

The dentofacial features of Sanjad–Sakati syndrome: a case report

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Summary. Sanjad–Sakati syndrome is an autosomal recessive disorder that was first reported by Sanjad *et al.* in 1988. It is characterized by congenital hypoparathyroidism, severe growth failure and dysmorphic features. The clinical features include deep set eyes, microcephaly, thin lips, depressed nasal bridge with beaked nose, external ear anomalies and learning difficulties. This report presents the case of a 4-year-old patient who was referred to our dental clinic because of pain in her mouth and poor dental health. Oral findings included micrognathic mandible and maxilla, microdontia, enamel hypoplasia as well as severely decayed teeth. Treatment was carried out under general anaesthesia to extract the most severely affected teeth and restore those which could be conserved. It was concluded that these patients have special dental needs; early diagnosis of the affected children is therefore important in order to commence preventive dental therapy and carry out appropriate dental treatment at the optimum time.

Introduction

Hypoparathyroidism is associated with a range of clinical conditions or syndromes characterized by parathyroid hormone deficiency, hypocalcaemia and hyperphosphataemia. Transient neonatal hypoparathyroidism is relatively common, usually occurs in association with prematurity, perinatal asphyxia and maternal diabetes, and usually resolves spontaneously in infancy [1]. Permanent congenital hypoparathyroidism is relatively rare. It may occur as an isolated condition, or with developmental anomalies, or as part of a complex syndrome such as DiGeorge's syndrome or Kenny–Caffey syndrome [2–8].

Sanjad–Sakati syndrome or Hypoparathyroidism–Retardation–Dysmorphism (HRD) (OMIM 241410) is an autosomal recessive disorder which was first reported in 1988 as a rare syndrome of congenital hypoparathyroidism associated with severe intrauterine and postnatal growth failure and dysmorphic features [9–12].

The clinical features include deep set eyes, microcephaly, micrognathia, thin lips, beaked nose,

depressed nasal bridge, external ear anomalies, small hands and feet, short stature and learning difficulties.

The patients typically present in the neonatal period with tetany, seizures or apnea due to hypocalcaemia. Patients have also been reported to have recurrent infections, probably due to immune deficits [10]. Most of the cases reported have been associated with parental consanguinity and have come from the Arabian peninsula [9,10,13–15].

A similar syndrome with the additional features of cortical thickening in the long bones with medullary stenosis and absent of diploic space in the skull has been classified as autosomal recessive Kenny–Caffey syndrome (AR-KCS; OMIM 244460). Although the skeletal findings seen in Kenny–Caffey syndrome have not been described in Sanjad–Sakati syndrome patients, the clinical similarities combined with the linkage of both conditions to chromosome 1q42–43 [8,12] suggested that the two are at least allelic disorders if not the same condition [7,8,11,12,16].

This report presents the oral/facial findings of a patient with Sanjad–Sakati syndrome who was referred to our dental clinic because of oral pain and poor dental condition.

Table 1. The biochemical data of a 4-year-old patient with Sanjad-Sakati syndrome.

	Calcium mmol/L	Phosphate mmol/L	Alkaline phosphatase IU/L	Parathyroid hormone pmol/L
Reference ranges	1.94–2.26 2.1–2.6	1.99–2.43 1.45–1.78	252 150–320	< 0.18 1.6–6.9

Case report

The patient was a 4-year-old Saudi girl, born to consanguineous parents (first degree cousins) and was a product of uneventful full term pregnancy and delivery. Her birth weight was 1800 g suggesting intrauterine growth retardation (IUGR). She was the first child of her parents and had no other siblings. The child had started to have signs and symptoms of hypocalcaemia (tetany and seizures) at the age of 7 days. Her initial biochemical investigations showed severe hypocalcaemia (serum calcium was 1.5 mmol/L) associated with high phosphate (2.44 mmol/L) and very low parathyroid hormone (0.18 pmol).

During clinical follow up she was treated with oral calcium supplements and a vitamin D preparation (Alfacalcidol – one alfa drop 0.8 µg once/day). Her serum calcium concentration ranged from 1.94 to 2.26 mmol/L, serum phosphate from 1.99 to 2.43 mmol/L but her parathyroid hormone remained persistently low (Table 1). There were no further seizures or tetany. The girl continued to have severe growth failure and failure to thrive. At the age of 4 years, her height was 72 cm (<< 3rd centile) and her weight was 5.8 kg (<< 3rd centile). This was marked as being underweight. There were no gastrointestinal symptoms. She was on Pediasure and Polycose supplements plus normal family diet.

The child had the characteristic facial features described previously in patients with Sanjad-Sakati syndrome: deep set eyes, microcephaly, prominent forehead, micrognathia, depressed nasal bridge with beaked nose, large and low set ears (Fig. 1, Table 2).

