

## Ocular Findings in Susac Syndrome

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### Abstract

**Purpose:** To describe retinal changes in Susac Syndrome (SS), report its evolution and the importance of follow-up in these patients.

**Setting:** The patient was being followed in the Ophthalmology Department of Irmandade Santa Casa de Misericórdia de São Paulo (ISCMSP) - São Paulo, Brazil.

**Methods:** Case report of a patient with SS and evaluation of alterations observed in ophthalmological, neurological and otological examinations.

**Discussion:** SS lesions can affect the vessels of the retina, brain and inner ear with artery occlusions. The patient in this case had mild changes in the structures that make up the triad of SS.

**Conclusion:** In order to avoid possible relapses of the disease and to maintain a good prognosis, it is necessary to observe regular follow-up and initiation of early treatment to prevent damages that compromise such structures and may cause irreversible deficits to the patient.

**Keywords:** Susac Syndrome, Branch retinal artery occlusion.

### Introduction

Susac Syndrome (SS) is a rare occlusive microangiopathy of the brain, inner ear, and retina [1-3]. The characteristic clinical triad, described by Susac, *et al.* in 1979, comprises encephalopathic or focal central nervous system (CNS) dysfunction, sensorineural hearing disturbances, and branch retinal artery occlusion (BRAO) [3,4].

It may affect more women than men (3:1), usually between the ages of 20-60 years [5]. The etiology is unknown, but they suspect it may be caused by an autoimmune reaction against the affected vascular structures due to recent detection of antiendothelial antibodies. Other hypotheses are that a hypercoagulable state or some previous viral infection may trigger an inflammatory process that could lead to the mentioned changes [6-8].

The most common form of ocular manifestation in SS is BRAO that may be recurrent, extensive or subtle; unilateral or bilateral. The encephalopathy can be manifested by headache, motor deficiencies, sensory deficiencies, aphasia and cognitive impairment caused by small strokes in the brain, involvement of the corpus callosum and multifocal supratentorial lesions. The low-frequency hearing loss

is usually bilateral, and can be associated with tinnitus and vertigo [8,9].

SS is often misdiagnosed or underdiagnosed because it has an extensive list of differential diagnoses such as multiple sclerosis (MS), acute disseminated encephalomyelitis (ADEM), vasculitis of the CNS, infectious encephalitis, cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy (CADASIL), Creutzfeldt Jakob disease, stroke, malignant tumors, migraine with aura, psychosis, or myopathy, encephalopathy, lactate acidosis, mitochondrial encephalomyopathy, lactic acidosis, and stroke-like episodes (MELAS), primary systemic vasculitis (Behçet's disease, Wegener's granulomatosis, Churg-Strauss syndrome, polyarteritis nodosa and Takayasu's arteritis), secondary vasculitis (systemic lupus erythematosus, sarcoidosis, Sjögren's syndrome, cryoglobulinemia), vasculitis related to infectious conditions (syphilis, Lyme disease, tuberculosis, herpes zoster, toxoplasmosis), arteriosclerosis, embolism, thrombophilic diseases, Menière's disease, Cogan syndrome, use of drugs and mitochondrial diseases [10-12].

There is currently no recommended treatment for SS. The proposed treatments are still controversial and depend on the clinical condition of the patient in question and the progression of the disease [13]. Most of the clinical cases previously reported

recommend the use of steroids in every patient, starting with a pulse of highly dosed methylprednisolone (1g/day for 5 days), followed by an oral dose of prednisone 1 mg/kg daily with a slow subsequent tapering, depending on the type and course of the patient's disease. The patients can use aspirin to reduce procoagulopathic states and immunosuppressive drugs can be introduced if necessary [14,15].

The prognosis depends on the evolution of the disease. Frequent ophthalmic and complementary examinations are important to follow the patient and prevent new relapses.

### Purpose

To describe retinal changes and report the importance of regular monitoring in a patient with Susac Syndrome.

### Case report

Case report of a 28 year old male Caucasian, who presented dizziness and pain in both eyes (OU) for 2 months. Associated with these symptoms was tinnitus and headache. He was evaluated in the Retina and Vitreous Department of Irmandade Santa Casa de Misericórdia de São Paulo – ISCMSP in October 2016. The best corrected visual acuity (BCVA) was 20/20 in OU. At the slit-lamp examination he had no chamber reaction or other significant changes OU. Intraocular pressure (IOP) was 14 mmHg OU. The funduscopy showed inferonasal BRAO and suffering from the layer of nerve fibers in the lower arch in left eye (OS), better observed during fluorescein angiography (figure 1).

