Brief Communication

All Madelung deformities are not endocrine

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ABSTRACT

Madelung deformity is a rare inherited disorder associated with endocrine disorders like Turner's syndrome, pseudohypoparathyroidism, but can be seen with short stature homeobox deficiency conditions such as Leri-Weill dyschondrosteosis (LWD) and Langers mesomelic dysplasia. It has also been reported following trauma to the distal radius epiphysis neoplasia mucopolysaccharidosis (MPS) and achondroplasia. Madelung deformity is an abnormality of distal radial epiphysis where in progressive ulnar and volar tilt of the articular surface occurring in association with distal subluxation of ulna. A 13-year-old girl was referred to us for evaluation of bilateral deformity of wrist and short stature. There was ulnar deviation and dorsal tilt of bilateral hands without history of pain to the joint trauma and family history of similar illness. On X-ray, wrist showed malformed distal radial epiphysis with dorsal and ulnar shift and with increased length of phalanges suggestive of Madelung deformity. X-ray spine was normal. Ultrasound abdomen showed normal uterus and ovary and her follicle stimulating hormone. Luteinizing hormone was normal and so was urine MPS screening. Based on the above points the diagnosis of LWD was made.

Key words: Achondroplasia, dyschondrosteosis, Madelung

Introduction

Leri-Weill's dyschondrosteosis (LWD) is anautosomal dominant condition with variable penetrance characterized by mesomelic short stature and with Madelung deformity.[1] It is due to deletion or mutation in Short Stature homeobox (SHOX) gene.[2] Here, there is deficient growth of a volar and ulnar aspect of distal radialphysis, triangulation of corpus with proximal and volar shift of the lunate.

Case Report

A 13-year-old girl referred to us with a history of deformity of bilateral forearm from the age of 5 to 6 years. There was no history of pain in the joint of hands, trauma to the wrist and no history of similar illness in the family.

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Patient was moderately built and significantly short for her age [Figure 1]. With height of 130 cm (less than 3rd percentile). Upper segment was 69 cm, lower segment was 61 cm, weight of 35 kg and body mass index of 20.71 kg/m^2. The arm forearm ratio was 1.78 suggestive of mesomelia. Secondary sexual characteristic Tanner-Whitehouse staging - B_4 (breast), P_4 (pubic hair), A_3 (axillary hair) her elder brother had normal height for age and no any hand deformities and so were her parents.

Investigation showed hemoglobin of 12.5 g/dl, liver function test and renal function test were normal Ca^{++} = 9.0 mg/dl alkaline phosphates 260 IU and phosphorus = 3.5 mg/dl. X-ray bilateral wrist showed malformed medial radial epiphysis with dorsal and ulnar shift and with increased length of phalanges suggestive of Madelung deformity [Figure 2]. Patient was evaluated for other causes of Madelung deformity. Her ultrasound abdomen showed no hepatomegaly or splenomegaly and had normal sized uterus and ovary X-ray spine and X-ray elbow were normal [Figure 3]. Her luteinizing hormone = −1.1 mIU/ml follicle stimulating hormone = 2.4 mIU/ml and karyotype was normal as was urine mucopolysaccharide screening.

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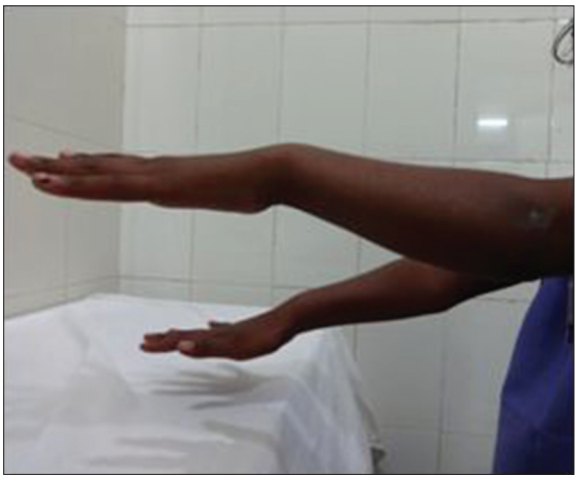

Figure 1:

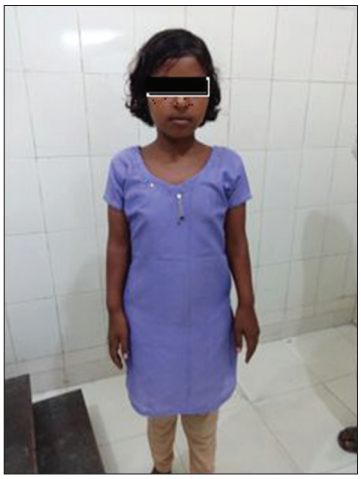

Figure 2:

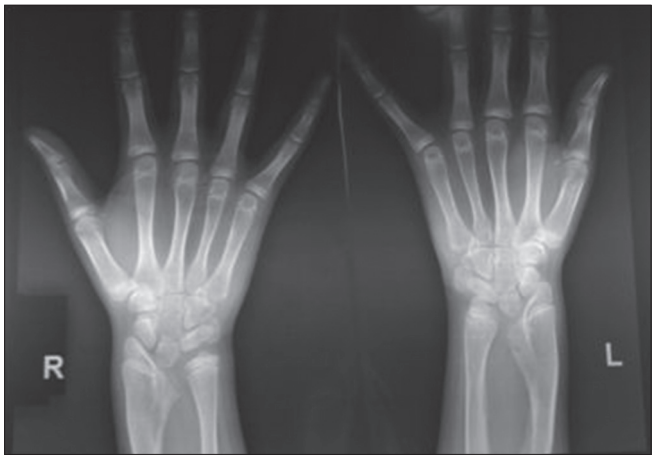

Figure 3: X-ray bilateral wrist

Based on the history, clinical examination, and investigations findings, the final diagnosis of LWD as a cause of Madelung deformity was made.

Discussion

LWD is characterized by the short stature and bilateral Madelung deformity. It is the most common cause of mesomelic deformity and is inherited as an autosomal dominant trait with variable penetrance.[2] It is often seen in girls and becomes apparent in late childhood or early adolescence. Madelung deformity, beside is also found in Turner's syndrome, pseudohypoparathyroidism, mucopolysaccharidosis and achondroplasia of distal radial epiphysis. It has also been reported following trauma to the wrist, infection and neoplasia.[3]

Madelung deformity was first described by Malgaigne in 1885 and later by Madelung in 1878 as a spontaneous forward subluxation of the hand. Madelung deformity is an ulnar and dorsal curvature of the distal radius due to deficient growth of the volar and ulnar aspect of distal radialphysis, increased inclination of the distal radial joint surface, triangulation of the corpus with proximal and volar migration of and a prominent dorsal subluxation of ulnar head. Recently, two subtypes of Madelung deformity have been described, one with short stature and mesomelia consistent with LWD and the other with severe involvement of the entire radius with limited range of motion of extremity, markedly bowed appearance of the forearm and conspicuous radiographic deformity of the forearm and distal radius.

The pathogenesis of LWD is linked to deletion or mutation in SHOX gene, present in the pseudoautosomal region of the sex chromosomes - Xp23 and Yp11. In early human embryos it is expressed in the developing limbs (particular elbow, knee, distal radius/ulnar and wrist) as well as first and second pharyngeal arches and plays an important role in bone growth as well as development. SHOX gene is also associated with short stature in Turner's syndrome and also some causes of growth retardation like LWS and Langers mesomelic dysplasia.[4-6]

Management is usually conservative. Persistent pain and or severe deformity call for orthopedic surgery involving radial osteotomy. In addition, ulnar shortening in skeletally immature patient or excision of distal ulnar head in skeletally mature patients are done. Surgical prophylaxis by distal resection of the abnormal part of distal radial epiphysis and its replacement by autologous fat (also known as physeolysis) has recently been shown to restore growth and minimized deformity.[7] Growth hormone supplementation is found to increase final height in Turner's syndrome, LWS but not in Langers mesomelic dysplasia.

Conclusion

All Madelung deformities are not endocrine condition. It is also found genetic condition like LWD. Hand deformity is corrected by orthopedic surgery and for SHOX gene related short stature Food and Drug Administration has recently approved growth hormone therapy.

References

1. Léri A, Weill JA. Une affection congénitale et symétrique dudéveloppement osseux. La dyschondrostéose. Bull Mem Soc Med Hop Paris 1929;53:1491-4.
2. Langer LO Jr. Dyschondrosteosis, a hereditable bone dysplasia with characteristic roentgenographic features. Am J Roentgenol Radium Ther Nucl Med 1965;95:178-88.
3. Clement-Jones M, Schiller S, Rao E, Blaschke RJ, Zuniga A, Zeller R, *et al*. The short stature homeobox gene SHOX is involved in skeletal abnormalities in Turner syndrome. Hum Mol Genet 2000;9:695-702.
4. Blaschke RJ, Rappold G. The pseudoautosomal regions, SHOX and disease. Curr Opin Genet Dev 2006;16:233-9.
5. Marchini A, Rappold G, Schneider KU. SHOX at a glance: From gene to protein. Arch Physiol Biochem 2007;113:116-23.
6. Lamb D. Madelung deformity. J Hand Surg Br 1988;13:3-4.
7. Vickers D, Nielsen G. Madelung deformity: Surgical prophylaxis (physiolysis) during the late growth period by resection of the dyschondrosteosis lesion. J Hand Surg Br 1992;17:401-7.

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