

# Susac's Syndrome: A Rare Presentation and Course



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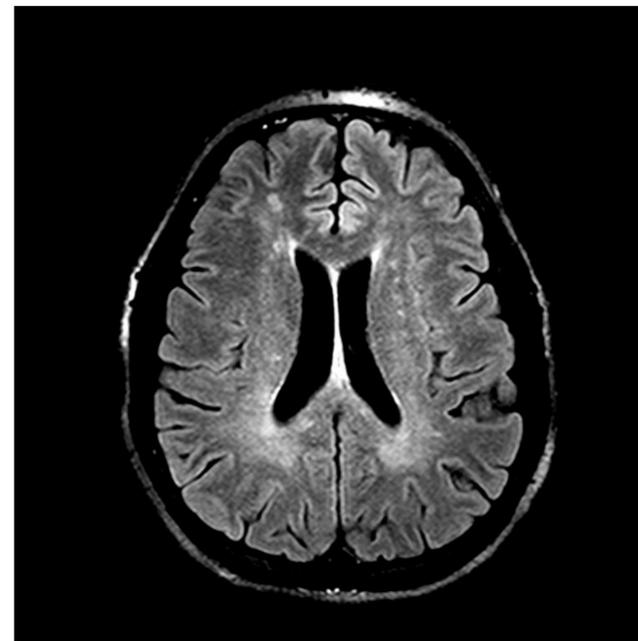
## Introduction

Susac's Syndrome is a rare immune-mediated microangiopathic disease affecting arterioles of the cochlea, brain, and retina, which manifest as a triad of hearing loss, encephalopathy, and visual changes.<sup>1</sup>

Diagnosis is based on clinical presentation, brain magnetic resonance imaging, retinal fluorescein angiography and audiometry.<sup>1</sup>

## Case Presentation

- A 32-year-old male presented for two months of bilateral hearing loss with tinnitus. He also reported new onset of anxiety, paranoia, and behavioral problems.
- History revealed exposure to a chemical spill of sodium borohydride and antibiotic treatment for a suspected unilateral ear infection.
- Prior medical history included tobacco use disorder. Physical examination was unremarkable. Testing for systemic lupus erythematosus, rheumatoid arthritis, vasculitis, tuberculosis, and syphilis were negative. CBC and CMP were within normal limits.
- Audiogram confirmed low-frequency bilateral symmetric sensorineural hearing loss.
- MRI of the brain showed multiple less than 5-millimeter diameter ill-defined T2 hyperintensities in bilateral frontoparietal deep white matter and corpus callosal cystic lesions.
- Underlying autoimmune etiology was suspected and he began a course of high dose corticosteroids.
- Subsequent ophthalmologic evaluation on fluorescein sonography found multiple bilateral peripheral branch retinal artery occlusions.
- With the complete clinical triad, Susac's syndrome was diagnosed.
- Symptoms were resistant to high dose corticosteroids.
- He was treated with intravenous immunoglobulins for six months. He saw some improvement with personality changes and coordination but still reported vertigo.
- Mycophenolate mofetil was initiated with subjective improvement of all symptoms.
- Subsequent ophthalmologic evaluation revealed near resolution of the retinal artery occlusions in both eyes on fluorescein angiography. Additionally, subsequent MRI imaging revealed stable areas of increased T2 and decreased T1 corpus callosal lesions with no new lesions.
- He still reports ongoing mild tinnitus.



## Images

*Brain MRI demonstrating corpus callosal cystic lesions, or "snowball" lesions, which are classic to Susac's syndrome. Additional findings include multiple less than 5 mm diameter ill-defined T2 hyperintensities in the bilateral frontoparietal deep white matter.*

## Discussion

Only a few hundred cases of Susac's Syndrome have been reported.

Classic diagnostic findings for Susac's Syndrome:

- 1) sensorineural hearing loss on audiometry
- 2) retinal artery occlusions on retinal fluorescein angiography
- 3) multi-focal lesions of corpus callosum on brain MRI<sup>1</sup>

Signs and symptoms can occur at different times.<sup>2</sup>

Vestibular-cochlear symptoms are often irreversible, while neurologic and ophthalmologic symptoms can be reversible.<sup>2</sup>

The underlying pathophysiologic mechanism is suspected to be complement activating IgG1 subclass anti-endothelial antibodies that damage precapillary arterioles of the brain, inner ear and retina (though they are only present in 30% of cases and are unknown if pathogenic or specific).<sup>1 2</sup>

Various therapeutic regimens have been described but standardized treatment is lacking.

The most common treatment options are high dose corticosteroids and intravenous immunoglobulins.<sup>2</sup>

## Conclusion

Diagnosis of Susac's Syndrome is challenging but early recognition and treatment are critical to protect from irreversible organ damage.<sup>2</sup>

As exemplified by this case, mycophenolate mofetil is an excellent alternative for refractory cases.

## References

<sup>1</sup> Pereira S, Vieira B, Maio T, Moreira J, Sampaio F. Susac's Syndrome: An Updated Review. *Neuroophthalmology*. 2020 May 1;44(6):355-360. doi: 10.1080/01658107.2020.1748062. PMID: 33408428; PMCID: PMC7746245.

<sup>2</sup> Monferrer-Adsuara C, Remolí-Sargues L, Hernández-Bel L, Gracia-García A, Hernández-Garfella ML, Cervera-Taulet E. Rituximab in the treatment of Susac's syndrome: Report of a case. *Eur J Ophthalmol*. 2020 May 7;1120672120924545. doi: 10.1177/1120672120924545. Epub ahead of print. PMID: 32380850.