Albright's hereditary dystrophy: brain stones

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DESCRIPTION

A 20-year-old woman presented with frequent cramps in extremities with carpopedal spasms and recurrent episodes of generalised tonic–clonic seizures for the past 10 years. Two years previously she had undergone cataract surgery with lenticular implants in both the eyes. There was no history of any prolonged drug intake, fever, toxin exposure, chronic illnesses in the past or any significant family history for similar illness.

Examination revealed short stature, rounded face with generalised skin darkening, poor dentition, patchy alopecia and brachydactyly (figure 1). She also had demonstrable Chvostek's Grade III and Trousseau sign. Her haemogram was normal and biochemistry revealed normal serum albumin, low serum calcium (8.0 mg/dL), elevated phosphates (7.5 mg/dL), elevated parathormone levels (200 pg/mL) and normal thyroid profile. A CT scan of the head showed calcification involving bilateral basal ganglia, thalamus, dentate nucleus and subcortical white matter (figure 2). Ultrasound abdomen revealed a left renal cortical cyst and nephrolithiasis. X-rays of both the feet showed shortening of fourth metatarsal (figure 3) along with osteodystrophic features.

The diagnosis of Albright's hereditary osteodystrophy was considered. This clinical entity was diagnosed by Albright *et al*.[1] Pseudohypoparathyroidism (PHP) type 1a is associated with constellation of clinical features collectively termed as Albright hereditary osteodystrophy (AHO). These features include short stature, rounded face, brachydactyly, brachymetacarpia, centripetal obesity, subcutaneous ossifications, basal ganglia calcifications and, in some cases, mental or developmental delay.[2] PHP is generally classified as types Ia, Ib, Ic and II according to different phenotypes and pathogenesis. AHO is often associated with pseudohypoparathyoidism, hypocalcaemia and elevated PTH levels (PHP Ia), as seen in our patient. Treatment of hypocalcaemia with calcium and vitamin D supplements is the mainstay, with routine screening of other associated endocrinopathy.

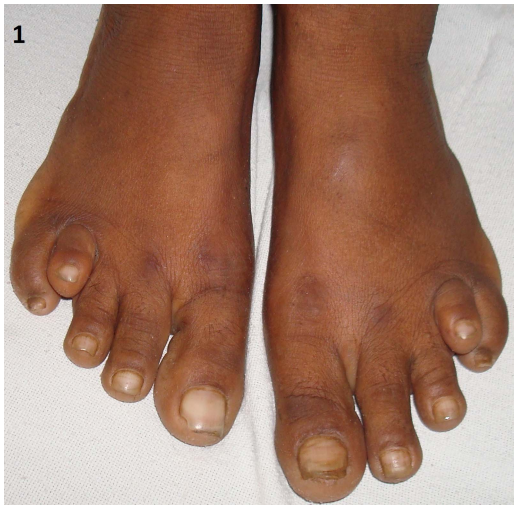

Figure 1 Foot photographs of the patient showing brachydactyly.

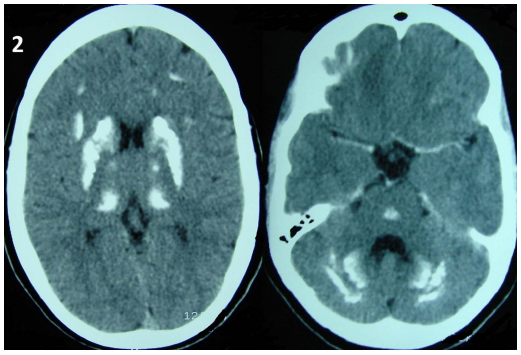

Figure 2 Non-contrast CT scan of the head showing bilateral basal ganglia, thalamic, pontine, dentate and subcortical calcifications.

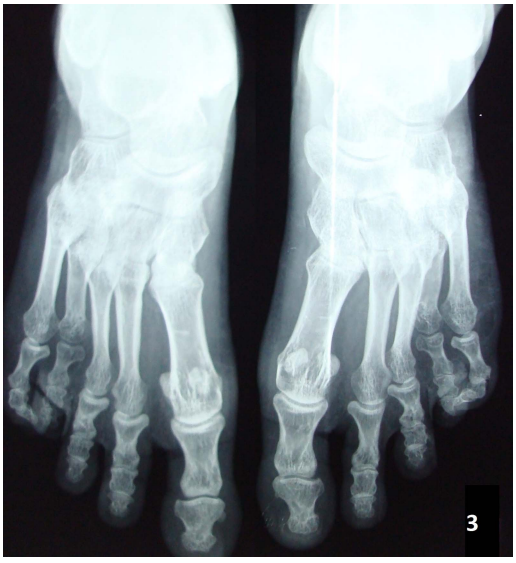

Figure 3 Digital X-ray of the feet (dorso-plantar view) showing bilateral shortening of fourth metatarsal.

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Learning points

- Albright hereditary osteodystrophy constitutes constellation of brachydactyly, tetany, subcutaneous and basal ganglia calcifications with pseudohypoparathyroidism (PHP) type Ia.
- Low serum calcium, high phosphates and raised parathormone levels suggesting parathormone resistance are characteristics of this syndrome.
- Immediate correction of hypocalcemia with calcium and vitamin D supplements is of paramount importance.

Competing interests None.

Patient consent Obtained.

Provenance and peer review Not commissioned; externally peer reviewed.

REFERENCES

1 Albright F, Burnett CH, Smith PH. Pseudohypoparathyroidism: an example of 'Seabright-Bantam syndrome'. *Endocrinology* 1942;30:922–32.
2 Levine MA. Pseudohypoparathyroidism: from bedside to bench and back. *J Bone Miner Res* 1999;14:1255–60.