CASE REPORT

Fahr's syndrome

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Abstract

Miss Merry, 17 years old girl hailing from Barobila., Patkelghata, Satkhira was admitted in Khulna Medical College Hospital on 27-06-2010 with the complaints of fever, convulsion and unconsciousness for 7 days. She has also some hearing impairment, behavioral abnormalities and stunted growth since her childhood. On examination she was deeply unconscious, anaemic and febrile. CT scan of brain reveals multiple bilateral symmetrical calcification seen in the brain parenchyma involving basal ganglia, thalamus, para ventricular region, and cerebellar nucleus. Multiple ill defined hypodense areas are seen in the both parieto-occipital region, suggestive of Fahr's syndrome with meningoencephalitis.

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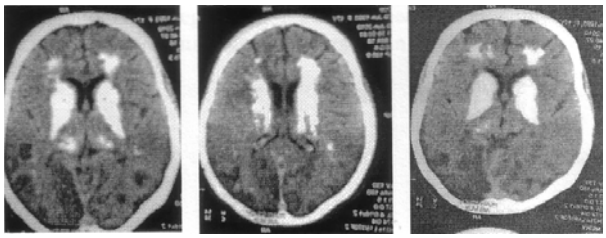
Introduction

Idiopathic Basal Ganglia Calcification, also known as Fahr's disease or Fahr's Syndrome or Bilateral StriatoPallidoDentate Calcinosis (BSPDC) is a rare, genetically dominant, inherited neurological disorder characterized by abnormal deposits of calcium in areas of the brain that control movement, including the basal ganglia and the cerebral cortex. The disease was first noted by German neurologist Karl Theodor Fahr in 1930.1 According to reports in medical literature, Fahr Disease is often familial. 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. Idiopathic calcification of the basal ganglia, also known as Fahr's disease, is a rare neurologic disorder of unknown etiology characterized by neuropsychiatric abnormalities.2 Parkinsonian or choreoathetotic-type movement disturbance, and extensive symmetrical calcification of the basal ganglia and dentate nuclei in the cerebellum. These symptoms cannot be explained by any other particular disorder of the calcium phosphorus metabolism or any other disease. Dementia is a well-recognized neuropsychiatric, manifestation of Fahr's disease. In addition, a schizophrenia-like psychosis characterized by paranoia, hallucinations, and delusions has been reported.3 There is no cure for

Fahr's syndrome, which worsens over time, nor is there a standard course of treatment.

Case report

Miss Merry, 17 years old girl hailing from Barobila, Patkelghata, Satkhira was admitted in Khulna Medical College Hospital on 27-06-2010 with the complaints of fever, convulsion and unconsciousness for 7 days. According to mother's statement she developed fever with unconsciousness 7 days back. Fever was high grade and continued in nature. She also developed convulsion for the same duration. The convulsion was generalized. She had vomiting and loose motion but not mixed with blood during the initial period of fever. There was no history of discharge from ear or cough. There was no history of contact with patients of pulmonary tuberculosis. She has also some hearing impairment, behavioral abnormalities and stunted growth since her childhood. She underwent appendicectomy 5 years back. Menarche is not yet established. She is immunized according to national immunization schedule. Her parents are alive and healthy. She has four brothers. Her father and grand mother is of low intelligence and have some behavioral abnormalities. On examination she was deeply unconscious, anaemic and febrile. On neurological examination- Plantar response was extensor, deep reflexes were exaggerated. Pupil were normal in size and reacting to light. Fundoscopic examination reveals no abnormalities. Examination of other systems reveal no abnormalities. Investigations reveals Hb- 62%, ESR- 140mm in 1St hour, TC-10700/cmm, DC-P-68%, L-27%, E-45%, M-O1%, B-0%, Urine analysis shows massive proteinuria, Pus cell 6-8 / hpf, RBC 0-2 / hpf, RBS- 7.11 mmol/L, S. calcium-11 mg/dl, S. phosphate- 4 mg/dl, S. creatinine- 1.2 mg/dl, Na+-132 mmol/L, K+-3.9 mmol/L, C1"- 102.9 mmol/L, S.T4-135.21 nmol/L, S.TSH- 0.62 mIU/L. CT scan of brain reveals multiple bilateral symmetrical calcification seen in the brain parenchyma involving basal ganglia, thalamus, para ventricular region, and cerebellar nucleus. Multiple ill defined hypodense areas are seen in the both parieto-occipital region, suggestive of Fahr's syndrome with meningoencephalitis.



