Fahr Disease : A Case Report

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Abstract

Fahr's disease is a rare autosomal dominant neurodegenerative disorder characterized by bilateral and symmetrical intracranial calcifications, primarily affecting the basal ganglia, thalamus, dentate nucleus, and centrum semiovale. It presents with progressive neurological and psychiatric symptoms, and computed tomography (CT) remains the most sensitive diagnostic modality. We report the case of a 9-year-old girl who presented with generalized seizures lasting less than 15 minutes without fever. Neurological examination revealed mild dystonia and dysarthria. Non-contrast brain CT scan demonstrated multiple bilateral calcifications in the basal ganglia, thalamus, and frontoparietal cortex. Laboratory results, including leukocyte, neutrophil, monocyte, and lymphocyte counts, were normal, effectively excluding infectious causes. Due to limited resources, further investigations such as MRI and metabolic studies (serum iron, TIBC, calcium, vitamin D) were not performed. The CT findings, along with normal laboratory results, strongly support the diagnosis of Fahr's disease. This case highlights the importance of considering Fahr's disease in pediatric patients with seizures and abnormal CT findings. Comprehensive evaluation of metabolic and genetic etiologies, as well as family screening, is recommended.

Keyword : *Fahr Disease, CT Scan, Basal Ganglia*

Abstrak

Penyakit Fahr adalah kelainan neurodegeneratif autosomal dominan langka yang ditandai dengan kalsifikasi intrakranial bilateral dan simetris, terutama memengaruhi ganglia basal, talamus, nukleus dentata, dan centrum semiovale. Penyakit ini muncul dengan gejala neurologis dan psikiatrik progresif, dan tomografi terkomputasi (CT) tetap menjadi modalitas diagnostik yang paling sensitif. Kami melaporkan kasus seorang anak perempuan berusia 9 tahun yang datang dengan kejang umum yang berlangsung kurang dari 15 menit tanpa demam. Pemeriksaan neurologis menunjukkan distonia ringan dan disartria. Pemindaian CT otak tanpa kontras menunjukkan beberapa kalsifikasi bilateral di ganglia basal, talamus, dan korteks frontoparietal. Hasil laboratorium, termasuk jumlah leukosit, neutrofil, monosit, dan limfosit, normal, sehingga secara efektif menyingkirkan penyebab infeksi. Karena keterbatasan sumber daya, penyelidikan lebih lanjut seperti MRI dan studi metabolik (serum besi, TIBC, kalsium, vitamin D) tidak dilakukan. Temuan CT scan, beserta hasil laboratorium normal, sangat mendukung diagnosis penyakit Fahr. Kasus ini menyoroti pentingnya mempertimbangkan penyakit Fahr pada pasien anak dengan kejang dan temuan CT scan abnormal. Evaluasi komprehensif terhadap etiologi metabolik dan genetik, serta skrining keluarga, direkomendasikan.

Kata Kunci : *Penyaki Fahr, CT Scan, Basal Ganglia.*

Introduction

Fahr's disease, a rare genetic disorder, is a neurodegenerative abnormality of bilateral calcium deposition in the brain parenchyma. It is a rare disorder with a prevalence of < 1/1,000,000 (Peters et al., 2020; Thillaigovindan, Arumugam, Rai, R, & Kesavan, 2019; Wazir, Ali, Mufti, Ahmad, & Ahmad, 2023). It was first reported by Karl Theodor Fahr, a German neurologist, in 1930 (Wazir et al., 2023). Calcium deposits in the brain are common incidental findings in traumatic and related to aging as hyperdense lesions. However, symmetrical bilateral calcium buildup in the brain tissue, mainly in the basal ganglia, thalamus, cerebral cortex, subcortical white matter, dentate nucleus, and cerebellum, unaccompanied by underlying pathology, is termed Fahr's disease.(Carecchio, Mainardi, & Bonato, 2023) It generally shows an autosomal dominant inheritance, and seven genes have been implicated in its genetic aetiology, of which four with dominant inheritance (SLC20A2, PDGFB, PDGFRB, XPR1) and three with recessive inheritance (MYORG, JAM2, CMPK2) (Magalhães, Alves, Paulino Ferreira, Alves, & Durães, 2024). The inheritance pattern is autosomal dominant, predominanting in the fifth and sixth decade of life while the symptoms are most similar to parkinsonism like bradykinesia, rigidity, tremor, hypophonia, hypomimia, mask-like facies, and a shuffling gait (Wazir et al., 2023).

