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Original Research Article

Current perspectives on Farh's disease: a glimpse from the internal medicine unit

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ABSTRACT

Background: Fahr's disease involves abnormal calcium deposits (calcium carbonate/phosphate) in the basal ganglia and other brain regions, primarily due to metabolic dysfunction, with rare autosomal dominant cases. These calcifications affect basal ganglia, thalamus, hippocampus, dentate nucleus, cerebral cortex, and subcortical white matter of the cerebellum. The aim of this study was to determine the prevalence of fahr's disease unrelated to secondary metabolic disorders (2017-2024).
Methods: We conducted a retrospective review of hospital records between 2017 and 2024 to identify patients with a clinical diagnosis of Fahr's disease or those presenting with neurological abnormalities such as seizures, altered mental status, involuntary movements, or intracranial calcifications detected during diagnostic evaluations. Inclusion criteria required cranial CT scans to systematically assess calcifications in predefined brain regions: basal ganglia, thalamus, hippocampus, dentate nucleus, cerebral cortex, or subcortical cerebellar white matter.
Results: Of 134 suspected Fahr's disease cases, only 8 were confirmed on tomography, not clinically with a prevalence of 0.06%. None showed clinical basal ganglia involvement, but CT revealed calcifications: 2 in basal ganglia, thalamus, hippocampus, and cerebellum; 4 in basal ganglia, thalamus, and hippocampus; and 2 in basal ganglia and cerebral cortex.
Conclusions: In our setting, Fahr's disease is generally underdiagnosed and is typically found incidentally through cranial CT scans. Its prevalence could potentially be even higher. Since most of these patients are asymptomatic, comprehensive extension studies are often not conducted for their evaluation. As a result, the current prevalence remains extremely low compared to other studies (around 1%).

Keywords: Basal ganglia, Brain calcifications, Cranial CT-scan, Fahr's disease, Prevalence

INTRODUCTION

Fahr syndrome, an uncommon neurological disorder, is distinguished by the build-up of calcium deposits in specific brain regions, specifically the basal ganglia and dentate nucleus. This condition manifests with a range of neurological symptoms, encompassing movement disorders, psychiatric manifestations, and cognitive decline. Movement disorders, notably tremors, stiffness, and gait difficulties, are the prevailing clinical features associated with Fahr syndrome. Additionally, psychiatric

symptoms, including anxiety and depression, are observed in approximately 40% of individuals affected.[1]

The underlying cause of Fahr syndrome remains incompletely elucidated; however, it is postulated to arise from a complex interplay of genetic and environmental elements. Within the realm of genetic contributions, multiple genes have been implicated in the pathogenesis of Fahr syndrome. Notably, the SLC20A2 gene, responsible for encoding a phosphate transporter, has garnered substantial attention. A novel missense mutation in the SLC20A2 gene was identified within a Pakistani family affected by Fahr syndrome. This novel discovery further bolsters the accumulating body of evidence linking the SLC20A2 gene to the onset and progression of Fahr syndrome.[2]

Fahr syndrome, being a rare condition, poses challenges in establishing a standardized therapeutic strategy. The management of Fahr syndrome primarily revolves around tackling its diverse array of symptoms and associated complications. Notably, addressing movement disorders often entails the utilization of medications such as levodopa or anticholinergics, whereas psychiatric symptoms may necessitate the administration of antidepressants or anxiolytics. A case report of a patient afflicted with Fahr syndrome exhibited notable movement disorders and cognitive decline. Through the implementation of levodopa and clonazepam, a remarkable amelioration of the patient's symptoms was achieved.[3]

This condition, manifests as an aberrant deposition of calcium within the brain, primarily affecting the basal ganglia and other deep-seated cerebral structures. Consequently, a wide spectrum of neurological symptoms emerges, encompassing movement disorders, cognitive decline, and psychiatric manifestations. Although the precise etiology of Fahr syndrome remains a subject of ongoing investigation, certain cases have unveiled the presence of genetic mutations. Diagnostic confirmation often relies on the utilization of advanced imaging modalities, such as computed tomography (CT) or magnetic resonance imaging (MRI) of the brain.[4]

