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CASE REPORT

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Intraoral findings of a patient with Nablus mask-like facial syndrome and dental treatment approaches: a case report and literature review

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Abstract

Nablus mask-like facial syndrome (NMLFS) (OMIM: 608156) is an extremely rare genetic syndrome first reported by Ahmad Teebi in 2000. Although it is a rare condition, it is characterized by distinctive facial features such as, expressionless facial appearance, tight, glistening facial skin, low anterior hairline, sparse eyebrows, small palpebral fissures (blepharophimosis), hypertolerism, bulbous nose with prominent columella, abnormally short nose and flat nasal bridge, abnormal ear configuration, bilateral longitudinal cheek dimples, everted lower lip, long philtrum, and maxillary hypoplasia. In addition, a happy and friendly disposition is considered to be the common symptom of this syndrome. Previous studies revealing the intraoral findings of this rare symptom are inadequate and the present report is the first one that presents a dental case involving Nablus syndrome in detail. The aim of this report is to contribute to the current literature through our oral findings in an NMFLS patient, presented at our clinic with toothache and through our treatment approach.

Keywords

Nablus mask-like facial syndrome (NMLFS); Microdeletion; Enamel hypoplasia; Caries lesion; Cleft palate

1. Introduction

Nablus mask-like facial syndrome (NMLFS) (OMIM: 608156) is an extremely rare syndrome first described by Ahmad Teebi in 2000. While examining 4-year-old boy in Nablus, Palestine, Teebi noticed that he had an abnormal facial appearance but his twin sister had a normal phenotype. In his later study, Teebi stated that there was no consanguineous marriage in the family and there was no remarkable complication during pregnancy and delivery [1]. He also observed that the boy had a long, expressionless facial appearance, tight, glistening facial skin, low anterior hairline, sparse eyebrows, small palpebral fissures (blepharophimosis), hypertolerism, bulbous nose with prominent columella, abnormally short nose and flat nasal bridge, abnormal ear configuration, bilateral longitudinal cheek dimples, everted lower lip, long philtrum, and maxillary hypoplasia. Similar facial appearances were also revealed in the subsequently-reported cases [1–10]. In addition to these findings, tendency to mental retardation, microcephaly, growth retardation, dental anomalies, cleft palate, abnormal muscle tone, camptodactyly, joint disorders and autistic behaviors were detected in the subsequent cases (**Supplementary Table 1**, Table 1) [2–5, 8, 11].

In a study conducted in 2006, genetic etiology of the syndrome was identified by comparative genomic hybridization technique, revealing a 4 Mb microdeletion at 8q21.3–8q22.1 region in two patients with NMLFS [2]. Subsequent genetic analysis studies also confirmed the micro-deletion, observed in the 8q22.1 region [4, 6]. Although deletions of 8q22.1 were previously observed together with the clinical features in NMFLS patients, individuals without the characteristic features of the syndrome have also been reported despite they had a deletion in this region [5, 11]. Deletion in the 8q22.1 region is necessary but not sufficient to develop NMFLS's typical features [5].

The aim of the case report is to contribute to the literature on this highly rare syndrome through the oral findings of an NMFLS patient who presented at our clinic with toothache and through our treatment approach.

2. Case Report

A 6-year-old boy with the previous diagnosis of NMFLS had a severe toothache and was admitted to Ataturk University, Faculty of Dentistry, Department of Pediatric Dentistry clinic in 2016. His anamnesis revealed no family history or consanguineous marriage, and no remarkable complication before or during the birth. In his official medical document for proof of disability, the patient was verified that he had sensorineural hearing loss, mild intellectual disability and developmental delay. The results of our extraoral examination such as tight and shiny facial skin, upwardly curved hairline, sparse eye-

TABLE 1. Reported cases of dental abnormalities.

