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CASE REPORT

Clinical and oral findings of a patient with Simpson–Golabi–Behmel syndrome

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Abstract
Background The Simpson–Golabi–Behmel syndrome (SGBS) is an overgrowth condition characterised by macrosomia, mental deficiency, large head, prominent skull sutures, midface deficiency, hypertelorism, broad nose, wide mouth, macroglossia, malocclusion, highly arched palate, and musculoskeletal and limb abnormalities. The aim of this case report is to present clinical and oral findings of an 8-year-old boy who had been diagnosed with SGBS.
Case report This patient had supernumerary nipples on the right side, cubitus valgus webbed fingers, scoliosis, umbilical hernia, a coarse face, macrocephaly, hypertelorism, a short broad nose, a wide mouth, a straight facial profile and hearing loss. The patient also had macroglossia, diastemas, over-retained primary tooth, absent mandibular permanent central incisors, and highly arched palate. Lateral cephalometric analysis revealed a large anterior cranial base, a large maxilla and mandible, a large inferior face height, and skeletal Class III jaw relationship.
Follow-up After extraction of the over-retained primary central tooth, a partial prosthesis was fabricated in order to maintain function. The patient has been recalled regularly at 6-month intervals for 2 years. Over the following years the prosthesis was replaced due to facial growth.
Conclusion Long term follow-up is essential for the patient with SGBS. Preventive dental care, including oral hygiene instructions, diet counselling and the use of fluoride has been implemented.

Keywords Simpson–Golabi–Behmel syndrome · Overgrowth syndrome · Oral findings

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Background

Simpson–Golabi–Behmel syndrome (SGBS) is an X-linked overgrowth disorder characterised by pre- and postnatal overgrowth, "coarseness" of face with macrostomia, macroglossia and dental malocclusion, highly arched palate, supernumerary nipples, hypospadias, polydactyly and fingernail hypoplasia, congenital heart defects, diaphragmatic hernia and enlarged viscera, including hyperplasia of the endocrine pancreas (Simpson et al. 1975; Golabi and Rosen 1984; Behmel et al. 1984; Gorlin et al. 1990; Neri et al. 1998). Intellectual disability is not constantly found and is usually mild (Young et al. 2006). There is an increased risk of embryonic neoplasia, including Wilms tumour, neuroblastoma and hepatoblastoma, with an overall tumour frequency of about 10 % (Li et al. 2001; Lapunzina 2005). Abdominal ultrasound examination should be performed every 3 or 4 months from birth until at least age 7 or 8 years, and yearly thereafter (Choyke et al. 1999; Lapunzina 2005). Abdominal ultrasound examination should assess for both Wilm's tumour and hepatic tumours. The incidence of SGBS is unclear because of the recent identification of the syndrome and the clinical overlap with other overgrowth syndromes like Beckwith Wiedemann syndrome (BWS) (Coppin et al. 1997) and Weaver syndrome (Kondo et al. 1991). SGBS is caused by mutation in the gene encoding glypican-3 (GPC3) on chromosome Xq26 (Veugelers et al.

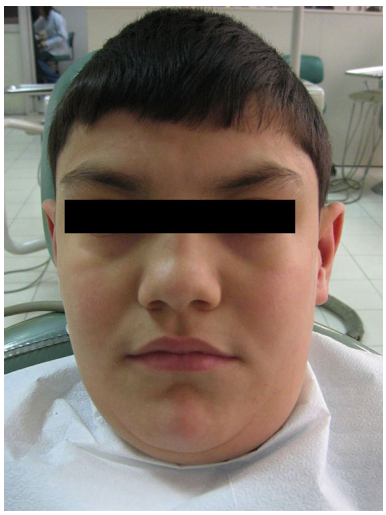

Fig. 1 Frontal facial photograph. Note a coarse face, macrocephaly, hypertelorism, broad nose, wide mouth and straight facial profile

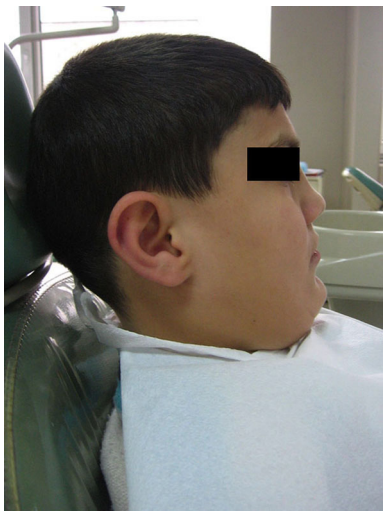

Fig. 2 Lateral facial photograph

2000; Li et al. 2001; Sakazume et al. 2007). The detection rate for GPC3 mutations and deletions in individuals with SGBS ranges widely from 37 % (7/19) (Li et al. 2001) to 70 %; (7/10) in the study of Veugelers et al. (2000) and 26/37 in the study of Lin et al. (1999). The diagnosis of SGBS is based on clinical findings, family history consistent with X-linked inheritance and molecular genetic testing of GPC3, the only gene currently known to be associated with SGBS (Pilia et al. 1996; Okamoto et al. 1999).

Very little information on clinical and oral findings of the syndrome can be found in the literature. In this case report, an 8-year-old boy who had been diagnosed with SGBS is presented.

Case report

The patient (an 8-year-old boy) had been diagnosed with SGBS at the Istanbul University, Faculty of Medicine and referred to the Istanbul University, Faculty of Dentistry, Department of Pedodontics for consultation of oral–dental findings.

