CASE STUDY

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

*1Dr. Venkata Pradeep Babu, 2Dr. Ankush Jajodia, 3Dr. Krishna Venivanapala, 3Dr. Pavani Medisetty, 4Dr. Rajat Bajaj, 5Dr. Krushna Chaudhary and 5Dr. Bharat Vyas Marla

1,5Department of Medical Oncology, Resident, Rajiv Gandhi Cancer Institute and Research Centre, New Delhi, India
2Department of Radiology, Rajiv Gandhi Cancer Institute and Research Centre, India
3Neurology Resident, Vydehi Institute of Medical Sciences and Research Centre, India
3Senior Resident, Siddartha Medical College, Vijayawada, India
4Senior Resident, Rajiv Gandhi Cancer Institute and Research Centre, India
5Consultant, Mythri hospital, Khammam, India

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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-social behavior, 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 pathophysiology 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
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 milestones 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
second 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.

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 (Yasuhiro 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,
infected 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.

REFERENCES

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disease) without neurological, cognitive and psychiatric
symptoms is not linked to the IBGC1 locus on chromosome
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