Case Reports	Dental Abnormality
Allanson, 2002	Partial anodontia.
Salpietro, Briuglia, Rigoli, Merlino, & Dallapiccola, 2003	High-arched palate. Thick and prominent tongue, with a short upper lip frenulum.
Shieh *et al.*, 2006-1	Retrognathia. Curve-shaped incisors.
Shieh *et al.*, 2006-2	The indented left upper incisor.
Mazziotti *et al.*, 2014	Missing upper lateral teeth. Openbite was also present from the premolars forward. Thick and prominent tongue, with a short upper lip frenulum.
Present Case Report	Submucous cleft palate. Thick upper alveolar ridge. Narrow vault. Anterior openbite. Anomalies in tooth positions. Congenitally missing upper lateral tooth in both primary and permanent teeth. Severe caries lesions with hypocalcifications in teeth. Macroglossia. Mucocele on mouth floor.

brows, small palpebral fissures (blepharophimosis), hypertolerism, bulbous nose with prominent columella, abnormal ear configuration, bilateral longitudinal cheek dimples, everted lower lip and maxillary hypoplasia were in parallel with the characteristic features of NMFLS (Fig. 1) as it was also reported in his official medical document for proof of disability. Furthermore, despite his mild mental retardation, we observed that the patient displayed an aggressive and agitated behavior.

Intraoral examination revealed severe caries lesions, hypocalcified areas in the primary teeth, missing upper primary lateral incisors, swelling and erythema associated with gingival inflammation, deep and dome-shaped palate and submucous cleft palate, thick upper alveolar ridge, narrow palatal vault, macroglossia and anterior openbite (Fig. 2). In addition, a 1 × 0.4 × 0.2 cm gray-white lump was observed in the lingual part of the right mandibular molars, on the floor of the mouth (Fig. 2).

Orthopantomography (OPG) taken during the radiographic examination demonstrated that the patient's dental development was normal but the upper permanent lateral incisors were also congenitally missing (Fig. 3).

Due to the patient's difficulty in cooperation, general anesthesia (GA) was considered to be necessary to perform his dental treatments such as the required fillings, extractions and endodontic procedures. After filling the patient's teeth #53, 51, 63 and 71 and performing pulpotomy for teeth #85 and 75, they were restored with stainless steel crowns and the remaining teeth #55, 54, 61, 64, 65, 72, 73, 74, 81, 82, 83 and 84 were extracted. In addition, the mass on the mouth floor was surgically removed and stored for pathological evaluation. The histopathological evaluation of the biopsy material revealed that the lesion was compatible with mucocele. After the extraction procedures, hemorrhage was managed and the extraction sockets were sutured (Fig. 4).

After performing the necessary post-anesthesia procedures, no complications were observed and the patient was discharged. In the follow-up two weeks later, the patient had no complaints (Fig. 5).

3. Discussion

Dental treatments under GA are highly important for the children who are uncooperative and in need of multiple dental procedures. The present case report highlighted the importance of GA in the dental treatment and follow-up of a 6-year-old boy with NMLFS, who could not be treated in the normal dental clinic conditions due to his lack of cooperation.

NMLFS is regarded to be one of the rarest conditions. Several patients have been diagnosed with this condition since it was first described by Teebi in 2000 and the intraoral findings of these cases are inadequate. Previous NMLFS cases in which intraoral findings have so far been reported are shown in Table 1 and the present case report is the first one presenting the intraoral findings of NMFLS more comprehensively than the others. However, it is also notable that the case report of an 11-year-old girl submitted by Mazziotti *et al.* [10] is compatible with the present report by revealing the missing upper lateral teeth, the presence of prominent diastema between the central incisors, macroglossia and anterior openbite. Present case report is considered to be different from the previous studies by being the first one to report severe caries lesions and hypocalcified areas in the primary teeth, missing primary and permanent upper lateral incisors, thick upper alveolar ridge, narrow palatal vault, and mucocele on the mouth floor.

