Susac’s syndrome in a patient with ulcerative colitis

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Abstract

The Susac’s syndrome is a rare disorder that was first described in 1979 and is characterized by a classic triad consisting in encephalopathy, visual impairment and sensorineural hearing loss. However, the etiology of the disease is still unclear. We report the case of a 29-year-old with ulcerative colitis treated with mercaptopurine, six months before to her admission started with personality changes attributed to symptoms of depression who subsequently present neurological symptoms characteristic of Susac’s Syndrome. In the literature there is no clear association between inflammatory bowel disease and Susac’s syndrome, but this case is presented in order to emphasize the simultaneous presentation of these two diseases with a tendency to vasospasm and an autoimmune pathogenesis.


Introduction

Susac’s syndrome is a rare disorder that was first described in 1979 by Susac et al.¹. It is characterized by a classic triad consisting of encephalopathy, visual deficit and sensorineural hearing loss². However, the etiology of the disease is not yet clear³. It is characterized by microangiopathy in the arterioles with retinal artery recurrent occlusion, and it affects mainly women and middle-aged people⁴. Although the understanding of its immunopathogenesis is incomplete, preliminary evidence suggests that antibodies to endothelial cells can play a highly important role in the development of this pathology, since it reflects the damage produced on endothelial cells⁵,⁶. The characteristics of endotheliopathy include endothelial cell swelling, to the point of occluding the lumen; endothelial cell degeneration, necrosis and detachment; intraluminal fibrin deposit; endothelial cell-deprived vessels, capillary drop-out; vascular ectasia; thickening of the basement membrane, reduplication and granular lamination; C5b-9 deposit in the blood vessels and perivascular lymphocytic infiltration⁷. Other authors have postulated that Susac’s syndrome is a special manifestation of primary vasospastic syndrome that cannot be attributed to other autoimmune disease, since these patients often have a history of cold hands, low blood pressure and migraine⁸. Most patients with ulcerative colitis and Crohn’s disease suffer acral vasospasm shown by capillaroscopy and increase in endothelin 1 values. This vasospasm can cause vascular retinal diseases and ophthalmic artery occlusion¹¹,¹². We have not found a clear association between inflammatory bowel disease and Susac’s syndrome in the literature, and this is why a case is presented in order to stress on the simultaneous presentation of both these diseases with a tendency towards vasospasm and an autoimmune pathogenesis the association of which is relatively rare.

Case report

We present the case of a 29-year-old female with ulcerative colitis, treated with mercaptopurine, who 6 months before her admission started with personality changes attributed to symptoms of depression who subsequently present neurological symptoms characteristic of Susac’s Syndrome. In the literature there is no clear association between inflammatory bowel disease and Susac’s syndrome, but this case is presented in order to emphasize the simultaneous presentation of these two diseases with a tendency to vasospasm and an autoimmune pathogenesis.
changes attributed to an episode of depression. Three months later, she experienced headache, pain and right ear hearing loss, which prompted her family doctor to prescribe paracetamol, with symptoms remitting. Five days prior to her admission she had episodes of confusion, headache, hypoacusis, disorientation and slow thinking.

On physical examination, the patient was disoriented in time and had difficulty to remember recent events. She had pyramidal syndrome on the right extremities, bilateral clonus, unstable gait and multiple petechial lesions that were observed on lower extremities. Cranial tomography was normal. Cerebrospinal fluid showed proteinorachia (160 mg/dL). Regular laboratory tests were normal.

Acute meningoencephalitis was integrated as diagnostic suspicion and empirical treatment for immunosuppressed patients with meropenem, vancomycin, ampicillin and acyclovir was started. During hospitalization she had delusions of grandeur (megalomania). Brain magnetic resonance imaging showed multiple hyperintense lesions distributed in the white matter of both hemispheres of the brain and cerebellum, including the corpus callosum (Fig. 1) and an ischemic infarction in the left cerebral hemisphere on the visual pathway and internal capsule (Fig. 2). The electrocardiogram discretely showed a deceleration, with frequent delta wave surges and sporadic sharp peaks in the temporal and frontal regions. There were no paroxysms or signs of status epilepticus, which suggested diffuse, unspecific cerebral compromise. The skin biopsy showed normal-looking epidermis and dermis with a discrete inflammatory infiltrate and lymphocytes, polymorphonuclear leukocytes and some eosinophils, around vascular structures with organized vascular thrombi. Ophthalmologic examination showed retinal edema and disperse vascular occlusions without areas of ischemia. Otolaryngologic examination showed sensorineural hearing loss of vascular etiology. The patient underwent all laboratory and imaging tests to rule out other etiologies (Table 1). Susac’s syndrome was diagnosed and treatment was started with corticosteroids, with the patient showing cognitive and neurological improvement and therefore being discharged from hospital.

