Madelung's Deformity: A Spectrum of Presentation

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Purpose: To evaluate the hypotheses that all Madelung's deformity subjects have dyschondrosteosis (defined as short stature and mesomelia, in addition to Madelung's deformity) and to evaluate the concept that Madelung's deformity may affect the entire radius.

Methods: A radiographic and medical records review was performed for 26 subjects (46 extremities) with Madelung's deformity. The radiographs were assessed for radius and ulna length, sagittal radial bow, severity of the Madelung deformity, and radiocapitellar joint space. The sagittal radial bow and the radiocapitellar joint space were used to classify subjects according to whether the Madelung deformity was limited to the distal radius or involved the entire radius.

Results: Thirty-one extremities in 18 subjects were classified as having a distal radius Madelung deformity and 15 extremities in 8 subjects were classified as having an entire radius Madelung deformity. The radius and ulna length and subject height were significantly decreased compared with age- and height-matched normal values in both groups; the entire radius group was more severely affected. In addition, the entire radius group had more severe deformities with respect to lunate subsidence and ulnar tilt. All of the entire radius subjects and 9 of 14 of the distal radius subjects had dyschondrosteosis.

Conclusions: Madelung's deformity presents as a spectrum. It may affect the entire radius or it may affect only the distal radius. Extremities with involvement of the entire radius have a shorter radius and ulna, decreased height, and a more severe deformity than extremities with involvement of only the distal radius. Additionally, most subjects with Madelung's deformity have dyschondrosteosis. (J Hand Surg 2007;32A:1393–1401. Copyright © 2007 by the American Society for Surgery of the Hand.)

Type of study/level of evidence: Diagnostic III.

Key words: Dwarfism, dyschondrosteosis, Leri-Weill, longitudinal dysplasia, Madelung's deformity.

Madelung's deformity, first described in 1855 by Malgaigne and 1878 by Madelung, is caused by the abnormal growth of the distal radius physis.[1–4] Madelung's deformity may be idiopathic, but genetic studies have demonstrated that it may be transmitted as an autosomal dominant trait with incomplete penetrance as well.[5,6] Clinical and radiographic features of Madelung's deformity have been well described: an ulnar and dorsal curvature of the distal radius (due to deficient growth of the volar and ulnar aspect of the distal radius physis), increased inclination of the distal radius joint surface, triangulation of the carpus with proximal and volar migration of the lunate, and a prominent dorsal subluxation of the ulnar head.[7–11]

Leri and Weill first described dyschondrosteosis as a developmental skeletal dysplasia characterized by mesomelia (ie, short forearm), short stature, and Madelung's deformity.[5,12–14] All 3 components must be present to classify the subject as having dyschondrosteosis. Several studies advance the concept that Madelung's deformity is always a component of dyschondrosteosis[12–14] and is never found in isolation. In contrast, other studies have concluded that not all Madelung's subjects have dyschondrosteosis and that Madelung's deformity may be an isolated condition.[8,9,15] This discrepancy in the literature suggests that the relationship between Madelung's deformity and dyschondrosteosis is not completely understood.

Most reports[16–20] have focused on the wrist deformity in Madelung subjects with limited consideration of the more proximal radius and elbow, forearm length, or subject height. It has been our observation that some Madelung subjects have abnormalities that are not limited to the distal radius physis but involve the entire radius.

The purpose of this study was twofold. First, we evaluated the hypothesis that there is a spectrum of involvement of the radius in Madelung's deformity and that, in some cases, the entire radius may be involved. Second, we investigated whether subjects with Madelung's deformity have dyschondrosteosis.

Materials and Methods

We performed a comprehensive medical record search for the years 1960 to 2006 for subjects with a diagnosis of Madelung's deformity. Institutional review board approval was obtained for this retrospective review. Inclusion criteria were a diagnosis of Madelung's deformity based on the classic description of distal radius and ulna abnormalities[7–11] and adequate preoperative radiographs of the wrist, forearm, and elbow. Subjects with idiopathic and familial Madelung's deformity were included; subjects with an acquired Madelung deformity due to trauma or infection were excluded.

