FRUCTOSE INTOLERANCE ASSOCIATED WITH CONGENITAL CATARACT*

Report of a Case

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Hereditary fructose intolerance is an inborn metabolic disorder, first described by Chambers and Pratt (1956) and Froesch et al. (1967). Since then more than 45 cases have been described in the literature. It appears that the disorder is not as rare as was believed initially. There is no case report from India. We describe a case of fructose intolerance associated with congenital cataract, a rare occurrence.

The metabolic disorder is characterised by symptoms of nausea, vomiting, malaise, convulsions and coma which follow after the ingestion of foods containing fructose. It may end fatally if undiagnosed and untreated early. If untreated cases survive, they continue to have hypoglycaemic symptoms in adult life (Swales and Smith 1960, Froesch et al 1963). With continued ingestion of fructose cirrhosis may develop (Jeune 1961, Perheentupa et al 1962). In some, mental retardation can occur. (Froesch et al 1957, Perheentupa et al 1962). In none of the cases was association of congenital cataract reported.

It was postulated by Froesch et al (1957) that this disorder is due to the absence of fructose-1-phosphate aldolase enzyme in the liver. It was later confirmed by Hers and Joassin (1961), Nikkila et al (1962), Froesch et al. (1963) and Black and Simpson (1967). The mode of inheritance seems to be of the autosomal recessive type.

Report of a Case

A male child, 10 weeks old, was referred to us with complaints of a cataract in one eye from birth. He was delivered by caesarian section at full term for foetal distress and weighed 3.0 Kg. at birth. A cataract in the right eye and a lower incisor tooth were present at birth. No other congenital defects were observed. The boy developed jaundice on the 3rd day, which cleared on the 21st day. The tooth was removed when the boy was 20 days old to prevent the child from accidental asphyxia. He developed diarrhoea and abdominal distension on the 8th day of birth, which continued on and off,
sometimes with vomiting. He had repeated attacks of upper respiratory infection. The liver was found to be enlarged 3 cm below the costal margin. The child was put on Lactodex for the first 3 days. Later for a week the child was given Ostermilk in addition. He was breast-fed only for one week. The child was put on Ostermilk exclusively from the 20th day. From the age of 4 weeks the boy received fruit juice with cane sugar regularly. An ophthalmologist reported the presence of a central cataract in the right eye on the 10th day.

There was a strong history of diabetes mellitus in the family on the paternal side. Two sisters were both normal.

The urine showed a reducing substance, positive to Seliwanoff’s and Foulger’s tests. It was laevorotatory and was identified as fructose and confirmed by a chromatogram. Quantitative estimation showed 510 mg of reducing substance in 100 ml. of urine. Total blood sugar was 118 mg.%. Quantitative estimation of fructose and fructose-1-phosphate aldolase activity in the liver biopsy material was not possible. However, in view of the symptoms of diarrhoea and vomiting, a diagnosis of fructose intolerance was made. The urine did not show any albumin.

The mother was advised to withdraw cane sugar (sucrose) and fruit juice from the child’s diet. The urine showed 230 mgs.% of reducing substance in the urine 24 hours after withdrawal of fructose from the diet. The urine became sugar free three days later. It was noticed that the cataract was getting opaque. At the age of 16 weeks, the cataract was absorbed in the centre, with a few opacities at the periphery of the lens. Light fixation was present.

The child is now 2 years 2 months old. He takes rice, meat and eggs. No fructose containing foods are given. There is no further clearance of opacities in the eye. All the milestones were delayed. The developmental quotient is about 50. The child is able to crawl, stand with support, utters a few monosyllables, plays well, recognises his mother but is not very active. The liver is not enlarged now. Weight gain is normal.

Comment

Signs and symptoms of fructose intolerance are varied. (Froesch et al. 1957, 1963, Nikkila et al 1962, Cornblath et al 1963, Black and Simpson 1967). Even in the same patient there can be remarkable variation in symptoms (Swales and Smith 1966). The affected child thrives well so long as he is breast-fed. Once the child is put on foods containing cane sugar or fructose gastrointestinal manifestations are seen and sometimes hypoglycaemic convulsions occur. The hypoglycaemia is said to be due to inhibition of hepatic glucose release and not due to increased insulin secretion (Froesch et al. 1963).

The interesting feature in our case is its association with a congenital cataract which has not been reported so far. Further there was a partial clearing of the cataract on therapy. Whether it was due to withdrawal of fructose from the food or was coincidental is not definite. Pirie and Van Heyningen (1964) found that the lens of diabetics, both cataractous