

Susac's syndrome: isolated retinal artery occlusion after 10 years of remission

Síndrome de Susac: oclusão isolada da artéria retiniana após 10 anos de remissão

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ABSTRACT | Susac's syndrome, or microangiopathy of the retina, inner ear, and brain, is a rare condition characterized by the clinical triad of encephalopathy, branch retinal artery occlusion, and sensorineural hearing loss. The complete triad has been documented in 85% of reported cases. At clinical onset, the most common manifestations are central nervous system symptoms, followed by visual symptoms and hearing disturbances. Although the clinical course of Susac's syndrome is usually self-limiting, fluctuating, and monophasic, clinical polycyclic and chronic courses have also been described. Likewise, recurrences of the full triad after more than 10 years of remission have been reported. We describe a 21-year-old woman who presented with branch retinal artery occlusions and magnetic resonance imaging findings compatible with Susac's syndrome without objective hearing loss. After 10 years of remission, the patient complained of visual field loss due to new retinal ischemia. Neither other symptoms nor neuroimaging or audiometry pathologic findings were found during the clinical course.

Keywords: Susac's syndrome/diagnosis; Ear diseases; Brain diseases; Hearing loss, sensorineural; Retinal artery occlusion

RESUMO | A síndrome de Susac, ou a micro angiopatia da retina, do ouvido interno e do cérebro, é uma condição rara caracterizada pela tríade clínica de encefalopatia, oclusão de ramo da artéria retiniana e perda de audição neuro-sensorial. A tríade completa é documentada em 85% dos casos registrados. No início dos sinais clínicos, a manifestação mais

comum relaciona-se ao sistema nervoso central, seguida por sintomas visuais e distúrbios auditivos. Apesar do curso clínico da síndrome de Susac ser usualmente auto limitante, variável e monofásico, cursos clínicos policíclicos e crônicos têm sido também descritos. Do mesmo modo, recorrências da tríade completa após mais de 10 anos de remissão têm sido relatadas. Descrevemos o caso de uma mulher de 21 anos que apresentava oclusões de ramos da artéria retiniana e imagens por ressonância magnética compatíveis com a síndrome de Susac, sem comprometimento objetivo da audição. Dez anos após a remissão, a paciente queixou-se de perda de campo visual devido a uma nova isquemia da retina. Nenhum outro sintoma, ou neuroimagem ou achado audiométrico patológico foi observado durante o curso clínico.

Descritores: Síndrome de Susac/diagnóstico; Otopatias; Encefalopatia; Perda auditiva neurosensorial; Oclusão da artéria retiniana

INTRODUCTION

Susac's syndrome is a rare disorder that is thought to be caused by autoimmune-mediated occlusions of microvessels in the brain, retina, and inner ear, leading to a characteristic clinical triad of encephalopathy, branch retinal artery occlusions, and neurosensory hearing loss^(1,2). It affects middle-aged women more often than men and is probably underdiagnosed because all three features may not be present at the onset, which complicates the diagnosis⁽³⁻⁶⁾. Since the first descriptions of Susac's syndrome in 1979⁽¹⁾, slightly more than 300 cases have been reported in the literature. Given the lack of systematic data on Susac's syndrome, the reports are vague with regard to age and gender differences, clinical patterns of presentation, prognosis, and treatment. The natural course of Susac's syndrome is somewhat unpredictable and is still a topic of considerable controversy.

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We present a case of recurrence of retinal artery occlusions in a patient with Susac's syndrome after 10 years of remission.

CASE REPORT

A 21-year-old Caucasian woman presented in 2006 with spotty visual field loss in her left eye. Three months earlier, she had experienced migrainous headaches accompanied by photophobia and sonophobia. Her best corrected visual acuity (BCVA) was 1 in the left eye and 0.8 in the right eye. The anterior segments of both eyes were unremarkable. Fundus examination of the left eye showed a superotemporal branch retinal artery occlusion (BRAO). Fluorescein angiography confirmed the presence of BRAO in her left eye and demonstrated arterial wall hyperfluorescence (AWH) along the left superotemporal artery (Figure 1).

