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CASE REPORT

A case report of Melkersson-Rosenthal syndrome in adolescence

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Abstract

Melkersson-Rosenthal Syndrome is characterized by the triad: recurrent orofacial edema, facial paralysis and fissured tongue. It is a rare disorder and it can manifest itself in oligosymptomatic or monosymptomatic form. In this report, a 13-year-old patient is presented with peripheral facial paralysis and daily headache since the age of eight, mainly in the temporal and frontal regions, in bilateral masseters and holocranial, with pain at night without warning signs. After the first episode of facial paralysis, she continued outpatient follow-up with partial improvement in eyelid movements and lip deviation. The diagnosis of Melkersson-Rosenthal Syndrome had been established due to the presence of persistent peripheral facial paralysis, in addition to two episodes of plicated tongue and orofacial edema in crises. The importance of carrying out early diagnosis and appropriate treatment to improve quality of life and investigation of an underlying systemic disease, which may or may not be present, is highlighted.

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INTRODUCTION

Melkersson-Rosenthal syndrome is a rare neurological disorder whose classic triad consists of recurrent orofacial edema, peripheral facial paralysis, and episodic fissured tongue[1]; however, it can also manifest in oligosymptomatic or monosymptomatic forms in crises[1,2]. Its pathogenesis is uncertain, limiting treatment, but it is known to be associated with hereditary, allergic, and infectious factors, and may also result from local trauma or parasympathetic nervous system dysfunction[1,3,4].

Typically, the first symptoms appear between the ages of 25 and 40, and the condition is rare in childhood and adolescence, with an average onset between 7 and 12 years in this group[1]. When the complete triad is present, the diagnosis is clinical, and additional tests are necessary to rule out differential and secondary diagnoses[5]. There is no consensus on the ideal treatment, but it generally consists of the administration of immunosuppressants and symptomatic medications[1].

Despite the rarity of the disease, it is essential that the medical community understand this syndrome in order to avoid underdiagnosis and consequent irreversible damage.

CASE DESCRIPTION

A 13-year-old female patient presented to a tertiary pediatric hospital with low back pain for 9 days. One week later, she developed biparietal headache and peripheral facial paralysis (House-Brackmann grade IV). Previously, she had chronic daily tension-type headaches since the age of 8. The neurological examination on admission revealed only peripheral paralysis, and a cranial tomography scan showed no abnormalities. Corticosteroid therapy with prednisone 40 mg/day for 5 days was prescribed, with partial resolution of the clinical condition. Two months after the initial admission, the patient experienced a new episode of facial paralysis and returned for treatment. In this situation, a cranial MRI was requested, which revealed asymmetry and faint paramagnetic contrast enhancement near the anterosuperior aspect of the left internal auditory canal, in the projection of the intracanalicular segment of the left facial nerve (Figures 1 and 2), corroborating the attending physician's hypothesis of an inflammatory/infectious process (facial neuritis) depending on correlation with clinical and laboratory data. The treatment chosen was discharge with a prescription for hydrocortisone for 5 days.

