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Shprintzen-goldberg craniosynostosis: craniofacial and oral characteristics, diagnosis, and clinical management of a very rare syndrome

Craniossinostose de shprintzen-goldberg: características craniofaciais e orais, diagnóstico e manejo clínico de uma síndrome muito rara

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ABSTRACT

Shprintzen-Goldberg craniosynostosis syndrome, characterized by craniosynostosis and marfanoid habitus, is a very rare entity described in 75 individuals worldwide. This study aimed to present a case report of a 6-year-old female Brazilian child with Shprintzen-Goldberg's craniosynostosis syndrome. We described the craniofacial and oral characteristics and its clinical management, comparing to the existing literature. The patient presented with intellectual disability, craniosynostosis, ocular proptosis, low-set anomalous ears, and other skeletal and connective tissue defects. Oral features included malocclusion, micrognathia, pseudo-cleft palate, dental caries, and inefficient biofilm control. The treatment started with guidance on cariogenic foods, oral hygiene, and an indication to discontinue bottle feeding. Subsequently, the carious lesions were restored with composite resins, and the patient was referred for orthopedic surgery, orthodontic treatment, speech therapy, and nutritional counseling. The patient was followed for 5 years. This case report emphasizes the importance of knowing the craniofacial and oral characteristics for the diagnosis and clinical management of a female child with a rare Shprintzen-Goldberg's craniosynostosis syndrome. The case also highlights the need for oral health care in individuals with intellectual disabilities.

Indexing terms: Craniosynostosis. Micrognathism. Rare diseases.

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RESUMO

A síndrome Shprintzen-Goldberg é uma entidade muito rara descrita em 75 pessoas, caracterizada por craniossinostose e habitus marfanóide. O objetivo deste estudo é apresentar o relato de um caso clínico de uma criança brasileira de 6 anos com síndrome da craniossinostose de Shprintzen-Goldberg, apresentando as características craniofaciais e orais e seu manejo clínico, comparando com a literatura. O paciente apresentava deficiência intelectual, craniossinostose, proptose ocular, orelhas anômalas de implantação baixa e outros defeitos esqueléticos e do tecido conjuntivo. As características orais incluíram má oclusão, micrognatia, pseudo fenda palatina, atividade de cárie e controle ineficiente do biofilme. O tratamento foi iniciado com orientações sobre alimentos cariogênicos, higiene oral e indicação de suspensão do uso de mamadeira. Em seguida, as lesões cariosas foram restauradas com resina composta e o paciente encaminhado para cirurgia ortopédica, tratamento ortodôntico, fonoaudiologia e nutricionista. O paciente foi acompanhado periodicamente por 5 anos. Este relato de caso enfatiza a importância do conhecimento das características craniofaciais e orais para o diagnóstico e manejo clínico de uma criança do sexo feminino com síndrome de craniossinostose de Shprintzen-Goldberg rara, destacando a necessidade de cuidados com a saúde bucal em indivíduos com deficiência intelectual.

Termos de indexação: *Craniossinostose; Micrognatismo. Doenças raras.*

INTRODUCTION

Shprintzen-Goldberg craniosynostosis syndrome (SGCS) is very rare. It was first described by Shprintzen and Goldberg in 1982 [1]. It is an autosomal dominant disorder and its etiology is related to pathogenic heterozygous variants in the SKI gene that interfere with transcriptional repression of transforming growth factor-β (TGF-β) signaling, causing changes in cell differentiation and maturation [2-4]. SGCS is characterized by craniosynostosis, hypertelorism, ocular abnormalities, cardiovascular abnormalities, some features of Marfan syndrome, downward slanting palpebral fissures, high-arched palate, micrognathia, arachnodactyly, camptodactyly, abdominal hernias, hypotonia, and differing degrees of intellectual disability [1,2,5-12]. To our knowledge, the literature has reported only 75 cases of SGCS, including 43 males and 32 females [1,2, 5-12].

