Familial Intracranial Calcification

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A 62 year old man presented with unsteadiness of gait, ataxia, frequent falls and abnormal movement of extremities. He had dragging of both feet and difficulty in walking since 5 years. Neurological examination revealed spastic dysarthria, choreoathetotic movement of upper limbs, face and tongue, bilateral rigidity, truncal ataxia, and bilateral positive cerebellar signs.

Computer tomography of brain revealed bilateral symmetrical calcification of basal ganglia (Fig. 1A), thalamus, cerebellum (Fig. 1B), frontal and parietal cortex. Serum calcium, phosphorus, and magnesium were normal. Assay for parathyroid hormone and thyroid function tests were normal.

His elder son had seizure disorder since 15 years of age. Computed tomography of brain of his elder son showed bilateral symmetrical basal ganglia calcification (Fig. 2). The final conclusion of Fahr syndrome was made on the basis of history, bilateral familial intracranial calcification and normal metabolic parameters like serum calcium, phosphorus, magnesium and parathyroid hormone.

Fahr’s disease is a rare degenerative neurological disorder characterised by the presence of abnormal calcium deposition and associated cell loss in the areas of the brain that control movement, including basal ganglia and cerebral cortex.1

The condition was first described by Fahr in 1930.2 This can be also be referred as ‘familial idiopathic basal ganglia calcifications,’ ‘bilateral striopallidodentate calcinosis,’ or ‘idiopathic nonarteriosclerotic intracerebral calcifications.’

Fahr’s disease is often familial. Familial Fahr’s Disease may be transmitted as an autosomal dominant inheritance or autosomal recessive trait.

Fahr’s disease is characterized clinically by seizures, extrapyramidal and neuro-psychiatric signs as a result of bilateral diffuse calcifications of the basal ganglia, dentate nucleus and white matter. The age at onset of clinical symptoms is 30 to 50 years. The patient usually present with parkinsonism, choreoathetosis, and cerebellar ataxia, pyramidal signs, psychiatric symptoms, and urinary incontinence.3

Calcification of the basal ganglia is observed as an incidental finding in approximately 0.7 to 1.2% of CT scans.4

The important causes for basal ganglia calcification are secondary hypoparathyroidism, pseudohypoparathyroidism, pseudo-pseudo-hypoparathyroidism, hypothyroidism, perinatal asphyxia, carbon monoxide intoxication, lead intoxication, Fahr’s syndrome (familial idiopathic symmetrical basal ganglia calcification), Cockayne’s syndrome, tubersclerosis, parkinsonism, vascular diseases, cerebral hemorrhage, radiation therapy and methotrexate treatment.

The important differential diagnosis of basal ganglia calcification of familial nature are, Fahr’s syndrome (Familial idiopathic symmetrical basal ganglia calcification), Cockayne’s syndrome, tubero sclerosis, and familial degenerative disorders. Aicardi-Goutières syndrome is an autosomal recessive encephalopathy which causes developmental arrest, basal ganglia calcification, and white matter disease in the presence of chronic cerebrospinal fluid lymphocytosis, and a raised level of cerebrospinal fluid interferon-alpha.

The treatment is usually symptomatic and there is no cure for the disease. Progressive neurological deterioration generally results in disability and death.

References