Sri Lanka Journal of Medicine **SLJM**

Case Report

Citation Alahakoon AMBD, Malalasekara S, Dissanayake AMSDM et al. 2026 Susac Syndrome in a Young Woman: A Case of Misdiagnosis and Irreversible Visual Sequelae. Sri Lanka Journal of Medicine, pp. 54-58.
DOI: https://doi.org/10.4038/sljm.v35i1.600

Susac Syndrome in a Young Woman: A Case of Misdiagnosis and Irreversible Visual Sequelae

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ABSTRACT

Susac syndrome is a rare autoimmune disorder defined by the triad of encephalopathy, branch retinal artery occlusions (BRAO), and sensorineural hearing loss. Its nonspecific presentation often leads to misdiagnosis and delayed treatment, resulting in irreversible complications. We report a 28-year-old woman with six months of progressive headaches, transient visual symptoms, and behavioral changes, initially diagnosed as migraine and functional neurological disorder. Further evaluation revealed right temporal quadrantanopia, right BRAO, and sensorineural hearing loss, confirming Susac syndrome. Immunosuppressive therapy improved headaches, behavior, and hearing; however, visual deficits were permanent. Early recognition and prompt treatment are essential to prevent lasting neurological and ophthalmic damage.

Keywords: Susac syndrome, visual impairment, headache

INTRODUCTION

Susac syndrome is an exceptionally rare autoimmune microvascular endotheliopathy characterized by a triad of encephalopathy, branch retinal artery occlusions (BRAO), and sensorineural hearing loss. Since its initial description in 1979, approximately 450 to 500 cases have been reported worldwide (2).

The time from symptom onset to a definitive diagnosis varies significantly. In a study of 16 patients, the diagnostic delay ranged from 2 weeks to 10 months (2). Another study reported a median time to diagnosis of 7 months, with a range from 0 to 126 months (3).

This delay is largely due to the syndrome's diverse and often nonspecific initial symptoms, which can lead to misdiagnosis. Notably, fewer than 15% of patients present with the full triad at disease onset, making early recognition particularly challenging (4). The absence of objective biomarkers for disease activity further adds to the diagnostic and management challenges.

We present a case of a 28-year-old woman who was misdiagnosed with migraine and functional neurological disorder for six months before a formal diagnosis of Susac syndrome was

Received: 2025-03-22 **Accepted revised version: 2026-01-07** **Published: 2026-04-22**

established. Further delays in treatment could have resulted in catastrophic neurological outcomes.

CASE REPORT

A 28-year-old female presented with a six-month history of episodic left-sided frontal headaches, which progressively worsened in frequency and duration. Initially, the headaches were accompanied by transient visual obscuration and blind spots, leading to a diagnosis of migraine by her general practitioner. An MRI of the brain performed at the onset of her illness was unremarkable.

The patient had multiple outpatient visits due to worsening visual symptoms and headaches, which were managed as migraine attacks. Over the past two months, her visual symptoms began to persist beyond the headache episodes. Additionally, she exhibited behavioral changes, including intermittent agitation and refusal to eat. Unfortunately, she did not respond to escalating migraine treatment. Given the presence of behavioral symptoms, a possible functional neurological disorder was considered, and she was referred to a psychiatrist.

She denied any fever, sick contacts, involuntary movements, psychotic symptoms, or recreational drug use.

At the end of the sixth month of illness, she presented to the emergency department. On admission, she was afebrile, with no neck stiffness, rashes, or orofacial dyskinesias. Her Glasgow Coma Scale score was 15/15, though she was intermittently disoriented to time and place.

A comprehensive neurological examination revealed right temporal quadrantanopia, right branch retinal artery occlusion, and left sensorineural hearing loss. There were no papilledema or any objective motor or sensory deficits.

Basic blood investigations performed on her, are as follows (Table 1).

