Intracranial Calcification

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Introduction

- Fahr's disease and Fahr's syndrome are rare disorders characterised by bilateral calcification of the basal ganglia.[1] Presenting features can include basal ganglia movement disorder, pyramidal signs, cognitive impairment, seizures, speech dysfunction, psychiatric presentations and sensory changes.[2]
- Fahr's disease, or familial primary brain calcification, can be of autosomal dominant or recessive inheritance and is associated with mutations in genes including *SCL20A2*, *PDGFB,* and *PDGFRB.*[3]
- Fahr's syndrome by contrast is associated with endocrine abnormalities such as hypoparathyroidism, metabolic abnormalities such as lipoproteinosis or mitochondrial myopathies, and infections such as intrauterine or perinatal brucellosis.[2]
- Whilst Fahr's disease is treated symptomatically, for example with anti-epileptics, anti-psychotics, or levodopa, Fahr's syndrome is managed by targeting the underlying abnormality.[2]

Case Report

- 58 year old female of Polish origin
- Presented after two tonic-clonic seizures
- Past Medical History:
  - 20 year history of muscle spasms, perioral and leg paraesthesia, as well as intermittent dysarthria and dysphagia
  - 20 year history of depression and anxiety
  - Previous seizures but had been seizure-free for the past 10 years
  - Investigated for the above symptoms in Poland 10 years ago. CT head showed bilateral basal ganglia calcification. Patient was subsequently told that her symptoms were due to a rare genetic disorder which could not be cured called Fahr's disease.
- No family history of similar symptoms
- On examination, Chvostek's and Trousseau's signs positive

Investigations

ECG

QTc 544ms (female: 350-450)

Biochemistry

cCa: 1.18mmol/L (2.20-2.60)

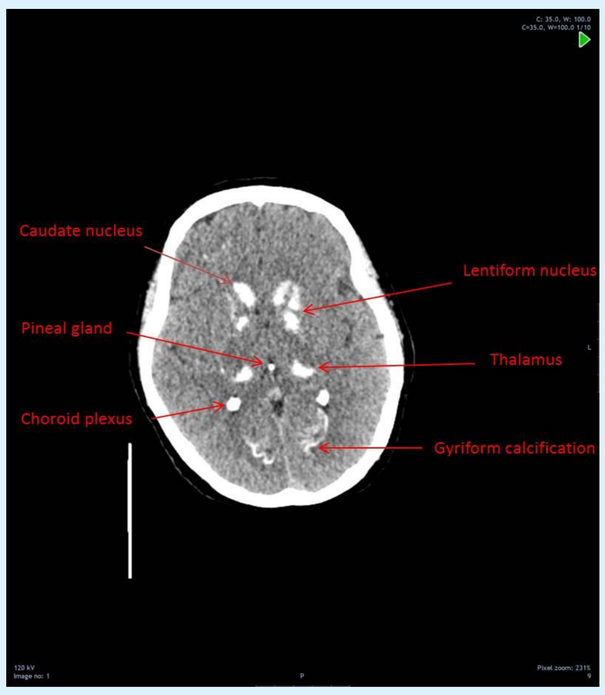
Phosphate: 2.19mmol/L (0.80-1.50)

PTH: <0.7pmol/L (1.6 -6.9)

25-OH Vitamin D: 50nmol/L (51-163)

Biochemistry in keeping with primary hypoparathyroidism

CT head



CT head: oblique axial (above left) and coronal (above right) images showing extensive dense bilateral basal ganglia, thalamic, and cerebellar dentate nucleus calcification.

In light of the investigation results above, a diagnosis of **Fahr's syndrome associated with idiopathic hypoparathyroidism** was made.

Management and Outcome

- Initial treatment was with intravenous calcium gluconate (20 ml of 10% calcium gluconate in 50 ml of 5% Dextrose IV over 10 minutes) followed by a calcium gluconate infusion.
- 48 hours following this treatment her corrected calcium had risen to 1.89mmol/L (2.20-2.60) and she remained seizure free. She was therefore switched to oral calcium (Sandocal® 1000 one tablet twice daily) and vitamin D (alfacalcidol 250 nanograms once daily) and discharged with endocrinology outpatient follow up.
- At her most recent outpatient review she denied any muscle spasms or paraesthesia and had been seizure free. There were no signs of neuromuscular excitability and her corrected calcium was stable. She reported feeling "better than ever" and that her "life has started again."

Conclusion

- Our case highlights the importance of excluding endocrine and metabolic abnormalities in all patients with basal ganglia calcification and neuro-psychiatric presentation before diagnosing Fahr's disease. In this case, lack of investigation for these abnormalities 10 years ago possibly resulted in a delayed diagnosis and treatment of idiopathic hypoparathyroidism.
- Treatment of Fahr's syndrome is directed at correcting the specific metabolic abnormality, for example correction of hypocalcaemia, which reduces seizure frequency.[2, 4] In contrast, only symptomatic therapies such as anti-epileptics or anti-psychotics can be used in Fahr's disease.[2] This therefore emphasises the importance of distinguishing between Fahr's disease and Fahr's syndrome as it affects both treatment and prognosis.

References

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