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(CASE REPORT)

A case of possible Susac syndrome with avascular necrosis of hip

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Abstract

Susac syndrome (SuS) is a rare autoimmune micro-angiopathic endotheliopathy. The diagnosis of SuS is a challenge to neurologists because of the diversity of symptoms and rarity of occurrence of classical triad of SuS. In this context, a plethora of immunological/demyelinating disorders can mimic the features of neurological manifestations of SuS. The detection of snowball corpus callosal lesion is unique to SuS. A high degree of suspicion is the key to diagnosis and early immunomodulator therapy can prevent catastrophic disabilities. The use of steroid-sparing medications should be advocated wherever possible. In this case report, we tried to explore a 20 year old female with headache, seizure, irritability, blurring of vision with right hip pain and emphasize the need for awareness of this rare entity along with high element of suspicion for detection of such disorders.

Keywords: Susac; Endotheliopathy; Snowball; Immunomodulator

1. Introduction

Susac syndrome (SuS) is an autoimmune microangiopathic disorder of brain, retina and inner ear. SuS is characterized by clinical triad of encephalopathy, branched retinal artery occlusion (BRAO) and sensorineural hearing loss.[1]It primarily affects women of 20-40 years age (female:male ratio 3:1) and is likely underdiagnosed due to its atypical clinical presentation: the three components of the triad do not always show at the same time, and there is frequently multisystem involvement that mimics other conditions, delaying the diagnosis.[2] Encephalopathy in the form of headaches, multifocal neurological deficits, and psychiatric manifestations appear early in the disease.[3]

John O. Susac first documented this syndrome in two young women with the characteristic clinical triad in 1979, but Hoyt named it Susac syndrome in 1986.[4] SICRET (small infarcts of cochlear, retinal, and encephalic tissue); RED-M (retinopathy, encephalopathy, deafness-associated microangiopathy); and retino-cochleo-cerebral vasculopathy are other acronyms used for SuS. The clinical presentation, evidence of BRAO, occurrence of typical features on Fluorescence angiography(FA), and distinctive findings on cerebral magnetic resonance imaging (MRI) that help differentiate SuS from other inflammatory entities such as multiple sclerosis and acute disseminated encephalomyelitis are often used to establish the diagnosis. The corpus callosum is typically affected during encephalopathy, and MRI demonstrates a distinctive pattern of small-to-large round white matter lesions ("snowballs") (sagittal T2 FLAIR) and linear defects ("spokes") in the corpus callosum's central fibres.[5] Biopsies of the brain indicate perivascular non-necrotic inflammation of the small vessels and microinfarcts, distinguishing it from vasculitis. During the encephalopathic phase, lymphocytic pleocytosis and high protein levels, as well as occasional elevation of myelin basic protein, are seen in the cerebrospinal fluid, without evidence of oligoclonal bands or an elevated IgG index. Immunosuppressive therapy is the mainstay of treatment in SuS. The clinical course can be monocyclic, polycyclic or chronic as per Rennebohm.[3] However, early treatment has favourable outcome.

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2. Case description

A 20-year-old girl presenting with chronic episodic headache for 2 years, intermittent irritability with multiple episodes of transient loss of consciousness for last 6 months. She also complains of severe right hip pain for last 2 months for which she experiences difficulty in walking. The headache is usually left-sided, severe, throbbing in nature with nausea, vomiting, photophobia and phonophobia. It lasts for 12 hours to 24 hours and decreases with NSAIDs. She experiences headache 2-3 times weekly for last 1 year without any change during menstrual cycle. During the last 6 months, her mother noticed intermittent irritability in her with occasional aggressive behavior. She also experienced 6 episodes of transient loss of consciousness and hyper-ventillation lasting for 2-3 minutes without any involuntary movements, tongue-bite, incontinence of urine or faeces, with preserved awareness during the episodes. She complains of occasional blurring of vision and paresthesia in both legs. For last 1 month, she complains of pain in right hip while walking. There was no history of fever, altered sensorium, loss of memory, dimness of vision, diplopia, difficulty in hearing or swallowing, facial weakness, etc. There is a history of acute onset paraparesis with urinary retention 2 years back which recovered over a month.

