Sanjad-Sakati Syndrome: Oral Health Care

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Significance of the Study

- This case report demonstrates the importance of recognizing orofacial features in the diagnosis of Sanjad-Sakati syndrome, and discusses the dental management considerations in affected patients.

Keywords

Sanjad-Sakati syndrome · Dental health care · Hypoparathyroidism · Mental retardation · Learning disability · Seizure disorder

Abstract

Objectives: The aim of this report is to describe the orofacial manifestations and dental management of a girl with Sanjad-Sakati syndrome. **Clinical Presentation and Intervention:** The facial features included microcephaly, thin lips, beaked nose, low set ears, and a retrognathic mandible. An oral examination revealed oligodontia/hypodontia, small dental arches, a high arched palate, and a deep overbite and increased overjet. Oral rehabilitation involved full coverage prosthetic crowns on the upper central incisors, stainless steel crowns on the lower molars, and removable partial prostheses to replace missing teeth. **Conclusion:** Recognition of orofacial features might help in the diagnosis of Sanjad-Sakati syndrome. Dental management of affected patients might be complicated by intellectual, neurological, and endocrine abnormalities.

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Introduction

Sanjad-Sakati syndrome (SSS), also known as hypoparathyroidism-retardation-dysmorphism syndrome, is a rare autosomal recessive disorder first described in 1988 in a group of Saudi children with congenital hypoparathyroidism, dysmorphic features, and learning disability [1]. The syndrome has been predominantly reported in Arab patients from the Middle East and North Africa, but is increasingly encountered elsewhere because of globalization and migration.

The characteristic clinical features of SSS are linked to mutations in the tubulin folding cofactor E (*TBCE*) gene on chromosome 1q42–43 [2]. Facial features of SSS include low set ears, deep set eyes, thin lips, micrognathia, and a beaked nose [3]. Orodonal manifestations of SSS include a high arched palate, microdontia, hypodontia, taurodontism, enamel hypoplasia, and malocclusion [4–6]; recognition of orodental manifestations might help distinguish SSS from other syndromes associated with congenital hypoparathyroidism, such as Di George, Kenny-Caffey, and Barakat syndromes [7–9]. Dental treat-

Fig. 1. Small hands with short and thin digits.

Fig. 2. Oral findings included small dental arches, increased overbite, plaque-induced gingivitis, and multiple carious and missing teeth.

Fig. 3. Small and pointed tongue.

Fig. 4. Orthopantomography showing multiple missing teeth, delayed dental development, taurodontism, and incomplete root formation of the lower first molars.

ment of SSS patients might be complicated by endocrinopathy, seizure disorder, and learning impairment [5].

Here, we describe the orodental manifestations of a 15-year-old girl with SSS. We also review the relevant literature, and discuss the dental management considerations.

Case Report

A 15-year-old girl presented with the chief complaints of toothache and multiple missing teeth with poor aesthetics. She was the second child born to consanguineous parents who have 2 other healthy daughters. She was born at full term by normal delivery with a birth weight of 2.3 kg, but was admitted to the hospital shortly after birth with tetany and repeated seizures. Clinical and laboratory evaluations at that time revealed hypocalcemia, hyperphosphatemia, and a low parathyroid hormone level. Her full body radiographic survey was normal; genetic testing revealed 12 bp (155–166 del) within the *TBCE* gene in exon 3 confirming the diagnosis of SSS [2]. She has mild learning disability, and she left school at the age of 14 years because of teasing and bullying by other students. Her current medications included alfacalcidol and calcium supplements.

A general examination showed delayed physical growth; her height was 112 cm, and weight 17 kg. She had small hands and feet with short and thin digits (Fig. 1). An extraoral head and neck examination revealed microcephaly, thin lips, beaked nose, low set ears, and a retrognathic mandible. An intraoral examination revealed oligodontia/hypodontia, poor oral hygiene with plaque accumulation, chronically inflamed gingivae, small dental arches, a high arched palate, and a deep overbite and increased overjet. There were multiple missing and carious teeth. The upper left central incisor (#21) was heavily decayed and tender to percussion, the lower left lateral incisor was badly broken (#32), and both lower first molars (#36 and 46) were carious and lingually tilted. Both

Fig. 5. Oral rehabilitation using stainless steel crowns, prosthetic crowns, and a removable partial denture.

