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Multimodal imaging in Susac syndrome with classic clinical triad presentation

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Abstract:

A 22-year-old male was referred for headaches, hearing impairment, and right eye scotoma. Branch retinal artery occlusion was revealed during the ophthalmological examination. Susac syndrome was suspected due to the symptoms described and the absence of cardiovascular risk factors. An extensive ophthalmological examination, including multimodal imaging was carried out, which is of special interest as it is considered to be a rare syndrome.

Keywords:

Branch retinal artery occlusion, multimodal imaging, Susac syndrome

Introduction

Susac syndrome is a rare immune-mediated microangiopathy with a prevalence of 0.14 cases/100,000 people^[1] that affects the endothelium of precapillary arterioles of the brain, retina, and inner ear causing ischemia.^[2] However, the clinical triad of encephalopathy, branch retinal artery occlusion and hearing loss appears in a minority of cases at the time of diagnosis, 13% showing most of the patients one or two symptoms, although almost every patient will develop the full triad overtime.^[1,2] Subtypes of Susac syndrome with neurological or ophthalmological manifestations alone have also been described with either a recurrent branch retinal artery occlusion form or encephalopathic form.^[2]

Neurological symptoms include migrainous headaches, cranial neuropathies, and cognitive deficits. Differential diagnosis with multiple sclerosis must be taken into account.^[3] Magnetic resonance imaging (MRI) typically reveals corpus callosum involvement shown as spoke-like

and snowball lesions. Increased protein levels and lymphocytic pleocytosis may be found in the cerebrospinal fluid.^[2]

Neurosensory hearing loss secondary to cochlear infarcts, as well as vertigo and tinnitus, is the most common otorhinolaryngological symptoms.^[2]

Different signs can be found during ophthalmological examination in Susac syndrome. The main ophthalmological symptom referred by patients is a visual field defect which is caused by branch retinal artery occlusion. Thickening and hyperreflectivity of inner retinal layers during the acute phase and atrophy of inner layers during the chronic phase can be appreciated in optical coherence tomography (OCT) imaging, as in any other retinal artery occlusions.^[4] OCT-A imaging is also a helpful tool to determine the extent of ischemia in Susac syndrome, normally showing an unaffected choriocapillaris vasculature.^[5] Arterio-arterial collaterals secondary to ischemic processes^[6] and recurrent branch retinal artery occlusions^[2] have been described overtime.

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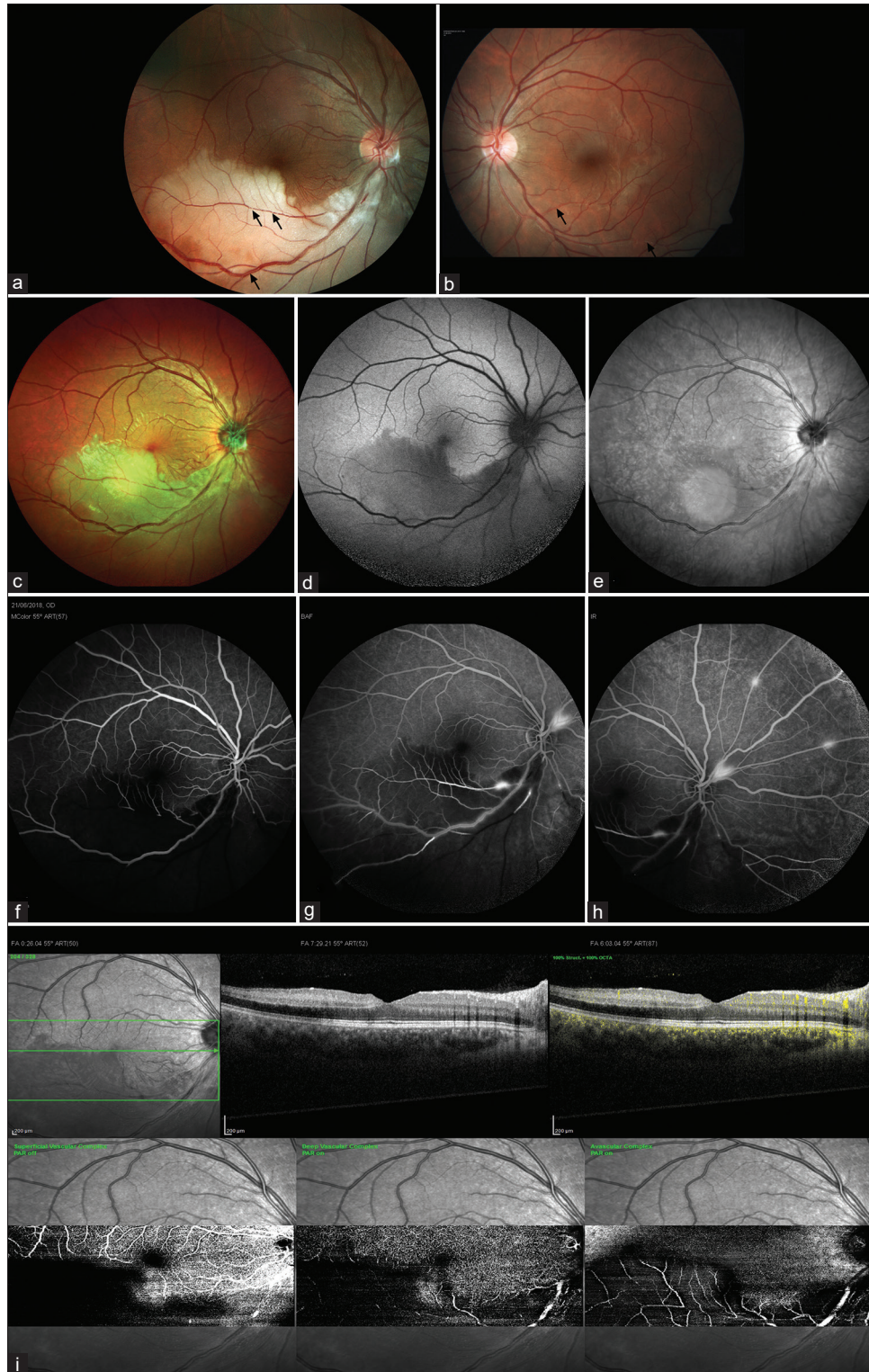


