

Susac Syndrome in a Pre Puberal Femele: Rare Case

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1. Abstract

Suac Syndrome is an autoimmune endotheliopathy with about 304 cases described until 2013. It charaterized by the triad of E-H-V (Encephalopathy, Hearing loss and Vision - branch retinal artery occlusions/BRAO [3]. The case report early-onset autoimmune neuropsychiatric disorder in a pre-pubertal 12 year old girl innitialy presenting with behavioral and emotional manifestations , firstly treated as a psychiatric disorder reaching the correct diagnosis of Susac Syndrome after a careful MRI analysis checking for specific findings of æoesnowbollsae in corpus callosum. The chek of sublte findigs in addition to unespecific perivenular and periventricular withe matter lesions in a young patient with confounding clinical presentation provided support for different treatment that prevented additional brain atrophy and decreased functionality.

2. Introduction

The present case report outlines the marked outcome change of a rare incapacitating neuropsychiatric disorder with unusual clinical presentation with a careful MRI analysis. It is the case of early-onset autoimmune neuropsychiatric disorder in a pre-pubertal 12 year old girl innitialy presenting with behavioral and emotional manifestations without clear cognitive and motor deficits firstly treated as a psychiatric disorder reaching the correct diagnosis of Susac Syndrome (SS) after a careful MRI analysis checking for rare although specific findings of "snowballs" in corpus callosum, together with the search for common images. The additional chek of sublte findigs in addition to unespecific perivenular and periventricular withe matter (WM) lesions in a young patient with confounding clinical presentation provided support for different treatment that prevented additional brain atrophy and decreased functionality.

SS is mostly described as an autoimmune endotheliopathy with about 304 cases described until 2013. It charaterized by the triad of E-H-V (Encephalopathy, Hearing loss and Vision - branch retinal artery occlusions/BRAO). Most patients present cognitive, mnemonic, behavioral and affective symptoms including

pseudobulbar affect (PBA) and abulia, and sometimes with more bizarre neuropsychiatric manifestations[1]. Clinically the diagnosis more difficult whith incomplete initial presentation of the triad [2]. Despite the rare incidente, SS must be considered in the differential diagnosis of several neurological, psychiatric and eye disorders, as well as ear, nose and throat (ENT) conditions, and is often misdiagnosed as multiple sclerosis (MS). Additionally, SS must be checked in severe disorders with prominent psychiatric symptoms with unespecific autoantibodies, such as schizophrenic disorders [3].

3. Case Description

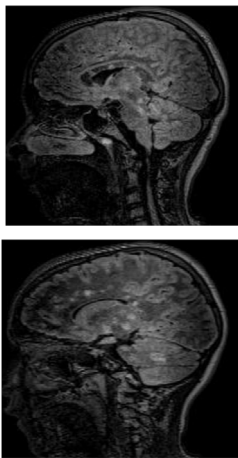
A twelve-year old girl with a 120-days course of sudden personality change, later evolving to mental confusion, irritability and learning disability, and about 45 days after onset with paranoid symptoms, increased holocranial headache more intense in the occipital region with vomiting, affective dullness and bradypsychism. 80 days after onset she evolved to pronounced walking deficits, bilateral lower limb weakness, speech and cognitive impairment, focal seizures with secondary generalization, attenuated after treatment with anticonvulsants.

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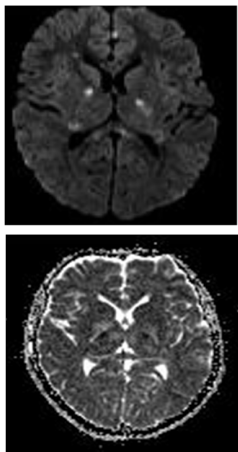
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On physical examination, the patient was aphasic, obtunded, isophotoreagent pupils, mouth masticatory movements, grade 3-4 tetraparesis, symmetric bilateral patellar hyperreflexia, and extensor cutaneous-plantar reflex.

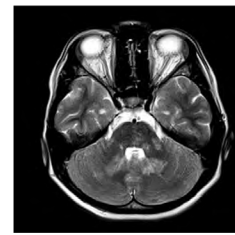
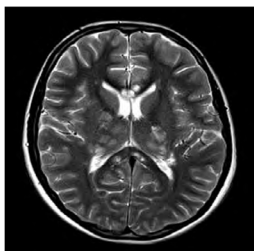
Blood serology was negative, Lumbar study with increased cellularity and proteins, negative for oligoclonal bands and neuronal N-metil-D-aspartate (NMDA) receptors. MRI (Figures 1- 8) revealed nodular lesions with increased signal in corpus callosum, corresponding to “snowballs”, periventricular white matter lesions and in middle cerebellar peduncles. Audiometry (Figure 9) detected moderate degree of bilateral sensorineural hearing loss. Retinal angiofluoresceinography (Figure 10) suggested residual arterial branch occlusion.



