Supernumerary Teeth and Dental Management in Sotos Syndrome

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ABSTRACT

Sotos syndrome is a genetic disorder characterized by early and excessive growth, acromegalic features, mental retardation, advanced bone age, and a distinctive facial appearance. The oral findings commonly seen with Sotos syndrome include: premature eruption of teeth; high, arched palate; pointed chin; and, more rarely, prognathism. The purpose of this case report was to review the diagnostic characteristics of Sotos syndrome, highlighting an interesting case involving six supernumerary teeth, and emphasize the importance of a multiprofessional dental intervention in combination with active family participation. Due to its rarity, many of the particularities of Sotos syndrome remain unknown. Close family and professional supervision are required to ensure that Sotos syndrome patients receive appropriate dental treatment.
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KEYWORDS: SOTOS SYNDROME, SUPERNUMERARY TEETH, RADIOGRAPHY FINDINGS, DENTAL MANAGEMENT

Sotos syndrome is a rare genetic disorder that was first described in 1964 by Juan Sotos et al. Since the discovery of this syndrome, it has been reported that haploinsufficiency of the gene for NSD 1 (nuclear receptor binding SET-Domain containing gene 1) is directly related to the pathophysiology of Sotos syndrome.[1]

Sotos syndrome is characterized by early, excessive growth associated with acromegalic features, microcephaly, nonprogressive cerebral disorders leading to mental retardation, prenatal overgrowth, advanced bone age, and a characteristic facial appearance.[1] The excessive growth is most pronounced in the first years of life and slows down, becoming relatively normal later in life. The patient, however, generally remains in the upper percentiles of the growth curve for their entire life. The prevalence of Sotos syndrome is not known, but it has been estimated that between 1 in 10,000 to 1 in 50,000 people may be affected by this disease.[2]

Most Sotos syndrome cases are sporadic, though several autosomal dominant familial cases have been reported in the literature.[3] There are generally no chromosomal abnormalities associated with Sotos syndrome, however, a few cases involving chromosomal lesions have been reported.[4] The diagnosis of Sotos syndrome is mainly based on clinical examinations and neuroimaging,[5] which, in the proper clinical context, can be used to differentiate this syndrome from other causes of mental retardation associated with macrocephaly. The neuroimaging findings support the hypothesis that Sotos syndrome results in delayed or disturbed development of the brain, specifically of the midline structures.[6] The differential diagnosis for patients with mental retardation and excessive growth include: autosomal dominant microcephaly, Weaver syndrome, Fragile X syndrome, Marfan syndrome, Bannayan-Riley-Ruvalcaba syndrome, and XYY syndrome.[2]

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Congenital hypotonia (high-arched palate, mild micrognathia, and anteverted nostrils) is often associated with Sotos syndrome and may result in asphyxiation and premature craniosynostosis.[5] Additional oral findings with Sotos syndrome include premature eruption of the teeth and prognathism.[1]

The purpose of this case report was to review the diagnostic characteristics of Sotos syndrome, highlighting an interesting case involving six supernumerary teeth, and emphasize the importance of an interdisciplinary dental intervention in combination with active family participation.

CASE DESCRIPTION

An 11-year-old Mestizo boy with Sotos syndrome was referred for dental treatment to the Peri-Peri Specialized Public Health Center, São Paulo, Brazil. He presented with a history of acute pain in the permanent mandibular left first molar (19).

DEVELOPMENTAL HISTORY

The patient was born vaginally at 9 months gestation with a weight of 3.56 g, a cephalic perimeter of 37 cm, a thoracic perimeter of 37 cm, and a height of 51 cm. His parents were nonconsanguineous and had 4 other healthy children before him. The pregnancy was complicated by gestational hypertension, which resulted in many hospital stays. The patient's motor function and developmental milestones were normal; he rolled over at 6 months and walked within 12 months.

At 2-years-old, the patient was noted to have a language delay and apparent cognitive dysfunction. Sphincter control only became possible when he was 4-years-old. He went to a normal school at that age. At the 14-years-old, he was still not able to read or write and exhibited impaired motor coordination. He was capable of participating in daily activities, however, such as going to school, shopping, and playing with his friends and sisters.

