Dysspondyloenchondromatosis in the newborn

Report of four cases

K. Kozlowski¹, K. Brostrom², J. Kennedy³, H. Lange⁴, L. Morris⁵

¹ Department of Radiology, Royal Alexandra Hospital for Children, Sydney, Australia
² Department of Radiology, Centralsygehuset Hillerod, Denmark
³ Department of Radiology, Christchurch Hospital, New Zealand
⁴ Gemeinschaftspraxis Lingen/Ems, Germany
⁵ Department of Radiology, Women’s and Children’s Hospital, Adelaide, Australia

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Abstract. Dysspondyloenchondromatosis is a rare form of generalised enchondromatosis with hypoplastic/dysplastic changes in the lower thoracic and upper lumbar spine. The disease presents at birth as neonatal dwarfism and is characterised later in life by marked shortening of stature, unequal length of the extremities and early development of kyphoscoliosis. We report four newborn babies – three boys and a girl – with dysspondyloenchondromatosis, who had skeletal survey performed shortly after birth. The condition can be established in the newborn, as the radiographic examination (skeletal survey) shows diagnostic radiographic findings.

Case reports

Case 1

A boy, the first child of healthy, normal, young Danish parents, was born at 39 weeks gestation following an uneventful pregnancy. Caesarian section was performed because of breech position. At birth his crown-rump length was 46 cm and his weight 3045 g. Short limbs with bilateral varus curving of the distal tibia and right foot were noted clinically, the latter varus position being correctable. These deformities were thought to be the result of intrauterine constraint. No other abnormality was seen.

The laboratory examinations carried out were: serum Ca, P, alkaline phosphatase, 1,25-dihydroxy-cholecalciferol, parathyroid hormone and the metabolic screening of urine. All of these were normal.

Skeletal survey on the first day of life showed shortening of all the long tubular bones with irregular, transradiant defects in the metaphyses. There was asymmetry of the long bones of the lower extremities, with the right femur and tibia being more severely affected. The knee epiphyses were not calcified. The short tubular bones were also shortened. The changes in the spine were a combination of hypoplasia and dysplasia. There was marked lumbar platyspondyly. The cervical spine showed delayed ossification. In the lumbar region there was widening of the interpediculate distances. The acetabula were triradiate. The acetabular roofs were horizontal. The iliac outlines were irregular. The ribs were slightly widened with cupping of their anterior osseous ends. The bones of

Table 1. Dysspondyloenchondromatosis: summary of clinicale data

<table>
<thead>
<tr>
<th>Case no.</th>
<th>Age</th>
<th>Sex</th>
<th>Birth length (cm)</th>
<th>Birth weight (g)</th>
<th>Abnormal features</th>
<th>Asymmetry of the lower extremities</th>
<th>Mental development</th>
<th>Hearing</th>
<th>Spine</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>5 years</td>
<td>M</td>
<td>46</td>
<td>3045</td>
<td>?</td>
<td>At birth</td>
<td>?</td>
<td>?</td>
<td></td>
</tr>
<tr>
<td>2</td>
<td>4½ years</td>
<td>M</td>
<td>47.7</td>
<td>3500</td>
<td>At 20 months</td>
<td>At 2½ years</td>
<td>Normal</td>
<td>Normal</td>
<td>At birth hypoplasia/dysplasia; minimal scoliosis at 9 months</td>
</tr>
<tr>
<td>3</td>
<td>15 months</td>
<td>F</td>
<td>39.5</td>
<td>2100</td>
<td>At birth</td>
<td>At birth</td>
<td>Developmental delay</td>
<td>Probably normal</td>
<td>At birth hypoplasia/dysplasia; scoliosis at 2½ years</td>
</tr>
<tr>
<td>4</td>
<td>7 years</td>
<td>M</td>
<td>46</td>
<td>3650</td>
<td>At birth</td>
<td>At 5 weeks</td>
<td>Normal</td>
<td>Normal</td>
<td>At birth hypoplasia/dysplasia; no scoliosis at 15 months</td>
</tr>
</tbody>
</table>

Correspondence to: K. Kozlowski, RAHC, Camperdown 2050, NSW, Australia
Case 2
A boy, the first child of healthy, young, unrelated Caucasian New Zealand parents, was born at full term after an uneventful pregnancy and assisted breech delivery. At birth his crown-rump length was 47.4 cm, and his weight 3500 g. He was noted to have short arms and legs, a barrel-shaped chest and a short neck.

The biochemical and metabolic screening tests were normal and specifically excluded hypophosphatasia. Skeletal survey on the first day of life showed changes similar to those in case 1, the only difference being that there was no long-bone asymmetry and the pelvic changes were slightly less marked.

Hypertelorism, prominent epicanthic folds and an alternating convergent squint were noted at 20 months. At a follow-up examination at the age of 2\(\frac{1}{2}\) years, a short left-sided scoliosis was present with discrepancy in the leg length, the left leg being shorter (Fig. 2). A review at the age of 4\(\frac{1}{2}\) years commented that his spinal and leg deformities were slightly more marked. He continued to make good intellectual progress.

Case 3
A girl, the fourth child of healthy, normal Caucasian Australian parents, was born at 40 weeks' gestation after an uneventful pregnancy. Ultrasound at term suggested only 34 weeks' gestation. Delivery was by caesarian section because of breech presentation. At birth, her crown-rump length was 39.5 cm and her weight 2100 g. Short limbs were noted. The baby had abnormal facies with uplifted palpebral fissures, telecanthus, epicanthus, epicanthus inversus, flat mid-face and abnormally folded ears. The maxillary alveolar ridges were hypoplastic and cleft palate was noted. There was also laryngotracheomalacia causing upper respiratory tract obstruction aggravated by glossoptosis. Tracheostomy was necessary.

Biochemical investigations and chromosome studies were all normal. Skeletal survey on the 4th day of life showed the same changes as cases 1 and 2. There was asymmetry of the lower extremities, the right being shorter. This was confirmed at follow-up examination at the age of 8 months (Fig. 3). Developmental delay was noted at follow-up examination at the age of 15 months.

Case 4
A boy, the second child of healthy, young unrelated German parents, was born at full term after an uneventful pregnancy. Caesarian section was performed because of prolonged labour. At birth his crown-rump length was 46 cm and his weight 3650 g. He was noted to have short extremities and abnormal facies. Asymmetry of the lower extremities - the left being shorter - was recognised at the age of 5 weeks.

Biochemical and metabolic screening tests were normal and specifically hypophosphatasia was excluded. Skeletal survey in the 5th week of life showed the same changes as in the other three cases (Fig. 4).

The child's motor development was slow. He sat at the age of 1 year and walked at 2 years. At the age of 3\(\frac{1}{2}\) years his height was 72 cm and weight 10.6 kg. His mid-face was flattened. There was hyperlordosis with scoliosis. The left lower limb was 5 cm shorter than the right. He showed some limitation in knee flexure and elbow extension. He was myopic. His mental development was normal.

Discussion
These four children presented at birth with short limb dwarfism. Lower extremity asymmetry was noted at birth in cases 1 and 3, at age 5 weeks in case 4 and at age...