Susac's syndrome – a case report

Clinical Case

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Abstract

Susac's Syndrome (SS) is an occlusive immune-mediated microangiopathy that preferentially affects cerebral, retinal and cochlear arterioles. Classically, it is associated with the triad of visual defects, encephalopathy and sensorineural hearing loss (SNH).
Diagnosis is based on clinical features and findings from fluorescein angiography (FA), brain magnetic resonance imaging (CE-MRI) and audiometry.
Treatment is based on high-dose corticosteroid therapy and must be started as soon as possible to prevent permanent sequelae. The clinical case presented is about a 34-year-old woman with multiple visits to the Emergency Department due to persistent headaches, psychomotor slowing and focal neurological deficits lasting 2 months. From the study carried out, multiple hyperintense lesions were found on T2 CE-MRI involving the corpus callosum, areas of retinal ischemia and hyperfluorescence on FA and bilateral moderate SNH at high frequencies on the audiogram.
The diagnosis of SS was assumed and immunosuppressive therapy was started, maintaining clinical and audiometric surveillance.
Keywords: Susac's Syndrome; encephalopathy; sensorineural hearing loss; retinal artery occlusion; audiometry

Introduction

Susac syndrome (SS) is a rare immune-mediated pauci-inflammatory microangiopathy characterized by microvascular occlusions, which primarily affects the cerebral, retinal, and cochlear arterioles.[1] It predominantly occurs in women aged between 20–40 years, with a female-to-male ratio of 3:1.[2]
SS is associated with a clinical triad of visual disorders, subacute encephalopathy, and sensorineural hearing loss (SNHL).[3] However, the presentation of SS can vary, and the complete triad is present in less than 20% of patients in the early stages;[2,4–6] thus, it requires a high index of suspicion. The syndrome typically begins with neurological symptoms, which generally dominate the clinical

picture. However, the severity of the disease is primarily attributed to retinal damage, which can present as decreased visual acuity, photopsia, or scintillating scotomas. Inner ear arteriopathy can affect the cochlear apex, resulting in sudden and sometimes bilateral SNHL. The diagnosis of SS is based on clinical history, fundoscopy, fluorescein angiography (FA), brain magnetic resonance imaging (MRI), and audiometry findings.

Fundoscopic examination reveals areas of retinal infarcts secondary to branch retinal artery occlusion (BRAO), with intact choroidal circulation and absence of inflammation.[1] FA allows the identification of Gass plaques and arteriolar wall hyperfluorescence in areas proximal to sites of occlusion and distant from bifurcations (in contrast to BRAO). These plaques indicate endothelial dysfunction and lipid extravasation from retinal vessels. They usually develop in the acute phase of SS, vary with the disease activity, and disappear in response to treatment.[6,7] Brain MRI typically shows white matter involvement, with hyperintense lesions in the tentorium cerebelli and corpus callosum on T2-weighted images ("snowball lesions"). These lesions represent micro-infarction areas, which subsequently progress to hypointense lesions on T2-weighted images ("holes").[8] Tonal and vocal audiometry often shows asymmetric bilateral SNHL predominantly at medium-low frequencies, accompanied by a speech discrimination deficit.[9] On laboratory analysis, SS can be associated with increased acute-phase reactants, and occasionally, increased titers of anti-nuclear and anti-phospholipid antibodies and factor VIII and von Willebrand factor, indicating underlying endothelial dysfunction.[4] The window of opportunity to prevent irreversible damage is short, posing a significant therapeutic challenge. SS treatment is based on high-dose intravenous corticosteroid therapy with gradual weaning, generally together with an immunomodulatory agent such as intravenous immunoglobulin. The duration and intensity of therapy depend on the patient's condition.[5,10] Patients with prothrombotic risk factors may benefit from concomitant antiaggregation and anticoagulation.[1]

This case report describes a case of SS, including the typical signs and symptoms, diagnostic steps, therapeutic interventions, and their impact on prognosis.