Upon physical examination, the patient was noted to have a prominent forehead and sparse hair, particularly in the frontal region. His skull was macrocephalic and dolicocephalic. His face gradually lengthened with time, resulting in a wide, prominent forehead and long, thin chin and jaw (Figure 1). He exhibited ocular hypertelorism, an interpupillary distance of 4 cm, divergent strabismus, intermittent horizontal nystagmus, myopia (9.5° in the right eye and 4.75° in the left eye; Figure 2), low set ears and auricular dysplasia (Figure 3), pulmonary stenosis, and delays in language acquisition. His nose was saddle-shaped and his palate was highly arched (Figure 4). According to his mother, his primary teeth began to erupt at 4 months and the eruption was complete when he was 1-year-old. Despite the growth of primary teeth, he had an apparent delay in the eruption of permanent teeth (Figure 5). At the time of this study, he was 14-years-old and his actual weight was 38 kg, his cephalic perimeter was 57 cm, and his height was 160 cm. At the initial visit at age 11 the patient complained of pain on tooth no. 19, which had to be endodontically treated.

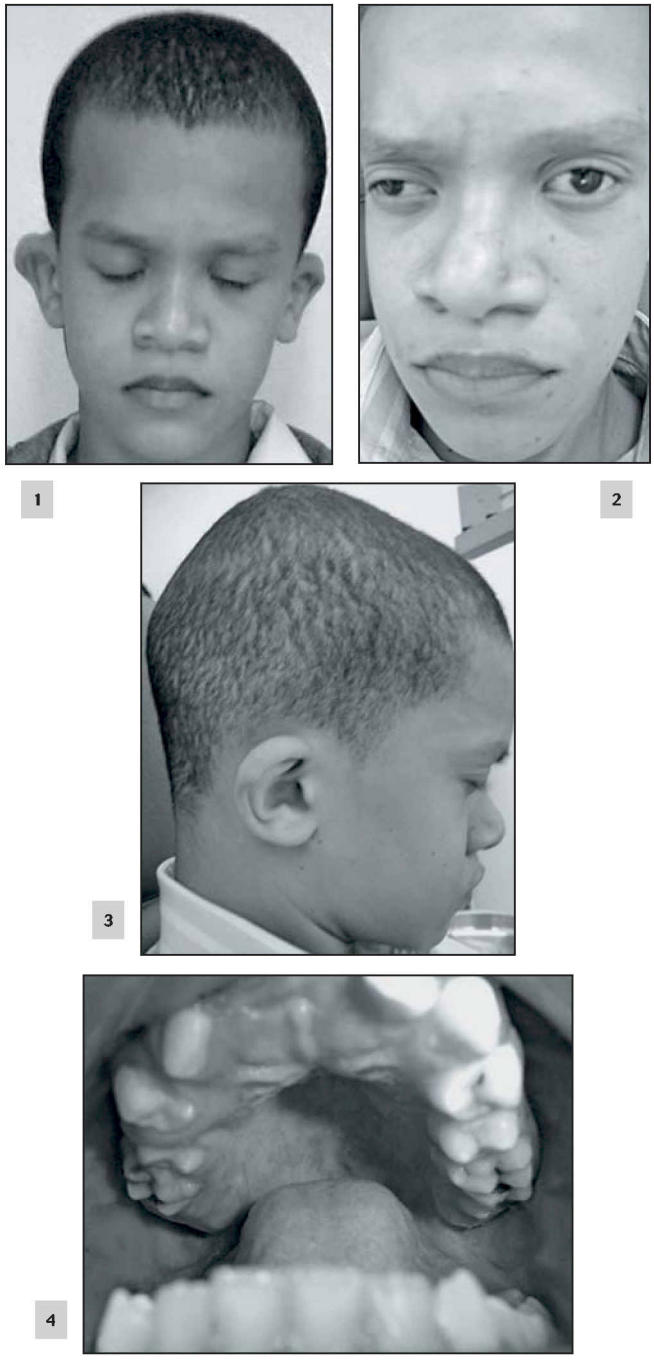


Figure 1. The patient at 11 years old, exhibiting auricular dysplasia, ocular hypertelorism, prominent forehead, sparseness of hair in the frontal region, macrocephaly, and dolicocephaly, resulting in a peculiar facial shape.

Figure 2. The patient at 14 years old, exhibiting ocular hyperte-lorism and divergent strabismus.

Figure 3. Lateral view of the patient. The peculiar head shape and low set ears are visible.

Figure 4. Highly arched palate.