The diagnostic criteria for Fahr's syndrome include bilateral striatopallidodentate calcification on neuroimaging, progressive cognitive dysfunction, and movement disorders without biochemical, infectious, toxic, and traumatic causes (Wazir et al., 2023). The demonstration of bilateral basal ganglia calcifications on brain imaging (CT scan) and the exclusion of secondary causes of calcium deposition in the brain. Abnormal clinical findings on examination, as well as a positive family history of brain calcifications, neurological or psychiatric disorders may be absent; therefore, the

diagnosis can be formulated on a radiological basis only after excluding secondary causes. These include persistent hypocalcemia due to altered calcium metabolism, infectious and mitochondrial diseases, and other rare neurodegenerative conditions. The differential diagnosis is based on the patient's age, clinical history, examination and laboratory findings. Laboratory screening should include a complete assessment of calcium metabolism (parathyroid hormone, vitamin D, calcium and phosphate levels), lactic acid and CPK (may be elevated in mitochondrial disease) along with a brain CT scan which is the gold standard radiological method for visualizing cerebral calcifications (Donzuso, Mostile, Nicoletti, & Zappia, 2019). Until now, computed tomography scan remains the best imaging modality for Fahr's disease (Thillaigovindan et al., 2019).

CASE PRESENTATION

A 9 years old girl with history of generalised seizure without fever duration below 15 minutes, after that fall to behind then get a hematoma in region parietal sinistra. History of seizure occur when 5 years old then treat medical treatment for 2 years. Clinical examination revealed normal with normal level of consciousness . There was mild degree dystonia, dysarthria. However, no evidence of any spasticity was noted. Rest of the neurological and physical examination was unremarkable.

Laboratory examination including Hemoglobin, MCV, MCH, MCHC, Leukocyte, Potassium , Chloride , Basophils, Segment Neutrophils, Monocyte and Lymphocyte were within normal limits while Platelets, Eosinophils and Sodium results low. Ig G and Ig M were negative . CT –Scan Non Contrast Brain revealed calcification multiple in basalis ganglia, Thalamus and cortical frontoparietal bilateral and hematoma extracranial region parietal sinistra.

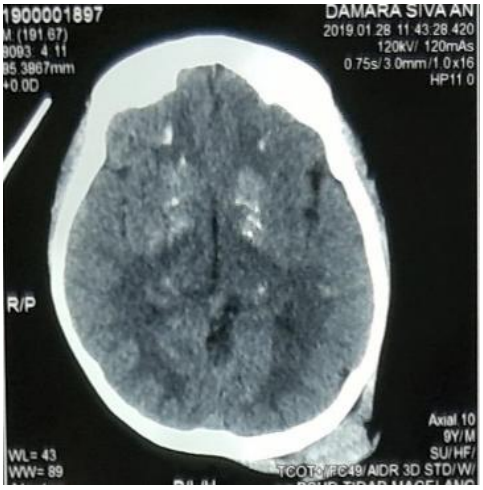

Figure 1 Calcification multiple in Basalis Ganglia, Thalamus and Hematom Extracranial region Parietal Sinstra

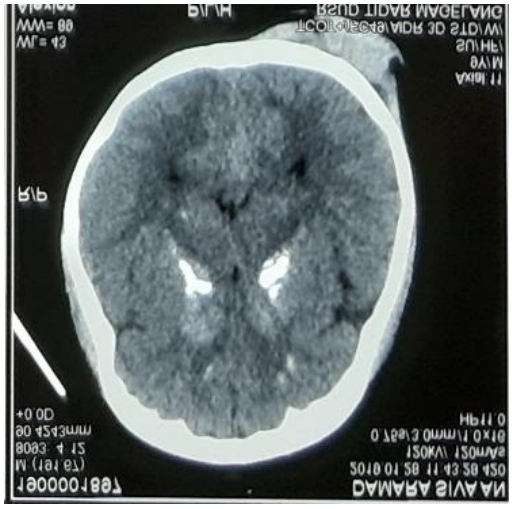

Figure 2 Calcification Multiple in Thalamus Bilateral and Cortical Frontal Bilateral

