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Crouzon syndrome in a fraternal twin: A case report and review of the literature

Xiao-Jing Li, Ji-Mei Su, Xiao-Wei Ye

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Abstract

BACKGROUND

Crouzon syndrome (CS; OMIM 123500) is an autosomal dominant inherited craniofacial disorder caused by mutations in the *fibroblast growth factor receptor 2* (*FGFR2*) gene. CS is characterized by craniofacial dysostosis, exophthalmos, and facial anomalies with hypoplastic maxilla and relative mandibular prognathism.

CASE SUMMARY

Our report involves a 6-year-old fraternal twin boy with many caries in the oral cavity who presented with characteristic features of CS based on clinical and radiographic examinations along with Sanger sequencing. The fraternal girl did not show any abnormalities indicating CS. Carious teeth and poor oral hygiene were managed promptly through administering appropriate behavior guidance, orthodontic treatment was planned, and preventive procedures were described.

CONCLUSION

CS could occur in a fraternal twin caused by a de novo mutation of the *FGFR2* gene. Oral hygiene instruction, preventive programs on oral hygiene, orthodontic treatment, and maxillary osteotomy were required for treatment.

Key Words: Crouzon syndrome; Craniosynostosis; *Fibroblast growth factor receptor 2*; Mutation; Fraternal twin; Case report

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Core Tip: Crouzon syndrome (CS) is an autosomal dominant inherited craniofacial disorder caused by mutations in *fibroblast growth factor receptor 2*, but approximately 50% of cases result from de novo mutations. This syndrome has been rarely seen and evaluated in fraternal twins, only one of whom has CS. We presented a 6-year-old fraternal twin boy diagnosed with CS who had many caries and enamel hypomineralization in the oral cavity. The boy's parents and his fraternal twin sister did not show any abnormalities indicating CS, so we hypothesize that the fraternal twin boy's gene mutation arose from a de novo mutation.

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INTRODUCTION

Crouzon syndrome (CS) is an autosomal dominant inherited disorder and characterized by calvarial deformities with craniofacial dysostosis, exophthalmos, and facial anomalies[1,2]. In 1912, the French physician Louis Edouard Octave Crouzon first reported this syndrome and identified a mother and her son with craniosynostosis that resulted in skull and facial deformities[3]. This syndrome is described as the mildest type of many craniosynostosis syndromes[3,4]. The birth incidence of CS is approximately 1:25000[4]. It is proven to be the most common craniosynostosis syndrome, as it accounts for nearly 4.8% of all craniosynostosis cases at birth[1,5]. It is more common in males than females (3:1)[6]. Mutations in the *fibroblast growth factor receptor 2* (*FGFR2*) gene on chromosome locus 10q25.3-q26 are related to CS[7]. Families with CS commonly suggest an autosomal dominant inheritance pattern[8,9]. The syndrome presents incomplete penetrance and variable expressivity leading to phenotypical characteristics varying from mild to severe[7,10]. About 50% of cases result from de novo mutations of the *FGFR2* gene [11].

CS is commonly observed at birth owing to characteristic facial and cranial deformities and a positive family history. The prominent manifestations of these patients can be detected upon physical examination. Brachycephaly, hypertelorism, proptosis, flattened forehead, beaked noses, and maxillary/midface hypoplasia are the most common features[3,12-14]. Hearing loss, strabismus (misaligned eyes), a cleft lip and palate, and dental problems can be also seen from these patients[12,15,16]. However, Crouzon syndrome patients have normal hands and feet in contrast to Apert syndrome patients and other craniosynostosis syndromes[17,18]. The different characteristics of the more common syndromic craniosynostosis syndromes are shown in Table 1[17,19-22].

The most common dental problems in craniosynostosis are supernumerary teeth, hypodontia, delayed eruption, and macrodontia[23,24]. We report a 6-year-old fraternal twin boy with CS who had many caries and enamel hypomineralization in the oral cavity. The boy's parents and his fraternal twin sister did not show any abnormalities indicating CS. Therefore, we hypothesize that the twin boy's gene mutation arose from a de novo mutation.

CASE PRESENTATION

Chief complaints

A 6-year-old Chinese boy was referred to the stomatology department of the Children's Hospital, Zhejiang University School of Medicine, with a chief complaint of dental caries.

History of present illness

Dental caries were observed since the patient was 4-years-old and gradually became progressively larger.

History of past illness

The patient underwent intracranial decompression surgery at the age of 1 due to hydrocephalus. Posterior cranial vault distraction was performed when he was 2-years-old. The patient underwent a ventriculoperitoneal shunt operation for hydrocephalus at the age of 3. One year later, the patient had cranial vault remodeling as a result of craniofacial dysostosis.

