IJPHS:
International Journal of Psychology and Health Science
GREENATION PUBLISHER
+62 812 1046 7572 http://greenpub.org greenation.info@gmail.com
DOI: https://doi.org/10.38035/ijphs.v4i3

Late-Onset Fahr Disease Presenting with Chronic Vertigo, Right-Sided Postural Instability, and Mild Cognitive Impairment: A Case Report

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Abstract: Fahr disease, currently recognized within the spectrum of primary familial brain calcification, is a rare neurodegenerative disorder characterized by bilateral intracranial calcifications, most commonly involving the basal ganglia, dentate nuclei, thalamus, internal capsule, and cerebral white matter. It usually presents with movement disorders, neuropsychiatric manifestations, seizures, gait disturbance, or cognitive decline. Chronic vestibular symptoms as the initial manifestation are uncommon and may delay diagnosis, especially in older patients with coexisting vascular brain changes. This report describes a late-onset case of Fahr disease presenting primarily with chronic vertigo, right-sided postural instability, and mild cognitive impairment. Case presentation: A 67-year-old woman presented with chronic spinning vertigo for one year, worsening over the last five months. The symptoms were prominent during positional change from lying to sitting and caused unsteady gait with a sensation of nearly falling. She also reported right-sided pulsatile tinnitus and hearing impairment. Neurological examination showed unidirectional nystagmus with a fast component to the left and right vestibulocochlear nerve dysfunction. Balance assessment revealed consistent right-sided deviation, including positive Romberg test, sharpened Romberg test, Fukuda stepping test, and tandem gait instability. Motor, sensory, autonomic, and limb coordination examinations were within normal limits. Cognitive screening using MoCA-INA showed mild cognitive impairment with a score of 22/30. Laboratory examination showed preserved renal function and normal glucose control, but hypercholesterolemia and elevated LDL cholesterol were present. Brain MRI showed mild periventricular leukoaraiosis, lacunar infarcts in the right cerebellum and left insular region, senile cerebral atrophy, and pathological calcification in the cerebellum and bilateral internal capsule regions, suggestive of bilateral striatopallidodentate calcinosis or Fahr disease. This case highlights chronic vestibular symptoms and right-sided postural instability as an unusual

initial presentation of late-onset Fahr disease. Mild cognitive impairment and vascular brain changes may coexist and contribute to clinical severity. Fahr disease should be considered in older patients with persistent vertigo, gait instability, cognitive decline, and bilateral intracranial calcification. Further evaluation of calcium-phosphate metabolism, parathyroid hormone, vitamin D, magnesium, family history, and genetic testing is needed to distinguish primary Fahr disease from secondary Fahr syndrome and to guide long-term management.

Keyword: Case Report, Fahr Disease, Primary Familial Brain Calcification, Chronic Vertigo, Basal Ganglia Calcification, Cognitive Impairment, Leukoaraiosis.

INTRODUCTION

Primary familial brain calcification (PFBC), historically known as Fahr disease, is a rare neurodegenerative disorder characterized by bilateral and usually symmetrical intracranial calcifications. These calcifications most often involve the basal ganglia, but they may also affect the dentate nuclei, thalamus, internal capsule, cerebral white matter, and other cortical or subcortical regions (Carecchio et al., 2023; DURANTE et al., 2021; Snijders et al., 2024). The term PFBC is now commonly used for genetically determined forms, while Fahr syndrome is often applied when a secondary cause can be identified, such as calcium-phosphate metabolism disorders, endocrine disease, infection, mitochondrial disease, toxic exposure, or other acquired conditions (Carecchio et al., 2023; Snijders et al., 2024).

The clinical spectrum of Fahr disease is broad. Patients may remain asymptomatic despite extensive calcification, while others present with movement disorders, cognitive decline, psychiatric manifestations, seizures, cerebellar signs, gait disturbance, and other neurological deficits (Carecchio et al., 2023; Lamessa et al., 2024; Sun et al., 2024). Recent genetic studies have identified several PFBC-associated genes, including SLC20A2, PDGFB, PDGFRB, XPR1, MYORG, JAM2, and CMPK2, which are involved in phosphate transport, neurovascular unit integrity, blood-brain barrier function, and abnormal mineral deposition in cerebral microvessels (Carecchio et al., 2023; Shen et al., 2021; Sun et al., 2024).