The general examination showed pulse of 70/min, regular, blood pressure 118/70 mm Hg, respiratory rate of 14/m, without evidence of icterus, edema, clubbing, cyanosis, rash, joint tenderness, hair fall, significant lymphadenopathy, thyroid swelling, or skin changes. Nervous system examination was unremarkable except for a limping gait. Local examination revealed right-sided hip joint tenderness with positive Faber's test. The neuro-ophthalmic examination showed normal pupillary size, shape, symmetricity and reaction to light with normal ocular movements and absence of relative afferent pupillary defect. The auditory functions were normal. On investigation, she had mild microcytic anemia with normal rest of hemogram, normal blood glucose, liver, renal, thyroid functions, electrolyte levels with hypovitaminosis D. Her vasculitic profile including anti-phospholipid antibody levels were normal. Her cerebrospinal fluid (CSF) examination showed 5 cells, all lymphocytes, protein 38mg/dL and sugar 76mg/dL. Her magnetic resonance imaging(MRI) of brain revealed T2/FLAIR peri-callosal white matter snowball hyperintensities with symmetric subcortical hyperintensities in T2/FLAIR without DWI restriction, suggesting demyelinating pathology (Figure-1). MRI of spine and orbit were unremarkable. The MR angiogram did not show any beading, attenuation or malformation. MRI of hip showed bilateral avascular necrosis of both hip joint (right>left)(Figure-2). Visual evoked potential was prolonged P-100 latency in right eye(110ms), with normal recording of left eye(106ms) [normative data suggesting VEP >108ms as prolonged value]. Fundoscopy revealed perivenous exudation(Figure-3). Fundus fluorescein angiography revealed hyper-fluorescence of the arterial vessel wall (Figure-4 A,B).Her CSF oligoclonal band andIgG index were normal along with negative anti-NMO and anti-MOG antibody assay. She has previously received prolonged oral corticosteroid therapy and azathioprine for 8 months but she discontinued treatment from herself 4 months prior to present admission. There was no evidence of new lesions or resolution of previous lesions in brain imaging for the last 2 years. She was diagnosed as possible Susac Syndrome and started on amitryptilline 10mg/day and naproxen 250 mg (sos). She showed marked improvement with ibandronate therapy from Orthopaedic team-work. Psychiatric counseling sessions were given in view of panic attacks. At discharge, she was clinically better with good response to headache treatment and behavioral aspects on counseling therapy.

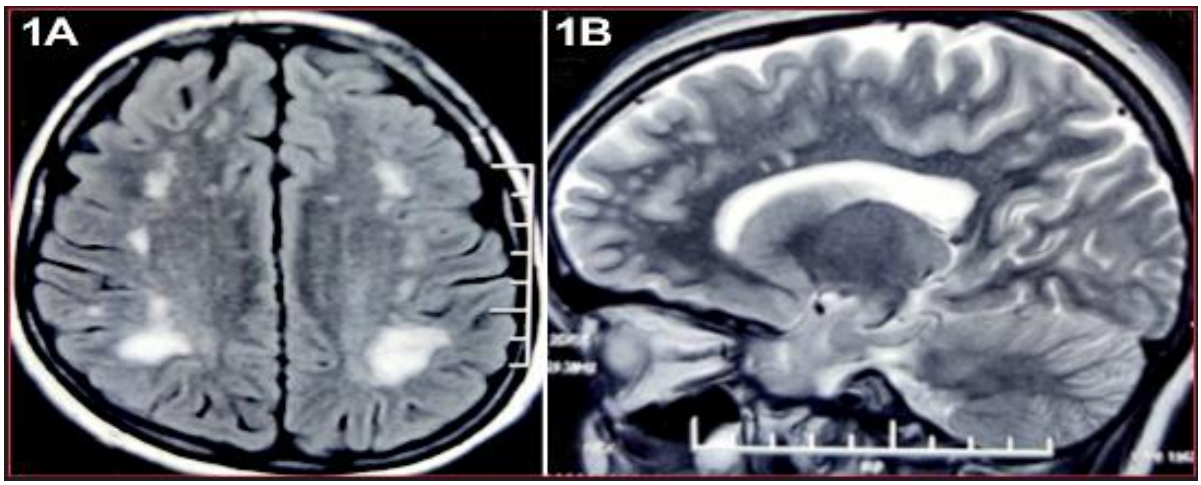

Figure 1 MRI brain showing symmetric subcortical hyperintensities in FLAIR sequence (A) and pericallosal snowball appearance T2 hyperintensities in sagittal view (B)