Several days later, the patient reported similar symptoms in her right eye with migrainous headache and paresthesias in both legs. She was found to have a BRAO along her right superotemporal arteriolar arcade. Visual field testing showed a total inferior field defect in her right eye with a small field remnant in her lower temporal quadrant. Her left eye had a small scotoma in the nasal quadrant and a superotemporal field defect. Audiometry did not reveal any sensorineural deficit.

Cranial magnetic resonance imaging (MRI) showed supratentorial, infratentorial, and callosal small and multifocal white matter lesions on T2-weighted and fluid attenuated inversion recovery sequences (FLAIR) with mild contrast enhancement more intense in the infratentorial structures (Figure 2). Cerebrospinal fluid analysis revealed hyperproteinorrachia and no presence of oligoclonal bands. An extensive systemic workup, which included serum markers of connective tissue diseases and serology, did not find any evidence of an autoimmune or infectious etiology.

Given the clinical, fundusoscopic, and neuroimaging findings, a diagnosis of Susac's syndrome was made. The patient received treatment with high-dose intravenous methylprednisolone (1 g) for 5 days and then prednisone 40 mg per day and anticoagulation therapy with acenocumarol. Prednisone was tapered as she received azathioprine 50 mg per day. The patient became pregnant 3 years later, and therefore the treatment was changed to acetylsalicylic acid 300 mg per day.

In 2016, after 10 years free of symptoms, the patient reported spotty temporal visual field loss in her right eye, spotty nasal visual field loss in her left eye, scintillating escotomas, and headache. No subjective episodes of deterioration of memory or hearing were reported. The anterior segments of both eyes and audiometry findings were unremarkable. Fundus examination and fluorescein

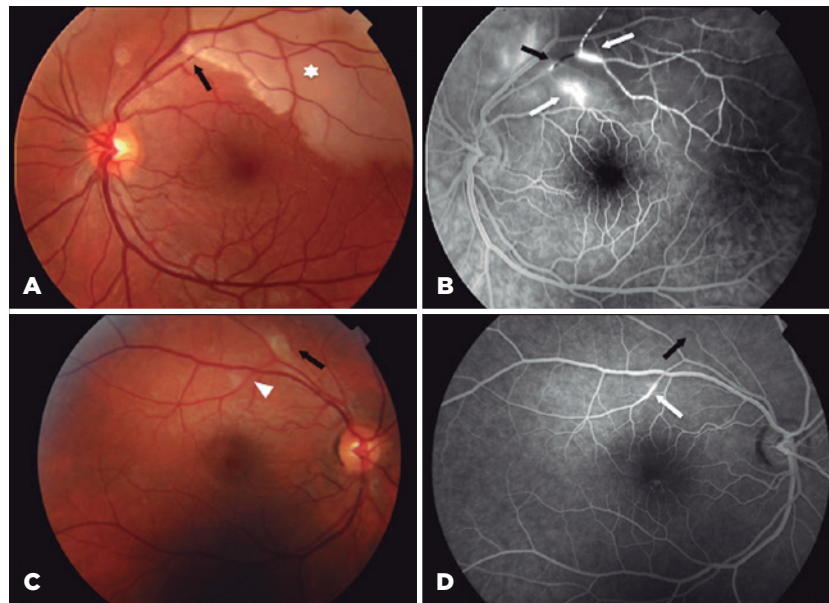


Figure 1. Optic fundus and fluorescein angiography at onset (A and B) and 10-year follow-up (C and D) showing temporal superior arteriolar occlusions (black arrows), retinal pallor (asterisk), segmental arteriolar whitening (white arrowhead), and arteriolar wall hyperfluorescence (white arrows).

angiography showed retinal ischemia (Figure 1). MRI did not show any pathologic changes (Figure 2). The patient was treated with photocoagulation in her left eye and prednisone 50 mg with slow tapering.