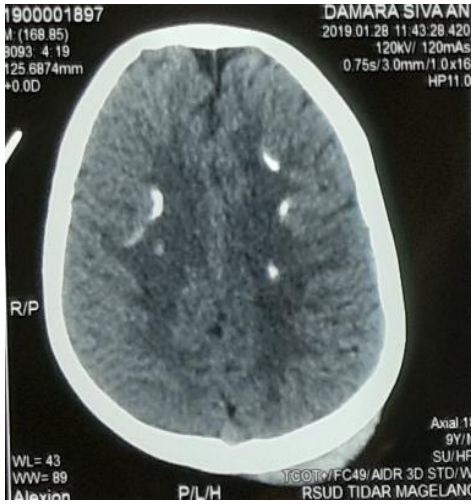

Figure 3 Calcification Multiple in Cortical Frontoparietal Bilateral

DISCUSSION

Fahr's syndrome is a disorder characterized by bilateral calcification of basal ganglia and the dentate nuclei. This intracranial calcification is usually bilateral and symmetrical, also called Bilateral Striopallido Dentate Calcinosis (BSPDC) or Idiopathic Basal ganglia

calcification (Wazir et al., 2023). The signs and symptoms of Fahr Disease include neurological manifestation (such as seizures and tetany), motor manifestation (includes lethargy, uncontrolled choreatic movement, and muscular cramps), and neuropsychiatric disease (eg, early dementia, cognitive impairment, behavioral and personality changes). This disease is most often associated with hypoparathyroidism. Other conditions include neuroferritinopathy, Kenny-Caffey syndrome type 1, intrauterine or perinatal infection (e.g., toxoplasma gondii, rubella), tuberous sclerosis complex, brucella infection have also been associated with Fahr's syndrome (Wazir et al., 2023). A study describe a 51-year-old woman with extensive bilateral brain calcifications in the basal ganglia, subcortical white matter, and cerebellum, who developed subacute cognitive and behavioral decline along with parkinsonism, despite showing normal calcium and parathyroid hormone levels (Al Ali et al., 2023b).

Fahr's disease, like Fahr's syndrome, has similiar clinical manifestations; however, it is a rare hereditary disease with an autosomal dominant inheritance pattern, presenting usually seen in the fourth to sixth decade of life along with no association with trauma, infection, chronic hypocalcaemia, e.g., vitamin D deficiency and renal failure, are associated with intracranial calcifications, metabolic and mitochondrial disease or any systemic disease (Peters et al., 2020; Wazir et al., 2023). Recent investigations reveal that patients with primary familial brain calcification exhibit diverse clinical profiles, including non-motor symptoms, symptom variability linked to intracranial calcification burden, and novel genetic and phenotypic findings across different populations (Balck et al., 2021; Bonato et al., 2024; Mathijssen et al., 2024; Sennfält et al., 2024). The clinical phenotype has been reviewed in a series of 516 genetically confirmed Fahr Disease patients, of which 67.6% were clinically affected. It suggests that the pathogenic

mechanisms involve dysfunction in the neurovascular unit, phosphate homeostasis imbalance, and mitochondrial impairment (Chen et al., 2023).

Mutation in specific genes such SLC20A2, PDGFRB, PDGFB, XPR1, MYORG, JAM2, CMPK2 and myogenic regulating glycosylase genes were observed in Fahr's disease (Magalhães et al., 2024; Tadic et al., 2015; Wazir et al., 2023; Xu et al., 2023). A study reporting four cases of brain calcifications from a tertiary care center, including one case of primary familial brain calcification with an SLC20A2 mutation and two cases associated with endocrine disorders. CT and MRI revealed calcifications in the basal ganglia and dentate nucleus, with one patient developing an acute ischemic stroke likely related to the calcifications (Srichawla, Andrade, & Kipkorir, 2023). Mutations in different Fahr Disease genes can be associated with specific radiological features. Biallelic mutations in MYORG and JAM2 have been associated with a more severe and widespread pattern of calcium deposition than dominant genes, with prominent cortical and cerebellar involvement (T. Zhao et al., 2023). A study report a patient presenting with asymmetric tremor, early-onset dementia, and extensive brain calcifications in whom genetic testing revealed a heterozygous splice-site mutation in SLC20A2 and a missense mutation in PSEN1, the latter associated with early-onset Alzheimer's disease (EOAD) (Hebestreit et al., 2023). Central pontine calcification is highly suggestive of MYORG mutations, which also cause significant levels of cerebellar atrophy and appear to produce a more severe phenotype (Chelban et al., 2020; Grangeon et al., 2019). Seizures, cerebellar and pyramidal signs are also frequently described (Yang, Li, Jiao, & Weng, 2022; Zhang et al., 2024). Dysarthria (even isolated) is almost always present in symptomatic MYORG mutation carriers who tend to show a phenotype dominated by progressive cerebellar signs with ataxia and cognitive decline. MYORG patients may also

