

# A Case of Susac Syndrome

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## Abstract

Susac syndrome is a readily recognized but often misdiagnosed disorder almost exclusively affecting women in the 20- to 40-year age range. Characterized by the clinical triad of encephalopathy, branch retinal artery occlusions, and sensorineural hearing loss, patients with Susac syndrome are often misdiagnosed with multiple sclerosis (MS). Unlike MS, however, the disease process extends over a 1- to 2-year period and then goes into remission. This presentation describes the progression of symptoms of a patient eventually diagnosed with Susac syndrome.

**Key Words:** Branch retinal artery occlusion, encephalopathy, sensorineural hearing loss, Susac syndrome

**Abbreviations:** ALD = assistive listening device, EEG = electroencephalogram, MRI = magnetic resonance imaging, MS = multiple sclerosis

Susac syndrome is a little known malady that was first described in the literature in 1979 by Susac et al (1979). Although many references have most often named the syndrome microangiopathy of the brain, the term “Susac syndrome” was coined in 1986 and has become the term of choice (Susac, 1994). The syndrome is characterized by the clinical triad of encephalopathy, branch retinal artery occlusions, and sensorineural hearing loss and seems to affect women in the 20- to 40-year age range. Since its initial description, additional cases fitting the clinical profile of Susac’s syndrome have been reported (Coppeto et al, 1984; Monteiro et al, 1985; Mass et al, 1988; Li et al, 1996; O’Halloran et al, 1998). It has been suggested that Susac’s syndrome is most often misdiagnosed as multiple sclerosis (MS), primarily due to white-matter defects on magnetic resonance imaging (MRI), but has also been misdiagnosed as cardiac emboli or Meniere’s disease (Mass et al, 1988).

The pathogenesis of Susac’s syndrome is unknown, and patients may initially present with any one of the triad of symptoms (Susac, 1994). Over the course of the illness, additional

symptoms appear and may vary and fluctuate. The branch retinal artery occlusions are usually bilateral and may be either extensive or subtle. If extensive, the patient will complain of visual disorder. Hearing is most often characterized by a bilateral low-frequency rising sensorineural loss with poor speech understanding. Encephalopathy usually evolves subacutely and is associated with psychiatric features of personality change and bizarre and paranoid behavior. In addition, cases have been reported in which patients have branch retinal artery occlusions with no hearing loss; bilateral sensorineural hearing loss and bilateral retinal artery occlusions with no neurologic disease; and branch artery occlusions, hearing loss with vertigo and tinnitus, and no neurologic involvement.

Clinical findings follow a pattern over the course of the syndrome. During the encephalopathic phase, the electroencephalogram (EEG) is diffusely slow. MRI shows numerous infarcts in the gray and white matter, leading to the diagnosis of MS. During the subacute phase, the areas of infarct enhance with gadolinium. In the chronic phase, atrophy ensues and becomes a residual finding.

A variety of therapies involving the use of steroids and immunosuppressant drugs have been used to treat Susac syndrome, but results have been mixed (Monteiro et al, 1985; Susac, 1994). Some individuals appear to improve, whereas others deteriorate. Generally, by the end of 2 years, the syndrome tends to go into remis-

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sion spontaneously. Residual effects may include mild to moderate dementia with some gait disturbance, hearing loss, and visual impairment.

The following presentation describes the course of illness of an individual diagnosed with Susac syndrome.

### CLINICAL PRESENTATION

**G**S is a 43-year-old female who first presented with brief vertiginous episodes associated with a sinus infection in August 1998. The vertigo was primarily experienced when she rolled over in bed and changed positions; meclizene did not relieve the symptoms. The vertigo became progressively more and more severe, and GS started experiencing "stiffness" in her left ear, which was also attributed to the sinus infection. On Labor Day weekend, GS experienced a severe vertiginous attack, which sent her to the emergency room.

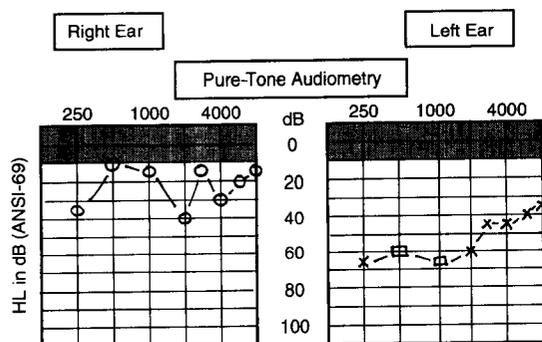
After this severe attack, GS continued experiencing episodic vertigo and noticed decreased hearing sensitivity in her left ear. A hearing test indicated hearing sensitivity grossly within normal limits for her right ear and a moderately severe rising to moderate sensorineural hearing loss for her left ear (Fig. 1). Speech understanding was poor for the left ear (44%) but was within normal limits for the right ear (100%). GS was placed on a sulfa drug, and within 24 hours of taking the drug, experienced a severe allergic reaction that caused a precipitous drop in her platelet count, mouth sores, and bruising all over her body. She was hospitalized and had a bone marrow biopsy to rule out leukemia. The biopsy was negative. At this time, she noticed decreased hearing sensitivity in her right ear.

