always gets the vein, and gets it easily. The child does not struggle much as he/she is comfortable in the parent's arms. This facilitates the job. The previously informed parents have no tension, as they witness everyhing and the job is over in a few minutes. This position obviates the need of a table or a bed or a procedure room for venous cannulation. It can be done in the consulting room or in emergency, anywhere.

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Glycogen Storage Disease Type II

Glycogen storage disease type II was the first lysosomal storage disease to be recognized. To date, few cases of GSD type IIa have been reported in the Indian literature(1). We report a similar case of GSD type IIa in a 14-month-old child proved by clinical, laboratory and histopathological features.

A 14-month-old first born female child of a non-consanguinous marriage was referred to us for generalized muscle weakness and intractable cardiac failure. She was born of a normal term delivery and was apparently normal at birth. There was no history of diminished fetal movements in the antenatal period. Parents initially noticed paucity of movements of the limbs by around one month of life. The weakness then progressed over the time. She had repeated aspiration of feeds since the age of 3 months. There was profound motor developmental delay since birth, but the social smile was attained at the normal age. There was no family history of a similar illness or any previous abortions in the mother. Examination showed a poorly nourished ill-looking child with central cyanosis and tachypnea. The respiratory excursions were shallow and the pulse was rapid and thready. The child was in a pithed frog position with minimal body movements and a weak inaudible cry even on painful stimuli. There was marked cardiomegaly. Liver was palpable 3 cm and firm. There was severe hypotonia, areflexia and muscle weakness. The chest *X*-ray revealed massive cardiomegaly. ECG showed short PR interval, inverted T-waves in the limb leads and tall QRS complexes. The serum CPK level was 222 IU/L (normal range 20-50 IU/L).

The child died 50 hours after admission due to cardiorespiratory failure. A post-mortem skeletal muscle biopsy showed marked infiltration with glycogen. This finding along with the clinical and laboratory features confirmed the diagnosis of GSD type IIa.

GSD type II is a rare inherited metabolic disorder with an incidence of one in more than 100,000(2). Three clinical phenotypes are recognized: the infantile (II*a*), childhood (II*b*), and the adult (IIc) types. In the infantile form (also known as Pompe's disease) affected infants are usually normal at birth. Skeletal muscle and cardiac involvement become clinically obvious within the first few months of life as muscle weakness and resistant cardiac failure, respectively. Both these progress relentlessly and death occurs before the age of two years due to cardiac and/or respiratory failure.

GSD type II is inherited as an autosomal recessive disorder involving the lysosomal glycogen degrading enzyme acid maltase or alpha-1,4-glucosidase. The gene for acid maltase is localized on chromosome 17. To date there is no specific treatment for the affected patients. Recent trials with high protein diet have yielded encouraging results in some studies(3), but not so in others(4). Bone marrow transplantation is another promising therapeutic approach which is beng evaluated(5). As it is a single gene disorder, gene therapy may prove to be useful in its management in the future, once the genome is fully mapped. Antenatal diagnosis is now possible by electron microscopy of chorionic villus fibrocytes obtained by chorionic villus biopsy at 10 weeks of gestation or by studying the enzyme activity in cultured amniotic fluid fibroblasts obtained by aminocentesis(6,7). This is especially useful in disease detection in subsequent pregnancies and to terminate if needed.

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