A UNIQUE CASE OF TUMEFACTIVE MULTIPLE SCLEROSIS RESULTING IN LIMB APRAXIA, CORTICAL BLINDNESS, AND ASTEREOGNOSIS

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Introduction

Tumefactive multiple sclerosis (TMS) is an atypical variant of multiple sclerosis (MS) that occurs in 1 to 3 out of 1,000 cases of MS and presents with space-occupying demyelinating lesions that can mimic intracerebral tumors and abscesses. The clinical presentation of TMS is highly variable depending on the affected area of the brain and is often related to the mass-effect of the lesion. The most common symptoms include headache, cognitive deficits, aphasia, apraxia, and seizures. Diagnosis of TMS is made through magnetic resonance imaging (MRI), positron emission tomography (PET) scan, and cerebral spinal fluid (CSF) analysis. Brain biopsy is not necessary for diagnosis of TMS but is used in some cases to confirm diagnosis and avoid mismanagement. MRI features include solitary or multiple lesions size >2 cm usually affecting the white matter with or without mass effect and incomplete ring enhancement lesion surrounded by vasogenic edema. Histological findings include areas of demyelination with hypercellularity, inflammatory infiltrates by myelin-containing foamy macrophages, reactive astrocytes that may contain multiple nuclei (Creuztfelt-Peters cells), and perivascular and parenchymal lymphocytic infiltrates with preservation of axons. Traditional medical management with steroids, interferon-beta, and glatiramer may be used, however, Fingolimod and Natalizumab are avoided in tumefaction. Even with medication, patients diagnosed with MS require lifelong rehabilitation for effective management of stress, lifestyle, diet, physiotherapy, fatigue, and depression. Rehabilitation in a TMS patient necessitates a more tailored approach with additional consideration for the neurologic and cognitive effects from the space-occupying lesions.

Case Description:

A 24-year-old female presented with progressive blurry vision, cognitive deficits, astereognosis, limb apraxia, and aphasia. Initial MRI of the brain demonstrated large demyelinating lesions in the white matter of bilateral temporal lobes and left parietal lobe. Given her presentation and MRI findings, she received 5 days of IV steroids with no improvement, followed by plasma exchange (PLEX). Subsequent MRI of the brain showed multifocal enhancing periventricular masses bilaterally suspicious for lymphoma. Positron emission tomography (PET) scan of the brain revealed mildly avid supratentorial periventricular lesions possibly representing demyelinating process due to distribution and low integral avidity. Cerebrospinal fluid (CSF) analysis from lumbar puncture was performed, which detected atypical B cells and positive oligoclonal bands suggestive of MS. Brain biopsy was performed with results incompatible with central nervous system tumor. The patient received another 5-day course of IV solumedrol, two courses of rituximab, another course of PLEX therapy, and was later transferred to an inpatient rehabilitation facility.

On physical exam, patient was alert and oriented x3, had intact cranial nerve testing from CN III to XII, 5/5 muscle grade in all extremities, and left knee reflex +3 without clonus. Sensation to pinprick and light touch were present in all extremities. Romberg was negative. Patient exhibited complete visual field deficits with retained ability to perceive light and moving objects, poor two-point discrimination in all extremities, poor proprioception, limb apraxia, and profound difficulty identifying objects placed in her hands.

During her rehabilitation, she displayed mild improvements in coordination, proprioception, cognitive function, visual perception, astereognosis, and her ability to perform activities of daily living (ADL's). Patient, however, was prematurely discharged from the facility Against Medical Advice due to family issues.

Image adapted from Wikipedia

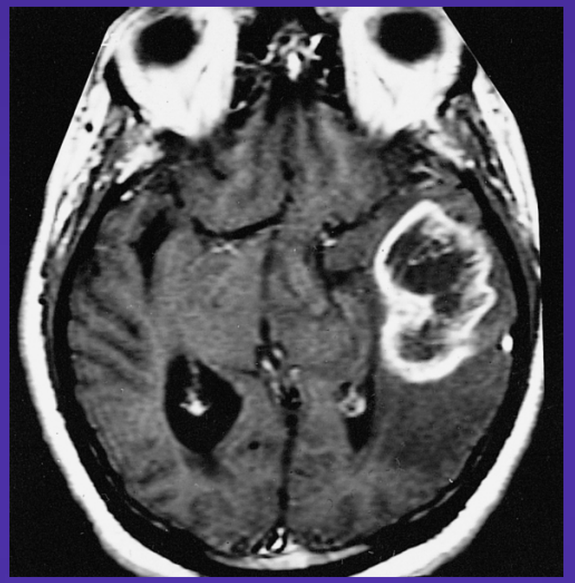

Fig A. T-2 Weighted MRI of brain showing the space-occupying lesion of Tumefactive MS

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Discussion:

Tumefactive MS represents a rare variant affecting a small percentage (0.1-0.3%) of MS patients. In addition to pharmacological therapy, rehabilitative measures such as exercise training, physical therapy, and gait training are essential for improving MS-related mobility disability through central and peripheral mechanisms. Tumefactive MS substantiates the need for more individualization of rehabilitation due to the varying presentation and long-term disability that may result. Astereognosis coupled with cortical blindness in this patient proved to make ADL training and basic tasks difficult to teach. Therapies were tailored to focus on object identification outside of traditional hand-to-object approaches, including verbal and tactile clues. Additionally, the use of text-to-voice technology demonstrates the integral function of technology in assisting patients with disability.

Optimal management recommended by the Consortium of Multiple Sclerosis Centers and International Multiple Sclerosis Cognition Society include: increased professional and patient awareness/education about prevalence, impact, and management of cognitive symptoms, a detailed assessment for adults who tests positive on initial cognitive screening or demonstrates significant cognitive decline, or neuropsychological evaluation for children with any unexplained change in school functioning. Assessment of cognitive function early in disease course is utilized not only to identify cognitive impairment but also to predict future impairments, limitations, and MS disease course. This patient received a neuropsychological evaluation to assess her verbal fluency, learning, and memory and insight into her communicative deficits.

Despite the patient's improvements in rehabilitation, she only participated in five out of the planned 14 to 21 days of inpatient therapy sessions, which limited the potential for observing even greater progress. Further outpatient rehabilitation and neuropsychological follow-up would be beneficial to aid her function and cognition. Adjunctive disease-modifying MS pharmacological therapy could also be administered to reduce the inflammatory response underlying the pathophysiology of MS.

Conclusion:

This 24-year-old female patient with tumefactive MS demonstrated unusual symptoms of cortical blindness, limb apraxia, and astereognosis, which presented a unique challenge requiring individualized and attentive care. Utilizing tactile and verbal cues may act as a surrogate to standard therapies and assist in ADL training despite these impairments. Based on her rapid improvement and functional efficiency improvement, this patient will benefit from continued therapy with a favorable prognosis.