The definitive diagnosis of Fahr syndrome depend predominantly on imaging findings, with CT and MRI emerging as the prevailing modalities for the detection of cerebral calcium depositions. The characteristic imaging hallmark of Fahr syndrome entails bilateral and symmetric calcification within the basal ganglia, notably the globus pallidus, discernible as areas of hyper density on CT and hyperintensity on T1-weighted MRI sequences. Furthermore, the calcification process can extend beyond the confines of the basal ganglia, affecting additional cerebral regions, including the thalamus, dentate nucleus, and cerebral cortex. Recent investigations have posited the potential utility of advanced imaging techniques, such as susceptibility-weighted imaging (SWI) and diffusion tensor imaging (DTI), in enhancing the diagnostic and therapeutic landscape of Fahr syndrome. SWI showcases its prowess by unveiling minuscule calcium deposits that may elude detection via CT or conventional MRI, while DTI facilitates the assessment of white matter tract integrity and the identification of alterations in brain connectivity. These cutting-edge methodologies may offer invaluable insights, particularly in scenarios featuring atypical imaging findings or when scrutinizing disease progression.[5,6]

The Fahr syndrome, presents typically in middle-aged to elderly individuals, around 40 years of age. Its prevalence ranges from 0.1 to 4.2 per 100,000 people, although it may be underestimated due to asymptomatic or undiagnosed cases. The disorder can be either familial or sporadic, with the familial form being more common. Inheritance follows an autosomal dominant pattern with varying penetrance. Notably, a Japanese study observed a higher prevalence of Fahr syndrome among elderly females, who also exhibited a higher prevalence of hypertension, hyperlipidaemia, and diabetes mellitus. In Italy, an increasing incidence of Fahr syndrome has been noted, likely attributable to improved diagnostic techniques and increased awareness. Neurological symptoms, such as movement disorders and cognitive impairment, are prominent, and bilateral basal ganglia calcification serves as a common imaging finding.[7,8]

This disease, is characterized by the presence of bilateral calcifications in the brain and associated neuropsychiatric symptoms. The condition presents with a range of symptoms, including movement disorders, cognitive impairment, psychiatric manifestations, and seizures. These neurological deficits tend to worsen over time and can have a significant impact on daily functioning. Onset of symptoms typically occurs between the ages of 30 and 50, and the syndrome is more prevalent in women than men. A recent study conducted in 2021 identified movement disorders, cognitive impairment, and psychiatric symptoms as the most common clinical features of Fahr syndrome. Furthermore, the study found that calcifications primarily affected the basal ganglia, thalamus, and cerebellum in a bilateral and symmetric manner. Another study from 2018 highlighted parkinsonism as the most frequent movement disorder observed in Fahr syndrome, followed by chorea and dystonia. The presence of seizures, particularly in patients with temporal lobe calcifications, was also noted.[9,10]

Fahr syndrome, a rare neurodegenerative condition, is characterized by the accumulation of calcium in the basal ganglia and cerebral cortex. While no specific laboratory tests exist for diagnosing Fahr syndrome, routine blood tests are typically normal and can help rule out other possible causes. Genetic testing can be performed to identify underlying genetic factors, including mutations in genes like SLC20A2, PDGFRB, and PDGFB. It can confirm the diagnosis and provide valuable information for family members, although it may not be necessary for all Fahr syndrome patients.[11,12]

In patients with Fahr syndrome, blood tests can be helpful in assessing calcium, phosphorus, parathyroid hormone, thyroid hormone, and other metabolic markers. These tests can reveal any abnormalities associated with the disorder. Genetic testing is valuable in identifying specific mutations, like SLC20A2 and PDGFRB, that are linked to Fahr syndrome. Imaging techniques such as CT-scans and MRIs are crucial for visualizing the extent of calcification in the basal ganglia and other brain areas, which is a key characteristic of the syndrome. Although there is no targeted treatment for Fahr syndrome, managing underlying conditions that contribute to its development, like hypoparathyroidism or Wilson's disease, is essential. Symptomatic treatment, including medication and therapies, can help alleviate seizures and movement disorders. Regular monitoring of metabolic and hormone levels is important for addressing potential complications associated with the syndrome.[13,14]