Thus, this study aimed to present a case report of a 6-year-old female Brazilian child with the rare Shprintzen-Goldberg's craniosynostosis syndrome, including the craniofacial and oral characteristics and its clinical management, comparing to the existing literature.

CASE REPORT

A 6-year-old female patient was referred for dental treatment at a specialized center. The child's parents signed a term of free informed consent to permit documentation of this clinical case. The diagnosis of SGCS was confirmed by a geneticist.

During anamnesis, the caregiver recalled that the child had respiratory problems but did not require continuous medication. The general physical examination verified that she had arachnodactyly, camptodactyly, congenital clubfoot, genu recurvatum, pectus excavatum, scoliosis, and dislocation and hypermobility of the joints. The presence of hypotonia, delayed development, intellectual disability, obstructive sleep apnea, and hyperelasticity of the skin, were also observed. During the craniofacial physical examination, craniosynostosis (dolichocephaly and trigonocephaly), a prominent forehead, ocular proptosis and ptosis, ocular hypertelorism, telecanthus, cleft of the lower eyelids, and posteriorly angled and low-set ears were observed (figure 1).

During the intraoral clinical examination, maxillary hypoplasia, dental malocclusion, and micrognathia were evident. The patient presented with a high-arched palate with prominent palatine ridges, pseudo-cleft palate, mouth breathing, plaque accumulation, caries lesions, dental restorations, and tooth loss (#51) due to dental trauma (figure 2).

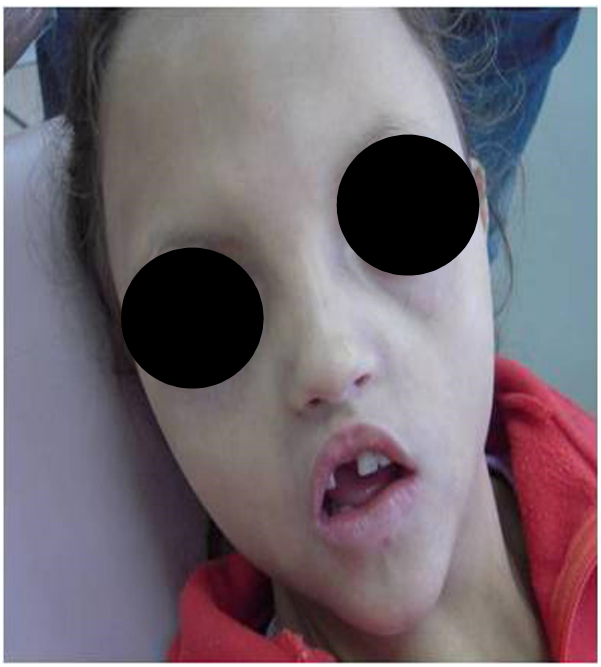

Figure 1 – The proband with Shprintzen-Goldberg craniosynostosis syndrome (SGCS) showing trigonocephaly, dolichocephaly, high prominent forehead, downward slanting palpebral fissures, ocular proptosis and ptosis, low-set ears, short philtrum, and carp-shaped mouth.

