MADELUNG'S DEFORMITY
A Clinical and Cytogenetic Study

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Madelung in 1879 described a painful deformity of the wrist which occurs in adolescence with no history of previous injury or infection. The distal growth plate of the radius, which is the primary site of the abnormality, has premature fusion of the antero-medial quadrant. As growth proceeds the characteristic deformity is produced which is now associated with the name of Madelung. The articular surface of the distal radius is tilted abnormally antero-medially, the ulna becomes disproportionately long and its distal epiphysis is distorted and subluxated posteriorly, and the lunate bone comes to lie deeply between the radius and ulna with consequent distortion of the proximal carpal row. Clinically the wrist has a characteristic appearance (Fig. 1). The lower end of the ulna is unduly prominent, and there may be quite marked shortening of the radius so that the carpal bones are deviated laterally. When the forearm and hand are viewed from the side the wrist lies in a plane anterior to the forearm.

This condition is part of an interesting group of epiphysial growth disturbances, the other main sites being the proximal tibial epiphysis and the proximal femoral epiphysis.

CLASSIFICATION
The term "Madelung's deformity" is now used to describe a variety of conditions at the wrist in which there is, to a greater or lesser extent, premature fusion at the distal growth plate of the radius with a consequent disproportionate overgrowth of the ulna. A classification from the etiological viewpoint includes: 1) Post-traumatic deformities—These lesions, although comparatively rare, occur most often after displacement of the distal radial epiphysis. The growth plate is usually damaged throughout its length and not just in one quadrant. 2) Bone dysplasia—The commonest of these is probably diaphyseal aclasis. Although the distal articular surface of the radius is tilted, the ulna is not usually lengthened. 3) Chromosome abnormalities—Kosowicz (1960) mentioned Madelung's deformity as occasionally occurring with other skeletal abnormalities in patients with gonadal dysgenesis or Turner's syndrome (Turner 1938). 4) Idiopathic—In this group the condition seems to occur in isolation and should be called "Madelung's disease" (Burrows 1937). Only patients in groups 3 and 4 are considered in this paper.

CLINICAL MATERIAL
Details of seven patients in the idiopathic group and one with probable gonadal dysgenesis seen in Jamaica are shown in Table I. All were female. In two patients the lesion was unilateral, but there was no obvious difference radiologically between the bilateral and
unilateral disease. A full skeletal survey of the patients in the idiopathic group revealed no other bony abnormality. Five of the patients were in late adolescence. One patient aged fifty-four attended hospital with a fractured humerus and the wrist deformities were noted incidentally. Female preponderance has been found in most previous reports, and, in fact, one may doubt whether the fully developed idiopathic condition ever occurs in the male.

The patient in Case 8 was a girl aged twenty-three who complained of "lumps" on the back of both wrists (Figs. 2 and 3). She was of normal intelligence and four feet six inches tall. The secondary sex characteristics were undeveloped and she had never menstruated. Radiographs of the knees showed beaking of the medial side of the upper tibial metaphysis,
most marked on the left (Fig. 4) and described by Kosowicz (1960) as occurring in some cases of gonadal dysgenesis. She also had the typical short fourth metacarpal bone of gonadal dysgenesis (Archibald, Finby and De Vito 1959), and a mild Madelung-type deformity (Kosowicz 1960) (Fig. 5). In addition she had symptomless congenital dislocation of the left hip (Fig. 6) which is an extremely rare condition in the Negro. It could be argued that a diagnosis of gonadal dysgenesis was not conclusive without laparotomy to inspect the internal gonadal tissues or examination of the urine for oestrogen or 17-ketosteroid content. Laparotomy was not considered justified and facilities were not available for estimation of urinary oestrogen or 17-ketosteroids at the time of this study. Nevertheless the authors submit that the diagnosis is virtually certain from the clinical, radiological and cytogenetic findings (Table II).