Diagnosis relies on clinical evaluation, neuroimaging, and laboratory tests. The symptoms of Fahr syndrome can vary and may include cognitive decline, movement disorders, seizures, and psychiatric symptoms. Neuroimaging techniques like CT-scan and MRI are essential for confirming the diagnosis, showing bilateral symmetric calcifications in specific brain regions. Laboratory tests help rule out other conditions related to calcifications by assessing calcium, phosphorus, and parathyroid hormone levels. Genetic testing is also valuable in identifying mutations in genes like SLC20A2, PDGFRB, XPR1, MYORG, and PDE1C, which are associated with Fahr syndrome. Genetic testing can provide insights into the underlying cause of calcifications and contribute to a more precise diagnosis.[15,16]

The differential diagnosis of Fahr syndrome poses a challenge due to its nonspecific symptoms and overlapping radiological features with other neurological disorders. To ensure accurate diagnosis, it is crucial to exclude conditions like infections, metabolic disorders, and tumors that can also cause basal ganglia calcifications. Blood tests, cerebrospinal fluid analysis, and imaging studies play a vital role in distinguishing Fahr syndrome from other potential causes. Two conditions that require differentiation from Fahr syndrome are hypoparathyroidism and primary familial brain calcification (PFBC). While both can present with basal ganglia calcifications and similar neurological symptoms, PFBC is a rare genetic disorder limited to brain calcifications, whereas hypoparathyroidism involves low parathyroid hormone levels and can lead to calcifications in various body parts.

To accurately diagnose Fahr syndrome, a comprehensive evaluation of clinical symptoms, imaging findings, and laboratory results is necessary to differentiate it from similar radiological conditions. Identifying the underlying cause of basal ganglia calcifications is essential for guiding appropriate treatment and management.[17,18]

Treatment for Fahr syndrome is primarily aimed at symptom control and addressing underlying conditions. While there is no specific cure, various approaches can help improve the quality of life for affected individuals. Seizures can be managed with antiepileptic medications, while psychiatric symptoms may benefit from psychotherapy and medications like antidepressants or antipsychotics. Rehabilitation therapies, such as physical, occupational, and speech therapies, can also play a valuable role in enhancing daily functioning. It's important to note that prognosis can be challenging to predict due to the variable nature of the syndrome, and treatment focuses on symptom management rather than complete resolution of the condition.[19,20]

There is a notable link between Fahr syndrome and hypoparathyroidism, with a prevalence rate of 29% observed in affected individuals. This finding underscores the importance of evaluating patients with Fahr syndrome for hypoparathyroidism. Early detection and treatment of hypoparathyroidism can potentially delay or prevent the onset of neurological symptoms associated with Fahr syndrome.[21]

There is a connection between Fahr syndrome and Parkinson's disease because of a higher occurrence of Fahr syndrome in individuals with Parkinson's disease compared to the general population. It is recommended an evaluation of patients especially those experiencing cognitive and psychiatric symptoms, for Fahr syndrome to ensure prompt diagnosis and appropriate care.[22] The objective of this study is determining the prevalence of fahr's disease, excluding secondary metabolic disorders.

METHODS

This retrospective study analyzed medical records of patients with suspected Fahr’s disease, the study was carried out at our institution, a tertiary referral center, and data were collected from the hospital registry of patients admitted between 2017 and 2024. Ethical approval was obtained from the Institutional Review Board. The inclusion criteria for patient selection were: patients with clinical suspicion of Fahr's disease based on symptoms such as movement disorders, cognitive impairment, and psychiatric manifestations, and patients with a history of metabolic disorders associated with basal ganglia calcifications. Patients with incomplete medical records or inadequate imaging data were excluded from the study. We conducted a thorough review of electronic medical records to identify eligible patients. Medical history, clinical presentation, and laboratory results were carefully examined to confirm the diagnosis of Fahr's disease.

From an initial cohort of 134 cases, only eight met diagnostic criteria for Fahr’s disease, all female, aged 34 to 62 years. We intentionally searched for cranial CT scans of the included patients. Two experienced radiologists independently analysed the CT images using a standardized protocol. The specific brain regions assessed

for calcifications included the basal ganglia (including the globus pallidus, putamen, and caudate nucleus), thalamus, hippocampus, dentate nucleus, cerebral cortex, and subcortical white matter of the cerebellum. The presence, location, and extent of calcifications were recorded and compared between the two radiologists to assess interobserver agreement. Descriptive statistics were used to summarize the demographic and clinical characteristics of the study population. The prevalence of Fahr's disease was calculated by dividing the number of confirmed cases by the total number of patients with clinical suspicion. The distribution of calcifications in different brain regions was analysed using percentages and presented in a tabular format. Interobserver agreement for the presence of calcifications was evaluated using Cohen's kappa coefficient.

