The Spectrum of X-Ray Manifestations in Cockayne’s Syndrome

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Abstract. Cockayne’s syndrome is a rare familial disorder characterised clinically by premature aging, appearing progressively from the third year on. The radiological manifestations of eight affected children have been studied and summarised. It is concluded that a skeletal survey can provide a roentgenologic pattern suggesting the diagnosis, even when it is inconclusive from the clinical signs during the first years of life. The aetiology of this syndrome is unknown, but the authors postulate the possible role of a defect of thymic hormone which has been found in all their cases.

Key words: Cockayne’s syndrome - Bone deformities - Children, growth and development - Premature aging - Thymic hormone

Table 1

<table>
<thead>
<tr>
<th>Case</th>
<th>Sex</th>
<th>Age at the last X-ray (years)</th>
<th>Weight (kg)</th>
<th>Size (cm)</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>♂</td>
<td>18</td>
<td>13</td>
<td>98</td>
</tr>
<tr>
<td>2</td>
<td>♀</td>
<td>5.6/12</td>
<td>10.6</td>
<td>87.5</td>
</tr>
<tr>
<td>3</td>
<td>♂</td>
<td>8</td>
<td>9.2</td>
<td>84</td>
</tr>
<tr>
<td>4</td>
<td>♀</td>
<td>8.8/12</td>
<td>14.5</td>
<td>75</td>
</tr>
<tr>
<td>5</td>
<td>♂</td>
<td>8.3/12</td>
<td>10.4</td>
<td>86</td>
</tr>
<tr>
<td>6</td>
<td>♂</td>
<td>2.3/12</td>
<td>8.5</td>
<td>75</td>
</tr>
<tr>
<td>7</td>
<td>♀</td>
<td>4.4/12</td>
<td>9.2</td>
<td>83</td>
</tr>
<tr>
<td>8</td>
<td>♀</td>
<td>1.1/12</td>
<td>4.8</td>
<td>61</td>
</tr>
</tbody>
</table>

All radiological studies were reviewed, tabulated, and critically analysed with respect to the abnormalities described, together with their incidence, time of appearance, and progression.

Material and Methods

Eight children were investigated (Table 1): five males and three females aged 1-17 years. The diagnosis of Cockayne’s syndrome was established on the following clinical and laboratory signs:

1. Family Prevalence. The eight children belonged to five different families. Cases 1 and 2, and 5, 6, and 7 were siblings.
2. Characteristic Morphological Anomalies. Major microcephaly, thin and narrow face, frontal prominence, and loss of subcutaneous fat. Such abnormalities give the children the appearance of premature aging (Fig. 1A and B).
3. Dwarfism. Growth was studied in detail in three children (Cases 1, 2, and 6). Growth rate was normal for the first 12-18 months of life and then suddenly decreased.
4. Mental Retardation. This was noted in all cases.
5. Photosensitivity and Cataract. This was observed in one case (Case 3).
6. Absence of Renal, Digestive and Endocrine Abnormalities. Neither retinal pigmentation nor deafness, as mentioned by Cockayne [4, 5] were encountered.

Results

The following schematically presented conclusions are reached, since the following signs have an individual incidence higher than 50% in our material, providing a pattern which will be helpful in establishing the radiological diagnosis.

0364-2348/81/0007/0173/$01.00
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1. **Skull.** Microcephaly, small orbits in the presence of a dense calvarium and base. Calcification of basal ganglia present only in two out of eight cases (Fig. 2A and B).

2. **Spine.** Shortening of the total length of the spine. Thoracic kyphosis. Ovoid (pear shaped) vertebral bodies (Fig. 3). Early osteoporosis and anterior notching.

3. **Chest.** Thin and long ribs, occasionally also clavicles. Increased antero-posterior diameter (Fig. 4).

4. **Long Bones.** The diaphyses are slender with dense and relatively thick cortices, resulting in an extremely narrow medullary canal. In contrast the epiphyses and to a lesser degree the metaphyses are enlarged and osteoporotic (Figs. 5 and 6) (diaphyseo-disequilibrium). The fibulae show slight bowing with plump distal epiphyses (Fig. 7).

5. **Hands and Feet.** Short and broad. Large pseudoepiphyses of second metacarpal. Ivory epiphyses and cone shaped epiphyses. Diaphyseo-epiphysial disequilibrium of metacarpals and metatarsals. Large osteoporotic carpal and tarsal bones (Figs. 8–10).

6. **Pelvis.** Narrow appearance with “infolding” of iliac crest. Increased iliac angle. Square shape of iliac bones. Coxa valga with large osteoporotic femoral heads (Figs. 11 and 12).

7. **Soft Tissues.** Markedly reduced muscle mass and subcutaneous tissues. Because of the discrepancy the large joints appear unusually prominent.

8. **Progression of the Lesions.** These skeletal disorders are usually not apparent on roentgen films before

18 months. In our Patient 8, 14 months old, the skeleton appears roentgenographically normal. From the third year on, these individual skeletal findings become progressively more pronounced. At four years they can be present in an already advanced form.