

SUSAC'S SYNDROME: A CASE REPORT

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Abstract

Susac's syndrome (SS) is a common infarct arteriolar occlusive disease. It consists of a triad of symptoms: encephalopathy, branch retinal artery occlusions (BRAO), and hearing loss. It usually affects women aged 20 to 40. The precise cause is not known, but the symptoms usually include hearing loss and vision loss.

We report a case of SS in a 52-year-old woman. She initially was admitted to the hospital with signs of loss of consciousness and hearing. An audiogram taken for the patient showed bilateral severe sensorineural hearing loss which affected all frequencies (right 82/80; left 110/78 dB). Speech reception thresholds (SDS) of 65 dB to 70 dB on the left to the right were observed and speech scores could not be obtained. A tympanogram of Curve Type A was obtained. The patient experienced sudden sensorineural hearing loss (SSHL) and treatment of SSHL was started immediately. Patients of long-duration stays at the clinic often experienced decreased vision and reduced coordination. Cranial magnetic resonance imaging (MRI), Axial T2-FLAIR images showed multiple hyperintense lesions involving cerebral white matter, and left internal-external capsule; sagittal T2W images showed hyperintense lesions involving periventricular cerebral white matter. No significant changes in hearing, but improvements in the patient's coordination and vision, were observed at a six-month follow up. The patient was given hearing aids at the follow up as well.

For all those patients with sudden hearing loss, confusion, and blurred vision, retinocochleocerebral disease that Susac's syndrome should be considered. Funduscopic eye examination, audiometry, and MRI are the first steps in diagnosis.

Case report (J Int Dent Med Res 2013; 6: (3), pp. 136-139)

Keywords: Susac's syndrome, branch retinal artery occlusions, retinocochleocerebral Vasculopathy.

Received date: 15 July 2013

Accept date: 20 October 2013

Introduction

Susac's syndrome (SS) is a common infarct arteriolar occlusive disease. It consists of a triad of symptoms: encephalopathy, branch retinal artery occlusions (BRAO), and hearing loss. It usually affects women aged 20 to 40, but men are also affected, and the overall age range

can extend from 9 to 72 years. The precise cause is not known, but the symptoms usually include hearing loss and vision loss. Detection of these symptoms that characterize the disease is a cause for investigation. Audiogram bilateral sensorineural hearing loss is especially prominent at low frequencies, and bilateral branch retinal artery occlusion in the distal region in funduscopy and fluorescein angiography, seen in white and gray matter, in the small, multi-focal increase in density in T2-weighted images of MR scan are also common.

Immunomodulators to antithrombotic drugs have been tested as treatments, but there is no definitive regime. After a period of activity following the course of the self-limiting disease,

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patients with signs of blindness, deafness, and dementia have returned to normal life in the late period¹⁻⁴.

Case Report

Here, we report a case of SS in a 52-year-old woman. She initially was admitted to the hospital with signs of loss of consciousness and hearing. She did not describe any additional systemic disease, except thalassemia major, in her history. Otoscope examination of the patient's eardrum and a standard ear, nose, and throat examination showed all normal. An audiogram taken for the patient showed bilateral severe sensorineural hearing loss which affected all frequencies (right 82/80; left 110/78 dB) (Figure.1).

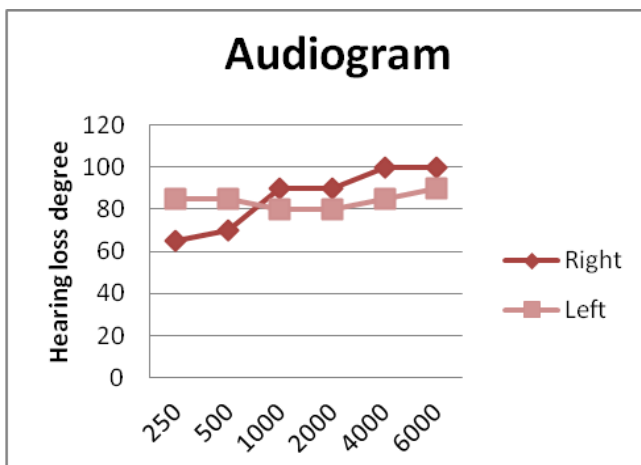


Figure 1. Audiogram shows sensorineural hearing loss.

Speech reception thresholds (SDS) of 65 dB to 70 dB on the left to the right were observed and speech scores could not be obtained. A tympanogram of Curve Type A was obtained. The patient experienced sudden sensorineural hearing loss (SSHL) and treatment of SSHL was started immediately. Patients of long-duration stays at the clinic often experienced decreased vision and reduced coordination.

Acute or subacute encephalopathy often manifests itself as a headache, impaired cognition and memory, ataxia, dysarthria, vertigo, or corticospinal tract dysfunction. Psychiatric features include personality changes and strange behavior, which also appeared. Ophthalmic and neurological consultations were requested. Cranial magnetic resonance imaging (MRI), Axial

T2-FLAIR images showed multiple hyperintense lesions involving cerebral white matter, and left internal-external capsule (Figure 2); sagittal T2W images showed hyperintense lesions involving periventricular cerebral white matter (Figure 3).

