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LETTERS TO THE EDITOR

Usefulness of magnetic resonance imaging in the diagnosis of Guillain Barré syndrome

Utilidad de la resonancia magnética en el diagnóstico del síndrome de Guillain Barré

Dr. Editor:

Guillain Barré syndrome (GBS) is an immune-mediated disorder of the peripheral nervous system characterized by acute polyradiculoneuritis. Clinically it is characterized by a sensory alteration, ascending paresis and areflexia. Sometimes, the symptoms are preceded by gastrointestinal tract infection. The diagnosis is clinical, and complementary explorations (lumbar puncture and neurophysiological study) support the diagnosis and rule out other etiologies. The primary role of magnetic resonance imaging (MRI) is not well established. We present two cases in which the MRI resulted being the key to the diagnosis.

Case 1

A 25-year-old man with no significant medical history, presented to the emergency room with weakness of both feet. The exploration showed distal paresis of both legs, grade 3/5, with exaggerated lower limbs osteotendinous reflexes and indifferent cutaneo-plantar reflex. The initial diagnostic suspicion was myelitis, so an MRI was performed, which showed an enhancement in conus medullaris and nerve roots (Fig. 1). A posterior lumbar puncture showed pleocytosis (8 cells/mm^3) and elevated cerebrospinal fluid (CSF) protein sprain (93.00 mg/dL), with negative serologies. The nerve conduction study reported prolonged F wave latency in right S1, with delayed ipsilateral sural conduction and mild S1 denervation in bilateral insertion study, all compatible with acute, sensitive polyneuropathy motor with mixed profile. The neurophysiological study realized one month later showed lower motor action potential values and signs of moderate denervation. A control study, six months after the onset of symptom, was normal. Anti-GM 1 and GM2 IgM anti-ganglioside antibodies were positive. On the basis of clinic manifestation, MRI, neurophysiological study and CSF, the final diagnosis was an acute idiopathic polyradiculoneuritis, acute motor axonal neuropathy (AMAN) variant. The patient improved with no specific treatment.

Case 2

A 51-year-old male, with no relevant medical history, presented to the emergency room with bilateral facial paresis and 24-h progressive dysphagia. The neurological examination showed bilateral facial paresis with other cranial nerves and osteotendinous reflexes preserved. The lumbar puncture showed pleocytosis (13 cells/mm^3, 100% mononuclear) with serologies, PCR for meningitis and CSF cultures, all negative. The study was completed with brain and cervical MRI with gadolinium, finding bilateral millimeter enhancement of both the VII and VIII cranial nerves in the internal auditory canals (Fig. 2). The anti-ganglioside antibodies (including GM1, GM2, GM3, GD1a, GD1b, GT1b and GQ1b) were negative. On the basis of the clinical manifestation, the lumbar puncture and the MRI the patient was diagnosed with acute idiopathic polyradiculoneuritis (facial diplegia variant). Finally, the patient was treated then with intravenous Immunoglobulin improving his symptoms.

Discussion

The diagnosis of GBS is established based on clinical manifestation and physical examination. The neurophysiological studies and cerebral fluid exam are used to confirm the diagnosis and rule out other etiologies. In the first case, we suspected a transversal myelitis. The MRI showed the enhancement in conus medullari and the root nerves, but not an inflammation of the medular spine. This result was essential to guide the posterior examinations, which confirmed the diagnosis of acute idiopathic polyradiculoneuritis, AMAN variant, due to neurophysiological study and the positivity of Anti-GM1 and Anti-GM-2 antibodies. In the diagnosis of GBS the role of MRI is not well established. There are few papers that describe changes in MRI in patients with GBS.[1,2] In a retrospective study of twenty-four children, Mulkey et al. discuss its importance in the differentiation of a myelitis or GBS, and how the enhancement of the lumbar nerves could be more sensitive than the neurophysiological study in early inflammation.[3] In the second patient, the MRI ruled out neoplastic or infectious causes, showing an enhancement of VII and VIII cranial nerves, also compatible with an inflammation of the nerves and the final diagnosis of facial diplejia,

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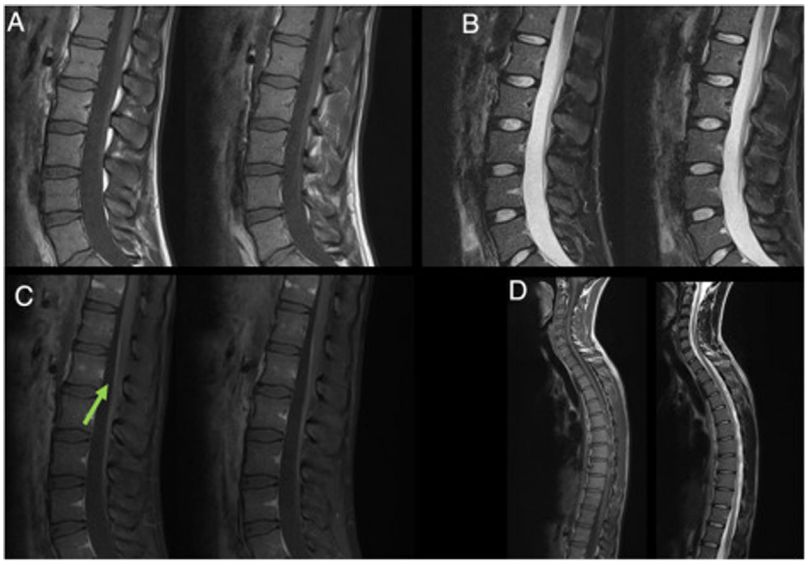

Figure 1 Lumbar MRI (A—C), and medullary MRI. (A) T1-weighted sequence without contrast. (B) STIR sequence. (C) T1-weighted sequence with gadolinium. (D) Medullary MRI. Left image T1 weighted sequence; right image T2-weighted sequence. The figure shows linear enhancement in conus medullaris and nerve roots (green arrow).

