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Clinical Image

Knuckle-Dimple-Dimple-Dimple Sign

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Clinical Image

A 15-year-old girl, second in birth order from consanguineous marriage, presented with progressive weight gain over five years. Upon examination, she had a round face, almond-shaped eyes, acanthosis nigricans, short stature (height SDS -2.5), and obesity (BMI 30.6 kg/m). Her hands and feet showed short, stubby fingers with positive Archibald sign, characterized by a "knuckle-dimple-dimple-dimple" pattern (Figure 1a). Her mother and maternal grandmother shared similar hand features, suggesting a familial inheritance. Radiographic evaluation of the hands revealed Type E brachydactyly based on the Tamtamy classification, with shortening of 3rd, 4th and 5th metacarpals and phalanges (Figure 1b). Biochemical testing showed normal calcium, phosphate, and Parathyroid Hormone (PTH) levels, ruling out true hypoparathyroidism. In presence of normal regulation of mineral ion homeostasis, short stature, obesity and typical Albright's Hereditary Osteodystrophy (AHO) feature, diagnosis of pseudo-pseudo hypoparathyroidism was stablished. This rare autosomal dominant disorder is caused by GNAS gene mutation.

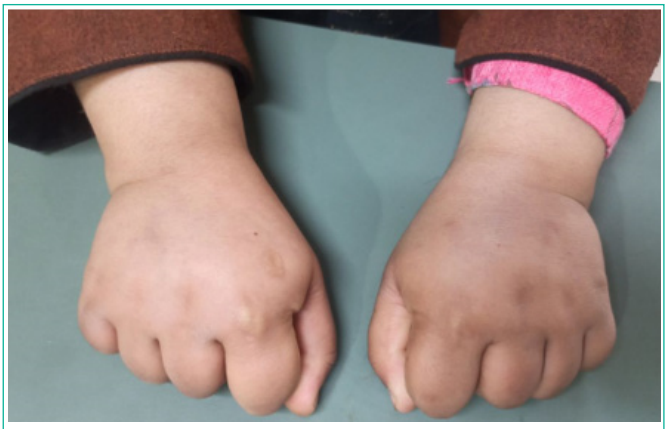

Figure 1a: Knuckle-dimple-dimple-dimple.

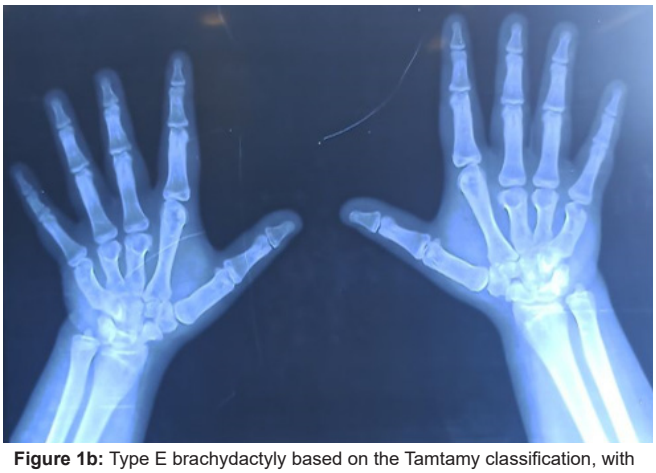

Figure 1b: Type E brachydactyly based on the Tamtamy classification, with shortening of the 3rd, 4th and 5th metacarpals.

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