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A familial case of Melkersson-Rosenthal syndrome

Melkersson-Rosenthal syndrome (MRS) is an idiopathic non-caseating granulomatous process, usually characterised by triad of symptoms, i.e. chronic swelling of the lips, peripheral facial palsy that tends to relapse, and in some cases furrowed tongue (*lingua plicata*) (1, 2, 3). One of its most common presentation is lip or cheek swelling alone and thus is described as monosymptomatic form of MRS (2, 4).

The prevalence of the disease is estimated at about 0.08% of general population. Literature review reports equal male and female ratios or slight female predominance, with no racial or ethnic predilection. The onset of MRS usually occurs in young adults. The etiology of the disorder is unknown, however possible association with some autoimmunologic and infectious diseases has been proposed. Familial cases of MRS were also reported, but most authors suggested a multifactorial origin including a genetic basis (1, 3).

The triad of symptoms: 1) persistent and recurrent orofacial swelling, 2) relapsing facial nerve palsy and 3) fissured tongue are accepted as the major features of MRS. The disease usually presents in a monosymptomatic or an oligosymptomatic form – only 8–25% of MRS patients have got a complete form.

The most common symptom of MRS is recurring orofacial swelling in which the upper lip and the cheek are more often affected – the monosymptomatc presentation of MRS involving only the lip is referred to as *cheilitis granulomatosa* or Miescher's cheilitis. A forehead, eyelids, or one side of the scalp may also be involved (less common). Swelling is reported in 90–100% of MRS cases. It is often the earliest manifestation of MRS. The swelling is asymmetrical, nonpainful, nontender, nonpitting, firm but not of hard consistency, erythematous, typically subsides completely in hours or days, and usually precedes facial paralysis by weeks, months or even years; each recurrent episode is believed to be more pronounced, of longer duration and may eventually become permanent. Regional lymph nodes are enlarged (usually minimally) in 50% of patients. Facial paralysis – reported in 30–50% of MRS cases is indistinguishable from Bell's palsy. The site of facial palsy often corresponds to the site of swelling, but this can be unilateral or bilateral, partial or complete. Similarly to swelling, its generally self-limiting but each recurring episode lasts longer and is more profound. Other cranial nerves (e.g. olfactory, auditory, glossopharyngeal, hypoglossal) are occasionally affected. Fissured tongue, also called *lingua plicata* or scrotal tongue, exists in 30–70% of MRS cases. Patients may lose the sense of taste and have decreased salivary gland secretion. The symptom is not specific as it occurs also in general healthy population, but is ten times more likely to be found among diagnosed MRS patients than in general population (1, 2, 4).

Differential diagnosis should include odontogenic infection, Crohn's disease, sarcoidosis, Ascher's syndrome, submucosal neoplasm, angioneurotic edema and erysipelas. Full confirmation of

the diagnosis of MRS requires positive clinical history and histopathologic assessment of edematous tissue (non-caseating epitheliod cell granulomas, multinucleate Langerhans-type giant cells, perivascular mononuclear infiltration and fibrosis), or clinical history (recurrent swelling) plus at least one of the two: idiopathic facial paralysis or fissured tongue (4, 5).

As the etiology of the disease is uncertain, the therapeutic management is difficult and treatment still remains largely symptomatic. Various therapeutic options have not produced consistent results. Radiotherapy, antihistamines, salazosulfapyridine, all antibiotics have been shown to be unpredictable in managing the MRS symptoms. Only long-time steroid therapy showed some efficacy in most cases of MRS (2, 4).