A total of 32 Madelung subjects were identified, and 26 met these criteria to form the study cohort. There were 24 female and 2 male subjects; 21 were white, 4 were African American, and 1 was Hispanic. The average age at the time of radiographic assessment was 16 years (range, 9–47 years); the majority of patients were skeletally mature and, therefore, were assessed as a single group. Twenty subjects had bilateral involvement, and 6 subjects were affected unilaterally for a total of 46 affected extremities. Six subjects had a parent with Madelung's deformity (4 mothers, 2 fathers). None of the subjects had Turner syndrome or Langer's syndrome, which carry manifestations of Madelung's deformity.

Radiographic

The senior author (C.A.G.) made the following measurements and observations from preoperative anteroposterior (AP) and lateral radiographs of the elbow, forearm, and wrist for all subjects included in this investigation. All radiographs were obtained in standard fashion at our hospital to allow direct measurement without magnification error.

Madelung's deformity distal radius to the distal radius versus Madelung's deformity affecting the entire radius. The development and morphology of the radius were assessed on AP and lateral elbow and forearm radiographs. Subjects with a radiographic deformity of only the distal radius were classified as having a distal radius Madelung deformity. Subjects with abnormal morphology of the radius, specifically noted as a radius bow on the lateral radiograph of more than 10° or a radiocapitellar joint space of 4 mm or more were considered to have an entire radius Madelung deformity. We also assessed and recorded the bow of the radius on the AP radiograph, the length of the radius, and the length of the ulna, but these features were not used to classify the deformity into these two groups.

Radial bow. The radial bow was measured for each extremity on both AP and lateral radiographs. The apex of the deformity was localized and the longitudinal axis of the radius proximal and distal to the apex was drawn, thus allowing an angular measurement.

Radiocapitellar joint space. The distance between the radial head and the capitellum was measured on the AP and lateral radiographs.

Radius and ulna length. Radius and ulna lengths were measured on the AP radiograph. Ulna length was the distance from the tip of the olecranon to the base of the ulna styloid (including ulnar epiphysis). Radius length was the measured distance from the most proximal portion of the radial head to the midpoint of the distal radial articular surface. The measured lengths were compared with age-matched normal subjects.[21] Additionally, we calculated the length of the radius and ulna as a percentage of height (see later) and compared these data with age-matched normal subjects.[21]

Severity of Madelung's deformity. McCarroll et al investigated 5 radiographic measurements (ulnar tilt, lunate subsidence, lunate fossa angle, palmar tilt, and palmar carpal displacement) to assess Madelung's deformity.[19] Ulnar tilt, lunate subsidence, and palmar carpal displacement were noted to be reliable and reproducible measurements for quantifying the severity of Madelung's deformity.[19] Negative values for lunate subsidence represent a lunate that is not subsided (ie, it remains distal to distal ulna), and a negative value for palmar carpal displacement indicates that the carpus is not palmarly displaced rela-

Figure 1. (A) Anteroposterior and (B) lateral forearm radiographs of an extremity with a distal radius Madelung deformity.

tive to the long axis of the ulna. These 3 measurements were assessed in order to compare the severity of distal radius and entire radius Madelung's deformity.

Medical Record Review

The medical records of all subjects were reviewed and the following data obtained. Patient height (obtained at the time the radiographs were taken) was measured, compared with age-matched normal subjects, and expressed as a percentile of normal.[21] Elbow, forearm, and wrist range of motion were obtained from the hand therapist evaluations (also obtained when the radiographs were taken). Range of motion data were compared with accepted normal values (rather than the opposite extremity due to the high incidence of bilaterality); normal motion was defined as elbow flexion 145°, elbow extension 0°, forearm pronation 75°, forearm supination 80°, wrist extension 75°, and wrist flexion 75°.

Statistical Analysis

Clinical and radiographic data were compared between distal radius and entire radius Madelung subjects. The Student's *t*-test was used to compare these groups. A p value <.05 was considered to be significant. All collected data were analyzed with use of Microsoft Excel (Microsoft, Redmond, WA).

Results

Distal Radius Versus Entire Radius Madelung's Deformity

We classified 31 extremities in 18 subjects as having a distal radius Madelung's deformity (Fig. 1) and 15 extremities in 8 subjects as having an entire radius deformity (Fig. 2). The 6 subjects with unilateral Madelung's deformity included 5 subjects with a distal radius deformity and one subject with an entire radius deformity. There were no instances of bilateral involvement in which one extremity was classified as entire radius and one as distal radius.

Radial Bow

The radial bow on the AP radiograph for entire radius Madelung subjects (20° ± 7) was not significantly different (p = .11) from the bow in distal radius Madelung subjects (17° ± 6) (Table 1). On the lateral radiograph, the bow of the radius was significantly greater in entire radius Madelung subjects (23° ± 12 vs 2° ± 4, p = .02).

