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. It is a feature of Albright’s Hereditary Osteodystrophy (AHO) phenotype known to be associated with both pseudohyoparathyroidism (PHP) as well as pseudopseudohyoparathyroidism (PPHP). The defect is due to tissue specific G protein alpha subunit (GαS) gene mutation. GαS 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. Tissue specific paternal imprinting is characteristic of PPHP. 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. 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. 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.

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Figure 1: Dimpling at knuckles on making a fist

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

We read with great interest your article entitled “A rare case of lingual thyroid with hyperthyroidism: A case report and review of the literature” by Jacob, et al. [1] the issue of May‑June 2012. We would be happy to make our modest contribution in the light of our humble experience. The literature review confirms the rarity of thyroid ectopy and the exceptional character of the association of this abnormal embryogenesis with hyperthyroidism.

The ectopic thyroid is the presence of thyroid tissue outside its normal seat secondarily to defective migration of thyroid diverticulum and whose pathogenesis is not fully elucidated. Clinical symptoms are variable from asymptomatic to complicated forms such as dyspnoea, dysphagia. Degeneration is exceptional. The patient may be euthyroid or hypothyroid or exceptionally hyperthyroid. Diagnosis uses 99m‑Tc or 123‑I thyroid scan which confirms the thyroid tissue, cervical computed tomography or magnetic resonance imaging or doppler. Simple monitoring is recommended in the case of euthyroidism, a hormone replacement therapy in the case of hypothyroidism, and antithyroid drugs, surgery, or radioiodine in the case of hyperthyroidism.

We had to take a child for a double localization, orthotopic, and ectopic lingual position of the thyroid gland with hypothyroidism which responded favorably to Levothyroxine treatment and no surgery was recommended. Moreover, Terris [4] in his work entitled “A new minimally invasive thyroidectomy lingual technique” has reported a case of lingual thyroid ectopy in a patient of 34 years, with dysphagia, who responded favorably to minimally invasive surgical treatment.

In the case reported by Jacob, et al., [1] the patient was in hyperthyroidism, which is, as reported by the author, an exceptional situation. The risks of surgery, namely the risk of bleeding and intubation, led the authors advocate radioiodine preceded by a medical preparation by antithyroid drugs. Regarding the therapeutic component, we adhere to the recommended course of action. In fact, it is very risky to operate this patient because, indeed, there is a hypervascular thyroid mass and hemostasis is difficult to achieve since it is the lingual artery to be ligated on both sides which can cause necrosis of the tongue. However, with regard to intubation, in our humble opinion, there is no indication against because there is the possibility of intubation by using nasal endoscope or tracheostomy under local anesthesia. We believe that the authors have opted for short‑term safety and they were right to do so but the risk of degeneration is always present and the possibility of hyperthyroidism recurrence will always be present with multiple risks including increased size of the gland with its mechanical complications. Overall, despite its rarity, the management of ectopic lingual thyroid is a challenge and must be individualized for each patient.

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