Motor manifestations are among the most recognized features of PFBC. Parkinsonism, tremor, dystonia, chorea, ataxia, dysarthria, and gait disturbance have been reported in different clinical settings (Carecchio et al., 2023; Sun et al., 2024). Balance impairment may result from combined basal ganglia and cerebellar involvement. A report described a patient with Fahr disease who showed mixed parkinsonian and cerebellar signs, with marked impairment in vestibular control during static balance assessment. This finding is clinically relevant because patients may complain of dizziness, instability, or falls before more typical extrapyramidal signs become obvious (Scarano et al., 2022).

Non-motor manifestations also have important diagnostic value. Cognitive impairment, psychiatric symptoms, sleep disturbance, autonomic symptoms, headache, and behavioral changes may occur in PFBC and may precede overt movement disorders (Bonato et al., 2024; Carecchio et al., 2023; M. Li et al., 2022; W. C. Li et al., 2024). A report stated that cognitive involvement and other non-motor symptoms are frequent in PFBC and should be assessed systematically (Bonato et al., 2024). Psychiatric presentations may mimic primary psychiatric disorders, as shown in cases with schizophrenia-like symptoms or long-term psychosis associated with Fahr disease (De Pieri et al., 2023; W. C. Li et al., 2024). These findings support the need for cognitive and neuropsychiatric screening in patients with unexplained intracranial calcifications.

Neuroimaging plays a central role in diagnosis. Brain CT remains the preferred modality for detecting calcification, but MRI can show abnormalities on susceptibility-sensitive sequences and may also identify associated white matter disease, lacunar infarcts,

cerebral atrophy, or other structural lesions (Carecchio et al., 2023; DURANTE et al., 2021). The diagnostic work-up should also exclude secondary causes. There was a report in 2024 that emphasized that infectious causes of basal ganglia calcification in adults should be considered based on clinical indication rather than tested routinely in all patients. Evaluation of calcium, phosphate, parathyroid hormone, vitamin D, renal function, infectious history, toxic exposure, and family history remains important to distinguish Fahr disease from Fahr syndrome (Snijders et al., 2024).

This case is notable because the patient presented primarily with chronic spinning vertigo, unilateral vestibular symptoms, right-sided postural instability, and mild cognitive impairment, rather than a typical movement disorder or seizure. MRI showed features consistent with bilateral striatopallidodentate calcinosis, mild periventricular leukoaraiosis, lacunar infarcts, and senile cerebral atrophy. This presentation highlights the diagnostic challenge of late-onset Fahr disease when vestibular complaints and vascular brain changes coexist. It also supports the need to consider Fahr disease in older patients with chronic vertigo, imbalance, cognitive decline, and bilateral intracranial calcification.

METHOD

A 67-year-old woman presented with chronic spinning vertigo that had started one year before admission and had worsened during the last five months. She described a sensation that the room was spinning around her. The symptom was most prominent when she changed position from lying down to sitting. The vertigo caused gait instability, unsteadiness, and a sensation of nearly falling while walking. She denied nausea, vomiting, headache, perioral paresthesia, facial deviation, slurred speech, dysphagia, hemiparesis, hemisensory loss, and bowel or urinary disturbance. She also reported right-sided pulsatile tinnitus and right-sided hearing impairment. She had previously consulted an otorhinolaryngologist, and no major otorhinolaryngological abnormality was reported.

The patient had also experienced progressive forgetfulness for the last six years. She often misplaced objects but could usually remember their location after some time. Her basic daily activities remained preserved. She could eat, drink, bathe, and perform self-care independently. Her medical history was notable for hypertension. However, she did not take antihypertensive medication regularly and only used it when she felt discomfort in the nape area. No family history of similar neurological or psychiatric illness was reported.

On physical examination, the patient was alert, with a Glasgow Coma Scale score of 15. Her blood pressure was 150/80 mmHg, pulse rate 70 beats/minute, respiratory rate 20 breaths/minute, and body temperature 36°C. Pupils were isochoric, 3 mm in diameter bilaterally, with normal light reflexes. Neurological examination showed unidirectional nystagmus with the fast component to the left. Cranial nerve examination showed right vestibulocochlear nerve dysfunction with right-sided tinnitus. Motor, sensory, and autonomic examinations were within normal limits.