He was born prematurely as the second child of consanguineous parents. Birth weight had been 3,500 g (>90th centile), length 51 cm (>90th centile) and head circumference 34.5 cm (90th centile). He had walked independently at 14 months; in addition his speech was delayed when compared with children of same age. The patient had supernumerary nipples on the right side, cubitus valgus webbed fingers, scoliosis and umbilical hernia. Clinical examination revealed a coarse facial appearance, macrocephaly, hypertelorism, broad nose, wide mouth, straight facial profile (Figs. 1, 2) and hearing loss. The child's mental development was judged to be normal. His maternal female first cousin was similarly affected

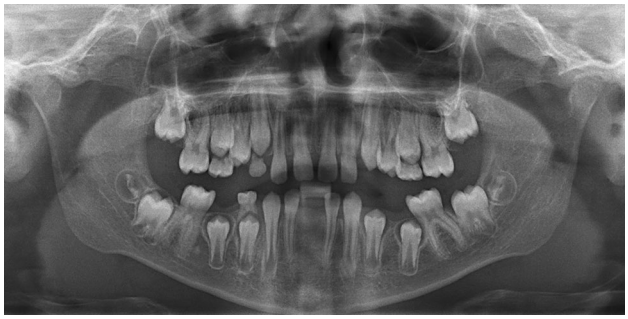

Fig. 3 Panoramic radiograph. Note congenitally missing mandibular permanent central incisors can be seen

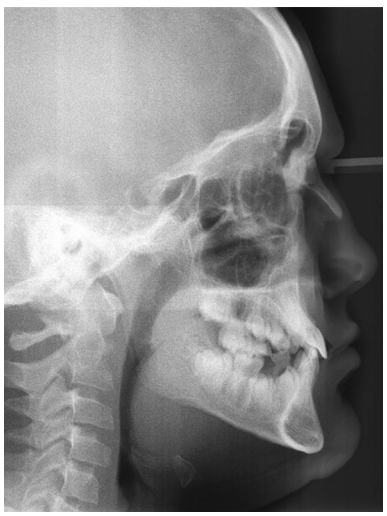

Fig. 4 Lateral cephalometric radiograph

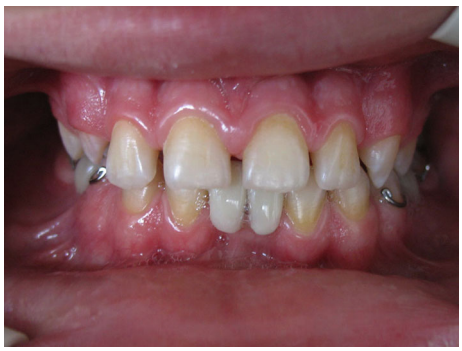

Fig. 5 Intraoral view after treatment

Oral examination revealed macrostomia, macroglossia, diastemas, an over-retained primary tooth and a highly arched palate. The panoramic radiograph (Fig. 3) revealed that the mandibular permanent central incisors were congenitally missing. Lateral cephalometric analysis (Fig. 4) revealed a large anterior cranial base [sella–nasion (SN) = 73 mm], a large maxilla [(Ptm'-A/palatal plane (PP) = 54 mm], a large mandible [gonion–menton (Go–Me): 74 mm], a large inferior face height [menton (Me)–PP = 70 mm], and a skeletal Class III jaw relationship [point A–nasion–point B line (ANB) = −2.6]. The hand–wrist radiograph on the right side revealed that bone age was 9 years old according to the method of Gruelich and Pyle (1959).

Treatment

The over-retained primary central tooth was extracted. A removable partial prosthesis was fabricated to maintain function and aesthetics (Fig. 5). The patient was referred to the orthodontic department for assessment. Treatment options for the patient with an overgrowth disorder would be the same as that of normal patients, although treatment plans should be tailored to the individual patient. Because patients are often affected psychologically by the unacceptable appearance of missing teeth, prosthodontics treatment is necessary to provide proper function and aesthetics.

Follow-up

The patient has been recalled regularly at 6-month intervals for 2 years. Over the following years, the prosthesis was replaced due to facial growth. There is sound evidence (Marinho et al. 2002, 2003; Azarpazhooh and Main 2008) that preventive dental visits improve oral health, and fluoride therapy decreases the rate of dental caries, particularly in high caries risk groups. The patient was given information about oral hygiene instructions, diet counselling and use of self-applied products at home. A professional dental prophylaxis was periodically performed and topical fluoride was applied at each follow-up visit to all teeth.

Conclusion

In summary, the patient had a coarse facial appearance, hypertelorism, broad nose, wide mouth and a straight facial profile. The patient also had an over-retained primary tooth, two congenitally missing teeth, a skeletal Class III jaw relationship with a large anterior cranial base, a large maxilla and a large mandible.

There is limited information referring to oral–dental features about SGBS in the literature. In a previous report of the craniofacial features in a patient with SGBS (Taniyama et al. 2003), the patient had similar facial profile as the present case, with five congenitally missing teeth. Further research should be carried out to propose that hypodontia may be a characteristic dental finding in patients with SGBS.

Although SGBS is not seen routinely in dental clinics, this case illustrates the importance of dental care in such a rare condition. Long-term follow-up of a patient who has overgrowth syndrome is essential.

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