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Fahr's Disease, A Case of Misdiagnosis for Decades

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Abstract

Fahr's Disease (FD) is a rare, genetically dominant pattern with incomplete age-related penetrance. Still, it could also be transmitted as an autosomal recessive or sporadic patterns. Fahr's disease is a neurological disorder depicted by abnormal calcifications in the brain. Although it is thought to be caused by congenital disorders, endocrine disorders, mitochondrial dysfunctions, or infections, the cause of Fahr's disease involves four genes with loss-function mutations.

Case report: Here, we present the case of a 39-year-old patient, a rare case in which the patient was misdiagnosed. Initially, the patient presented with a history of essential tremors, frequent falls, knee pain, back pain, lower extremity paresthesia and weakness, urge incontinence of urine, anxiety, depression, and morbid obesity. The patient was diagnosed with Turner's, Leukodystrophy, and Mitochondrial Disease despite the patient having multiple brain magnetic resonance imaging (MRI) showing diffused white matter. The patient has never had an official testing or diagnosis of Fahr's disease, and her symptoms have been managed with primidone, baclofen, gabapentin, and selective serotonin reuptake inhibitors (SSRI).

Keywords: ***Fahr's disease; Neurodegenerative disease; Basal ganglia calcifications; Familial basal ganglia calcifications; Psychotic symptoms***

1. Introduction

Fahr's disease, aka bilateral strio-pallido-dentate calcinosis or primary brain calcification or calcinosis nucleorum, is a rare, genetically dominant neurological disorder characterized by abnormal calcification of basal ganglia and other areas such as the

thalamus, hippocampus, dentate nucleus, cerebral cortex, cerebellar subcortical white matter that control movement [1]. This abnormal calcium, copper, zinc, magnesium, or iron deposition is hypothetically thought to cause a locally altered blood–brain barrier which results in defective iron transport and free radical production to cause brain tissue damage that slowly extends to the entire neurons [2].

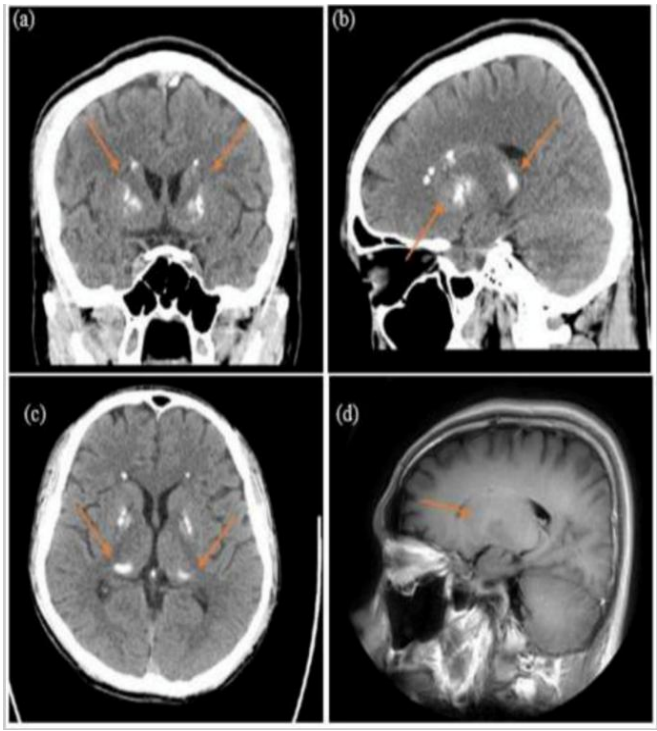

FIG. 1. **Basal ganglia calcification in the brain computed tomography: (a) coronal plane; (b) sagittal plane; (c)transverse plane. Basal ganglia calcification in the brain magnetic resonance imaging: (d) sagittal plane. https://www.ncbi.nlm.nih.gov/pmc/articles/PMC11148442/figure/f1/**

Four genes that plays a role as molecular basic of Fahr's disease are SLC20A2 loss of function mutation on chromosome 8p (40%), gene PDGFB on chromosome 22q (11%), XPR1 mutation on chromosome 1q (2%), and PDGFB mutation on chromosome 5q (2%). Other loci have linked to Fahr's disease include IBGC1 locus at chromosome 14q, a locus at chromosome 2q, and another locus at chromosome 8 [2].

The prevalence of the disease is less than 1/1,000,000,000 and most commonly presents in people aged 40-50 [3,4]. Patients are often in good health in their youth and tend to develop progressive neurodegenerative diseases later in adulthood.

Clinical presentations and manifestations of Fahr's disease can include movement disorder (parkinsonism, bradykinesia, rigidity, tremor, hypophonia, shuffling gait, dystonia, slurred speech, clumsiness, choreoathetosis) to neuropsychiatric symptoms (depression, apoplexia, dementia, subcortical dementia resembling in Wilson disease and Huntington disease) to other central nervous system manifestations (loss of consciousness, seizures, spasticity, speech impairment, myoclonus, papilledema, chronic headache, vertigo, urinary urgency or incontinence, impotence, severe hypertension) [5,6].

