

Case Reports

Fahr's Disease: A Rare Neurodegenerative Disorder in Children

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Introduction:

Fahr's disease is a rare degenerative neurological disorder characterized by the presence of abnormal calcium deposits and associated cell loss in the areas of brain that control movement, including basal ganglia and cerebral cortex¹⁻⁴. The condition is often referred to as idiopathic basal ganglia calcification, because there is no apparent explanation for such calcification. Fahr's disease was first noted by German neurologist Karl Theodor Fahr in 1930. The pathogenesis is not known, but may be secondary to impairment of the blood brain barrier or to a neuronal calcium phosphorus metabolism disorder¹. According to reports in medical literature, Fahr's disease is often familial^{5,6}. It is believed to have autosomal dominant inheritance but a few cases have been reported to have autosomal recessive inheritance and even some sporadic cases have been reported in literature. The association between the abnormal phenotypes and abnormal genes remain unclear despite the recent mapping to chromosome 14q of a susceptible locus for Fahr's disease⁷. The onset is frequently seen in the fourth and sixth decades of life, though occasional cases have been reported in children⁵.

Case report:

A 2 year-9 month old male child, immunized as per EPI schedule, only issue of non-consanguineous parents presented with delayed developmental milestones. He achieved his neck control at 4 months of age. He attained his sitting with support at the age of 2 years (fig:1) and could not stand or walk. The child also developed seizure for one episode which involved the face and upper limbs with no neurological deficit. He uttered only Ma... Ba... Da... etc. There was no family history of any similar illness or any other neurological disorder. His birth history was uneventful. General examination was normal. CNS

examination showed increased tone in all four limbs especially lower limbs. Deep tendon reflexes were exaggerated with bilateral Babinski sign. Sensory function and cranial nerves were intact. Slit lamp and fundus examination were normal. All the other systems were normal on examination. Blood chemistry revealed normal serum levels of calcium, phosphorus, alkaline phosphatase and vitamin D. And the serum parathormone level was 13pg/dl (normal range; 9-65pg/dl). Mantoux test was negative. TORCH titre was done and found negative. Chest x-ray did not reveal any abnormality. Plain radiographs of the skull demonstrated irregular calcifications in bilateral frontoparietal region of skull. A contrast enhanced CT scan of brain was done which revealed bilaterally symmetrical non-enhancing hyperdense lesions of calcification, involving basal ganglia, thalami and subcortical white matter (fig:2). The patient could not afford MRI study. The CT scan findings when correlated with clinical history and normal blood chemistry were suggestive of Fahr's disease. His seizure was controlled with carbamazepine and put on a regular outpatient follow up. Only the conservative treatment was advised to avoid respiratory tract infection. Physical and speech therapy was advised.



Fig.1: A 2yr-9month child can sit only with support.

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Fig.-2. CT scan image showing multiple symmetric calcifications in basal ganglia, thalamus, subcortical white matter.

Discussion:

Fahr's disease or familial idiopathic basal ganglia calcification is characterized by bilateral basal ganglia calcification. The most common site of calcification is the globus pallidus. However, additional areas of calcification are putamen, caudate nucleus, internal capsule, dentate nucleus, thalamus, cerebellum and cerebral white matter. The calcium deposits occur in the extracellular and extravascular space often surrounding the capillaries^{8,9}. It is not clear whether the calcification in Fahr's disease is a metastatic deposition, secondary to local disruption of blood brain barrier, or is due to disorder of neuronal calcium metabolism. The clinical evolution is that of a degenerative disorder, rather than a developmental disorder. The core clinical features are dysarthria, extrapyramidal signs, ataxia, and progressive deterioration of mentality, loss of motor accomplishments, symmetrical spastic paralysis, athetosis, and eye impairment due to optic atrophy.

Radiological diagnosis could be the starting point in guiding the clinician to the possibility of Fahr's disease.

CT scan is considered to be the best modality of investigation in the diagnosis of Fahr's disease where unenhanced CT reveals dense calcifications within the basal ganglia, subcortical white matter of the posterior parietal lobes, and the dentate nuclei of the cerebellum. On magnetic resonance imaging (MRI), the signal may be variable. On T1 weighted images, low signal is due to the low proton density of calcium and other mineral ions present in higher concentration. However, they might present hyperintense signal, due to proteins and mucopolysaccharides binding the mineral ions. The calcification might also be undetected on MRI when they are in an intermediary stage¹⁰.

Fahr's disease, however, needs to be distinguished from Fahr's syndrome in which basal ganglia calcification is secondary to some other disorders, such as hypoparathyroidism. Basal ganglia calcification may also be seen in various other conditions like CMV infection, toxoplasmosis, neurobrucellosis, tuberculosis, astrocytomas, calcified infarct, pseudohypoparathyroidism, hyperparathyroidism, hypervitaminosis D, mitochondrial encephalopathies, and leukodystrophic diseases¹.

There is neither a cure for Fahr Disease, nor a standard course of treatment. Case reports have suggested that haloperidol or lithium carbonate may help in patients with psychotic symptoms¹¹. The prognosis is variable and hard to predict.

Conclusion:

In our case, the child presented with developmental delay and seizure. All the common causes of Fahr's syndrome were excluded and CT scan showed classical symmetrical basal ganglia involvement. This rare case of idiopathic Fahr's disease in a child, which has never been reported in Bangladeshi literature, has been brought out to highlight this unusual condition and its differentiation from the commoner Fahr's syndrome. Patients diagnosed with idiopathic Fahr syndrome should be monitored, and regular neuropsychiatric evaluation should be performed.

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