Case Report

Multiple sclerosis with megacystic presentation: A case report

Andrés Felipe Herrera Ortiz, MD\textsuperscript{a,b,}\textsuperscript{*}, Santiago Aristizabal, MD\textsuperscript{a,b}, Juan Guillermo Arángula, MD\textsuperscript{b}, Valeria del Castillo, MD\textsuperscript{b}, Juan Calderon, MD\textsuperscript{b}, Nury Tatiana Rincón Cuenca, MD\textsuperscript{c}, María José Abuchar, MD\textsuperscript{b}, María Mónica Yepes, MD\textsuperscript{a,b}, Angela Guarnizo, MD\textsuperscript{a,b}

\textsuperscript{a}Radiology, Fundación Santa Fe de Bogotá, Bogotá, Colombia
\textsuperscript{b}Universidad El Bosque, Bogotá, Colombia
\textsuperscript{c}Fundación Universitaria de Ciencias de la Salud (FUCS), Bogotá, Colombia

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A B S T R A C T
Multiple sclerosis is a frequent condition where the diagnosis relies on clinical presentation, neurologic examination, cerebro spinal fluid markers, and diagnostic imaging tests; however, atypical variants of the disease can lead to misdiagnosis in some scenarios. Herein, we describe a case of a 24-year-old patient with multiple sclerosis with megacystic plaques, in which appropriate interpretation of the imaging findings lead to a proper diagnosis and treatment.

\textsuperscript{*}Corresponding author.
E-mail address: afherreraor@gmail.com (A.F. Ortiz).
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Introduction

Multiple Sclerosis (MS) is an inflammatory disorder of the central nervous system presented in roughly 2.8 million people worldwide; the disease predominantly affects women in 69% of cases, and the mean age of diagnosis is 32 years old \cite{1}. MS is characterized by focal lymphocytic infiltration that leads to damage to myelin and axons \cite{2}. In the early stages, the inflammation is transitory, and remyelination may occur; however, as the disease progresses, extensive and chronic neurodegeneration arises \cite{2}. Patients with MS usually manifest with acute neurological deficits, which can have a broad spectrum of symptoms depending on the region of the central nervous system involved. The most frequent areas affected are the juxtacortical region, optic nerves, brainstem, and spinal cord \cite{2}.

MS with megacystic plaques is an atypical presentation of the disease described only in a few case reports; therefore, it represents a diagnostic challenge leading to confusion with...
Case presentation

A 24-year-old male patient with a history of type 1 diabetes presented to the emergency department with a 7-day history of cervicalgia, oblique diplopia, and right conjugate gaze worsened with long distances. On physical exam, the patient showed right eye abduction limitation, left paresis, and paresthesia.

Head computed tomography was requested, only showing an expansile cystic lesion with a maximum diameter of 3 cm in the right frontal lobe without any associated mass effect or surrounding edema; the initial diagnostic suspicion was a neuro infection (Fig. 1). Brain magnetic resonance imaging (MRI) with spectroscopy was ordered to assess these findings better, showing multiple periventricular high signal foci perpendicular to the body of the lateral ventricles on T2 weighted sequences and a big cyst in the right frontal lobe. Spectroscopy demonstrated a slight reduction in n-acetyl aspartate (NAA) and an increase in lactate, lipids, and choline, without significantly altering the choline/NAA relationship (Fig. 2).

Due to the clinical picture and the MRI findings, the diagnosis of a demyelinating disease with megacystic manifestation was considered. A lumbar puncture was performed, showing type II oligoclonal bands, which confirmed MS diagnosis.

Once the diagnosis was performed, treatment was established with methylprednisolone 1 gr/daily for 5 days, with substantial improvement of the diplopia, paresis, and paresthesias; discharge was given with a neurology control appointment and natalizumab 300 mg/monthly (Previously negative JC virus test).

Discussion

MS with megacystic presentation is a rare and challenging diagnosis reported only in a few case reports [4]. Atypical inflammatory demyelinating disorders are characterized by aggressive demyelination presenting as large solitary lesions with more than 2 cm in diameter, heterogeneous appearance, and margins that vary from well to ill-defined [5,6]. These atypical manifestations may skew the diagnostic impression to other differentials, including infection (bacterial, fungal, or parasitic), inflammatory or demyelinating disorders (Neuromyelitis Optica, acute disseminated encephalomyelitis, sarcoidosis, Behcet disease), and neoplasia (primary or metastatic), leading in some scenarios to unnecessary brain biopsies [4,7].