Our patient was also medically reported to have experienced hearing loss, which was another significant finding in the present report. We reviewed the literature and found that hearing loss had been reported only by Jamuar *et al.* [9] in the case of a 14-year-old male patient. However, hearing loss, which was previously reported in only one NMLFS case as a symptom, it has been presented as a finding of the different

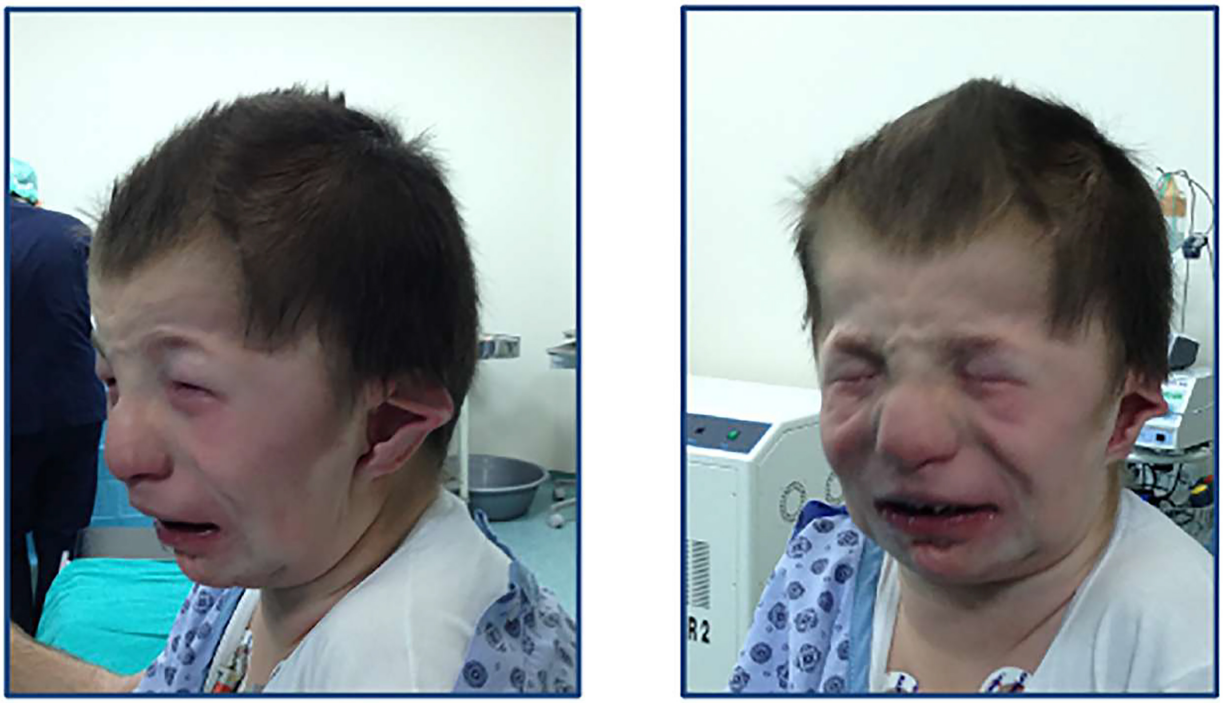

FIGURE 1. Full face photo of a 6-year-old male patient diagnosed with NMLFS, exhibiting the characteristic facial features of the syndrome.

