IJHS

Case Report

Riga-Fede disease associated with Fabry's disease and Niemann-Pick C disease in a boy with microcephaly: A case report

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ABSTRACT

A 16-month-old Saudi boy has microcephaly and three rare genetic diseases [Riga Fede disease (RFD), Niemann-Pick C disease, and Fabry disease. In the published literature, there is no reported case with these four associations, especially RFD affection of the dorsal surface of the tongue. It is also a clear demonstration of how the proper diagnosis and treatment could provide a better quality of life, ease pain, resources, and money saving. The difficult RDF ulcer was resolved by a conservative treatment, which was accomplished by smoothing sharp edges and removing mamelons of the lower primary incisors using a diamond bur in a high-speed dental handpiece. The aim of this report is to present and discuss steps of diagnosis and the effect of misdiagnosis on the management of a very rare case.

Keywords: Angiokeratoma, case report, Fabry disease, microcephaly, Niemann-Pick C disease, Riga-Fede disease

Introduction

Riga-Fede disease (RFD) is a rare disease that affects infants and could be associated with neurological and genetic disorders. It is described as benign traumatic ventral tongue ulceration caused by repeated forward and backward movements of the tongue over the lower incisors.[1]

Niemann-Pick C disease (NP-C) is a rare inherited neurovisceral atypical lysosomal lipid storage disorder initiated by mutations in genes that caused damaged intracellular lipid running and build-up of glycosphingolipids and cholesterol in the brain and other tissues.[2,3]

Fabry disease (FD) is a rare an X-linked lysosomal disorder caused by the deficient activity of the enzyme a-galactosidase A. Angiokeratomas, pain crisis, and cornea verticillata are early manifestations of FD.[4]

To date, there is no previous published case with these rare genetic associations; RFD, NP-C, FD, and microcephaly. In addition, it is the first reported case of a RFD affected the dorsal surface of the tongue.

The aim of this report is to present and discuss steps of diagnosis and the effect of misdiagnosis on the management of a very rare case.

Case Report

A special needs 16-month-old Saudi boy. The chief complaint was a large ulcer on his tongue that caused pain, bleeding overnight, lack of sleep, and feeding difficulties. The mother reported that she noticed the ulcer 2 months ago and clinically examined by eight separate dentists. One of them thought that the lesion was a fungal infection and prescribing Daktarin oral gel 40 mg but in spite 1 month of treatment, the ulcer got bigger with more feeding and sleeping difficulties.

The mother also reported that the child was having a repetitive forward and backward movement of the tongue. The parents were very anxious as some dentists informed them that this lesion could be cancer.

Regarding familial history, the parents are first cousins and the mother has hypothyroidism. This boy was her only child after two abortions; one born with anencephaly (absent brain). The medical history revealed that he was born preterm (36 weeks) with microcephaly. After delivery, he was hypoactive, hypotonic and had respiratory distress, neonatal convulsion, and poor sucking reflexes. Before their visit to me, they had visited 12 local, national, and international medical centers for clinical examinations and laboratory investigations [Table 1].

On the first visit, he was not responding, and no eye contact. It was difficult to open his mouth as he had a strong bite and

Table 1: Medical investigations

Age	Consultant	Type of investigation	Result/recommendation
6 days	Radiologist	Brain CT scan	Evidence of diffuse hypoattenuating pattern of both cerebral white matter with a definition of gray-white matter interface/for brain MRI assessment
1 month	Radiologist	Brain MRI	Brain malformation (cerebellopontine hypoplasia)
6 months	Audiologist	Hearing assessment	Hearing loss/using binaural hearing aids
6 months	Neurologist	Clinical consultation	Convulsion controlled by phenobarbitone 5 mg/kg/day
7 months	Neurologist	EEG	Generalized epilepsy
9 months	Audiologist	Auditory brainstem response	Bilateral severe to profound hearing sensitivity in the frequency range 2–4 kHz/hearing aids fitting
12 months	Urologist Hematology	Kidney function tests Liver function tests	Normal Normal Normal
12 months	Neurologist	EEG	Normal, no evidence of epileptic activity
12 months	Radiologist	Neck CT scan	Enlarged adenoid
13 months	Audiologist	Hearing assessment Immittance measurements	Complete hearing loss in the left ear, profound hearing loss in the right ear Normal pressure with low compliance in the left ear and flat curve with normal ear canal volume in the right ear/using binaural hearing aids
15 months	Neurologist	Muscles, skin, and genetic tests	Neiman-Pic disease type C and Fabry's disease

CT: Computed tomography, EEG: Electroencephalogram, MRI: Magnetic resonance imaging