Personal and family history

His medical and developmental history showed that he was born at full-term by cesarean delivery after

Table 1 Clinical characteristics of the most common syndromic craniosynostosis syndromes

	Crouzon	Apert	Pfeiffer	Muenke	Saethre-Chotzen
Gene	FGFR2	FGFR2	FGFR1/ FGFR2	FGFR3	TWIST1
Craniofacial phenotype	Brachycephaly; shallow orbits; ocular proptosis; midface hypoplasia	Turribrachycephaly; large anterior fontanelle with bitemporal widening and occipital flattening; shallow orbits with ocular proptosis and horizontal grooves above supraorbital ridges; mild hypertelorism and down-slanting palpebral fissures; "parrot beak" nasal deformity	Turribrachycephaly; midface hypoplasia; proptosis; hypertelorism; strabismus; down-slanting palpebral fissures; beaked nasal deformity	Craniosynostosis of coronal sutures; uncommon to have midface hypoplasia	Brachycephaly or acrocephaly or anterior plagiocephaly; low frontal hairline; eyelid ptosis; facial asymmetry; deviated nasal septum; ear deformities; parietal foramina
Oral and dental phenotype	Constricted, high-arched palate; anterior open bite; Missing teeth; supernumerary; delayed tooth eruption	High arched or cleft palate; severe midface hypoplasia; anterior open bite; tooth agenesis; supernumerary teeth; dental fusion; delayed tooth eruption	Anterior open bit and bilateral posterior cross-bites; hypodontia; microdontia; dilacerations; radicular dentin dysplasia	High arched palate; the lowest incidence of cleft palate	Cleft palate
Limb phenotype	Normal hands and feet	Severe syndactyly (mitten hand); poor joint mobility	Broad thumbs; broad great toes; variable feature, partial soft-tissue syndactyly of the hands; ankylosed elbows; hydrocephalus	Thimble-like middle phalanges	Brachydactyly; syndactyly; radioulnar synostosis; Hallux valgus
Other phenotype		Low-set ears	Hearing loss; visceral anomalies	Hearing loss	Hearing loss; congenital heart malformation; short stature
Cognition	Normal	Intellectual disability	Normal or nearly normal		Normal

FGFR: Fibroblast growth factor receptor.

an uneventful pregnancy with a 48 cm height and 2.35 kg weight, along with his fraternal twin sister with a 48 cm height and 1.8 kg weight. The family history of going back for three generations was negative for similar conditions, and his fraternal twin sister had no medical issues.

Physical examination

Physical examination showed normal height (108 cm) and weight (20.4 kg) compared with that of children of the same age. Clinically normal hands and feet were found. Extraoral examination revealed a prominent forehead, eyelid ptosis, ocular proptosis, shallow eye sockets, hypertelorism, midface hypoplasia, and a retrusive upper lip and protrusive lower lip (Figure 1).

Intraoral examination showed that the patient was in early mixed dentition, with all deciduous canines and molar teeth present in the maxillary and mandibular arch. The chronology of eruption and eruption status was in accordance with the child's age (Figure 1). The oral hygiene of the child was poor with scattered pigmentation on the teeth, and permanent teeth 36 and 46 and deciduous teeth 54, 55, 65, 74, 75, and 85 were mostly decayed. The eruptive permanent teeth, including 11, 21, 36, and 46, showed enamel hypomineralization. The maxillary arch was constricted, and the anterior cross bite and open bite are shown.