Figure 2. (A) Anteroposterior and (B) lateral forearm radiographs of an extremity with a longitudinal Madelung deformity. Note the bowing of the radius (more severe on the lateral radiograph) and the abnormal relationship between the radial head and the capitellum.

Table 1. Comparison of Deformity Between Distal Radius and Entire Radius Madelung Subjects

	Radius Bow (°)	Lateral Radius Bow (°)	Radial Head–Capitellum Distance AP (mm)	Radial Head–Capitellum Distance Lateral (mm)
18 distal radius Madelung subjects				
Average	17	2	3	3
SD	6	4	1	1
Range	5–30	0–10	2–5	2–5
8 entire radius Madelung subjects				
Average	20	23	13	10
SD	7	12	4	6
Range	5–33	10–36	2–18	1–20
p Value	.11	.02	.02	.001

Radiocapitellar Joint Space

The radial head–capitellum distance was significantly greater in entire radius Madelung subjects on both AP (13 mm vs 3 mm, $p = .02$) and lateral radiographs (10 mm vs 3 mm, $p = .001$) (Table 1).

Radius and Ulna Length

Radius and ulna length values are noted in Tables 2 and 3. In general, both distal radius and entire radius Madelung length values were less than normal values; entire radius values were less than distal radius values.

The average radius length in distal radius Madelung extremities was 18 cm ± 2, significantly ($p < .05$) different from age-matched normals (22 cm ± 2) (Table 2). The average ulna length in distal radius Madelung extremities was 20 cm ± 2, significantly ($p < .05$) less than age-matched normals (24 cm ± 2). The average radius and ulna lengths in entire radius Madelung extremities were 14 cm ± 3 and 17 cm ± 2, respectively; these were both significantly ($p<.05$) shorter than those of age-matched normal subjects (radius, 22 cm ± 2; ulna, 24 cm ± 2) (Table 3). The average radius (14 cm ± 3) and ulna (17 cm ± 2) length of entire radius Madelung extremities were significantly ($p < .05$) shorter than the average radius (18 cm ± 2) and ulna (20 cm ± 2) lengths of distal radius Madelung extremities.

The average radius and ulna length as percentage of height for both distal radius and entire radius Madelung subjects was significantly less than that of age-matched normal subjects (Tables 2 and 3). Entire radius Madelung extremities had a radius/height average percentile of 9.4 cm ± 1.7 and ulna/height average percentile of 12 cm ± 1; these were significantly ($p < .05$) smaller than average radius/height percentile average (12 cm ±1) and ulna/height percentile average (14 cm ± 1) for distal radius Madelung subjects.

In the distal radius Madelung group of 31 extremities, 5 extremities had a radius length greater than the 10th percentile and 6 extremities had an ulna length greater than the 10th percentile compared with age-matched normals, as well as compared with normal subjects by percentile of radius and ulna/height ratios (Table 2). There were two subjects with bilateral involvement in which 1 forearm was short and the other was of normal length. All radius and ulna lengths in the entire radius Madelung extremities were below the 10th percentile compared with age-matched normal subjects and compared with normal subjects by percentile of radius and ulna/height ratios (Table 3).

Severity of Madelung's Deformity

The range for ulnar tilt was 7° to 72°, the range of lunate subsidence was (–)7 mm to 35 mm, and the range of palmar carpal displacement was (–)22 mm to 40 mm (Table 4). Lunate subsidence was significantly greater ($p < .02$) in entire radius Madelung subjects (7 mm ± 10) compared with distal radius Madelung extremities (3 mm ± 7). Ulnar tilt (31° ± 18 vs 48° ± 14) ($p < .001$) was also significantly greater in entire radius Madelung extremities. There was no significant difference in palmar carpal displacement between distal radius and entire radius Madelung subjects (22 mm ± 7 vs 17 mm ± 18) ($p = .16$).

Patient Height

The average percentile height for all subjects with Madelung's deformity was below normal (Table 5), averaging the 19th percentile (range, 3rd to 90th).

