Jarcho-Levin Syndrome

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The Jarcho Levin syndrome also known as spondylo thoracic dysplasia is a condition manifested by malformations of the ribs and vertebrae. Described for the first time by Jarcho and Levin in 1938(1), there are now 57 reported cases in literature(2) whereas from India there has been only one case report(3). This is an autosomal recessive condition and may be associated with neural tube defects (NTD)(4).

Here we describe a case of this syndrome, associated with neural tube defect and suggest that in all cases of NTD this syndrome should be thought of. This would result in better genetic counselling regarding recurrence risk, management and prognosis.

Case Report

A young iren-consanguinous couple was referred to us at 32 weeks of gestation because a routine ultrasound had detected fetal hydrocephalus. A repeat ultrasound at our centre confirmed this and in addition also detected a thoracic meningomyelocele together with abnormal configuration of ribs. The patient underwent termination. The fetus was female, still born with a birth weight of 1200 g. She had a short trunk, protuberant abdomen and a mid thoracic meningomyelocele.

Radiographic evaluation revealed hemi-vertebrae and hypoplastic vertebrae in the mid thoracic and lower lumbar regions. The ribs had abnormal shape and configuration bilaterally *(Fig. 1)*.

The autopsy showed bilaterally bilobed but not hypoplastic lungs. There was no other abnormal finding.

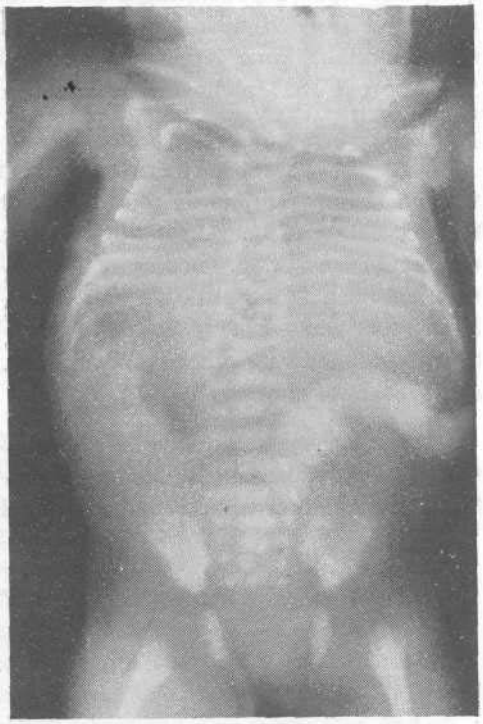

Fig. 1. X-ray of fetus. Note the vertebral anomalies and bizarre shape of ribs.

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The patient was counselled regarding the autosomal recessive inheritance of this syndrome, the 25% recurrence and the futility of periconceptional vitamin supplementation for prevention.

A year later this couple had a normal child with no congenital malformations.

Discussion

We have described a case of the Jarcho-Levin Syndrome which had a neural tube defect associated with vertebral and rib anomalies. Not all cases of this syndrome are associated with NTD and anomalies of the urogenital system and limbs have also been seen(5,6).

However, it is important to rule out the syndrome when encountering a child with NTD. An isolated neural tube defect is multifactorial, having a recurrence risk of 5% whereas the Jarcho-Levin syndrome has a much higher recurrence of 25%.

With the advent of periconceptional vitamin therapy (primarily folic acid) for the prevention of neural tube defects this distinction is even more necessary. Prevention has been found effective only in isolated NTD and is not advocated in conditions associated with other malformations, *e.g.,* Meckel's syndrome(7).

It is thus suggested that all cases of neural tube defects should undergo *(i)* detailed examination to rule out associated malformations, *(ii)* radiological evaluation for defects of the ribs and vertebrae, and *(iii)* autopsy of still borns to document anomalies of urogenital tract.

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