

# Melkersson-Rosenthal syndrome associated with Down syndrome

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**Abstract.** – We present a clinical case of Melkersson-Rosenthal (M-R) syndrome associated with Down syndrome. No evidence of this association is described in the literature. We also present a technique for the macrocheilia treatment of lower lip caused by M-R syndrome in a patient with Down syndrome. This patient during pediatric age had many events of facial nerve paralysis and edematous episodes of lower lip with unknown etiology. This technique is based on a wedge full thickness central excision of the lower lip and on a transversal lozenge excision in the vermilion portion with orbicular muscle to reverse the chin-labial corner. The results are an agreeable aspect of the lip and physiological digestive and phonetic processes. The technique is safe and simple and the aesthetic functional result is very good. In our case, the postoperative complication is caused by an insufficient collaboration of the patient and it is solved in three weeks.

#### Key Words:

Down syndrome, Melkersson-Rosenthal syndrome, Macrocheilia, Macroglossia, Hypertrophy of lingual papillae, Facial nerve paralysis.

## Introduction

An excessive prominence of the lower lip represents an aesthetically and a functionally impairment that is always present in patients affected by Melkersson-Rosenthal (M-R) syndrome<sup>1,2</sup>. Etiology of this syndrome is unknown and the pathogenesis is uncertain. Clinical features include edema of lips, recurrent and transitory paralysis of facial nerve, taste and visus abnormalities and feverish symptoms<sup>3,4</sup>.

In this paper we report a case of M-R syndrome in a Down patient with macroglossia, cheilitis granulomatosa and intermittent facial

nerve paralysis. M-R syndrome associated with Down syndrome is a very rare event. In fact, in the literature we have not seen any description of clinical cases. Surgical correction of labial defect represents the only solution for this problem when we also have a serious pathologic contest. We present a technique that is an efficient system to resolve the labial defect in three dimensions: width, height and projection. The results are an agreeable aspect of lip and a physiological function of digestive and phonetic processes.

## Case Report

We observed a 23 years old white male patient with Down syndrome affected by M-R syndrome (Figure 1). The general condition resulted normal but during pediatric age he had many edematous episodes of the lower lip and many events of facial nerve transient paralysis with unknown etiology. In familiar anamnesis there were no significant signs of pathology. However, in objective exam we observed a macrocheilia of the lower lip, scrotum, tongue and hypertrophy of lingual papillae (Figure 2). Ultrasound and colour duplex scanning of the lower lip have shown an echogenic homogeneity similar to the upper lip and an increase of vascularization without hemo- or lymphoangioma. The histological exam of the lower lip revealed a deposition of collagen fibres inside the corion associated with a lymphocyte infiltrate and rare foreign bodies granulomas. Orbicularis muscle was dissociated by a large amount of connective tissue with focal deposition of collagen fibres. The blood tests were negative for allergic or systemic disorders.

According to history anamnesis, clinical aspects and histological exam, we concluded for a diagnosis of M-R syndrome. The surgical treatment was important to correct aesthetic facial defect in lower lip.

Infact, macroglossia pushing the corrispondent lip can cause a pseudo-expansion of the same lip.



**Figure 1.** A 23 years old white male patient with Down syndrome affected by M-R syndrome.

The lower lip enlargement was corrected in his three dimensions lines by a modification of Conway and Mouly procedures<sup>5</sup>. Briefly, we removed a cuneiform central part of lower lip in full thickness. In the same time we removed a transversal lozenge of mucous tissue in the vestibular region including a portion of orbicular muscle along his principle axis (Figures 3 and 4). In this way we reversed the lip keeping his sphincteric function. Therefore, two mucous flaps of vermillion based on inferior labial arteries were mobilized for closure of the middle of the lower lip. With this method we reversed the chin-labial corner and we were able to reduce the height and thickness of the lip. In the same time



**Figure 2.** Macrocheilia of the lower lip, tongue and hypertrophy of lingual papillae.



**Figure 3.** Drawing of the transversal lozenge of mucous tissue and orbicular muscle with cuneiform central part.

we corrected the macroglossia, taking away its tip. Moreover, with a wedge full thickness central excision of lower lip, we reduced the widthness. Finally, remodelling was fundamental to complete the surgical treatment, suturing in vicryl 3/0 and prolene 5/0 for the sutures of lip. (Figures 5 and 6). During the postoperative course the only complication was a little aperture of vermillion shutting, but it healed in three weeks for second intention. This problem was caused by an insufficient collaboration and macroglossia of patient associated with Down syndrome.

With this technique we could have a good lip projection and a recovery of digestive and phonic processes (Figures 7 and 8).



**Figure 4.** Cuneiform full tickness central part excision of the lower lip.



**Figure 5.** Mobilization of two mucous flaps of vermilion for closure of the middle of the lower lip.

### Discussion

Melkersson<sup>6</sup> was the first who described this syndrome in 1928; also Rosenthal<sup>7</sup> in 1931 reported other cases.

The most common autosomal aneuploidy syndrome in term infants is Down syndrome (DS). The most serious consequence of DS is mild-to-moderate mental retardation (average IQ, about 50). Forty per cent of patients with DS have a congenital heart defect, most frequently ventricular septal defect or atrioventricular defect, although other congenital heart defect may occur. Thyroid disease, hearing loss, and celiac disease are common. Most person with DS have a trisomy 21 as a result of a new mutation nondisjunctional event.



**Figure 6.** Suture of lip.



**Figure 7.** Postoperative frontal view.

The macrocheilia of M-R syndrome associated with Down syndrome is a rare event. We have not found any case of this association in the literature. The diagnosis is very difficult and in the past was always deferred. In fact, the pre-surgical ecographic exam and the post-surgical hystologic exam are two important elements for the diagnosis. Causes of this syndrome are unknown. Some Authors say that it is an inherited and acquired disease of autonomous nervous system with an allergic granulomatous reaction against different circulating aspecific antigen. Instead, others associate this syndrome with some forms of Herpes viruses infection<sup>8</sup>, with Ehlers-Danlos syndrome<sup>9</sup> or also with a gastrointestinal disease like Crhon disease or simply like occasional constipation and diarrhoea<sup>10</sup>.



**Figure 8.** Postoperative lateral view.

The aesthetic problem of MR syndrome is important for the psychological impact of the patient in society and in his family. So the target of surgical correction is to re-establish form and function in the same time. The only vermilionectomy may not solve the problem but with this technique reconstructing the lip in three dimensions and restoring the function of orbicularis muscle are easy. Postoperative problems may be only caused by an insufficient collaboration of patient associated with Down syndrome. It has also been observed a high variability of answer for several pharmacologic therapies. In fact, the general use of corticosteroid in the average age is significative, but there are important results with intralesional injection of triamcinolone associated with oral somministration of clofazimine<sup>11</sup>, an anti-parassitary drug with an anti-leprotic and anti-inflammation activity. However, the first choice treatment is surgical and we think that our technique is satisfactory for aesthetic obtained result.

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