Fahr's Syndrome

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Introduction

Fahr syndrome is a rare degenerative neurological disorder characterized by abnormal deposits of calcium and associated cell loss in parts of brain that control movements including basal ganglia and cerebral cortex.

German neurologist Karl Theodor Fahr first describes this condition in 1930. It may be autosomal dominant and autosomal recessive. However, some cases are reported sporadic. The calcifications in brain are bilaterally symmetrical in basal ganglia and cerebral cortex.

Presence of bilaterally symmetrical basal ganglia calcification with neuropsychiatric symptoms and hypoparathyroidism is known as Fahr's syndrome.

Presence of bilaterally symmetrical basal ganglia calcification with neuropsychiatric symptoms and familial association is known as Fahr disease.

Case report

A 45 yrs right handed female patient presented in casualty of Dr. PDM medical college, Amravati with pertubation, uneasiness, irritability, unexpected anxiety, chocking sensation while breathing, headache, auditory hallucinations & delusion of reference and abnormal involuntary jerky movements of hands and feet.

Detailed history reveled there were similar episodes on and off since 18 yrs whose frequency was increasing from 1-2 episodes per year to present episodes of 10-12 per year. She also gives history of gradual increase in tremors, pill-rolling movements of fingers, muscle rigidity, and shuffling gate. Tremors and pill rolling movements became more obvious during the attack. She also gives past history of tetany on 2-3 occasions. There were no history of fever, convulsions, drug intake, and exposure to toxins, trauma to head or history of similar complaints in family. She has undergone bilateral cataract extraction 15 yr back. She did not have a history of DM/HTN/IHD/Asthma.

She was married for 12 yrs and had two live issues- 10 yr and 8 yrs. Her obstetric history was unremarkable and those attacks were not exacerbated during pregnancy.

Her general examination was normal except mild pallor. In CNS examination, patient was conscious, oriented but her mood was agitated and anxious. She had good self-care and normal speech pattern and her interest to her environment was normal. Her memory, perception, orientation, and attention were within normal limits. Quantitative and qualitative judgment was normal.

Her emotional state was distorted as anxietic her thought content was focused on the pre occupation of her illness and her conduct was anxietic and disturbed. The patient had auditory hallucinations & delusions of reference .There were no sensory or motor deficit except for tremors, muscle rigidity, dystonia and choreoathetosis.

Laboratory investigations – CBC, ESR, AST, ALT, FBS, Sr. sodium, Sr. potassium were normal except Hb%- 9.6 gm%, Sr. Calcium (ionized) – 1.72 mg/dl (normal 3.5-4.9) and corrected after treatment is 1.96 mg/dl., Sr. PTH - <3 (normal 12-72).

EEG/ECG was normal. Fundus examination showed Early Papilledema

Clinical diagnosis of Organic neurogenic disorder was put forth.

CT scan done – which reveals bilateral symmetrical cerebellar, basal ganglia & deep nuclei calcifications.

Treatment – patient was treated with IV Calcium Gluconate 10ml in D5% 12hrly for 3 days the switch to oral calcium 500 mg BD. And Haloperidol 0.25 mg HS.

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Her clinical symptoms responded well to above treatment in 3 days. Her anxiety and tremors were decreased.

Discussion

Fahr's syndrome is a rare idiopathic disorder with age of onset for symptoms 3rd or 4th decade. There is no sex predilection. Pathologically, Calcium deposition is selective for small capillaries & small vessels of white matter. There is endothelial, stromal vascular cell & interstitial calcification Region ischemia is regarded as precipitating cause. There is abnormality in calcium metabolism & local inflammatory process. Calcifications could be primary – autosomal dominant / recessive trait & familial disease.

Diagnosis of Fahr's syndrome is by exclusion only as there were many differential diagnoses for intracranial calcifications. Nevertheless, radiological bilateral symmetrical basal ganglia calcifications with typical neuropsychiatric symptoms and hypoparathyroidism should clinch the diagnosis of Fahr's syndrome.