CASE STUDY OF A PATIENT DIAGNOSED WITH MRS

Here we present the case of familial MRS. We treated a 28-year-old woman who suffered from unilateral facial palsy with ipsilateral cheek oedema. She had had the first episodes of facial palsy, and facial oedema at the age of 13, and now it was her eighth episode. Each of the episode lasted longer and was more pronounced. Swelling was nonpainful and preceded current facial paralysis by 3–4 days. The onset of facial palsy and oedema started approximately one month before admitting to hospital, and a few days after upper respiratory ailment. History of alcohol, tobacco, or drug abuse was negative. Physical examination revealed well developed, well nourished female in no acute distress with swelling of the right intraoral area and right cheek edema (Fig. 1) and fissured tongue (Fig. 2). Neurological examination revealed facial palsy (paralysis on the right side of the face, along with sagging eyebrow and difficulty in closing the eye) (Fig. 3); other cranial nerves as well as further neurological examination were normal. Standard laboratory work-up revealed no inflammation features. Determinations of *Borrelia burgdorferi* IgG and IgM serum antibodies using enzyme-linked immunosorbent assay in serum as well as in cerebrospinal fluid were negative. Another lab examination was also unremarkable. Brain imaging studies (CT, MRI) were also non-contributory, showing bilaterally symmetric, normal facial nerves images.

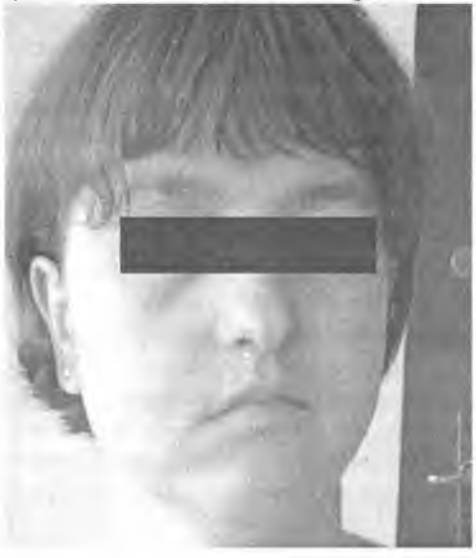

Fig. 1

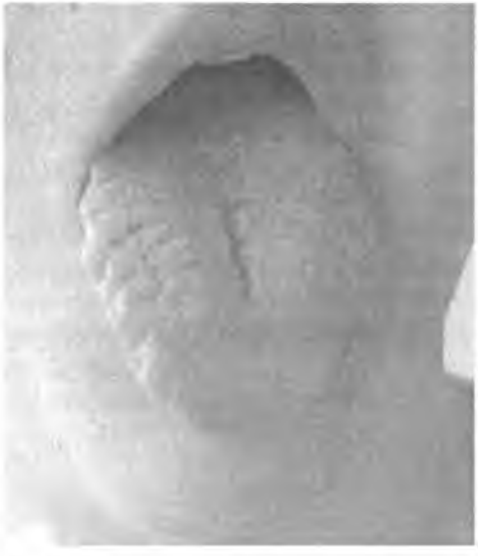

Fig. 2

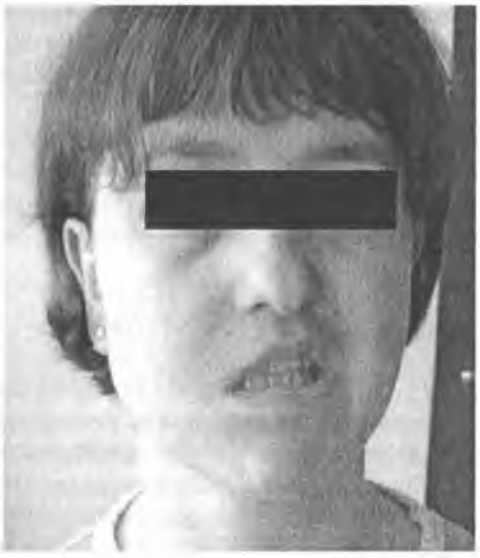

Fig. 3

Oral administration of prednisone 1 mg/kg/day for 2 weeks, intravenous lincomycin in the dose of 1200 mg/day, massages and electrical stimulation yielded significant, almost complete improvement of the facial palsy and cheek oedema.

Careful analysis of a family history revealed that both brothers of the patient had the oligosymptomatic form of MRS. They had had unilateral facial palsy, one with transient course, currently without any symptoms and one with persistent sign of facial nerve impairment, but they neither developed swelling nor furrowed tongue and they were never treated. In conclusion, we report the familial case of an incomplete form of MRS. It is also worthy to notify that though the origin of disease is still unknown, the steroids seem to be the most effective therapy.

