Original article

Radiological and clinical analysis of Madelung's deformity in children

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A B S T R A C T

Introduction: Madelung's deformity is a bone dysplasia that occurs predominantly in adolescent females, characterized by early epiphyseal growth arrest in the medial part of the distal radius. This leads to an upward and medial displacement of the radial joint surface, restricting range of motion.

Objectives: The objective of this study was to determine whether there was a link between clinical and radiological data in children with Madelung's deformity and to test the hypothesis of a relation between the deformity and a genetic mutation.

Methods: A retrospective study recruited 13 patients with Madelung's deformity, with a mean age of 13.2 years (range, 8–18 years). Assessment comprised level of pain, range of motion and grip force, with standard AP and lateral wrist X-rays. Every patient except one underwent molecular genetic screening, adhering to current recommendations.

Results: Pronation-supination, radial inclination and grip force were significantly impaired compared to normal results. All X-ray measurements were significantly abnormal, except for the lunate-covering ratio. Genetic mutation (SHOX) was systematic in the 12 patients screened.

Discussion: Radiological deformity did not correlate with functional disturbance or pain. Non-acquired Madelung's deformity requires molecular screening for SHOX or XO mutation, which definitively diagnoses Léri-Weill dyschondrosteosis or Turner syndrome.

Conclusion: A larger series is necessary to confirm these preliminary results, which nevertheless suggest that non-acquired Madelung's deformity is not isolated but syndromic. Early detection of Léri-Weill or Turner syndrome is essential, due to their therapeutic specificities.

Level: IV.

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1. Introduction

Madelung's deformity was in fact first reported by Malgaigne in 1855. In 1878, Madelung gave a precise description, with suggested etiology and treatment [1–3]. Etiology remains unknown, but various hypotheses have been made. Early epiphyseal growth arrest is to be distinguished according to whether it is congenital or acquired (post-traumatic, infectious, etc.) [4].

The present study of the congenital (as opposed to acquired) forearm and wrist malformations known as Madelung's deformity sought to determine precise radiographic identification and measurements to assess possible syndromic associations.

Léri-Weill syndrome, or dyschondrosteosis (SHOX gene mutation), and Turner syndrome (X0 karyotype) have been reported as being potentially associated, although the correlations were not clearly assessed. Léri-Weill syndrome is a mutation of the SHOX gene, lying in the pseudo-autosomal regions of the X and Y chromosomes, with dominant autosomal transmission. Patients present a highly characteristic phenotype, associating moderate mesomelia with Madelung's deformity [4,5]. Turner syndrome comprises small stature and varying bone abnormalities.

The hypothesis of a correlation between radiologic Madelung's deformity and SHOX gene or X0 mutation was tested, and the correlation estimated.

2. Materials and methods

A retrospective analysis, including all congenital Madelung's deformity patients followed-up in our center between 2005 and 2013, was jointly performed by the Children's Orthopedic Surgery
and Clinical Genetics Departments. There were 13 patients, with ages ranging from 8 to 18 years (mean, 13.2 years); age, gender, deformity size and uni-/bi-laterality were noted.

Morphotype (mesomelic) was systematically described, with any associated upper- or lower-limb deformities. All patients except one underwent genetic examination for molecular analysis to screen for SHOX gene and/or X0 mutation. This analysis adhered to current ethics committee recommendations, with the parents’ written consent.

All cases of Madelung’s deformity clearly identified as acquired on history-taking were excluded.

Study variables included pain on Visual Analog Scale (VAS), grip force (in kg) on Jamar dynamometer, and joint range of motion (°) on goniometer: flexion/extension, pronation/supination radial/ulnar inclination. Subjective esthetic discomfort was also assessed.

Radiographic analysis comprised objective criteria, assessed on AP and lateral wrist views:

- lunate fossa angle (normal < 40°) on AP view: angle between the ulnar axis and the line through the radial lunate fossa (Fig. 1);
- radial height (normal, 10 mm) on AP view (Fig. 2);
- lunate-covering ratio (radial coverage of the lunate fossa) (normal, 70–100%) on AP view (Fig. 3);
- ulnar head dorsal translation index (normal, 0 mm) on lateral view: distance between the ulnar and radial axes (Fig. 4).