show a phenotype similar to progressive supranuclear palsy (PSP) with vertical gaze palsy, progressive cognitive decline and early falls in the context of rigid-akinetic parkinsonism (Chelban et al., 2020; Grangeon et al., 2019; Taglia et al., 2019). JAM2 mutation carriers exhibit a higher TCS as compared to MYORG patients, with severe and confluent cortical calcification involving the occipital, temporal, frontal and parietal cortices bilaterally (Cen et al., 2020). Several heterozygous CMPK2 carriers from two recently published Chinese families showed small, punctate calcifications in the GPi on brain CT scans. Radiological findings have also been reported in two clinically symptomatic carriers of a single JAM2 mutation although genetic analysis failed to identify a second pathogenic variant (Khojasteh et al., 2024; M. Zhao et al., 2022). The prevalence of Fahr syndrome is <1/1,000,000, making it a rare neurodegenerative disorder (Wazir et al., 2023). Recent studies have highlighted the genetic heterogeneity of primary familial brain calcification (PFBC), with several reports identifying novel variants in PDGFB and PDGFRB linked to distinct clinical manifestations. Case reports describe patients carrying frameshift and splice-site mutations in SLC20A2 and PDGFB, as well as truncating PDGFB variants associated with small vessel disease, underscoring the complex vascular involvement in PFBC (Al Ali et al., 2023a; Keogh et al., 2015; Shen et al., 2021; Yektay Farahmand et al., 2024).

The diagnostic workup of Fahr Disease begins with recognizing the clinical manifestations and identifying bilateral basal ganglia calcification on CT/MRI brain (Wazir et al., 2023). Investigation for parathyroid disorders (for primary and secondary hypoparathyroidism) and abnormalities of calcium metabolism should be performed to rule them out, as basal ganglia calcification associated with them is a common entity in adults (Wazir et al., 2023). Further investigations should be performed to rule out other

conditions such as hypoparathyroidism, pseudohypoparathyroidism, mitochondrial myopathy, brucellosis, neurocysticercosis, toxoplasmosis, and tuberous sclerosis complex (Wazir et al., 2023). In the case of Fahr's disease, family history will indicate the inheritance pattern, along with molecular genetic testing, should be performed to observe for genetic mutation (Peters et al., 2020). Lastly, physiological basal ganglia calcifications should be distinguished from pathological calcifications. These have been detected in up to 20% of CT scans, with their predilection involvement of globus pallidus, pineal gland, falx, and choroid plexus (Wazir et al., 2023).

Since there is currently no cure or therapy to slow the progression of calcification, treatment focuses on symptom management with, for example, anti-Parkinson's drugs, antidepressants, and antipsychotics. A deeper understanding of the genetics and pathophysiological mechanisms behind the disease may provide new insights into the treatment of patients with Fahr's disease and ectopic (brain) calcification outside of Fahr Disease (Nicolas, Charbonnier, Campion, & Veltman, 2018; Peters et al., 2020). The first potential treatment for patients with Fahr disease was bisphosphonate therapy. Newer nitrogen-containing bisphosphonates, such as alendronate, primarily inhibit osteoclasts. Although used for the treatment of osteoporosis, older non-nitrogen-containing bisphosphonates were initially shown to prevent heterotopic mineralization. The bisphosphonate etidronate is a molecular homologue of the circulating calcification inhibitor, inorganic pyrophosphate (PPi). The potential of etidronate for the treatment of vascular calcification has been convincingly demonstrated in patients with calcification disorders, such as common arterial calcification of infancy and pseudoxanthoma elasticum. A second candidate therapy could be vitamin D. A study suggested that the SLC20A2 gene is regulated by vitamin D and vitamin D reduces calcification in the brain.

They found vitamin D deficiency in patients with Fahr's disease. It was suggested that under normal circumstances, vitamin D can bind to the promoter region of the SLC20A2 gene and increase SLC20A2 mRNA. Therefore, this mechanism could be a possible treatment for patients with Fahr's disease (Peters et al., 2020). Third, some of the mutations found in Fahr disease resemble mutations in a specific subgroup of patients with cystic fibrosis who have nonsense mutations that result in a truncated, nonfunctional protein. Treatment with ataluren or PTC124 ensures that the initial stop codon is not read and translation occurs at the normal stop codon. This results in a full-length, functional cystic fibrosis transmembrane conductance regulator product (Peters et al., 2020).

CONCLUSION

In summary CT scan findings in the form of multiple calcifications in the basal ganglia, thalamus and cortical frontoparietal bilateral as well as laboratory results with normal results can indeed strengthen the diagnosis. Further investigation of metabolic disorder and organic etiologies in patients presenting with neuropsychiatric symptoms, family members of patients with Fahr disease is recommended.

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