Table 2. Radius and Ulna Lengths of Distal Radius Madelung Subjects Compared With Age-Matched Normal Subjects

			Study Patient				Age-Matched Normal Subjects				Percentiles			
					% of Height				% of Height				% of Height	
Subject	Age	Side	Radius Length (cm)	Ulna Length (cm)	Radius Length	Ulna Length	Average Radius Length	Average Ulna Length	Average Radius Length	Average Ulna Length	Radius Length	Ulna Length	Radius Length	Ulna Length
1	15+6	L	23	25	14	16	24	26	14	15	35th	50th	40th	70th
2	12+9	R	20	22	13	14	22	24	14	15	<10th	<10th	<10th	<10th
		L	17	20	11	13	22	24	14	15	<10th	<10th	<10th	<10th
3	36	R	20	17	NA	NA	23	25	NA	NA	<10th	<10th	NA	NA
4	15	R	20	22	13	14	24	25	14	15	<10th	<10th	<10th	<10th
5	15+6	R	19	21	12	13	24	26	14	15	<10th	<10th	<10th	<10th
		L	19	20	12	13	24	26	14	15	<10th	<10th	<10th	<10th
6	10+6	R	17	18	12	13	20	21	13	14	<10th	<10th	<10th	<10th
		L	20	22	15	16	20	21	13	14	85th	85th	90th	90th
7	16+9	R	20	23	13	14	23	25	14	15	<10th	<10th	<10th	<10th
	14+3	L	21	24	13	15	23	25	14	15	10th	30th	<10th	<10th
8	14+9	R	18	20	12	14	23	25	14	15	<10th	<10th	<10th	<10th
		L	19	21	13	14	23	25	14	15	<10th	<10th	<10th	<10th
9	13+5	R	15	20	10	13	23	24	14	15	<10th	<10th	<10th	<10th
		L	17	20	11	13	23	24	14	15	<10th	<10th	<10th	<10th
10	15	L	21	23	13	14	24	25	14	15	<10th	<10th	<10th	<10th
11	9+6	R	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA
		L	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA
12	9+7	R	19	20	13	14	17	19	13	14	90th	70th	60th	30th
		L	17	20	11	13	17	19	13	14	<10th	70th	<10th	30th
13	17	R	20	20	14	14	23	25	14	15	<10th	<10th	10th	<10th
		L	20	23	13	15	23	25	14	15	<10th	<10th	<10th	60th
14	9+9	R	16	18	13	14	17	19	13	14	10th	20th	50th	60th
15	33	R	17	19	NA	NA	23	25	NA	NA	<10th	<10th	NA	NA
		L	15	17	NA	NA	23	25	NA	NA	<10th	<10th	NA	NA
16	12+8	R	17	19	12	13	22	24	14	15	<10th	<10th	<10th	<10th
		L	16	19	11	13	22	24	14	15	<10th	<10th	<10th	<10th
17	10+4	R	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA
		L	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA
18	12+3	R	17	19	12	14	21	23	14	15	<10th	<10th	<10th	<10th
		L	13	16	9	12	21	23	14	15	<10th	<10th	<10th	<10th
Average			**18**	**20**	**12**	**14**	**22**	**24**	**14**	**15**				
SD			**2**	**2**	**1**	**1**	**2**	**2**	**1**	**1**				
p Value			**<.05**	**<.05**	**<.05**	**<.05**								

NA, not available

Table 3. Radius and Ulna Lengths of Entire Radius Madelung Subjects Compared With Age-Matched Normal Subjects

			Study Patient				Age-Matched Normal Subjects				Percentiles			
					% of Height				% of Height				% of Height	
Subject	**Age (years + months)**	**Side**	**Radius Length (cm)**	**Ulna Length (cm)**	**Radius Length**	**Ulna Length**	**Average Radius Length**	**Average Ulna Length**	**Average Radius Length**	**Average Ulna Length**	**Radius Length**	**Ulna Length**	**Radius Length**	**Ulna Length**
1	12+9	R	14	17	9	12	22	24	14	15	<10th	<10th	<10th	<10th
		L	13	17	9	12	22	24	14	15	<10th	<10th	<10th	<10th
2	14+1	R	13	18	9	12	23	25	14	15	<10th	<10th	<10th	<10th
3	8+6	R	10	13	8	11	16	18	13	14	<10th	<10th	<10th	<10th
		L	10	13	8	11	16	18	13	14	<10th	<10th	<10th	<10th
4	14+11	R	15	19	9	12	24	25	24	15	<10th	<10th	<10th	<10th
		L	14	19	9	12	24	25	24	15	<10th	<10th	<10th	<10th
5	12+5	R	14	17	11	14	22	24	14	15	<10th	<10th	<10th	<10th
		L	15	17	12	14	22	24	14	15	<10th	<10th	<10th	<10th
6	13	R	12	16	8	10	22	24	14	15	<10th	<10th	<10th	<10th
		L	10	15	7	10	22	24	14	15	<10th	<10th	<10th	<10th
7	13+3	R	13	18	9	12	22	24	14	15	<10th	<10th	<10th	<10th
		L	19	21	13	14	22	24	14	15	<10th	<10th	<10th	<10th
8	46+6	R	15	19	10	12	23	25	14	15	<10th	<10th	<10th	<10th
		L	17	20	11	12	23	25	14	15	<10th	<10th	<10th	<10th
Average			**14**	**17**	**9**	**12**	**22**	**23.5**	**15**	**15**				
SD			**3**	**2**	**2**	**1**	**2**	**2.3**	**3**	**1**				
p Value			**<.05**	**<.05**	**<.05**	**<.05**								

