Letter to the editor

Parry-Romberg syndrome in an 11-year-old female with neurological manifestations without brain imaging abnormalities

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1. Introduction

Parry-Romberg syndrome or progressive hemifacial atrophy is a rare clinical entity, aetiology unknown, characterized by skin, fat tissue, muscle and sometimes adjacent bone atrophy. Lesions are usually localized to one side of the face, following a trigeminal dermatomal distribution [1]. Neurological and ophthalmological manifestations are the main complications of this disease. They can be as diverse as epileptic seizures [2], ophthalmologic abnormalities, hemiparesis, learning difficulties [3] or intracranial calcifications [4]. Parry-Romberg syndrome frequently begins before the age of 20 [5], with increased severity at younger patients. Serologic abnormalities suggest a pathogenic role for autoimmunity [1].

2. Case report

An 11-year-old girl, black race, Guiné-Bissau resident, consulted us due to left hemifacial progressive atrophy with 8 years of evolution. Until the age of three, the patient was healthy and had a normal development. She then presented progressive atrophy of the left hemiface and developed seizures, which were controlled with oral carbamazepine 500 mg/day that the child took ever since. Physical examination showed left paramedian atrophy of the face, folded skin without thickening or discoloration. The neurological and ophthalmologic exams were normal. With the exception of positive anti-nuclear antibodies (ANA) (titer: 1/80) and a weak positive rheumatoid factor (29.2 UI/mL), laboratorial studies were normal. Anti-topoisomerase and anti-centromere antibodies were negative.

A brain CT-scan and MRI (Fig. 1) were performed to study the central nervous system: no anomalies were found. Electroencephalogram showed no significant changes. A CT-scan of the face (Fig. 2) showed facial asymmetry with hypoplasia of the left side and marked enophthalmia. Since facial damage was established without noticeable progression in the last years or organs currently affected, no medication was initiated. The patient awaits plastic surgery evaluation for facial reconstruction.

3. Discussion

This patient has the typical Parry-Romberg syndrome depiction, with both neurological and ophthalmological complications. Epilepsy is the most common neurological problem associated with Parry-Romberg syndrome [2,6]. A case was reported [2] of a

Fig. 1. MRI of the brain showed no anomalies apart from marked facial asymmetry with hypoplasia of the left side.

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3-year-old girl with Parry-Romberg syndrome and seizures, without intracranial changes on CT-scan. However, the neurologic findings preceded skin changes, while in our case, they were concomitant. Unlike our patient, at 5 years of age, CT-scan showed multiple parenchymal calcifications and white matter lesions of the frontoparietal lobes. Brain MRI and CT-scan allow evaluation of cerebral abnormalities, especially MRI due to its higher sensitivity [3]. Our case shows that albeit using brain MRI, some patients do not evidence brain lesions, even after 8 years of disease duration with neurological symptoms. Both ANA and rheumatoid factor were positive in our patient, although with low values. In a serologic study of 14 Parry-Romberg patients [1], 8/14 had positive ANA, 5/14 had positive rheumatoid factor, and only 2/14 had positive anticientromere antibody. Our results also suggest that autoimmunity could play a pathogenic role in this condition.

There is no standard therapeutic option [7] or clear evidence of change in the outcome and deformations with any medication. Corticosteroids and/or methotrexate are prescribed if there is organ involvement or an early stage disease, although no benefit was shown in clinical trials [8].

Disclosure of interest

The authors declare that they have no conflicts of interest concerning this article.

References


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