

Fahr's Syndrome – Clinico-Radiological Diagnosis of A Rare Condition.

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Abstract: Fahr's syndrome is a rare inherited neurodegenerative disease, characterized by the presence of bilateral and symmetrical intracerebral calcifications due to unknown etiology. It usually manifests as headache, seizures and movement difficulties. Here we discuss a patient who presented with seizure disorder and headache for the past 2 weeks. Patient was referred to our department for MRI study of the brain which revealed features suggestive of Fahr's disease with bilateral symmetrical basal ganglia calcifications and conditions such as hypoparathyroidism and tuberous sclerosis was excluded. This case further aims to substantiate the clinical and radiological features of this disease.

Keywords: Fahr's disease, Fahr's syndrome, intracerebral calcification, Magnetic resonance imaging (MRI), Hypoparathyroidism.

I. Introduction

Fahr's disease also known as idiopathic striopallidodentate calcinosis is a rare inherited neurodegenerative disease with a prevalence of < 1 / 1,000,000. Fahr's disease is characterized by presence of bilateral and symmetrical intracerebral calcifications in the basal ganglia, thalamus, dentate nucleus and centrum semiovale due to unknown etiology.

II. Case Report

A 44 year old female, who was a known case of seizure disorder for 32 years presented with complaints of uncontrolled seizure activity with headache, slurring of speech, difficulty in walking, bilateral ptosis and vision impairment for past three months. She was also a known case of Diabetes mellitus, hypertension and primary amenorrhea with no family history of neurological disorder.

On physical examination, patient was conscious but disoriented to time and place with dysphonia. Patient's blood pressure was under control. Neurological examination showed less visual acuity on both sides, increased hypertonicity and impaired cerebellar tests (co-ordination, gait and Rhomberg's test).

Patient was referred for MRI brain, which revealed multiple punctate T1 hyperintense and T2/FLAIR hypo to isointense lesions involving the bilateral ganglio-capsular region (caudate nucleus, lentiform nucleus, thalamus and internal capsule) and subcortical white matter bilaterally (Fig:1). The lesions show blooming on gradient recalled echo(GRE) (Fig:2). With these findings Fahr's disease was considered. Mineralizing microangiopathy is less likely.

Computed tomography was done, which showed dense calcifications involving bilateral ganglio-capsular and bilateral cerebellar hemispheres (Fig:3), thus confirming the magnetic resonance imaging findings. X-ray skull AP and lateral showed abnormal calcifications (Fig:4).

Blood investigations showed decrease in hemoglobin (8gm/dl), reduced serum calcium (5.8mg/dl) (hypocalcemia), increase in phosphorus (10.7mg/dl), decreased parathyroid hormone (9.6pg/ml). Other routine blood investigations and blood sugar levels were within normal limits.

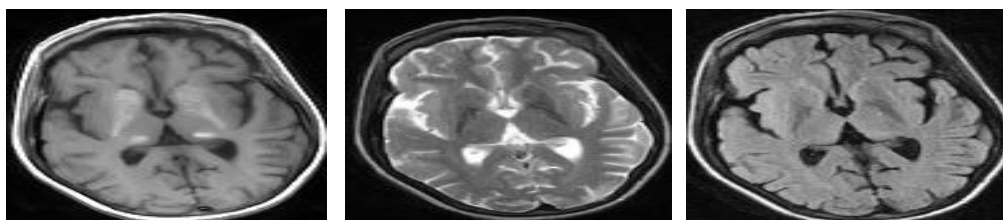


Figure. 1: Magnetic Resonance Imaging (MRI) T1, T2 and FLAIR axial sections: Multiple punctate T1 hyperintense and T2/ FLAIR hypointense signals noted involving bilateral ganglio-capsular region.

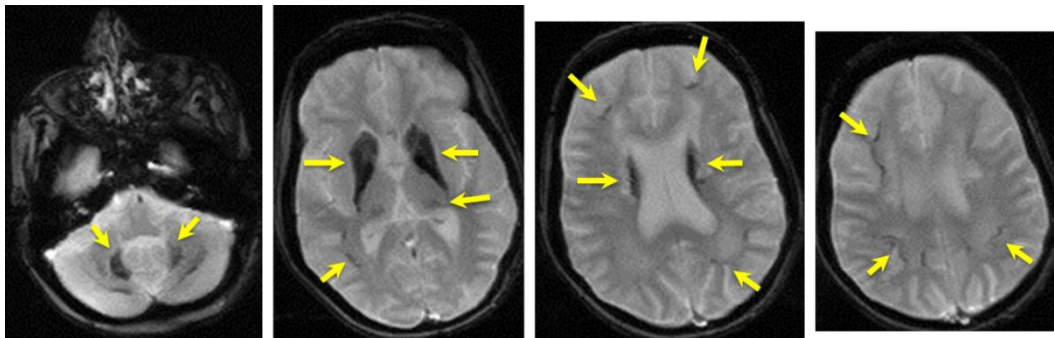


Figure. 2: Gradient Recalled Echo (GRE) Axial: The lesion shows blooming on GRE images in the ganglio capsular region (the caudate nucleus, lentiform nucleus, thalamus & internal capsule were seen to be involved). Blooming was also noted in the bilateral subcortical white matter (involving the fronto-parietal and occipital regions) and in both the cerebellar hemispheres.