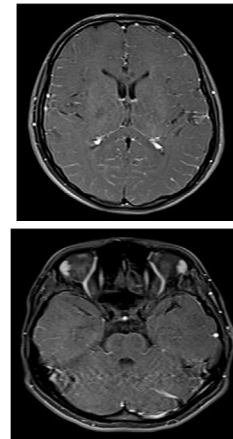
Figures 1e 2: (Sagittal FLAIR):3 to 7 mm hypersignal nodular lesions at periventricular white matter, middle cerebellar peduncles, and corpus callosum,characterized as “Snowballs”.



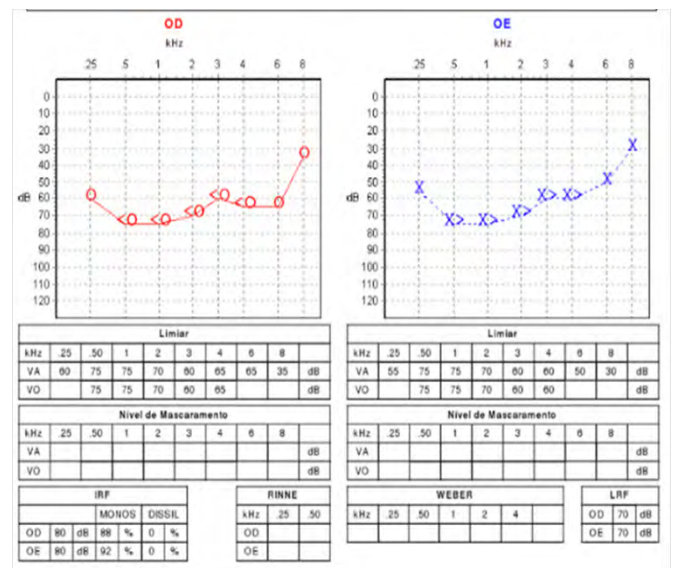
Figures 3: (Axial Diffusion); 4 (Axial ADC map) Restriction to diffusion of water molecules from nodular lesions.



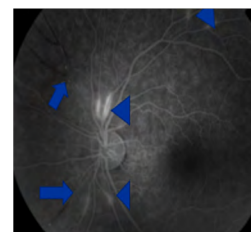
Figures 5,6 (Axial T2): Lesions with T2 hypersignal in corpus callosum, posterior arm of internal capsule, thalamus (PALIC) with upper and middle cerebellar peduncles and bilateral dentate nuclei involvement.



Figures 7,8: (Axial T1 with Gadolinium Fat Suppression) with no contrast enhancement of lesions.



Figures 9: Audiometric findings of Bilateral Sensorineural Hearing Loss/ Moderate degre.



Figures 10: Retinal angiofluoresceinography: Arrows: multiple retinal arteriolar obstructions. Arrowheads: focal areas of hyperfluorescence in arterial walls.



Figure 11 (Sagittal T1): Cavitated lesions on the corpus callosum and brain and cerebellar volume reduction.

At hospital admission the patient received standard dose immunoglobulin. At discharge, she had less confusion and partially recovered motor deficits, persistent patellar hyperreflexia and Babinski Reflex.

Patient received 5 cycles of cyclophosphamide along 6 months. Final assessment evidenced improved strength and gait, seizure control, grade 5 global strength, bilateral Babinski, diffuse hyperreflexia, walking without assistance with enlarged base, and mild cognitive impairment (MCI). Final assessment: MRI (**Figure 11**) with cavitory lesions in corpus callosum with diffuse Brain Volume (BV) reduction.

4. Discussion

Susac's Syndrome occurs in young women between the ages of 20 and 40, and more eventually in men. The age range in both sexes is from 16 to 58 years, with male to female ratio of 3:1 [4]. Affected patients have multiple bilateral branch retinal artery occlusions, progressive hearing loss, and variable neurologic findings including but not limited to psychiatric symptoms and encephalopathy [5]. Adequate diagnosis and management often require a multidisciplinary effort involving neurology, neuroophthalmology, otolaryngology, neuroradiology, and rheumatology [6].

Brain Magnetic Resonance Imaging (MRI) has an important role in assessment of suspected SS [7]. The most important MRI sign and typical for SS is the snowball lesion in the center of the corpus callosum, reported in 78% of patients [8]. Early findings include contrast enhancement around small vessels representing perivascular leakage and frequently with leptomeningeal contrast enhancement. Other typical MRI findings are multiple small central holes giving a riddled aspect of corpus callosum. The most important differential diagnoses are multiple sclerosis (MS) and acute disseminated encephalomyelitis (ADEM). In the case, the initial hypothesis was MS, and the test for an "exotic" differential diagnosis (unusual early onset, unusual early behavioral abnormalities precluding the typical "triad" provided correct diagnosis and early appropriate therapy that reduced the severity of permanent sequels expected in this disease. Diagnostic problems like the above, specially when the most frequent diagnosis fit with the initial Clinical and Radiological presentation is

common, leading to delayed treatment and increased morbidity. In the case, It would be expected severe visual loss (32%), hearing loss (90%), and neurological deficits (56%) [9]. Otherwise, rapid diagnosis in this girl led to early and continued immunosuppressive therapy, with almost complete recovery, coincident to previous studies in similar conditions [10]. Foremost, similar methodology looking for additional diagnostic alternatives (traditional, deadly and exotic alternatives [11]) provide dramatic reduction of diagnostic bias in Medicine.

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