Medicina (Kaunas) 2013;49(1):15–7

Susac's Syndrome: A Case Presentation and Radiological Approach to this Rare Autoimmune Endotheliopathy

Goda Randakevičienė[1], Rymantė Gleiznienė[1], Rūta Nylander[2]
[1]Department of Radiology, Medical Academy, Lithuanian University of Health Sciences, Lithuania,
[2]Department of Radiology, Uppsala University Hospital, Sweden

***Key Words**: Susac's syndrome; microangiopathy; magnetic resonance imaging; corpus callosum; lesions.*

***Summary.** Susac's syndrome is an uncommon neurologic disorder, consisting of the clinical triad of encephalopathy, branch retinal artery occlusions, and hearing loss. We report a case of a sudden vision and hearing impairment in a 35-year-old female patient. Magnetic resonance imaging of the brain revealed multiple lesions in the white matter and the corpus callosum, typical of Susac's syndrome.*

Introduction

The clinical triad of encephalopathy, branch retinal artery occlusions, and hearing loss was first described by Susac et al. in 1979: they presented a case of an unusual microangiopathy of the brain and retina and sensorineural hearing loss in 2 women. Both of the patients had developed dementia and spasticity and had had changes in personality and paranoid psychosis (1).

Susac's syndrome is also known as a retinocochleocerebral vasculopathy, RED-M (retinopathy, encephalopathy, and deafness associated with microangiopathy) and SICRET (small infarctions of cochlear, retinal, and encephalic tissue) syndrome. In its partial forms, the clinical triad may be incomplete, making this relatively new and rare syndrome even harder to diagnose. Therefore, it is often misdiagnosed as multiple sclerosis (MS) or acute disseminated encephalomyelitis (ADEM) (2).

Being familiar with the syndrome and keeping in mind distinctive white matter lesions in the central part of the corpus callosum, leptomeningeal enhancement occurring in about one-third of Susac's syndrome cases (but never in MS or ADEM) (3), and typical clinical manifestations should make the right diagnosis easy to derive.

Case Report

A 35-year-old woman with a history of an abrupt hearing impairment in the right ear 4 months ago presented with sudden vision deterioration in the right eye. She had been suffering from severe migraine for years and had a history of meningitis at the age of 10 years.

On an ophthalmological examination, the visual field loss of the right eye was observed; ophthalmoscopy revealed multiple branch retinal artery occlusions. The findings of neurological examination were consistently normal except for a migrainous headache. Routine laboratory blood results were insignificant.

CT of the head revealed no major pathological changes except for a cystic gliotic lesion – an old, small lacunar infarct in the left corona radiata (Fig. 1).

Magnetic resonance imaging (MRI) of the brain without and with contrast was performed: nonspecific hyperintense lesions on T2-weighted and T2-weighted/FLAIR images as well as linear like defects, "spokes," located bilaterally in the periventricular white matter were observed (Fig. 2).

Hyperintense, irregularly shaped white matter lesions without contrast uptake were observed in the central fibers of the corpus callosum (Figs. 3 and 4).

Based on the findings of ophthalmologic (branch retinal artery occlusions, visual field defects) and otolaryngological examinations (sensorineural hearing loss) and radiological imaging (characteristic lesions on the MR image of the head), the diagnosis of Susac's syndrome was made.

Discussion

Susac's syndrome is a rare autoimmune microangiopathy of the brain, retina, and inner ear. It predominantly affects young persons with a male-to-female ratio of 1:3. The age range extends from 16 to 58 years (3), but women aged between 20 and 40 years are most vulnerable (4).

Clinically the diagnosis is difficult to make due to syndrome polymorphism, especially when the clinical triad is lacking; in most of cases, the 3 elements are not all present at the onset of the disease and may occur days, months, or even years later. In partial forms of this syndrome, 1 or 2 components remain silent all the time.

The syndrome has a pathognomonic radiological feature on MRI: idiosyncratic callosal white matter lesions, which together with clinical symptoms can permit an immediate diagnosis (5).

Correspondence to G. Randakevičienė, Department of Radiology, Medical Academy, Lithuanian University of Health Sciences, Eivenių 2, 50028 Kaunas, Lithuania
E-mail: goda.vilu@gmail.com

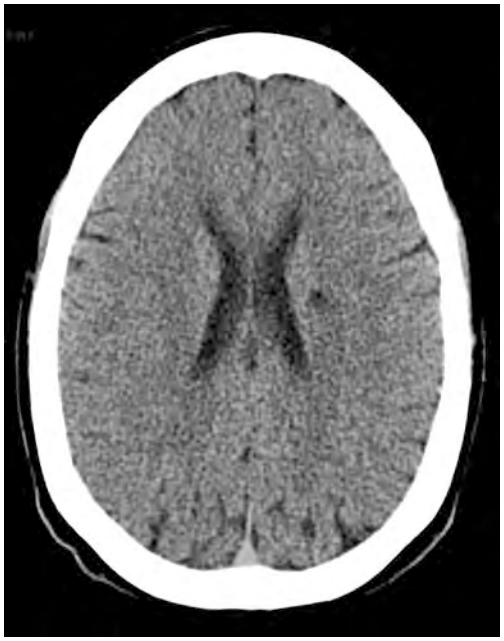

Fig. 1. A computed tomography scan
A small hypodense lesion in the left corona radiata.