The usual statistical tests were not applied, due to the small size of the series. For each clinical and radiological variable, a significance threshold was set at ≥ 20% difference from normal values.

3. Results

Finally, 13 patients were included; all were female, with a mean height of 139 cm (range, 115–153 cm); i.e., 1 standard deviation from normal-for-age values. All had bilateral wrist deformity. Three were at end of growth. All reported subjective esthetic blemish.

SHOX gene mutation was found in 9 patients. Three others had Turner syndrome, confirmed on molecular analysis (X0). Molecular analysis could not be performed in 1 patient, who had, however, a clinical presentation typical of Léri-Weill syndrome, with mesomelia (Fig. 5). The molecular mutation rate in the 12 patients tested was thus 100%.

Only 2 patients (15.4%) reported VAS > 3, with no associated radiographic feature. Mean grip force was 10 kg, compared to a mean 23 kg normal value depending on age; this difference was significant. Regarding range of motion, only radial inclination and pronation-supination were significantly impaired. Table 1 shows mean values.

All radiographic variables were significantly impaired, except lunate-covering ratio, which was nevertheless below normal. Table 2 shows mean values.
Table 1
Mean clinical values.

<table>
<thead>
<tr>
<th></th>
<th>Flexion</th>
<th>Extension</th>
<th>Radial inclination</th>
<th>Ulnar inclination</th>
<th>Pronation</th>
<th>Supination</th>
</tr>
</thead>
<tbody>
<tr>
<td>Normal (degrees)</td>
<td>80</td>
<td>90</td>
<td>20</td>
<td>40</td>
<td>90</td>
<td>90</td>
</tr>
<tr>
<td>Madelung's deformity (degrees)</td>
<td>70</td>
<td>77.5</td>
<td>14.7</td>
<td>37.5</td>
<td>70</td>
<td>50</td>
</tr>
<tr>
<td>Difference (degrees)</td>
<td>10</td>
<td>12.5</td>
<td>5.3</td>
<td>2.5</td>
<td>20</td>
<td>40</td>
</tr>
<tr>
<td>% difference</td>
<td>12.5</td>
<td>13.88</td>
<td>26.5</td>
<td>6.25</td>
<td>22.22</td>
<td>44.44</td>
</tr>
<tr>
<td>Significance (20%)</td>
<td>Negative</td>
<td>Negative</td>
<td>Positive</td>
<td>Negative</td>
<td>Positive</td>
<td>Positive</td>
</tr>
</tbody>
</table>

Table 2
Mean radiographic values.

<table>
<thead>
<tr>
<th></th>
<th>Lunate fossa angle</th>
<th>Radial height</th>
<th>Lunate-covering ratio</th>
<th>Ulnar head dorsal translation index</th>
</tr>
</thead>
<tbody>
<tr>
<td>Normal</td>
<td>&lt; 40 degrees</td>
<td>10 mm</td>
<td>70–100%</td>
<td>0 mm</td>
</tr>
<tr>
<td>Madelung's deformity</td>
<td>50.4 degrees</td>
<td>17.3 mm</td>
<td>58.9%</td>
<td>7.8 mm</td>
</tr>
<tr>
<td>Difference</td>
<td>10.4 degrees</td>
<td>7.3 mm</td>
<td>11.1%</td>
<td>7.8 mm</td>
</tr>
<tr>
<td>% difference</td>
<td>26</td>
<td>73</td>
<td>15.8</td>
<td>–</td>
</tr>
<tr>
<td>Significance (20%)</td>
<td>Positive</td>
<td>Positive</td>
<td>Negative</td>
<td>Positive</td>
</tr>
</tbody>
</table>

Fig. 4. Ulnar head dorsal translation index: distance between the ulnar and radial axes on lateral view.

Fig. 5. Patient not tested on molecular analysis but presenting clinical mesomelic dwarfism.

