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CASE STUDY

A RARE CASE OF BILATERAL STRIATOPALLIDODENTATE CALCINOSIS IN A YOUNG BOY PRESENTING WITH SEIZURES

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ABSTRACT

Idiopathic calcification of basal ganglia and cerebellar dentate nucleus is synonymous with Fahr's disease, which is often an autosomal dominant, rarely occurring neurodegenerative disorder. Few sporadic autosomal recessive cases have also been described. Symptomatology of the Fahr's disease ranges from movement disorders, Parkinsonism like symptoms to cognitive defects. Majority of the cases reported were in adults with typical presentation. Here we report a rare occurrence of Fahr's disease in a young boy presented only with seizures.

Key words: Calcification, Basal Ganglia, Seizures.

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INTRODUCTION

Idiopathic symmetric calcification of basal ganglia, cerebellar dentate nuclei of Globus pallidus region and centrum semiovale is synonymous with Fahr's disease (Smits *et al*., 1983). Various types of movement disorders and dementia are its prominent clinical features (Shakibai *et al*., 2005). Extrapyramidal and pyramidal tract symptoms such as gait disturbances, paresis, spasticity, chorea, tremors, dystonia, myoclonia and parkinsonism like symptoms have been associated with this disease. Cognitive impairment, behavioral disturbances such as apathy, intermittent disinhibition, anxiety, ritualistic and anti-socialbehavior, irritability, frequent mood changes and psychosis are the psychological disturbances attributed to Fahr's disease (Manyam *et al*., 2001; Lam *et al*., 2007). It has been demonstrated that Fahr's disease is autosomal dominant, most likely linked to chromosome 14q and is frequently familial (Victor and Ropper; Geschwind *et al*., 1999). Some sporadic autosomal recessive cases have also been reported (Geschwind *et al*., 1999). There have been studies associating TULIP1 coding in Fahr's disease but these are far from being conclusive (Brodaty *et al*., 2002; Oliveira *et al*., 2009; Oliveira *et al*., 2007). The patho physiology of Fahr's disease is less understood but it has been hypothesized that one or the combination of the following factors such as- local disruption of blood- brain barrier, metastatic calcification secondary to disturbances in calcium metabolism resulting from hypoparathyroidism, hyperparathyroidism, pseudo hyperparathyroidism, anoxia, irradiation, toxins and systemic disorders (Malik *et al*., 2004). If it is metastatic calcification indeed, particular proclivity for above detailed structures needs also to be elicited. We report a singular case of idiopathic Fahr's disease in a young male with seizures, which is an uncommon presentation.

CASE REPORT

An eighteen years old male patient presented to the casualty with recurrent episodes of tonic-clonic convulsions. He was a

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known case of complex partial seizures with secondary generalization on antiepileptic medications since last five years. His symptoms were insidious in onset, gradually progressed over years. Seizures occurred randomly once or twice in 2-3 days. There was no history of fall, headache, neck rigidity, diplopia, vomiting, post-ictal weakness, behavioral disturbances, movement disorders or gait disturbances. He was reasonably well at his studies. He was born to non consanguineous parents and his birth was by normal vaginal delivery and there were no neonatal complications. He had attained his mile stones normal for his age. There was no history suggestive of previous illness other than seizures. There was no history of similar complaints or parkinsonism like features and behavioral disturbances in his family members. On examination, his vitals were within normal limits. His BMI was 18.75. General survey was unremarkable, except for hypertrichosis all over the body which might be secondary to the long term use of phenytoin. His IQ was normal for his age. His mini mental status examination was normal. He had attained secondary sexual characters normal for his age.

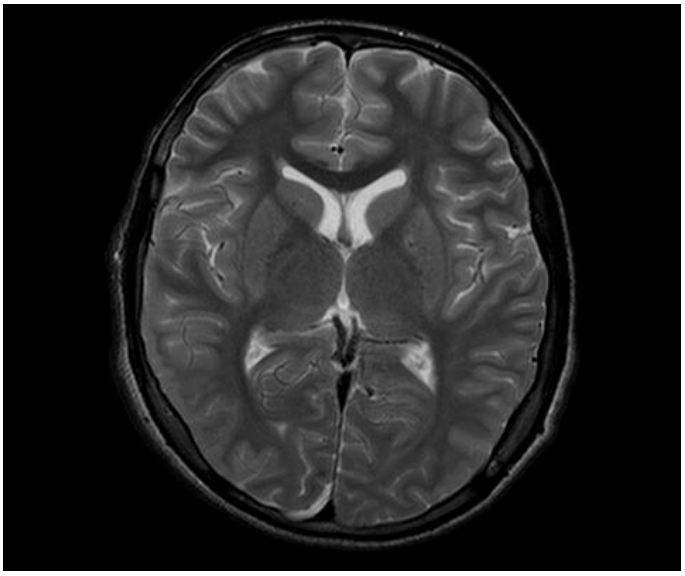

Figure 1. MRI brain showing T2W hypo intense areas involving bilateral head of caudate and lent form nuclei