Case report

A 34-year-old obese woman presented with a two-month history of multiple visits to the emergency service (ES) due to a holocranial headache, which was refractory to analgesia. The headache was associated with phono- and photophobia and focal neurological deficits, including left facial and upper limb paresthesia, diplopia, and ataxia. Subsequently, the patient developed uncontrollable vomiting, imbalance, unilateral hearing loss, and tinnitus, which led to progressive functional impairment. She had no significant personal or family history and used no medications except for combined oral contraceptives. During the initial evaluation at the ES, the patient was found to have marked psychomotor retardation, and her preliminary neurological examination supported the clinical description, with no campimetry changes or cranial nerve deficits. Vestibular examination showed no spontaneous or gaze-evoked nystagmus or skew deviation, and the head impulse test was negative. Romberg's test showed imbalance to the right, without any other consistent changes. Tuning fork test and otoscopy revealed no significant findings. Complementary diagnostic tests revealed no significant changes. The electroencephalogram indicated moderate to severe encephalopathy, and brain MRI ruled out cerebral venous thrombosis or space-occupying lesions. Lumbar puncture showed hyperproteinorrachia without any oligoclonal bands, and microbiological culture was negative. Brain MRI demonstrated multiple hyperintense lesions in the corpus callosum on T2-wighted imaging, no contrast uptake, and diffusion restriction (Figure 1). Fundoscopy revealed BRAO, and FA demonstrated flow interruption in multiple vessels and vascular wall hyperfluorescence distant from the

Figure 1
Brain magnetic resonance imaging (MRI) in axial section shows multiple lesions involving the corpus callosum and internal capsule (arrowheads), no contrast uptake on T2-wighted image (A), and diffusion restriction (B).

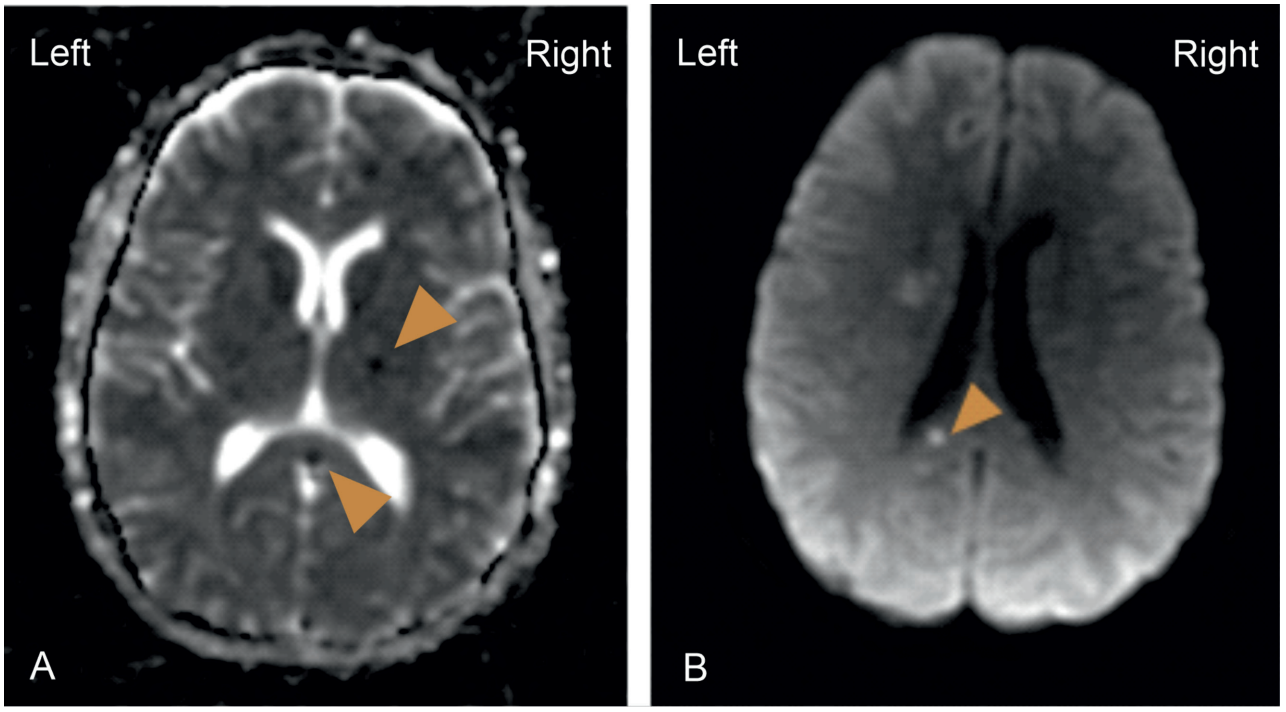
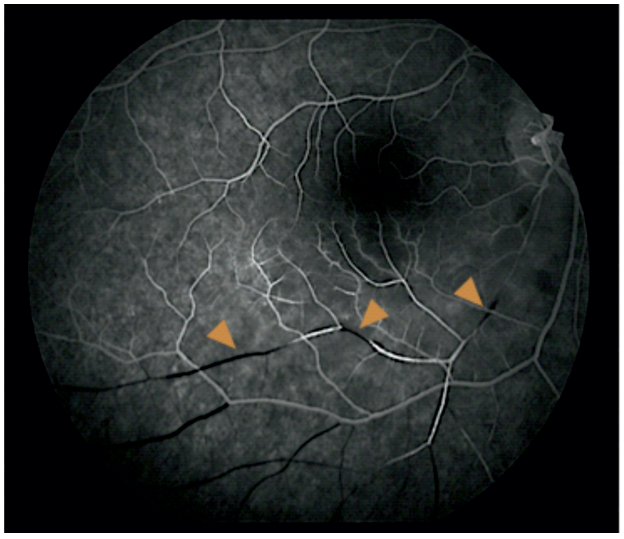