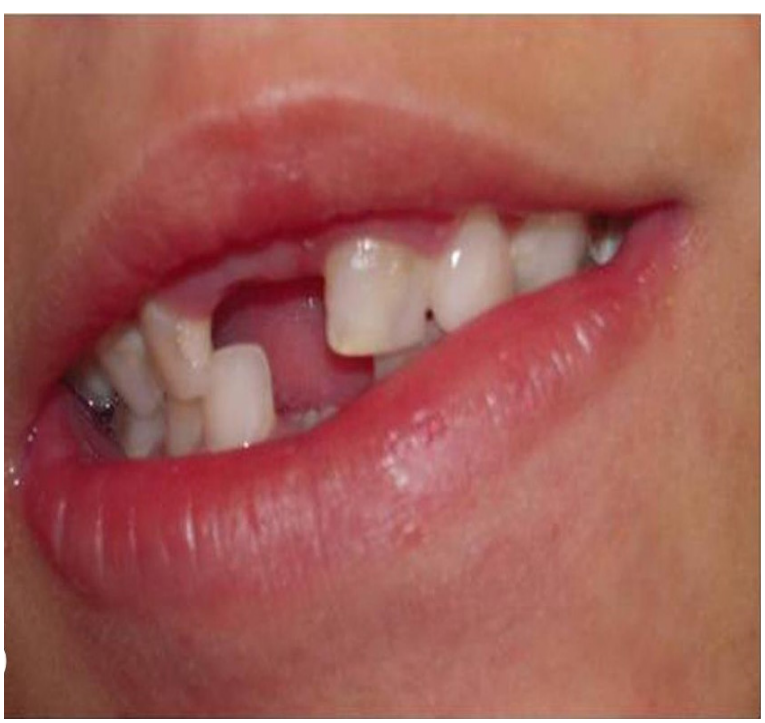

Figure 2 – Clinical examination showing plaque accumulation and higher caries risk.

The use of bottle feeding, difficulty in swallowing solid food due to malocclusion, occasional gagging, and a very cariogenic diet, were confirmed. A three-dimensional computed tomography was obtained (figure 3), in which the presence of dolichocephaly, midfacial hypoplasia, and micrognathia was evident.

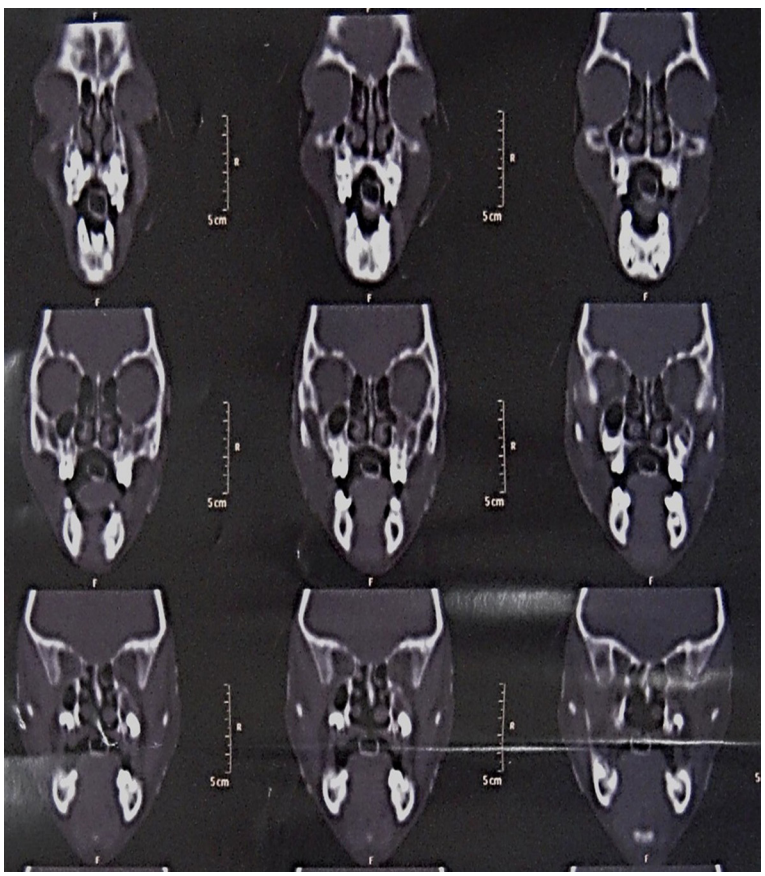

Figure 3 – Three-dimensional computed tomography showing the presence of dolichocephaly, midfacial hypoplasia and micrognathia.

Table 1 shows the craniofacial and dental features and other characteristics of patients with SGCS as described in the literature [1,2,5-12] and compared with the patient in this clinical report.