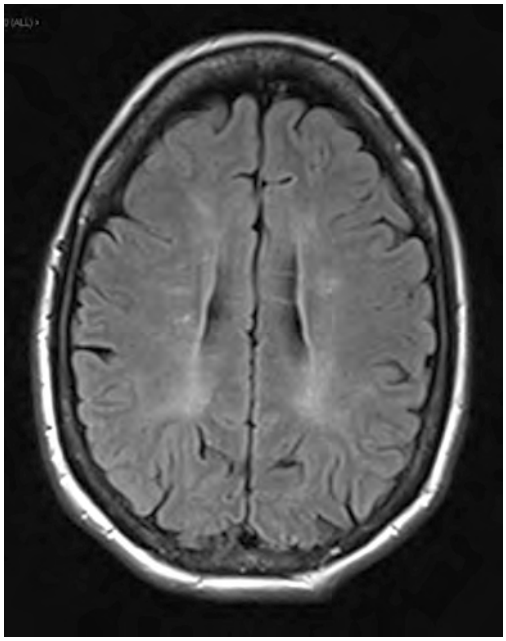

Fig. 2. A magnetic resonance imaging, T2-weighted/FLAIR axial image
Multiple hyperintense white matter lesions (linear shaped "spokes") are seen bilaterally in the corona radiata.

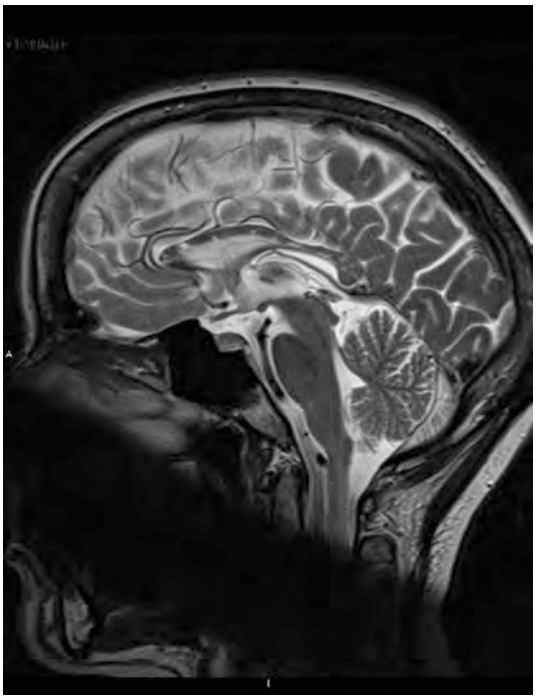

Fig. 3. A magnetic resonance imaging, T2-weighted sagittal image
Lesions in the central fibers of the corpus callosum.

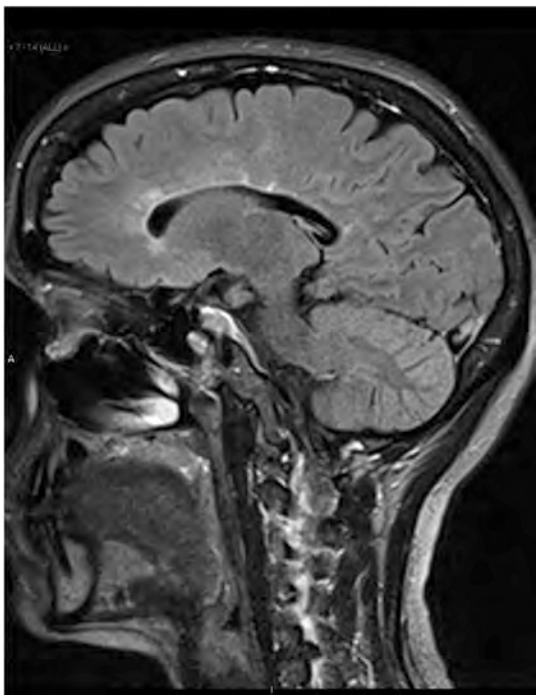

Fig. 4. A magnetic resonance imaging, T2-weighted/FLAIR sagittal image
Irregularly shaped lesions in the central fibers of the corpus callosum.

The manifestation of encephalopathy is polymorphic: it may present with a headache as the major feature, which is an almost constant complaint. Disorientation, confusion, and memory and psychiatric disturbances are often observed (3, 4).

Branch retinal artery occlusions, usually bilateral, may be the presenting features of Susac's syndrome. However, in most of the cases, being difficult to diagnose, they are not always observed at the onset of the disease. Thus, examinations by neuroophthal-

mologists or retinal specialists should be repeated at frequent intervals. Branch retinal artery occlusions, if not found at the beginning, may develop later in the course of the disease (3).

Cochleovestibular symptoms, such as hearing loss being most often bilateral and associated with tinnitus and vertigo (6), occur due to the microinfarcts of the inner ear (cochlea and semicircular canals) (3).