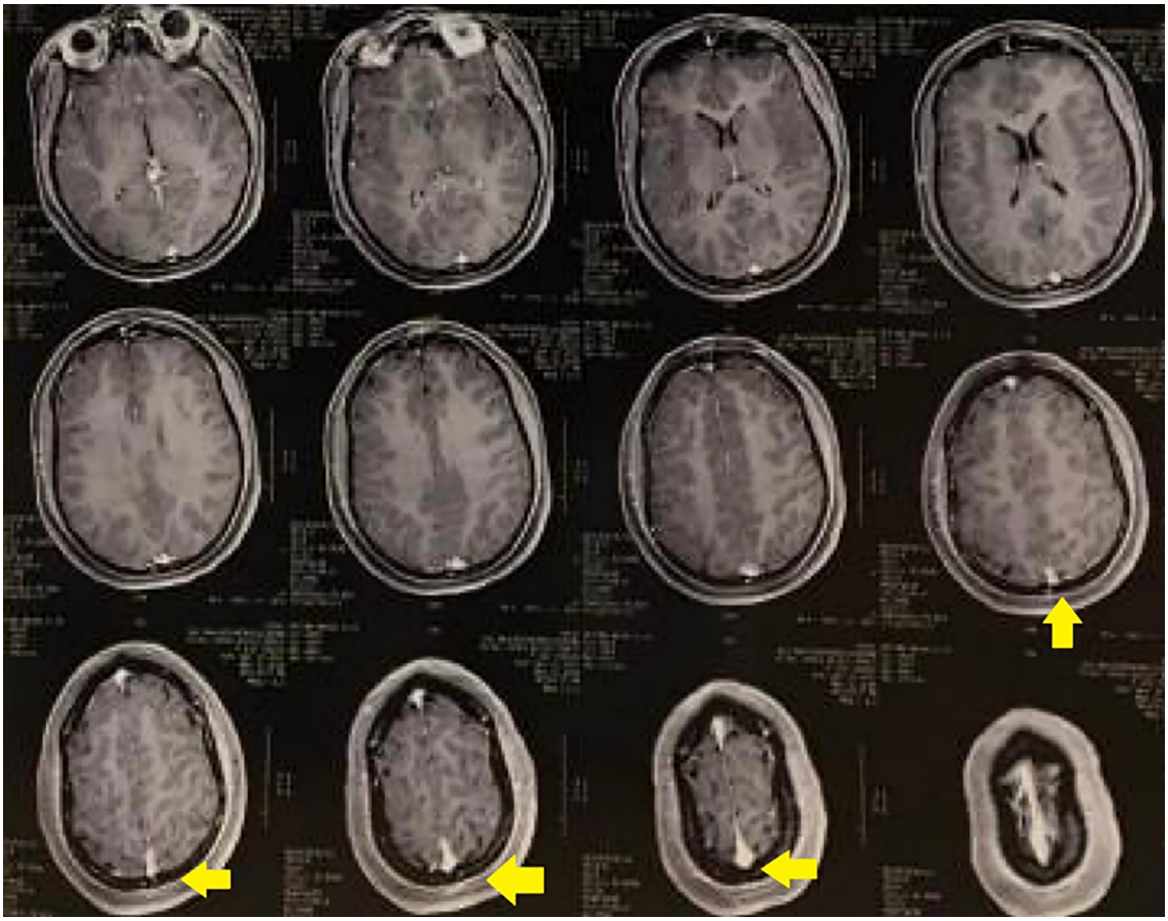


Figure 1. Asymmetry and faint uptake of paramagnetic contrast medium near the anterosuperior aspect of the left internal auditory canal, in the projection of the intracanalicular segment of the left facial nerve.

