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Case Report

Fahr's syndrome presenting as focal seizures in an adolescent: a case report

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ABSTRACT

Fahr's syndrome refers to a rare syndrome which is characterized by symmetrical and extensive intra-cranial calcification. It mostly presents in age group 30-40 years or in older adults, although few cases have also been reported in paediatrics. The usual presentation is in the form of movement disorders, cognitive impairment, neuro-psychiatric manifestations etc. Here we report a case of a 14-year-old adolescent male who came to us with complaints of chronic headache and recurrent episodes of focal seizures. His cranial radio-imaging revealed bilateral basal ganglia calcification. On metabolic workup, he was found to have hypocalcemia and secondary hyperparathyroidism, thus confirming the diagnosis of Fahr's syndrome.

Keywords: Fahr's syndrome, Basal ganglia calcification, Movement disorders, Hypocalcemia

INTRODUCTION

Fahr's disease is a rare neuro-degenerative disorder which is characterized by abnormal calcified deposits in the basal ganglia and cerebral cortex. It was first described by a German neurologist Karl Theodor Fahr in 1930 in an 81-year-old patient with epileptic seizures and diffuse calcification of basal ganglia.[1] It is a rare disease with a prevalence of <1/1,000,000.[2,3] The etiology is unknown in patients who present with idiopathic basal ganglia calcification, but it may be secondary to certain metabolic or infectious diseases. The pathogenesis is not known, but it may be secondary to defective iron transport and free radical production, which leads to tissue damage and initiation of calcification.[4] Fahr's disease usually manifests in the 3rd or 4th decade of life, but few cases have been reported in chilhood.[5] Most cases present with extra-pyramidal movement disorders. But there may be other manifestations like seizures, dementia, cerebellar dysfunction, cognitive decline, parkinsonism, neuro-psychiatric symptoms etc.[6] We report a 14-year-old male child who presented to us with complaints of chronic headache and recurrent episodes of left sided focal seizures and was found to have the features of Fahr's syndrome on evaluation.

CASE REPORT

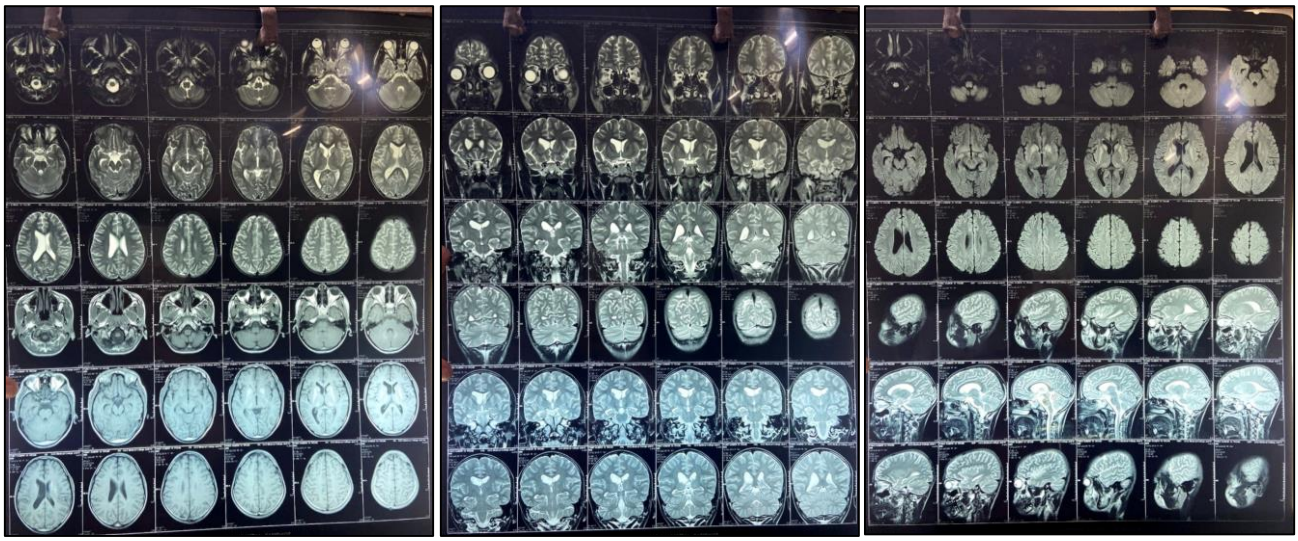
A 14-year-old male child, born of non-consanguineous marriage, presented to Emergency with complaints of headache on and off for the past 6 months and recurrent episodes of left sided focal seizures for the past 2 months. The headache was insidious in onset, generalized, non-progressive, not associated with any diurnal variation, no associated photo or phonophobia and used to get relieved with oral medication (paracetamol). There was no associated vomiting, fever, visual disturbances or altered consciousness. He also had history of left side abnormal body movements (tonic-clonic) involving upper limb more than lower limb and were not associated with secondary generalization. These episodes lasted for 2-3 minutes and used to occur every 10-15 days. There was