RESULTS

A total of 134 patients suspected of having Fahr's disease were identified during the study period (Table 1). Among them, only eight cases were confirmed as Fahr's disease, all of which were female patients aged between 34 and 62 years. We intentionally sought to identify neuropsychiatric disturbances (including personality changes, disorientation, dementia, and psychosis), motor symptoms (such as spasticity, athetosis, and dystonia), epilepsy, and cognitive impairments, but none of these patients exhibited the typical clinical symptoms associated with basal ganglia involvement. However, upon analysing cranial CT scans, distinct patterns of involvement were observed in the identified cases, manifestations that result from the abnormal and symmetrical calcium deposits characteristic of this condition. Two patients showed calcifications in the basal ganglia, thalamus, hippocampus, and subcortical white matter of the cerebellum. Four patients presented calcifications in the basal ganglia, thalamus, and hippocampus. Lastly, two patients displayed calcifications in the basal ganglia and cerebral cortex. These findings provided valuable insights into the distribution of calcifications in brain regions associated with Fahr's disease (Figure 1).

Table 1: Demographic characteristics of population studied.

Characteristic	N (%)
Sex	
Male	48 (35.8)
Female	86 (64.2)
Comorbidities	
Type 2 diabetes	42 (31.3)
Hypertension	36 (26.9)
Dyslipidemia	15 (11.2)
Ischemic heart disease	12 (9.0)
Obstructive sleep apnea	9 (6.7)
Chronic obstructive pulmonary disease	7 (5.2)
Cardiomyopathy	4 (3.0)
Systemic lupus erythematosus	2 (1.5)

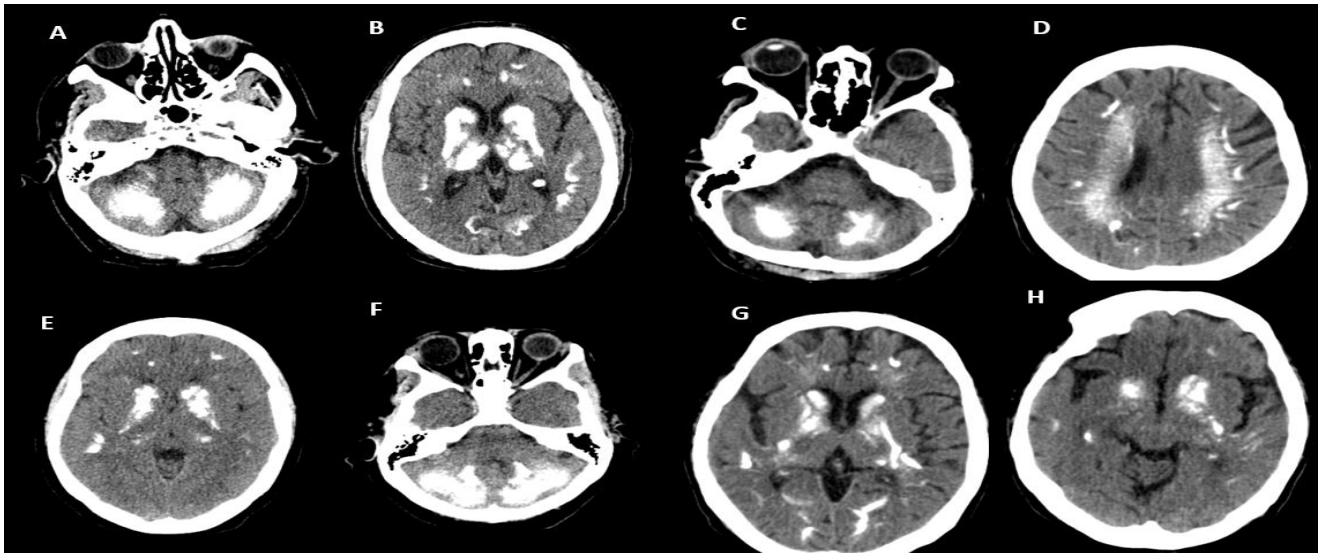

Figure 1: Brain CT scans demonstrating calcification patterns in Fahr's disease (A, C, F) axial CT images showing calcifications in the cerebellar lobes; (B, E, G, H) axial CT images revealing bilateral thalamic calcifications; (D) axial CT image depicting disseminated calcifications predominantly in the right cerebral lobes.

