Thorax (1972), **27**, 100.

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

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TABLE I

FREQUENCY OF RIB ANOMALIES

Author	No. of Chest Radiographs	Percentage of all Rib Anomalies	Percent of Aplasia of First Rib			Percent of Hypoplasia of First Rib			
			Bilateral	Right	Left	Bilateral	Right	Left	Total
Sycamore (1944)	2,000	2·80	0	0	0	0·20	0·10	0·20	0·50
Etter (1944)	40,000	1·35	0	0	0	0	0·06	0·12	0·20
Pionnier and Depraz (1956)	10,000	5·72	0·01	0·01	0	0·38	0·26	0·38	1·02

TABLE II

SKELETAL MANIFESTATIONS OF DOWN'S SYNDROME[2]

Anatomical Location	Skeletal Features	Authors
Skull	Brachycophaly, low cribiform plate, increased interorbital distance, absence of frontal sinuses	Benda (1946); Spitzer *et al.* (1961)
Pelvis	Horizontal sloping acetabulae, flaring of iliac wings, small tapering ischial rami, coxa valga	Caffey and Ross (1958)
Hand	Shortening of middle phalanx of 5th finger	Hefke (1940)
Cervical position of spine	Atlanto-axial dislocation, disc space narrowing, and end plate irregularities	Tishler and Martel (1965); Martel and Tishler (1966)
Lumbar position of spine	Frequent incomplete fusion of vertebral arches, vertebral bodies relatively increased in vertical diameter and decreased in anteroposterior diameter	Mautner (1950); Rabinowitz and Moseley (1964)
Manubrium sterni	Development of multiple epiphyseal centres	Currarino and Swanson (1964); Horns and O'Loughlin (1965)
Ribs	Eleven pairs of ribs	Beber *et al.* (1966); Getlik (1966)

[2] Adapted from Curtis, Blank, and Fisher (1968).

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.

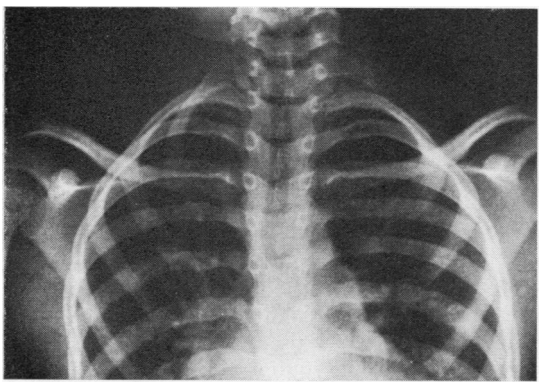

FIGURE. *Radiograph showing hypoplastic left first rib.*

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