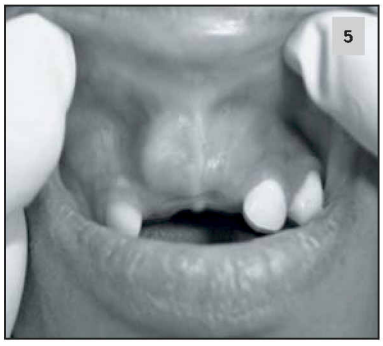
DENTAL TREATMENT

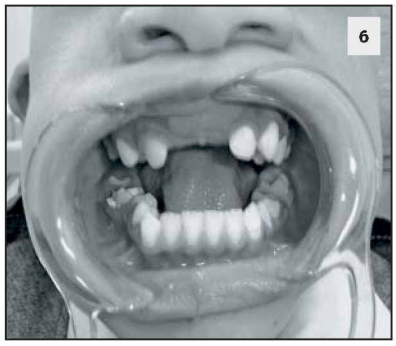
Seven days after this initial procedure, it was necessary to realize the pulpectomy of tooth no. 30 due to pulpitis. Complete endodontic treatment was only made possible after physiotherapy involving stimulation of the oral motor area, which increased the mouth opening (from 25-30 mm; Figure 6).

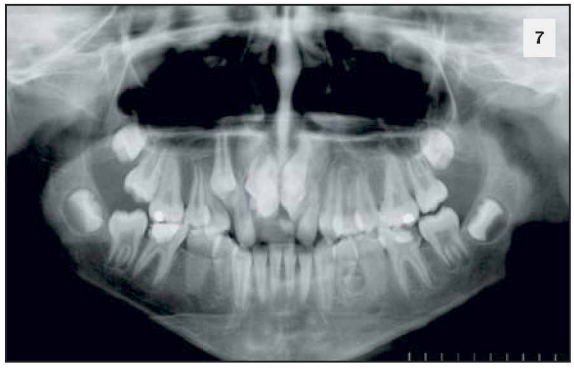
All the exercises were performed with the supervision of a professional phonologist, who instructed the mother to massage the patient's masseter muscle using up to down movements for relaxation of the muscle. The patient was also asked to keep his mouth open as wide as he could for a few minutes every day, as often as possible and to stimulate his vestibular gingiva with the tongue. No etiologic factors could be specifically related to the patient's difficulty opening his mouth. Neurological and psychological support was also used to diminish anxiety. The patient demonstrated a collaborative attitude throughout his visit to the dental office.

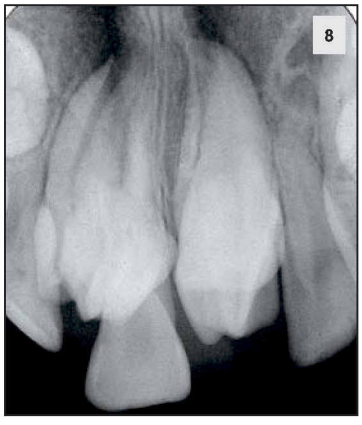
Carious lesions were found in the following teeth: permanent maxillary first molars (3 and 14); permanent maxillary left second molar (15); and permanent mandibular left second molar (18). They were filled with conventional amalgam (Standalloy SF, Degussa, Brazil). All of the mandibular and maxillary premolars (4, 5, 12, 13, 20, 21, 28, and 29) were treated with a resinous fluoride sealant (Bioseal Biodinâmica, Ibiporã, PR, Brazil). Very poor dental hygiene was noted upon examination, as the patient was accustomed to brushing his teeth by himself. To address this, information about proper oral hygiene was provided to the patient's mother and sisters, since the patient was not able to brush his teeth in a satisfactory manner. Since then, these family members are responsible for examining the patient's mouth after his brushing to ensure that proper oral hygiene measures have been taken. The family also agreed to bring the patient for periodical control examinations and to attend all oral preventive health programs offered by the public health department of the institution.

