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Psychiatric Manifestation In A Fahr's Disease

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Abstract

Background

Fahr's disease or Idiopathic Basal Ganglia Calcification is a rare autosomal dominant neurodegenerative disorder, with prevalence <1/1,000,000 and common among 3rd to 4th decade of life. Caused by abnormal calcium deposition in basal ganglia, dentate nucleus, thalamus, cerebral cortex, subcortical white matter, cerebellum, and hippocampus. Characterized by rigidity, tremor, choreathetosis, hypokinesia, ataxia, seizure, psychosis, mood disorders, anorexia nervosa, mania, dementia, fugue state, cognitive dysfunction, abnormalities in thyroid, parathyroid, calcium, sodium and phosphate metabolism, .

Case Presentation:

23yr-old-female presented with disorganized, withdrawn, disinhibited, threatening and wandering behaviour, associated with suspiciousness towards others, unprovoked anger outburst, sleep disturbance, and staring mirror with repetitive stereotypic hand movements for past 2years, episodic course with inter-episodic spontaneous recovery. No significant past history, nor genetic loading. Extremely good functioning pre-morbidly. General physical examination were normal. Neurological examination revealed rigidity. Mental status examination revealed eye contact not sustained, rapport established with difficulty, retarded psychomotor activity, speech decreased in rate/tone/volume; increased reaction time, constricted affect, delusion of persecution present. Attention aroused but concentration wasn't sustained. Provisional diagnosis was psychosis unspecified. Managed with Risperidone (2mg), subsequently patient developed dystonia hence switched over to Aripiprazole (5mg), with Trihexyphenidyl (4mg) and clonazepam (0.5mg) and reported 50% improvement in symptoms. MRI revealed bilateral symmetrical T1 hyper-intensity, involving bilateral globus pallidus, substantia nigra and along dentate nucleus. Blooming on SWI sequence (suggestive- Fahr's disease). Medication stopped without advice resulting in worsening of symptoms, hence restarted and attained remission. Thyroid-Function-Test & Parathyroid-hormone were normal.

Conclusion:

40%of Fahr's disease present with psychotic features and behavioral disturbances as they share common pathway of sub-arachnoid space dilatation. Hence a probable differential diagnosis in atypical psychotic presentation with motor abnormalities. This case emphasizes role of neuroimaging to rule out structural disorders in atypical psychotic presentation, which helps in optimizing antipsychotics in short and long term to prevent long term neurological disorder.

Keywords: Fahr's disease, basal ganglia calcification, autosomal dominant neurodegenerative disorder

Introduction

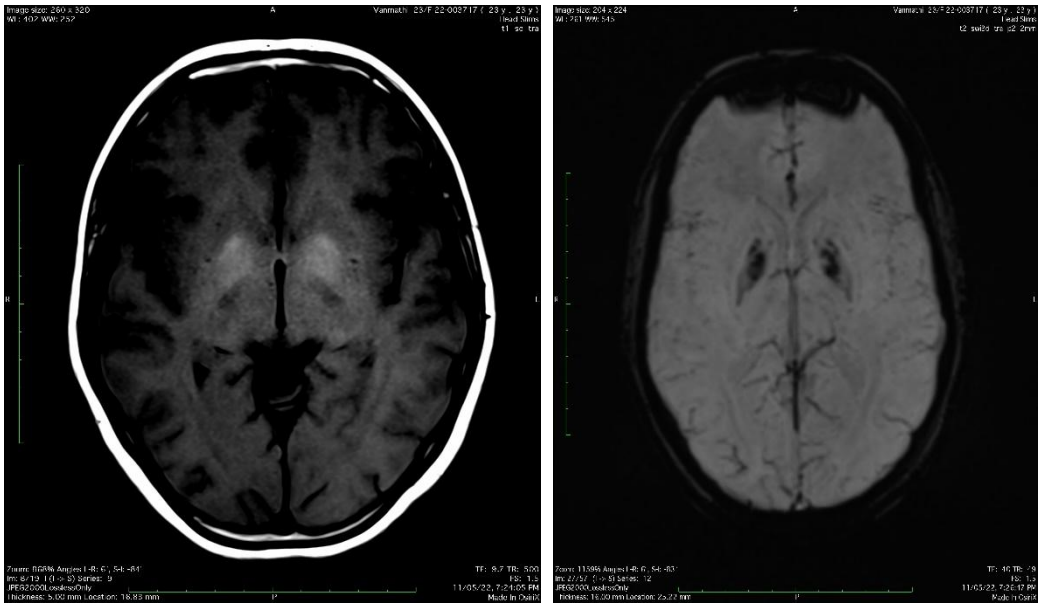
Fahr's disease or Idiopathic Basal Ganglia Calcification is a rare autosomal dominant neurodegenerative disorder. [1] It is also known as Fahr's syndrome, striopallidodentate calcification, or calcinosis nucleorum. [2] It has a prevalence of <1/1,000,000. It is common among 3rd to 4th decade of life. It is caused by abnormal, progressive and gradual deposition of calcium in basal ganglia, dentate nucleus, thalamus, cerebral cortex,