Balance testing showed a consistent right-sided postural tendency. The Romberg test with eyes closed was positive, with falling to the right. The sharpened Romberg test also showed falling to the right. The Fukuda stepping test demonstrated deviation to the right. Tandem gait testing showed instability and falling to the right. Limb coordination testing was within normal limits.

Cognitive screening using the Indonesian version of the Montreal Cognitive Assessment showed cognitive impairment, with a total score of 21/30 and an adjusted score of 22/30 after adding one point for education below 12 years. The impaired domains included visuospatial/executive function, attention, abstraction, language fluency, and delayed recall. Orientation remained intact.

Laboratory evaluation showed no major hematological abnormality. Hemoglobin was 13.8 g/dL, hematocrit 41.0%, leukocyte count 6.7 × 10^3/µL, and platelet count 285 × 10^3/µL. Liver function tests were within reference limits, with AST 23 U/L, ALT 20 U/L, and gamma-glutamyl transferase 39 U/L. Fasting glucose was 97 mg/dL, HbA1c was 5.3%, creatinine was 0.72 mg/dL, and estimated glomerular filtration rate was 87 mL/min/1.73 m^2. Lipid profile showed hypercholesterolemia, with total cholesterol 219 mg/dL and direct LDL cholesterol 165 mg/dL. HDL cholesterol was 51 mg/dL and triglycerides were 133 mg/dL. Uric acid was 3.7 mg/dL. Urinalysis showed slightly turbid yellow urine, positive leukocyte esterase, 5-10 leukocytes per high-power field, and positive bacteria, while nitrite, albumin, glucose, ketone, bilirubin, and blood were negative.

Brain magnetic resonance imaging without gadolinium contrast was performed using axial, coronal, and sagittal sequences, including T1-weighted, T2-weighted, FLAIR, DWI/ADC, GRE, SWI, and MR angiographic sequences. The MRI showed widened cortical sulci, widened Sylvian and interhemispheric fissures, and features of cerebral atrophy. Mild diffuse periventricular white matter abnormalities appeared hyperintense on T2-weighted and FLAIR sequences, consistent with mild periventricular leukoaraiosis. Small well-defined lesions measuring less than 15 mm were identified in the right cerebellum and left insular region. These lesions were hypointense on T1-weighted and FLAIR sequences, hyperintense on T2-weighted images, showed no blooming artifact on GRE, and did not show restricted diffusion on DWI/ADC. These findings were consistent with lacunar infarcts.

Susceptibility artifacts were observed on SWI in the cerebellum and bilateral internal capsule regions, consistent with pathological calcification. The radiological impression was mild periventricular leukoaraiosis with lacunar infarcts in the right cerebellum and left insular region, pathological calcification in the cerebellum and bilateral internal capsules suggestive of bilateral striatopallidodentate calcinosis or Fahr disease, and senile cerebral atrophy. No calvarial fracture was seen.

Based on the chronic vestibular symptoms, right-sided vestibulocochlear dysfunction, right-sided postural instability, mild cognitive impairment, MRI evidence of bilateral intracranial calcification, and associated lacunar infarcts with leukoaraiosis, the patient was considered to have late-onset Fahr disease presenting with chronic vertigo, balance impairment, and cognitive decline.

RESULT AND DISCUSSION

Clinical features

Fahr disease, currently often discussed under the broader term primary familial brain calcification (PFBC), has a wide clinical spectrum. Patients may present with movement disorders, cerebellar symptoms, psychiatric symptoms, seizures, headache, cognitive decline, or gait disturbance. Some patients remain asymptomatic despite extensive calcification, while others develop progressive neurological impairment. This broad phenotype often delays diagnosis, especially when the first symptoms are nonspecific, such as dizziness, imbalance, or mild cognitive complaints (Adhikari et al., 2023; Aghemo et al., 2023; Iqbal et al., 2022).

The present case showed an atypical but clinically relevant phenotype. The patient presented mainly with chronic spinning vertigo, postural instability, right-sided deviation on balance testing, and mild cognitive impairment. These features are important because Fahr disease is commonly recognized through movement disorders or neuropsychiatric symptoms, but cerebellar and vestibular-like complaints may dominate the early clinical picture. A similar pattern has been reported in patients with mixed motor, cerebellar, and cognitive dysfunction, suggesting that brain calcification can disrupt cortico-subcortical and cerebellar networks involved in posture, gait, executive function, and memory (Almutairi et al., 2025; Slaven et al., 2024).