Figura 2
FA showing inferior temporal branch retinal artery occlusion (arrowheads) associated with retinal ischemia



occlusion zones (Figure 2). Tonal and vocal audiogram revealed bilateral SNHL with left predominance at higher frequencies (pure tone average [PTA] of 46 dB) (Figure 3). The possibility of SS was considered based on the clinical, imaging, and audiometric findings.

Treatment with intravenous methylprednisolone 1 g was initiated, which was switched to oral prednisolone 1 mg/kg/day after 5 days, acetylsalicylic acid, and intravenous immunoglobulin. The patient was closely monitored to assess the clinical progress.

Audiometry was repeated after the first week of treatment, and revealed increased thresholds and hearing loss at medium-low frequencies (PTA of 66 dB) (Figure 4).

After two weeks of hospitalization, the patient was discharged with regular clinical and audiometric follow-up. She was also recommended 20 hyperbaric oxygen therapy (HBOT) sessions, with subsequent audiometry showing thresholds overlapping those observed prior to treatment. Initially, the patient showed clinical improvement, but subsequent neurological worsening in the first month after discharge required intensification of the immunosuppressive therapy. Currently, the patient is taking rituximab and weaning off oral prednisolone, with partial neurological improvement; however, the otologic symptoms persist despite the treatment.

