CASE REPORT

A Rare Cause of Headache in A Middle-Aged Patient

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ABSTRACT

Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy (CADASIL) is a genetic disease that commonly presents with migraine with aura, ischemic episodes, cognitive decline, and psychiatric manifestations. It is caused by mutations in the NOTCH3 gene with autosomal dominant inheritance. We report a 45-year-old male who presented with recurrent episodes of migraine over the last fifteen years and a history of migraine in his family members. A review of his imaging suggested CADASIL as a possible diagnosis. He underwent genetic testing which showed a positive NOTCH3 mutation. This report aims to highlight the importance of considering CADASIL as a cause of familial migraine.

KEY WORDS: CADASIL, Migraine, Stroke.

History

This 45-year-old patient born of a non-consanguineous marriage presented to the outpatient department with a history of recurrent episodes of hemicranial headache associated with preceding aura ,nausea and photophobia. On questioning, there was a history of similar headaches in his father and elder sister. Clinical examination revealed no focal neurological deficits. Considering the clinical history suggestive of migraine headaches and positive family history, the possibilities of Familial Hemiplegic Migraine and CADASIL were also considered. Imaging done - MRI Brain -showed multiple hyperintense areas on T2 FLAIR sequences involving the subcortical and periventricular white matter (Figure 1) including the anterior temporal lobes. Diffusion weighted imaging (DWI) showed no areas of restriction while susceptibility weighted imaging (SWI) sequences (Figure 2) showed evidence of micro bleed in right basal ganglia.

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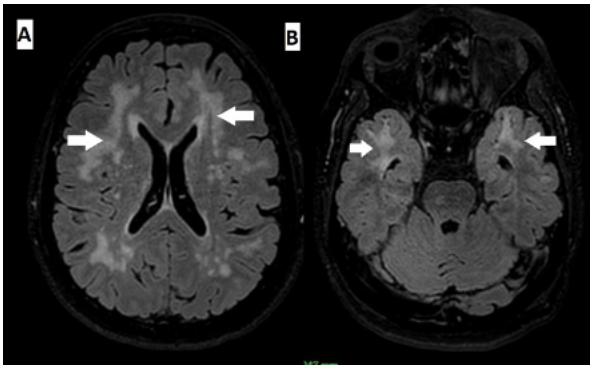

Figure 1: MRI T2 FLAIR sequences showing A. multiple hyperintensities involving the subcortical and periventricular regions B. Typical involvement of anterior temporal poles

Discussion

Considering the clinical history and imaging findings, the possibility of a CADASIL was considered. The patient underwent a clinical exome sequencing which was positive for NOTCH3 mutation on chromosome 19, confirming the diagnosis. CADASIL is the most common monogenic disorder presenting in young adults in their thirties with episodes of migraine headache with aura.[1] This is followed by recurrent episodes of subcortical stroke which can lead to progressive cognitive decline.[2]

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Less common features include depression, mood disturbances, seizures, encephalopathy and parkinsonism.[3] Imaging shows characteristic features on MRI and diagnosis is made by genetic analysis showing the characteristic NOTCH3 mutation.[4] The overall life expectancy is reduced in these patients due to multiple strokes causing cognitive decline, and cardiac events. Early diagnosis and initiation of anti-platelet agents can help in stroke prevention though effect on long term outcomes in still unclear.

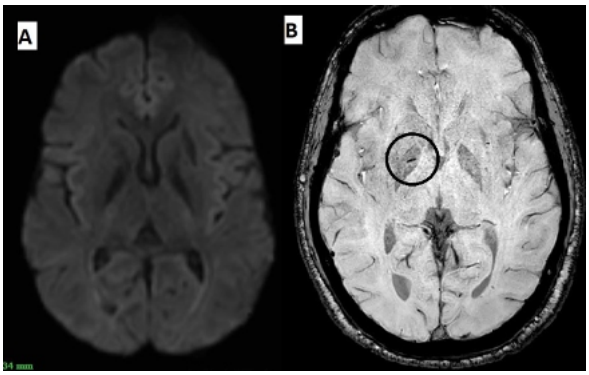

Figure 2: A. Diffusion weighted imaging (DWI) showing no areas of restriction B. Susceptibility weighted imaging (SWI) showing microbleed in right basal ganglia

Abbreviations

- CADASIL - Cerebral Autosomal Dominant Arteriopathy with Subcortical Infarcts and Leukoencephalopathy
- MRI - Magnetic Resonance Imaging
- FLAIR - Fluid Attenuated Inversion Recovery
- SWI - Susceptibility Weighted Imaging

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Conflict of interest

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