Susac Syndrome: A Clinical Triad…Eventually

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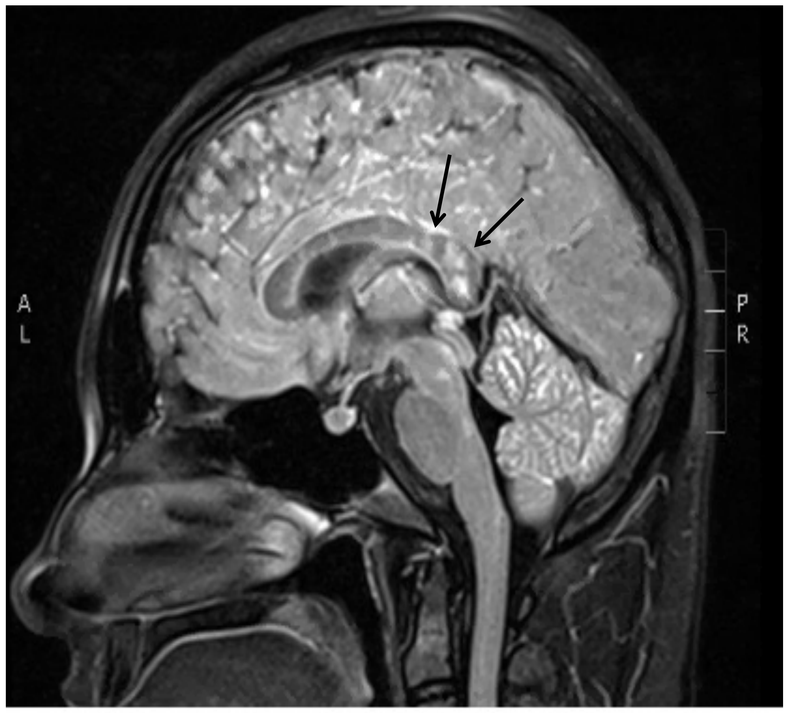
Introduction

Susac syndrome is a rare autoimmune microangiopathy that causes microvascular occlusions in the brain, retina and inner ear. The disease predominantly affects young females and manifests with a clinical triad of encephalopathy, visual disturbances, and hearing loss. The exact pathogenesis for Susac syndrome is unknown and there are no specific biologic markers. Diagnosis is based on the clinical history and specific characteristic imaging findings[1].

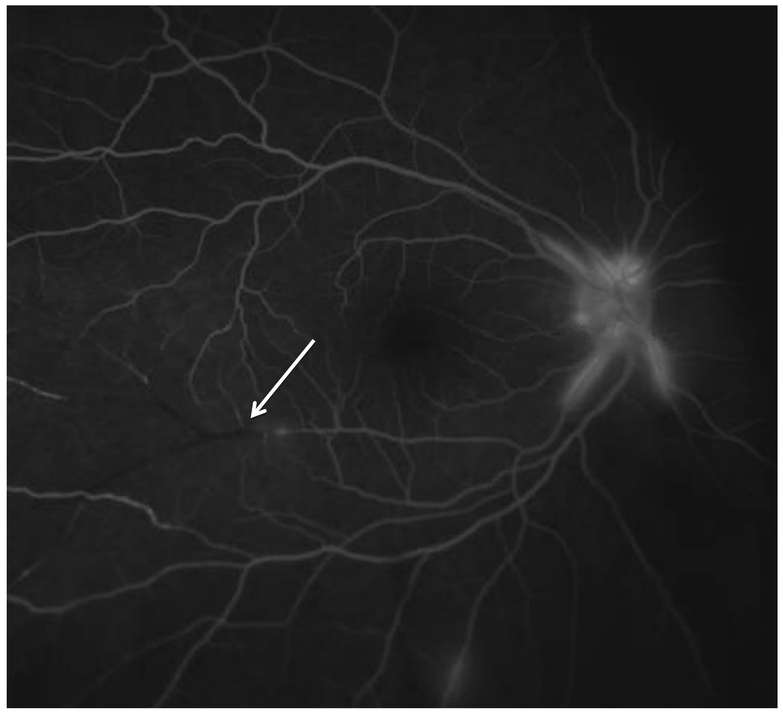
Case Presentation

An 18-year-old previously healthy female presented with four weeks of worsening headache, ataxia, and confusion after a trip to Mexico. On exam she was alert but disoriented, minimally verbal with inappropriate laughter. Neurologic exam was limited due to poor understanding but there was no obvious focal deficit, dyskinesia or neck stiffness. The remainder of the physical exam was normal. CBC and CMP were normal. CSF demonstrated 7 WBCs (82% lymphocytes, 13% monocytes, 2% eosinophils, 3% neutrophils) and elevated protein of 247 mg/dl without oligoclonal bands. MRI brain is shown below. CT angiogram was normal.

Imaging



MRI Brain: abnormal leptomeningeal enhancement of the cerebellum and brainstem, scattered T2 hyperintensities of the subcortical white matter and corpus callosum.



Fluorescein Angiography: areas of capillary drop out and retinal non perfusion within the mid-peripheral retina in both eyes

Hospital Course

Extensive workup for infectious, rheumatologic, and paraneoplastic conditions was nondiagnostic. She was treated empirically with high dose steroids and IVIG with slow improvement in her mentation. After three weeks in the hospital, family noticed she was rubbing her eyes and seemed to have trouble hearing. An eye exam with fluorescein angiography (FA) revealed areas of retinal non-perfusion consistent with branch retinal artery occlusion (BRAO). Audiometry found significant bilateral hearing loss. Based on these findings a diagnosis of Susac syndrome was made. She was started on a slow steroid taper, monthly IVIG, azathioprine and aspirin. Follow up six months later showed improved cognition; she was living at home with parental supervision and helping with light chores. She received bilateral cochlear implants for the hearing loss.

Discussion

Diagnosing Susac syndrome can be challenging because of its rarity, its clinical heterogeneity and the lack of specific disease biomarkers. Only 13% of patients have the full clinical triad at presentation, although 85% will develop it eventually[2]. Headache and encephalopathy are the most common CNS manifestations, but ataxia, upper motor neuron signs and frank psychosis can occur[3]. As in this case, there is often significant time lapse before the clinical triad is fulfilled; the average delay is 21 weeks[2]. This often leads to late diagnosis, misdiagnosis, and delayed initiation of therapy. While there are no specific biomarkers for Susac syndrome, there are characteristic imaging findings that are key to diagnosis; 98% of reported cases have corpus callosum lesions on MRI, and 99% that undergo FA have BRAOs[3]. Treatment consists of high dose steroids and IVIG, with additional immunosuppressive agents for refractory disease[3].

Take Home Points

- Susac syndrome manifests with a clinical triad of encephalopathy, visual disturbances, and hearing loss
- It is important to be aware of Susac syndrome in the differential diagnosis for encephalopathy
- Consider ophthalmoscopic evaluation early in the workup of encephalopathy if the etiology remains unclear

References

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