Tumefactive Demyelination of 20 Patients: Retrospective Case Series and Review of Literature

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ABSTRACT

Tumefactive demyelination, characterized by lesions >2cm, is a rare presentation of an inflammatory demyelinating disease. Theses clinical and radiographic features can mimic neoplasms (glioma or lymphoma) or infections (cerebral abscesses, parasitic cysts) on MRI. These overlapping characteristics makes the diagnosis difficult in the very few patients presenting with these lesions. Our objective in this case series is to look at the clinical presentations of tumefactive demyelination. An analysis of about 20 cases will be conducted, looking at patient demographics, clinical presentation, diagnoses, MRI, treatment methods, and follow-up.

INTRODUCTION

Tumefactive demyelination (See figure 1 for diagram and explanation of demyelination) is an atypical presentation of an inflammatory demyelinating disease (IDD) in the central nervous system (CNS) characterized by pseudo tumoral lesions (Figure 2), also known as tumefactive demyelinating lesions (TDL). Lesions are characterized as tumefactive when they are greater than 2cm in diameter[1][2][3].

The features of TDL's can often mimic a glioma (Figure 3), cerebral abscess (Figure 4), or other infections of the CNS[4]. In these cases, it is the neurologist's job to further deduce the cause of such lesions, creating a diagnostic challenge. In certain cases, a biopsy may be necessary in order to eliminate differential diagnosis of neoplasms or infections.

Multiple Sclerosis (typical MS lesion seen in Figure 5) is the most common cause of tumefactive demyelination[2] [5]. For those with a tumefactive lesion as their first presentation, about 2/3 will go on to follow a relapsing remitting course of MS[5]. Frequency of TDL's in cases of MS is estimated to be around 2-3 per 1000 individuals[6].

Depending on the location and size of the lesions, symptoms can include: headaches, cognitive abnormalities, mental confusion, aphasia, apraxia, visual field defects, and seizures[2].

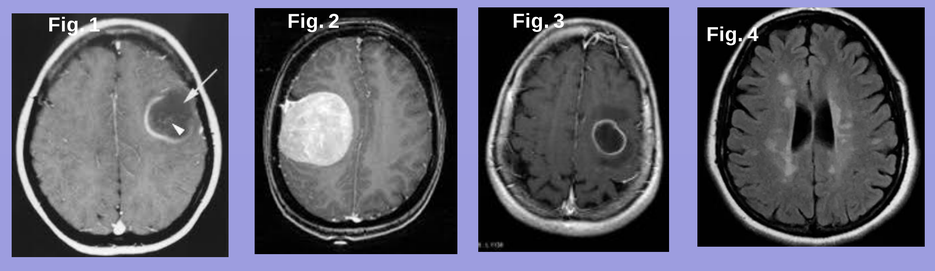

Figure 2. Open Ring Tumefactive Demyelination
Figure 3. Cerbral Glioma
Figure 4. Cerebral Abcess
Figure 5. Typical MS lesions

STATEMENT OF PURPOSE

This retrospective case analysis hopes to further contribute to the growing library of research dedicated to tumefactive demyelination, to continue to take steps towards reducing rates of misdiagnosis and use of unnecessary, invasive diagnostic tests, as well as improving treatment management for patients affected by this rare demyelinating disorder.

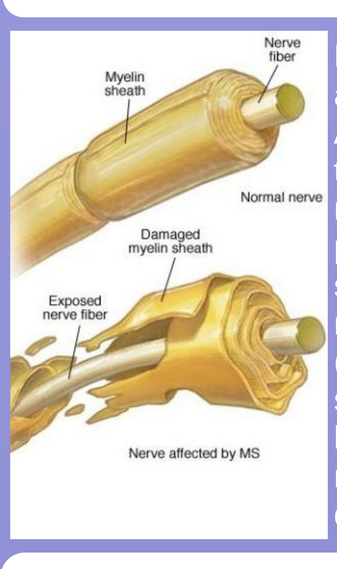

Figure 1. a healthy axon and a demyelinated axon. Axons are the connective tissues that run between neurons. Myelin is the protective coating that surrounds axons. When myelin is damaged (demyelination), electrical signals cannot be sent between neurons causing motor and cognitive disfunction.

METHODOLOGY

This study will be a retrospective chart review of patients with identified tumefactive demyelinating lesions. The source of records to be reviewed will be from the NYU Multiple Sclerosis Care Center. Study population will include people ages 18-60 seen at NYU Langone Medical Center with radiologically or biopsy proven tumefactive demyelinating lesions (See Figure 6 for biopsy results evident of TDL). An estimated 20 patient charts are expected to be reviewed. Radiographic and clinical data will be collected for all eligible patients. Current and previous medical status will have been established by neurologists at NYU Langone Health. Lesion enhancement patterns will be identified as megacystic, infiltrative, ring-like, balo-like, and non-specific based on the MAGNIMS recommended classifications for atypical demyelinating lesions.

PLANNED ANALYSIS

Patients will be separated into 4 distinct categories: those with TDL at onset, those with TDL at onset who developed MS, those with MS who developed a TDL and those with a clinically isolated TDL. From there, a statistical analysis of patient demographics (age of TDL onset and biological sex), presence of demyelinating disease prior to TDL onset, lesions (location, size, pattern of enhancement, edema and mass effect), clinical presentation (symptoms), biopsy and cerebrospinal fluid analysis, final diagnosis, choice of treatment, and long-term follow-up (when available) will be completed.

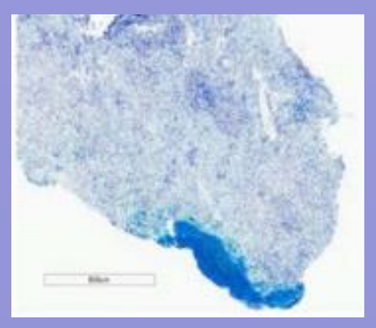

Figure 6. Luxol Blue Histochemical Stain (a histological test done following biopsy to prove or deny myelin loss) shows an almost complete loss in myelin[6]

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