid milk products, sausage and candies. Because of liver involvement, patients need fat soluble vitamin supplements.

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