Tumefactive Multiple Sclerosis: a Case Analysis
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Abstract

Multiple sclerosis is a chronic autoimmune disease that over time destroys the connections between neurons. The disease presents itself differently and uniquely among all cases. The pathology of the disease is not linear and can be quite difficult to diagnose and treat. One particularly rare case of MS is tumefactive MS characterized by large areas of destroyed tissue. This study would look at the clinical presentations of tumefactive MS patients and how to effectively look at epidemiology and pathology.

Introduction

Multiple Sclerosis (MS) is a complex chronic autoimmune disease of the central nervous system (CNS), characterized by demyelination of neurons, forming lesions, plaques and scarring of the myelin sheath. These plaques cut off electrical impulses being sent between neurons to communicate actions, resulting in a wide variety of disabilities. Early symptoms can include fatigue, vision problems, cognitive issues, and in severe chronic cases, can lead to paralysis. This process of demyelination is followed a process of remyelinating, where the site “heals” itself (and symptoms temporarily go away) but not without leaving residual damage (Fig. 1 and 2). There is a spectrum of types of MS, the most common being Relapsing Remitting, which effects about 90% of multiple sclerosis patients. Women seem to be at greater risk for developing the disease with a 3:2 ratio, with MS affecting roughly 2.5 million people worldwide. The specific cause of multiple sclerosis is unknown; however, several genetic and environmental factors have been linked to the disease: chronic stress, smoking, lack of vitamin D, exposure to the Epstein-Barr virus, etc. The typical age of onset is around 27 years old, but diagnosis can generally occur anywhere from 15-40 years old. Tumefactive MS is the presence of a lesion (where the myelin has been damaged or destroyed), that is greater than 2 centimeters in diameter. Misdagnosis is very common with a lesion like this, because not only can it mimic a malignant glioma or cerebral abscess, but also on biopsy can be misidentified as a neoplasm. Because of the overly common misdiagnosis, tumefactive MS is not treated properly and can develop severe symptoms quickly.

Review of Literature

A Case Control Study of Association between Socio-Demographic, Lifestyle, and Medical History1 Ghadirian, P, et al.
Roughly 400 people, half being controls, and half having MS, were looked at for the influences of genetic and environmental links of developing MS. A significant risk was observed between developing Multiple Sclerosis and smoking 20-40 cigarettes a day. Cases of patients with exposure to house animals like cats, dogs, and birds for several years prior to diagnosis were higher compared to the controls (Fig. 3). The reasons for this could be that these animals are involved in exogenous causative agents of disease. Concordance was also seen as a possible links, due to its possible disruption of the blood-brain barrier, which is how immune cells get to the neuron (Fig. 4).

Na MRI reveals persistent sodium accumulation in tumefactive MS lesions2 Huhn, Konstantin, et al.
Shared features between enlarged MS lesions, tumefactive demyelination and “Balo-like” lesions make them hard to differentiate on MRI. NaMRI is a way of looking at sodium accumulation. MS patients have displayed higher sodium levels within acute inflammatory lesions. This study looked at their relationship to tumefactive lesions. Tumefactive demyelination showed elevated sodium levels, consistent with other research, however, these enlarged lesions showed the highest levels of sodium accumulation in contrast. With intervention, acute MS lesions begin to decline until plateau is reached, whereas even 5 weeks after intervention of the tumefactive lesions, sodium levels stayed elevated despite reconstruction of the blood-brain barrier.

Fig. 3 and 4 show the results that many of the variables tested, (including exposure to disease, having domestic animals, and smoking) are correlated with the development of multiple sclerosis.

Fig. 5 shows the linear regression evaluated between childhood stress and age of onset of MS.

Fig. 6 shows the scatter of immunolabelling in the superior aspect of the left parietal lobe.

Fig. 1: The healthy brain, vs. a brain with damaged neurons from MS. The whiter areas show the sections scans, also known as “slices.”

Fig. 2: A comparison between a healthy brain, vs. a brain with damaged neurons from MS. The whiter areas show the sections scans, also known as “slices.”

Fig. 3: A Case Control Study of Association between Socio-Demographic, Lifestyle, and Medical History.1 Ghadirian, P, et al.

Fig. 4: A Case Control Study of Association between Socio-Demographic, Lifestyle, and Medical History.1 Ghadirian, P, et al.

References


Potential Methodology

With the patient charts and work done through the Multiple Sclerosis Comprehensive Care Center (a part of the NYU Langone Medical Center), an analysis of cases will be conducted, looking at patient demographics, diagnoses, MRI, and biopsy. From this, we will look into the relationships displayed between several different factors such as: initial diagnosis and/or potential misdiagnosis, size of lesions and plaques, and other variables that may come up.

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