Immunological investigations showed that her immunoglobulins, complements and T lymphocyte function were all normal suggesting no immunodeficiency in this child, although she had a history of recurrent *Pseudomonas* ear discharges and frequent hospitalizations for chest infections. There was no congenital heart abnormality. Neurological examination was unremarkable although she had severe speech delay and mental retardation. Her hearing and vision were normal. She had started to walk unsupported at the age of 2 years. There were no skeletal defects.



Fig. 1. The characteristic facial features for the Sanjad-Sakati patient: deep set eyes, microcephaly, prominent forehead, micrognathia, depressed nasal bridge with beaked nose, large and low set ears, thin lips.

There were no reports of other family members being similarly affected. The case was diagnosed as Sanjad-Sakati syndrome by her paediatrician based on her clinical features, biochemical results and consanguineous family history.

Oral findings

The patient was referred to our dental clinic complaining of severe pain in her mouth and with poor dental condition. On examination the patient had micrognathic mandible and maxilla, thin lips, high

Table 2. Clinical findings, dysmorphic features and oral findings in Sanjad–Sakati syndrome (Sanjad *et al.* 1991).

Clinical findings	
	Congenital hypoparathyroidism
	Hypocalcaemia
	Hyperphosphataemia
	Extreme growth failure (intrauterine and postnatal)
	Learning difficulties
Dysmorphic features	
	Microcephaly
	Prominent forehead
	Deep set eyes
	Beaked nose
	Depressed nasal bridge
	External ear anomalies
	Small hands and feet
Oral findings	
	Micrognathic maxilla and mandible
	Thin lips
	High arched palate
	Microdontia
	Enamel hypoplasia

**Fig. 2.** The oral features of the 4-year-old child with Sanjad–Sakati syndrome showing: micrognathic mandible and maxilla, thin lips, enamel hypoplasia and severely decayed teeth.

arched palate, microdont teeth, enamel hypoplasia and severely carious teeth (Fig. 2, Table 2). Radiographic examinations were not possible.

Treatment was carried out under general anaesthesia to extract the most severely carious teeth and to restore others. Oral hygiene instruction as well as dietary advice was given. The need for regular dental visits was stressed.

Discussion

Sanjad–Sakati syndrome is a newly described disorder consisting of congenital hypoparathyroidism

Table 3 Differences between DiGeorge's syndrome and Sanjad–Sakati syndrome.

DiGeorge's syndrome	Sanjad–Sakati syndrome
Congenital heart disease	No congenital heart disease
Cellular immunodeficiency	No cellular immunodeficiency
No severe growth failure	Severe growth failure both IUGR and Postnatal
Dysmorphic features	Different dysmorphic features

associated with severe growth failure and dysmorphic features.

The condition was first described by Sanjad and Sakati in 1988 [9]. Most of the cases reported have been from the Middle East in children of consanguineous parents. Some had affected siblings who died in infancy. Reported cases have ranged from the age of 4 months to 12 years [10,13–15].

The autosomal recessive form of Kenny–Caffey syndrome is also clinically manifest as growth retardation, craniofacial anomalies, small hands and feet, hypocalcaemia, hypoparathyroidism, in addition to radiological evidence of cortical thickening in the long bones with medullary stenosis and absent diploic space in the skull.

Both conditions (AR-KCS and Sanjad–Sakati syndrome or HRD) have been mapped to the same chromosome and share an ancestral haplotype, suggesting a common founder mutation and both are chaperon diseases caused by a genetic defect in the tubulin. It has therefore been suggested that these syndromes are similar or allelic [7,8,11,12].

The condition has also been found to have some clinical resemblance to DiGeorge's syndrome which is characterized by congenital hypoparathyroidism, T cell immunodeficiency (causing chronic infections, diarrhoea or skin rashes), congenital cardiac anomalies and dysmorphic facial features [17]. However in Sanjad–Sakati syndrome there are different dysmorphic features as well as severe growth failure both intrauterine and postnatal (Table 3) [10].

Dysmorphic features are common in many genetic conditions and syndromes but the facial manifestations seen in our patient were found to be particularly consistent with previous reported cases of Sanjad–Sakati syndrome [10,13,14]. A common feature of Sanjad–Sakati syndrome patients is the tapering of the mandible towards the chin which is a unique feature of this syndrome.

The child in our case had severely decayed teeth and poor dental condition, this could have been due

to several factors. Enamel hypoplasia, which is a defect of the enamel with reduced enamel thickness, is commonly found in hereditary and environmental conditions [18,19]. Mineralization disturbances in the dental enamel have been previously reported in patients with similar disorders such as DiGeorge's syndrome [20]. The enamel disturbances found in our patient were thought to be a probable result of the hypoparathyroidism and hypocalcaemia found in this syndrome [10,13,14]. The parents reported that the child's teeth had possessed rough discoloured surfaces since the time of eruption confirming the developmental nature of the enamel defect seen. These enamel defects may better retain dental plaque, increasing the risk for dental caries.

