

## Leri-Weill Syndrome in an Adult Nigerian: A Rare Case Report

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### Abstract

### Case Report

Leri-Weill syndrome was first described in 1929 by Leri and Weill. It is an autosomal dominant skeletal dysplasia characterised by mesomelic short stature with bilateral Madelung deformity. Leri-Weill syndrome is one of the rarest causes of dwarfism and usually presents in late childhood. Because of its rarity, it is not usually considered as a differential cause of dwarfism. We report a rare case of Leri-Weill syndrome presenting in an adult Nigerian female. We also described the characteristic radiographic features of madelung deformity.

**Keywords:** Leri-Weill, Madelung deformity, Skeletal, dysplasia, Dwarfism.

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## INTRODUCTION

Leri-Weill syndrome (LWS) also called Leri-Weill dyschondrosteosis was first described in 1929 by Leri and Weill [1, 2]. It is an autosomal dominant skeletal dysplasia characterised by mesomelic short stature with bilateral Madelung deformity (MD) [2, 3]. The characteristic features of MD include; lateral and dorsal bowing of the distal radius, proximal displacement (subsidence) of the lunate and positive ulnar variance. It is also important to note that Madelung deformity can occur in isolation (isolated idiopathic Madelung deformity) which presents with wrist deformity and no other clinical characteristic. Leri-Weill syndrome is usually seen in females and becomes apparent before puberty with the deformity affecting both wrists with asymmetric severity [4-6].

The pathogenesis of LWS is linked to deletion or mutation of short stature homeobox (SHOX) gene present on sex chromosomes. Heterozygous mutation of SHOX gene is commonly seen in LWS with cosegregation of Madelung deformity and mesomelic short stature [7]. Consequently, it results to growth arrest of the medial radial physis with resultant increase volar tilt and increase inclination of the distal radius and proximal migration of the lunate.

Though LWS is a congenital abnormality that tends to present in late childhood or early adolescence with wrist pain and deformity which is usually bilateral. The pain and/or deformity are the common provoking symptoms that will make the patient to present to the hospital. The clinical features of LWS include the following; Short stature, bilateral wrist deformity, Wrist pain, bowing of the distal forearm, deformity of the palm, reduction in grip strength, and limitation of movement (supination, pronation and extension of the wrist).

The radiographic appearances may include; Increased volar tilt of the distal radius, Increased inclination of the radial articulating surface, premature fusion of the medial half of the distal radial epiphysis, complete or partial loss of the medial half of the distal radial epiphysis, triangulation of the lateral remnant epiphysis with apex pointing medially, dorsal displacement of the distal ulnar, Ulnar variance, dislocation of the distal radioulnar joint, widening of the interosseous space, triangulation and wedging of the carpal bones between the abnormal distal radioulnar joint and decreased carpal angle. Other radiographic features include; Ulnar tilt  $\geq 33^\circ$ , Lunate subsidence  $\geq 4\text{mm}$ , Lunate fossa angle  $\geq 40^\circ$  [8, 9].

Computed tomography may be helpful in characterising the features better than plain radiograph

especially the 3-D reconstructed image might help delineate the articulation of the deformed wrist [10]. MRI may demonstrate thickening of the radiolunate ligament (Vickers ligament).

A rare case of Leri-Weill syndrome in a Nigerian adult female is presented in this report. To the best of our knowledge, Leri-Weill Syndrome has not been reported in Nigeria or Sub Saharan Africa.

## CASE REPORT

H.M.A was a 20 years old female who presented to the General outpatient department (GOPD) of the Federal teaching hospital Gombe (F.T.H.G) on account of 1 year history of progressive deformity of both wrists with associated pain. The pain was said to be worsening with normal daily activities. Patient also complained of limitation of movement of both wrist joints. No deformity, pain or limitation of movement in the lower limbs. There was no previous history of trauma to the wrist joints or forearms. There was also no family history of wrist deformity or short stature.

On physical examination, the patient was of short stature with a height of 112cm (< 5 percentile). There was dorsal prominence of the ulnar head bilaterally with the right side worse than the left and postero-lateral bowing of the distal forearms were also noted. Significant ulnar tilt of the hand and kyphosis of the carpus were noted bilaterally. There was limitation of movement affecting supination, pronation and extension elicited at both wrist joints. Flexion and extension at the elbow joints were normal. The lower limbs were grossly within normal limits.