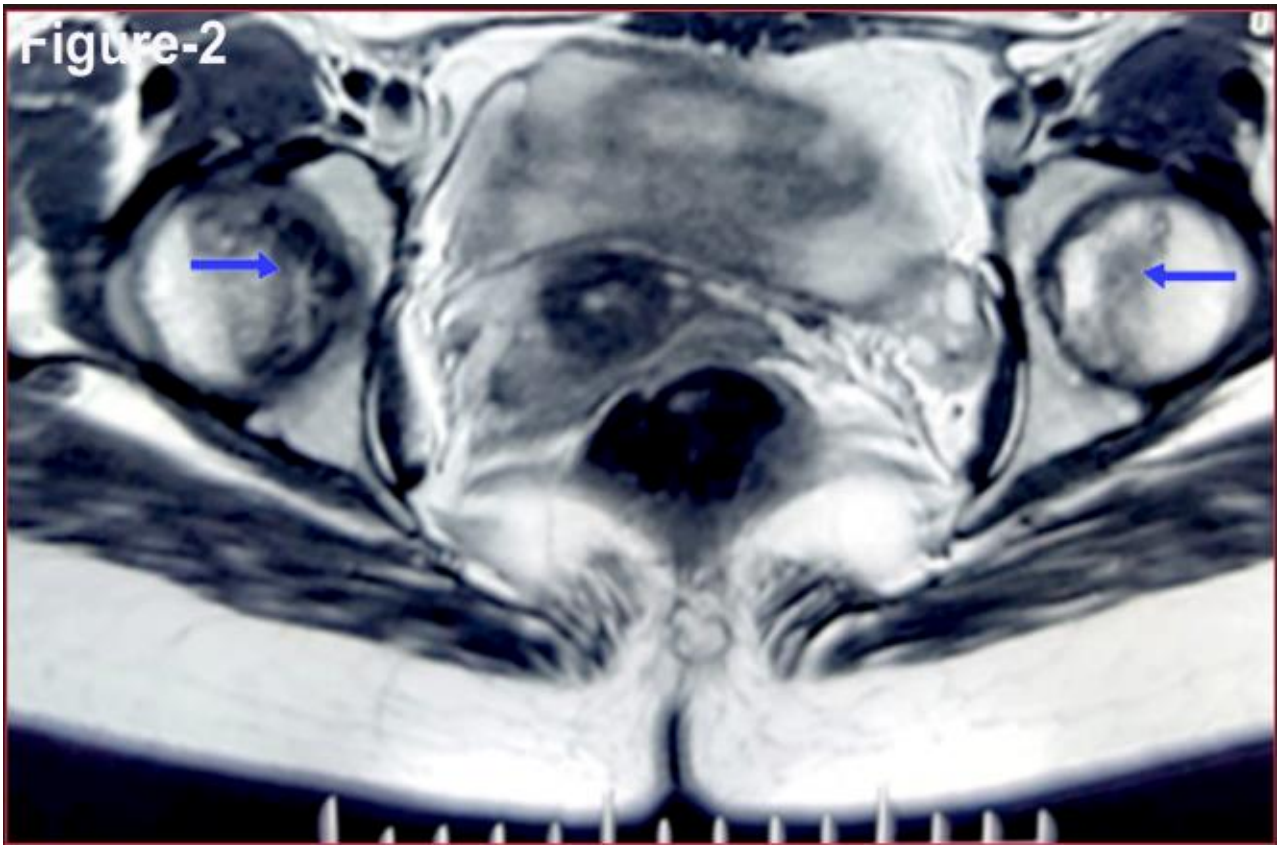

Figure 2 MRI hip joint showing bilateral asymmetric T2 hypointensities (blue arrow) of medial portion of head of femur suggesting avascular necrosis of head of femur