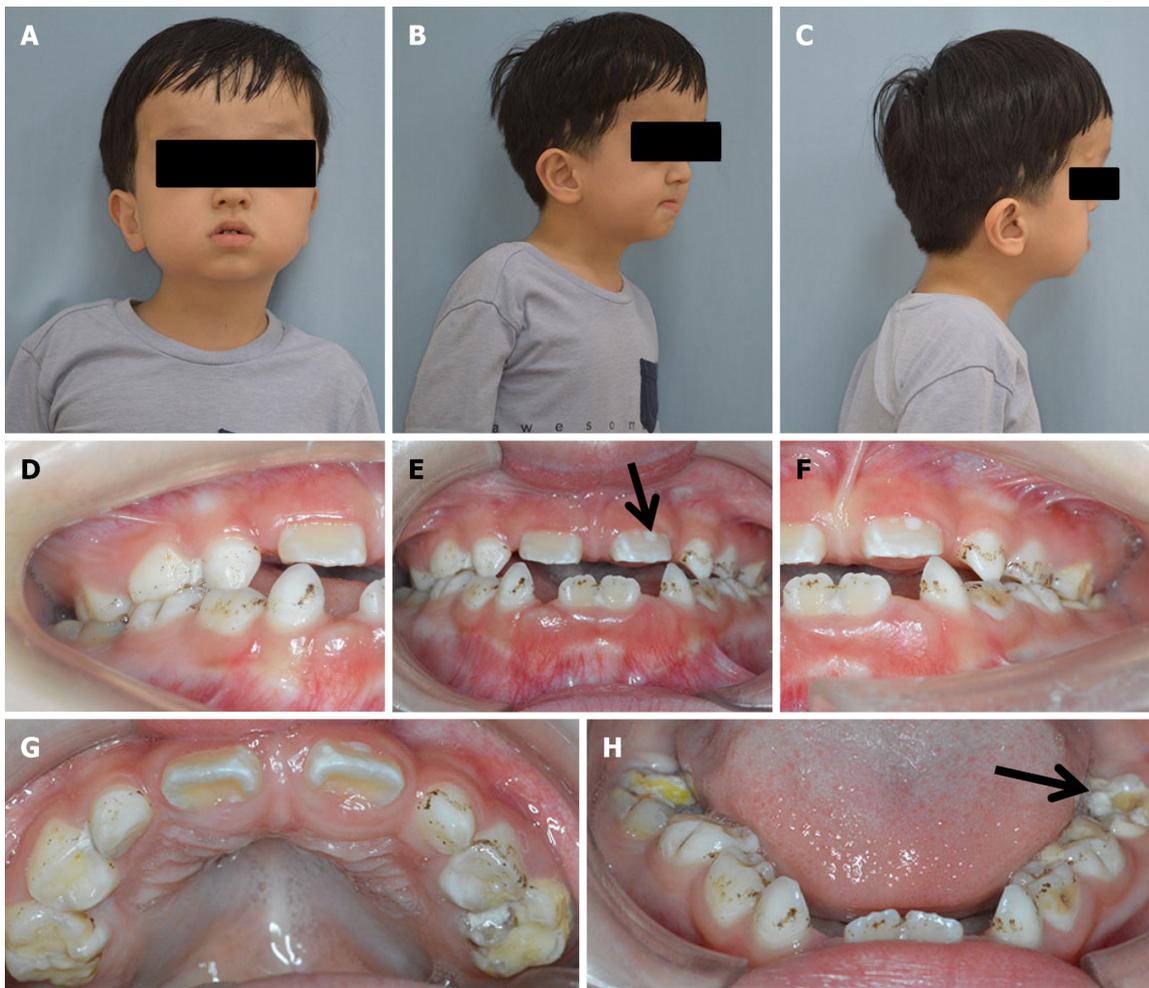
Laboratory examinations

Sanger sequencing showed that the pathogenic variant c.1026C>G (p.Cys342Trp) was present on the *FGFR2* gene for the patient. The mutation was not present in any of the family members.

Imaging examinations

The panoramic radiograph showed that there were no supernumerary teeth or normal permanent tooth germ development. Deciduous carious teeth 55, 64, 65, 75, and 85 had once been restored and displayed further second caries at the time of the study. Permanent teeth 36 and 46 and deciduous teeth 54 and 74 were decayed (Figure 2A).

Based on the lateral cephalometric analysis, a severe skeletal Class III relationship was diagnosed with Sella-Nasion-A point angle: 63.8°, Sella-Nasion-B point angle: 71.6°, and A point-Nasion-B point angle: -7.8° (Figure 2B). A hypodivergent facial pattern was shown with Sella-Nasion/Gonion-Gnathion: 35.1°. The cephalometric analysis also revealed the maxillary skeletal size (condylion to point A) was diminished. The upper central incisors leaned toward the lip (U1/Sella-Nasion: 112.8°).



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Figure 1 Extraoral and intraoral presentations of the patient. A-C: Extraoral photos showed a prominent forehead, ocular proptosis, midface hypoplasia, retrusive upper lip, and protrusive lower lip; D-H: The intraoral photos showed that oral hygiene was poor with scattered pigmentation on the teeth. Teeth 36, 46, 54, 55, 65, 74, 75, and 85 were decayed, and teeth 11, 21, 36, and 46 showed hypomineralization (black arrows).

