· Case report ·

Retinocochleocerebral microangiopathy with sudden peripheral visual field defects and hearing loss: a case of Susac-syndrome

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Abstract

• AIM: To report a case of Susac-syndrome (SS) in a 25 years old female with sudden peripheral visual field defects and hearing loss.

• METHODS: A 25 years old female with sudden peripheral visual field defects and hearing loss was reported, and the correlations between retinal findings in ultra-widefield-angiography and presented visual field defects were documented.

• RESULTS: Our patient presented the characteristic clinical triad of SS. Certain correlations between retinal findings in ultra-widefield-angiography and visual field defects could be shown.

• CONCLUSION: Ultra-widefield-angiography has been proven to be a valuable tool to detect and follow peripheral lesions of SS in clinical management.

• KEYWORDS: Susac-syndrome; widefield angiography; optomap; fundus imaging; retinal microangiopathy

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INTRODUCTION

Susac-syndrome (SS) is a microangiopathy, characterized by the clinical triad of acute or subacute encephalopathy, sensorineural hearing loss and branch retinal artery occlusion (BRAO)[1]. Since the first description by Susac *et al*[1] in 1979 about hundred cases have been reported[2-5]. Although the understanding of SS's pathogenesis is incomplete, there is growing evidence that it presents an immune-mediated endotheliopathy causing small infarcts in the brain, the cochlea, and the retina[6].

CASE REPORT

A 25-year-old afro-american female, mother of two young children (3 and 5 years old) was referred to our department due to a painless decrease in visual acuity and visual field defects in the right eye. At initial presentation the ophthalmologic history was unremarkable and no treatment had been administered. The patient reported hearing loss in the right ear starting about two years ago and suffering from increasing anxiety since months. Her general medical history showed a mild hypothyroidism treated with L-thyroxin and genital herpes infection several months ago. Laboratory work-up was normal for sedimentation rate, lipids, renal and hepatic parameters, uric acid, CRP, protein C, S, antithrombin III, homocystein, serum glucose, antinuclear antibodies (ANA), antineutrophil cytoplasmic autoantibodies (ANCA), rheumatoid factor, and antiphospholipid antibodies. Sickle cell anemia was excluded. Basal TSH was reduced to 0.15mU/L, TSH-receptor-Ab slightly elevated to 1.1κU/L, and thyreoglobulin-Ab elevated to 80κU/L. Serologic tests for detection of HSV, VZV, CMV, EBV, toxoplasmosis, lues, and Lyme disease did not reveal any signs for acute, chronic or previous infection. Visual acuity was 20/20 on both eyes, but Goldmann perimetry revealed extensive visual field defects in the inferotemporal periphery of the right eye and smaller defects in the nasal periphery of the right eye and in the infero- and superotemporal periphery of the left eye (Figure 1A). The anterior segment of both eyes was completely quiet and the vitreous showed no cells. Fundus examination revealed discrete hard exudates temporally and nasally and conspicuous peripheral ischemic retinal areas in the right eye. In the left eye more distinct ischemic retinal lesions could be seen in the temporal periphery. HRA fluorescein angiography was performed and showed peripapillary arterial hyperfluorescence in the left eye and non-perfusion in the temporal periphery of the right eye but it was not possible to obtain sufficient images for follow-up. The patient was referred to the department of neurology, where a complete neurological examination including MRI of the brain and examination of the cerebrospinal fluid was performed. MRI scan showed multiple distinct hyperdense lesions within the dorsal corpus callosum, but no typical signs of an acute inflammatory affection of the brain or multiple sclerosis. Besides the hypoacusis of the right ear and the visual field defects, clinical neurological examination and analysis of the cerebrospinal fluid did not reveal further abnormalities. Audiometry revealed a severe loss over all frequencies and absence of the stapedial reflex response in the right ear. The left ear showed no abnormalities. Given some experience with the disease at our department

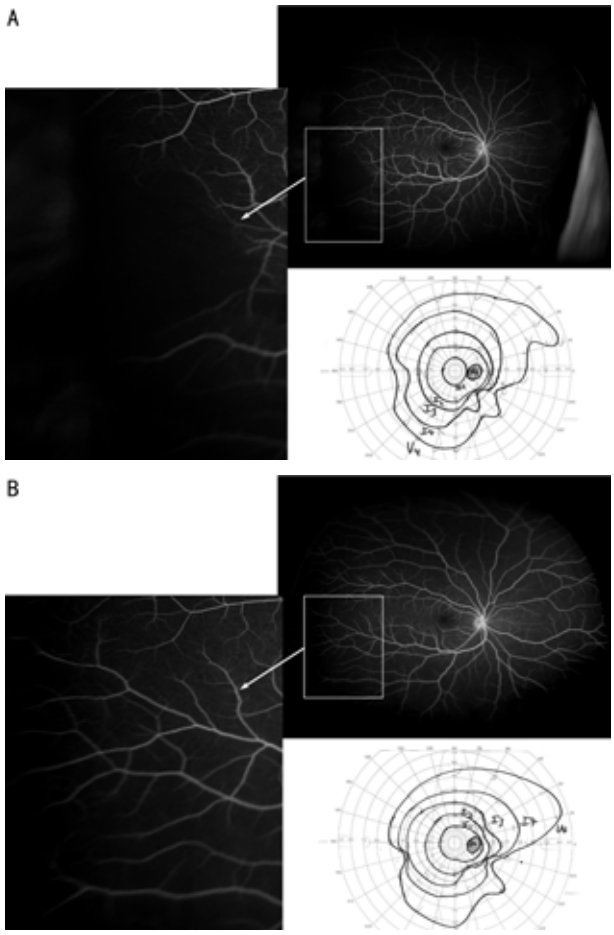

Figure 1 Optomap widefield angiography and Goldmann perimetry
A: Before treatment; B: Six weeks after systemic steroid treatment