Discussion

We present a case of Susac’s syndrome, a disease that was first described in two women with encephalopathy, retinal arterial occlusions and deafness. It is underdiagnosed and predominantly affects middle-aged healthy women. In our case, the classic triad was present, it is not this way in all cases. Vasospastic occlusions can play a crucial role in the pathogenesis of this syndrome. These occlusions are also observed in patients with ulcerative colitis, and our hypothesis is that this condition might increase the risk for syndromes such as vasospastic syndrome and Susac’s syndrome, secondary to endothelial dysfunction and the pro-coagulant state observed in these patients. However, there are no studies supporting this hypothesis and we didn’t find any other case reporting both conditions together.

This patient was previously treated with mesalazine and infliximab. Treatment with 5-aminosalicylic acid-derived agents has been associated with adverse effects such as myocarditis, nephritis and vasculitis,
and therefore id cannot be associated with the real pathology. Tumor necrosis factor inhibition with infliximab has been reported to be beneficial for the treatment of this syndrome.

Another data that is important mentioning is that our patient was immunodepressed at the onset of symptoms, and taking into account that the main theory about Susac’s syndrome pathogenesis is that it is an autoimmune disease, this drives us to consider that this is not the only implicated mechanism. In our opinion, this is the first published case of this syndrome in a patient of these characteristics who was receiving immunosuppressant therapy.

Susac’s syndrome can mimic several diseases and, as in our study, it is necessary for exhaustive studies to be carried out in order to exclude other conditions’ encephalopathy symptoms15. This syndrome can be masked by visual and auditory compromise, as in our patient, and lead to rather think in a neuropsychiatric process. Hearing loss is often acute, unilateral and associated with tinnitus, vertigo and nystagmus21,22. Audiometry demonstrates sensorineural hearing loss attributed to cochlear infarction22.

There are several treatments that have been used for the management of this syndrome; corticosteroids, immunosuppressant agents, platelet antiaggregants, antithrombotic agents, intravenous immunoglobulins, plasmapheresis and even hyperbaric oxygen, but the rareness of the disease make it very difficult for their efficacy to be assessed2. It appears that in patients whose diagnosis and treatment is delayed, permanent morbidity is higher in terms of neurological deficit and visual and hearing loss21.

Table 1. Summary of differential diagnoses that were ruled out prior to establishing the Susac’s syndrome diagnosis

<table>
<thead>
<tr>
<th>Tests</th>
<th>Result</th>
</tr>
</thead>
<tbody>
<tr>
<td>Other autoimmune conditions (lupus, arthritis and spondylitis)</td>
<td>Negative</td>
</tr>
<tr>
<td>Onconeural antibodies</td>
<td>Negative</td>
</tr>
<tr>
<td>Serology for cytomegalovirus, Epstein-Barr, human immunodeficiency virus, Treponema pallidium, hepatitis A, B and C virus and Toxoplasma gondii</td>
<td>Negative</td>
</tr>
<tr>
<td>Cryoglobulins</td>
<td>Negative</td>
</tr>
<tr>
<td>Blood and cerebrospinal fluid cultures</td>
<td>Negative</td>
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<tr>
<td>Polymerase chain reaction in cerebrospinal fluid for cytomegalovirus, Epstein-Barr, human herpes virus 7 and 8 and enterovirus</td>
<td>Negative</td>
</tr>
<tr>
<td>Electrocardiogram, echocardiogram, chest radiography</td>
<td>Normal abdomen and chest tomography</td>
</tr>
</tbody>
</table>

References