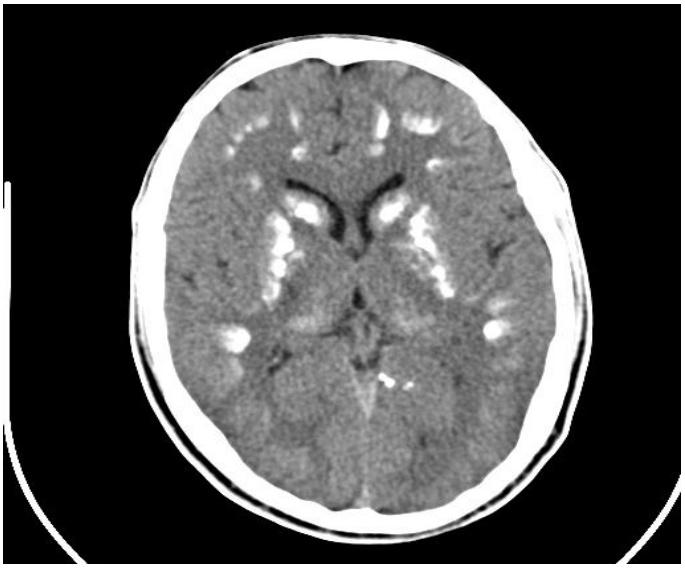

Figure 2. Plain CT Brain Showing dense symmetrical calcification of bilateral caudate and lent form nuclei

His systemic examination was un remarkable Routine investigations such as- CBP, Peripheral smear, ESR, Blood sugar, Serum Creatinine, Electrolytes, Serum Calcium & Phosphate, Urine examination, Stool microscopy, Chest X Ray and ECG were within normal limits. He was screened for HIV, Hepatitis B and Hepatitis C, Tuberculosis, Malaria and other infectious diseases and all of them were negative. Assays of thyroid and parathyroid hormones were normal. His Vitamin D assay and Serum ceruloplasmin were normal. Cranial MRI scan (Figure 1) revealed multiple T1W hyper intense and T2W is to mildly hypo intense areas involving bilateral dentate nuclei, bilateral head of caudate nuclei, bilateral lentiform nuclei, bilateral thalami and bilateral frontal and parietal gyri. Lesions are profoundly hypo intense on gradient sequences and have shown near symmetry. Complimentary plain cranial CT (Figure 2) revealed dense symmetrical calcifications involving the regions of brain parenchyma as described in the MRI findings. Patient was diagnosed to be suffering from Fahr's disease based on the above radiological features.

DISCUSSION

Clinical diagnosis of Fahr's disease is based on clinical, radiological features and by the exclusion of other causes. None of the symptoms classically described in literature detailing Fahr's disease were present in this patient (Sobrido *et al*., 2007). Diagnosis was arrived at only after reviewing the radiological reports. Absence of family history further confirms that this is a sporadic, probably autosomal recessive case of Fahr's disease (Geschwind *et al*., 1999). Age of onset of the disease in this case is much earlier than that of most of the cases described in the literature (Yasuhiko *et al*., 2005). The differential diagnosis of Fahr's disease includes- hypoparathyroidism, pseudo hypoparathyroidism, hyperparathyroidism, other causes resulting in metastatic calcification such as- neoplasms, cerebrovascular lesions, infectious diseases such as- toxoplasmosis, syphilis and inflammatory conditions such as- SLE, which have been ruled out in this case (Goodwin, 2006). Prenatal or genetic testing is not available yet for counseling for Fahr's disease. There is neither a cure nor a definitive treatment strategy. This patient was treated symptomatically and antiepileptics have been stepped up to the maximum tolerable doses. It's hard to predict the prognosis in this case at this stage.

Conclusion

The age of onset of the Fahr's disease can be much earlier i.e. in teens and behavioural disturbances and movement disorders may not be associated always with this disease. The extent of calcium deposition in the brain need not correlate with the symptomatology. The prognosis of this disease is variable and is very difficult to predict. Secondary causes of abnormal calcium deposition in brain need to be searched for before establishing the diagnosis. Fahr's disease may be added to the differential diagnosis of seizures.

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