Hypoplasia of the left first rib in a child with Down’s syndrome and an endocardial cushion defect

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A case is reported of a child with Down’s syndrome having a hypoplastic left first rib and an endocardial cushion defect. To our knowledge, a similar entity has not been reported.

Congenital anomalies of the ribs have been an area of interest for the thoracic surgeon, orthopaedic surgeon, paediatrician, and radiologist. Fusion of adjacent ribs and bifid ribs are a common radiographic finding, but absence or rudimentary formation occurs less frequently (Ehrenhaft, Rossi, and Lawrence, 1966). The incidence of absence and rudimentary formation of the first rib has been reported to vary between 1.0% and 0.2% (Table I).

In the field of mental retardation, children with Down’s syndrome are sometimes referred to as the ‘unfinished children’ because they have so many developmental anomalies. Among these anomalies are several skeletal manifestations of the syndrome (Table II). The two common thoracic anomalies are accessory sternal ossification and 11 pairs of ribs (Currarino and Swanson, 1964; Getlik, 1966). Beber, Litt, and Altman (1966) found 11 pairs of ribs.

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TABLE I
FREQUENCY OF RIB ANOMALIES

<table>
<thead>
<tr>
<th>Author</th>
<th>No. of Chest Radiographs</th>
<th>Percentage of all Rib Anomalies</th>
<th>Percent of Aplasia of First Rib</th>
<th>Percent of Hypoplasia of First Rib</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td></td>
<td></td>
<td>Bilateral Right Left</td>
<td>Bilateral Right Left Total</td>
</tr>
<tr>
<td>Sycamore (1944)</td>
<td>2,000</td>
<td>2.80</td>
<td>0 0 0</td>
<td>0.20 0.10 0.20 0.50</td>
</tr>
<tr>
<td>Etter (1944)</td>
<td>40,000</td>
<td>1.35</td>
<td>0 0 0</td>
<td>0 0.06 0.12 0.20</td>
</tr>
<tr>
<td>Pionnier and Depraz (1956)</td>
<td>10,000</td>
<td>5.72</td>
<td>0.01 0.01 0</td>
<td>0.38 0.26 0.38 1.02</td>
</tr>
</tbody>
</table>

TABLE II
SKELETAL MANIFESTATIONS OF DOWN’S SYNDROME

<table>
<thead>
<tr>
<th>Anatomical Location</th>
<th>Skeletal Features</th>
<th>Authors</th>
</tr>
</thead>
<tbody>
<tr>
<td>Skull</td>
<td>Brachycephaly, low cribiform plate, increased interorbital distance, absence of frontal sinuses</td>
<td>Benda (1946); Spitzer et al. (1961); Caffey and Ross (1958)</td>
</tr>
<tr>
<td>Pelvis</td>
<td>Horizontal sloping acetabulae, flaring of iliac wings, small tapering ischial rami, coxa valga</td>
<td></td>
</tr>
<tr>
<td>Hand</td>
<td>Shortening of middle phalanx of 5th finger; Atlanto-axial dislocation, disc space narrowing, and end plate irregularities</td>
<td></td>
</tr>
<tr>
<td>Cervical position of spine</td>
<td>Frequent incomplete fusion of vertebral arches, vertebral bodies relatively increased in vertical diameter and decreased in anteroposterior diameter</td>
<td></td>
</tr>
<tr>
<td>Lumbar position of spine</td>
<td>Development of multiple epiphyseal centres</td>
<td></td>
</tr>
<tr>
<td>Manubrium sterni</td>
<td>Eleven pairs of ribs</td>
<td></td>
</tr>
</tbody>
</table>

Adapted from Curtis, Blank, and Fisher (1968).
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Ribs in 38% of their cases and suggested that the aplasia may be the result of the absence of a dorsal vertebra. The incidence of an endocardial cushion defect in patients with Down's syndrome is approximately 14% (Rowe and Uchida, 1961). We wish to present the case of a child with Down's syndrome having hypoplasia of the left first rib and an endocardial cushion defect. To our knowledge, a similar case has not been reported.

CASE REPORT

C. W., a 10-year-old girl with Down's syndrome, was noted to have a heart murmur at 6 years of age. She was admitted to the University of Chicago Hospitals in May 1970 for cardiac catheterization. The haemodynamic studies demonstrated an atrial septal defect, a ventricular septal defect, mitral regurgitation, and pulmonary hypertension. Chromosome studies showed her to have trisomy 21. On chest radiography she was found to have a rudimentary left first rib (Figure). There was, however, no visible chest deformity and her pectoral muscles were intact.

REFERENCES


FIGURE. Radiograph showing hypoplastic left first rib.
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