Table 4. Severity of Madelung's Deformity for All, Distal Radius and Entire Radius Madelung's Deformity Subjects

	Ulna Tilt (°)	Lunate Subsidence (mm)	Palmar Carpal Displacement (mm)
All 26 subjects			
Average	43	3	20
SD	17	7	12
Range	7–72	(–)7–35	(–)22–40
McCarroll range[19]	14–73	(–)5–20	10–36
18 distal radius Madelung subjects			
Average	48	1	22
SD	14	4	7
Range	24–72	(–)6–8	11–40
8 entire radius Madelung subjects			
Average	31	7	17
SD	18	10	18
Range	7–68	(–)7–35	(–)7–35
p Value*	.001	.02	.16

(–) implies lunate is entirely distal to distal ulna.
*Comparing distal radius and entire radius groups.

Subjects with distal radius Madelung's deformity were in the 24th percentile (range, 3rd to 90th) and subjects with an entire radius Madelung's deformity were in the 9th percentile (range, 3rd to 25th); there was a significant difference between the groups ($p < .05$). Twelve of the distal radius Madelung subjects were below the 25th percentile for height; 7 were below the 10th percentile. In contrast, all 8 entire radius Madelung subjects were below the 25th percentile for height; 6 were below the 10th percentile.

Dyschondrosteosis

All 8 subjects in the entire radius group may be retrospectively diagnosed with dyschondrosteosis as all were of short stature and had a markedly short forearm in addition to their Madelung deformity. Fourteen of the 18 subjects in the distal radius group had sufficient data to allow an accurate assessment of percentile height and forearm length. Nine of these subjects may be retrospectively diagnosed with dyschondrosteosis. Four of the other 5 subjects were of normal percentile height (the other subject was of short stature but had a normal-length forearm). Interestingly, in 2 of these subjects, one forearm was short whereas the other was of normal proportion.

Table 5. Comparison of Percentile Height Between Distal Radius and Entire Radius Madelung's Deformity Subjects

	No. Subjects	
Percentile Height	Distal Radius	Entire Radius
0–5	3	5
6–10	4	1
11–15	3	1
16–20	0	0
21–25	2	1
26–50	2	0
51–75	0	0
76–100	2	0
Unknown	2	0

Range of Motion (Table 6)

Elbow extension averaged 2° in distal radius Madelung extremities, significantly straighter ($p = .0003$) compared with the average 18° in subjects with entire radius Madelung's deformity. Entire radius extremities had less pronation (69° vs 47°, $p = .0001$) and supination (72° vs 59°, $p = .01$) compared with distal radius extremities. Wrist extension averaged 53° in distal radius extremities and 32° in entire radius extremities ($p = .0001$). There was no significant difference between groups for elbow flexion and wrist flexion ($p > .05$).

