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CASE REPORT

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Susac syndrome with cognitive impairment - case report of 31-year-old woman

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Abstract

Introduction: Susac syndrome (SuS) is a rare immune-mediated disease caused by occlusions of microvessels in the brain, retina and inner ear. It is characterized by the clinical triad of encephalopathy, visual disturbances and hearing loss. The diagnosis of SuS is based mainly on the clinical symptoms and the supporting tests in which characteristic changes occur in the MRI.

Case report: Here, we present a case of a patient with possible SuS with psychiatric symptoms. A 31-year-old woman was admitted to the Department of Psychiatry due to deteriorating mental health for several weeks manifested as a negative mood and suicidal thoughts. During hospitalization, a neurological consultation was conducted, in which the patient was identified as conscious, psychomotor sluggish, with impaired verbal contact and persistent hearing and vision impairment. Cross-sectional assessment of cognitive functions revealed that the patient had a generalized syndrome of neuropsychological deficits, which confirms the diagnosis of dementia.

Discussion: This case summary provides an example of a woman diagnosed with SuS manifested as a cognitive impairment with associated vision and hearing deterioration. It is worth emphasizing the fact that such presentation of the triad of at disease onset is rare. The characteristics of the organic changes in the brain described in the MRI probably explain the symptoms described in that case. The prognosis of SuS depends on early diagnosis and treatment.

Conclusions: A diagnosis of SuS should always be considered in the presence of nonspecific neuropsychiatric symptoms and progressive multifocal neurological symptoms, hearing loss, and visual impairment. An important fact is that the typical triad of SuS symptoms in most cases does not occur simultaneously, which makes the diagnostic process very difficult and may lead to misdiagnosis.

Keywords: Susac's syndrome, cognitive impairment, hearing loss, visual disturbances encephalopathy

Streszczenie

Wstęp: Zespół Susaca (SuS) to rzadka choroba o podłożu immunologicznym, spowodowana niedrożnością naczyń w mózgu, siatkówce i uchu wewnętrznym. Charakteryzuje się kliniczną triadą objawów - encefalopatią, zaburzeniami widzenia i utratą słuchu. Rozpoznanie SuS opiera się głównie na objawach klinicznych i testach pomocniczych, w których występują charakterystyczne zmiany w MRI.

Opis przypadku: Przedstawiamy przypadek pacjenta z SuS z towarzyszącymi zaburzeniami poznawczymi. 31-letnia kobieta została przyjęta do Kliniki Psychiatii z powodu pogarszającego się stanu zdrowia psychicznego objawiającego się obniżonym nastrojem i myślami samobójczymi. W trakcie hospitalizacji przeprowadzono konsultację neurologiczną, w której pacjentkę opisano jako spowolniałą psychoruchowo, z zaburzonym kontaktem werbalnym oraz utrwalonym upośledzeniem słuchu i wzroku. Przekrojowa ocena funkcji poznawczych wskazała na występowanie u pacjentki uogólnionego zespołu deficytów neuropsychologicznych, co potwierdza rozpoznanie otępienia. Charakterystyka zmian organicznych w mózgu opisane w MRI

prawdopodobnie wyjaśniają objawy opisane w tym przypadku.

Dyskusja: Rokowanie w SuS zależy od wczesnej diagnozy i leczenia. Rozpoznanie SuS należy zawsze brać pod uwagę w przypadku niespecyficznych objawów neuropsychiatrycznych i postępujących wieloogniskowych objawów neurologicznych, utraty słuchu i zaburzenia widzenia.

Podsumowanie: Warto podkreślić fakt, iż typowa triada objawów SuS w większości przypadków nie występuje jednocześnie, co bardzo utrudnia proces diagnostyczny i może prowadzić do błędnej diagnozy.

