

Case Reports

Zellweger syndrome (cerebro-hepato-renal syndrome)

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A syndrome with distinctive clinical features affecting brain, liver and kidneys was described by Bowen et al in 1964¹ and Smith et al. in 1965². In 1973 Goldfisher et al³ has reported that peroxisomes were absent in the liver and kidneys of affected children. More recently lack of dihydroxyacetone phosphate acyltransferase (DHAP-AT) a peroxisomal enzyme with a major role in glycerol ether lipid synthesis has been documented. This condition appears to be quite rare and incidence has been estimated as 1 in 100000 live births⁴.

Case Report

A baby boy was delivered vaginally at 38 weeks of gestation at Base Hospital, Negombo. It was a breech delivery and the Apgar score was 8 at 1 minute. Antenatal period had been uneventful. Baby was admitted to Teaching Hospital Ragama on the 3rd day with a history of poor sucking and lethargy. The parents are first cousins. Mother was 26 years old and father was 33 years old. They have been married for 8 years. This was the fourth pregnancy. C 1 is a normal boy of 6 years. C 2 a girl, died on the 16th day and C 3 a boy, died at 3 months of age. Both were vaginal deliveries and C 3 was a breech delivery. They also had been lethargic and had feeding difficulties since birth. C 3 had been diagnosed to have a congenital heart disease (? septal defect). Both had recurrent seizures.

Birth weight of the baby was 2.3kg, length was 49cm and the head circumference was 31cm. He was lethargic and markedly hypotonic on admission. He was afebrile, had large anterior and posterior fontanelles and a widely separated sagittal suture.

Baby had a high forehead, an absent orbital ridge, low set ears and mild micrognathia (Figure 1).

He also had a comparatively long body (49cm), short fingers and a single palmar crease on the right hand.

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Figure 1

In the lower limbs there was limited extension at the knees and equino varus deformity of the left foot (Figure 2).

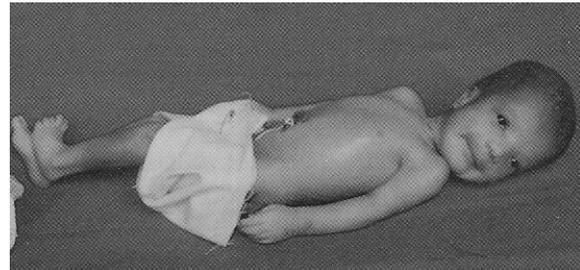


Figure 2

Heart and lungs were clinically normal and liver was just palpable

Investigations

Full blood count, blood sugar and liver function tests were normal. Ultrasound scan of the brain was normal.

Ultrasound scan of the abdomen showed abnormalities in the kidneys. They were more echogenic than normal and there was mild dilatation of the pelvicalyceal system of both kidneys. There

was a suggestion of a small cyst in the lower pole of the right kidney.

The x ray of the knees showed typical scimitar shaped calcification of patellae (Figure 3).

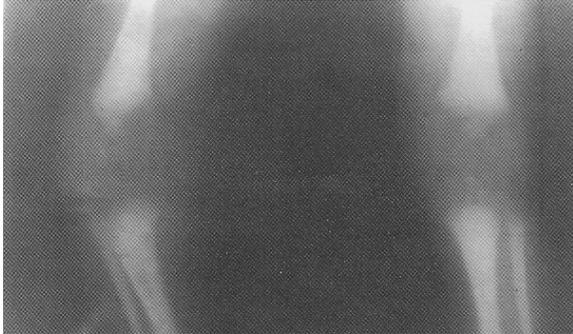


Figure 3

The baby was given gavage feeds and she developed seizures on the 4th day which became more frequent the next day. On the 6th day the parents took the baby against medical advice.

Discussion

The cerebro-hepato-renal syndrome of Zellweger is an extremely rare condition, not documented in Sri Lanka. It can be fairly easily diagnosed by a clinician who is familiar with this condition. Some patients may be mistakenly diagnosed clinically as Down Syndrome.

The combination of severe hypotonia and cerebral dysfunction manifested as lethargy, poor feeding and early onset of seizures in a baby with characteristic high forehead, long face and long trunk and hypotonia should point to a diagnosis of Zellweger syndrome. The diagnosis can be confirmed by radiological features of knee which shows typical scimitar shaped calcification of patella and tri-radiate cartilage in the acetabulum in the hips. Majority of the babies have been delivered by breech. Patent ductus arteriosus and septal defects have been described in some of these children. 97% of the babies have renal cysts. This is a fatal autosomal recessive disorder where majority of children have succumbed to the illness during the 1st few month of life.

References

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