Fig. 6. Erythematous candidiasis (denture stomatitis).

upper first molars (#16 and 26) had been restored with stainless steel crowns at the age of 8 years because of severe hypoplasia (Fig. 2). No sinus tracts or soft tissue abnormalities were identified. The tongue was small and pointed (Fig. 3). Orthopantomography revealed delayed dental development, incomplete root formation of lower first molars, taurodontism of upper and lower first molars, absence of third molar dental follicles, and multiple missing teeth (Fig. 4). Saliva appeared to be of normal quantity and consistency.

A comprehensive treatment plan was formulated and discussed with the girl and her family. The disease control phase of our treatment plan involved extraction of the unrestorable lower left lateral incisor (#32), root canal therapy of a nonvital upper left central incisor (#21), scaling and polishing of the teeth, oral hygiene instructions, and dietary counseling. Carious teeth were restored using resin-bonded restorations.

The rehabilitation phase involved full coverage prosthetic crowns on the upper central incisors, stainless steel crowns on the lower molars, and removable partial prostheses to replace missing teeth in the upper and lower arches (Fig. 5). The patient showed good cooperation with the dental team and was tolerant to dental treatment; therefore, all procedures were performed under local anesthesia. The patient was scheduled for regular follow-up visits.

At the 1-year follow-up, there was no evidence of active dental diseases, and the dentures were satisfactory and well tolerated. However, erythematous candidiasis was evident underneath the upper denture because the patient refused to take it out at night (Fig. 6). Miconazole oral gel was prescribed, and the patient was given instructions on denture hygiene.

Discussion

SSS has been predominantly reported in the Middle East and North Africa, probably due to the deep-rooted norm of consanguineous marriage in the region. The exact prevalence of this rare syndrome is unknown, but estimates reported a prevalence of 1–2.5 per 100,000 births in Saudi Arabia, and 7–18 per 100,000 live births in Kuwait [7]. The patient in this report demonstrated the characteristic features of SSS, namely infantile hypoparathyroidism, learning disability, and dysmorphic features, including short stature, microcephaly, retrognathia, and small hands and feet [1–6]. Features of SSS are very similar to Kenny-Caffey syndrome, but the latter is characterized by osteosclerosis, cortical thickening, and medullary stenosis of tubular bones [5]. Our patient had normal full body radiographic surveys taken shortly after birth and at 3 years. Interestingly, both syndromes are caused by mutations in the *TBCE* gene, suggesting that SSS might be an allelic variant of Kenny-Caffey syndrome [8]. The *TBCE* gene encodes tubulin-specific chaperone protein E, which is required for the dimerization of α - and β -tubulin and polymerization of microtubules [7].

Oro dental manifestations of SSS have been sparsely described in the literature [3–6]. Reported features were similar to those found in our patient. Nontreated hypocalcaemia during the different stages of tooth development might contribute to the enamel hypoplasia, delayed eruption, and cessation of root formation observed in SSS [5]. The caries might be attributed to enamel hypoplasia, the use of sweetened medications over a long period, poor oral hygiene, and infrequent dental visits for preventive measures [5]. Parents of children with SSS should be educated about potential oral health consequences, and patients should be provided with preventive measures, including dietary counseling, oral hygiene instructions, topical fluoride application, and frequent recall visits. Oral rehabilitation in these patients might be challenging because of the small dental arch, associated dental anomalies, and learning impairment.

Our patient showed good cooperation with the dental team and all dental procedures were adequately performed under local anesthesia. General anesthesia and

airway management in SSS might be complicated by dwarfism, a retrognathic mandible, microstomia, seizures, and endocrine abnormalities [10]. It has been suggested that some patients with SSS have an increased predisposition to recurrent pneumonia and other bacterial infection; prophylactic antibiotics might therefore be necessary before extensive dental surgery under general anesthesia [5, 10].

Conclusion

SSS is a rare autosomal recessive disorder with characteristic facial features. Recognition of orofacial features might help in the diagnosis. Dental management of affected patients might be complicated by intellectual, neurological, and endocrine abnormalities.

Disclosure Statement

The authors declare no conflicts of interest.

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