Laboratory investigations including PCV, FBC, RBS and urinalysis were all normal. Anteroposterior (AP) radiograph of both wrists (Fig. 1A) showed bilateral increased inclination of the articulating surface of radius and triangulation of the distal radial epiphysis with its apex pointing medially. The medial aspect of the distal radial epiphyses was not

visualised. There was distortion of the proximal row of the carpus with reduction in carpal angle and wedging of the carpus into the widened distal radioulnar joint. There was ulnar variance of both wrists.

Lateral radiograph of both wrists (Fig. 1B) demonstrated bilateral posterior bowing of the radius with associated arching of the carpus volarly in continuation of the dorsal bowing of the radius. Bilateral posterior displacement of the ulnar was also noted. The ulnar tilt measurements (Fig. 2A) were 44.8° and 51.1° on right and left sides respectively (normal < 33°). Increased in the lunate fossa angles were noted which measured 56.8° and 48.0° on the right and left respectively (Fig. 2B). Marked lunate subsidence was also noted bilaterally measuring 14.4mm and 9.5mm on right and left respectively (normal < 4mm) (Fig.3A). Bilateral palmar carpal displacement was observed and measured 25.8mm and 21.1mm on the right and left respectively (Fig. 3B). The radiographs of the foot and elbow were within normal limits.

Based on the history, clinical features and radiographic findings, diagnosis of LWS was made.

## DISCUSSION

Leri Wiell syndrome is a rare form of dyschondrosteosis which is associated with mesomelic short stature. Patient usually presents with pain and deformity of both wrists. It is an autosomal dominant dyschondrosteosis with variable penetrance which is commoner in females and present commonly in late childhood. The case reported on was a female adult who presented with pain and deformity of both wrists with associated short stature which is in conformity with classical features of the disease. However there was no family history of the condition which may be due to the variable penetrance of SHOX gene mutation or the milder form of the disorder exist in the parents and siblings which is asymptomatic and can only be detected Incidentally [11,12].



**Fig-1: (A) Anteroposterior radiograph of both wrists showing bilateral increased inclination of the articulating surfaces of radius and triangulation of the distal radial epiphysis with its apex pointing medially (white arrows). Note the distortion of the proximal row of the carpus with reduction in carpal angle and wedging of the carpus into the widened distal radioulnar joint. Ulnar variance is seen bilaterally. (B) Lateral radiographs of both wrists**

**demonstrating bilateral posterior bowing of the radius with associated arching of the carpus volarly in continuation of the dorsal bowing of the radius. Bilateral posterior displacement of the ulnar was also noted**



**Fig-2: (A) AP radiograph showing measurement of the ulnar tilt which measured 44.8° and 51.1° on right and left respectively. (B) AP radiograph showing increase in lunate fossa angle noted which measured 56.8° and 48.0° on right and left respectively**



**Fig-3: (A) AP radiograph showing marked lunate subsidence noted bilaterally measuring 14.4mm and 9.5mm on right and left respectively. (B) Lateral radiograph showing palmar carpal displacement measured 25.8mm and 21.1mm right and left respectively**

LWS is a congenital disorder but tends to present in late childhood with bilateral wrist pain and/or deformity. However, in this case report the patient presented in adulthood.

The differential diagnosis of LWS include; isolated idiopathic Madelung deformity, isolated short stature, Turner's syndrome, Langer syndrome, trauma, and infection, multiple exostosis, Ollier's disease, and reverse Madelung deformity.

Plain radiographs, anteroposterior and lateral radiographs of the wrists and forearms are diagnostic for Madelung deformity. Short stature can be evaluated by accurate assessment of longitudinal growth with critical analysis of appropriate growth data for age and sex. The radiographic findings in this case were classical features of Madelung deformity and the patient's height of 112cm was less than 5 percentile. MRI may be useful in early detection by demonstrating thickening of the radiolunate ligament in early childhood even before the development of osseous deformity in suspected cases. MRI may play a role in early detection of the disorder. However, it was not done for this patient.

The management of LWS is often conservative especially in those presenting with mild deformity. Persistent pain and severe deformity may warrant surgical treatment. Operative procedures that will reduce deformity and pain include; radial osteotomy, Vicker's ligament release, ulnar resection and ulnar osteotomy. Growth hormone therapy may improve longitudinal growth. The patient in this case report opted for conservative management and she was responding to treatment satisfactorily at follow up.

## CONCLUSION

LWS is a rare form of dyschondrosteosis which is characterised by short stature and bilateral Madelung deformity. The unique appearances of Madelung deformity seen on radiographs were diagnostic. These were demonstrated by the index case presented.

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