Table 1: Basic Blood Investigations

Investigation	Value	Reference Range
Heamoglobin	112	100-140 g/l
Platelets	356	150-150 /mm^3
White cell count	7.8	4-11 / mm^3
C- Reactive Protein	<0.5	<0.5 g/dl
Erythrocyte Sediment Rate	45	<10$mmhr^{-1}$
Alanine Transferase	24	25-55 U/L
Aspartate Transferase	18	25-22 U/L
Glama Glutamic Transferase Acid	45	55-120 U/L
Alkaline Phosphatase	34	25-65 U/L
Albumin	32	30-50 g/dl
Globulin	28	20-40 g/dl
Serum sodium	138	135-145 mmol/l
Serum potassium	4.5	3.5-5.5 mmol/l
Serum Creatinine	67	35-90 U/L
Thyroid Stimulating Hormone	3.2	2-5 U/L
Thyroxine	12 .4	4-15 U/L

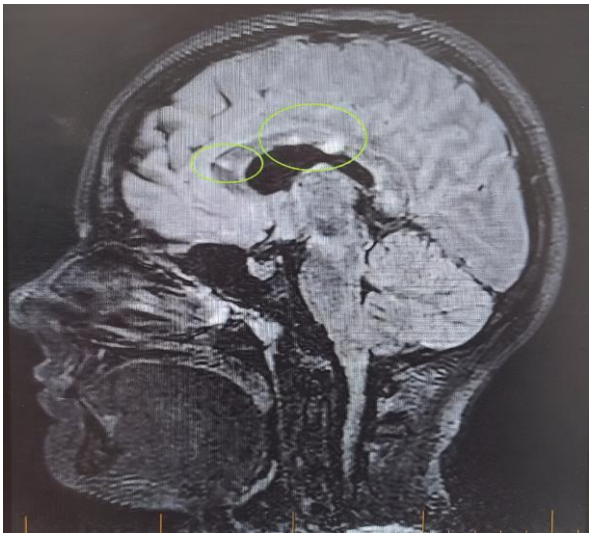

Figure 1: Magnetic Resonance Imaging (MRI) of the brain

Magnetic Resonance Imaging (MRI) of the brain was performed and reported as "widespread focal areas of restricted diffusion throughout the supratentorial and infratentorial brain suggestive of encephalitis. The appearances are atypical for cerebral vasculitis in terms of the distribution" (Figure 1).

A lumbar puncture was performed, which revealed elevated protein with normal cell count and CSF-sugar levels.

Protein: 1000 mg/L (150 – 500)
Glucose 4.3 mmol/L (2.2 – 3.9)
Cell count:
WBC's 05 x10^6/L (Mononuclear 100)
RBC's 1 x10^6/L

An extensive infective, immune, vasculitis, thrombophilia and paraneoplastic panels were performed on her blood sand CSF samples (Table 2).

Table 2: Infective, immune, vasculitis, thrombophilia and paraneoplastic panels

Panel	Investigations included in the panel
Infective panel - Negative	Serum - Treponema pallidum antibody, QuantiFERON tuberculosis, retroviral antibodies, Hepatitis serology, JC virus DNA, toxoplasma serology, varicella zoster serology, cysticercosis serology, brucella serology, taeniasis serology, Q fever serology, melioidosis serology, cryptococcal serology, cytomegalovirus serology CSF - Varicella zoster DNA, bartonella Henslee DNA, cryptococcal antigen, enterovirus RNA, herpes simplex 1 /2 DNA, JC virus DNA, mycobacterial examination, VDRL (venereal research laboratory)
Vasculitis panel - negative	Serum - antineutrophil cytoplasmic antibodies (ANCA- PR3, MPO)
Immune Panel - Negative	Serum - Anti-Neuromyelitis Optica antibody, Anti-MOG antibody, NMDA receptor antibodies, anti-VGKC (voltage gated potassium channel) antibody, ENA screen, antinuclear antibody, antimitochondrial antibody, oligoclonal assessment CSF - Anti-Neuromyelitis Optica antibody, Anti neuronal antibody (Anti-Hu, anti Ri, anti-Yo, Anti-PCA-2),
Thrombophilia screen - Negative	Serum - Antithrombin 3, Protein-c, protein-s, lupus anticoagulant screen, factor V Leiden, anti-cardiolipin antibody, anti-beta2 glycoprotein antibody,

She underwent pure tone audiometry, which revealed sensorineural hearing loss at 0.25-0,.5kHz Hz in the left ear. A fluorescein angiogram confirmed a branch retinal artery occlusion of the right eye.