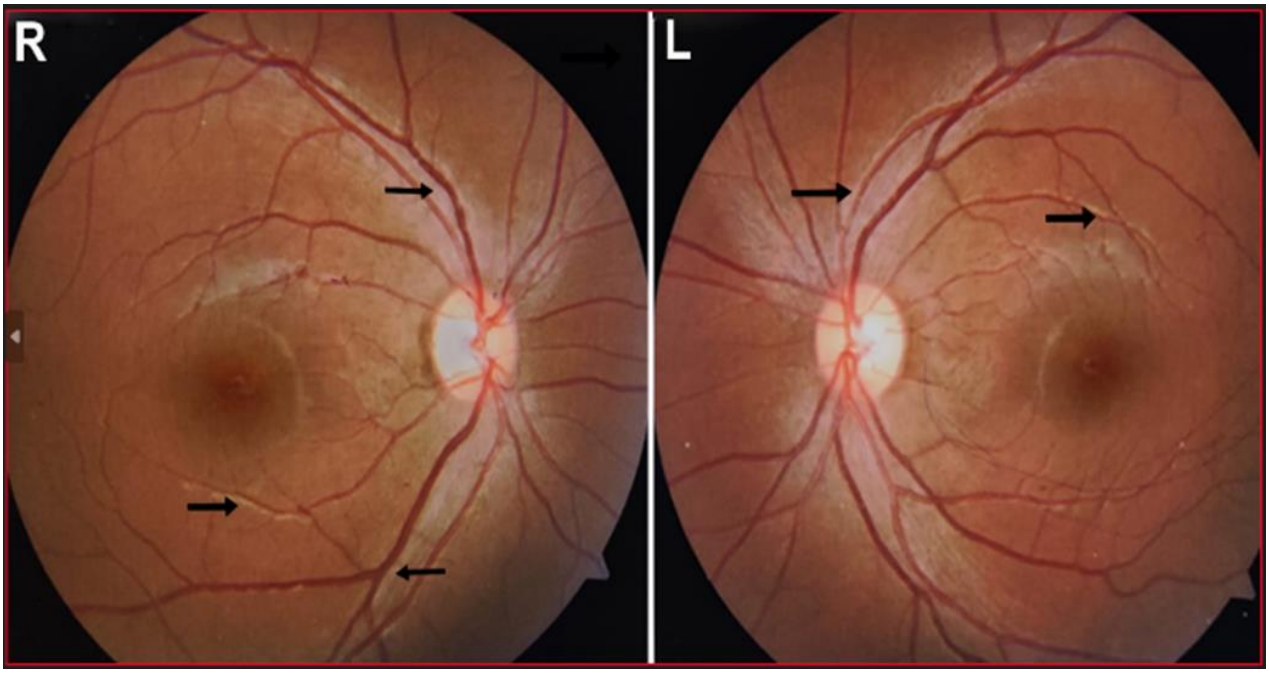

Figure 3 Fundus photography showing perivascular exudation (black arrow) in both sides

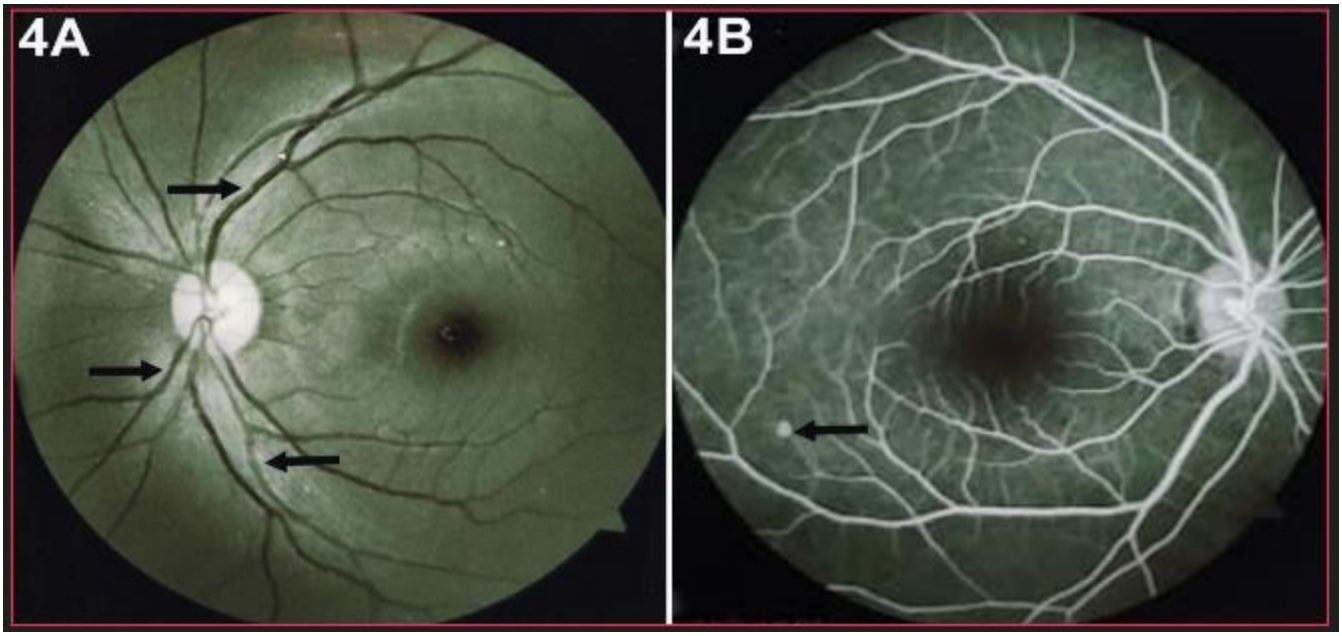

Figure 4 Fundus fluorescein angiography showing perivascular exudation(4A) and hyperfluorescence of the arterial vessel wall (4B)

