Susac syndrome – single center case presentation and first time report of new potential diagnostic tool.

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INTRODUCTION

Susac syndrome (SuS) is an autoimmune endotheliopathy, which affects precapillary segments of arteries in the brain, cochlea and retina. resulting in a clinical triad of encephalopathy, sensorineural hearing loss, and visual loss [1-3]. SuS is a rare and often underdiagnosed condition with about 450 cases reported worldwide. It affects young people, especially women [4]. Clinical presentation is variable [2,5-7] and not all patients exhibit the whole triad at the onset of symptoms [2,4-9]. In addition to clinical variability, the disease course and aggression vary widely [1]. In mild cases, peak disease severity lasts only for several months, and the dysfunction is reversible. On the other hand, prolonged course over several years with devastating long-term or even fatal consequences has been reported as well[1,8,10,11]. Only one case of SuS has been reported from the Czech Republic so far. This paper describes further 2 patients admitted to our Neurology department within 6 months – the first one presenting with mild symptoms, the second one at the other one end of the clinical spectrum including very rare cardiac involvement[6], where all of the available treatment options (corticosteroids, cyclophosphamide, intravenous immunoglobulins, plasmapheresis, rituximab) were used to achieve remission.

CASE 1

- 61-year-old woman
- clinical presentation: acute onset of rotatory vertigo, vomiting, and confusion lasting for several hours.
- neurological examination: partial amnesia without any focal neurological deficit
- MRI: several lesions (Figure 1-A, 1-B) in the corpus callosum and in the left precuneus with diffusion restriction
- black blood MRI (Figure 1-C.I) : inflammatory microangiopathy, with gadolinium enhancement of the small vessel walls, dot-like infarctions and focal leptomeningeal enhancement.
- CSF: mild mononuclear pleocytosis (8 cells/µl), no oligoclonal bands were found.
- Audiometry: sensorineural hearing loss on the right ear.
- Ophthalmology: normal
- Fundus fluorescein angiography (FAG): regional hyperfluorescence and peripheral vessel segmentation (Figure 1-D).
- Treatment : pulse of methylprednisolone (total dose of 5 g), high-dose oral prednisone (1 mg/kg) with a slow taper, azathioprine (100 mg per day)