DISCUSSION

In 1986, Hoyt proposed the eponym "Susac's syndrome" in reference to the original description by Dr. Susac in 1979 of two women with encephalopathy, deafness, and retinal arteriolar branch occlusions^(1,2). Susac's syndrome typically follows one of three major courses: a monophasic, polycyclic, or chronic continuous course. In the majority of patients, the disease follows a self-limiting monophasic course with residual deficits and no recurrences. Some patients have several recurrences followed by variable and unpredictable periods of remission, which may persist for 2 to 4 years. The chronic continuous course shows some variation in the severity of symptoms, with no definitive remission^(2,7).

To date, only three cases of clinical recurrence after more than 10 years of remission have been reported. All of them had the classic triad of brain, cochlear,

and retinal effects at onset. In the case presented by Robles-Cedeño et al., the syndrome recurred with gait impairment and worsened visual acuity due to new activity of the brain lesions after 12 years of remission⁽⁸⁾. Petty et al. reported a patient with encephalopathy, ataxia, tinnitus, and hearing loss 18 years after her first symptom⁽⁹⁾. Feresiadou et al. reported a case of Susac's syndrome with a previous clinical outburst 23 years before. The retroactive character of this case impeded the complete ophthalmologic study that could have demonstrated arterial occlusions during the first presentation⁽¹⁰⁾.

We report a 10-year prospective follow-up of a patient diagnosed with Susac's syndrome. After 10 years, there has been only one retinal ischemic relapse, with no neurologic or hearing damage. The absence of neurologic signs and symptoms during follow-up, in addition to the unremarkable findings on MRI, lumbar puncture, and blood tests, excluded other neurologic and systemic mimics, such as multiple sclerosis and central nervous system vasculitis. This unusual presentation supports the finding that the polycyclic course of Susac's syndrome may last more than 10 years and may exclusively affect

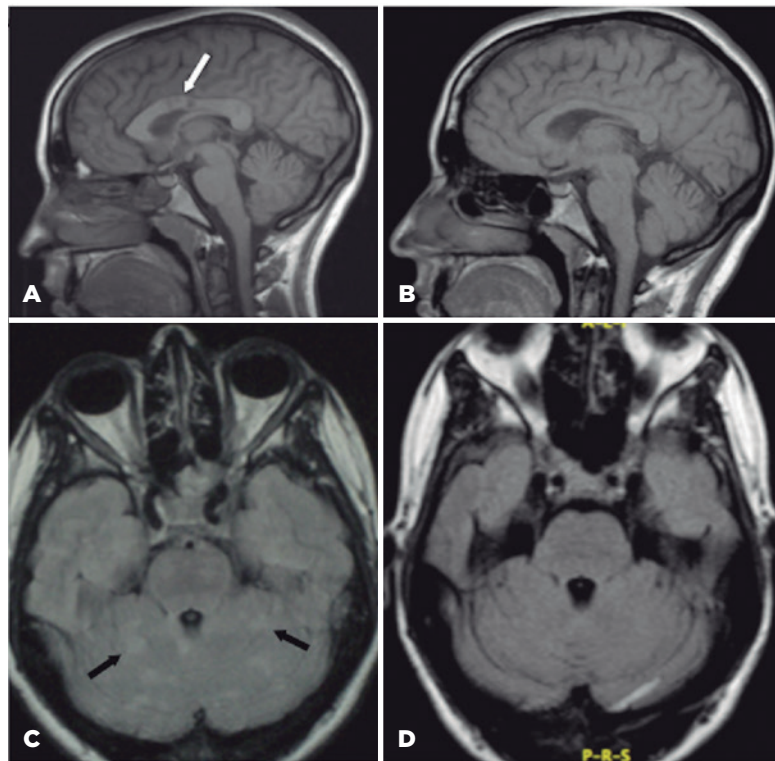


Figure 2. Sagittal and axial T2 FLAIR at onset (A and C) and 10-year follow-up (B and D): black holes indicate central and full-thickness corpus callosum lesions (white arrows) and hyperintense posterior fossa lesions (black arrows).

the retina. Ophthalmologists and neurologists should be aware of this possible course in order to ensure a close follow-up of patients with Susac's syndrome.

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