Discussion

Previous descriptions of Madelung's deformity have concentrated on the bony abnormality at the distal radius and wrist. In their classic description of the 12 features of Madelung's deformity, Dannenberg et al[7]

Table 6. Average Elbow, Forearm, and Wrist Motion in All, Distal Radius and Entire Radius Madelung's Deformity Extremities

	Elbow Extension (°)	Elbow Flexion (°)	Forearm Pronation (°)	Forearm Supination (°)	Wrist Extension (°)	Wrist Flexion (°)
All extremities						
Average ± SD	8 ± 13	143 ± 4	61 ± 19	68 ± 16	46 ± 17	62 ± 12
31 Distal radius Madelung extremities						
Average ± SD	2 ± 5	143 ± 4	69 ± 11	72 ± 14	53 ± 16	64 ± 10
15 Entire radius Madelung extremities						
Average ± SD	18 ± 17	142 ± 4	47 ± 23	59 ± 16	32 ± 9	59 ± 17
p Value*	.0003	.25	.0001	.01	.0001	.14
Normal motion	0	145	75	80	75	75

*Comparing distal radius and entire radius groups.

focused on the wrist but did mention that the entire diaphysis of the radius may be involved. The deformity at the radiocapitellar joint and the increased sagittal bow of the radius were not included among the 12 features. Subsequent reports on Madelung's deformity[17–20] also focus on the wrist deformity.

We identified 2 groups of subjects with Madelung's deformity, those with an involvement of the distal radius only (69%) and those with involvement of the entire radius (31%). Utilizing the criteria of McCarroll et al[19] to quantify the deformity, the severity of the wrist deformity was significantly worse in the entire radius group. The subjects in the entire radius group had a more significant sagittal bow to the radius and an increased radiocapitellar distance. Clinically, subjects in both groups had a loss of motion in pronation, supination, and wrist extension; however, the subjects in the entire radius group had a more severe loss of motion and, additionally, had a loss of elbow extension. Additionally, 5 of the 6 subjects with Madelung's deformity affecting only 1 side had the less severe distal radius deformity.

The identification of patients with deformity involving the entire radius has clinical importance. Functionally, the entire radius patients have decreased elbow, forearm, and wrist motion compared with other Madelung patients. Aesthetically, the patients will have a shorter and more curved forearm rather than only involvement of the distal radius. Furthermore, surgical procedures are typically focused on the distal forearm.[18] Both the patient and the surgeon should be aware that such procedures in the entire radius group are technically more difficult and not likely as effective in correcting the functional and aesthetic abnormalities.

The three criteria for the diagnosis of dyschondrosteosis,[5,7–9,12–14] or Leri-Weill syndrome, are Madelung's deformity, mesomelia, and short stature.[5,12–14] All the subjects in our study group had Madelung's deformity. All 8 subjects in the entire radius group had dyschondrosteosis. Nine of the 14 (64%) subjects in the distal radius group with complete data had dyschondrosteosis. Considering these data together, 17 of 22 (77%) of the total subjects (with complete data) fit the classic definition of Leri-Weill syndrome. This finding is in agreement with some previous authors who noted that most subjects with Madelung's deformity were likely to have dyschondrosteosis.[12–14]

The other 5 patients may also have dyschondrosteosis but without the typical presentation. For example, 2 of the 5 subjects had the unusual finding of 1 short forearm and 1 forearm of normal proportion. These variable findings in height and degree of mesomelia do not exclude the diagnosis of dyschondrosteosis as previous investigations have reported phenotypic variation in subjects with Leri-Weill syndrome. The Madelung's deformity is constant but, whereas most patients demonstrate short stature and mesomelia, there has been variability in height and forearm length.[22,23]

It should be noted that, at this time, there is a good but imperfect confirmatory genetic test for Leri-Weill syndrome. The SHOX-DNA-Dx genetic test (Esoterix, Inc., Austin, TX) uses polymerase chain reaction amplification to detect both deletions and mutations in the SHOX gene. Genetic abnormalities in the SHOX pseudoautosomal gene (short stature homeobox-containing gene) may be related to Turner syndrome, Leri-Weill syndrome, and short stature.[19]

Most of the subjects in this investigation did not have genetic testing as they were treated prior to the availability of this test. The findings of this investigation suggest that Madelung's deformity is, most commonly, a genetic condition. Confirmatory testing will be helpful for clinician and family alike.

There are several weaknesses of this study. First, radiographic measurements were performed at a single time point. Because multiple subjects had surgical correction for their Madelung deformity and others did not get routine follow-up radiographs of their extremities at each clinic visit, we were unable to assess the change in extremity deformity over time. Second, humerus and lower-extremity radiographs were not routinely obtained in the majority of the subjects, and we were unable to use these for assessment of mesomelia or short stature; however, we believe that the use of radius and ulna length relative to height and comparing these data with normal values is an effective assessment tool.