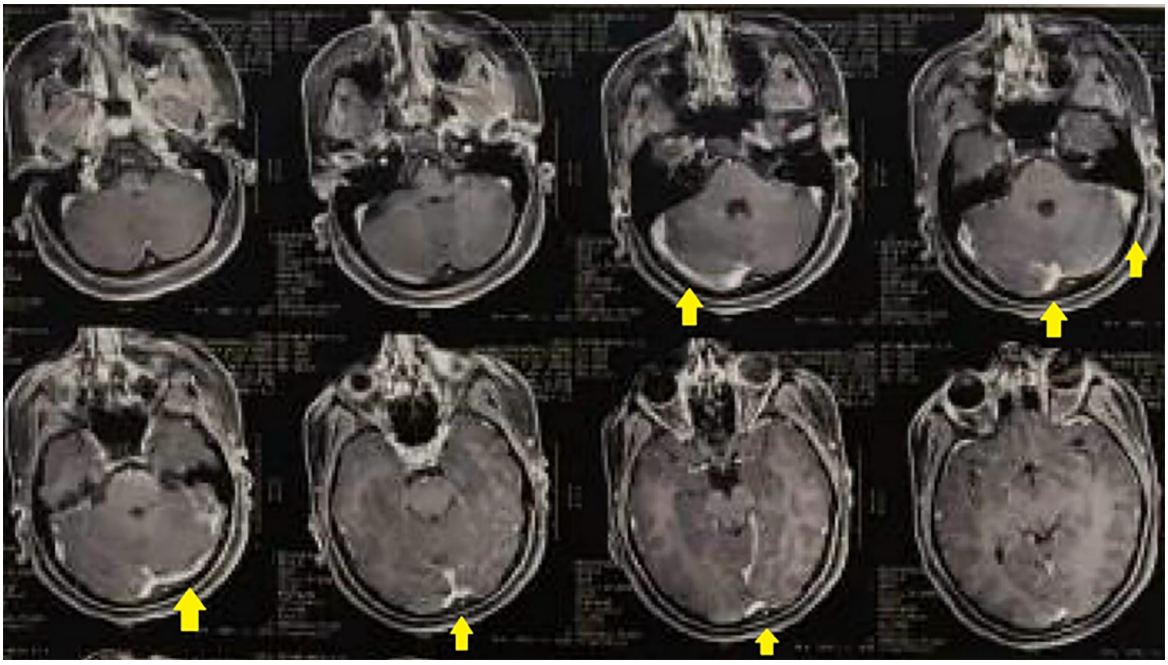


Figure 2. Asymmetry and faint uptake of paramagnetic contrast medium near the anterosuperior aspect of the left internal auditory canal, in the projection of the intracanalicular segment of the left facial nerve.

Three months later, the patient returned with episodes of fainting, nausea, orofacial edema, especially in the upper lips, and daily headaches since May of the same year. The patient also had a plicated tongue noted on physical examination, but the facial paralysis and eye pain had not improved. The treatment prescribed included 600 mg/day of carbamazepine, 50 mg/day of sertraline, lubricating eye drops, and an ophthalmic ointment containing retinyl acetate, methionine, and chloramphenicol. Due to the recurrent episodes of facial paralysis and orofacial edema, along with the finding of a plicated tongue, the hypothesis of RMS was raised.

Seven months after the last emergency room visit, the patient returned to the pediatric neurology clinic complaining of orofacial edema and bifrontal headache, associated with photophobia and nausea, with no improvement in the facial paralysis. Investigations for autoimmune diseases were negative. Between appointments, there was significant dissatisfaction with the lack of cure or complete remission of symptoms, despite the use of multiple pharmacotherapies. The pharmacotherapy was adjusted to gabapentin 900mg/day, cyclobenzaprine 5mg/day, and fluoxetine 40mg/day due to the low response of neuropathic pain to carbamazepine , in addition to medications that addressed the acquired comorbid psychological condition. The patient remained on the same medications until the age of 18, with progressive improvement in the facial paralysis , mainly in the upper face, but still with mild sequelae in the lower region — rhythm deviation and asymmetry—noted with a forced smile (House-Brockmann II) . The last therapeutic regimen was the most effective, with improvement in complaints and remission of the recurrent condition, and she was referred to the Neurology service at the age of 18 for follow-up in another service.

DISCUSSION

In only one-third of cases of the syndrome, the clinical picture will consist of the classic triad, as symptoms can occur in isolation, with an interval ranging from months to years. The main finding is orofacial edema ipsilateral to the paralysis, and when present, it has the following characteristics: non-erythematous, asymmetrical, non-pruritic, and painless; typically observed on the upper lip, but can affect the cheeks, lower lip, nose, eyelids, and temporal region[2]. Facial paralysis can be unilateral or bilateral, complete or incomplete, and, as the disease progresses, becomes more frequent, potentially affecting the chorda tympani nerve, culminating in hearing impairment. Fissured tongue, or plicata, is an anatomical variation with no pathological significance[4]. Patients with this condition may also present with migraine, involvement of other cranial nerves, hyperacusis, ophthalmological changes and dysfunction of the salivary and lacrimal glands, especially after repeated attacks[4,6].

Clinical diagnosis can be made when two or three elements of the triad are present. If only one is present, a mucocutaneous biopsy is necessary, usually of the lips or eyelid, with evidence of granulomatous cheilitis or perivascular lymphocytic infiltrate. The complexity of the clinical presentation, combined with the unknown etiology, makes diagnosis

difficult, resulting in many cases being underdiagnosed, and the average time between onset and diagnosis is 4 to 9 years.

