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Clinical picture

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Knuckle–dimple sign

A 30-year-old man was noted to have short fourth and fifth metacarpals (Figure 1) on routine examination. Biochemically, he had hypocalcemia, hyperphosphatemia, high parathyroid hormone and normal renal function. He was obese [body mass index (BMI) = 33]. He carried a diagnosis of pseudohypoparathyroidism. Brachydactyly is one of the commonly recognized signs of Albright's hereditary osteodystrophy (AHO),[1] along with short adult stature, obesity and developmental delay. Short metacarpals are not pathognomonic of AHO and can be seen in many other conditions. AHO was first described by Albright and his colleagues in 1942. AHO has been genetically linked to *GNAS1* gene.[2] Brachydactyly most commonly involves the fourth and fifth metacarpals, but can also be seen in other metacarpals and metatarsals.[3] Affected knuckles dimple when the fist is clenched causing the 'Knuckle–dimple' sign (Figures 2 and 3). Patients with AHO features can have either pseudohypoparathyroidism or pseudopseudohypoparathyroidism based on the presence of end-organ resistance to parathyroid hormone.

Figure 1. Bilaterally shortened fourth and fifth fingers seen in AHO.

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Figure 2. Dimple seen instead of fourth knuckle causing 'knuckle–dimple' sign.

Figure 3. Dimple seen instead of fourth knuckle causing 'knuckle–dimple' sign.

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