no associated gait abnormality reported by parents. There was no history of any psychiatric manifestations. There was no history of head trauma in the past. The child was developmentally normal and immunized as per age. There was no family history of similar illness and he had a younger sibling aged 10 years who was asymptomatic. He had sought treatment from local practitioner but no anti-epileptic was started. Last episode of seizures occurred 2 days prior to the current admission. On examination, the child was well looking with normal vital parameters. His weight was 38 kg (10th-25th centile) and height was 155 cm (25th-50th centile). His higher mental functions were intact there were no neuro-cutaneous markers and no facial dysmorphism. Cranial nerve examination was normal, including vision and hearing evaluation. There were no signs suggestive of meningeal irritation or cerebellar dysfunction. Motor system examination revealed mild spasticity of left upper limb more than left lower limb. Rest of the motor and sensory examination was within normal limits.

Routine investigations were done with normal septic workup and non-contributory CSF examination. Electroencephalographic recording was suggestive of intermittent right temporal discharges against a normal background. Metabolic profile showed hypocalcemia (serum calcium 6.0 mg/dl, ionized calcium 3.2 mg/dl), mild hyperphosphatemia (serum phosphorous 5.8 mg/dl), normal serum magnesium and albumin levels and 25 hydroxy vitamin D levels in the insufficient range (13.5 ng/ml).

Ultrasonography of kidney, ureter and bladder was not suggestive of any stones. Skeletal survey was also done and there was no evidence of fracture. Serum parathormone evaluation was done by chemi-luminescence assay and it was found to be high (128.6 pg/ml against a reference range of 14-72 pg/ml). Cranial imaging was done in view of focal seizures which revealed hyperintensities in bilateral basal ganglia, thalami and dentate nuclei suggestive of mineralization (calcification).

To evaluate for other causes of intra-cerebral calcification, CMV IgG and HIV serologies were negative. The patient was diagnosed clinically and radiologically to have features suggestive of Fahr's syndrome. He was started on valproate after which the seizures were controlled. Vitamin D and calcium supplements were also started in therapeutic doses and patient was discharged in stable condition.



a) **b)** **c)**

Figure 1 (a-c): Cranial imaging.

DISCUSSION

Bilateral striopallido dentate calcinosis, which is otherwise known as idiopathic basal ganglia calcification, is a neuro-degenerative disorder popularly known as Fahr's disease. It is distinct from Fahr's syndrome, which is thought to be due to some underlying disorder.Exact etiology of the disease is not known. It maybe idiopathic primary basal ganglia calcification (Fahr's disease) or secondary to certain endocrine disorders (hypoparathyroidism, pseudohypoparathyroidism, hyperparathyroidism etc), infections (HIV, EBV etc), radiotherapy, chemotherapy etc (Fahr's syndrome). Some cases have an autosomal dominant mode of inheritance.[7] A loss of function mutation in the gene coding for type 3 sodium dependent phosphate transporter 2 (SLC20A2) located on chromosome 8 has been implicated in the pathophysiology of this disease.[8-10]

Most common presentations as per the Fahr's disease registry are movement disorders, accounting for 55% cases. Other neurological manifestations include cognitive impairment, speech disorders, cerebellar signs, psychiatric features, gait abnormalities, seizures etc.[11] The imaging findings of symmetrical and extensive

cerebral calcification are usually typical, which were also seen in our case. The clinical diagnosis of Fahr's disease requires a combination of clinical features, brain imaging and exclusion of other causes of intra-cranial calcification. Basal ganglia calcification may occur as a consequence of several other known genetic, infectious and metabolic conditions.[12] In our case, infections like CMV, HIV, toxoplasmosis and neurocysticercosis were eliminated after workup. Other causes include hypothyroidism, hypervitaminosis D, tuberous sclerosis, cerebral hemorrhage, SLE, radiotherapy etc. which were also not seen in the present case. However, our patient had hypocalcemia, insufficient vitamin D levels and mild hyperphosphatemia on metabolic workup.

This led to secondary elevation of serum parathyroid hormone levels, leading to secondary hyperparathyroidism. The treatment includes mainly symptomatic support. Our patient did well with sodium valproate and calcium and Vitamin D supplements. Treatment of underlying process may lead to some improvement in patients with neuro-psychiatric features. Prognosis is variable, unpredictable and is not related to the extent of cerebral calcification.[13]

CONCLUSION

Fahr's disease/syndrome should be considered as a differential diagnosis in children presenting with seizures, movement disorders or cognitive decline. Classical radiological finding of bilateral basal ganglia calcification should prompt search for underlying infectious, metabolic or vascular etiology so as to differentiate Fahr's syndrome from other causes of cerebral calcification. Patients of Fahr's syndrome presenting in paediatric age group should be followed up regularly for development of neuro-psychiatric manifestations.

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