

Years of Frustrating Symptoms, Multiple Tests: Neurosarcoidosis is the Etiology – A Case Report

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Abstract

Neurosarcoidosis is a rare, chronic inflammatory disease that affects the nervous system. It is diagnosed in 5 to 10% of sarcoidosis patients, but has been found in 25% of autopsies. This suggests that a large percentage of patients are asymptomatic or misdiagnosed. Diagnosis is difficult due to variable expressivity and overlap of symptoms with other neurological disorders, such as multiple sclerosis. Moreover, diagnosis proves challenging because currently, the most specific test for neurosarcoidosis requires a histological biopsy confirmation of the affected neural tissue. This presents with its own complications. Due to the lack of fixed diagnostic methods and symptom overlap, a multidisciplinary approach is recommended in diagnosing a patient with possible neurosarcoidosis. This case describes the difficulty in making the diagnosis and the persistence of the patients physicians.

Keywords: Neurosarcoidosis • Inflammatory disease • Multiple sclerosis

Introduction

Neurosarcoidosis, a neurological manifestation of systemic sarcoidosis, is a devastating disease. The vague and ambiguous symptoms of neurosarcoidosis require extensive effort and evaluation. With symptoms resembling Multiple Sclerosis (MS), a family history, and the increased incidence of MS in women, the initial diagnosis for our patient pointed to MS. A diagnosis of exclusion, sarcoidosis can be difficult to diagnose. Multiple tests, including but not limited to imaging, laboratory tests, and EMGs, must be done to rule out other more common diseases like Parkinson's, Alzheimer's, or other movement and sensory disorders. Lastly, the high risk neural biopsy, a confirmatory test for neurosarcoidosis, is rarely done. Luckily, for this patient, as the disease progressed, symptoms changed, astute physicians reevaluated the initial diagnosis and concluded the problem was neurosarcoidosis and not MS.

Case Presentation

In 2010, a 60-year-old female presented with paraesthesia of her face, arms, and legs associated with lethargy and weakness. Cardiac evaluation noted idiopathic total ventricle occlusion of unknown etiology, and a pacemaker was placed. She now noted paresthesias along the right temporal region of her scalp and legs, impacting her walking and worsening with any activity over fifteen minutes diagnosed as neurogenic claudication. About this time her sister was diagnosed with MS. She continued to live with these unexplained, recurrent episodes of numbness and tingling. An evaluation for a sudden twenty-five-minute episode of right hemiparesis, involving her head, face, trunk, and limbs in 2014 led to an unremarkable head CT and a concern for a possible Transient Ischemic Attack (TIA). Her evaluation included a CT angiogram, Sedimentation Rate (ESR), Creatinine Phosphokinase (CPK), and

an EMG with nerve conduction studies. With normal results ruling out vascular stenosis, temporal arteritis, lumbosacral polyradiculopathy, polyneuropathy, myopathy, and mononeuritis multiplex.

Without remarkable findings, the neurologist discussed more possibilities with the patient regarding the etiology of her symptoms, including the remote chance of spinal stenosis, seizure events, or migraine aura without headache. The patient proceeded with a trial of antiseizure medication followed by a tricyclic antidepressant to determine if the symptoms were from migraine auras. Neither was helpful which led to a CT myelogram to look for spinal stenosis. Though the test did note severe degenerative changes involving the lumbar spine, there was no significant spinal canal compromise, negating the spinal stenosis theory. Four months later, she presented with acute diplopia. The CT of the orbits noted abnormal, wispy attenuation of the intraorbital retrobulbar or intraconal fat suspicious for orbital pseudotumor, lymphoma or sarcoidosis. After four years of symptoms, this was the first suggestion of sarcoidosis. Systemic sarcoidosis was diagnosed after a chest CT demonstrated granulomas and lumbar puncture demonstrated inflammatory spinal fluid, ruling out multiple sclerosis due to the lack of oligoclonal bands and an IgG index. With a presentation of transverse myelitis with a sensory level in the mid thorax later in her course, her diagnosis of neurosarcoidosis was confirmed, almost 5 years after her initial symptoms.

Discussion

The clinical manifestation of neurosarcoidosis varies because multiple levels of the central nervous system may be afflicted. The symptoms may be monophasic disease presenting with only cranial nerve damage to a more widespread disease leading to peripheral neuropathy and myopathy [1]. Moreover, an accurate diagnosis proves challenging because the only current definitive test for neurosarcoidosis requires histological confirmation from a biopsy of the affected neural tissue. Due to their location, neural biopsies are not as easily performed and are typically considered in those who have an established diagnosis of systemic sarcoidosis [2]. However, systemic sarcoidosis can also be a challenging disease to diagnose. Neurosarcoidosis can be asymptomatic in up to 40% of patients [2]. As in this patient, it is likely that if a patient with subclinical sarcoidosis that has not been diagnosed, presents with neurological symptoms, more common neurologic disorders will appropriately be considered. Blood tests and imaging are components of the diagnostic process, but may fail to distinguish neurosarcoidosis from other similar-presenting diseases. Imaging, such as chest CTs, PET scans,

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or abdominal ultrasounds, may be nonspecific for neurosarcoidosis, but can certainly help confirm systemic sarcoidosis. MRIs are useful, though findings may resemble those found in Multiple Sclerosis (MS) and thus may elicit some confusion and ambiguity. Although blood tests are of limited benefit for diagnosis, evaluation of Cerebrospinal Fluid (CSF) is useful [3]. The ambiguity of imaging, the need for histopathological biopsy, and the possible utility of CSF evaluation demonstrate the necessity of a neurosarcoidosis diagnosis utilizing various diagnostic modalities.

Conclusion

Though the process was time-consuming and frustrating, the patient eventually received closure with her diagnosis, allowing her to access accurate information, specialized physicians, and support groups. For years, the physical and emotional toll of her symptoms weighed on her without any offered rationale or community. However, once reaching a diagnosis, she was able to obtain support from others with sarcoidosis and physicians who could

help her manage her symptoms as they present. Her experience highlights the importance of diagnosis both clinically and personally. Though physicians could have treated symptoms as they manifested, the patient's comfort and reassurance came from a named diagnosis for the neurological symptoms she experienced.

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