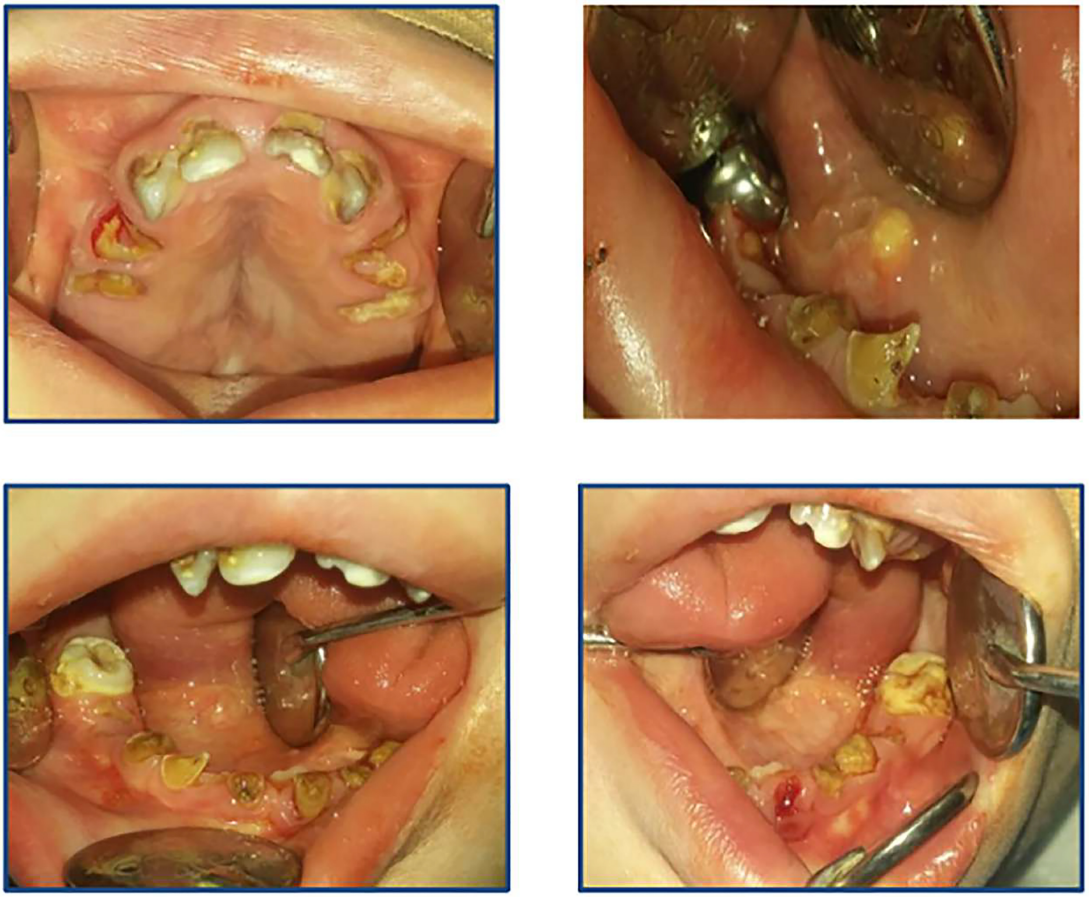

FIGURE 2. Severe caries lesions, hypocalcified areas on the primary teeth, missing upper primary lateral incisors, gingival inflammation-associated swelling and erythema, deep and dome-shaped palate, cleft submucous palate and a mass on the floor of the mouth.