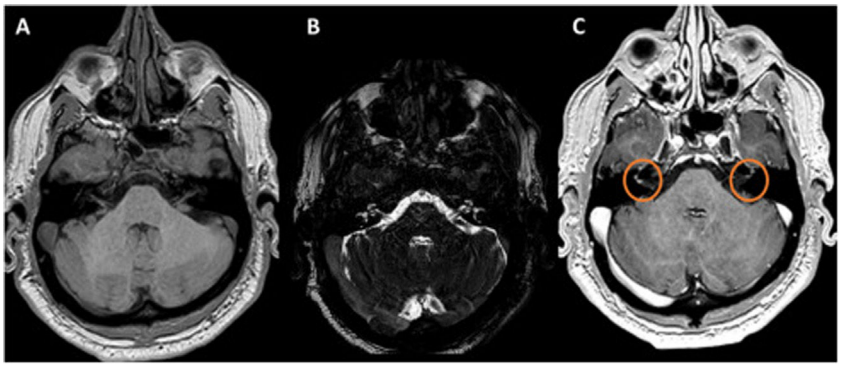

Figure 2 Brain MRI. (A) T1-weighted sequence. (B) Fat suppression T2-weighted sequence. (C) T1-weighted sequence with gadolinium. The figure shows millimeter enhancement of both the VII and VIII cranial nerves.

an unusual variant of GBS.[4] The MRI helped the diagnosis of GBS and this case report supports another previously described.[5—7]

We present images already described, but unusual in clinical practice. In our patients the MRI has been the key to the diagnosis of the pathology described. We think that it is an important exam to perform in doubtful cases or those in which the neurophysiological studies does not show abnormalities[8]; it should be taken into account in the diagnosis criteria.

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Diagnostic confusion of demyelinating lesions and incidental diagnosis of a new pathogenic mutation of the *FLNA* gene☆

Confusión diagnóstica de lesiones desmielinizantes y diagnóstico incidental de una nueva mutación patogénica del gen *FLNA*

Dear Editor:

The increased availability and technical quality of magnetic resonance imaging (MRI) has made it possible to achieve an earlier and more precise diagnosis of demyelinating lesions, such as those seen in multiple sclerosis (MS). However, it has also broadened the range of findings that may lead to a misdiagnosis of MS and, consequently, to unnecessary treatment. A multicentre study conducted in the United States gathered patients misdiagnosed with MS and analysed the possible causes.[1] With the exception of neuromyelitis optica spectrum disorders, almost all definitive diagnoses (conversion disorders, migraine, fibromyalgia, etc) lacked a specific biomarker, as is also the case with MS. Up to 24% of patients misdiagnosed with MS were diagnosed by a neurologist specialising in the disease, and up to 70% received disease-modifying treatments (occasionally with significant iatrogenic effects) and the incorrect diagnosis was maintained for years. One of the most significant factors in misdiagnosis was the misinterpretation of MRI findings, which may be explained by several reasons.[2] Therefore, the new 2017 McDonald criteria[3] highlight the importance of clinical symptoms, describing red flags and situations in which criteria should be applied with caution (patients who are asymptomatic or present atypical symptoms), and recommend expanding the analytical and radiological workup in these cases.[4]

We report the case of a 19-year-old woman with migraine and no other neurological symptoms, who was referred to our MS unit due to the detection of subcortical and periventricular white matter lesions potentially suggestive of MS in brain MRI studies performed to diagnose her migraines. The patient had history of Caroli disease (congenital ectasia of the intrahepatic bile ducts) and Laubry-Pezzi syndrome (congenital heart malformation characterised by high interventricular communication and secondary aortic valve insufficiency). Results of the neurological examination were strictly normal, with no evidence of previous episodes suggesting MS relapses. CSF analysis revealed normal cytobiochemical and immunological results. Given the diagnostic uncertainty, the radiological images were reassessed in detail (Fig. 1). The subcortical lesions were non-specific and were considered an incidental finding in a patient with history of migraine.[5] The periventricular lesions presented lower signal intensity on T2-weighted sequences (isointense with regard to the cerebral cortex) and presented a nodular morphology, protruding into the lateral ventricles, with no contrast uptake. They were finally classified as periventricular cortical heterotopia. The patient had no relevant family history. In the second examination, we observed joint hypermobility and hypertelorism.

The patient was referred to the clinical genetics department in view of her multiple congenital malformations. A first array comparative genomic hybridisation study was conducted after the patient received proper genetic counselling, showing no noteworthy alterations; a clinical exome sequencing study identified a probably pathogenic novel variant of the *FLNA* gene in heterozygosis. This gene encodes the filamin A protein (FLNA), which binds to actin and many other ligands with a range of cellular structural functions.[6] The variant consisted of a deletion of 4 nucleotides (c.6981_6984del) in exon 42, which presumably leads to a frame shift with the appearance of a premature stop codon (p.T2328Ffs*9). Pathogenic variants of the *FLNA* gene have been described in association with cardiac valvular dystrophy,[7] nodular periventricular heterotopia,[8] and otopalatodigital spectrum disorders with characteristics of Ehlers-Danlos syndrome (skin hyperlaxity and joint hypermobility).[9,10] As the patient presented these symptoms, this novel variant was classified as probably pathogenic. *FLNA* presents an X-linked inheritance pattern,

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