

## Susac's syndrome: First from India and youngest in the world

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A two and half year old female was admitted at the emergency room suffering from gradually worsening headache followed by nausea. The child presented with reduced level of consciousness and bilateral hypoacusis. The patient was lethargic. Ophthalmic examination showed branch retinal artery occlusion (BRAO). This finding was crucial to the diagnosis of Susac's syndrome (SS), a rare autoimmune disease characterized by, endotheliopathy of retina, encephalic tissues and cochlea. Magnetic resonance imaging of the brain also showed typical features. Thorough blood investigations did not reveal any other abnormality. Patient was treated with immunosuppressive to prevent her from developing severe sequelae of this disease. The child showed dramatic improvement in her systemic condition within 48 h of starting the treatment. This is the youngest ever and first case report from India.

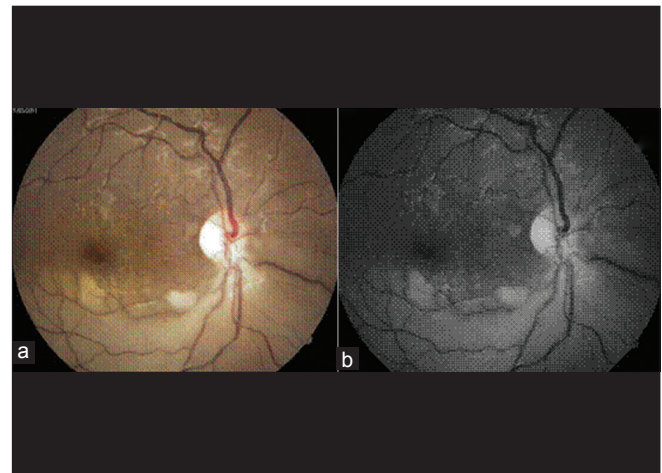
**Key words:** Branch retinal arterial occlusion, encephalopathy, retinal vasculitis, sensorineural hearing loss, susac's syndrome

Susac's syndrome was first described by Susac *et al.*, in 1979.<sup>[1]</sup> It involves a clinical triad that includes encephalopathy, retinal arterial branch occlusions, and bilateral hearing loss.<sup>[2]</sup> It commonly affects women in the 20-40 years age group.<sup>[3,4]</sup> The underlying process is believed to be an immunological reaction, leading to small vessel vasculitis,<sup>[5]</sup> causing microinfarcts in the retina, brain and the apical turn of the cochlea. Early recognition of this syndrome is important because treatment with immunosuppression may minimize permanent cognitive, audiological and visual sequelae. This syndrome has a good prognosis when treated early.

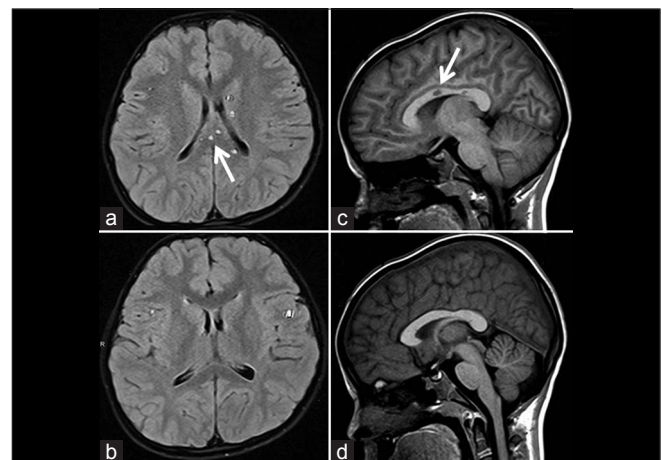
### Case Report

A two and half year old female was admitted at the emergency room suffering from gradually worsening headache followed by nausea. The child was lethargic, and had reduced level of consciousness with hypoacusis. Laboratory studies were

negative for antinuclear antibodies, hepatitis, Epstein-Barr polymerase chain reaction, cardiolipin antibodies, and herpes infection. Cerebro Spinal Fluid (CSF) analysis was negative for oligoclonal bands and nonreactive for the Venereal Disease Research Laboratory (VDRL) test. However, patient's level of consciousness and age did not allow measurement of visual acuity or hearing loss. Color fundus and red free photography was performed, which showed inferotemporal retinal arteriolar branch occlusion in the right eye [Figs. 1a and b]. Optical coherence tomography of right macula showed no significant change. Magnetic resonance imaging (MRI) revealed multiple foci of increased FLAIR T2 signal intensity in the periventricular white matter and corpus callosum [Fig. 2a] and T1 image showed lesions with decreased intensity in corpus callosum [Fig. 2c]. She was treated with intravenous prednisolone one gram/kilogram body weight for 3 days. The response to steroid was remarkable. The symptoms of encephalopathy subsided within 48 h. She was started on oral steroid (prednisolone) 10 mg/day for 10 days, which was tapered gradually. Repeat MRI, 4 weeks later, showed almost



