Fahr's, Falls, and TBI: A Case Report

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Introduction

Fahr's disease (FD), also known as idiopathic basal ganglia calcification, is a neurodegenerative disorder characterized by calcium deposits in the striopallidodentate area. The prevalence is <1/1,000,000, with higher incidence among males and typical onset in the 4th to 6th decades of life [1]. Symptoms include fatigue, dysarthria, dysphagia, migraines, seizures, vertigo, dementia, and psychiatric symptoms, with parkinsonian features occurring later. Diagnosis involves multiple criteria and exclusion of other diseases. Neuroimaging includes CT head without contrast and MRI of the brain, with CT being superior in measuring extent of calcifications. Genetic testing can detect presence of mutations of the SLC20A2 or PDGFRB genes. Blood and urine analysis should be done to assess calcium metabolism and other heavy metals. Currently, there is no cure for FD. Symptom management includes medications for seizures, headaches, and psychiatric symptoms, whereas therapy can address ambulation and activities of daily living [2].

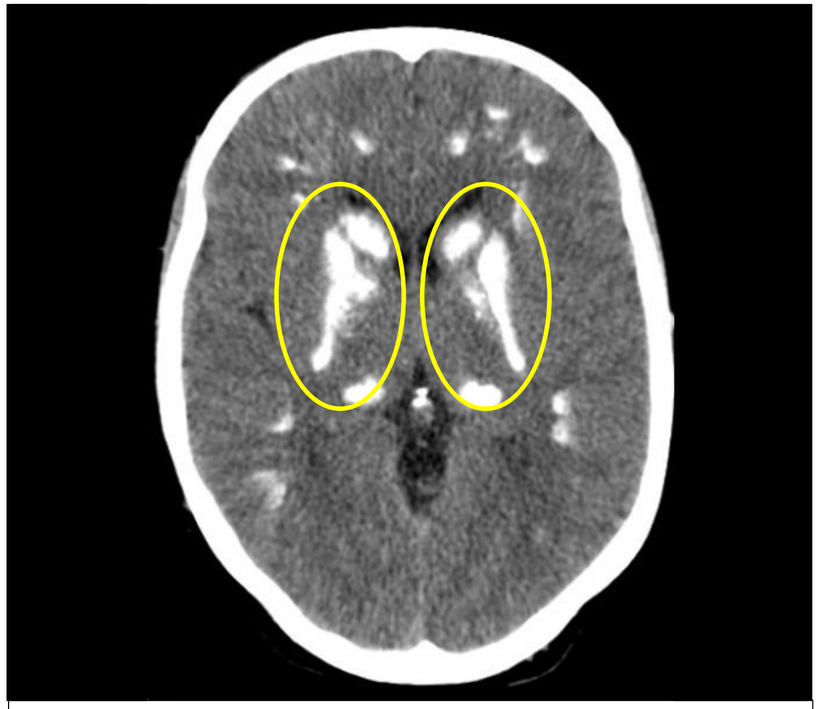

Figure 1: CT head without contrast – Multiple calcifications found within the basal ganglia, supra- & infratentorial white matter regions [3].

Case Description

A 67-year-old man with past medical history of migraines and ADHD was admitted to acute rehabilitation after a fall that caused a subarachnoid hemorrhage. Prior MRI and CT with outpatient neurology revealed incidental calcium deposition in subcortical white matter, basal ganglia, and cerebellum since 2009; however no additional workup had yet been pursued. Over the last year, he developed dysarthria and shuffling gait, while his migraines, attention, and memory all worsened. Due to an increasing number of falls, he started attending physical therapy to improve balance and ambulation. Further follow-up was delayed due to COVID-19 and he sustained the SAH. In rehabilitation, the patient demonstrated mild mobility and ADL impairment, but greater deficiencies in cognition, attention, and memory. Twice, rehabilitation was interrupted due to episodes of confusion and transient global aphasia. The patient was transferred to acute care as there was concern for stroke or seizures, but all workup was negative, and the patient returned to baseline the following day. He would describe these episodes as "debilitating migraines," and stated they had been occurring with mounting frequency. Management of symptoms proved difficult. Carvidopa-levodopa was started for Parkinsonian features and provided minimal benefit. For seizure prophylaxis, he was started on topiramate, which prevented further migraines but worsened his cognition. Upon discharge, only the patient's ambulation was improved, and he still had major deficits with cognition. Ultimately, no alternative diagnosis was found, and the conclusion was FD.

Discussion & Conclusion

Discussion: Our patient was noted with calcifications at age 56. His initial symptom of difficulty concentrating was attributed to ADHD, and his consistent headaches were thought to be simple migraines. Ten years later, progression was noted with increasing falls due to difficulty balancing and shuffling gait, along with evidence of coarse, progressive, bilateral, symmetrical basal ganglia calcification. Diagnosis of FD should be considered if some or all the following symptoms are present: basal ganglia movement disorder, pyramidal signs, cognitive impairment, gait disorder, cerebellar abnormalities, speech dysfunction, psychiatric presentations, and sensory changes [2].

Conclusion: This case demonstrates the necessity for early diagnosis and research of FD. Prompt recognition could lead to better treatment, prevention of complications, and enhanced education. This would improve outcomes and avoid unnecessary diagnostic tests and medication trials. Furthermore, increased knowledge of the disease could prevent unnecessary transfers and rehabilitation interruptions. With improved diagnostic ability and management, these patients could return closer to their baseline functional status.

References

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