In conclusion, the results of this study indicate that most patients with Madelung's deformity will also have the extreme short stature and mesomelia consistent with Leri-Weill syndrome. Additionally, there is a subgroup of Madelung's deformity patients who have severe involvement of the entire radius. These patients have decreased range of motion of the extremity, a more bowed appearance to the forearm, and more notable radiographic deformity of the forearm and distal radius.

Received for publication March 16, 2007; Accepted in revised form August 17, 2007.

This investigation was performed at St. Louis Shriners Hospital for Children, St. Louis, MO.

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0363-5023/07/32A09-0012$32.00/0
doi:10.1016/j.jhsa.2007.08.012

References

1. Beals RK, Lovrien EW. Dyschondrosteosis and Madelung's deformity. Report of three kindreds and review of the literature. Clin Orthop Relat Res 1976;116:24–28.
2. Dobyn J. Madelung's Deformity. New York: Churchill-Livingston, 1993.
3. Madelung O. Die spontane Subluxation der Hand nach vorne, verh. Dtsch gese Chir 1878;7:259–276.
4. Nielsen JB. Madelung's deformity. A follow-up study of 26 cases and a review of the literature. Acta Orthop Scand 1977;48:379–384.
5. Dawe C, Wynne-Davies R, Fulford GE. Clinical variation in dyschondrosteosis. A report on 13 individuals in 8 families. J Bone Joint Surg 1982;64B:377–381.
6. Stetten D. Idiopathic progressive curvature of the radius, or so-called Madelung's deformity of the wrist (carpus varus and carpus valgus). Surg Gynecol Obstet 1909;8:4–31.
7. Dannenberg M, Anton J, Spiegel M. Madelung's deformity: consideration of its roentgenological diagnostic criteria. Am J Roentgenol Rad Therapy 1939;42:671–676.
8. Felman AH, Kirkpatrick JA Jr. Madelung's deformity: observations in 17 patients. Radiology 1969;93:1037–1042.
9. Golding JS, Blackburne JS. Madelung's disease of the wrist and dyschondrosteosis. J Bone Joint Surg 1976;58B:350–352.
10. Henry A, Thorburn MJ. Madelung's deformity. A clinical and cytogenetic study. J Bone Joint Surg 1967;49B:66–73.
11. Tachdjian M. Madelung's deformity. Philadelphia: Saunders, 1990.
12. Herdman RC, Langer LO, Good RA. Dyschondrosteosis. The most common cause of Madelung's deformity. J Pediatr 1966;68:432–441.
13. Langer LO Jr. Dyschondrosteosis, a hereditable bone dysplasia with characteristic roentgenographic features. Am J Roentgenol Radium Ther Nucl Med 1965;95:178–188.
14. Leri AaW J. Une affecion congenitale et symetrique du development osseus: la dyschondrosteose. Bull Et Mem Soc Med D Hop De Paris 1929 ;53:1491–1494.
15. Plafki C, Luetke A, Willburger RE, Wittenberg RH, Steffen R. Bilateral Madelung's deformity without signs of dyschondrosteosis within five generations in a European family—case report and review of the literature. Arch Orthop Trauma Surg 2000;120:114–117.
16. Bruno RJ, Blank JE, Ruby LK, Cassidy C, Cohen G, Bergfield TG. Treatment of Madelung's deformity in adults by ulna reduction osteotomy. J Hand Surg 2003;28A:421–426.
17. Carter PR, Ezaki M. Madelung's deformity. Surgical correction through the anterior approach. Hand Clin 2000;16:713–721.
18. Harley BJ, Carter PR, Ezaki M. Volar surgical correction of Madelung's deformity. Tech Hand Up Extrem Surg 2002;6:30–35.
19. McCarroll HR Jr, James MA, Newmeyer WL III, Molitor F, Manske PR. Madelung's deformity: quantitative assessment of x-ray deformity. J Hand Surg 2005;30A:1211–1220.
20. Watson HK, Pitts EC, Herber S. Madelung's deformity. A surgical technique. J Hand Surg 1993;18B:601–605.
21. McCammon R. Human growth and development. Springfield, IL: Thomas, 1970.
22. Ross JL, Scott C Jr, Marttila P, et al. Phenotypes associated with SHOX deficiency. J Clin Endocrinol Metab 2001;86:5674–5680.
23. Schiller S, Spranger S, Schechinger B, et al. Phenotypic variation and genetic heterogeneity in Leri-Weill syndrome. Eur J Hum Genet 2000;8:54–62.