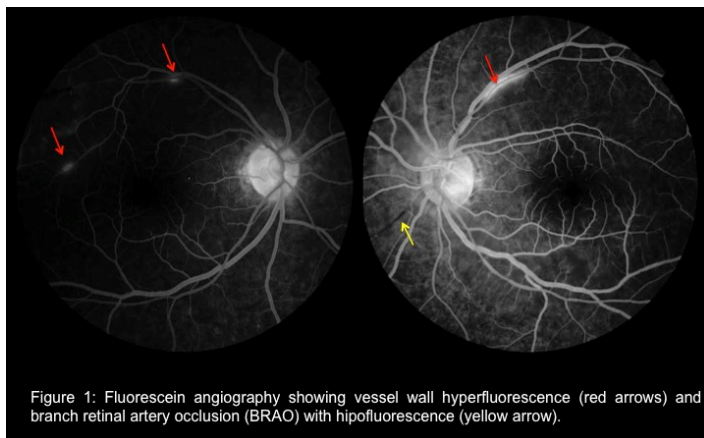


Figure 1: Fluorescein angiography showing vessel wall hyperfluorescence (red arrows) and branch retinal artery occlusion (BRAO) with hypofluorescence (yellow arrow).

Retinography, fundus autofluorescence (FAF), fluorescein angiography (FA), optical coherence tomography (OCT) and visual field were performed for documentation and follow-up of the case. Several laboratory tests were requested to exclude infectious causes, hematological disorders and other inflammatory causes. According to clinical characteristics and ophthalmic examination, an SS diagnosis was suspected and the patient was referred to the neurology and otorhinolaryngology departments for investigation. They performed audiometry, puncture of cerebrospinal fluid, Doppler ultrasound and brain magnetic resonance imaging (MRI). The patient was periodically evaluated every week and it was decided to introduce oral corticosteroid (prednisone 1mg/kg/day) in slow regression. He evolved with significant improvement of the initial funduscopy aspect and is now waiting for the result of the laboratory tests for the introduction of the immunosuppressive therapy with azathioprine.

### Discussion

As SS is a diagnosis of exclusion, the detection is sometimes made considerably late. SS can cause serious damage to the brain, eye and ear. Early diagnosis and detection of the disease stage is therefore extremely important in order to avoid preventable sequelae.

There are studies that suggest that involvement of the brain, retina and cochlea occur in SS is due to their similar embryological origin. The retina and the inner ear present barriers similar to the blood-brain barrier, whose endothelium may have common structural and functional properties [16-18]. The patient in this case did not represent a typical course of the disease, but had triad factors that fit in SS.

From the beginning of this patient's follow up until the completion of the complementary tests and evaluation with the other specialties, he evolved with a slight worsening in funduscopy in the right eye (OD) with superotemporal BRAO and superficial microhemorrhages in the periphery (figure 2). In OS, there was improvement of the inferonasal BRAO and the areas of suffering of the layer of nerve fibers. In OS, there was improvement of the inferonasal BRAO and the areas of suffering of the layer of nerve fibers.

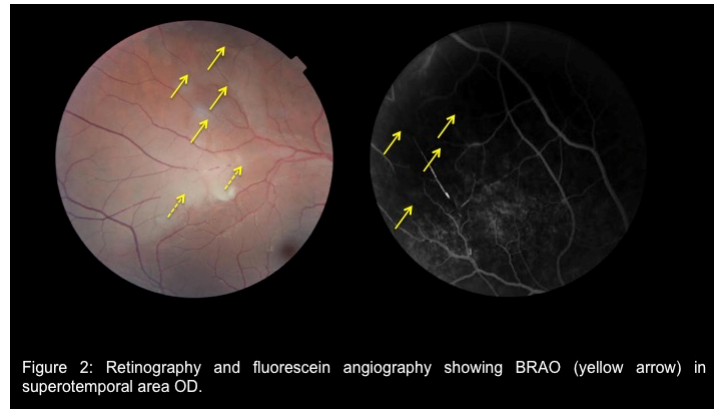


Figure 2: Retinography and fluorescein angiography showing BRAO (yellow arrow) in superotemporal area OD.

A complete neurological investigation was performed and the cerebrospinal fluid revealed a slight pleocytosis and elevated protein but no oligoclonal bands. At the MRI he had small callosal commissure with atrophy and periventricular "snowball"-like lesions (figure 3). Doppler ultrasound was normal.