According to Seewan et al., atypical inflammatory demyelinating disorders suggested by MRI can be classified into 4 morphological subtypes: infiltrative, Baló-like, ring-like, and megacystic (Table 1) [6,8]. Megacystic type presents with large cystic-like lesions with more than 3 cm in diameter, well-defined margins, and incomplete rim enhancement, as seen in our patient.

It is essential to highlight that in patients with MS, these atypical morphological appearances do not occur in isolation, and other “typical” lesions are associated in more than 50% of patients, as shown in this case report, in which the megacystic appearance was associated with periventricular high signal foci “Dawson fingers” in FLAIR sequences [8].

Even though the MS megacystic type is usually associated with “typical” MS lesions, differential diagnosis remains quite
Table 1 – Imaging characteristics of atypical inflammatory demyelinating disorders subtypes [adapted from Wallner et al. [8].]

| Morphological subtypes | Imaging appearance                                                                                                    | T1       | T2       |
|------------------------|-----------------------------------------------------------------------------------------------------------------------|----------|----------|
| Infiltrative           | Large irregular ill-defined areas of T2 abnormality with or without contrast medium uptake.                         | Hypointense | Hyperintense |
| Baló-like              | Multiple concentric rings                                                                                           | Hypointense | Hyperintense |
| Ring-like              | Round (≥ 2 cm in diameter) lesions with incomplete rim-like enhancement surrounded by an ill-defined zone of T2 hyperintensity suggestive of edema. | Hypointense | Hyperintense |
| Megacystic             | Large (≥3 cm in diameter) cyst-like lesion with incomplete rim enhancement.                                         | Hypointense | Hyperintense |

Fig. 2 – Brain MRI and MR spectroscopy. Axial T2W (A), axial FLAIR (B), and axial T1W post gadolinium (C) images show a high T2 signal oval lesion in the right inferior frontal gyrus involving the juxtacortical white matter without associated edema and open ring enhancement (green arrows). Notice the enhancing T1 and FLAIR high signal lesion in the periventricular white matter (white arrows). Axial and sagittal FLAIR images (D and F) demonstrate multiple high signal foci in the periventricular white matter (orange arrows). MR Spectroscopy (E) shows a slight reduction in NAA and an increase in lactate, lipids, and choline without significantly altering the relationship of Choline/NAA.

tricky; therefore, it is crucial to identify clues that aid in the distinction of this entity from other pathologies to narrow the differential diagnosis. The absence of ring-enhancing lesions with well-defined margins, lack of surrounding edema, and lack of mass effect are characteristics that go against an infectious process. [4]. On the other hand, factors that can help to differentiate brain tumors from megacystic MS are that brain neoplasms tend to compromise the cortical zone, and contrast enhancement usually has an irregular rim-like appearance. In contrast, MS megacystic lesions are characterized by a smooth incomplete rim-like enhancement, as shown in this case [6]. In all cases, it is crucial to remember that clinical data, physical exam, and cerebro spinal fluid analysis are essential in the diagnosis.

Advanced imaging modalities such as MRI spectroscopy multivoxel analysis in patients with MS usually shows a
slight reduction in NAA and an increase in lactate, lipids, and choline, reflecting an inflammatory process. This pattern is not pathognomonic of MS and can be seen in other diseases such as brain neoplasms; however, according to Llufriu et al., spectroscopy can be used to measure the degree of NAA depletion, which moderately correlates with subsequent development of physical disability [9]. In this case, our patient showed a slight decrease in NAA, which suggest a good prognosis and recovery.

Despite the large size of MS megacystic lesions, the long-term prognosis is favorable; however, factors such as age, ethnicity, disease burden, and disease activity may influence the outcome [8]. Our patient showed a significant recovery short time after receiving treatment with corticosteroids despite having a megacystic lesion, which is in line with current literature [8].

Conclusions

Demyelinating disease should be included in the differential diagnosis of cystic lesions in the brain without surrounding edema. Careful attention must be paid to clinical data, cerebrospinal fluid analysis, and imaging findings, in order to suggest the diagnosis and determine the proper treatment.

Patient consent

Informed consent was obtained from the patient.

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