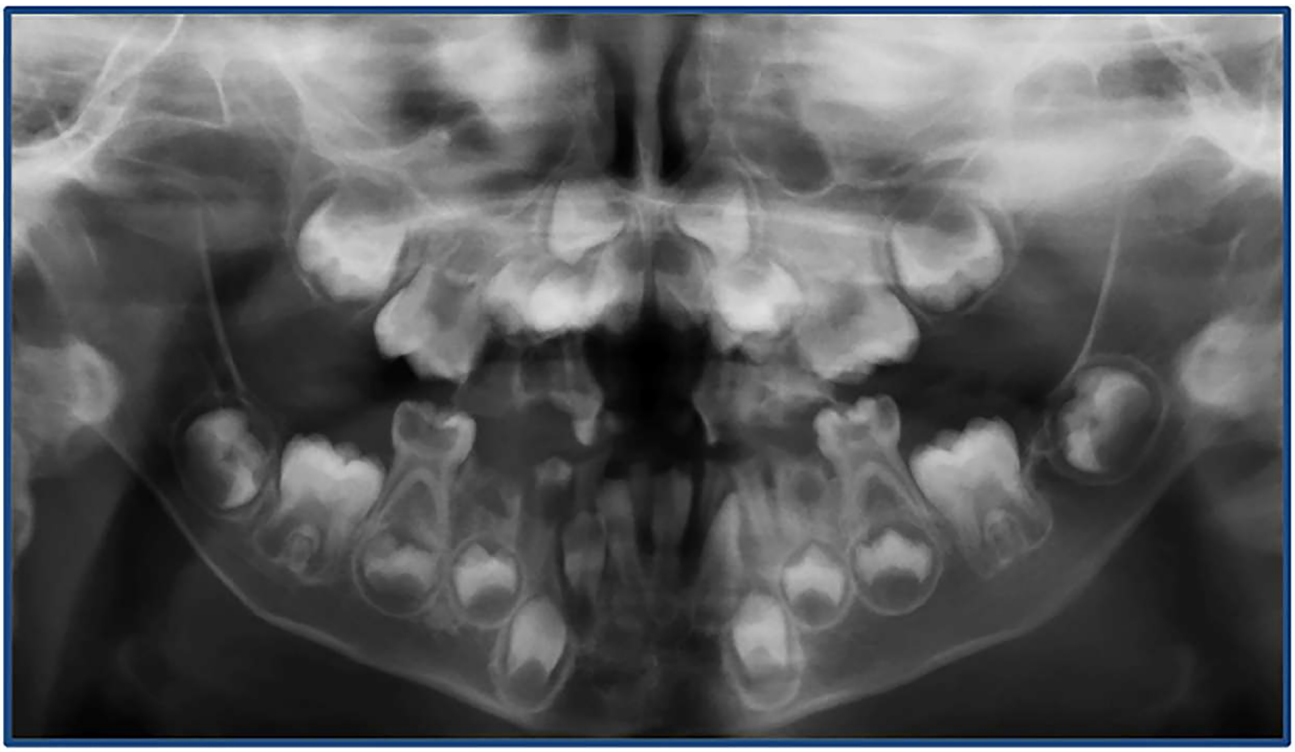

FIGURE 3. The patient's OPG showing the missing primary and permanent lateral teeth, severe caries lesions, and premature loss of primary teeth.

