Albright's dimpling sign

Sir,

A five-years-old girl presented to our hospital for an upper respiratory tract infection. She was second born to non-consanguineous marriage by spontaneous vaginal delivery at term with normal birth weight, normal antenatal, neonatal and postnatal period. On examination she was stocky, appeared normal in height (107 cms). Her weight was 25 kg (>97th percentile on the WHO weight for age chart) and her body mass index was 21.74 (>97th centile for age).Incidentally on examination she was found to have brachycephaly, hypertelorism, epicanthal folds, depressed nasal bridge short stubby fingers. Dimpling of hand at the site of knuckles on making a fist [Figure 1] due to short metacarpals of 3rd, 4th and 5th finger [Figure 2] with sparing of index finger was evident. Short metacarpals and dimpling of knuckles was first described by Fuller Albright and is also referred to as the Albright`s dimpling sign.[1] It is a feature of Albright`s Hereditary Osteodystrophy (AHO) phenotype known to be associated with both pseudohypoparathyroidism (PHP) as well as pseudopseudohypoparathyroidism (PPHP). The defect is due to tissue specific G protein alpha subunit (Gsα) gene mutation. Gsα gene mutations inherited from the mother cause Albright`s Hereditary Osteodystrophy (AHO) phenotype and resistance to action of thyrotropin, parathormone and gonadotropin and is seen in PHP.[2] Tissue specific paternal imprinting is characteristic of PPHP.[3] It differs from PHP in not having resistance to parathormone and other hormones.

In our case, to differentiate the aforesaid conditions, serum calcium, phosphorus, alkaline phosphatase and parathormone levels were obtained but were found to be normal. Hence a diagnosis of PPHP was made. There were no subcutaneous calcifications noted on physical examination which are commonly seen in PHP rather than PPHP.[4] Intracranial calcifications were ruled out by a normal CT brain. Both parents and the elder sibling were normal and did not share the same phenotype. Brachydactyly occurs in these children due to premature closure of epiphysis in the metacarpals. For affected families, pre-implantation genetic diagnosis is available to identify severe phenotype.[5] In our case the family was completed and consent for a genetic diagnosis was denied. Looking at a child`s knuckles is a valuable clinical sign not to be forgotten in a busy outpatient practice.

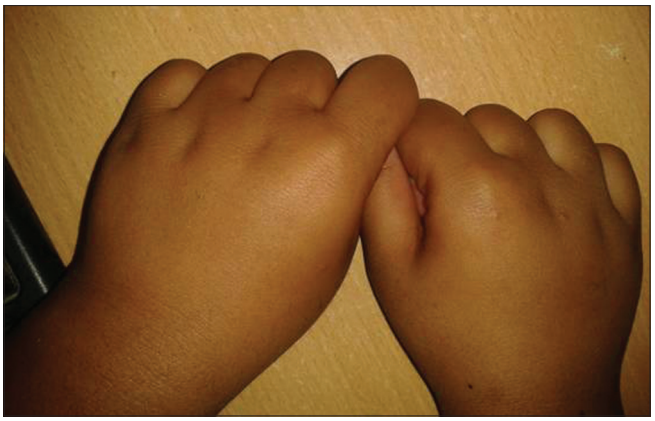

Figure 1: Dimpling at knuckles on making a fist

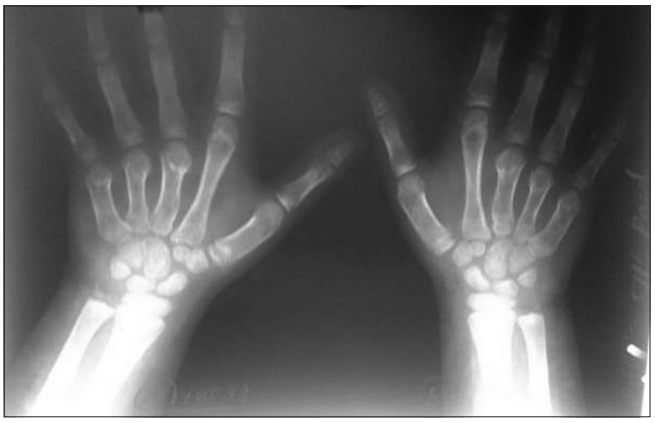

Figure 2: Radiograph showing short 3rd, 4th and 5th metacarpals

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Quick Response Code:	Website: www.ijem.in
	DOI: 10.4103/2230-8210.109694