The severe allergic reaction was addressed and her overall physical condition improved. She reported that hearing in both ears seemed to fluctuate but was improving. Over the month

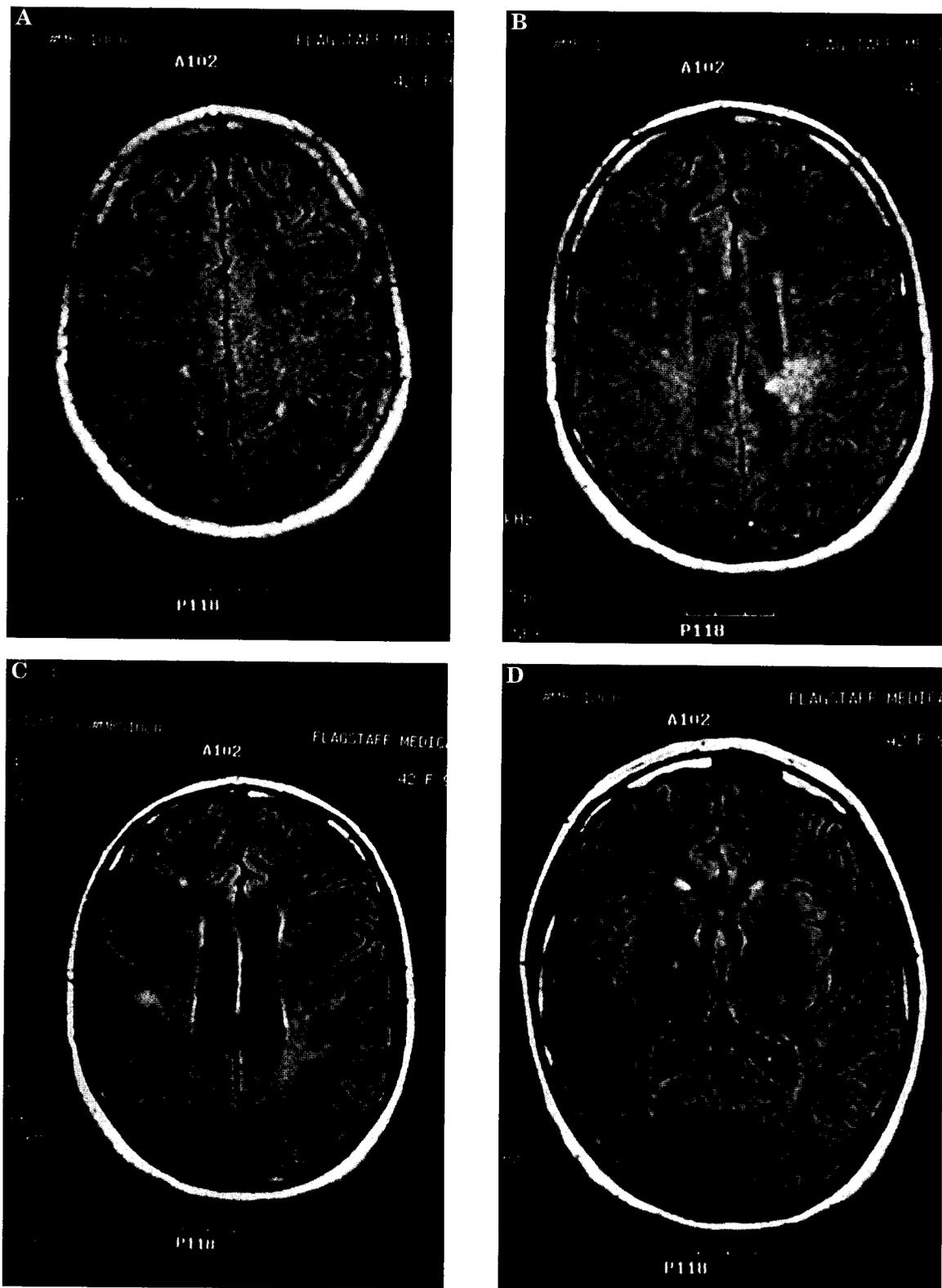
of September, she continued experiencing brief periods of episodic vertigo. She also began experiencing a general weakness. Because of her symptoms and deteriorating physical condition, an MRI scan was ordered. Results showed multiple diffuse areas of increased intensity with involvement of the corpus callosum, findings suggestive of MS (Fig. 2). However, GS did not experience typical MS symptoms of visual disturbance, diplopia, loss of vision, bowel or bladder complaints, and paresthesias or weakness. Because the MRI showed no brainstem involvement, cochlear symptoms were attributed to possible atypical Meniere's disease.