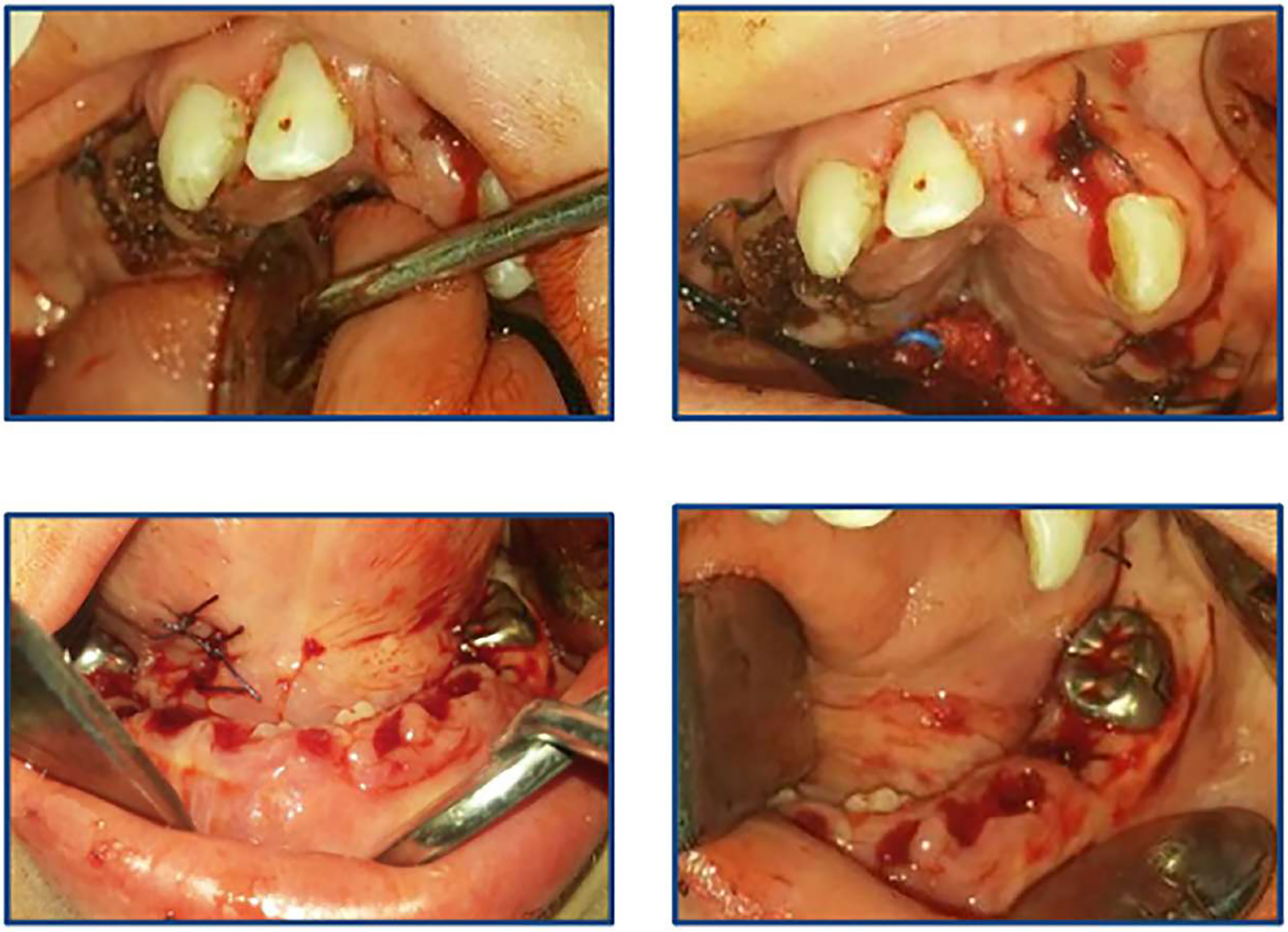

FIGURE 4. Post-treatment intraoral photograph of the patient.

