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Wrist Radiography for Hand Bone Age Tells A Lot; A Girl with SHOX Deficiency

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Madelung's deformity (MD) occurs as a result of premature closure of the medial and volar aspects of the distal radial physis.[1] It is more frequent and severe in girls, and usually develops in middle/late childhood.[2] MD is one of the most characteristic features of the short-stature homeobox gene (SHOX) deficiency, which causes short stature.[3] Radial bowing is one of the well-known radiological futures. On the other hand, there are three typical radiological sign of the hand radiograph for SHOX deficiency; triangularization, pyramidalization of the os lunatum, and radiolucency at the distal radius.[4]

In the evaluation of a 9-year-old girl who was investigated for precocious puberty, her height measurement was 18th percentile. On the wrist X-ray taken for the determination of the bone age of the patient, there was an appearance compatible with MD (**Figure 1**). In the genetic studies of the patient with MD, normal female karyotyping (46, XX) was demonstrated by Trypsin G banding Technique. Heterozygous SHOX deletion was detected by Fluorescence In Situ Hybridization technique using a probe specific to the SHOX gene region (Xp22.33).

Interpreting the direct X-ray is important in recognizing the MD. Thus, it will be easier to detect SHOX gene deletion in the etiology of short stature patients with this deformity.

Figure 1: Madelung deformity detected on left wrist radiograph: radial bowing, premature fusion of the distal radial epiphysis

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