Table 1 – Craniofacial, dental, and other characteristics of patients with Shprintzen-Goldberg craniosynostosis syndrome (SGCS) as described in the literature [1,2,5-12] and compared with the patient in this clinical report.

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	Yes	No	Present case
CRANIOFACIAL FEATURES			
Craniosynostosis	67%	13%	+
Dolichocephaly	82%	9%	+
Hydrocephalus	13%	12%	-
Anterior fontanelle increased	17%	5%	-
Prominent, high forehead	36%	8%	+
Ocular proptosis	57%	13%	+
Ptosis	20%	20%	+
Ocular hypertelorism	84%	9%	+
Telecanthus	10%	-	+
Downward slanting palpebral fissures	80%	10%	+
Strabismus	30%	17%	-
Low-set ears	59%	3%	+

Table 1 – Craniofacial, dental, and other characteristics of patients with Shprintzen-Goldberg craniosynostosis syndrome (SGCS) as described in the literature [1,2,5-12] and compared with the patient in this clinical report.

	Yes	No	Present case
CRANIOFACIAL FEATURES			
Posterior angulation of ears	16%	3%	+
Malformation of the ears	22%	13%	-
Exophthalmos	30%	3%	+
DENTAL FEATURES			
Maxillary hypoplasia	41%	1%	+
High palate	67%	-	+
Prominent palatine crest	16%	4%	+
Bifid uvula	7%	24%	-
Dental malocclusion	16%	9%	+
Micrognathia	86%	5%	+
Cleft palate/Pseudo	10%	12%	+
Dental anomaly	5%	3%	-
Mouth breathing	38%	14%	+
Dislocation of joints	5%	14%	+
Articular hypermobility	49%	7%	+
Joint contractures	46%	26%	+
OTHER CHARACTERISTICS			
Hypotonia	55%	4%	+
Intellectual disability	84%	9%	+
Hernias	51%	20%	-
Obstructive sleep apnea	13%	26%	-
Skin hypermobility	24%	41%	+
Myopia or astigmatism	30%	25%	+
Mitral valve prolapse	31%	51%	-
Thin ribs	13%	-	-
Osteopenia	11%	3%	-
Cryptorchidism	11%	1%	-
Pairs of ribs	7%	-	-

Note: + present / - not present.

The first phase of care consisted of orientation concerning cariogenic foods, discontinuation of the use of bottle feeding, and motivating the family members. Then, the restorative phase was initiated, which involved composite resin restorations. The patient was referred for orthopedic surgery, orthodontic treatment, speech therapy, and nutritional counseling, to help improve her quality of life and reduce the risk of dental caries.

The patient was followed periodically for 5 years, with monthly visits to the dentist (figure 4).

Each follow-up included topical application of fluoride, dental sealants, oral hygiene with fluoride dentifrice, and gingival health maintenance.

DISCUSSION

A comparison of the physical characteristics reported in the literature was performed on 75 patients with SGCS (43 males and 32 females) and compared with the clinical case described. A higher prevalence was verified among males

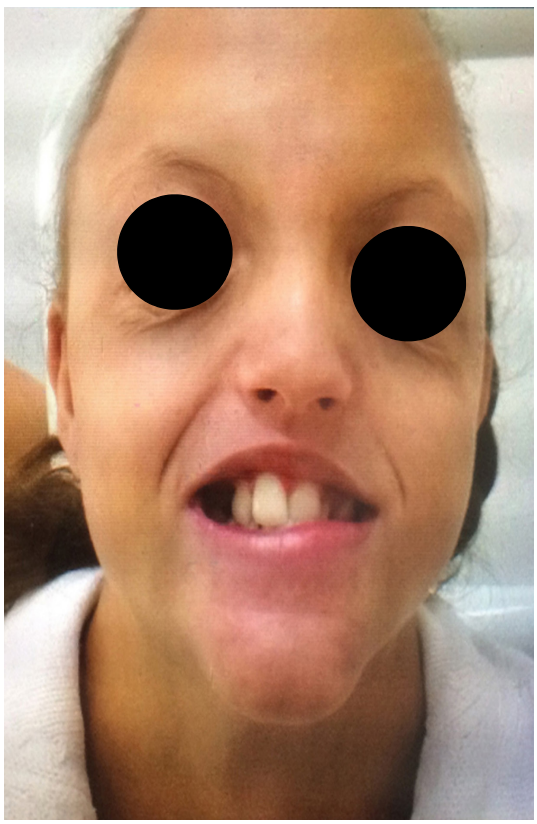

Figure 4 – The patient after 5 years of dental care, with erupted permanent teeth.