During the month of October, approximately 2 months after presentation of her initial symptoms, GS's condition seemed to stabilize, although she reported that her hearing continued to fluctuate. During this time, she began experiencing auditory hallucinations, constantly hearing repetitive music that could be "rock, heavy metal, or classical." She underwent a complete neurologic evaluation, including evoked potential testing, a repeat MRI, and a lumbar puncture to rule out MS. Results from auditory brainstem response audiometry, visual evoked potential testing, median nerve somatosensory evoked potential evaluation, and tibial nerve somatosensory evoked potential evaluation were all within normal limits. A repeat MRI scan continued to show white-matter lesions with involvement of the corpus callosum, typical of MS. However, a lumbar puncture found the presence of oligoclonal bands in both blood and cerebrospinal fluid, a finding not specific for MS.

In early November, a diagnosis of possible atypical MS was entertained, and it was decided that GS would be treated symptomatically. However, in mid-November, her condition deteriorated. Audiometric testing showed a moderate low-frequency sensorineural hearing loss, rising to a mild sensorineural hearing loss above 2000 Hz for the right ear and a severe to moderately severe sensorineural hearing loss in the left ear (Fig. 3). Speech understanding for monosyllabic words presented monitored live voice indicated extreme communication difficulties in both ears (RE: 40%, LE: 40%). GS also began noticing that she "was seeing funny." She was hospitalized for evaluation and found to have retinal branch artery occlusion of the right eye, resulting in loss of vision in the lower medial part of her right visual field. During this hospitalization, a series of tests were performed to rule out Refsum's disease, sarcoidosis, metabolic disorders,



**Figure 1** Audiogram from September 1998.



**Figure 2** MRI images: A, FLAIR axial cuts above the lateral ventricles showing hyperintensities in the bilateral cerebral hemispheres; B, FLAIR axial image just at the top of the lateral ventricles showing white-matter hyperintensities posterior to the left lateral ventricle; C, FLAIR axial image above the basal ganglia showing white-matter hyperintensities in the right frontal and bilateral parietal lobes; D, FLAIR axial image through basal ganglia showing areas of hyperintensity in the white matter of the right frontal lobe, right occipital lobe, and left parietal lobe as well as in the basal ganglia bilaterally.

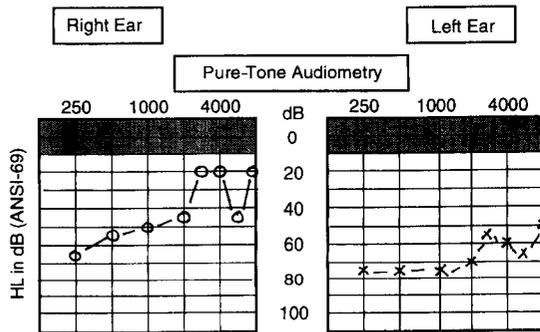


Figure 3 Audiogram from November 1998.

and porphyria. All results were negative. A repeat MRI was unchanged. A carotid Doppler was normal. A complete work-up for vasculitis, including a conventional angiogram and serum studies, was performed with no significant findings. With this plethora of tests, a disease profile did not emerge. Many abnormalities were ruled out, but no definite abnormality was identified. Diagnosis of possible MS or atypical Cogan's syndrome was considered. GS was placed on high-dose prednisone and aspirin, and her condition stabilized.

In December, GS was still on aspirin therapy and began tapering off the prednisone. She reported that her hearing was improving, especially in her right ear, and the auditory hallucinations were diminishing. Physically, she continued feeling weak. In mid-month, she developed a severe cold and sinus infection and was prescribed an antibiotic. While being treated for the cold, she woke up disoriented and weak, requiring support to walk. She exhibited a receptive and expressive aphasia and found it difficult to talk, and speech attempts revealed that her speech was slurred. She reported that she could hear people ask questions but was unable to respond verbally. She was unable to follow commands and unable to write. She was immediately hospitalized for possible stroke secondary to vasculitis versus MS presentation. A repeat MRI showed no evidence of stroke; white-matter lesions were unchanged. Selective bilateral carotid and vertebral arteriography showed no evidence of aneurysm or vasculitis, normal basilar artery, and normal selective cerebral arteriography. EEG recordings revealed mild to moderate generalized cerebral dysfunction, which is a nonspecific finding associated with toxic/metabolic encephalopathy, hypoxia/ischemic encephalopathy, trauma, hemorrhage, infection, and neurodegenerative disorders. The stroke-like

symptoms persisted for approximately 24 hours and then subsided. GS was discharged from the hospital and continued taking aspirin; prednisone was tapered.

