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CASE REPORT

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Asphyxiating thoracic dysplasia

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Summary

Background: Asphyxiating Thoracic Dysplasia is the most frequent form of Small Thorax – Short Rib Syndromes.

Case report: Asphyxiating Thoracic Dysplasia in two patients with different clinical course is reported.

Conclusions: Radiographic examination is the only method to diagnose Asphyxiating Thoracic Dysplasia with certainty. The correct diagnosis is important for prognostication and genetic counseling. It also excludes the necessity of further, often expensive investigations.

Key words: Bone dysplasia • asphyxia • thorax • pelvis

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Background

Asphyxiating Thoracic Dystrophy (ATD) (MIM 208500) is a relatively common bone dysplasia occurring in about 1:100.000-130.000 births.

The major distinctive clinical features are long, narrow thorax, short extremities, short hands, progressive renal disease and hepatic fibrosis [1, 2].

The major characteristic radiographic findings consist of small thorax, triradiate acetabulum, short long tubular bones, and short middle and distal phalanges with cone shaped epiphyses [1, 2].

ATD is inherited as a recessive trait with significant variability of clinical course, and fairly constant radiographic presentation. Between the severe cases which are stillborn or die soon after the birth [2, 3], and the asymptomatic patients diagnosed accidentally [4] there is a whole range of "between cases" of variable severity. Recurrent respiratory infections and progressive renal disease are constant features of the "in between cases".

We report two patients with ATD, one asymptomatic and one with severe, clinical course and fatal outcome.

Case report

PATIENT 1

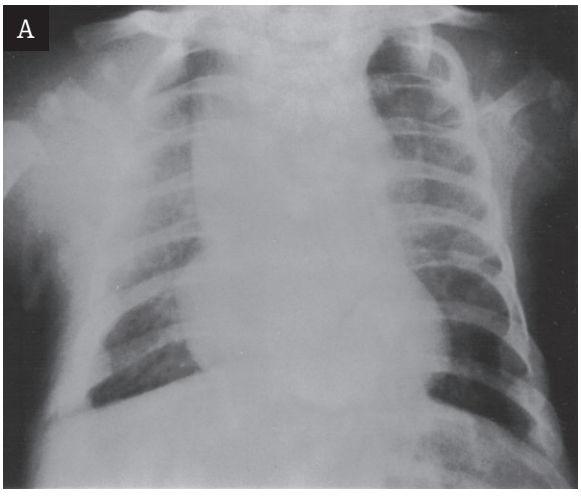
This boy was born after an uncomplicated pregnancy and delivery. There was no evidence of respiratory distress but because of small thorax skeletal survey was performed and the diagnosis of ATD established on the basis of chest and pelvic X-rays (Fig. 1A&B).

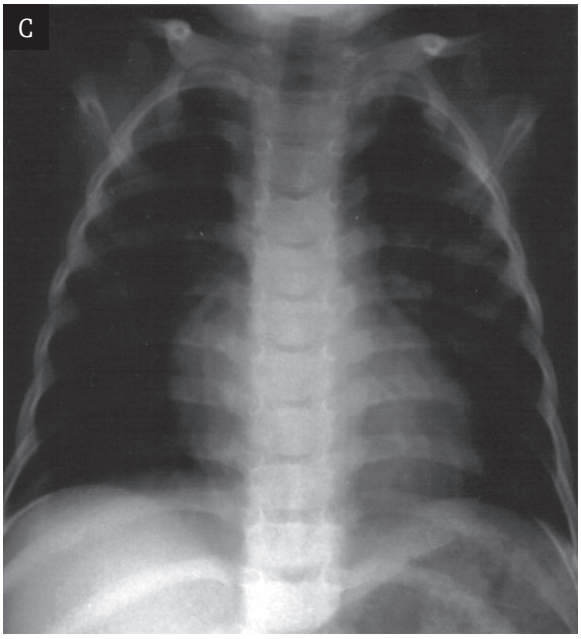
At the age of 3 months he was admitted to the Mater Hospital in Brisbane with clinical features suggesting hepatomegaly. There was no evidence of chest problems. The routine blood and urine examinations and liver function tests were normal. Ultrasound of the abdomen was normal. Hepatomegaly was spurious secondary to small thorax.

