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Sprengel's Deformity with Absent Ribs

Harmesh Singh

Sprengel's deformity is an uncommon congenital anomaly(1); but is one of the common anomalies of the shoulder(2,3). It consists of elevation and upward rotation of the scapula(4). It is more common in girls and on left side(1).

Sprengel's deformity is frequently associated with abnormalities of the cervical spine such as Klippel-Feil syndrome and congenital scoliosis, as well as cervical ribs and rib fusions(5). Other associated anomalies include cervical spina bifida, congenital kyphosis, syringomyelia, platybasia, situs inversus, mandibulofascial dysostosis, ipsilateral shortened humerus and clavicular anomalies and rarely renal anomalies(1,6,7). Hypoplasia and anomalies of the upper thoracic ribs are reported in Sprengel's deformity(4). There are no reports in literature describing absent ribs in patients with Sprengel's deformity. We report a case of Sprengel's deformity with absent ribs.

Case Report

The patient was a 2-month-old, Sikh, male child, product of a nonconsanguinous marriage after an uneventful 38 weeks gestation born to a primigravida mother. The parents have history of abnormal movements of the chest during respiration. There was no other associated complaint.

The examination revealed higher right shoulder and scapula than on the opposite side. On looking at from the rear, the inferior angle was displaced towards the spine. On attempted abduction of the arm, the lower angle of the right scapula did not rotate outwards as it should in normal. Kyphosis and scoliosis were present. On palpation right upper thoracic ribs were not felt. There was indrawing of the right upper chest wall during inspiration.

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General physical and other systemic examination was within normal limits. There was no other obvious anomaly.

The roentgenogram (*Fig.*) revealed a typical Sprengel's deformity of the right scapula with absent second to sixth ribs, fusion of seventh and eighth ribs on the same side. Vertebral anomalies were seen in the form of scoliosis, hemivertebra, spinabifida, and fusion of 7th and 8th thoracic vertebrae. Parents were refused admission and did not come for follow-up.

Discussion

The frequent association of deformities of the occiput and the base of the skull, the cervical and the upper thoracic spine, and the ribs and the surrounding muscle tissues indicates that the causative factor(s) is operative during early embryonic life, most particularly at the time of development of the cervical spine and the upper limb buds(6). The elevation and upward rotation of the scapula is due to failure of the normal descent of the scapula from its embryonic position to its normal location(5).

The report of Sprengel's deformity of right scapula in a male child is unusual, in the literature it is reported that the deformity is more common on left side with a female to male ratio of 3 : 1(1). All other anomalies detected in the present case are documented in the literature but absent ribs in Sprengel's deformity has not previously been reported.

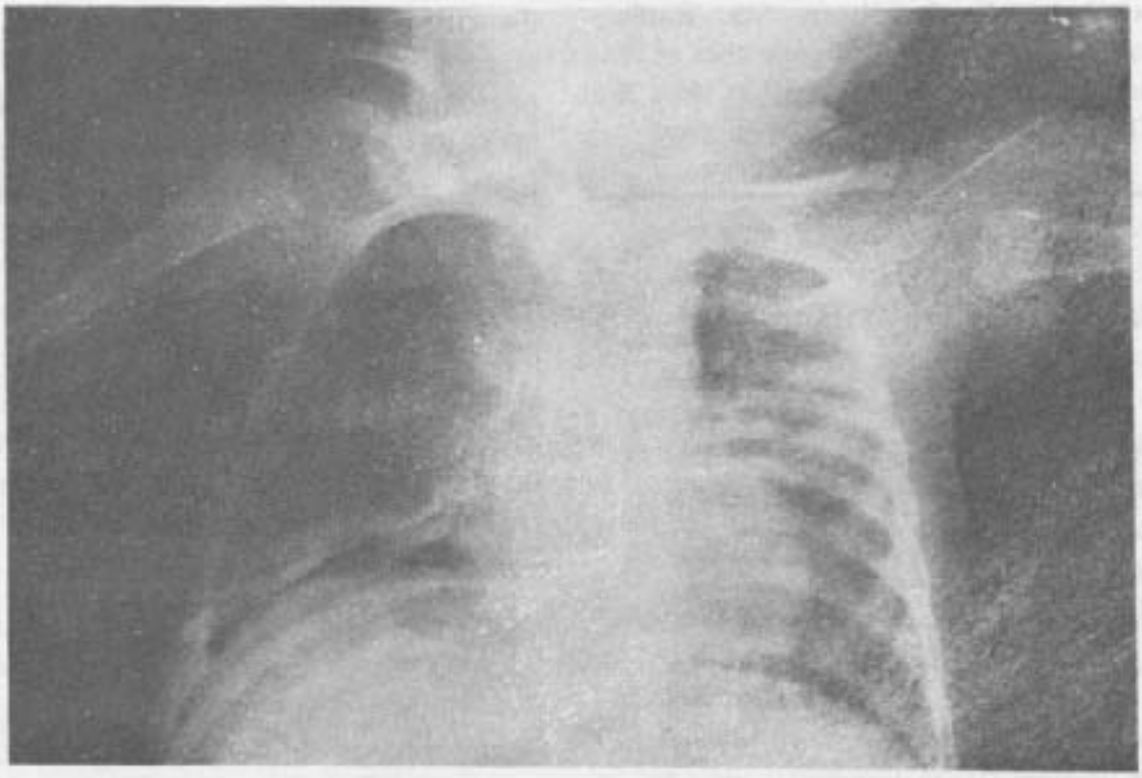

Fig. Roentgenogram showing Sprengel's deformity of right scapula, absent right upper thoracic ribs and other anomalies.

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Thanatophoric Dwarfism

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Until recently thanatophoric dwarfism, a short-limbed dwarf condition, was misinterpreted as severe immature form of achondroplastic dwarfism. In 1967, Maroteux *et al.*(1) stated that thanatophoric dwarfism was discrete entity; the condition is rare with a prevalence of 1 in 6400 population.

Case Report

A stillborn baby was delivered to a primigravida at 40 weeks gestation. During the antenatal period the height of uterus was less than period of gestation and the presentation was breech. *In utero*, ultrasonography showed a single live fetus with biparietal diameter of 92 mm, hyperextended spine with platyspondyly, narrow thorax with pulmonary hypoplasia, normal abdominal contents, posterior fundal position of placenta, very small upper and lower extremities with bowing of long bones and excessive liquor. A diagnosis of thanatophoric dwarfism and asphyxiating thoracic syndrome was considered.

There was no history of abortion, dwarfism in parents or in other family members, consanguinity or exposure to potential teratogenic agents. Mother's 'TORCH' test was positive for Rubella and CMV antibodies. Radiographic examination of the stillborn showed disproportionately short extremities in relation to trunk. The skull was large showing frontal bossing, spine showed platyspondyly with H-shaped vertebra, the thorax was triangular in shape, tubular bones were short with bowing of long bones and slight metaphyseal irregularity and flaring.

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