subcortical white matter, cerebellum, and hippocampus. Lenticular nucleus, chiefly the globus pallidus are most frequently involved. Calcification may begin at areas external to the basal ganglia.[2]

It is characterized by rigidity, tremor, choreathetosis, hypokinesia, ataxia, seizure, psychosis, mood disorders, anorexia nervosa, mania, dementia, fugue state, cognitive dysfunction, and abnormalities in thyroid, parathyroid, calcium, sodium and phosphate metabolism. [2]

Case Report

A 23yr-old-female presented with disorganized behaviour like locking up kitchen utensils in bedroom cupboard, withdrawn, disinhibited, threatening & wandering behaviour, associated with smiling to self, suspiciousness towards others, unprovoked anger outburst, sleep disturbance, & staring mirror for abnormally long hours with pill rolling like repetitive stereotypic hand movements for past 2years, episodic course with inter-episodic spontaneous recovery. No significant past history, no genetic loading of psychiatric disorders & good functioning pre-morbidly. General physical examination: CNS: mild tremors, mild rigidity, other findings were within normal limits. Mental status examination: eye contact not sustained, rapport established with difficulty, retarded psychomotor activity, speech decreased in rate/tone/volume; increased reaction time, constricted affect, delusion of persecution present. Attention aroused but concentration wasn't sustained. MRI revealed bilateral symmetrical T1 hyperintensity, involving bilateral globus pallidus, substantia nigra and along dentate nucleus. Blooming on SWI sequence(suggestive of Fahr's disease); Thyroid-Function-Test & Parathyroid-hormone were within normal limits. Provisional diagnosis of psychosis unspecified was made & initially managed with Risperidone(2mg), subsequently patient developed dystonia hence switched over to Aripiprazole(5mg), with Trihexyphenidyl(4mg) and clonazepam(0.5mg) and reported 50% improvement in symptoms. Medication stopped without advice resulting in further worsening of symptoms, hence restarted and attained remissions.