4. Discussion

Madelung’s deformity is usually diagnosed at 8 to 13 years of age, with onset of painful or painless progressive wrist deformity or in the context of known familial history [1]. It is 3 times as frequent in girls as boys; the present series included no boys. It is defined as early epiphyseal growth arrest in the medial part of the distal radius, resulting in anterior and medial displacement of the radial joint surface, radius bowing, with the carpus frontally triangular and laterally subluxated forward, and posterior projection of the ulnar head [6,7]. It restricts the range of motion of the wrist, especially in pronation-supination and radial inclination, as can be seen from the present results, due to disorganization of the distal radio-ulnar joint and verticalization of the radial joint surface. The other radiographic deformities, such as excessive anteversion or reduced lunate coverage, however important they may be in themselves, did not affect the other ranges of motion. Clinical tolerance is generally good, despite significantly impaired grip force. In the present results, there was no correlation between pain variables and severity of radiographic deformity. Standard X-ray is thus insufficient in these patients for exploring pain, which does not exclusively implicate the deformity: pain may be caused by changes in other structures, possibly ligamentous, and MRI or CT-arthrogram, which were not performed in the present series, could shed light on the origin of pain.

Although aspect and clinical impact may be non-determining, AP and lateral wrist X-ray is enough for diagnosis. McCaroll et al. [8] and Turder et al. [7] describe various useful diagnostic radiographic indices: ulnar inclination (angle between the longitudinal axis of the ulna and the tangent to the proximal scaphoid and lunate bone surfaces (positive if >33°), lunate bone collapse (positive if >4 mm), lunate fossa angle (positive if >40°) and palmar carpal displacement (positive if >20 mm). However, they considered only lunate fossa angle to be contributive in diagnosing early forms. We therefore used this index to analyze childhood deformity, and determined other radiographic criteria adapted to early forms and thus to childhood deformity: radial height, lunate-covering ratio, and lunate dorsal translation index. These correspond to abnormalities with earlier onset than for the other indices described in adults, which are established forms, whereas in children, the deformity becomes progressively more severe, precisely because of the medial epiphyseal growth arrest. Early radiological diagnosis seems to us to be primordial: from the present results, it should lead to
molecular analysis which, if positive, would indicate medical management of the mesomelia.

Madelung's deformity is a rare pathology, and etiology remains unclear. The present series included no isolated congenital deformities, such as have exceptionally been reported [2,4]: all the present patients presented syndromes, either Léri-Weil or Turner, as was systematically confirmed on molecular analysis in all 12 cases.

The present findings are in line with those of Lukas et al. [2], who found 17 dyschondrosteoses in 22 patients with typical Madelung's deformity (77%). However, no genetic analysis was performed in their study: dyschondrosteosis was diagnosed from the association of Madelung's deformity, mesomelic micromelia and small stature, and the few isolated cases may have been minor phenotypic forms of dyschondrosteosis or of Turner syndrome.

In our opinion, isolated congenital Madelung's deformity does not exist. This idea is founded on the studies by Rosilio et al. [9] and Salmon-Musial et al. [10], who examined Madelung's deformity in two patient populations, with and without SHOX mutation: Madelung's deformity was significantly more frequent in the former (50 vs 9.2%, \( P < 0.01 \)). They did not, however, test for \( X0 \) mutation, and the patients without SHOX mutation may have had overlooked the Turner syndrome.

In Léri-Weil and Turner syndrome, mesomelia may be slight and overlooked [2]. Madelung's deformity may be the only presenting symptom. Genetic screening for SHOX or \( X0 \) mutation should therefore, we consider, be systematic in case of Madelung's deformity, especially in children of significantly small stature, as treatment by growth hormone is effective in both Turner syndrome, Léri-Weil dyschondrosteosis.

5. Conclusion

Analysis of the present series highlighted the difficulty of correlating not only radiological and clinical data but also genotype and phenotype: radiological deformity did not systematically correspond to functional disorder or pain, and the functional impairment found in certain cases would require complementary analysis of structural lesions, preferably by MRI. In non-acquired Madelung's deformity, molecular study is necessary, especially in children, to screen for SHOX or \( X0 \) mutation, due to the implications for subsequent treatment. In case of positive screening, parents should be educated regarding the mode of transmission of the disease, its consequences and the possibilities of specific treatment of small stature if the family so wish.

Disclosure of interest

The authors declare that they have no conflicts of interest concerning this article.

References