The patient's MoCA-INA score of 22/30 supports the presence of cognitive impairment. This finding is clinically meaningful because cognitive symptoms are increasingly recognized as part of the PFBC spectrum. Cognitive dysfunction may involve executive function, attention, memory, processing speed, and visuospatial ability. Yoon et al. (2024) described PFBC with XPR1 mutation presenting with cognitive dysfunction, while Almutairi et al. (2025) reported progressive motor and cognitive decline in Fahr disease. These reports support the interpretation that the patient's cognitive impairment may relate to the underlying calcification, small vessel disease, or both (Almutairi et al., 2025; Yoon et al., 2024).

The patient also had hypertension with irregular treatment and hypercholesterolemia, with total cholesterol of 219 mg/dL and LDL cholesterol of 165 mg/dL. These findings are not primary risk factors for Fahr disease, but they are relevant vascular risk factors. They may explain the coexistence of periventricular leukoaraiosis and lacunar infarcts. Therefore, the clinical picture in this case likely reflects an overlap between PFBC-related network dysfunction and vascular brain injury.

Etiology and histopathology

Fahr disease refers to idiopathic or genetic bilateral brain calcification, whereas Fahr syndrome is usually used when a secondary cause is identified. Secondary causes include hypoparathyroidism, pseudohypoparathyroidism, calcium-phosphate metabolism disorders, chronic renal disease, infection, mitochondrial disease, autoimmune disease, toxic exposure, and other acquired conditions. This distinction is clinically important because some secondary causes are treatable (Berrabeh et al., 2023).

Genetic predisposition remains a major etiological factor in PFBC. Several genes have been associated with PFBC, including SLC20A2, PDGFB, PDGFRB, XPR1, MYORG, JAM2, and CMPK2. SLC20A2 is one of the most frequently reported genes and is related to impaired phosphate transport. Reports of SLC20A2-associated PFBC show variable phenotypes, including psychiatric symptoms, familial basal ganglia calcification, recurrent psychosis, parkinsonism, and cognitive involvement (Bu et al., 2022; M. Li et al., 2022). Other genes may affect phosphate homeostasis, neurovascular integrity, and blood-brain barrier function. PDGFRB-related PFBC may present with variable neuropsychiatric or systemic features, while MYORG-related PFBC is commonly associated with autosomal recessive inheritance and broader calcification patterns (Al Ali et al., 2023; Cao et al., 2024; Song et al., 2023; Uno et al., 2020).

Histopathologically, PFBC is characterized by abnormal mineral deposition within cerebral microvessels and perivascular regions. Calcification may involve capillaries, small arteries, arterioles, venules, and adjacent neural tissue. The basal ganglia, thalamus, dentate nuclei, subcortical white matter, and cerebellum are common sites. These deposits are often composed of calcium-phosphate material and may impair local microcirculation, neuronal-glial interaction, and neurovascular coupling (Berrabeh et al., 2023; Bu et al., 2022; M. Li et al., 2022; Song et al., 2023). These mechanisms may explain why patients develop movement disorders, cognitive decline, psychiatric symptoms, and cerebellar or vestibular manifestations.

In this patient, no family history of similar neurological or psychiatric illness was reported. Routine laboratory findings showed preserved renal function, normal fasting glucose, normal HbA1c, and no major liver function abnormality. However, calcium, phosphate, parathyroid hormone, magnesium, and vitamin D levels were not documented in the available data. Therefore, the present case can be described as radiologically suspected Fahr disease or bilateral striatopallidodentate calcinosis, but secondary Fahr syndrome cannot be fully excluded until calcium-phosphate metabolism and parathyroid status are evaluated.

Diagnosis

The diagnosis of Fahr disease requires integration of clinical features, neuroimaging findings, laboratory exclusion of secondary causes, and family or genetic assessment when available. Neuroimaging is central. Computed tomography is generally more sensitive for intracranial calcification, but MRI can demonstrate susceptibility artifacts, associated white matter abnormalities, lacunar infarcts, and brain atrophy. In this case, MRI showed pathological calcification in the cerebellum and bilateral internal capsule regions, mild periventricular leukoaraiosis, lacunar infarcts in the right cerebellum and left insular region, and senile cerebral atrophy. These findings support bilateral striatopallidodentate calcinosis consistent with Fahr disease.