Słowa kluczowe: Zespół Susaca, zaburzenia poznawcze, ubytek słuchu, encefalopatia

Introduction

Susac syndrome (SuS) is a rare immune-mediated disease caused by occlusions of microvessels in the brain, retina and inner ear. SuS was first described by John Susac in 1979. It is characterized by the clinical triad of encephalopathy, visual disturbances and hearing loss [1-3]. The disease is probably caused by an autoimmune process leading to microangiopathy of precapillary arterioles of the brain, retina and inner ear [4]. It should be emphasized that the clinical symptoms are variously presented and the entire triad occurs in less than 20% of patients at the onset of the disease [4-5]. There is limited data available regarding the prevalence and incidence of SuS. The annual incidence was recently estimated in Austria to be 0.024/100000. The largest case series in Israel includes 10 patients diagnosed over a 26-year period [6]. Most of the patients affected by SuS is between 20 and 40 years old. A higher incidence was noted in women than in men [7]. The diagnosis of SuS is based mainly on the clinical symptoms and the supporting tests in which characteristic changes occur in the MRI. The most difficult element of the triad in SuS to recognize are visual disturbances because they are usually characterized by reduced visual acuity and mild visual field disturbances. Here, we present a case of a patient with possible Susac's syndrome with psychiatric symptoms [1-3].

Case report

A 31-year-old woman was admitted to the Department of Psychiatry due to deteriorating mental health for several weeks manifested as a negative mood. The patient reported suicidal thoughts - she thought about jumping out of the building. In addition, aggression towards family members and prolonged sleep latency.

At the time of admission to the ward, the patient was loud and aggressive with incomplete allopsychic orientation, in a dysphoric mood and with increased psychomotor drive. She did not utter delusional content and denied hallucinations. In the mini-International Neuropsychiatric Interview (MINI) scale, she obtained 0 points, which means a low risk of suicide.

At the beginning of hospitalization the patient had increased psychomotor drive, alternating mood and maladjusted affect. She was presenting an accelerated way of thinking and dissociation. The patient was presenting persecutory delusions which were multi-threaded, lengthy, bizarre, illogical explanations of cause-and-effect relationships and dissimulation. The patient denied experiencing any unusual experiences, such as visual and auditory hallucinations. She was ignoring her current life situation (unemployment) and denied the need for hospitalization and treatment. She was presenting a delusional attitude towards the family.

Due to the pharmacotherapy and psychotherapy the improvement of condition has been observed, such as remission in psychotic symptoms, psychomotor alignment, relief of anxiety and general improvement in the functioning of the patient.

During further hospitalization the patient was calm, keeping her affect adjusted with clear awareness, balanced mood and psychomotor drive. Psychotherapy and motivating interviews aimed at improving cooperation in treatment were carried out. Due to the persistent lack of cooperation, the drug - aripiprazole was included in the form of a depot.

During hospitalization, a neurological consultation was conducted, in which the patient was identified as conscious, psychomotor sluggish, with impaired verbal contact and persistent hearing and vision impairment. Besides, a physical examination without deviations. Laboratory tests were also conducted - morphology, immunochemical and biochemical tests. There was no significant aberration.

Cross-sectional assessment of cognitive functions revealed that the patient had a generalized syndrome of neuropsychological deficits, which confirms the diagnosis of dementia, as in the July 4 study. On the Addenbrooke's Cognitive Examination-Revised (ACE-R) scale, the patient obtained a total of 59 points, i.e. over 20 below the lowest cut-off point on this scale.

Proportionally the lowest results were for the memory and verbal fluency subscale. Also, the performance of the bvrt test, in which the patient did not correctly reproduce a single pattern and made a total of 15 errors, clearly indicates advanced disorders of learning and control of their own cognitive activity. These results demonstrate the progressive nature of the cognitive

impairment associated with the underlying disease and justify the diagnosis of moderate dementia.

