A case of dyschondrosteosis from Roman Britain

Editor—Dyschondrosteosis (Leri-Weill syndrome) is an autosomal dominant condition in which mesomelic short stature is associated with Madelung deformity of the radius. The expression of the condition is variable but consistently more severe in females and generally first appears in late childhood. Patients with dyschondrosteosis (DCS) have been found to have deletions and mutations of the short stature homeobox containing gene (SHOX). Homozygous mutations at the DCS locus result in the more severe Langer type of mesomelic dwarfism.

I report here a case of DCS discovered in a skeleton from the Romano-British period, which may be the earliest case known at present.

The skeleton was one of a small number of isolated burials excavated in 1998 from Crowland Road, Gloucester. The burials were in shallow graves, typically 0.4 m below the modern ground level and associated with structures dating to the 2nd or 3rd centuries AD. None of the burials was orientated west-east and therefore may be of pagan rather than Christian origin. Unfortunately the skeleton was damaged by earth moving equipment when the site was stripped and some further damage was sustained during excavation. Nevertheless, a photograph taken when the skeleton was still in situ clearly shows a Madelung deformity of the right forearm (fig 1).

During recovery, some of the bones were damaged so that the amount of the skeleton which was available for examination in the laboratory was less than shown in fig 1. Nevertheless, it was clear that the skeleton was that of a robust male judging from the morphology of the pelvis and the skull. The left side of the skeleton had suffered particularly badly when the topsoil had been stripped off and only fragments of the left leg bones had survived. In addition, the left humerus had been broken and only a fragment of the left radius was recovered. Many of the vertebrae had been damaged when being removed from the soil and so had most of the proximal carpals, including the lunates, and some of the tarsals and other small bones of the hands and feet. The skull had been shattered but most of the individual elements seemed to be present.

The man was aged between 20 and 25 years of age at the time of death judging from the state of dental eruption, the expected mesomelic impairment. The height of the man, calculated from the combined maximum lengths of the vertebrae both to confirm the sex and to see if it was substantially shorter than expected, was 1.56 (± 0.3) m; for comparison, the mean height of males from a number of Romano-British contexts is 1.70 (± 0.3) m (author’s unpublished data).

The lengths of the intact long bones and the estimated length of the left humerus are shown in table 1, with expected lengths for males from the Romano-British period. The length of the left humerus is very close to expected, but the right is clearly substantially shorter. The lengths of the radius and ulna are much shorter than expected and the right ulna is longer than the ipsilateral radius.

In the lower limb, although the right femur is substantially shorter than expected, the length of the ipsilateral tibia is relatively much shorter, showing the expected mesomelic impairment. The height of the man, calculated from the combined maximum lengths of the right femur and tibia was 1.56 (± 0.3) m; for comparison, the mean height of males from a number of Romano-British contexts is 1.70 (± 0.3) m (author’s unpublished data).

An attempt was made to extract DNA from one of the vertebrae bodies both to confirm the sex and to see if it was possible to carry out studies of SHOX mutations, but unfortunately no DNA had survived in this skeleton (G M Taylor, personal communication).

Examples of dwarfism are rarely encountered in human remains and the total number of reported cases is probably less than 20. The earliest known case seems to be that of an adolescent male from the Upper Palaeolithic in Italy, which was thought to represent acromesomelic dysplasia. Other cases include achondroplasia, cleidocranial dysostosis,
pituitary dwarfism, and two cases of spondyloepiphysial dysplasia congenita with mild coxa vara from a Bronze Age site in Sweden which were thought to have been related. The present case is the first case of DCS to be described in archaeological material as far as I am aware.

Criteria for the classification of DCS have been published by a number of authors. Langer suggested that shortening of the radius, triangulation of the distal radial epiphysis, and wedging of the carpal bones were pathognomonic of Madelung deformity and he considered that Madelung deformity and DCS were the same condition. Other authors have added additional criteria for the diagnosis of DCS, however. Mesomelia is a sine qua non of the condition and the shortening of the limbs is usually more extreme in the arms than in the legs. It is noted that the ulna is equal to or greater in length than the radius and that the fibula is equal to or shorter in length than the tibia. The present case meets many of the criteria necessary for the diagnosis of DCS although unfortunately many of the carpal bones were not recovered during the excavation and the fibulae were too damaged to be able to estimate their length relative to the tibiae. Given the changes in the distal articulation of the radius, it seems reasonable to suppose that the carpal bones would have been wedged between the abnormal articulation of the radius and the ulna as typically described. The changes in the humerus have occasionally been described in DCS and do not militate against the diagnosis. An exostosis on the proximal end of the tibia, such as was seen in this case, is another inconstant finding in DCS and spur formation may accompany other forms of mesomelic shortening of the upper limbs. Taken together, the morphological features of this skeleton seem adequately to confirm the diagnosis of DCS.

This man was in his late teens or early twenties when he died judging from the fact that the iliac crest was still fusing and that the clavicle was unfused. DCS is not known to

### Table 1
Lengths of long bones (mm) from skeleton with dyschondrostosis

<table>
<thead>
<tr>
<th>Bone</th>
<th>Observed length</th>
<th>Expected length* (SD)</th>
<th>Ratio O/E</th>
</tr>
</thead>
<tbody>
<tr>
<td>Left humerus</td>
<td>310†</td>
<td>327 (17)</td>
<td>0.95</td>
</tr>
<tr>
<td>Right humerus</td>
<td>246</td>
<td>333 (19)</td>
<td>0.74</td>
</tr>
<tr>
<td>Right radius</td>
<td>175</td>
<td>245 (15)</td>
<td>0.71</td>
</tr>
<tr>
<td>Left ulna</td>
<td>186</td>
<td>264 (15)</td>
<td>0.70</td>
</tr>
<tr>
<td>Right ulna</td>
<td>200</td>
<td>267 (16)</td>
<td>0.75</td>
</tr>
<tr>
<td>Right femur</td>
<td>413</td>
<td>456 (25)</td>
<td>0.91</td>
</tr>
<tr>
<td>Right tibia</td>
<td>302</td>
<td>366 (23)</td>
<td>0.83</td>
</tr>
</tbody>
</table>

*Expected lengths based on observations from male skeletons from various Romano-British sites (author's unpublished data).
†Estimated.

Figure 2 Right radius and ulna showing Madelung deformity and shortening of distal radioulnar joint. The head of the radius was lost during excavation and the postmortem break in the olecranon process has been repaired.

Figure 3 Right humerus showing shortening and abnormality of head. The postmortem break in the shaft has been repaired.
be associated with premature death and several cases have been reported in adults.\(^1\)\(^2\)\(^3\) There were no signs on the skeleton to suggest what the cause of death might have been, although this is, unfortunately, frequently the case when examining skeletal material.

The examination of human remains has relevance to clinical practice since it may provide new information about the pathology of a condition, as in the case of leprosy, for example.\(^4\)\(^5\) It may indicate ways in which the expression of a particular disease has changed over time and perhaps relate these to social or environmental factors, such as in osteoarthritis,\(^6\) or it may reveal clues to the antiquity of a disease within a population. The antiquity of the present case shows that the genotype necessary for the expression of DCS has been within the British population for not less than 1700 or 1800 years. The further study of human remains from other sites may show that its first appearance can be backdated even further.

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