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Case Report

Susac syndrome: A rare presentation

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ABSTRACT

Susac syndrome is an autoimmune microangiopathy characterized by a triad of encephalopathy, sensorineural hearing loss and varying degrees of branch retinal artery occlusion. A young male patient is described with features of focal neurological lesions and small superficial retinal haemorrhages and areas of retinal arteriolar endothelial inflammation appearing as Gass plaques. Susac syndrome presents with varying features involving brain, inner ear and retina. Fundus Fluorescein angiography should be performed in all patients with an unexplained encephalopathy to look for characteristic arteriolar wall changes.

1. Introduction

Susac syndrome, which was originally described by J.O. Susac in 1979 is a rare occlusive micro-angiopathy of the brain, inner ear and retina. The characteristic clinical triad which includes encephalopathic or focal CNS disturbances, varying degrees of sensori-neural hearing loss and partial or complete Branch retinal artery occlusion(BRAO).

The specific symptoms, severity and outcome of Susac syndrome vary from one person to another. The clinical triad of encephalopathy, hearing loss and BRAO are not always present at the time of onset and all three do not necessarily develop in all cases. Susac syndrome primarily affects young women around the age of 20-40 yrs. Women are affected three times more often than men. [1]

The exact etiology of susac syndrome is unknown but recent detection of anti-endothelial antibodies suggest the autoimmune nature of the disease. [2] Other suggested hypotheses include a hypercoagulable state or some viral infection as a trigger for the inflammatory process.

2. Case Report

A 40-year old male patient was referred to us from the Department of neurology for ophthalmic examination. The patient was conscious, well oriented but gives history of forgetfulness, slurred speech and occasionally disturbance in gait. Patient's family members also complained about the fluctuations regarding mental status of the patient, particularly having spells of agitated behavior. There was no disturbance in hearing.

The patient as such had no visual complaints. His visual acuity was 6/6 both eyes and Slit lamp biomicroscopic examination didn't reveal any anterior segment findings. His Intra Ocular Pressure (IOP) was also normal, 16mmHg both eyes.

On fundus examination few superficial hemorrhages were seen superior to macula in right eye (OD). There were multiple small grey-yellow lesions temporal to macula in both eyes suggestive of Gass plaques.

Fundus photograph of right eye (Figure 1) showing few superficial hemorrhages(Red Arrow) and Gass plaques (Black arrow) temporal to macula ; (Figure 2) showing Gass plaques temporal to macula in left eye (Black arrow).

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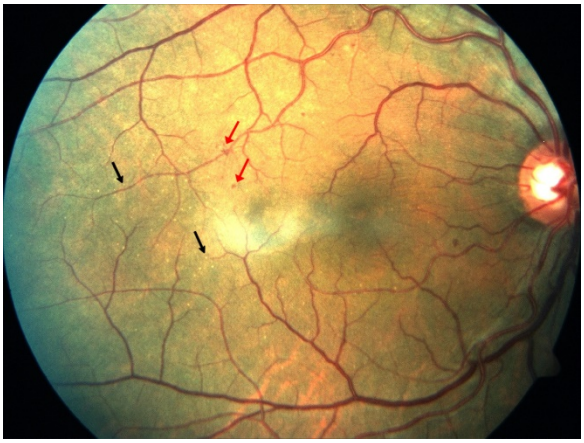
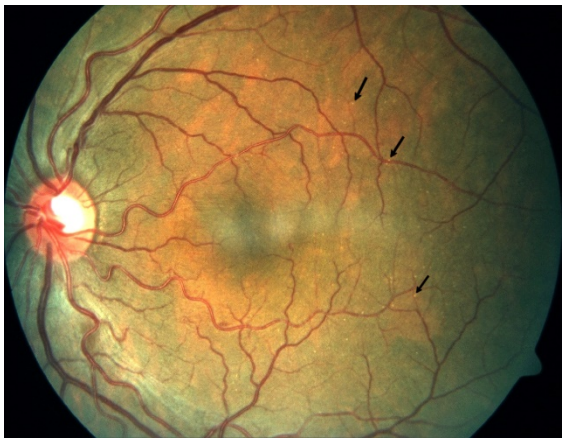


Fig. 1:



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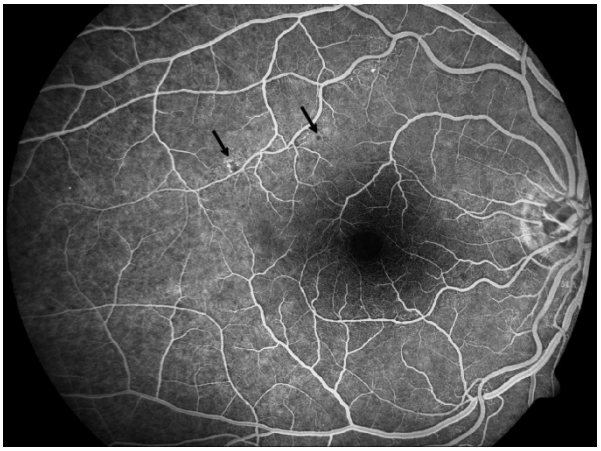

Fig. 3:

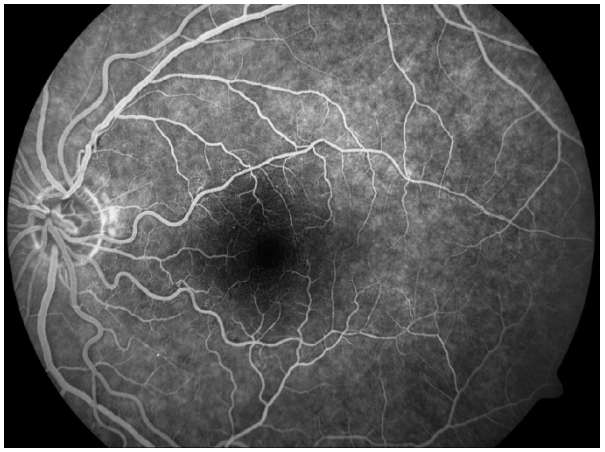

Fig. 4:

Fundus fluorescein Aangiography (FFA) was done which revealed areas of blocked fluorescence (Black arrow) corresponding to areas of superficial hemorrhages in Right eye (Figure 3), normal FFA in left eye (Figure 4).

MRI scans were showing few periventricular snowball like lesions (Red arrow) in corpus callosum suggesting small areas of infarcts (Figure 5).

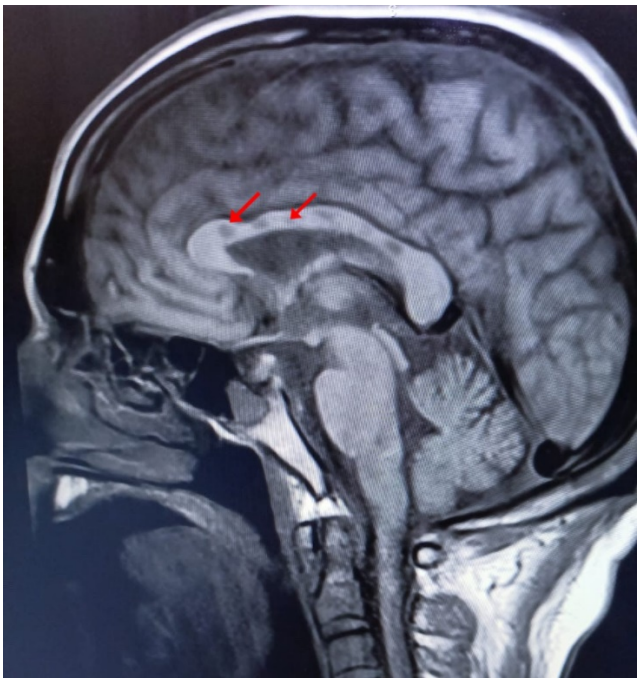

Fig. 5:

Lumbar puncture as done by the neurologists revealed no abnormality.

Keeping in view the neurological symptoms of the patient and the MRI findings along with the presence of few superficial retinal hemorrhages and Gass plaques, a

diagnosis of SUSAC Syndrome was made and the patient was sent back to neurology department for initiation of systemic steroid therapy.

3. Discussion

The diagnosis of Susac syndrome requires a high index of suspicion because a high percentage of patients, about 97% do not follow the clinical triad at the time of onset of symptoms. The complete triad may develop after a delay of weeks to more than 2 years.

Neuropsychiatric disturbances may be seen in 75% cases. Only in 10% cases illness is revealed by hearing/cochlear or ophthalmic symptoms. This condition is often misdiagnosed or under diagnosed because of the features related to other diseases like Multiple sclerosis, Migraine with aura, lupus erythematosus, acute disseminated encephalomyelitis, Meniere's disease, thromboembolic states, lactic acidosis.[3,4]

Studies have suggested that involvement of brain, retina and cochlea in susac syndrome is due to their common embryonic origin. Also that retina and the inner ear have barriers similar to the blood-brain barrier, thus acting as a possible substratum for multiple infarcts in these three regions.[5,6] Besides, complete neurological and ENT workup, a detailed ophthalmological examination is essential for making up the diagnosis of Susac syndrome.

Patients with susac syndrome can present with multiple peripheral retinal arteriolar branch occlusions that can be seen on ophthalmoscopic examination or retinal fluorescein angiography. The occlusions may be quite extensive or may be very subtle. Segmental loss of vision in one or both eyes and visual scintillating scotoma are typical visual complaints. Some patients do not complain of visual deficit, despite the typical arteriolar occlusive process. This may be due to either encephalopathy or the retinal infarct being too small or too peripheral. Occlusions at the posterior pole cause profound visual loss, whereas more peripheral lesions might be associated with no subjective visual symptoms. Fundoscopy may show occlusion of the branches of the central retinal artery; the fundi are sometimes considered normal when the occlusions are confined to the small arterioles in the retinal periphery. Retinal fluorescein angiography is the best method for detecting the retinal arteriolar occlusions. It may also show leakage of dye. Arteriolar wall hyperfluorescence has been noted before the occurrence of retinal arteriolar occlusion. This can be taken as an indicator of active disease and hasten preventive treatment.[7]

Few patients might present with small yellowish refractile dots called as Gass plaques as seen in our patient. These plaques are sites of inflammation in the endothelium and do not have to be associated at the location of BRAO.

4. Conclusion

The diagnosis of susac syndrome should be suspected in cases of unexplained neuropsychiatric symptoms and Ophthalmological examination including fundus fluorescein angiography would help in the diagnosis.

5. Source of Funding

None.

6. Conflict of Interest

None.

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