DISCUSSION

cytopenias with marrow fibrosis Susac syndrome are a disorder believed to be mediated by an autoimmune response targeting endothelial cells, resulting in a characteristic clinical trial of encephalopathy, visual disturbances due to branch retinal artery occlusions (BRAOs), and sensorineural hearing impairment. First described by Dr. John O. Susac in 1979, this syndrome predominantly affects young women, with a female-to-male ratio of approximately 3:1 (5).

The exact pathogenesis of Susac syndrome remains incompletely understood, but current evidence suggests an autoimmune-mediated endotheliopathy targeting the microvasculature of the brain, retina, and inner ear. This immune response leads to occlusion of small arterioles, resulting in ischemic damage to the affected tissues (6).

Patients typically present with a combination of neurological, visual, and auditory symptoms. Encephalopathy may manifest as headaches, cognitive dysfunction, psychiatric symptoms, and, in severe cases, focal neurological deficits or seizures. Visual disturbances include BRAOs, which may lead to scotomas or visual field deficits. Sensorineural hearing loss, often accompanied by tinnitus and vertigo due to involvement of the cochlear microvasculature, is a well-recognized auditory complication.

The onset of these symptoms can be sequential or simultaneous, and not all patients exhibit the complete triad at initial presentation, posing diagnostic challenges.

Due to its rarity and frequent misdiagnosis, the exact prevalence remains uncertain. However, a study conducted in Austria estimated an annual incidence of 0.024 per 100,000 individuals (7). In contrast, a case series from Israel reported a 5.4-fold increase in incidence compared to expected rates, suggesting regional variability or increased recognition of the condition (8).

Maintaining a high index of suspicion and employing a comprehensive diagnostic approach are crucial for prompt diagnosis and management of Susac syndrome. A diagnostic criterion, including clinical, radiological, and laboratory domains, was proposed by Kleffner et al. in 2016 (9), to aid in diagnosing probable and definitive Susac syndrome. These criteria include Magnetic Resonance Imaging (MRI) findings, which typically reveal multifocal supratentorial lesions with characteristic involvement of the corpus callosum, fluorescein angiography findings of BRAOs and other retinal microvascular changes, and audiometry findings indicating sensorineural hearing loss.

Given the absence of randomized controlled trials, treatment guidelines are primarily based on cumulative clinical experience and expert consensus. The overarching goal is to suppress the autoimmune response to prevent further endothelial damage. Early and aggressive immunosuppressive therapy is advocated to mitigate disease progression and reduce the risk of permanent deficits.

A commonly recommended initial treatment regimen includes high-dose corticosteroids, such as three consecutive daily pulses of 1,000 mg methylprednisolone, followed by a course of oral prednisone. In more severe or refractory cases, additional immunosuppressive agents like mycophenolate mofetil, azathioprine, cyclophosphamide, or rituximab may be employed. The choice of agent and duration of therapy should be individualized based on disease severity and patient response (10).

Studies have shown that relapses can occur when corticosteroid dosages are tapered prematurely, underscoring the need for sustained immunosuppressive therapy. Moreover, delays in treatment initiation have been associated with the development of new clinical symptoms and progression of existing deficits (10). Therefore, early and accurate diagnosis is crucial for initiating appropriate immunosuppressive therapy to prevent irreversible neurological damage leading to dementia, hearing loss, or vision impairment.

Author declaration

Acknowledgement
Authors thank all the staff engaged in diagnosis and management of this patient members of this patient who consented for this publication.

Authors' contributions:
AMBDA: Conceptualization, design, data collection, interpretation, manuscript writing and editing; SM: Conceptualization, design, data collection, interpretation, manuscript writing and editing.

Conflicts of interest:
The authors declare that there is no financial or non-financial conflict of interest.

Funding statement:
Self-funded.

Ethics statement:
Written informed consent was obtained from the patient before they participated in the case, ensuring their understanding of the purpose, procedures, and potential implications involved.

Statement on data availability:
All data are available upon request.

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