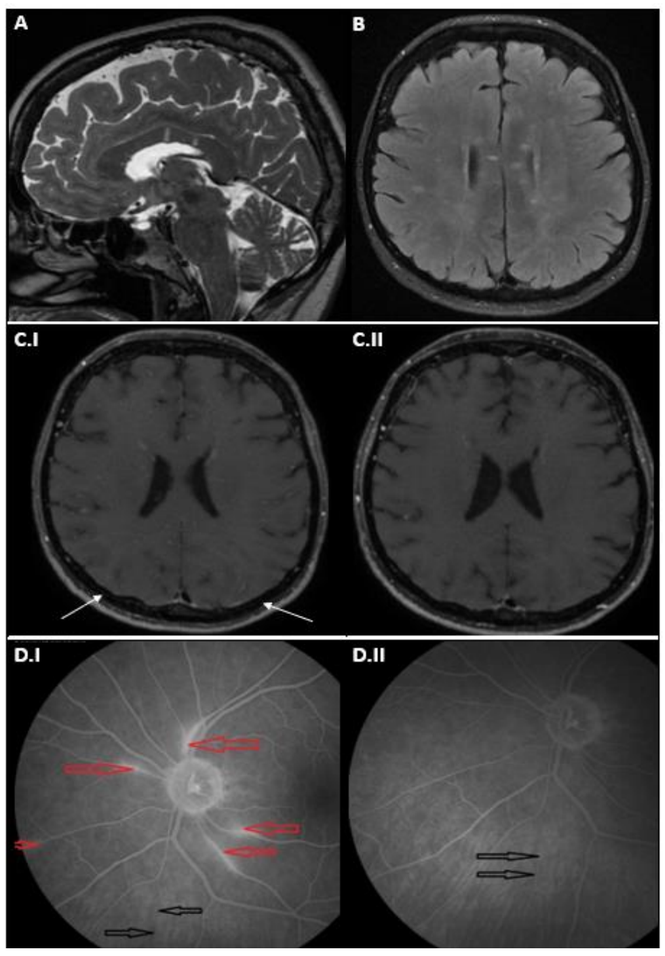

Figure 1

CASE 2

- 32-year-old woman
- clinical presentation: recurrent weakness and paresthesia of left-side limbs
- neurological examination: severe encephalopathy with multiple domain cognitive impairment, left-sided central hemiparesis and tactile neglect syndrome, ECG monitoring showed persistent asymptomatic bradycardia
- MRI: multiple areas of restriction of diffusion predominantly in the corpus callosum, in bilateral subcortical white matter and in the cerebellum (Figure 2- C.I)
- Black blood MRI: gadolinium enhancement of small vessel walls (Figure 2-C.I), focal leptomeningeal gadolinium-enhancement in the posterior fossa
- CSF: pleocytosis and elevated protein concentration; no oligoclonal bands were present
- Audiometry: confirmed subclinical sensorineural hearing loss in the left ear
- Fundoscopy: intraretinal hemorrhage and peripheral artery occlusion in the left eye
- FAG: multiple peripheral artery occlusions in both eyes, but no areas of hyperfluorescence (Figure 2- A.I-II)
- Treatment: two pulses of methylprednisolone (5 g for the first pulse and 7 g for the second pulse after a clinical and radiological relapse), taper (80 mg of prednisone per day), plasmapheresis, cyclophosphamide (12 mg/kg iv), IVIG, rituximab

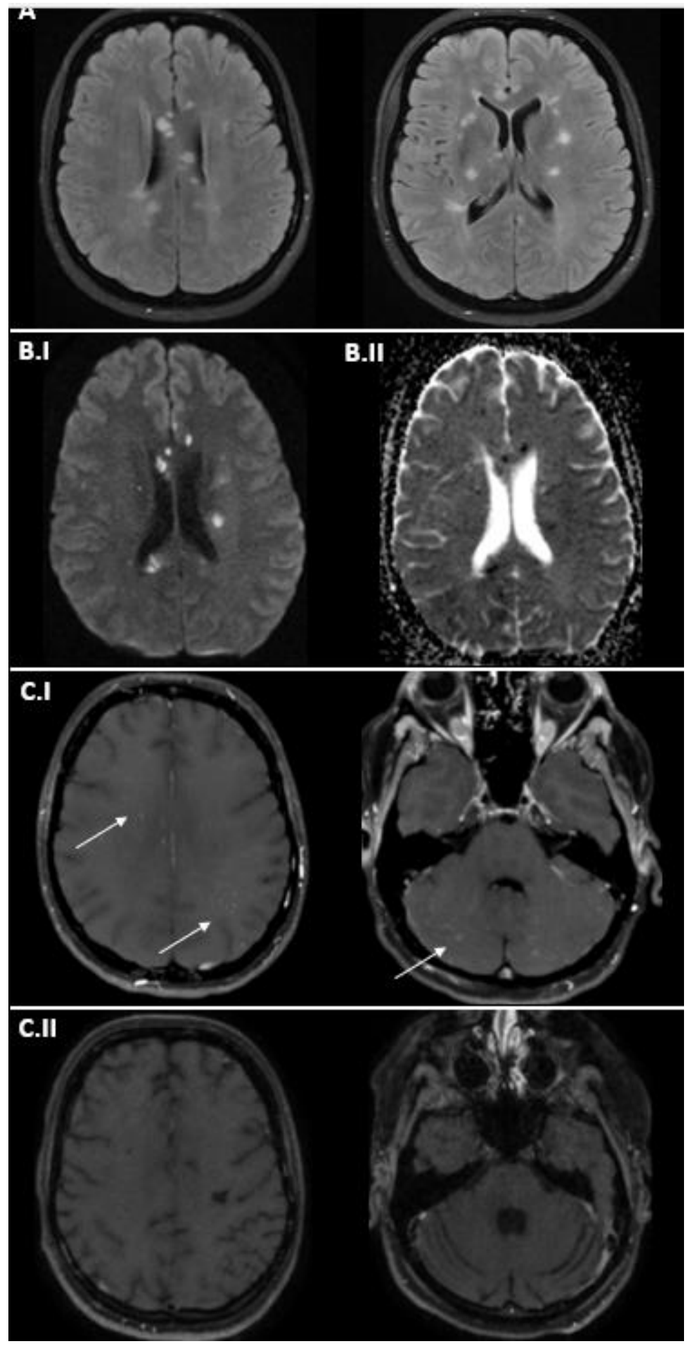

Figure 2

CONCLUSION

Diagnosis of Susac syndrome can be very challenging due to its rarity and broad clinical variability. We highlight the benefit of black blood MRI of the vessel wall, that can be very useful diagnostic tool and provides us also important information according to disease activity and treatment efficacy. Despite its rarity, the awareness of Susac syndrome may be of uttermost importance since it ultimately is a curable condition. If diagnosed in a timely manner, early intervention can substantially improve the outcomes of our patients.

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