The prevalence of Fahr's disease in our study population was 0.06%. It is noteworthy that these diagnoses were incidental findings on computed tomography scans, as none of the patients exhibited symptoms indicative of Fahr's disease at the time of diagnosis. This highlights the importance of thorough imaging examinations to identify asymptomatic cases and underscores the need for greater awareness and vigilance when interpreting CT scans. The incidental nature of the diagnoses raises intriguing questions about the potential clinical implications and long-term prognosis of individuals with asymptomatic Fahr's disease. Further research is required to determine the significance of these incidental findings and their impact on the health and overall well-being of affected individuals.

It is essential to acknowledge the limitations of our study, including its retrospective nature and the relatively small number of confirmed Fahr's disease cases. Additionally, the absence of clinical symptoms in the diagnosed patients calls for caution when generalizing our findings to a broader population.

DISCUSSION

Fahr’s disease, or PFBC, is a rare neurological disorder characterized by bilateral calcifications in the basal ganglia and other brain regions, often presenting with heterogeneous clinical manifestations. Our study identified a prevalence of 0.06% among 134 suspected cases, all of which were incidentally detected via cranial CT scans without overt clinical symptoms at the time of diagnosis. This finding contrasts with prior reports suggesting a prevalence closer to 1%, raising critical questions about underdiagnosis and the natural history of this condition.[23]

In our population, Fahr's disease is a condition that is often underdiagnosed and frequently discovered incidentally through cranial CT scans. The prevalence of Fahr's disease in our study reflects the limited diagnostic process carried out in asymptomatic patients, resulting in a lower prevalence compared to other studies where more exhaustive evaluations were conducted. The incidental detection of Fahr's disease through cranial CT scans highlights the potential for an even higher prevalence if these imaging studies were routinely performed in patients with metabolic disorders or relevant clinical histories. The importance of incidental findings in the diagnosis of Fahr's disease cannot be underestimated, as it emphasizes the need for greater vigilance among healthcare professionals and the implementation of standardized diagnostic protocols.

Low prevalence in our cohort compared to other studies reflects several factors. First, selection bias may play a role, as our cohort was derived from a hospital registry of symptomatic or high-risk patients, whereas population-based studies might capture broader demographics. Second, differences in diagnostic criteria could explain the discrepancy, as some studies include secondary calcifications due to metabolic disorders like hypoparathyroidism, while we focused on idiopathic cases. Third, variations in imaging sensitivity must be considered; while CT scans remain the gold standard for calcification detection, early or minimal deposits might only be visible on susceptibility-weighted MRI. Notably, all confirmed cases in our study were asymptomatic, reinforcing the hypothesis that Fahr’s disease has a prolonged subclinical phase, as previously reported in genetic studies where 20–30% of mutation carriers lacked symptoms.[24] This asymptomatic presentation poses significant challenges for timely diagnosis and underscores the importance of neuroimaging in identifying subclinical cases.

The anatomic distribution of calcifications in our cohort aligns with established patterns in the literature. All cases showed basal ganglia involvement, while 75% exhibited thalamic calcifications and 50% had hippocampal deposits. Two cases also demonstrated cerebellar white matter involvement; a pattern associated with autosomal dominant mutations in SLC20A2.[25] The absence of clinical correlates despite these radiologic findings suggests potential neuroplasticity mechanisms that compensate for calcification-related damage until a critical threshold is reached. Alternatively, genetic heterogeneity may contribute to phenotypic variability, as mutations in PDGFB or MYORG have been linked to milder presentations. Advanced imaging techniques like SWI and DTI could provide further insights into subclinical damage, as these modalities have detected microstructural white matter changes in asymptomatic carriers.[26] The discordance between radiologic severity and clinical presentation remains one of the most intriguing aspects of Fahr’s disease and warrants further investigation through longitudinal studies combining advanced neuroimaging with detailed clinical assessments.