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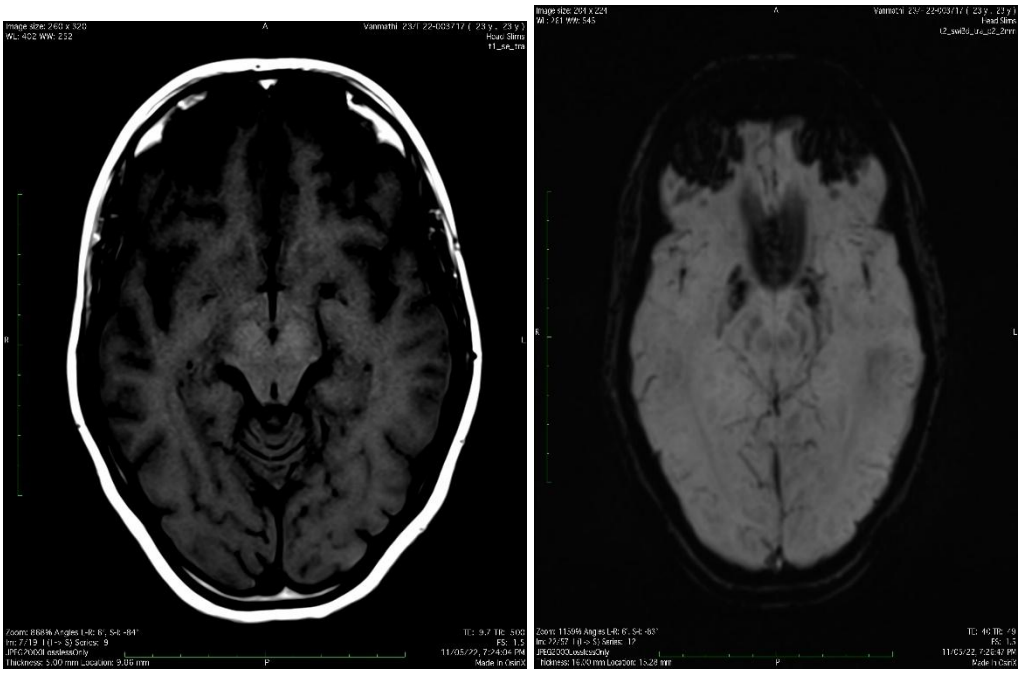

Discussion

Fahr's disease is arare autosomal dominant neurodegenerative disorder, with a prevalence of <1/10,00,000. With an incidence ranging from 0.5-10% in various studies. More commonly seen among 3rd to 4th decade of life.

Pathogenesis involves the calcification in vessel wall & peri-vascular space due to defective iorn transport & free radical production, resulting in initiation of calcification and nidus formation, leading to progressive mineralization, compressing vessel lumen & impairing the blood flow.[2]

Studies have revealed that autosomal dominant idiopathic basal ganglia calcification is caused by mutation of SLC20A2 gene in certain families. It was also found to be linked to IBGC1 and IBGC2 mapped on chromosome 14 and 2.[1] Loss of function mutation of SLC20A2, which is a gene encoding for type III sodium dependent phosphate transporter 2 (SLC20A2), which is present on chromosome 8 is involved in the pathophysiology of fahr's disease. Locus at 14q (IBGC1) is commonly involved.[2]

Neurological manifestation associated with fahr's disease include rigidity, tremor, choreathetosis, hypokinesia, ataxia, seizure.[3] Psychiatric manifestation associated with fagr's disease includes psychosis, mood disorders, anorexia nervosa, mania, dementia, fugue state & cognitive dysfunction. [4] Endocrinal manifestation- abnormalities in Ca2+ metabolism, PTH, hypothyroidism, thyroid adenoma, grave's disease, hyponatremia & phosphate metabolism. [4]

40% present of Fahr's disease present with psychotic features as they share common pathway of disruption of subcortex especially in limbic system and sub-arachnoid space dilatation. [5,6] When basal ganglia involvement increases, the susceptibility to antipsychotic induced extra-pyramidal symptoms subsequently raises.

Various differential diagnosis for Fahr's disease includes latent tetany, parathyroid dysfunction, mitochondrial disease, calcified angiomas, Addison's disease, infections, encephalitides, any other disease involving calcification of brain regions. [2]

Management of fahr's disease involves initial radiological workup. Computed tomography is preferred for localizing and assessing the extent to which calcification had occurred.

Conclusion

Fahr's disease is a differential diagnosis in an atypical psychotic presentation with motor abnormalities. This case emphasizes role of neuroimaging to rule out structural abnormalities in any atypical psychotic presentation, & hence optimize treatment plan with short & long term goals to prevent further deficits, and optimize the use of psychotropic especially antipsychotic to prevent long term neurological side effects.

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