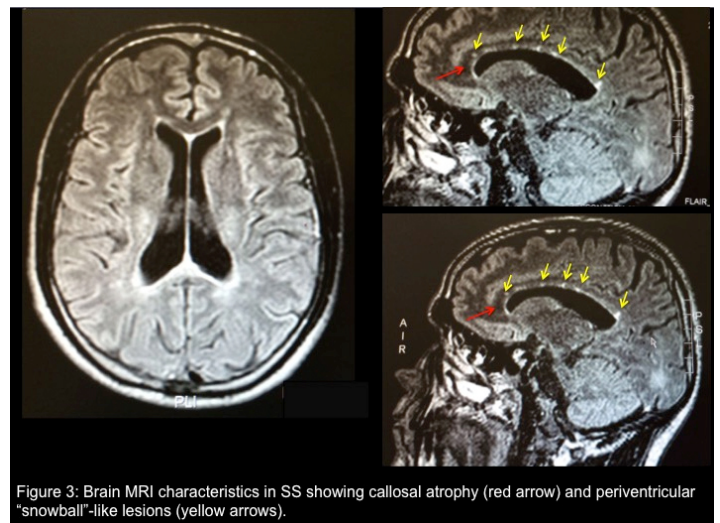


Figure 3: Brain MRI characteristics in SS showing callosal atrophy (red arrow) and periventricular "snowball"-like lesions (yellow arrows).

The otorhinolaryngologists evaluation showed none of the typical alterations in the audiometry of SS patients, since the patient only had a slight decrease in the high frequencies (acute).

He had no changes in OCT, FAF and visual field exams OU. The FA showed contrast block and hypofluorescence in areas of the occlusion of a retinal artery in superotemporal region OD and hyperfluorescence with leakage of the vessel wall in upper arch OU.

Three months after starting treatment with oral corticosteroids the patient evolved with improvement of the fundoscopic aspect and the areas of BRAO and maintained good visual acuity (figures 4 and 5).

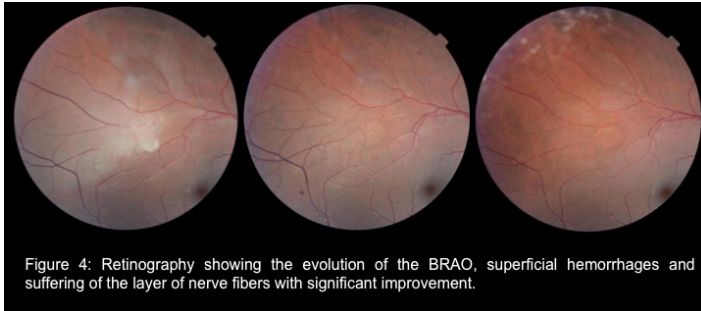


Figure 4: Retinography showing the evolution of the BRAO, superficial hemorrhages and suffering of the layer of nerve fibers with significant improvement.

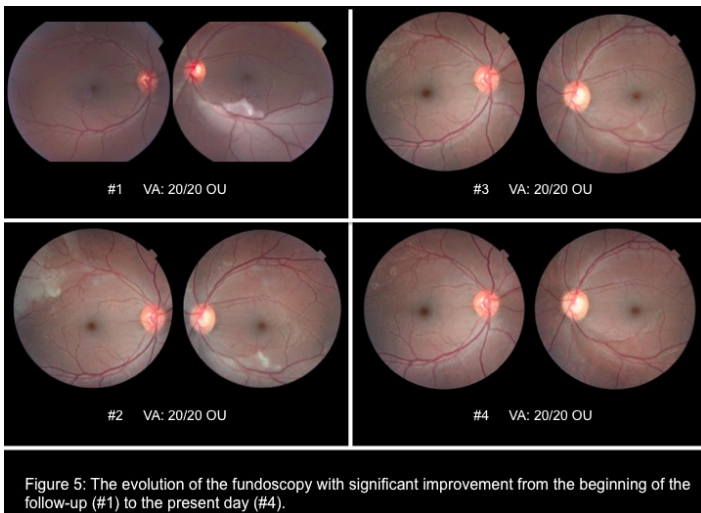


Figure 5: The evolution of the funduscopy with significant improvement from the beginning of the follow-up (#1) to the present day (#4).

## Conclusion

The course of the SS is impossible to predict and the patient may present recurrences through the follow-up. Therefore, periodic monitoring of these patients is extremely important in order to avoid possible damages resulting from the disease.

In this specific case, the patient had a good evolution because the diagnosis was promptly made. The other extraocular manifestations were in the initial phase with a good prognosis after prompt treatment.

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