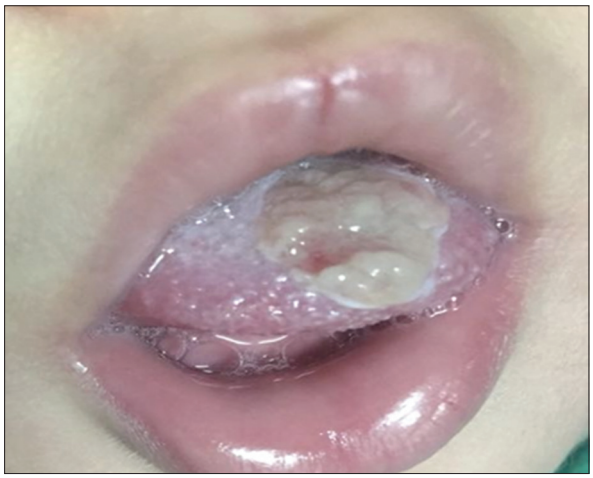

Figure 1: Riga-Fede disease ulcer before the treatment

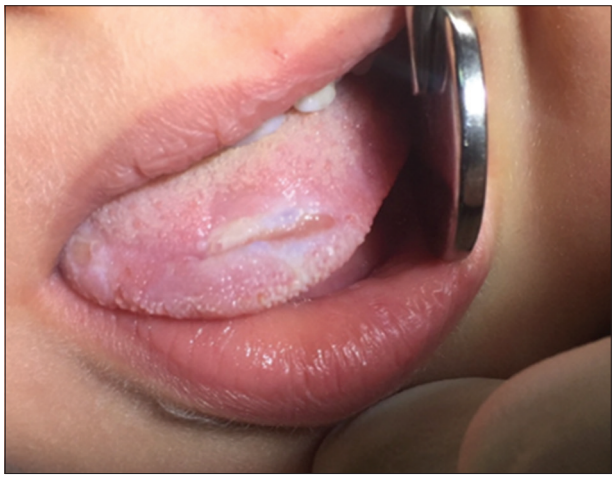

Figure 2: Riga-Fede disease ulcer after the treatment

uncontrolled tongue thrust. The intraoral examination revealed an ulcer approximately measuring about 3 cm × 2.5 cm on the dorsal surface of the tongue extended from the tip to the middle of the tongue [Figure 1]. Smoothing the sharp edges by removing mamelons of the lower primary incisors using a diamond bur in a high-speed dental handpiece was completed. A month later, the RFD ulcer had healed successfully [Figure 2]. After 1 year, a follow-up visit disclosed that the boy had stopped the habit of repetitive tongue thrust and no signs of ulcer reoccurrence. A written informed consent had been obtained from the child's parents.

Discussion

Although 60% of the lesions occur on the ventral surface of the tongue, it could be report on other areas such as palate, lip, vestibular mucosa, the floor of the mouth, and gingiva.[1] RFD had been reported previously with the association of microcephaly in only one case,[5] other cases described the occurrence of RFD due to neonatal/natal teeth[6,7] or as a result of tongue tie.[8] To date, there is no reported case of RFD on the dorsal surface of the tongue. In this case, the RFD affected the dorsal surface – instead of the ventral surface – of the tongue.

RFD treatment could be performed by covering the teeth with Stomahesive wafer strip, but it was not available in the country. Parents had spent loads of money and effort to help their baby have better health. However, they were content by the final outcomes. Furthermore, the boy needs to continue his medical follow-ups and management by his neurologist and audiologist.

Conclusion

The difficult RDF ulcer had been resolved by a conservative treatment (smoothing the sharp edges) which was successful and could be suggested as a possible treatment for similar future conditions. It is apparent that young children are unable to report their medical problems or pain as it's the case in adults. Moreover, most of the general dental practitioners' diagnosis is not definitive. Therefore, these cases need to be referred to a pediatric dentist to avoid the consequences of misdiagnosis and malmanagement.

Availability of Data and Material

The data used in this study are available and will be provided by the corresponding author on a reasonable request.

Patients Consent

Written patient's consent was taken to participate in this study.

Competing Interest

None.

Funding Statement

None.

References

1. Baroni A, Capristo C, Rossiello L, Faccenda F, Satriano RA. Lingual traumatic ulceration (Riga-Fede disease). Inter J Dermatol 2006;45:1096-7.
2. Vanier MT. Niemann-Pick disease Type C. Orphanet J Rare Dis 2010;5:16.
3. Patterson M, Hendriksz C, Walterfang M, Sedel F, Vanier M, Wijburg F. Recommendations for the diagnosis and management of Niemann-Pick disease Type C: An update. Mol Genet Metab 2012;106:330-44.
4. Tarabuso AL. Fabry disease. Skinmed 2011;9:173-6.
5. Baghdadi ZD. Riga-Fede disease: Association with microcephaly. Int J Paediatr Dent 2002;12:442-5.
6. Costacurta M, Maturo P, Docimo R. Riga-Fede disease and neonatal teeth. Oral Implantol 2012;5:26-30.
7. Buchanan S, Jenkins S. Riga-Fedes syndrome: Natal or neonatal teeth associated with tongue ulceration. Case report. Aust Dent J 1997;42:225-7.
8. Narang T, De D, Kanwar AJ. Riga-Fede disease: Trauma due to teeth or tongue tie? J Eur Acad Dermatol Venereol 2008;22:395-6.