Upon dental examination, the absence of the permanent maxillary central incisors and an expansion of the vestibular plate in the region where these teeth should be was noted (Figure 5). A panoramic radiograph revealed the presence of 3 mesiodens, additional maxillary right and left supernumerary premolars, and a mandibular left supernumerary premolar (Figures 7 and 8). This was the first case of supernumerary teeth in his family. The patient was referred to an oral surgery service for the extraction of the supernumerary teeth. After the surgery, slight mobility of the permanent maxillary right central incisor (8) was noted, possibly because









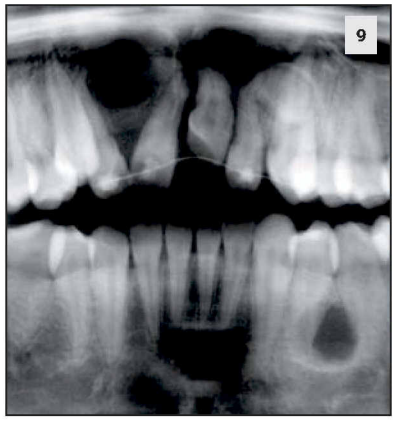


Figure 5. Absence of the maxillary central incisors and expansion of the vestibular plate caused by mesiodens.

Figure 6. Limited mouth opening; physiotherapy support involving stimulation of the oral motor area was necessary.

Figure 7. The patient's first panoramic radiograph. Maxillary right and mandibular left supernumerary premolars and mesiodens can be seen.

Figure 8. A periapical radiograph revealed three mesiodens. The maxillary right central incisor has just erupted. The maxillary right supernumerary premolar and the initial formation of a maxillary left supernumerary premolar are also visible.

Figure 9. Close view of a post-operative panoramic radiograph. The mesiodens and the premolar supernumerary teeth were extracted. Teeth in the area of surgery were held in place by an orthodontic wire until normalization of periodontal tissue. Note the initial formation of another maxillary left supernumerary premolar.

the supernumerary mesiodens were located near this tooth's root. A semi-rigid retaining appliance with an orthodontic wire and photopolimerizable resin was placed to stabilize the tooth during the time required for total bone healing (6 months). A panoramic radiograph was obtained as a baseline postoperative image. It showed the formation of an additional supernumerary tooth in the maxillary left premolar region (Figure 9). Upon removing the retaining appliance, normal periodontal tissue was present throughout the maxillary anterior segment. Currently, new teeth are erupting in reasonable position.

Since the patient presented with pulmonary stenosis, the dental extractions and endodontic treatments were performed according to the American Heart Association's (AHA) 1997 specifications for prevention of bacterial endocarditis in patients with acquired valvular dysfunction.[7]

The patient has been followed for 3 years since the initial consultation, and no additional caries have been detected. The maxillary incisors show normal periodontal tissue since the mesiodens surgery. The patient has come to the dental office once a month to help control plaque and check his oral hygiene, which has been much improved with the help of his mother and sisters. During the plaque and oral hygiene exams, endocarditis prophylaxis is no longer given because the recent guidelines from the AHA excludes valvular dysfunction as a high risk factor for infective endocarditis.[8]

The patient's family consented to the reporting of this case and the researchers also obtained the approval of the Ethics Committee of Ibirapuera University, São Paulo, Brazil.

DISCUSSION

The craniofacial characteristics of Sotos syndrome, when taken individually, are relatively nonspecific. Head circumference is almost invariably large at birth and generally remains above the 97th percentile throughout growth.[9] The patient's hands and feet are also very large. Children exhibit long, thin faces, broad foreheads, sparse frontotemporal hair, downward-slanting palpebral fissures, and malar flushing. The facial features in adults are somewhat characteristic, with the most distinctive feature being the prominence of the chin. In adults, the forehead may not appear as broad, although the hairline usually remains high.[10] In addition, patients typically exhibit macrodolicocephaly, hypertelorism, anteverted nostrils, mild micrognathia, a highly arched palate, and other signs of congenital hypotonia, including a flushed (ruddy) nose and perioral/cheek region and relatively large ears.[5] The patient in this case report demonstrated many of these nonspecific characteristics (Figures 1-4). The combination of the previously enumerated characteristics results in the characteristic appearance of Sotos syndrome.

Magnetic resonance imaging findings from Sotos syndrome patients consistently include prominent extracerebral fluid-filled spaces, modest thinning of the corpus callosum, enlarged ventricles, particularly in the trigone region, and a persistent cavum septi pellucidi and cavum vergae.[6] These characteristics were also observed in the present patient. According to Schafer et al (2007),[6] Sotos syndrome children have normal-sized brains encased within a larger-than-normal head and a unique combination of abundant ventricular and extracerebral fluid space that is not commonly seen in other macrocephalic syndromes. Although seizures have been described in 50% of Sotos syndrome patients,[1] half of these seizures were related to febrile episodes, though this patient has never presented with this symptom.