The diagnosis of Fahr's disease is diagnosed based on neuroimaging findings in brain computed tomography (CT) scan, magnetic resonance imaging (MRI) brain, fluorodeoxyglucose-positron emission tomography (FDR-PET), plain skull radiograph, and clinical symptoms. Molecular genetic testing may be considered in familial cases [7].

The disease's prognosis is unpredictable. There is no correlation between the age of onset, symptoms at onset, or extent of calcifications in the brain and the severity of the disease. Penetrance is age-dependent, with a 95% occurrence by age 50 [1].

2. Case Presentation

Part of the patient's history of present illness was obtained from the mother. The rest of the history was from chart review.

A 39-year-old female patient with a past medical history of essential tremors, frequent falls, knee pain, back pain, lower extremity paresthesia and weakness, urge incontinence of urine, anxiety, depression, morbid obesity, gastroesophageal reflux disease without esophagitis, chronic hearing loss of right ear, hereditary hemochromatosis, vocal cord paralysis status post intubation, subglottic stenosis, severe persistent asthma without complication, hypertension, and restrictive lung disease, was brought to the emergency department by her mom after a fall. On arrival, the patient was alert, awake, and oriented. She had no neurological deficits from the fall. Per the mother, this morning, when the patient was getting out of the shower, she slipped and fell, which was unwitnessed. The patient hit her head without loss of consciousness or urinary incontinence. There were no other concerns. The patient didn't articulate well and couldn't provide a history.

Per the mother, family history was significant for developmental delay and early onset hearing loss in the patient's mother, two maternal aunts, and a maternal uncle. The patient reached most of her milestones late in infancy. She has been ill since childhood. Doctors had told them that the patient had Turner's syndrome, Leukodystrophy, and Mitochondrial Disease but had never genetically been tested for either one. The patient was managed with Primidone 50 mg, gabapentin 300 mg, solifenacin 5 mg, baclofen 10 mg tablet, fluoxetine 20 mg capsule, bupropion 150 mg tablet, lidocaine 5% transdermal patch daily, and other hypertension medications.

During the physical exam, the patient was not distressed, and vitals were within normal limits. There was a rounded 2 cm - 3 cm hematoma to the occiput without tenderness. The rest of the physical and neuro exams were non-significant.

A head computed tomography without contrast was obtained to evaluate for skull fracture and intracranial pathology. The results showed evidence of Fahr's disease without any concussion or pathology from the fall. This comprehensive evaluation ruled out any immediate concerns from the fall. Fahr's disease appeared to be chronic and was getting worse. The patient and family weren't aware of the diagnosis. The patient was discharged home on the same day and she was recommended to follow up with her primary teams and discuss the computed tomography findings.

3. Discussion

The case emphasizes the importance of early diagnosis, management, and differential diagnosis for Fahr's disease. Basal ganglia calcifications are also present in other familial and non-familial pathologies that must be excluded before diagnosing primary familial basal ganglia calcification, Fahr's disease [8].

Fahr's disease is a neurological condition characterized by abnormal calcification deposits of the basal ganglia and other areas of the brain controlling movement [9]. It is inherited in an autosomal dominant pattern, autosomal recessive pattern, or sporadic [10].

Fahr's disease can present early or late onset. In the early onset cases, patients often present with psychiatric symptoms. In the last onset, patients usually present with movement disorders followed by dementia-like [11].

Non-familial cases of basal ganglia calcifications could be results of endocrine disorders (parathyroid hormone, hypoparathyroidism, pseudohypoparathyroidism, pseudo-pseudohypoparathyroidism) [12,13], infections (intrauterine and perinatal infections of herpes, cytomegalovirus, rubella, toxoplasmosis, HVI/AIDS), congenital disorders (Cockayne syndrome/Neill-Dingwall syndrome [12], Aicardi-Gouteres Syndrome, tuberous sclerosis, Coats plus syndrome), adult-onset disorders (neuroferritinopathy, spinocerebellar ataxia type 20, Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy), dermatological diseases (Lipoid proteinosis, Dyskeratosis Congenita), and other causes [14,15].

Although the patient presented with symptoms of Fahr's disease and was misdiagnosed since childhood, it could have been due to overlapping neurodegenerative and neuropsychiatric features with other diseases [4]. Although the patient presented with essential tremors, frequent falls, knee pain, back pain, lower extremity paresthesia and weakness, urge incontinence of urine, anxiety, depression, morbid obesity, other conditions may also present with these. The patient had been managed with bupropion 150 mg tablet, fluoxetine 20 mg capsule for anxiety and depression, primidone 50 mg tablet, baclofen 10 mg tablet, and gabapentin 300 mg capsule for movement symptoms [16].

