Snowballs on a spring day: a case report

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OBJECTIVES

Susac syndrome is a rare autoimmune cerebrovascular disease characterized by the clinical triad of encephalopathy, branch retinal artery occlusion (BRAO), and sensorineural hearing loss. Early diagnosis and aggressive treatment are crucial to improve outcomes and minimize permanent disability.[1-3]

MATERIALS

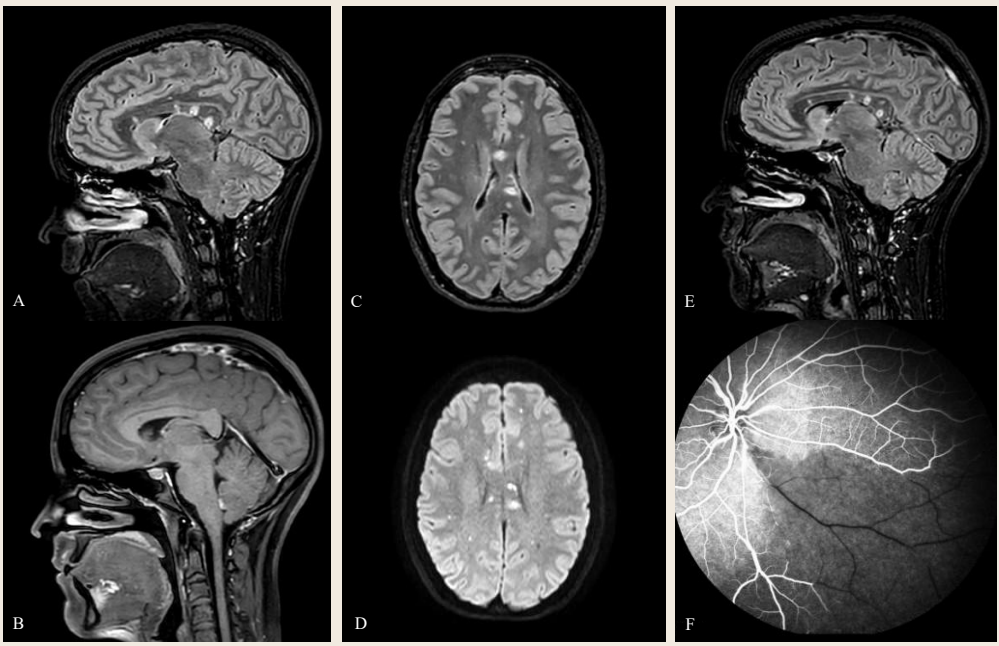
We analysed signs and symptoms, CNS imaging, CSF findings, blood tests, and neurophysiological findings of a young patient admitted to our department.

METHOD

We present the case of a 21-year-old female with an unremarkable past medical history admitted to the neurological department in March 2025. About a week before the hospitalization, the patient reported new onset refractory mild-intensity headache, drowsiness, asthenia, ears fullness and mild impairment in memory and attention domains. Her neurological examination showed attention and short-term memory deficits, partial disorientation in time, postural instability and fluctuating ataxic marks. Primary brain CT and CTA scans were unremarkable. A lumbar puncture was performed and CSF analysis showed protein level of 212 mg/dL, with normal glucose, cell count and microbiological analysis.

RESULTS

A first electroencephalography (EEG) showed a continuous theta-alpha background with bilateral frontally predominant polymorphic delta waves. Brain MRI revealed multiple 'snowball-like' lesions in both supratentorial and infratentorial white matter, mostly pronounced in the callosum corpus. These findings raised suspicion of Susac syndrome. An intravenous steroid bolus therapy was started, followed by oral tapering. Spine MRI was unremarkable. A follow-up brain MRI showed an improvement in the number and size of the lesions. A later post-therapy EEG was similar to the admission one. Neuropsychological testing revealed a mild pathological performance for the lexical tests, verbal learning and working memory. The audiometric exam revealed an initial neurosensorial loss on low frequencies. OCT with fluoroangiography identified multiple peripheral BRAO. Visual evoked potentials and brainstem auditory evoked responses were within normal limits. A diagnosis of definite Susac syndrome was formulated.[3] The patient was discharged on oral steroid and single antiplatelet therapy.



The figures displayed show: T2/FLAIR sagittal section (A), T1 with contrast sagittal section (B), T2/FLAIR axial section (C), DWI axial section MRI at the admission (D); T2/FLAIR sagital section post-treatment MRI after two weeks (E); peripheral BRAO at the OCT with fluoroangiography (F).

DISCUSSION

This case report underscores that the classic clinical triad of Susac syndrome is infrequently observed at disease onset. Brain MRI is a crucial diagnostic tool, often raising suspicion of the condition, which must be confirmed through additional investigations. Robust evidence supporting an optimal immunotherapeutic regimen remains limited. [1-3]

CONCLUSION

Susac syndrome is a rare autoimmune microangiopathy of the CNS. It should be included in the differential diagnosis for young female patients presenting with encephalopathy and sensorineural hearing loss. Randomized controlled trials are necessary to establish the most effective treatment strategies for this population.

BIBLIOGRAPHY

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