In the white matter of both hemispheres of the brain in the periventricular location, poorly delimited high-signal areas are visible in T2-weighted and fluid-attenuated inversion recovery (FLAIR) images of an ambiguous nature, requiring differentiation between ischemic and chronic lesions and demyelinating lesions. Low-height corpus callosum with diffuse hypoxic-ischemic high-signal areas. Attention is drawn to the age-inadequate disappearance of the gray matter of both cerebellar hemispheres. Bilateral complexes of nerves VII and VIII, unchanged on the MRI image. The ventricular system is symmetrical, not widened, not displaced. There were no symptoms of increased intracranial pressure. No pathological contrast enhancement was found after intravenous injection of a paramagnet. MRI shows normal meningeal morphology.

On the day of discharge, the patient remained clearly conscious, autopsychically and allopsychically oriented correctly, in logical verbal contact, adjusted in behavior, in an even mood and psychomotor drive. She denied the presence of productive symptoms, negated the occurrence of suicidal thoughts and tendencies, self-aggressive and heteroaggressive tendencies.

Discussion

This case summary provides an example of a woman diagnosed with Susac's syndrome manifested as a cognitive impairment with associated vision and hearing deterioration. It is worth emphasizing the fact that such presentation of the triad of symptoms (central nervous system, eye and ear symptoms) at disease onset is rare. Dörr et al. found that only 13% of patients with available data had the characteristic clinical triad of Susac syndrome at disease onset [8].

The characteristics of the organic changes in the brain described in the MRI (callosal lesion, T2-hyperintense lesions of white matter) probably explain the symptoms described in that case. One of the most characteristic signs of Susac syndrome in MRI study is involvement of corpus callosum [9]. Analysis of available clinical trials shows that callosal lesions occur in 78% of SuS cases diagnosed with MRI [8]. The encephalopathic changes can manifest as memory impairment, confusion, behavioral disturbances, ataxia, dysarthria, paranoid psychosis, occasional mutism, and headaches [10]. On the other hand, Machado et al. did not find strong association between MRI markers and cognitive and behavioral measures. This fact may question the pathogenesis underlying the brain tissue lesion in this disorder [11].

Due to the probable immune-mediated etiology of SuS with precapillary arterioles damage of the brain, retina and middle ear, the key is to apply early immunomodulatory treatment that significantly improves the prognosis. First-line recommended drugs are glucocorticosteroid. Among the alternatives we can distinguish IVIG or plasmapheresis of immunosuppressive drugs such as azathioprine, cyclosporine, cyclophosphamide, mycophenolate mofetil, methotrexate and rituximab [12]. Vodopivec et al. additionally mention antiplatelet therapy and nifedipine as a supportive therapy [13]. Rennebohm et al. in updated guidelines for treatment of Susac syndrome underline the fact that the clinician must balance fear of inadequately suppressed disease with fear of potential side effects of aggressive immunosuppression [14].

SuS requires differentiation from multiple sclerosis and acute disseminated encephalomyelitis due to the similarity of autoimmune etiopathogenesis, clinical symptoms and the presence of multifocal lesions in brain MRI [15]. The prognosis of SuS depends on early diagnosis and treatment. There are case reports in which the early implementation of treatment resulted in a significant improvement in the clinical condition, leaving only a slight neurological deficit. However, in most cases, appropriate therapy is significantly delayed, leading to persistent symptoms damage to the nervous system, such as cognitive impairment, hearing impairment and visual disturbances [7].

Conclusions

A diagnosis of SuS should always be considered in the presence of nonspecific neuropsychiatric symptoms and progressive multifocal neurological symptoms, hearing loss, and visual impairment. Rarity of the disease and the lack of specific diagnostics are the reasons for the lack of optimal treatment of SuS. An important fact is that the typical triad of SuS symptoms in most cases does not occur simultaneously, which makes the diagnostic process very difficult and may lead to misdiagnosis. The above-mentioned arguments confirm that Susac syndrome requires further development of pathogenesis, which will allow for the development of optimal diagnostics and treatment strategies.

Conflict of interest

The authors have declared no conflict of interest.

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