The main diagnostic challenge in this case is the coexistence of calcification and vascular brain changes. Lacunar infarcts and leukoaraiosis may be related to chronic hypertension and dyslipidemia. However, PFBC itself has also been linked with microvascular pathology and small vessel disease. Several reports stated that small vessel disease in PFBC with truncating PDGFB variants, supporting the concept that neurovascular dysfunction may be part of the disease mechanism in some patients. Reports of ischemic stroke or cerebellar infarction in patients with Fahr disease or Fahr syndrome also indicate that vascular events can coexist with or complicate intracranial calcification (Mishra et al., 2025; Slaven et al., 2024).

The laboratory evaluation in this patient is useful but incomplete for etiological classification. Normal creatinine and eGFR make chronic kidney disease less likely as a major contributor. Normal fasting glucose and HbA1c reduce the likelihood of diabetic microangiopathy as the main driver of the MRI findings. However, the lack of serum calcium, phosphate, parathyroid hormone, magnesium, and vitamin D data limits definitive separation between primary Fahr disease and secondary Fahr syndrome. Hypoparathyroidism is an important and treatable cause of Fahr syndrome. Therefore, these tests should be included before concluding that the case is idiopathic or genetic (Berrabeh et al., 2023).

Treatment

There is currently no established disease-modifying treatment for Fahr disease or PFBC. Management is mainly symptomatic and individualized. Treatment should address the dominant clinical problems, such as vertigo, gait instability, cognitive impairment, movement disorder, psychiatric symptoms, seizures, and vascular risk factors. In this case, fall prevention, vestibular rehabilitation, gait training, cognitive monitoring, and control of hypertension and dyslipidemia are clinically relevant.

For patients with parkinsonism, dopaminergic therapy may be considered, although responses vary. For psychiatric symptoms, low-dose antipsychotics have been reported in selected cases, including SLC20A2-associated recurrent psychosis (Uno et al., 2020). However, antipsychotics should be used cautiously in older patients because they may worsen extrapyramidal symptoms or increase fall risk. For seizures, standard antiseizure medication can be used according to the seizure type. If secondary Fahr syndrome is identified, treatment should target the underlying disorder. In hypoparathyroidism-related Fahr syndrome, correction of calcium, phosphate, vitamin D, and parathyroid-related abnormalities may improve symptoms or prevent progression (Berrabeh et al., 2023).

In the present patient, treatment should not focus only on Fahr disease. The MRI findings of lacunar infarcts and leukoaraiosis, combined with hypertension and elevated LDL cholesterol, indicate a vascular prevention priority. Blood pressure control, lipid management, assessment for antiplatelet indication, and lifestyle intervention should be considered according to standard stroke prevention principles. Because the patient has

tinnitus and hearing impairment with right-sided vestibulocochlear dysfunction, audiometry and vestibular testing may also help determine whether the vertigo is purely central, peripheral, or mixed.

Complications and prognosis

The prognosis of Fahr disease is variable. Some patients remain stable for years, while others develop progressive movement disorders, cognitive decline, psychiatric symptoms, seizures, dysarthria, ataxia, or disability. Extensive calcification does not always correlate linearly with symptom severity, which explains the clinical heterogeneity described in recent case reports (Adhikari et al., 2023; Aghemo et al., 2023). Late-onset cases may be misdiagnosed as degenerative dementia, parkinsonism, vascular cognitive impairment, psychiatric disease, or vestibular disease, especially when CT or MRI is not performed early (Iqbal et al., 2022).

This patient has several potential risks. First, chronic imbalance and right-sided postural instability increase the risk of falls. Second, MoCA-INA 22/30 suggests cognitive impairment that may progress over time. Third, lacunar infarcts and leukoaraiosis indicate cerebral small vessel disease, which may contribute to cognitive decline and increase future stroke risk. Fourth, the presence of intracranial calcification raises the possibility of future motor, psychiatric, or seizure manifestations. Reports of progressive motor and cognitive dysfunction, ischemic stroke, cerebellar infarction, and psychiatric presentations in Fahr disease support the need for longitudinal follow-up (Almutairi et al., 2025; Mishra et al., 2025; Slaven et al., 2024; Uno et al., 2020).