Neuroimaging findings in brain computed tomography scan, magnetic resonance imaging brain, Fluorodeoxyglucose (PDG)-positron emission tomography (PET), plain skull radiograph, and clinical symptoms are the primary tools for diagnosing Fahr's disease. In addition, molecular genetic testing may be considered in familial cases [17].

However, there is currently no definitive treatment available for Fahr's disease. Symptoms can typically be managed with antileptics for seizures, pain killers, anticholinergics for incontinence, SSRIs for psychiatric symptoms, and neuroleptics for movement disorders. Physical and occupational therapy are important for patients to improve muscle strength, posture stability, gait, and spasticity [18].

4. Conclusion

Patients with Fahr's disease can present with a wide range of features, symptoms, and physical findings. Diagnosis of Fahr's disease can be mainly based on bilateral basal ganglia calcification on brain CT scan, MRI brain, FDG-PET, or plain skull radiograph. Other genetic testing and lab investigations are used to support the diagnosis. There is currently no available treatment for Fahr's disease. Management of Fahr's disease symptoms needs multiple disciplines to help improve the quality of life of patients suffering from the disease. It is important for families with risks to have genetic counseling, prenatal counseling, and annual surveillance.

5. Contributors

The authors wrote and edited the manuscript. The authors approved the final version of this manuscript and is responsible for all aspects of this study.

6. Competing Interests

None stated.

REFERENCES

1. Amisha F, Munakomi S. Fahr Syndrome. [Updated 2023 Aug 13]. In: StatPearls [Internet]. Treasure Island (FL): StatPearls Publishing; 2024 Jan-. Available from: https://www.ncbi.nlm.nih.gov/books/NBK560857/
2. Geschwind DH, Loginov M, Stern JM. Identification of a locus on chromosome 14q for idiopathic basal ganglia calcification (Fahr disease). Am J Hum Genet. 1999;65(3):764-72.
3. Li WC, Hsieh YC, Chen PT, et al. Idiopathic young-onset Fahr's disease with schizophrenia-like presentation: a case report. Front Psychiatry. 2024;15:1391607.
4. Saleem S, Aghemo K, Salmanzadeh R, et al. Fahr's syndrome: literature review of current evidence. Orphanet J Rare Dis. 2013;8:156.
5. Puvanendran K, Wong PK. Idiopathic familial basal ganglia calcification associated with juvenile hypertension. J Neurol Neurosurg Psychiatry. 1980;43(3):288.
6. Weisman DC, Yaari R, Hansen LA, et al. Density of the brain, decline of the mind: an atypical case of Fahr disease. Arch Neurol. 2007;64(5):756-7.
7. Aghemo K, Salmanzadeh R, DeAngelo O, et al. Advanced early-onset fahr's disease: A case report. Cureus. 2023;15:e39495.
8. Ukai K, Kosaka K. Diffuse neurofibrillary tangles with calcification (Kosaka-Shibayama disease) in Japan. Psychiatry Clin Neurosci. 2016;70(3):131-40.
9. Förstl H, Krumm B, Eden S, et al. Neurological disorders in 166 patients with basal ganglia calcification: a statistical evaluation. J Neurol. 1992;239(1):36-8.
10. Mohapatra S, Satapathy A. A case of schizophrenia like psychosis due to fahr's disease. Indian J Psychol Med. 2016;38:155-6.
11. Ostling S, Andreasson LA, Skoog I. Basal ganglia calcification and psychotic symptoms in the very old. Int J Geriatr Psychiatry. 2003;18(11):983-7.
12. Bruyn GW, Bots GT, Staal A. Familial bilateral vascular calcification in the central nervous system. Psychiatr Neurol Neurochir. 1964;67:342-76.
13. Maeda K, Idehara R, Nakamura H, et al. Anticipation of familial idiopathic basal ganglia calcification? Intern Med. 2012;51(8):987.
14. Raymond AA, Zariah AA, Samad SA, et al. Brain calcification in patients with cerebral lupus. Lupus. 1996;5(2):123-8.
15. Revesz T, Fletcher S, al-Gazali LI, et al. Bilateral retinopathy, aplastic anaemia, and central nervous system abnormalities: a new syndrome? J Med Genet. 1992;29(9):673-5.

16. Takashima S, Becker LE. Basal ganglia calcification in Down's syndrome. J Neurol Neurosurg Psychiatry. 1985;48(1):61-4.
17. Zisimopoulou V, Siatouni A, Tsoukalos G, et al. Extensive bilateral intracranial calcifications: a case of iatrogenic hypoparathyroidism. Case Rep Med. 2013;2013:932184.
18. Di Biase L, Munhoz RP. Deep brain stimulation for the treatment of hyperkinetic movement disorders. Expert Rev Neurother. 2016;16(9):1067-78.