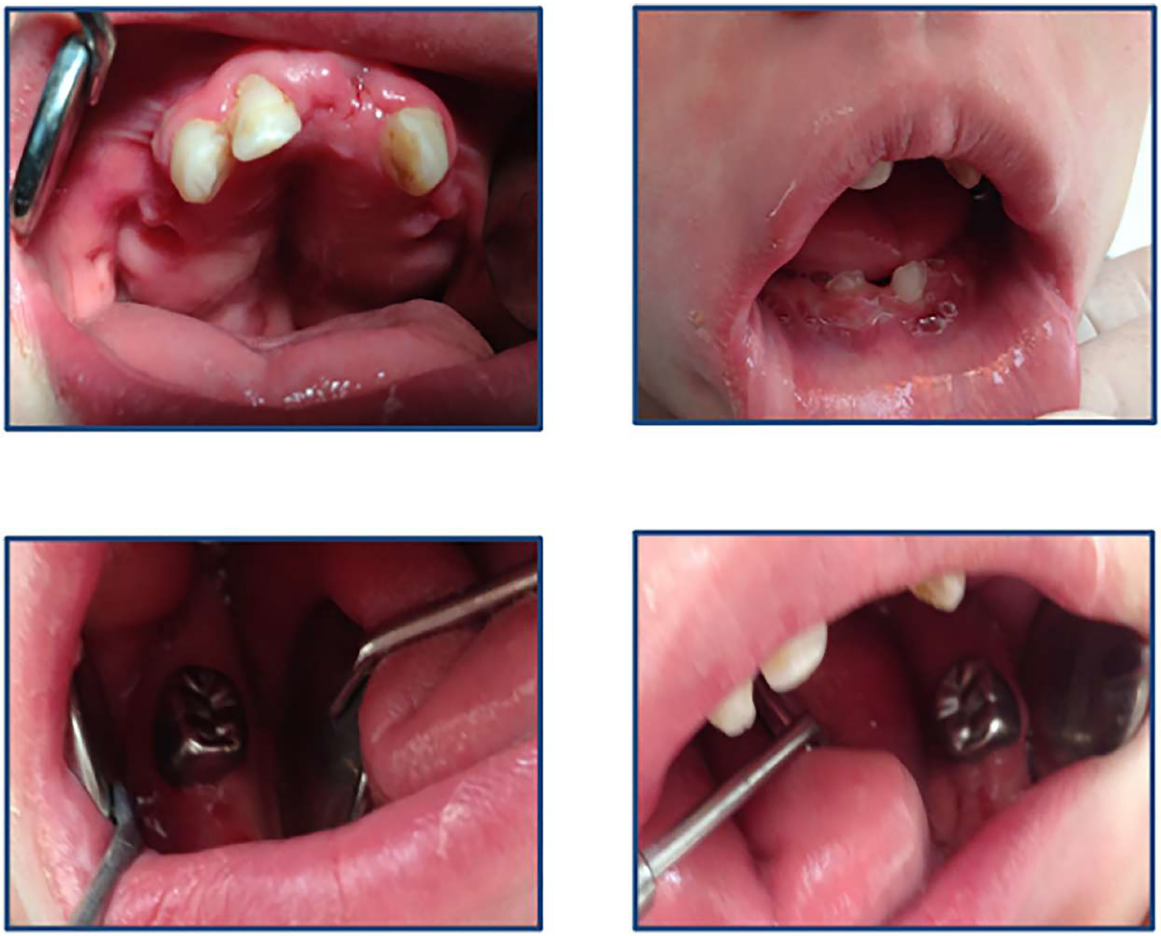

FIGURE 5. Healed extraction sockets and improved erythema and gum swellings two weeks after the dental treatment.

syndromes similar to the NMLFS in some case reports and studies [12, 13].

Although in the first NMFLS case report, Teebi initially remarked that the patient affected with the syndrome had no mental retardation [1] that was later regarded as one of the distinctive diagnoses of the syndrome, subsequent reports revealed that the patients were likely to have moderate or mild mental retardation or learning disability [2, 3, 14]. Furthermore, a happy and friendly disposition was also considered to be a common finding in this syndrome [4, 10]. However, it can be said that our patient exhibited more aggressive and agitated behaviors rather than a happy and friendly disposition and we thought that this attitude probably resulted from the psychological problems caused by his abnormal facial appearance.

The genetic etiology of NMLFS was first reported by Shieh *et al.* [2] using the genomic hybridization technique in their study in 2006, and a 4 Mb microdeletion was pointed out in the 8q22.1 region. In 2009, Raas-Rothschild *et al.* [4] confirmed that the genetic etiology of the syndrome resulted from microdeletion in the chromosome 8q22.1. In another case report presented a year later, a 6-year-old male patient was described to have a sad disposition and autistic behavior disorder rather than facial anomalies and a happy disposition that were the characteristic features of the syndrome despite the presence of a microdeletion in the 8q22.1 region [11]. Case reports, which have been submitted later also presented the individuals who developed no typical distinctive features of the syndrome despite the presence of microdeletion and thus, it was argued that microdeletion in the 8q22.1 region was necessary for the emergence of the syndrome, but it might not be sufficient to develop the typical distinctive features of the NMLFS [5, 15, 16]. It is currently a well-known fact that microdeletion in this region is a key factor for the occurrence of the syndrome, but its role has not yet been fully elucidated. Since there is insufficient data on this very rare syndrome in the literature each new case report will be significant for the clarification of phenotypic and genotypic features of NMLFS and will also be important for the dentists' treatment approaches.

4. Conclusions

Although only few previous reports have provided limited findings of dental abnormality, the present case report is considered to be the first to present comprehensive oral findings observed in NMLFS patients. Multidisciplinary evaluation of oral findings of the patients with NMLFS plays a key role in the patients' oral health, function, phonation, aesthetics and in treatment approaches as well.

AVAILABILITY OF DATA AND MATERIALS

The data presented in this study are available on reasonable request from the corresponding author.

AUTHOR CONTRIBUTIONS

SD and AB—designed the research study. AB—performed the research. SD, AB and FS—analyzed the data. AB and FS — wrote the manuscript.

ETHICS APPROVAL AND CONSENT TO PARTICIPATE

This report was approved by the ethics committee of Atatürk University Faculty of Dentistry with the decision #76 and session # 09.2022 dated 22.09.2022. Permission has been obtained from the patient's parents for presentation.

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CONFLICT OF INTEREST

The authors declare no conflict of interest.

SUPPLEMENTARY MATERIAL

Supplementary material associated with this article can be found, in the online version, at https://oss.jocpd.com/files/article/1653601477579423744/attachment/Supplementary%20material.docx.