Figure 3
Tonal and vocal audiogram show bilateral SNHL with left predominance

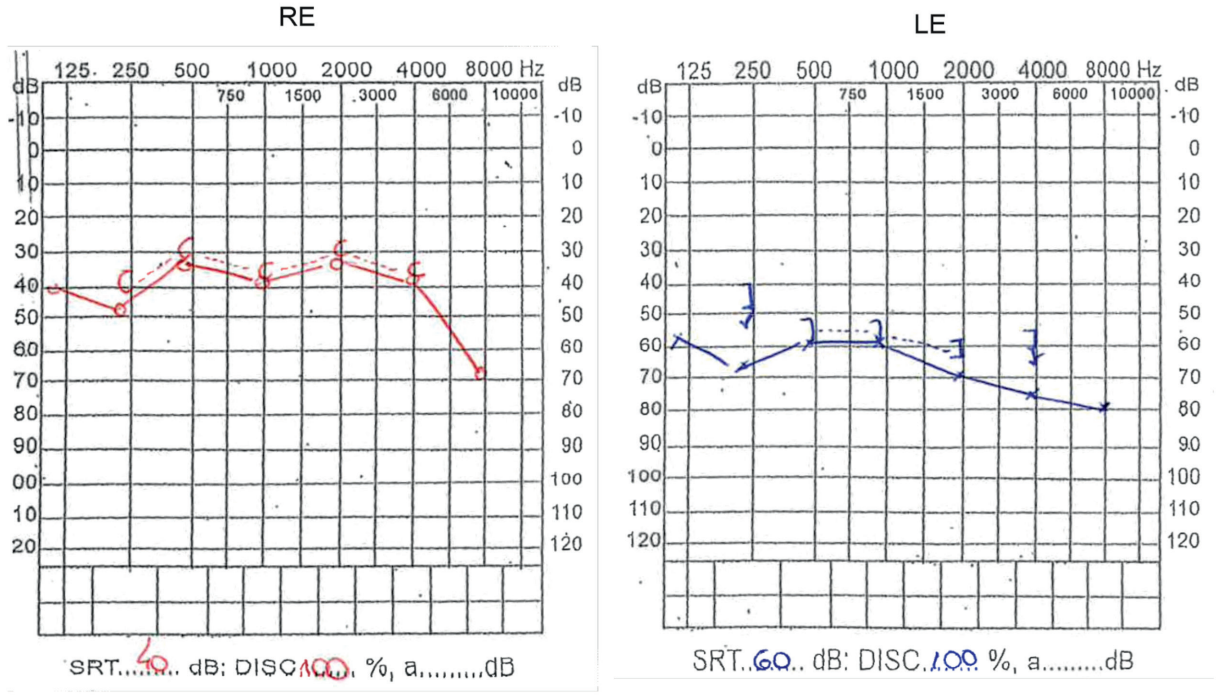
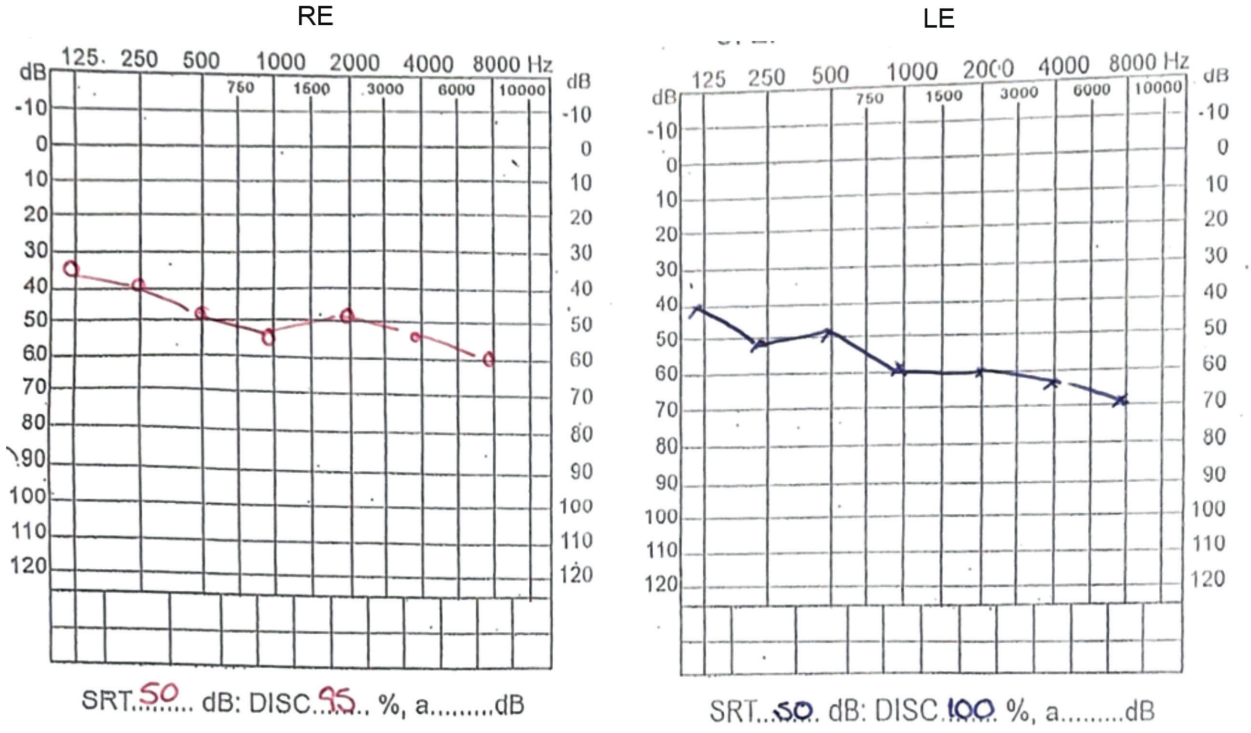


