Case Report

Fahr's syndrome

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ABSTRACT

Fahr's syndrome is a rare, neurological disorder characterized by abnormal calcified deposits in basal ganglia and cerebral cortex. We report a 48 year old post-menopausal woman who presented with episodes of generalised tonic clonic seizures and abnormal posturing of hands and feet. She had carpopedal spasms and decreased alertness. MRI Brain showed extensive basal ganglia calcifications and laboratory investigations were suggestive of hyperparathyroidism, hypocalcaemia and hyperphosphatemia. We treated her with calcium supplements which showed dramatic improvement. The current case and its review of literature is an attempt to remark on the current substantial evidence surrounding this disease.

Introduction :

Fahr's syndrome is a rare, neurological disorder characterized by abnormal calcified deposits in basal ganglia and cerebral cortex. Calcified deposits are made up of calcium carbonate and calcium phosphate, and are commonly located in the basal ganglia, thalamus, hippocampus, cerebral cortex, cerebellar subcortical white matter and dentate nucleus. It is a rare inherited or sporadic neurological disorder with a prevalence of <1/1,000,000[1-3]. It was first described by German neurologist Karl Theodor Fahr in 1930[4]. It is characterized by abnormal deposition of calcium in areas of the brain that control movements including basal ganglia, thalamus, dentate nucleus, cerebral cortex, cerebellum, subcortical white matter, and hippocampus[5].

Case Report :

48-year-old, postmenopausal female presented with complaints of recurrent episodes of generalized tonic clonic seizures associated with tongue bite. She had abnormal posturing of hands on and off, extreme fatigue since 10 days. She had similar complaints in the past and was started on anticonvulsants since two years. H/o discontinuation of anticonvulsants since 15 days prior to admission. There was no past history of thyroid surgery, trauma or head injury. Her family history was not significant. Her vitals were stable. She had recurrent carpopedal spasms & trousseau's sign was positive. There were no dysmorphic features, mucocutaneous candidiasis, or vitiligo. Patient was conscious but had decreased alertness and was slow in comprehension. There was decreased attention span and overall sluggishness in her movements. She was responding to verbal commands, rest of the neurological exam was normal. Investigations revealed normal complete blood picture. Liver function tests were normal with normal albumin 5.2 gm%. KFT was within normal limits. Total serum calcium 6.7 mg/dL, Ionized calcium 2.52 mg/dl,Serum Parathyroid hormone 6.4 pg/mL were all found to be low. Phosphate level was raised; 6.3 mg/dL. Serum TSH, Vitamin D and Magnesium levels were normal. Initially MRI brain was done, as patient presented with seizures which revealed extensive basal ganglia calcifications on susceptibility weighted imaging. As calcifications are best seen on CT Brain, this was also done showing clear calcifications, also covering frontal and occipital areas. Our patient was diagnosed to have Fahr's syndrome secondary to idiopathic hypoparathyroidism. Patient was started on anticonvulsants and calcium supplements. This led to drastic improvement in her cognitive abilities and reduced overall sluggishness. Patient is being followed for the last 6 months and has no episode of seizures or abnormal posturing of extremities since then.

Discussion :

Fahr's syndrome is defined by basal ganglia calcifications. It can occur in multitude of conditions. Fahr's syndrome can occur in endocrinal

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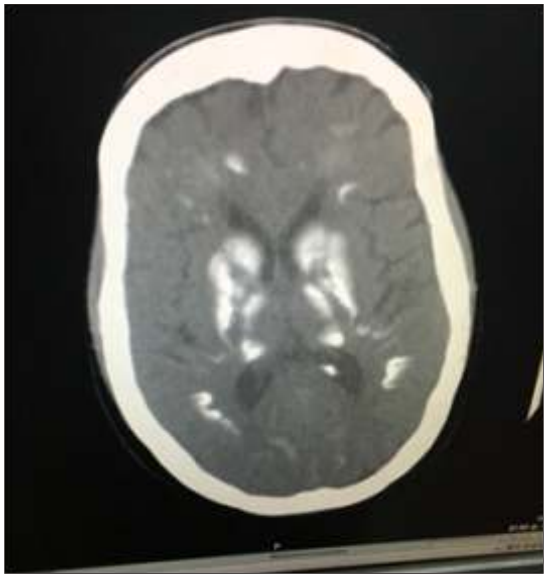

Figure 1 : CT image showing bilateral symmetric calcifications throughout the basal ganglia and the bilateral gangliocapsular region involving the lentiform nucleus and the caudate

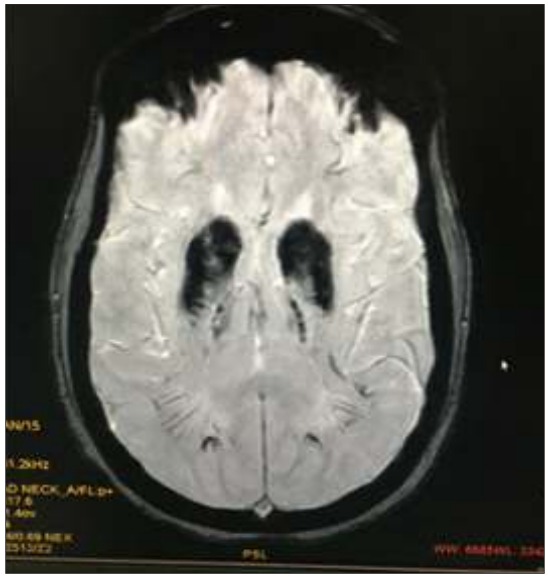

Figure 2 : Specific sections of susceptibility weighted imaging specific for calcifications

conditions like secondary hypoparathyroidism, idiopathic hypoparathyroidism, pseudohypoparathyroidism, pseudopseudohypoparathyroidism and Hyperparathyroidism[5]. Other important causative conditions include Fahr's disease, mitochondrial myopathies, dermatological abnormalities and infectious diseases. Fahr's disease is one of the cause of this syndrome. It is a rare, autosomal dominant inherited neurological disorder characterized by primary basal ganglia calcification.

CT scan is the best modality to diagnose this condition. Abnormal deposits of calcium occur in areas of the brain that control movement., primarily in the basal ganglia and also in other areas such as the cerebral cortex. Clinical manifestations of this disease incorporate a wide variety of symptoms, ranging from neurological symptoms of extrapyramidal system to neuropsychiatric abnormalities of memory and concentration to movement disorders including Parkinsonism, chorea and tremors amongst others. Diagnostic criteria for this disease consist of bilateral calcification of basal ganglia, progressive neurologic dysfunction, absence of biochemical abnormalities, absence of an infectious, traumatic or toxic cause, significant family history in Fahr's disease. Imaging modalities for the diagnosis include CT, MRI, and plain radiography of skull. Other investigations include blood and urine testing for hematologic and biochemical indices. Disease is as yet incurable but management and treatment strategies mainly focus on symptomatic relief and eradication of causative factors.

Conclusion :

Fahr's syndrome is a rare condition, with very little review of literature available on this date; however certain evidence is present to suggest that early diagnosis and treatment can reverse the calcification process leading to complete recovery of mental functions.

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