Significant Facial Deformity With Multiple Dysfunctions In A Patient With A 15-Year-Undiagnosed Melkersson Rosenthal Syndrome

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Case Presentation

35-year-old man presented with 15-year facial deformity with recurring lower facial edema, orthopnea, muffled voice. Physical exam demonstrates severe sagging, loose cheek and prominent drooping, edematous lower lip, muffled voice and social anxiety features. Genetic testing was negative for neurofibromatosis. Nerve conduction study presented facial, and trigeminal nerve palsy. Histopathology showed lymphocyte infiltration. After two months of high dose IV corticosteroid, antibiotics, vitamin, and psychotherapy, his face and lower lip were significantly reduced. He was able to sleep on his back after 10 years of sleeping in a seated position. He then underwent facial surgery to reduce edematous facial tissue and planned for another cosmetic surgery to improve his appearance.

Figure

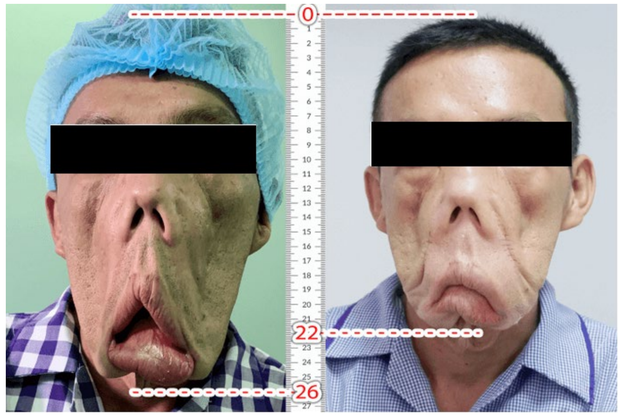


Figure 1

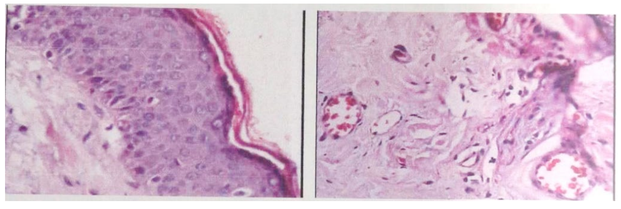
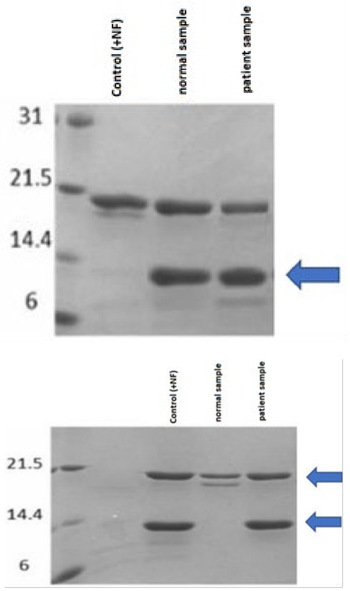


Figure 2



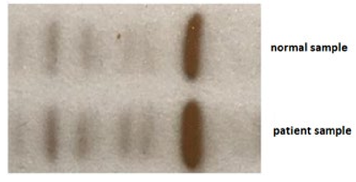


Figure 3

Figure 1 – before (left) and after 2-month-corticoid therapy (right);
Figure 2 – Lymphocyte infiltration in the dermal connective tissue;
Figure 3 – genetic testing

Literature Review

Melkersson-Rosenthal syndrome (MRS) is a rare, neurocutaneous syndrome with classic triad of recurring oro-facial edema, facial paralysis and fissured tongue. The pathogenesis is unknown. MRS is confirmed when the patient has at least two clinical features of classic triad or chronic granulomatous inflammation with at least one clinical entity. Common chronic inflammatory and neurocutaneous diseases should be excluded such as neurofibromatosis, inflammatory bowel disease, sarcoidosis, and hereditary Gelsolin amyloidosis. Clinical exam and genetic analysis could confirm the diagnosis. Corticosteroid, short term antibiotics, vitamin are the mainstay of treatment.

Unique Aspect

Identifying MRS is challenging in the patient with facial deformity due to long-term untreated MRS. Physical and mental aspect were evaluated to achieve the optimal outcome

Conclusions

Significant chronic facial deformity due to recurring facial edema and nerve palsy can be challenging for MRS diagnosis. Its complication may affect patient's physical and social functioning. Comprehensive management including appropriate medical and cosmetic treatment can improve the outcome of the patient.

References

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Banks T, Gada S. A comprehensive review of current treatments for granulomatous cheilitis. The British journal of dermatology 2012;166:934-7