Investigation of other causes consists of a neurological examination focusing on the cranial nerves, otoscopy, and visual and auditory acuity. Suggested laboratory tests include complete blood count, blood glucose, evidence of inflammatory activity, renal and liver function, cranial CT and MRI, chest X-ray, angiotensin-converting enzyme levels, antinuclear antibodies, antineutrophil cytoplasmic antibodies, thyroid function, ophthalmological evaluation, visual and auditory evoked potentials, and facial nerve conduction studies. All of this workup was performed on the patient in this case, who showed no signs of an underlying secondary disease, and long-term follow-up with no new symptoms. Detailed investigations, more dependent on the underlying secondary cause, according to each patient, include tuberculin skin test, karyotype, colonoscopy for inflammatory bowel disease, absence of C1 inhibitor for hereditary angioedema, and next-generation sequencing[1].

Due to the rarity of the disease and the complexity of establishing the diagnosis, RMS remains a diagnosis of exclusion. It is essential to rule out congenital conditions associated with facial nerve paralysis, such as Möbius syndrome, Goldenhar syndrome, congenital pseudobulbar palsy, and Arnold-Chiari syndrome. Obstetric trauma resulting in facial paralysis should also be considered, especially in cases involving forceps use, fetal macrosomia, prematurity, or cesarean delivery. Additionally, it is necessary to rule out infectious diseases that cause unilateral or bilateral facial nerve paralysis, such as those caused by the *Herpes simplex virus, Varicella-zoster virus, Epstein-Barr virus* , and *Cytomegalovirus* , as well as infections with *Mycobacterium tuberculosis* and *Treponema pallidum* . Lyme disease, particularly in endemic areas, should be investigated as a common cause of acute facial paralysis in children. Furthermore, granulomatous diseases, such as sarcoidosis and Crohn's disease, can also mimic RMS. Finally, it is crucial to rule out causes of orofacial edema: hereditary or allergic angioedema, granulomatosis with polyangiitis; causes of plicated tongue: vitamin B12 deficiency[4].

Although several theories exist to explain the etiology of RMS, its pathophysiology remains uncertain, and consequently, treatment remains questionable. Corticosteroid therapy for the management of orofacial edema and facial paralysis is proposed due to the immune dysregulation and allergic tendency associated with the syndrome , with an improvement of approximately 50-80% and a reduction in recurrence of 60-75%. Corticosteroids are used for up to 14 days, with pulse therapy only in severe cases[1]. Other symptomatic approaches have been proposed, such as intralesional triamcinolone acetonide or lidocaine, antibiotics, fluoxetine, other immunosuppressants, immunobiologicals, and, in some selected cases, surgical treatment. Further studies are needed to demonstrate the efficacy of each of these approaches[1,2].

In this case report, the patient was diagnosed with oligosymptomatic RMS, presenting with the complete triad only once, and was treated with chronic pain medications and corticosteroids. In addition to pharmacotherapy, multidisciplinary monitoring is required, especially with the mental health team, due to the potential for compromised imaging.

CONCLUSION

Melkersson-Rosenthal syndrome, despite its rarity, presents a complex clinical picture that can hinder diagnosis and appropriate management, especially in pediatric patients, the population with the lowest incidence of cases. This patient's case demonstrated the importance of a comprehensive approach, considering the variety of symptoms that can manifest, from facial paralysis to orofacial edema and fissured tongue. These symptoms may not be synchronous with long-term presentations, requiring longitudinal follow-up.

Treatment, although still under debate, benefits from the use of corticosteroids and other symptomatic interventions, proving effective in many cases, with the aim of minimizing irreversible complications. Although the syndrome typically presents episodically with complete remission of symptoms, residual symptoms and sequelae may occur in those affected, which can be minimized with early monitoring and treatment, including motor rehabilitation. Therefore, it is important to ensure that there is no delay in diagnosis and access to both pharmacological and non-pharmacological therapies.

Furthermore, multidisciplinary monitoring, especially regarding mental health, is essential to improve patients' quality of life and minimize the emotional impacts of the condition, especially in adolescence, a phase in which aesthetics begin to be valued as a means of fitting in. Delayed treatment can result in relapses and serious social problems arising from facial changes, sometimes permanent and resulting, as in the case presented. Continued education of the medical community about RMS is crucial to reduce underdiagnosis and promote early interventions, preventing permanent sequelae and improving the prognosis for those affected by this syndrome.

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