Figure 1: (a) Right eye fundus: inferior temporal branch occlusion associated with retinal ischemic whitening and multiple Gass plaques (black arrows). (b) Left eye fundus: Gass plaques with no signs of ischemia (black arrows). (c) Right eye multicolor image. (d) Right eye autofluorescence: inferior temporal hypofluorescence. (e) Right eye infrared reflectance image. (f-h) Right eye fluorescein angiography: hypofluorescence of the ischemic area and patched arterial wall hyperfluorescence. (i) Right eye optical coherence tomography: hyperreflective inner retinal layers in structural optical coherence tomography and decreased vascular perfusion in optical coherence tomography-A

Gass plaques can be seen in fundus examination as yellowish deposits in the arterioles and represent areas of endothelium disruption.^[7] Unlike embolisms,

they can be found not only in arteriolar bifurcations but also between them, and not only limited to the ischemic area.^[6] These plaques are demonstrated

using fluorescein angiography as arteriolar wall hyperfluorescence.^[2]

Optic disc angiopathy has also been described associated with Susac syndrome in a minority of cases.^[8]

Case Report

A 22-year-old Caucasian male presented with a 2-month history of headaches, left hearing impairment, and a superior scotoma in his right eye. His past medical history was unremarkable.

Visual acuity was 1.0 in both eyes using Snellen's visual acuity chart. Anterior segment slit-lamp examination showed no pathological signs, and intraocular pressure was not altered. Fundus examination revealed multiple yellowish refractile lesions distributed along retinal arterioles, which corresponded to Gass plaques [Figure 1a and b]. Inferior temporal retinal whitening corresponding to branch retinal artery occlusion was present in his right eye. A small flame-shaped hemorrhage was observed within the ischemic area [Figure 1a].

The clinical triad of neurological symptoms, hearing loss, and branch retinal artery occlusion was highly suggestive of Susac syndrome, so the extensive examination was carried out. Multimodal imaging including multicolour images and autofluorescence [Figure 1c-e] revealed retinal pigment epithelium alterations in the ischaemic area. Fluorescein angiography showed diffuse areas of hypofluorescence caused by arteriolar occlusions in the inferior temporal retina of his right eye, and patched arterial wall hyperfluorescence was seen in both eyes, being, therefore, findings consistent with our suspected diagnosis [Figure 1f-h]. Right eye structural OCT imaging showed hyperreflective inner retinal layers

in the ischemic area and OCT-A imaging revealed decreased vascular perfusion with no-flow areas in superficial and deep vascular complexes corresponding to the affected area [Figure 1i].

Underlying causes of retinal artery occlusion, which needed urgent intervention, were studied. Blood pressure was 123/78 mm Hg; electrocardiography and carotid Doppler ultrasound did not show pathological findings. Extensive laboratory workup including complete blood count, D-dimer, antinuclear antibody, prothrombin time/partial thromboplastin time, antithrombin III activity, and factor V Leiden were reported as normal.

The further medical examination was carried out by the Otorhinolaryngology and Neurology departments. Audiometry revealed left neurosensory hypoacusis [Figure 2], and MRI imaging showed white matter disturbances affecting corpus callosum compatible with Susac syndrome [Figure 3].

