

## Susac Syndrome

Category: Medicine & Medical Specialties, Poster Presentation

Disclosure: The authors did not report any financial relationships or conflicts of interest

[Supplemental Video](#)

Presenting Author: Aswani Thurlapati, MD, Internal Medicine Resident PGY2, Department of Internal Medicine, LSU Health Shreveport, Shreveport, Louisiana

Coauthors: Phani P Morisetti MD, Associate Professor, Department of Nephrology and Internal Medicine, LSU Health Shreveport, Louisiana

### Introduction

With an unknown prevalence, Susac syndrome is a rare autoimmune endotheliopathy that causes micro-ischemic damage to vessels in the brain, ears, and eyes. It is mainly seen in young adults, with a median age of onset of 32 years. Here, we report a case of Susac syndrome diagnosed in an elderly female.

### Case Presentation

History: Our patient is a 62-year-old female, with significant history of type 2 diabetes mellitus, essential hypertension, stable ascending aortic aneurysm, and pituitary adenoma status-post resection, presented to the emergency department with acute onset of worsening weakness of bilateral lower extremities, ataxia with recurrent falls, and bilateral hearing loss for one week prior to presentation. During the hospital course, she became confused and disoriented to time and place. She eventually developed transient blurring of vision. There was no reported history of recent travel, fever, rash, headache, urinary or bowel incontinence, seizures, loss of consciousness, chest pain, or shortness of breath.

Physical Exam: On the physical exam, the patient had a flat affect and was alert and oriented to name. Weber's test is localized to the midline and Rinne's test showed bilateral air conduction greater than bone conduction, suggestive of bilateral sensorineural hearing loss. All cranial nerves were intact. Motor strength, power, tone, reflexes, sensations, and coordination were intact.

Differential Diagnosis: Guillain-Barre syndrome, brain mass, brain abscess, encephalitis, inflammatory myopathy, peripheral neuropathy, idiopathic intracranial hypertension

Tests and results: Patient's blood counts, serum chemistries, liver and renal function tests were within normal values. Blood alcohol level, urine drug screen, heavy metal screen, and urine fractionated porphyrin were negative. Acute hepatitis panel, RPR, HIV, COVID-19, and diphtheria antibody, and anti-nuclear antibody screen are negative. MRI brain showed minimal chronic microvascular ischemic changes. MRI cervical, thoracic, and lumbar spines were all unremarkable. CSF showed RBC 14,100 cu/mm, WBC 0 cu/mm, glucose 54 mg/dl, and protein 74 mg/dl. CSF culture, herpes PCR, VDRL, and paraneoplastic panel were negative. Electromyography and nerve conduction study revealed no electrophysiological evidence of inflammatory or degenerative myopathy. Electroencephalogram was normal. Serum protein electrophoresis was unremarkable. Rheumatoid factor, anti-citrullinated peptide antibody, Lyme antibody, acetylcholine receptor ganglionic neuronal antibody, CRMP-5 IgG antibody, neuronal V-G K+ channel antibody, NMO antibody, N-type and PQ-type calcium channel antibodies, AGNA type 1,2,3 antibodies, PCA 1,2,Tr antibodies, amphiphysin antibody, striated muscle antibody, and ganglioside antibody were all negative.

### Diagnosis

As the majority of the tests was unrevealing, the patient was initially treated as a variant of Guillain-Barre syndrome with 5 doses of intravenous immunoglobulin, but had no improvement. She also underwent a course of plasma exchange therapy without any improvement. Later, due to sudden onset of transient vision loss, neuro-

ophthalmology was consulted. Patient was noted to have focal retinal whitening at the proximal trunk of the inferotemporal vascular arcade with several faint peripapillary cotton wool spots suggestive of retinal vasculitis. Due to the patient's triad of retinal changes, bilateral sensorineural hearing loss, and confusion, the patient was ultimately diagnosed with Susac syndrome.

#### Management

Patient received an infusion of cyclophosphamide 800mg along with mesna, which improved her symptoms gradually over a week. She is scheduled for outpatient infusion in two weeks with a repeat in 6 months. Patient discharged to a nursing home for rehabilitation and assistance.

#### Learning Objectives

- Identify and raise awareness of rare diseases which we may encounter in our clinical practice
- Discuss the various neurological testing performed prior to coming to a diagnosis
- Describe the physical features, presentation, and management of Susac syndrome
- Raise the importance of ophthalmological evaluation and its key role in identifying Susac syndrome