SYMPTOMS AND SIGNS

Most patients complained of aching pain on using the wrist and of the unsightly deformity. One patient said that the pain had preceded the deformity by some years. Four patients found

### TABLE I
**DETAILS OF PATIENTS**

<table>
<thead>
<tr>
<th>Case number</th>
<th>Sex</th>
<th>Age in years</th>
<th>Side</th>
<th>Comments</th>
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<tr>
<td>1</td>
<td>Female</td>
<td>14</td>
<td>Left</td>
<td>—</td>
</tr>
<tr>
<td>2</td>
<td>Female</td>
<td>13</td>
<td>Left</td>
<td>—</td>
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<tr>
<td>3</td>
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<td>15</td>
<td>Bilateral</td>
<td>—</td>
</tr>
<tr>
<td>4</td>
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<td>17</td>
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<td>—</td>
</tr>
<tr>
<td>5</td>
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<td>23</td>
<td>Bilateral</td>
<td>—</td>
</tr>
<tr>
<td>6</td>
<td>Female</td>
<td>54</td>
<td>Bilateral</td>
<td>Noted incidentally</td>
</tr>
<tr>
<td>7</td>
<td>Female</td>
<td>16</td>
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<td>Short stature</td>
</tr>
<tr>
<td>8</td>
<td>Female</td>
<td>23</td>
<td>Bilateral</td>
<td>Short stature, Turner's syndrome</td>
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</table>

### TABLE II
**SEX CHROMATIN AND CHROMOSOME COUNTS**

<table>
<thead>
<tr>
<th>Case number</th>
<th>Sex chromatin per cent positive</th>
<th>Chromosome counts</th>
<th>Comments</th>
</tr>
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<tbody>
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<td></td>
<td>44 or less</td>
<td>45 46 47 Total cells</td>
<td></td>
</tr>
<tr>
<td>3</td>
<td>28</td>
<td>3 0 12 0 15</td>
<td>Poor culture presumptive 46XX</td>
</tr>
<tr>
<td>4</td>
<td>20</td>
<td>0 1 19 0 20</td>
<td>46XX</td>
</tr>
<tr>
<td>5</td>
<td>17</td>
<td>0 0 20 0 20</td>
<td>46XX</td>
</tr>
<tr>
<td>7</td>
<td>31</td>
<td>1 2 26 1 30</td>
<td>46XX</td>
</tr>
<tr>
<td>8</td>
<td>6, 12, 7</td>
<td>10 30 19 1 60</td>
<td>45XO/46XX slight chance of a small 47XXX line</td>
</tr>
</tbody>
</table>
Case 8—The pelvis showing the old-standing congenital dislocation of the hip.

Case 8—To show the wrists and hands, with the short fourth metacarpal bones and mild Madelung's deformities.
the deformity the worst feature. On examination, besides the typical deformity already described, wrist movements were characteristically abnormal: dorsiflexion was very limited whereas palmar flexion was increased; pronation was blocked by the lower end of the ulna, but supination was usually full.

**RADIOLOGY**

In the seven patients in the idiopathic group the radiological appearances were uniform. Figures 7 and 8 show a typical example. In the antero-posterior radiograph the lower end of the ulna is enlarged and distorted and the epiphysis is abnormally dense; the growth plate is sloping. The proximal carpal row, instead of being smoothly convex proximally is V-shaped with the lunate bone forming the apex. The growth plate of the distal radius slopes sharply in a proximo-medial direction at about its centre and becomes fused. At the site of fusion on the medial cortex there is a characteristic "beak" closely resembling that seen in an analogous position in tibia vara. In the lateral radiograph there can be seen the increased anterior inclination of the articular surface of the radius: this is continued through the lunate bone. The articulation of the radius and lunate bone being abnormally proximal, it is obscured by the radial styloid in the lateral view. The defect in the radius shows as a triangular and relatively translucent area.

**CYTOGENETIC STUDIES**

Because of the association of Madelung's deformity with short stature in two patients it was felt worthwhile to investigate the possibility of abnormality of the sex chromosomes. Buccal smears and chromosome studies were done in five patients. Buccal smears were stained with thionine and at least 100 cells were counted. The results are given in Table II. Chromosome studies were made on cultured lymphocytes from peripheral blood. The method used was
the microtechnique of Arakaki and Sparkes (1963). Air dried smears were stained with orcein from all patients except those from Case 8, which were stained with Giemsa (Table II). As can be seen, four patients had sex chromatin and chromosome counts within the range for the normal female, though it might be argued that insufficient cells were counted in the patient

in Case 3 to exclude mosaicism. Unfortunately the latter patient did not return for a repeat culture as requested. The patient in Case 8, however, is of the greatest interest. As was suspected from her clinical appearance, there was sex chromosome mosaicism, with the major line being 45XO in the peripheral blood (Figs. 9 and 10). There was also the possibility of a further small line of 47XXX cells which we have been unable to confirm as yet.