REFERENCES

[1] Teebi AS. Nablus mask-like facial syndrome. American Journal of Medical Genetics. 2000; 95: 407–408.

[2] Shieh JTC, Aradhya S, Novelli A, Manning MA, Cherry AM, Brumblay J, *et al*. Nablus mask-like facial syndrome is caused by a microdeletion of 8q detected by array-based comparative genomic hybridization. American Journal of Medical Genetics Part A. 2006; 140A: 1267–1273.

[3] Salpietro CD, Briuglia S, Rigoli L, Merlino MV, Dallapiccola B. Confirmation of Nablus mask-like facial syndrome. American Journal of Medical Genetics. 2003; 121A: 283–285.

[4] Raas-Rothschild A, Dijkhuizen T, Sikkema-Raddatz B, Werner M, Dagan J, Abeliovich D, *et al*. The 8q22.1 microdeletion syndrome or Nablus mask-like facial syndrome: report on two patients and review of the literature. European Journal of Medical Genetics. 2009; 52: 140–144.

[5] Allanson J, Smith A, Hare H, Albrecht B, Bijlsma E, Dallapiccola B, *et al*. Nablus mask-like facial syndrome: deletion of chromosome 8q22.1 is necessary but not sufficient to cause the phenotype. American Journal of Medical Genetics Part A. 2012; 158A: 2091–2099.

[6] Barber JCK, Maloney VK, Huang S, Bunyan DJ, Cresswell L, Kinning E, *et al*. 8p23.1 duplication syndrome; a novel genomic condition with unexpected complexity revealed by array CGH. European Journal of Human Genetics. 2008; 16: 18–27.

[7] Allanson JE. A second family with blepharo-naso-facial syndrome. Clinical Dysmorphology. 2002; 11: 191–194.

[8] Overhoff J, Rabideau MM, Bird LM, Schweitzer DN, Haynes K, Schultz RA, *et al*. Refinement of the 8q22.1 microdeletion critical region associated with Nablus mask-like facial syndrome. American Journal of Medical Genetics Part A. 2014; 164: 259–263.

[9] Jamuar SS, Duzkale H, Duzkale N, Zhang C, High FA, Kaban L, *et al*. Deletion of chromosome 8q22.1, a critical region for Nablus mask-like facial syndrome: four additional cases support a role of genetic modifiers in the manifestation of the phenotype. American Journal of Medical Genetics Part A. 2015; 167: 1400–1405.

[10] Mazziotti S, D'Angelo T, Ascenti G, Blandino A. Facial abnormalities in Nablus mask-like facial syndrome: multidetector computed tomography findings. Journal of Oral and Maxillofacial Surgery. 2014; 72: 1579–1584.

[11] Jain S, Yang P, Farrell SA. A case of 8q22.1 microdeletion without the Nablus mask-like facial syndrome phenotype. European Journal of Medical Genetics. 2010; 53: 108–110.

[12] Sommer A, Bartholomew DW. Craniofacial-deafness-hand syndrome revisited. American Journal of Medical Genetics. 2003; 123A: 91–94.

[13] Sachdev M, Rastogi A, Singh A, Kumar K, Kapoor S, Bansal Y, *et al*. Phenotypic overlap between blepharo-naso-facial syndrome and Nablus mask-like syndrome. Report from the first Indian family. Ophthalmic Genetics. 2013; 34: 65–68.

[14] Turan B, Akinci MA, Esin IS, Dursun OB. Nablus mask-like facial syndrome with moderate developmental delay. The Eurasian Journal of Medicine. 2020; 52: 229–230.

[15] Allanson J, Smith A, Forzano F, Lin AE, Raas-Rothschild A, Howley HE, *et al*. Nablus syndrome: easy to diagnose yet difficult to solve. American Journal of Medical Genetics Part C: Seminars in Medical Genetics. 2018; 178: 447–457.

[16] Debost-Legrand A, Eymard-Pierre E, Pebrel-Richard C, Gouas L, Goumy C, Giollant M, *et al*. A new case of 8q22.1 microdeletion restricts the critical region for Nablus mask-like facial syndrome. American Journal of Medical Genetics Part A. 2013; 161: 162–165.

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