CASE REPORTS

Madelung Deformity - A Case Report with Literature Review.

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Abstract:
Madelung deformity (MD) is a rare autosomal dominant disorder characterized by epiphyseal growth plate disturbance in distal ulnar sidewhereina progressive ulnar and volar tilt of itsarticular surface occurs in association with a dorsal subluxation of the distalulna.Diagnosis of the disease is by correlation of clinical data with radiological findings and genetic transmission. In this study, radiographic findings of a 16 years old female patient progressive bilateral wrist joint deformity and bowing of distal end of forearm without significant functional impairment and systemic complaints and diagnosed as Primary Madelung deformity, is reported.

Keyword: Wrist Joint, Deformity, Bilateral, Madelung

Introduction:
Madelung deformity (MD) is a rare inherited disorder involving the epiphyseal growth plate of distal radial end and consequent deformity as unaffected radial and dorsal portions of the growth plate continue to grow, accounting for 1.7% of all congenital hand anomalies. It can be bilateral in 50-66% of patients. It often occurs as rare congenital deformity and does not usually manifest until 10-14 years and is primarily found in females. One third of cases of MD are transmitted in an autosomal dominant fashion with variable penetrance. It may also be seen as an acquired consequence of trauma to the growth plate (e.g. Salter V fracture). Henry and Thorburn classified MD into 4 different etiologic groups: Posttraumatic, Dysplastic, Chromosomal or genetic (Turner syndrome), and idiopathic or primary. The underlying cause of this is unclear, with possibilities including local vascular insufficiency, trauma, infection (osteomyelitis) or muscular disorders. And sometimes MD associated with Leri-Weill syndrome (autosomal dominant dyschondrosteosis & mesomorphic dwarfism), Turner syndrome, nail-patella syndrome, diaphyseal aclasis (hereditary multiple exostosis), Hurler mucopolysaccharidosis, achondroplasia. Madelung deformity was first described Malgaigne in 1885 and later in 1878 by Otto Wilhelm Madelung, German surgeon (1846-1926) as “Spontaneous forward subluxation of the hand”. It was defined in terms of radiological findings and genetic transmission. Prompt and correct diagnosis is very important in order to ensure correct treatment and to provide the necessary consultation services to families. Herein, we present a case of MD in a teenage girl presented with progressive bilateral wrist joint deformity and bowing of distal end of forearm without significant functional impairment and systemic complaints and that was diagnosed with radiological and clinic findings, and typed by literature review.

Case Report:
A 16-year-old Muslim girl, second of two siblings of consanguineous birth to normal parents, hailing from Korban Ali’s Bari, Jamalpur, Shenbagh Thana, Noakhali and presented with short stature and progressive bilateral wrist joint deformities.

She was alright 1 year back, then she developed gradual onset, progressive, painless dinner fork disfiguration of right wrist with associated outward bowing of right forearm and little bit difficulties in writing & gripping. Now for last 5 months it’s also involved her left hand with sparing spine & lower limb.

She had no joint pain, morning stiffness, sensory complains and no history of preceding trauma or surgery or delay in achieving mile stone of development.
She was well cooperative and mentally & emotionally stable with normal vital signs. She was 141 cm tall with US: LS ratio of 0.9 and total arm span 113 cm & OFC 53 cm.

She had mesomelic shortening of both upper limbs with lumbar lordosis (Fig. 1). The limbs showed widening of wrist with garden spade deformity, fixed pronator deformity & bowing and bilateral cubitus valgus with normal warmth, bulk of muscle & no reddening & tenderness on palpation (Fig. 2). There was no limitation in movement of joints & spine and gait with normal other systemic examination without any organomegaly.

X-ray of hand and wrist were diagnostic of Madelung deformity with lunate subsidence 22 mm (normal < 4 mm), lunate fossa angle 45° (normal < 40°), palmar carpal distance 21 mm (normal < 21 mm) (Fig. 3, 4). The present case had ulnar tilt 11° which is not fit radiologic criteria of Madelung deformity (e°33° in Madelung deformity).

All routine investigations were within normal limit. Some second line investigations also performed and the findings were within normal limit like Serum

**Fig.-1:** Showing mesomelic shortening of both upper limbs with lumbar lordosis.

**Fig.-2:** Showing widening of wrist with garden spade deformity with radial bowing & bilateral cubitus valgus predominantly in right than left.

**Fig.-3:** X-ray of hand and wrist shows lunate subsidence 22 mm, lunate fossa angle 45° which are diagnostic for Madelung deformity.
calcium 8.7 mg/dl, Serum Inorganic Phosphate 4.5 mg/dl, Serum Alkaline Phosphatase 70 U/L, Serum PTH 51.4 pg/ml, normal Ultrasonography of whole abdomen without any organomegaly and normal thyroid status with Serum TSH 1.51 µIU/ml, F T4 1.18 ng/dl & F T3 3.14 pg/ml.

She was diagnosed as a case of Madelung deformity of right wrist and initiation of similar deformity in left wrist. Orthopedics consultation was advised and preparation of surgery was undertaken. But the patients and attendant did not give consent for operative procedure and discharged from hospital with her own deformity.

**Discussion:**

The Madelung deformity is relatively uncommon, with prevalence less than 2% and female predominance with female: male ratio is 3 to 5.10, 11. Typically, the deformity is present bilaterally and seldom manifests clinically before the age of 7 years and typically middle to late adolescent onset of the disorder may be linked to the adolescent growth spur.2-5 The long-standing and progressive radial deformity gradually worsens until it is suddenly exacerbated by the increased growth rate, often occurring concurrently with a premature physical fusion. The present case had support the above statements.

Radiographic features of MD are characterized by.

1. dorsal and radial bowing of the radius
2. exaggerated palmar (up to 35°) and ulnar tilt (up to 60°) of the radio carpal articulation
3. failure of ossification of the ulnar side of the distal radial epiphysis
4. exaggerated radial inclination
5. Decreased carpal angle below 118°; normal from 118° to 139°.
6. carpal subluxation in a palmar and ulnar direction
7. lunate is gradually forced to the apex of the V-shaped radioulnarocarpal joint
8. “V-shaped” proximal carpal row = herniated proximal carpal row.
9. dorsal subluxation of the distal ulnar and positive ulnar variance
10. wedging of the carpus between the radius and ulna.

Management is usually conservative. Persistent pain and/or severe deformity call for orthopedic surgery involving radial osteotomy. In addition, ulnar shortening in skeletonally immature patients or excision of distal ulnar head in the skeletonally mature is done. Surgical prophylaxis by resection of the abnormal part of the distal radial epiphysis and its replacement by autologous fat (also known as asphyxiolysis) have recently been shown to restore growth and minimize deformity.14-17 In our patients she was asymptomatic but her deformity was rapidly developing and also creates social stigma. That’s may be the reason why orthopedics department wanted to operate on her.

![X-ray of hand and wrist](image.png)

**Fig.-4:** X-ray of hand and wrist shows palmar carpal distance 21 mm and dorsal & radial bowing of radius with exaggerated radial inclination.
Conclusion:
Madelung deformity is a rare condition that affects the structure and function of the wrist. One third of disease is transmitted as autosomal dominant fashion and is commonly linked to several heritable factors. Sometime local trauma may also be responsible. Conservative approach in asymptomatic patient is the key but improved surgical technique for symptomatic patients with mature bone are rewarding now a days.

Conflict of Interest: None

References:
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