MRI is the neuroimaging study of choice for Susac's syndrome (2). MRI shows a cortical, deep gray matter involvement (70%) and leptomeningeal enhancement (33%) (3). According to Susac et al. (3, 4, 6), MRI findings always include distinctive white matter lesions of the corpus callosum; the central fibers of the corpus callosum are the most susceptible region in the course of this disease. MRI scans in Susac's syndrome show a characteristic pattern of small-to-large round white matter lesions, "snowballs," and linear defects, "spokes," found in the central fibers of the corpus callosum (7). As the acute changes resolve, central callosal "holes" develop (4). The number of lesions evident on the MR image is not related to the degree of encephalopathy (3).

MRI findings, seen in patients with Susac's syndrome, are often attributed to MS or ADEM. However, the lesions found in patients with Susac's syndrome differ from those in a demyelinating disease: the callosal white matter lesions typically involve the central fibers with relative sparing of the periphery (6). In MS and ADEM, in contrast to Susac's syndrome, the callosal involvement is seen on the undersurface of the corpus callosum at the septal interface. In MS or ADEM, no leptomeningeal enhancement is found; in the case of Susac's syndrome, the leptomeningeal involvement is found in 33% of cases (3, 4). Deep gray matter lesions occurring in Susac's syndrome and never in MS may help differentiate Susac's syndrome from MS; the lesions may occur in ADEM too, but in this case, the clinical features and history of the previous viral infection or vaccination are usually helpful in differentiating these two entities.

Susac's syndrome responds well to immunosuppressive therapies: a combination of steroids, cyclophosphamide, and intravenous immunoglobulin. Aspirin is a useful additive (4). Based on the findings of light microscopy in the brain biopsy material analogous to an antiendothelial cell antibody (AECA)-mediated microvascular injury, it is postulated that Susac's syndrome is a distinct autoimmune endotheliopathy syndrome associated with AECAs (8).

According to Rennebohm et al., Susac's syndrome and dermatomyositis are very similar immunopathogically; therefore, the immunosuppressive treatment protocol and recommendations for Susac's syndrome similar to the ones for dermatomyositis are suggested (7, 9).

The disease usually has a monophasic, self-limited course (3) and lasts from months to years. According to O'Halloran et al., the mean duration of the illness is 46.7 months (10).

The course and outcomes of Susac's syndrome are variable. Timely and correct treatment is important avoiding residual disabilities of dementia, deafness, and blindness.

Conclusions

Comparison of the articles on Susac's syndrome published recently shows that the incidence of Susac's syndrome all around the world is dramatically increasing most likely because of the increasing apprehension of it. Unfortunately, not all specialists are familiar with this rare and comparatively new syndrome yet; therefore, it is often misdiagnosed as multiple sclerosis or acute disseminated encephalomyelitis.

By reporting this case, we would like to make this syndrome more evident to clinicians and radiologists. Being acquainted with the typical clinical and radiological presentation of Susac's syndrome, specialists should suspect and make an early and accurate diagnosis. Timely and correct treatment of Susac's syndrome can stop the progression of symptoms and markedly improve patient's recovery.

Statement of Conflict of Interest

The authors state no conflict of interest.

References

1. Susac JO, Hardman JM, Selhorst JB. Microangiopathy of the brain and retina. Neurology 1979;29:313-6.
2. Saenz R, Quan AW, Magalhaes A, Kish K. MRI of Susac's syndrome. AJR Am J Roentgenol 2005;184:1688-90.
3. Susac JO. Susac's syndrome. AJNR Am J Neuroradiol 2004; 25:351-2.
4. Susac JO, Egan RA, Rennebohm RM, Lubow M. Susac's syndrome: 1975-2005 microangiopathy/autoimmune endotheliopathy. J Neurol Sci 2007;257:270-2.
5. Rennebohm RM, Susac JO, Egan RA, Daroff RB. Susac's syndrome – update. J Neurol Sci 2010;299:86-91.
6. Susac JO, Murtagh FR, Egan RA, Berger JR, Bakshi R, Lincoff N, et al. MRI findings in Susac's syndrome. Neurology 2003;61:1783-7.
7. Rennebohm RM, Egan RA, Susac JO. Treatment of Susac's syndrome. Curr Treat Options Neurol 2008;10:67-74.
8. Magro CM, Poe JC, Lubow M, Susac JO. Susac syndrome: an organ-specific autoimmune endotheliopathy syndrome associated with anti-endothelial cell antibodies. Am J Clin Pathol 2011;136:903-12.
9. Rennebohm RM, Lubow M, Rusin J, Martin L, Grzybowski DM, Susac JO. Aggressive immunosuppressive treatment of Susac's syndrome in an adolescent: using treatment of dermatomyositis as a model. Pediatr Rheumatol Online J 2008;6:3.
10. O'Halloran HS, Pearson PA, Lee WB, Susac JO, Berger JR. Microangiopathy of the brain, retina, and cochlea (Susac syndrome). A report of five cases and a review of the literature. Ophthalmology 1998;105:1038-44.

Received 8 September 2012, accepted 22 January 2013