Since this hospitalization, GS's condition has stabilized and begun to improve, and she is experiencing increased stamina and strength. She is still missing part of her visual field in her right eye but has, fortunately, experienced no further deterioration. Although she reports that her hearing fluctuates, repeat testing has shown that hearing has remained essentially unchanged, showing a moderate low-frequency sensorineural hearing loss, rising to a mild sensorineural hearing loss above 2000 Hz for the right ear and a severe to moderately severe sensorineural hearing loss in the left ear (Fig. 4). Speech understanding scores indicate severe difficulty understanding speech in both ears (RE: 68%, LE: 20%). The subjective fluctuations in her hearing may be related to fluctuations in speech comprehension.

## COMMENTS

In summary, GS is a 43-year-old female with branch retinal artery occlusion, cerebral white-matter changes on MRI, and bilateral sensorineural hearing loss. The triad of symptoms support a diagnosis of Susac syndrome. Approximately 46 cases of Susac syndrome have been reported in the literature (O'Halloran et al, 1998); however, because of the extreme difficulty in diagnosing the syndrome, it is possible that individuals with Susac syndrome have been misdiagnosed as atypical MS. Steroids have not been shown to be effective therapy for this disorder, so the improvement observed in GS may indeed be fortuitous. Certainly, immunomodulating therapies for MS (interferon- $\beta$ -1a [Avonex],

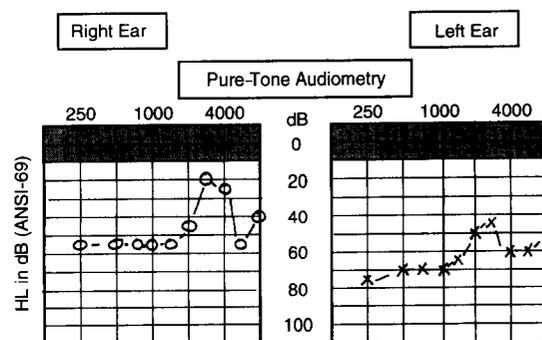


Figure 4 Audiogram from October 1999.

interferon- $\beta$ -1b [Betaserone], and glatiramer acetate [Copaxone] are not indicated.

GS has been an audiologic mystery. Throughout the course of her illness, she has reported fluctuations in her hearing, with better hearing some days and poorer hearing other days. However, these subjective fluctuations cannot be documented with repeat audiologic measures. Over the past 11 months, pure-tone thresholds and speech understanding scores have remained essentially unchanged. It is possible that this varying comprehensibility with no change in objective audiometric findings is due to mild encephalopathy. Full neuropsychometric testing has been arranged to explore this possibility.

GS has been involved in extensive aural rehabilitation counseling regarding amplification. Before hearing aids were considered, realistic expectations of amplification were discussed, and the limitations imposed by her poor speech understanding scores were explained. GS had hoped that hearing aids would "cure" her hearing disability, so it was necessary to explain that to optimize auditory input, she would need to develop her speechreading skills and incorporate communication strategies. Once GS had an understanding of amplification, binaural digital hearing aids were dispensed. Although she reports that in most situations she can hear people when she is wearing her aids, she has great difficulty understanding people because speech is "muffled" and "not tuned." Some days she reports that she is able to understand most women, whereas other days she cannot understand even familiar women. She always reports difficulties understanding men. Different settings have been programmed into her digital hearing aids, but an optimum fitting has not been established. It is possible that the subjective fluctuations she reports unaided are also evident in the aided condition. Efforts continue to be made for an acceptable fitting, and counsel-

ing regarding realistic expectations of hearing aid use is being reinforced.

GS has experienced success using assistive listening devices (ALDs). She uses an amplifier on her telephone and reports few communication problems. She has started using an infrared system at church and reports that when she looks at the minister and listens through the ALD, she can follow most of the service. GS is extremely motivated to maximize auditory input and experience successful communication.

GS continues to be followed medically by her neurologist, ophthalmologist, and otolaryngologist. Her symptoms seem to have stabilized, and, in many ways, her physical condition is improving. She is beginning to work part time at home and is hoping to eventually be fully employed.

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