As a result of the neuroanatomical defects described above, mental deficiency is present in 80% to 85% of patients. Some Sotos syndrome patients do not have global mental retardation, but do have deficits in language, mathematics, and visual motor coordination.[11] In this case, the patient's language deficit was clear, and was the main cause of his anxiety and social interaction. In the preliminary case history, it was also discovered that his visual problems, such as nystagmus, strabismus, and severe myopia, were important factors in his behavioral problems. The patient also presented with photophobia, which necessitated the use of sunglasses during dental interventions. Although his mother did not acknowledge his mental deficiency, psychological analyses confirmed a small intelligence deficit.

Generally, the pregnancy is described as normal in mothers of Sotos syndrome children. Toxemia or preeclampsia, however, have been described as potential associated factors in several cases,[12] including the present case. An apparent increase in the incidence of spontaneous abortions has also been described in mothers who have a Sotos syndrome child.[5] Since Sotos syndrome infants are large for gestational age, many of them exhibit feeding difficulties.[1]

Some Sotos syndrome children have been shown to develop a larger-than-normal appetite later in life, and may become overweight as adults.[5] Despite this patient's normal weight at the time of the case study, his family should be alerted that his condition may predispose him to overeating.

Primary teeth may erupt early and dental age appears to be advanced in Sotos syndrome children.[5] In this case report, the patient's primary teeth began to erupt at 4 months old and were completely in place by 1-year-old. He had a significant delay, however, in the eruption of permanent teeth. The supernumerary teeth, especially the 3 mesiodens, appeared to be the cause of this delay (Figure 5). He presented with 3 other supernumerary premolars, which also had to be extracted (Figures 7-9). Supernumerary teeth are encountered in many syndromes as a consequence of dental lamina hyperactivity or horizontal proliferation.[13] In 1985, Bale et al.[14] described a patient who presented with a supernumerary tooth in the maxillary midline and a prominent lateral palatine

ridge. Furthermore, in 2001, Inokushi et al.[15] described a Sotos syndrome patient who lacked mandibular premolar teeth. Another group[16] described a case report of a Sotos syndrome patient with hypodontia. In 2007, Takei et al.[17] also described a patient who lacked several teeth.

Dental treatment was almost impossible in this case due to limited mouth opening by the patient. This is consistent with the findings of Cole et al. (1994),[1] who previously described Sotos syndrome children as having small mouths and hoarse, low-pitched voices. In this case, physiotherapy helped to increase the patient's range of mouth opening. Speech, math, and fine motor skills are also traditionally delayed in these children,[1] and therapy targeting these deficiencies in this case as well as others has helped improve the patients' outcomes. This case demonstrates the importance of an integrated treatment plan, including not only the input of other health professionals, but also a substantial involvement of the family.[18]

For Sotos syndrome patients who present with congenital heart disease, an echocardiogram is always indicated. Pulmonary stenosis was detected in this patient, and at the beginning of his dental intervention, endocarditis prophylaxis was given according to the AHA specifications in place at the time.[7] It is important to note that, at the time of this report, endocarditis prophylaxis is no longer given. The most recent AHA guidelines now recommend prophylaxis only for cardiac conditions with the highest risk of adverse outcomes from endocarditis—such as previous infective endocarditis, cardiac valve replacement, and surgically constructed pulmonary shunts or conduits.[8]

The family understood the importance of multidisciplinary assistance and was present for all consultations (dentist, phonologist, stomatologist, psychologist, and geneticist). They also followed all the dental and phonological recommendations. When the patient was referred to our service, his oral health was very poor. With help from his family, over the last 3 years he finally realized that his syndrome did not have to include oral pathologies such as caries or gingivitis.

Calm behavior is frequently seen in Sotos syndrome children,[5] and the number of children who present with behavioral disturbances is comparable to that of other pathologies involving mental deficits. Thus, the procedures for dental treatment management for this case were essentially the same as those employed in the case of a normal child. Other Sotos syndrome patients, however, may display anxious behavior (panic or phobic reactions) or irritability,[19] so the child must always be closely supervised during all procedures. Since the time of the patient's first dental intervention, we have emphasized the importance of close dental follow-up to his family, and the patient has greatly benefited from this supervision.

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