Overall, this case highlights three important clinical messages. First, Fahr disease should be considered in older patients with chronic vertigo, gait instability, and cognitive impairment when neuroimaging shows bilateral intracranial calcification. Second, vascular risk factors should be assessed because leukoaraiosis and lacunar infarcts may coexist with PFBC and worsen neurological outcomes. Third, secondary causes must be excluded before labeling a case as idiopathic Fahr disease. In this patient, further evaluation of calcium-phosphate metabolism, parathyroid hormone, vitamin D, magnesium, family history, and possible genetic testing would strengthen diagnostic certainty and guide management.

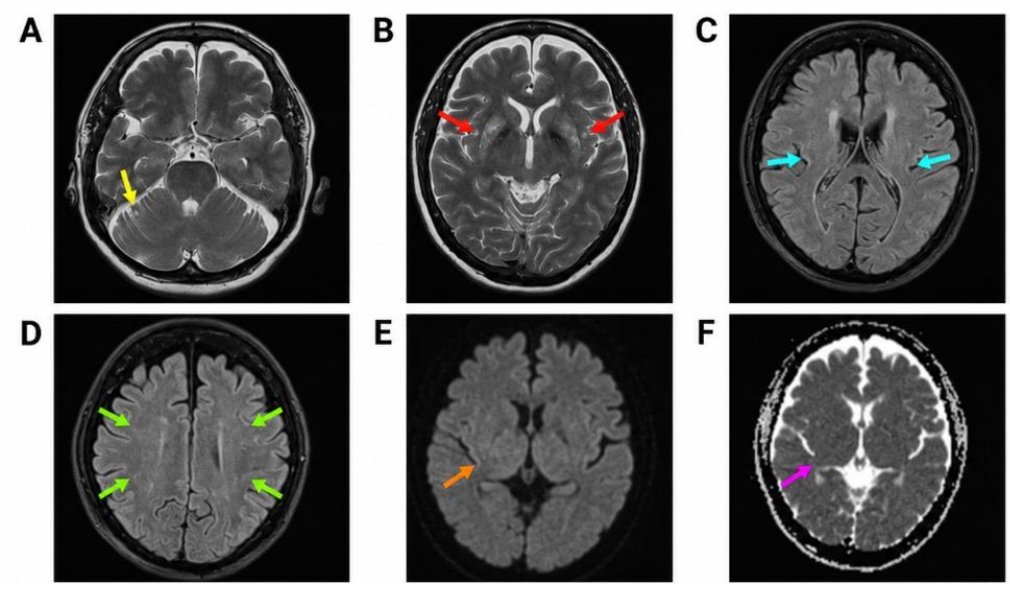

Figure 1. Brain magnetic resonance imaging findings.

Figure 1. Brain magnetic resonance imaging findings. (A) Axial T2-weighted image shows a small lacunar lesion in the right cerebellar region. (B) Axial T2-weighted image shows abnormal signal changes involving the basal ganglia and internal capsule region, corresponding to pathological intracranial calcification. (C) Axial FLAIR image shows mild periventricular white matter hyperintensities consistent with leukoaraiosis. (D) Axial FLAIR image shows additional supratentorial white matter hyperintensities. (E) Diffusion-weighted imaging shows no evidence of acute restricted diffusion. (F) Apparent diffusion coefficient map confirms the absence of restricted diffusion

CONCLUSION

This case highlights chronic vestibular symptoms, right-sided postural instability, and mild cognitive impairment as an unusual initial presentation of late-onset Fahr disease. Brain MRI supported bilateral striatopallidodentate calcinosis with coexisting leukoaraiosis, lacunar infarcts, and cerebral atrophy. The patient's MoCA-INA score suggested cognitive involvement, while hypertension and hypercholesterolemia may have contributed to vascular brain injury. Fahr disease should be considered in older patients with persistent vertigo, gait instability, cognitive decline, and bilateral intracranial calcification. Further evaluation of calcium-phosphate metabolism, parathyroid hormone, vitamin D, magnesium, family history, and genetic testing is important to distinguish primary Fahr disease from secondary Fahr syndrome and to guide long-term management.

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