Figure 4
Tonal and vocal audiogram after the first week of treatment



Discussion

In this case, the presence of a persistent headache, encephalopathy, focal neurological deficit, and SNHL, which were not explained by a lesion in a single vascular territory, raised the suspicion of SS, which was confirmed by FA and brain MRI findings. According to the literature, headache is the most common prodromal symptom of SS, occurring in 80% of the cases, and often precedes encephalopathy by several months.[1] Subsequently, the patient can present with altered levels of consciousness, memory changes, cranial nerve palsies, behavioral changes, seizures, and dementia.[4] Analyzing the cerebrospinal fluid and differentiating SS from central nervous system disorders with a similar presentation, such as multiple sclerosis and acute disseminated encephalomyelitis, is crucial.[9]

Hyperproteinorrachia and lymphocytic pleocytosis have been observed in SS, with oligoclonal bands being generally absent,[4] as observed in the present case. While retinal damage has a major impact on the quality of life, symptoms may be minimal when the peripheral retina is affected,[1] as in the present case.

SNHL at higher frequencies observed in this case contrasts with the typical involvement of the cochlear apex and lower-frequencies described in the literature.[4,7] Conversely, the peripheral vestibular system may be affected, leading to vertigo and tinnitus,[1] making differentiation from Ménière's syndrome difficult.[11] The natural progression of SS varies, with some cases following a monophasic, self-limited course with complete resolution within two years. However, more severe forms can include a polycyclic relapse-remission or a continuous course with prolonged symptoms and permanent sequelae.[1–2, 5]

Therefore, despite an uncertain diagnosis, early treatment initiation can delay SS progression and prevent permanent sequelae.

In case of relapse or disease refractory to initial therapy, alternative immunosuppressants like mycophenolate mofetil, cyclophosphamide, or rituximab, may be prescribed, as was done in the present case. HBOT has shown promise in the treatment of BRAO and cochlear ischemia. While visual disorders can be resolved or alleviated with treatment, SNHL is often permanent,[1,5,11] as the cochlea is more susceptible to irreversible ischemic injury and has a smaller window of opportunity for intervention.[6] In these cases, hearing aids or cochlear implantation may be indicated.

Despite the generally favorable prognosis of SS, several cases of recurrence have been reported, sometimes recurring several years after the initial diagnosis and occasionally associated with pregnancy.[11,12] Therefore, these patients should be followed up with regular clinical, imaging, and audiometric evaluations.

Conclusion

SS can manifest as SNHL with an unclear cause. Although it is a rare condition, it can have disabling symptoms and a significant impact on the quality of life of affected individuals; thus, it is considered an important condition in clinical practice.

Conflicts of Interest

The authors declare that there is no conflict of interests regarding the publication of this paper.

Data Confidentiality

The authors declare having followed the protocols in use at their working center regarding patients' data publication.

Protection of humans and animals

The authors declare that the procedures were followed according to the regulations established by the Clinical Research and Ethics Committee and to the 2013 Helsinki Declaration of the World Medical Association.

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Availability of scientific data

There are no datasets available, publicly related to this work.

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