The diagnostic challenges highlighted by our study are multifaceted. Current diagnostic frameworks require both radiologic evidence of bilateral calcifications in specific brain regions and exclusion of secondary causes such as metabolic disorders, infections, or toxins. However, our findings demonstrate several limitations of this approach. First, asymptomatic cases are likely to be missed without routine neuroimaging, leading to underestimation of disease prevalence. Second, the overlap with age-related calcifications, particularly in the pineal gland, can create diagnostic confusion. Third, the clinical relevance of incidentally detected calcifications remains uncertain, making it difficult to counsel patients about prognosis. Genetic testing for mutations in SLC20A2, PDGFRB, and other associated genes could help resolve some of these diagnostic uncertainties, but cost and accessibility currently limit its widespread use in clinical practice. Furthermore, the absence of clear genotype-phenotype correlations complicates the interpretation of genetic results, as the same mutation can lead to vastly different clinical presentations even within the same family.[27] These

challenges underscore the need for standardized diagnostic protocols that incorporate both imaging and genetic criteria, as well as clinical follow-up to monitor for symptom development in incidentally identified cases.

The clinical implications of our findings are significant for both patient management and future research directions. For clinical practice, our results suggest that screening with cranial CT or MRI should be considered in several scenarios. Patients presenting with movement disorders, psychiatric symptoms, or cognitive decline of unclear etiology warrant neuroimaging evaluation for possible Fahr's disease. Those with a family history of neurological disorders or brain calcifications should also be assessed, given the autosomal dominant inheritance pattern seen in many cases. Additionally, a thorough metabolic workup including calcium, phosphorus, parathyroid hormone, and thyroid function tests is essential to rule out secondary causes of basal ganglia calcification. For asymptomatic patients with incidentally detected calcifications, management decisions are more complex. While some experts recommend regular neurological follow-up to monitor for symptom development, others argue that the unpredictable natural history makes such monitoring of uncertain value.[28] In symptomatic patients, treatment remains supportive and symptom-specific. Levodopa may provide benefit for parkinsonian features, while antiepileptics are used for seizure control and psychotropic medications for psychiatric manifestations. However, response to these therapies is often partial and unpredictable, highlighting the need for more targeted treatment approaches.

From a research perspective, our study identifies several important knowledge gaps that future investigations should address. Prospective longitudinal studies are needed to better define the natural history of Fahr's disease, particularly in asymptomatic individuals with radiologic evidence of calcification. Such studies could help determine the rate of symptom development, identify predictors of clinical progression, and establish evidence-based guidelines for monitoring incidentally detected cases. Additionally, research into potential biomarkers of disease activity or progression could significantly improve clinical management. Cerebrospinal fluid analysis, advanced neuroimaging parameters, or blood-based markers might help stratify risk and predict clinical course. Considering this, our study has several limitations that must be acknowledged. The retrospective design introduces potential selection bias and limits our ability to establish causal relationships. The relatively small number of confirmed cases reflects the rarity of the condition but restricts the generalizability of our findings. The lack of genetic confirmation in our cohort is another limitation, as molecular characterization could have provided additional insights into phenotype-genotype correlations. Furthermore, the absence of long-term follow-up data prevents us from drawing conclusions about the clinical evolution of our asymptomatic cases. Future studies should address these limitations through prospective designs, incorporation of genetic testing, and extended follow-up periods.

CONCLUSION

Fahr's disease, a rare neurological disorder characterized by abnormal intracranial calcifications, remains underdiagnosed in clinical practice, as evidenced by our retrospective study. Among 134 patients with suspected Fahr's disease, only 8 cases (0.06%) were confirmed, all of which were incidental findings on cranial CT scans, with no overt clinical symptoms at the time of diagnosis. The calcifications predominantly involved the basal ganglia, thalamus, hippocampus, and cerebellar white matter, aligning with established neuroanatomical patterns of the disease.

The strikingly low prevalence in our cohort, compared to rates of approximately 1% reported in other studies, underscores the challenges in diagnosing Fahr's disease, particularly in asymptomatic individuals. This discrepancy highlights the critical role of neuroimaging in identifying subclinical cases and suggests that the true prevalence may be higher if systematic screening were implemented. Furthermore, the absence of symptoms in our diagnosed cases raises important questions about the natural history of Fahr's disease and the potential for delayed neurological manifestations.

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