TREATMENT

In most of the patients, symptoms were not sufficient to warrant treatment. Conservative measures are of little avail and for those in whom pain or deformity is a handicap, some form of surgery is advisable. Pain is probably caused in two ways: by the mechanical derangement of the radio-ulnar joint and by degenerative changes in the opposing articular surfaces of the lunate bone and radius where they tend to be compressed on dorsiflexion of the wrist. This was clearly seen at operation on one patient in whom the articular cartilage was severely fibrillated and eroded.

Several types of operative procedure have been described in the literature. Burrows (1937) suggested an operation in which the juxta-epiphysial part of the shaft of the ulna and the epiphysial disc were excised: a linear osteotomy of the distal radius completed the procedure. Initially the result was good but no account has been found of this technique having been used elsewhere. The usual operation is subperiosteal resection of the distal one inch of the ulna with reconstruction of the medial ligament of the wrist (Darrach 1936). This will restore pronation but does not prevent radio-carpal articular damage and there is a possibility of the carpus progressively subluxating medially off the lower end of the radius.

The procedure used in two patients in this series was arthrodesis of the radio-carpal joint by the wedge technique in addition to excision of the distal end of the ulna. This operation was attributed to Brockman by Evans (1955). It is here suggested as the best operation for idiopathic Madelung's deformity. The unsightly "knob" is removed and the threat of painful osteoarthritis in the radio-carpal joint is prevented. A fair range of flexion and extension may be maintained by preservation of the mid-carpal joint. One patient (Case 1) operated on in 1957 by this method was reviewed in 1964. She had a strong, painless wrist with full
pronation and supination, 60 degrees of palmar flexion and 10 degrees of dorsiflexion (Figs. 11 and 12). The second patient (Case 3) was operated upon in 1964 and has so far had a successful result.

**FIG. 11**

Case 1—Antero-posterior and lateral radiographs of the wrist seven years after wedge arthrodesis.

**FIG. 12**

**DISCUSSION**

Madelung's deformity, like tibia vara, appears to be more common in the West Indies than elsewhere although it is still rare. Langenskiöld and Riska (1964) reported seventy-one cases of tibia vara from Finland and it would be interesting to know the incidence of idiopathic Madelung's deformity in that country. The radiological appearances in the radius are very similar to those found in the proximal tibia in tibia vara as shown by Blount (1937), Golding and McNeil-Smith (1963) and Langenskiöld and Riska (1964); in this condition the posteromedial quadrant of the epiphysis is affected so that the deformity is produced by the continued growth of the unaffected part. However, at the wrist severe varus is prevented by the buttressing effect of the ulna.

The underlying basis of idiopathic Madelung's deformity has never been explained. Brailsford (1953) suggested a possible localised osteochondrodystrophy and drew a parallel with infantile coxa vara. The similarity of these three conditions is striking, but the part played by abnormal weight-bearing stresses on the growth plate of the proximal tibia and femur cannot be operative in idiopathic Madelung's deformity.

It is well recognised that other abnormalities of the bones and joints besides Madelung's deformity may accompany Turner's syndrome. There have been several reports of large series of cases of gonadal dysgenesis with extensive study of the clinical and cytogenetic manifestations and the radiological findings (Levin 1962; Astley 1963; Jones, Ferguson-Smith and Heller 1963; Lemli and Smith 1963; Ferguson-Smith, Alexander, Bowen, Goodman, Kaufman, Jones and Heller 1964; Engel and Forbes 1965). Among a wide range of skeletal abnormalities the authors have been unable to find any record of congenital dislocation of the hip which, as has been mentioned, is extremely rare in the Negro.
Madelung's Deformity

It is possible that the association of Turner's syndrome and Madelung's deformity is a fortuitous one. Family histories of these patients (as far as they can be ascertained) do not suggest X-linked inheritance, because the only patient with a positive family history (maternal grandmother of the patient in Case 7) has normal males in the intervening generation. It is of interest that all the patients in this series are females and that two of them are of short stature. This suggests a sex-modified inheritance. It is, therefore, reasonable to consider the possibility of Turner's syndrome when investigating girls of short stature with Madelung's deformity.

SUMMARY

1. A classification of Madelung's deformity on an etiological basis is suggested.
2. Seven patients with "idiopathic" Madelung's deformity and one occurring in association with Turner's syndrome are discussed.
3. Cytogenetic studies of five patients are recorded and their significance discussed.
4. Arthrodesis of the radio-carpal joint by the "wedge" technique is preferred for those patients whose symptoms warrant treatment.

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REFERENCES


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