Radiograph of the chest taken at the age of 3 years was grossly normal (Fig. 1 C). An older sister is also affected with similar, moderate, asymptomatic form of the disease.

PATIENT 2

This girl was born after a normal pregnancy and delivery. Birth weight was 3560g. Her thorax was small and narrow.





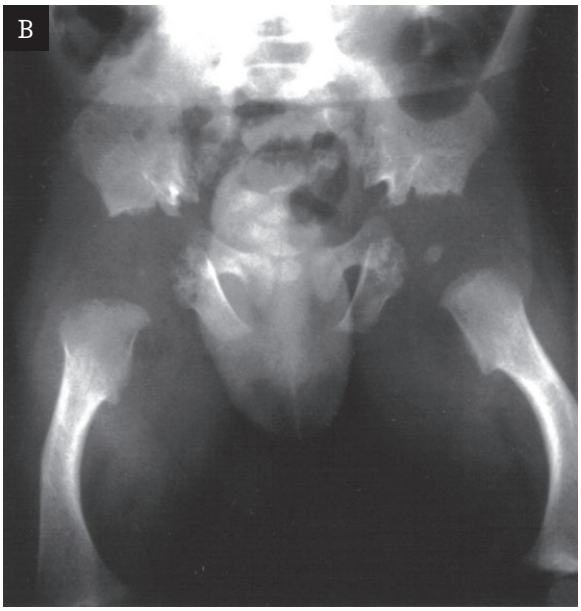


Figure 1 A-C. Patient 1. A&B Newborn **A**. Small thorax slightly narrowed in the upper part. Short, anteriorly widened ribs. **B**. Flat, triradiate acetabula. Short, bowed femora with widened ends. Advanced ossification of the capital femoral epiphyses. **C**. 3 year-old. Grossly normal chest.

Severe respiratory distress was present immediately after birth. She was admitted to intensive care unit but died at the age of 5 days.

Radiographic examination on the second day of life documented narrow, rectangular chest, characteristic pelvis and upper limb changes (Fig. 2 A-C). Postmortem bone biopsy disclosed changes consistent with ATD.

Discussion

Both our patients presented with a small thorax. Patient 1 had an asymptomatic clinical course, whereas Patient 2 with smaller and narrowed thorax succumbed to respiratory insufficiency after the first few days of life. The size and shape of the thorax is probably an important prognostic sign.

The differential diagnosis of ATD is with Chondroectodermal Dysplasia, Barnes syndrome, Short Rib Polydactyly syndromes, Neonatal Schwachman syndrome and Localised Chest dysplasia.

Major clinical signs of **Chondroectodermal dysplasia** are hexodactyly, ectodermal dysplasias and cardiac defects. Deformity of the tibia and hamate-capitate bone fusion are major radiographic signs [5]. Some authors presume that ATD, Chondroectodermal dysplasia and renal-hepatic-pancreatic dysplasia form part of a disease spectrum, rather than being distinct conditions [6].

Barnes syndrome – thoraco-laryngo-pelvic dysplasia – is characterized by small thorax, small pelvis and laryngeal stenosis. The appearances of the pelvis are different from ATD and the hands are normal [7].

Thoraco-pelvic dysplasia shows close resemblance in the appearance of the chest and pelvis to Barnes syndrome but there is no laryngeal stenosis [7].

Short Rib Polydactyly syndromes are characterized by more severe chest narrowing and severely dysplastic long bones. Still birth or early fatal outcome is the usual clinical course [6, 8].

Shwachman syndrome may present at birth with respiratory distress and small thorax.

Although radiographic appearances of the chest may be confusing, normal pelvis X-rays exclude ATD and Barnes syndrome. Long bones in neonatal Shwachman syndrome may show some metaphyseal cupping, which may cause confusion with neonatal rickets [9].