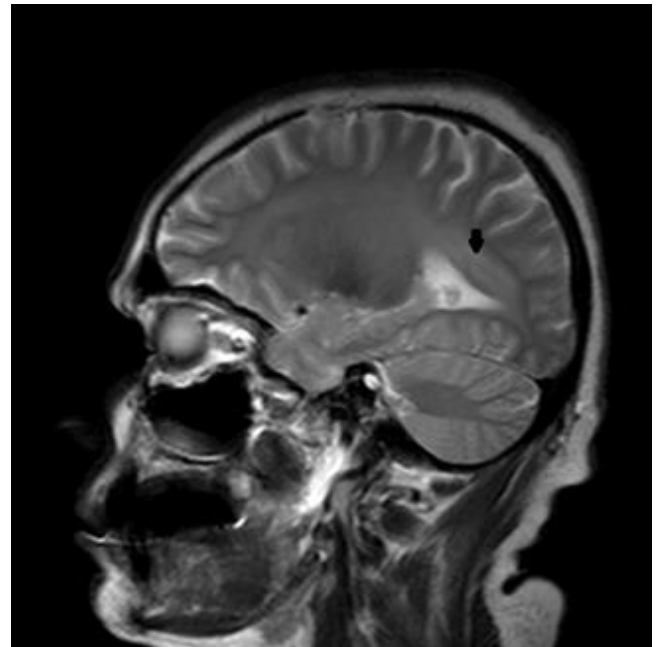


Figure 2. Axial T2-FLAIR images shows multiple hyperintense lesions involving cerebral white matter (black arrows), and left internal-external capsule (arrows).

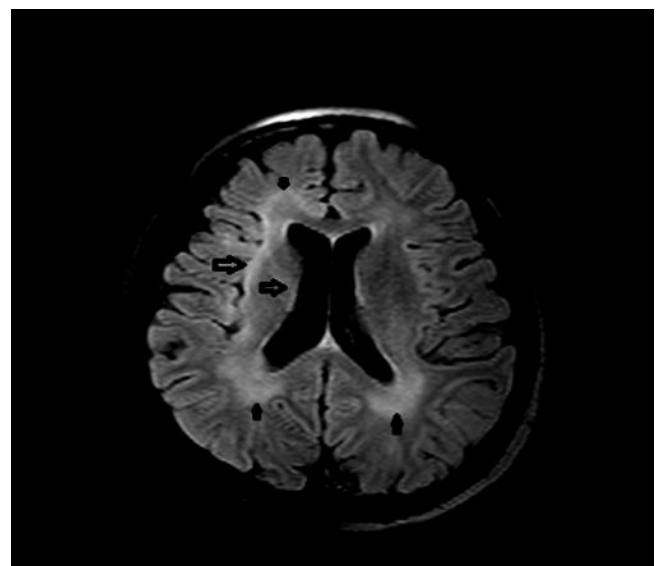


Figure 3. Sagittal T2W images shows hyperintense lesions involving periventricular cerebral white matter (arrow).

Slow wave activity was observed in the EEG (Alpha rhythm). Prominent vascular

abnormalities were not found in the CT angiography. Both blood levels of antithrombin (56.2 %) and hemoglobin (7.82 Ü) were low. The Von Willebrand factor was high (298.2%). The antinuclear antibody (ANA) and other immunologic tests were negative.

There was observed discontinuity in the left temporal retinal arteriolar vascular when a funduscopy eye examination was performed. The council of neurology and ophthalmology established, as a result of correct diagnosis based on the above triad of clinical symptoms in conjunction with the MRI and fundoscopic findings (Figure.4), that Susac's syndrome was present and followed this diagnosis with medical treatment. When SS is diagnosed, the standard therapy includes intravenous steroid.

No significant changes in hearing, but improvements in the patient's coordination and vision, were observed at a six-month follow up. The patient was given hearing aids at the follow up as well.

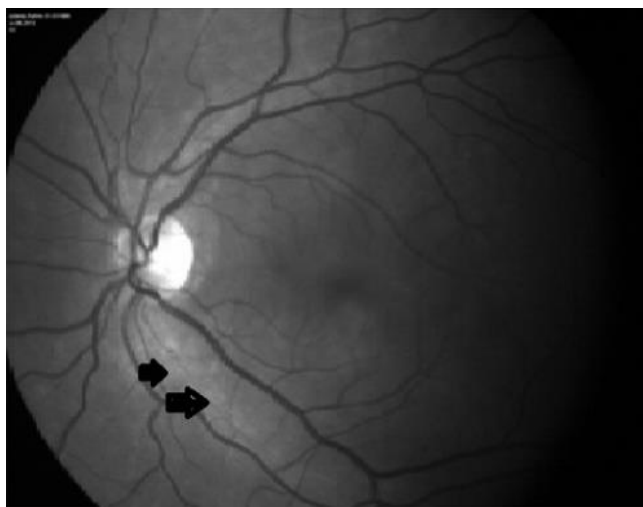


Figure 4. Funduscopy shows discontinuity in the left temporal vascular retinal arteriolar.

