Fahr's disease

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DESCRIPTION

An 18-year-old girl with a history of hypocalcaemic seizure disorder, diagnosed at 14 years, refractory to treatment with phenytoin, phenobarbital and valproate, presented with three new episodes of generalised tonic–clonic seizures. In addition to some cognitive impairment, on examination she had a positive Trousseau sign (flexion of the wrist and metacarpophalangeal joints when blood pressure cuff if inflated above the systolic blood pressure), and brisk reflexes with ankle clonus. Laboratory studies revealed calcium 6.5 mg/dL (8.5–10.5 mg/dL), phosphorus 6.5 mg/dL (3–4.5 mg/dL), parathyroid hormone 134 pg/dL (17–70 pg/dL) and 25-hydroxyl vitamin D 32 ng/dL (30–65 ng/dL). CT of the brain revealed bilateral symmetrical calcification of the caudate nucleus, lenticular nucleus (putamen+globus pallidus), thalamus (figure 1) and the dentate nucleus of the cerebellum (figure 2). She was diagnosed with striatopallidodentate calcinosis (or Fahr's disease) secondary to hypocalcaemia due to pseudohypoparathyroidism type Ib. Her calcium levels were corrected, and she followed up with 24 h urinary calcium levels for titration of therapy.

Fahr's disease is a neurodegenerative syndrome associated with symmetric intracerebral calcifications in the basal ganglia and adjacent parenchyma, and cognitive, neuropsychological and movement disorders. In 1850, Delacour first described vascular calcifications of the basal ganglia and Bamberger-described histopathological entity.[1] While bilateral striatopallidodentate calcinosis is commonly referred to as 'Fahr's disease', there are 35 additional names used in the literature for the same condition. Fahr's name became associated with all forms of bilateral calcifications in the basal ganglia and other parts of the brain, despite the fact that he was not the first to describe calcification in the brain. Secondary bilateral calcification is also reported in a variety of genetic, developmental, endocrine, metabolic and infectious conditions.[2] This is an atypical presentation of striatopallidodentate calcinosis which is almost always a movement disorder with or without cognitive impairment.

Findings of low calcium and high phosphate levels are consistent with true hypoparathyroidism. However, elevated parathyroid hormone levels reflect hormone resistance, hence the name pseudohypoparathyroidism. There are four types of pseudohypoparathyroidism types Ia, Ib, II and pseudopseudohypoparathyroidism. Our patient had type Ib pseudohypoparathyroidism as there was an absence of the Albright phenotype (short fourth and fifth metacarpals that is a classic symptom for type Ia).[3]

Learning points

- Know the presentation of Fahr's disease.
- Recognise secondary causes of bilateral striatopallidodentate calcinosis.
- Understand the biochemical abnormalities in psuedo-hypo-parathyrodism.

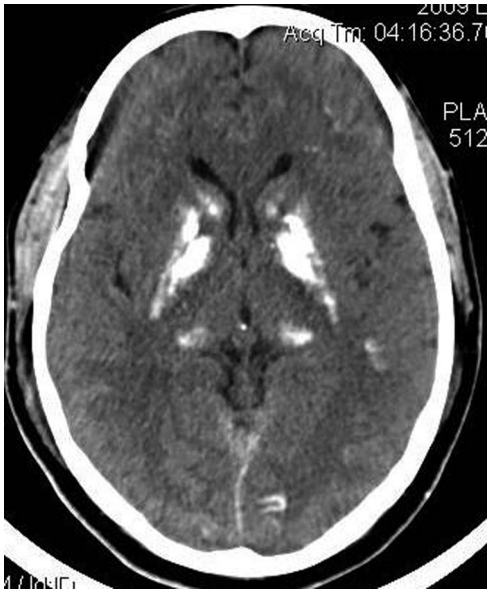

Figure 1 CT of the brain demonstrating bilateral symmetrical calcification of the caudate nucleus, lenticular nucleus (putamen+globus pallidus), thalamus.

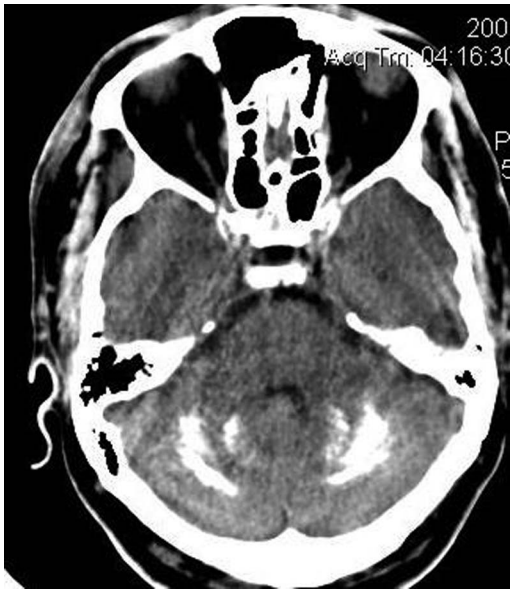

Figure 2 Dentate nucleus of the cerebellum on CT scan.

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