The condition should be differentiated for intracranial calcifications. These include

1. Idiopathic
2. Physiological
3. Congenital
4. Inflammation / infection
5. Trauma
6. Toxic
7. Endocrine

   Hypo / hyperPTH

   Hypothyroidism
8. Metabolic

Leigh disease, Mitochondrial cytopathy, Fahr's syndrome.

Prognosis is highly variable and very hard to predict. There is no correlation between age, sex, amount of calcification, and neurological deficit.

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Fig1. Bilateral dentate nuclei (Cerebellar) calcifications and bilateral basal ganglia calcifications.

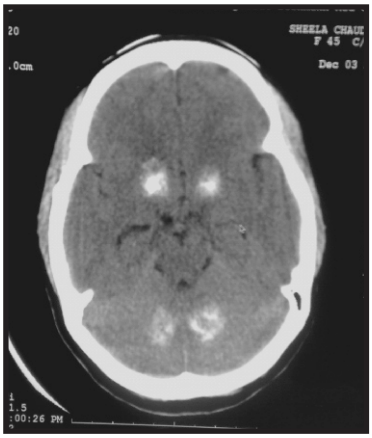

Fig 2. Bilateral dentate nuclei (Cerebellar) calcifications

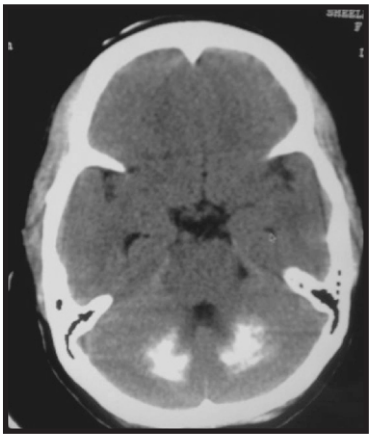

Fig 3. Bilateral basal ganglia calcifications

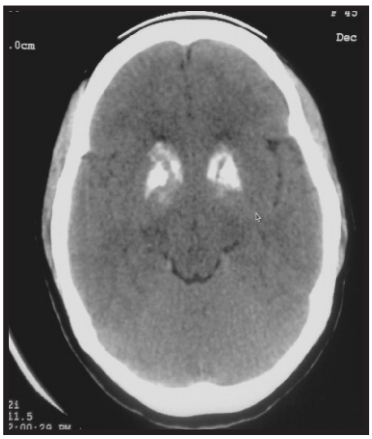

Fig 4. Bilateral basal ganglia calcifications & Calcifications of internal capsules & lateral thalami

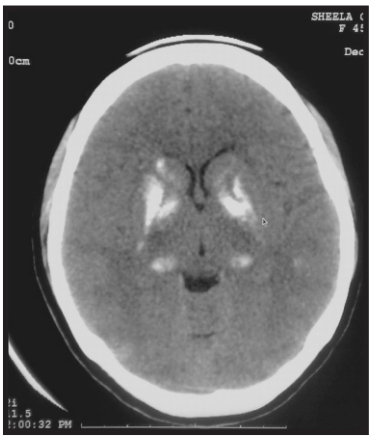

Fig 5. Calcifications of lateral thalami & Calcification of deep nuclei

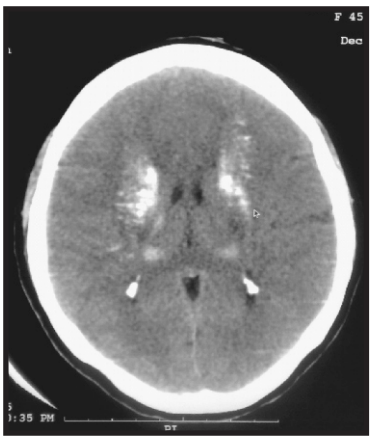

Fig 6. Calcification of deep nuclei