As well as the enamel defect, because of the frequent infections that the child suffered, both sugared medicines and more cariogenic dietary habits with frequent snacks meals and drinks were consumed. It was also expected that oral hygiene procedures may not have been carried out properly when the child was ill and the infection itself may have affected salivary function. All of these factors may contribute to impaired oral health and poor dental condition.

Conclusion

This case report highlights some of the dentofacial features of Sanjad-Sakati syndrome. The report describes a 4-year-old child with this condition who presented with severe dental caries. Patients with this syndrome have special dental needs and early dental consultation is important in order to introduce effective caries preventive measures and dental therapy. Unfortunately, the condition carries a poor prognosis with death normally occurring as a result of recurrent infections and pneumonia. Early recognition of the disorder may reduce morbidity and mortality of the affected children.

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Résumé. Le syndrome de Sanjad-Sakati est une maladie autosomique récessive décrite pour la première fois par Sanjad et al en 1988. Il est caractérisé par un hypoparathyroïdisme congénital, un

déficit de croissance sévère et des dysmorphoses. Les signes cliniques comprennent des yeux enfoncés, une microcéphalie, de fines lèvres, une ensellure nasale déprimée avec un nez en bec d'oiseau, des anomalies de l'oreille externe et un retard mental. Cet article décrit le cas d'un patient âgé de 4 ans adressé à notre clinique dentaire pour des douleurs buccales et une hygiène buccale déficiente. Les caractéristiques buccales incluaient une micrognathie maxillaire et mandibulaire, une microdontie, des hypoplasies de l'émail de même que des dents très cariées. Le traitement a été effectué sous anesthésie générale afin d'extraire les dents les plus cariées et restaurer celles qui pouvaient l'être. En conclusion, ces patients présentent des besoins dentaires spécifiques. Un diagnostic précoce est donc important afin de mettre en place une action de prévention dentaire et d'effectuer les traitements dentaires appropriés au moment optimal.

Zusammenfassung. Sanjad-Sakati Syndrom ist eine autosomal rezessive Erkrankung, die von Sanjad et al. 1988 erstmals beschrieben wurde. Sie ist charakterisiert durch angeborenen Hypoparathyreoidismus, ausgeprägten Minderwuchs und Dysmorphien. Klinisch zeigen sich tiefsitzende Augen, Mikrocephalie, dünne Lippen, ein tiefsitzender Nasenansatz mit einer gebogenen Nase, Anomalien der äußeren Ohren und mentale Retardierung. In diesem Fallbericht wird über eine vierjährige Patientin berichtet, die aufgrund von Schmerzen im Mundbereich und ihres Zahnstatus in die Zahnklinik überwiesen wurde. Der Mundbefund zeigte mikrognathe Ober- und Unterkiefer, Mikrodontie, Schmelzhypoplasie und ausgeprägte Zahnzerstörungen. Die Behandlung wurde unter Vollnarkose durchgeführt mit Extraktion der am schwersten betroffenen Zähne und der Wiederherstellung der erhaltungswürdigen Zähne. Es wurde gefolgert, dass betroffene Patienten spezielle dentale Betreuung benötigen, die frühe Diagnosestellung ist wichtig, um rechtzeitig präventive Maßnahmen zu ergreifen und ggf. Zahnbehandlungen zu einem optimalen Zeitpunkt auszuführen.

Resumen. El síndrome de Sanjad-Sakati es una alteración autosómica recesiva que fue descrita por primera vez por Sanjad y col. en 1988. Se caracteriza por hipoparatiroidismo congénito, disminución severa del crecimiento y características dismórficas. Las características clínicas incluyen ojos hundidos, microcefalia, labios delgados, puente nasal deprimido

con nariz curva, anomalías externas de las orejas y retraso mental. Este informe presenta el caso de una paciente de 4 años que fue referido a nuestra clínica dental a causa de un dolor en boca y mala salud dental. Los hallazgos bucales incluyeron micrognatismo mandibular y maxilar, microdoncia, hipoplasia del esmalte así como dientes con caries severa. El tratamiento se realizó bajo anestesia general para extraer los dientes con caries más severa y restaurar aquellos que podían ser conservados. Se concluyó que estos pacientes tenían necesidades dentales especiales, el diagnóstico precoz de los niños afectados es por lo tanto importante para comenzar el tratamiento preventivo y poder llevar a cabo el tratamiento dental apropiado en el momento óptimo.

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