CT axial images show calcifications in the bilateral basal ganglia

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Discussion

The disease was first noted by German neurologist Karl Theodor Fahr in 1930. Fahr's Syndrome is a rare degenerative neurological disorder characterized by calcifications and cell loss within the basal ganglia. The calcium deposits in the brain may occur before the onset of the symptoms, usually in the third decade of life. Although it may also be evident in childhood4,5 and with advancing age the amount of calcification increases. In Fahr's disease the mineral deposits tend to be selective for small capillaries and small vessels of white matter, which is different from that in atherosclerosis.6 The calcification may include endothelial and stromal vascular cells as well as the interstitium. However, the local circulatory disturbances such as regional ischemia7 have been regarded as the primary event precipitating the deposition of calcium as well as other minerals. Other contributed factors are abnormality in the calcium metabolism8 or local inflammatory process.7 Also, the calcification could be a primary event occurring without preceding circulatory dysfunction, since a significant familial type suggests either autosomal recessive or dominant inheritance.4 Brain calcification without symptoms such as the small calcifications in the basal ganglia, and less commonly in the dentate nucleus of the cerebellum can occur in elderly

patients. According to reports in medical literature, Fahr Disease is often familial. 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 Disease.9

Symptoms may include motor function deterioration, dementia, mental retardation, spastic paralysis, dysarthria, stiffness of the limbs, ocular problems and athetosis. Features of Parkinson's disease such as tremors, rigidity (resistance to imposed movement), a mask-like facial appearance, shuffling gait, and a "pill-rolling" motion of the fingers may also occur in individuals with Fahr's syndrome. Other symptoms may include dystonia (disordered muscle tone), chorea (involuntary, rapid, jerky movements), and seizures. Onset of the disorder may occur at any time from childhood to adulthood. Fahr syndrome thus involves abnormalities of the neurologic system (cerebral calcification, dementia, spastic paraplegia, athetosis), skull (microcephaly, i.e. an abnormally small head), eyes (glaucoma, optic nerve atrophy, retinitis pigmentosa), and a significant hormone problem, namely hypoparathyroidism (the parathyroid gland regulates calcium). The disease is inherited as an autosomal recessive trait in which both parents carry a Fahr gene and each of their children (boys and girls alike) stands a 1 on 4 (25%) risk of receiving both Fahr genes and therefore having this dreadful disease.

Fahr's disease is also characterized by neuropsychiatric abnormalities, Parkinsonian or choreoathetotic-type movement disturbance, and extensive symmetrical calcification of the basal ganglia and dentate nuclei in the cerebellum.2,6 These symptoms cannot be explained by any other particular disorder of the calcium phosphorus metabolism or any other disease.8 Dementia is a well-recognized neuropsychiatric manifestation of Fahr's disease. In addition, a schizophrenia like psychosis characterized by paranoia, hallucinations, and delusions has been reported.3 The pathophysiology of psychosis in Fahr's disease remains unknown, though previous studies have found a decreased cerebral blood flow matching the distribution of calcification or decreased perfusion in the cortex, which may reflect secondary deficits due to calcification. Radiological diagnosis could be the starting point to guide the clinician for possibility of Fahr's disease. The differential diagnosis includes but not limited to; Parkinson's disease, Huntington's disease, progressive supranuclear palsy, Wilson's disease, spasmodic torticollis, oligodendroglioma, lowgrade astrocytoma and arteriovenous malformation. Therefore Fahr's Disease or Bilateral StriatoPallidoDentate Calcinosis (BSPDC) is a diagnosis of exclusion.12,13

There is no cure for Fahr's syndrome, which worsens over time, nor is there a standard course of treatment. The process of calcification cannot be stopped or reversed. Treatment is directed toward minimizing symptoms. Where possible, clinicians focus on alleviating its various mental and physical effects. These may vary to some degree depending on the individual, even among members of the same family. Case reports have suggested that haloperidol or lithium carbonate may help with psychotic symptoms, while antidepressant medications are often used to combat depression.10 Ear infections associated with Fahr disease can be treated with antibiotics and pain medication.The prognosis for any individual with Fahr's Syndrome is variable and hard to predict, and is often poor. There is no reliable correlation between age, extent of calcium deposits in the brain, and neurological deficit. Since the appearance of calcification is agedependent, a CT scan could be negative in a gene carrier who is younger than the age of 55.11 Progressive neurological deterioration generally results in disability and death.

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