**Figure 1:** (a and b) Fundus photo of right eye showing inferotemporal branch retinal arterial occlusion. Right eye red free (green) photo



**Figure 2:** (a-d) Magnetic resonance imaging FLAIR T2 image showing multiple well-defined hyperintense lesions in periventricular and callosal area. Magnetic resonance imaging T1 image showing hypointense lesions in center of corpus callosum. Magnetic resonance imaging, 4 weeks later showing almost complete disappearance of lesions

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complete disappearance of the foci [Figs. 2b and d]. Child did not return for subsequent follow-up visits.

## Discussion

To our knowledge, this is the first case report of SS in India. Since the first description of Susac Syndrome in 1979, hundreds of patients with SS have been reported, mainly in Western countries.<sup>[1]</sup> This syndrome is also known as “SECRET syndrome” (small infarctions of cochlear, retinal, and encephalic tissue) or “RED-M” (microangiopathy with retinopathy, encephalopathy, and deafness).<sup>[3,4]</sup> Most of the patients do not have the clinical triad at the onset of symptoms, but rather occurrences of one or more of the components of the triad. The condition is self limiting, chronic in nature with frequent remissions and exacerbations. Headache is most common presenting feature of encephalopathy and is a sign of central nervous system involvement. Other features include cognitive disturbance, memory loss, weakness and hyperreflexia. Ophthalmic involvement is seen as branch retinal arterial occlusion, which may present with symptoms like scotoma or impaired vision. Hearing loss is often the presenting feature and may be acute, unilateral or bilateral, and sometimes accompanied by vertigo, tinnitus, nausea, and vomiting. The cochlear hearing loss is usually greatest for low to moderate frequency tones,<sup>[6]</sup> and may be due to microinfarction of the apical turn of the cochlea. MRI changes consist of small well-demarcated, spherical, high-signal-intensity lesions located in the body and splenium of the corpus callosum on FLAIR and T2 sequences.<sup>[4]</sup> Eluvathingal *et al.*, reported a case of Susac’s syndrome in a 9-year-old girl to increase the awareness among pediatric radiologists of this entity, which is usually not considered as a differential diagnosis of multifocal white matter involvement in this age group.<sup>[7]</sup> MRI findings are commonly confused with demyelinating processes, typically multiple sclerosis or acute disseminated encephalomyelitis.<sup>[3,4]</sup> Saliba M *et al.*, reporting a 14-year-old girl with diagnosis of Susac’s syndrome state that it usually affects young women in young adulthood and is extremely rare during childhood.<sup>[8]</sup> The important differentials are multiple sclerosis, aseptic meningitis, systemic lupus erythematosus, Behcet’s and complicated migraine, sarcoidosis, tuberculosis, syphilis and lymphomas. A most recent study says that, the commonest cause of retinal arterial occlusion in young adults of Indian origin was found to be hyperhomocysteinemia. The highly prevalent nutritional deficiency in the Indian subcontinent is likely responsible for the hyperhomocysteinemia. In contrast, coagulation disorders, cardiac abnormalities, hemoglobinopathies and oral contraceptive use were cited as the major causes of retinal arterial occlusions in the Western population.<sup>[9]</sup>

Exact pathogenesis of this disorder is unknown, retinal microangiopathy and brain biopsies suggest a small vessel vasculitis leading to arteriolar occlusion and microinfarction of cerebral, retinal and cochlear tissue. Accumulation of amorphous material in the basal lamina, loss of viable endothelial cells, and capillary dropout suggest that SS may be an endotheliopathy.<sup>[10]</sup> Although, Susac’s syndrome is a self-limiting disease, but if not diagnosed and treated early it might lead to permanent hearing<sup>[11]</sup> and visual loss. Susac *et al.*, recommended the treatment, which includes pulse of methylprednisolone, intravenous immunoglobulin, oral prednisone and cyclophosphamide.<sup>[6]</sup> Similar strategy, using pulse steroid, helped our case to show considerable improvement.

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