3. Discussion

The diagnosis of Susac syndrome(SuS) is a challenge to neurologists because of the diversity of symptoms and rarity of occurrence of classical triad of SuS(13%).[6] In our case, she was diagnosed as Possible SuS as per diagnostic criteria proposed by Kleffner et al.[7]

Headache(migranous or oppressive type), seizure, cognitive impairment, focal neurological deficits and psychiatric illness can manifest eventually on the timeline of disease progression, and at times monophasic, further increasing the diagnostic dilemma. BRAOs manifesting with scotoma or photopsia with diffuse bilateral occlusion of retinal vessels might be delayed in onset and accumulate over time. On the contrary, a plethora of immunological disorders including multiple sclerosis, acute disseminated encephalomyelitis, Neuromyelitis optica spectrum disorder, Systemic lupus erythematosus, CNS vasculitis, CADASIL etc. can manifest such neurological involvement. In our case, the 20- year-old girl with background history of chronic episodic migraine, one episode of seizure, irritability, blurring of vision with classical corpus callosal lesions in imaging, perivenular exudation in fundoscopy and retinal fluorescein changes lead to the diagnosis of SuS. Despite some clinical manifestations of spinal cord dysfunction in SuS, no spinal cord lesions have been seen on MRI of SuSpatients as evident in our case.[8]

Due to the sheer rarity and lack of knowledge about its patho-physiology, there are no prospective or randomized controlled studies of treatments for SuS. The postulate of an autoimmune inflammatory endotheliopathic etiology supports empirical therapeutic options. At present state, she developed avascular necrosis of both hip due to prolonged steroid therapy. The use of steroid-sparing immunomodulators is better tolerated and safe for chronic use in such disorders. The disease is strongly recommended to be rapidly and completely suppressed to prevent the target organs from irreversible damage causing blindness, dementia and deafness.[9]

Treatment is determined by the severity of CNS involvement, which is divided into extremely severe, severe, moderate, and mild manifestations, according to the most recently published treatment guidelines.[10] Intravenous pulsed methyl prednisolone, followed by high-dose oral prednisone combination with intravenous immunoglobulin, cyclophosphamide, mycophenolate mofetil, tacrolimus, and rituximab, is usually required for extremely severe CNS involvement. Treatment parameters for severe CNS disease are similar to those for extremely severe disease, with the difference that cyclophosphamide is not required. Intravenous pulsed methyl prednisolone is used to treat moderate CNS illness, followed by high-dose oral prednisone combination with intravenous immunoglobulin, mycophenolate mofetil, and rituximab. Intravenous pulsed steroids are an option in mild CNS cases. Intravenous immunoglobulin, mycophenolate mofetil, and rituximab are coupled with high-dose oral steroids. Treatment for all patients, regardless of severity, usually lasts more than two years. Treatment with intravenous pulses of methyl prednisolone, followed by oral prednisone and a prednisone taper, is initiated in individuals with a predominant clinical presentation of BRAO.

Mycophenolate mofetil and intravenous immunoglobulin are also indicated. The prophylactic use of anticoagulants or aspirin is not effective in SuS and hence not advocated unless associated with anti-phospholipid antibodies.[11]

4. Conclusion

Susac syndrome is a treatable autoimmune disease with endotheliopathy affecting brain, retina and inner ear. The classic triad of this rare disorder is less than 15% in clinical practice. Hence, a strong index of suspicion is required to diagnose SuS. The early initiation of immunomodulators gives favorable outcome and can prevent permanent damage of vital organs.

Compliance with ethical standards

Acknowledgment

I acknowledge the patient and the parents for giving consent for publishing data for medical learning.

Disclosure of conflict of interest

Authors have declared that no conflict of interests exist.

Statement of informed consent

Informed consent was obtained from all individual participant included in the study.

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