Localised Chest dysplasias do not show any other bony abnormalities (10). Small thorax is a common feature of many bone dysplasias. Other clinical and radiographic

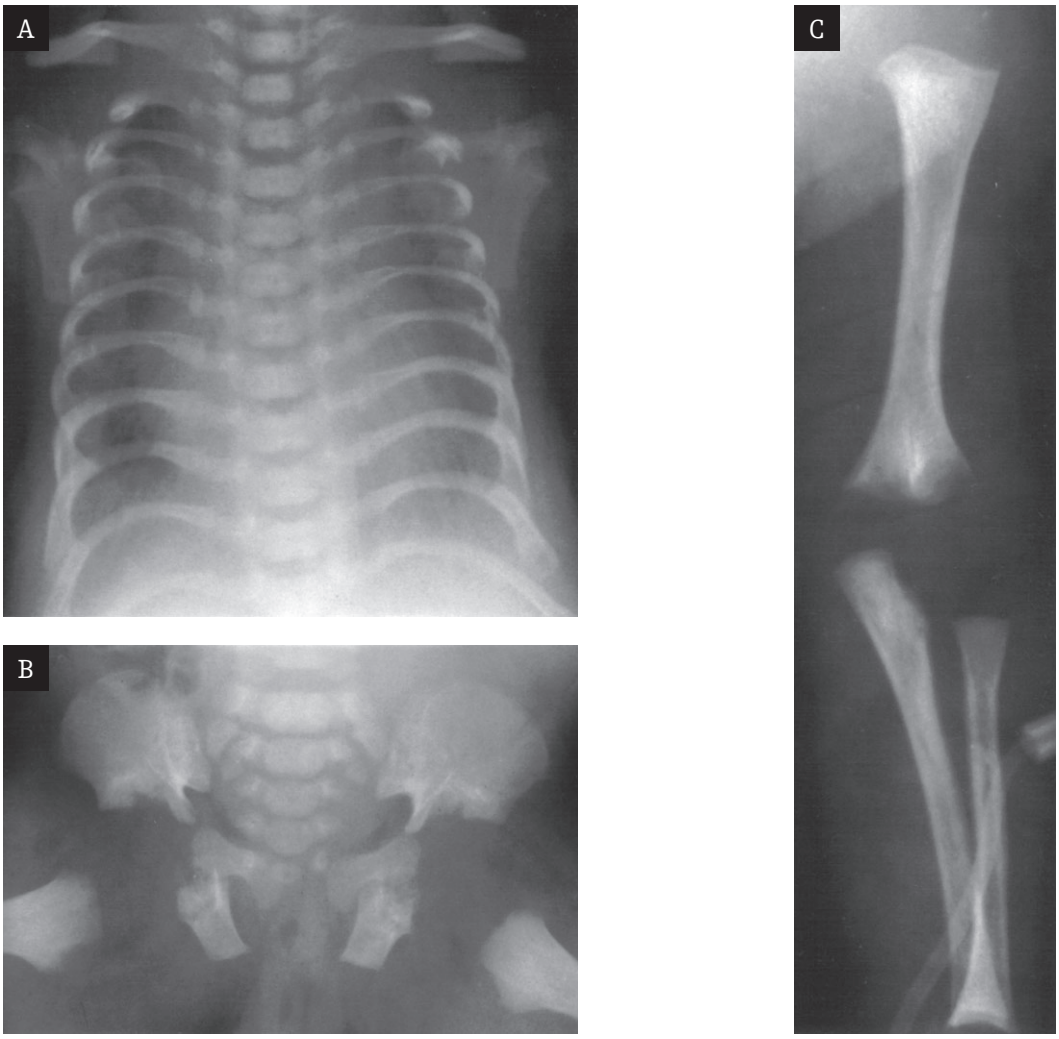


Figure 2 A-C. Patient 2. Newborn **A**. Narrow, rectangular chest, short anteriorly widened ribs. **B**. Flat, triradiate acetabula. **C**. Slightly dysplastic long bones. Cupped distal end of the forearm bones. Irregular distal humeral metaphyses.

characteristics allow easy confirmation or elimination of ATD. Ultrasound examination about 18-20 weeks of pregnancy can establish prenatal diagnosis of ATD [11].

The chromosomal location and gene defect of ATD are unknown.

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