[1,5,7,8]. However, there were several reports of females with this syndrome [6,7]. This is probably a casual finding since the two genes linked to the syndrome are autosomal, i.e., they show no sex preference among those affected. Likewise, because it is a Mendelian entity with autosomal dominant inheritance, finding an important discrepancy between the sexes would be unexpected.

In general, the main craniofacial and skeletal characteristics described in the literature were identified in our proband [1,2,5-12]. This shows an important pattern related to this syndrome's features, which must be carefully evaluated by different health professionals.

Dental characteristics, such as maxillary hypoplasia [1,5-8,10], dental malocclusion [6,7], micrognathia [1,5-8,10], high-arched palate with prominent palatine ridges [1,5,7], pseudo-cleft palate (1), and mouth breathing [1,5,6,8,9], have been reported in the clinical diagnosis of SGCS. These dental characteristics were observed in the present case, corroborating the findings in the literature. It is important to mention that the dental approach to patients with SGCS should involve the integration of a multidisciplinary dental team (e.g., pediatric dentistry, special care needs, and orthodontics) to prevent dental diseases and manage dental changes, thereby maintaining good oral health and improving patient´s quality of life.

This SGCS case report described caries risks and caries activity. These may be related to the difficulty in biofilm control, probably due to arachnodactyly, micrognathia, and malocclusion. Moreover, the patient's parents showed low adherence to maintaining a sugar-restricted diet and avoiding the intake of soft drinks and other sugar-rich beverages. It is easy to inculpate sugar intake. However, doing so draws attention away from other important problems. In this case, to prevent gagging, a soft food diet was the child's main source of nutrition.

Caries risk assessment models currently consider a combination of factors, including diet, fluoride exposure, host susceptibility, and oral microflora, as well as cultural, social, and behavioral factors [13]. The highest risk of dental disease

has been reported in children from low-income families, among minorities, and those with special needs [14]. Moreover, the American Academy of Paediatric Dentistry affirms that children with special healthcare needs are in the risk category of moderate caries risk [15]. It is possible to improve the patient's quality of life, while concurrently reducing their risk of caries by developing a dental care plan adapted to a patient [16] that includes hygiene guidelines for caregivers and referring the patient to other health professionals, following a multidisciplinary management approach.

The recognition of clinical deviations based on the external morphological phenotype by the dentist is crucial for determining the etiologic factors, therapeutic planning, establishing patient prognosis, and genetic counseling. The best dental care for patients with SGCS should begin early in life. by clarifying their special oral hygiene needs with the parents to avoid unnecessary invasive treatment and reduce the risk of oral diseases.

CONCLUSION

This case report emphasizes the importance of knowing the craniofacial and oral characteristics for the diagnosis and clinical management of a female child with the very rare Shprintzen-Goldberg's craniosynostosis syndrome. The case also highlights the need for oral health care in individuals with intellectual disabilities.

Collaborators

DM VIEIRA, concepts literature search, definition of intellectual content, experimental study, manuscript preparation. FG SILVA, manuscript preparation, manuscript review. MB DINIZ, concepts, manuscript preparation, manuscript editing, manuscript review. MCD FERREIRA, experimental study, manuscript review. MTBR SANTOS, manuscript preparation, manuscript editing, manuscript review. RO GUARÉ, concepts literature search, definition of intellectual content, experimental study, manuscript preparation, manuscript review.

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