the patient's symptoms and findings led to the diagnosis of SS. Treatment with a daily dose of 100mg prednisolone *p. o.* and ASS 100 per day was initiated. To better investigate the peripheral perfusion of the retina, a widefield angiography using a wide-field scanning laser ophthalmoscopy (Optomap P200MA, Optos, Dunfermline, UK) was performed. This wide-field examination revealed leakage from arterial walls and multiple vascular occlusions of periperal arterial branches and extensive areas of non-perfusion in the nasal and temporal periphery of the right eye (Figure 1A). Less pronounced similar lesions were observed in the left eye. The angiographic findings from both eyes correlated well with the visual field defects detected by Goldmann perimetry. After 3 days of treatment, prednisolone was reduced, as the funduscopic aspect of both eyes seemed to stabilize. One week after first presentation funduscopy still showed some hard exudates. Visual acuity was 25/20 on both eyes, and the patient still recognized the peripheral visual field loss. Six weeks after initial presentation, visual acuity remained stable at 25/20 in both eyes. The patient described some improvement in visual field disturbance. She also reported that her anxiety had become worse and weight gained was beyond 5kg. A follow-up wide-field angiography was performed that day. Angiography revealed that the majority of the previously non-perfused areas in the temporal and nasal periphery of both eyes were re-perfused now. However, some new foci of leakage, but no new areas of non-perfusion could be seen in the superior area of the right eye (Figure 1B). Goldmann perimetry revealed persisting visual field defects in the temporal and nasal periphery of both eyes (Figure 1B). After 6 months the retinal situation had stabilized in funduscopy and visual field defects remained the same. Psychiatric treatment could improve depressive symptoms with no further medication.

DISCUSSION

SS is a rare microangiopathy affecting brain, cochlea, and retina. Monocyclic, polycyclic and chronic continuous courses have been described, but in most cases it is thought to have a fluctuating self-limiting course[7,8]. Nevertheless, there are cases with a historiy over decades and residual cognitive, visual and hearing deficits have been reported[6]. The exact pathogenesis remains unclear, preliminary evidence suggests that SS represents an immune-mediated endotheliopathy, which affects the microvasculature of the brain, retina and the inner ear[6]. Most authors agree that treatment of SS requires immunosuppression. Therefore, steroidal therapy is the mainstay of treatment[6]. Other immunosuppressive drugs, such as azathioprime or cyclophosphamide, together with anticoagulation and/or aspirin have been described as useful[3,9-11]. One woman was reported to suffer from the first symptom of a SS at the age of 55[12]. In general the age of onset is between 20 and 40 years[5]. Women are more often affected than men (sex ratio 3∶1). Initially, in 97% of all patients SS does not manifest itself as the classic triad (encephalopathy, sensorineural hearing loss and BRAO)[11,13]. Therefore, the occurrence rate for this disease is estimated to be significantly higher than those cases reported and symptoms are often misdiagnosed. Differential diagnoses include multiple sclerosis, ADEM (acute disseminated encephalomyelitis), viral and bacterial infections, as well as primary or secondary vasculopathies[14,15]. In our case the diagnosis of SS could be made in collaboration with the neurology department and due to the fact, that our department had already seen SS patients before. Therefore, an early treatment was started. Nonetheless, the patient experienced hearing loss as initial feature two years before retinal and neurological manifestation. The typical ophthalmologic findings in SS are branch retinal artery occlusions (BRAO). In most previous cases patients were suffering from acute loss of central vision. In contrast, our patient had a visual acuity of ≥20/20 in both eyes. Nevertheless, distinct peripheral visual field defects due to peripheral BRAO existed. Cases with vascular occlusions located in the periphery often remain clinically asymptomatic or, as in our case, result in symptomatic visual field defects. With standard fluorescein angiography it is difficult to document changes in the outer periphery. In our case ultra-wide-field angiography with Optomap has been proven to be a valuable tool to detect and follow peripheral lesions. This helped with clinical management of this rare disease.

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视网膜耳蜗脑微血管病并突发周边视野缺损及听力损失:Susac 综合征 1 例

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摘要

目的:报告 1 例 25 岁女性患 Susac 综合征(SS),突发周边视野缺损及听力丧失。

方法:报告 1 例 25 岁女性患者突发周边视野缺损及听力丧失,并记录视网膜超广角血管造影检查结果之间的相互关系以及所展现的视野缺损。

结果:患者显现出 SS 特有的临床三联征。我们可以看出视网膜超广角血管造影检查结果和视野缺损之间的一些相互关系。

结论:经证明超广角血管造影术是一个在临床治疗中检测和追踪 SS 周围病变的有价值的工具。

关键词:Susac 综合征;广角血管造影术;欧宝全景照相系统;眼底成像;视网膜微血管病