The patient was referred to the neurology department and was initially treated with oral prednisolone, acetylsalicylic acid, and flunarizine. Two months after the diagnosis, he started receiving rituximab and intravenous immunoglobulins. No ophthalmological treatments were implemented.

The patient has not presented further ophthalmological, neurological, or otorhinolaryngological manifestations. The visual field examination remains stable, and a permanent superior defect in his right eye persists.

Discussion

Definite Susac syndrome diagnosis has to include neurological symptoms and alterations in the corpus

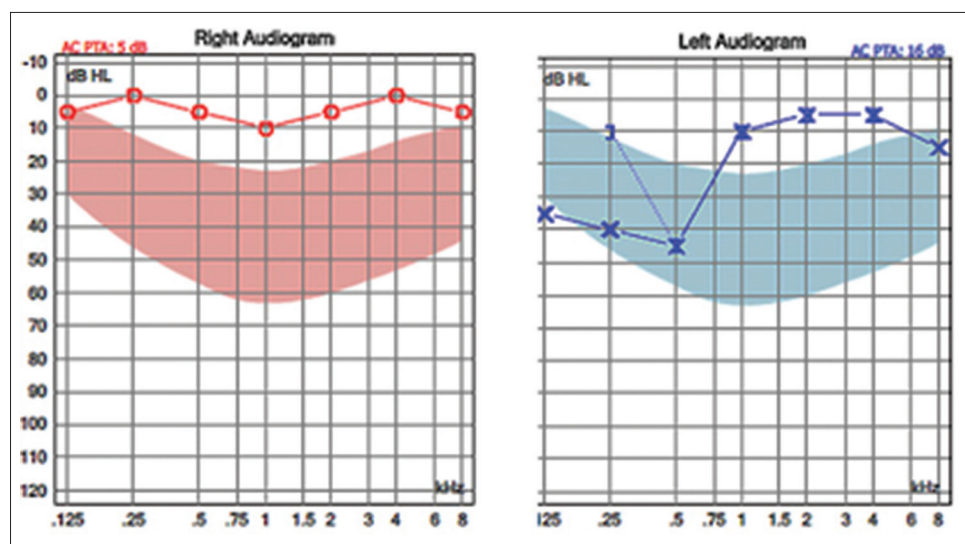


Figure 2: Audiogram showing neurosensory left hypoacusis

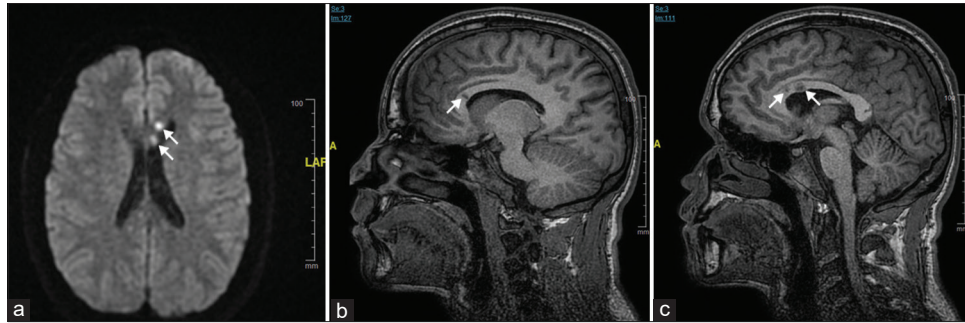


Figure 3: Magnetic resonance imaging showing snowball-like lesions in the corpus callosum (white arrows). (a) T2. (b and c) T1

callosum seen in neuroimaging; branch retinal artery occlusion or arterial wall hyperfluorescence in fluorescein angiography; and vestibulocochlear symptoms supported by an audiogram.^[9] Our patient meets all of the required criteria to establish a certain diagnosis.

This case is of special interest as the three classical clinical manifestations were found at the time of diagnosis, which added to its low prevalence, makes it infrequent in the medical literature. It is vital for ophthalmologists to be aware of this triad and carefully evaluate patients with branch retinal artery occlusions, especially when there is the absence of cardiovascular risk factors, as early diagnosis will allow appropriate treatment to be established as soon as possible.

In addition, an in-depth ophthalmological examination was key to the diagnosis, and all classical findings (Gass plaques, branch retinal artery occlusion, and arteriolar wall hyperfluorescence) are shown, not only using classical ophthalmological examination techniques (fundus photography and fluorescein angiography) but also using multimodal imaging and OCT-A. This is particularly relevant as these new imaging techniques could substitute fluorescein angiography as a main diagnostic and prognostic test in many vascular pathologies and more cases are needed to better understand the extent to which these imaging modalities can give us valuable information in a noninvasive way.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient has given his consent for his images and other clinical information to be reported in the journal. The patient understands that his name and initial will not be published and

due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Nil.

Conflicts of interest

The authors declare that there are no conflicts of interest in this article.

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