

**MELKERSSON-ROSENTHAL SYNDROME: A CASE REPORT****Wafa Baya\*, Jihed Anoun, Imen Ben Hassine, Anis Mzabi, Amel Rezgui and Chedia Laouani Kechrid**

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**ABSTRACT**

Melkersson-Rosenthal syndrome (MRS) is a rare orofacial granulomatosis characterized by a clinical triad, including lip edema, in its complete form. We report the case of a young woman who was suffering from paroxysmal lip edema for years. Examination objectified a left facial palsy and an edematous fissured lingual hypertrophy. Based on these signs and the normality of the biological tests, the diagnosis of MRS was retained. The patient was treated with oral corticosteroids with a minor improvement.

**KEYWORDS:** Lip edema - orofacial granulomatosis - Melkersson-Rosenthal syndrome.**INTRODUCTION**

Macrocheilia and lip swelling can be challenging. Vast pathological varieties must be considered including orofacial granulomatosis (OFG). One of these OFG is Melkersson-Rosenthal syndrome (MRS) which is a rare disorder characterized by a clinical triad.<sup>[1]</sup> This triad is the complete form of this syndrome although it's the less frequent presentation found in less than one third of the cases.<sup>[2]</sup>

Herein, we report a new case of a young woman presenting with the complete triad of MRS.

**CASE REPORT****Figure 1: Examination objectifying labial edema with left facial palsy.****Figure 2: Examination objectifying the edematous fissured lingual hypertrophy.**

A 26-year-old woman with a history of urinary lithiasis complained, since 2014, of persistent labial edema predominant in the upper lip with episodic aggravation. These episodes occur without a triggering factor and no allergic signs. No systemic symptoms were found. Examination noted an elastic edema of the upper lip (Fig. 1) with left facial palsy (Fig. 1 and 2) and edematous fissured lingual hypertrophy (Fig. 2). These signs preceded lip swelling as reported by the patient.

Complete blood count, erythrocyte sedimentation rate, C-reactive protein, electrolyte panel and thyroid function were unremarkable. Immunological screening was negative. C3, C4, C1q and total IgE dosage was normal. Chest X-ray was normal. Salivary gland biopsy showed discreet nonspecific lymphocytic infiltrates without granuloma. With the presence of the three clinical signs (lasting lips swelling, facial palsy and fissured tongue), Melkersson-Rosenthal syndrome was diagnosed. Oral corticosteroids were prescribed with a minor reduction of the swelling. Associated peripheral hypothyroidism with negative antibodies was objectified (TSH = 6 mUI/l). No

treatment was prescribed and clinical and biological surveillance was recommended.

## DISCUSSION

The first description of MRS dates back to 1928<sup>[3]</sup> and 1931.<sup>[4]</sup> Since then, this entity has been variously discussed in medical literature. No clear pathogenesis has been highlighted. Many theories have been suggested<sup>[5]</sup> including genetics, especially based on familial cases<sup>[6,7]</sup>, allergies, infections and inflammation MRS is considered an autosomal dominant disease with a variable expressivity.<sup>[8]</sup>

MRS is a rare neuro-muco-cutaneous non-caseating granulomatous disease with an estimated incidence of 0.08%.<sup>[9]</sup> It remains an exclusion diagnosis. Sarcoidosis and Crohn disease must be excluded.<sup>[10]</sup> Diagnosis is clinical. Histological findings are not needed in the diagnosis establishment.<sup>[10,11,12]</sup> The classic triad including recurrent or persistent oro-facial edema, episodic peripheral facial palsy and lingua plicata, is less frequent than oligosymptomatic or monosymptomatic presentations.<sup>[1]</sup> Its prevalence varies from 8% to 18%.<sup>[13]</sup> This leads to misdiagnosis and frequently delayed diagnosis.<sup>[14]</sup> Our patient presented the three signs progressively and additively. The fissured tongue was the first sign noted after episodes of facial palsy occurred then became persistent. Lip swelling was the last sign to appear. In fact, symptoms can present at different times.<sup>[15,16]</sup> This disease affects young adults in their second and third decades.<sup>[1]</sup> Pediatric cases have been reported.<sup>[17,18]</sup> Few cases have been reported in Tunisia. Mansour et al<sup>[7]</sup> recently presented a familial occurrence of MRS with three members affected with variable presentations. Five cases have been reported by Toumi and al treated differently.<sup>[18]</sup> In fact, no specific therapy has been established for this condition. Many treatments have been used in this disease with variable results such as corticosteroids, dapsone, antibiotics (penicillin, tetracycline, erythromycin, clindamycin, clofamazine), sulfasalazine, hydroxychloroquine, ranitidine, diphenhydramine, thalidomide, methotrexate and biologic agents.<sup>[2,19-21]</sup> Results are correlated to their anti-inflammatory and immuno-modulatory effect.<sup>[22]</sup> Surgical procedures of nerve decompression and reduction cheiloplasty are used for aesthetic results.<sup>[2,19,22]</sup>

Peripheral hypothyroidism was also diagnosed in our patient. Pathological associations to MRS have been reported in various case reports especially autoimmune and inflammatory diseases.<sup>[17,23-25]</sup> Thyroid disorder was essentially represented by Hashimoto's thyroiditis.<sup>[17]</sup> Our patient had negative antibodies.

## CONCLUSION

MRS is a rare granulomatous disease. Its pathogenesis is still unknown. Professionals might be misled by atypical symptoms and/or associations. Thus, delayed diagnosis

is frequent. In our case, it was established four years after the disease's onset.

Better knowledge of MRS clinical and histological characteristics as well as its different forms, especially the complete triad, is imperative to minimize physical and psychological repercussions and to prescribe early therapeutic intervention.

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