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SUMMARY

Melkersson-Rosenthal syndrome (MRS) is usually characterised by triad of symptoms, i.e. chronic swelling of the lips, peripheral facial palsy that tends to relapse, and in some cases furrowed tongue (*lingua plicata*). The etiology of the disorder is unknown, however the possible association with some autoimmunologic and infectious diseases has been proposed. Familial cases of MRS were also reported, but most authors suggested a multifactorial origin including a genetic basis. We present here the case of familial, possibly inherited MRS. We treated the 28-year-old woman who suffered from unilateral facial palsy with ipsilateral cheek oedema. She had had the first episode of facial palsy, and facial oedema at the age of 13, and now it was her eighth episode. Physical examination revealed swelling of the right intraoral area, the fissured tongue, and right-sided peripheral facial palsy with some taste disturbances. Standard laboratory work-up revealed no inflammation features. Determinations of *Borrelia burgdorferi* IgG and IgM serum antibodies using enzyme-linked immunosorbent assay in serum as well as in cerebrospinal fluid were negative. Brain imaging studies (CT, MRI) were also non-contributory. Oral administration of prednisone 1 mg/kg/day for 2 weeks, antibiotics, massages and electrical stimulation yielded significant, almost complete improvement of the facial palsy and cheek oedema. Careful analysis of a family history revealed that both brothers of the patient had the oligosymptomatic form of MRS. They had had unilateral facial palsy, one with transient course, currently without any symptoms and one with persistent sign of facial nerve impairment, but they neither developed swelling nor furrowed tongue and they were never treated. In conclusion, we report the familial case of an incomplete form of MRS. It is also worthy to notify that though the origin of disease is still unknown, the steroids seem to be the most effective therapy.

Rodzinna postać zespołu Melkerssona-Rosenthala

Zespół Melkerssona-Rosenthala (MRS) charakteryzuje triada objawów: przewlekły obrzęk warg, obwodowe porażenie nerwu twarzowego oraz charakterystyczne, głębokie bruzdy na języku. Etiologia choroby jest nieznana. Rozważa się się czynniki autoimmunologiczne, infekcyjne oraz genetyczne, chociaż większość autorów sugeruje udział wieloczynnikowy. Prezentujemy przypadek kliniczny rodzinnego występowania MRS. W Klinice Neurologii leczono 28-letnią kobietę z powodu jednostronnego niedowładu nerwu twarzowego z tożstronnym obrzękiem policzka. Pierwszy epizod uszkodzenia nerwu twarzowego wystąpił w wieku 13 lat, a obecny był ósmym. Badaniem stwierdzało się obrzęk prawego policzka, głębokie bruzdy języka oraz obwodowe porażenie nerwu twarzowego z zaburzeniami smaku. Standardowe badania laboratoryjne (krew, badanie płynu mózgowo-rdzeniowego) nie wykazały cech infekcji. Nie stwierdzono obecności przeciwciał skierowanych przeciwko *Borrelia burgdorferi*. Badania neuroobrazujące nie ujawniły patologii wewnątrzczaszkowej. Chora była leczona prednizonem w dawce 1 mg/kg/day przez dwa tygodnie oraz antybiotykami. Stosowano również masaż oraz stymulację elektryczną. Zastosowane leczenie spowodowało prawie całkowite ustąpienie obrzęku policzka oraz objawów uszkodzenia nerwu twarzowego. Dokładny wywiad rodzinny wykazał obecność oligosymptomatycznego obrazu zespołu MRS u obu braci pacjentki. Obaj w przeszłości przebyli jednostronny obwodowy niedowład nerwu twarzowego, bez ewidentnego obrzęku warg lub policzka. Podsumowując, prezentujemy rodzinne występowanie zespołu MRS. Ponieważ etiologia choroby jest wciąż nieznana, steroidy wydają się terapią z wyboru.