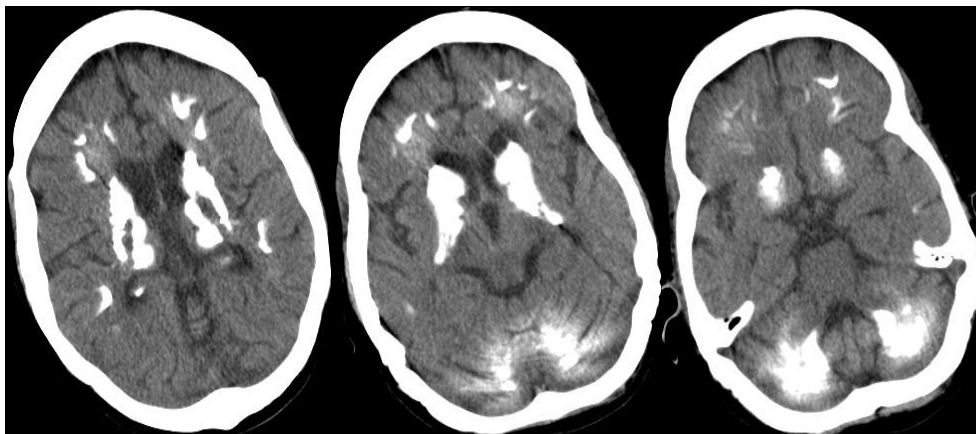


Figure. 3: CT brain plain (axial sections): Dense calcifications noted involving bilateral ganglio-capsular, subcortical white matter regions and both cerebellar hemispheres.

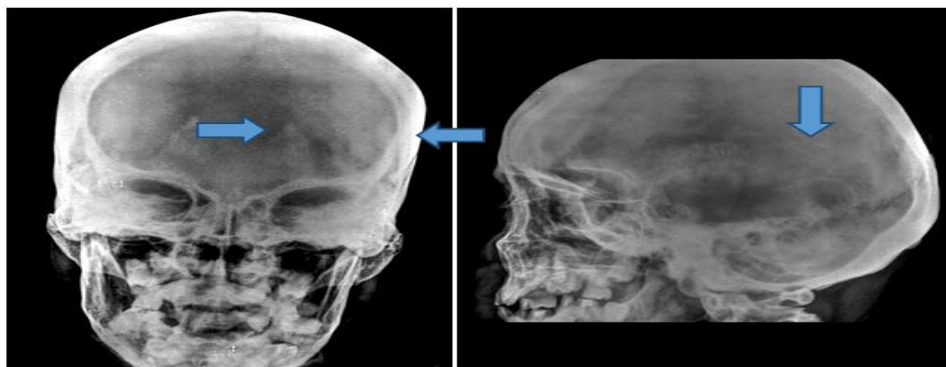


Figure. 4: X-ray skull AP and Lateral : Abnormal calcifications identified.

III. Discussion

Fahr's disease is also referred by other names like Familial idiopathic basal ganglia calcification/Bilateral striopallidodentate calcification/Idiopathic familial cerebrovascular ferrocalsinosis.

Basal ganglia calcifications are of 2 types

- **Primary**(idiopathic) - Fahr's Disease
- **Secondary**(mainly endocrine) - Fahr's Syndrome

Fahr's syndrome (FS): Fahr's syndrome includes Fahr's disease and secondary causes of striopallidodentate calcinosis [1]. It is a rare, chronic, slowly progressive, genetically dominant, inherited

neurodegenerative disorder with incidence of < 1 / 1,000,000 with male female ratio (2:1). It is characterized by extensive bilateral deposition of calcium in the basal ganglia, thalamus, cerebral cortex, dentate nucleus, cerebellum subcortical white matter, and hippocampus associated with endocrine disorders like hypoparathyroidism.

Clinical manifestations occur usually in the fourth or fifth decade of life and includes neuropsychiatric manifestations with gradually progressive cognitive impairment[2,3]. Most of these conditions are systemic diseases and the reason for the focal accumulation of calcium in basal ganglia is unknown. The most common neurological signs are headache, vertigo, movement disorders, syncope and seizures. Other specific neurological deficits consist of paresis, spasticity, gait disturbance, speech disorders, coma, dementia, Parkinsonism, chorea, tremor, dystonia, myoclonia and orthostatic hypotension[4].

There are various differentials for bilateral basal ganglia calcifications which are broadly classified as idiopathic, toxic, infectious, metabolic and inherited conditions. Physiological calcification due to aging is the most common cause of basal ganglia calcifications[5]. Close differential for Fahr's syndrome with diffuse bilateral symmetric striopallidodentansosis is primary hypoparathyroidism[6,7]. Other close differentials are lupus, tuberous sclerosis, Alzheimer's disease, myotonic muscular dystrophy and mitochondrial encephalopathy[8]. Laboratory investigations should include blood calcium and parathormone levels in addition to other routine blood investigation which helps in differentiating idiopathic Fahr's syndrome from secondary conditions like hypoparathyroidism.

IV. Conclusion

Although rare, Fahr's disease and Fahr's syndrome are conditions that should be kept in mind in all the cases of progressive neuropsychiatric disturbances and seizure disorders, particularly if they present in the fourth or fifth decade of life. In such cases, it is important to perform radiological investigations & laboratory tests for the purpose to find out any metabolic abnormalities. In cases of Fahr's syndrome due to hypoparathyroidism, the neurological and psychiatric symptoms usually improve with normalization of plasma calcium and phosphorus levels, so timely diagnosis and follow up is important.

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