Discussion

As with all diseases, diagnosis is achieved via an examination of medical history, physical examinations, and diagnostic laboratory and radiological studies combined together, as with a puzzle. Because SS is considered a rare disorder, the diagnosis may be more difficult. SS is a rare but important differential diagnosis in numerous neurological, psychiatric, ophthalmological, and ear, nose and throat disorders³.

In SS, occlusions of microvessels, presumed to be mediated by an autoimmune response to an as-yet unknown antigen, lead to a characteristic clinical triad of central nervous system dysfunction, branch retinal artery occlusions, and sensorineural hearing impairment. Improved understanding of this disorder is crucial, therefore, to ensure that patients receive appropriate treatment and care. Most existing studies on SS are based on reports of single patients and small case studies. Dörr et al.³ completed a review, the goal of which was to extend these previous, primarily anecdotal findings by collecting data from all 304 cases of Susac's syndrome that have been published worldwide. Although a detailed pathomechanism remains to be elucidated, SS is associated with a microangiopathy of the brain, retina, and cochlea^{2,3,4}.

The associated hearing loss is caused by cochlear impairment and has been attributed to infarction of the cochlear apex in the vascular region of small end arteries^{7,8}. Audiometry reveals sensorineural hearing loss that is usually in the low- to mid-frequency range. Audiometry reports (when documented) revealed sensorineural hearing loss, more commonly bilateral than unilateral, in almost every patient³. Although hearing loss is included in the triad of SS, in fact, there are no details reporting types or analyses of hearing in the existing reviews of SS³ or other case reports. A flat-type audiogram is available in the case of the 52-year-old woman whose hearing loss affected all frequencies. Although the hearing test was repeated four times in one year, no changes in audiometry were identified. The patient's hearing loss was found to be irreversible. These results may be associated with the patient's advanced age and with the thalassemia major present as a co-morbid.

Audiometry procedures should be performed in every case as early as possible during the course of the disease, as even normal findings can provide important information for the evaluation of subtle changes in these measures during follow up.

The associated retinopathy is characterized by multiple peripheral retinal arteriolar branch occlusions that can be seen during ophthalmoscopic examination or retinal fluorescein angiography. In this case, retinal fluorescein angiography wasn't performed after recognition of the brain MRI findings because

informed consent from the patient could not be obtained. But discontinuity was observed in the left temporal retinal arteriolar vascular during a fundusoscopic eye examination (Figure 3). Visual acuity may normal or reduced, and retinal fluorescein angiography may observed as normal, even when branch retinal artery occlusion and arterial wall hyperfluorescen_e exist.

There were multiple hyperintense lesions involving cerebral white matter, and left internal-external capsule and hyperintense lesions involving periventricular cerebral white matter in the MRI images. Observed signal changes with the thinning along corpus callosum were also present. The most striking MRI finding in patients with SS is that the characteristic callosal lesions, the centrum semiovale, internal capsule, periventricular white matter, brainstem, cerebellum, cerebral and cerebellar peduncles, basal ganglia, and thalamus may all be involved^{6,7,7,9,10}.

Based on the findings described above (the presence of SSHL, reduction of visual acuity, and encephalopathy related to psychotic disorders), SS was diagnosed by a multidisciplinary team.

Both the cause and the pathogenesis of this microangiopathy remain unclear. High Von Willebrand levels and low level of antithrombin do not support the clotting factors of pathogenesis. Elevation of a blood-clotting protein, factor VIII, has been suggested as a finding of potential pathophysiological relevance in Susac's syndrome¹⁰. The present patient had a negative value for antinuclear antibody (ANA), while a mild elevation of ANA titre was evident in 22 of the 304 patients included in the Dörr et al. study³. However, the simple detection of elevated ANA alone is not sufficient for the diagnosis of Susac's syndrome¹².

In patients with suspected Susac's syndrome, diagnostic procedures in addition to those described above are often performed, including comprehensive screening for autoantibodies and clotting abnormalities, CSF analysis, EEG, cerebral catheter angiography (CAA), and leptomeningeal biopsy. Whether such tests can provide clinically relevant information remains unclear. Despite a considerable number of patients being subjected to invasive diagnostic procedures, such as brain biopsy or cerebral angiography, in many cases these procedures did not aid the diagnosis³.

Therefore, the appearance of the full triad of symptoms is important for diagnosis.

Conclusions

For all those patients with sudden hearing loss, confusion, and blurred vision, retinokohleoserebral Susac's syndrome should be considered. Fundusoscopic eye examination, audiometry, and MRI are the first steps in diagnosis.

Declaration of Interest

The authors report no conflict of interest and the article is not funded or supported by any research grant.

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