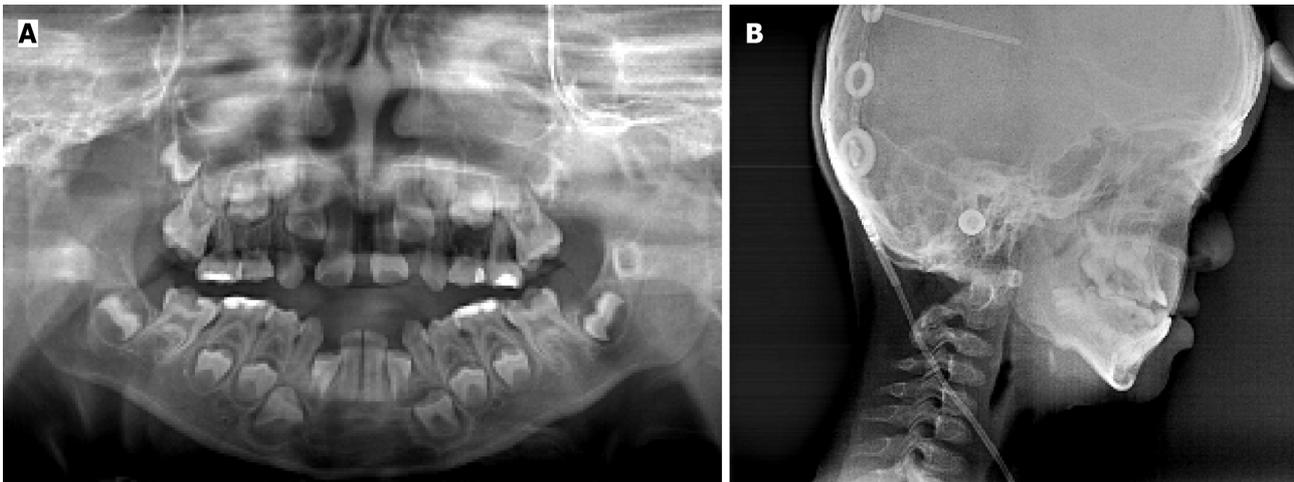
FINAL DIAGNOSIS

The final diagnosis of the presented case was CS, caries, and enamel hypomineralization.

TREATMENT

A comprehensive treatment plan was made. The initial treatment involved improving the patient’s oral hygiene and restoring his carious teeth. The dental caries in teeth 54, 55, 64, 65, 74, 75, 85, 36, and 46 were treated with resin-based restorations. Topical fluoride was regularly applied to enamel hypomineralization in teeth 11, 21, 36, and 46 to promote enamel remineralization. The Skeletal Class III malocclusion was treated by orthodontic treatment and orthognathic surgery. Multidisciplinary approaches involving pediatric dentists, dental surgeons, oral and maxillofacial surgeons, and orthodontists were indispensable for this treatment.

At the first dental visit, the patient was nervous and fearful and was unwilling to open his mouth. Appropriately administered behavior management and guidance[25,26], including show-and-tell tactics, positive reinforcement, sound communication, and modeling, were used to manage the patient’s anxieties. Oral examination, radiographic assessment, oral prophylaxis, and polishing of the teeth with the rubber cup were performed during the first dental appointment. For the latter dental appointments, carious restoration was successfully completed utilizing show-and-tell tactics, positive reinforcement, and distracting the patient by showing movies during the procedure. After the procedure, the patient was praised and given a post-visit sticker, and he was agreeable for a future visit.



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Figure 2 Panoramic and lateral cephalometric radiograph of the patient. A: The panoramic radiograph showed normal permanent tooth germ development; B: The lateral cephalometric radiograph showed a skeletal Class III relationship.

OUTCOME AND FOLLOW-UP

Three months later, the oral hygiene of the patient was good, and topical fluoride was applied in teeth 11, 21, 36, and 46 to promote enamel remineralization.

DISCUSSION

Craniosynostosis results from the premature fusion of one or multiple cranial sutures, resulting in restricted growth of the skull, brain, face, and central nervous system development[4]. CS is the most common syndromic craniosynostosis and is caused by the mutation of *FGFR2*[17]. *FGFR2* belongs to a family of four *FGFRs*. The *FGFR* family plays a primary role in the growth and differentiation of mesenchymal and neuroectodermal cells by binding to FGF and initiating signal transduction. Additionally, the *FGFR* family regulates cranial suture fusion on a macroscopic level[7].

For our patient the c.1026C>G (p.Cys342Trp) mutation on the *FGFR2* gene caused a cysteine-to-tryptophan substitution at amino acid 342. Mutations at amino acid 342 can cause CS and Pfeiffer syndrome. Normal hands and feet can be observed with CS, which differentiates it from Pfeiffer syndrome. The loss of this cysteine residue is one of the most common mutations in CS patients and has previously been reported in a Chinese family, a Japanese sporadic case, and 4 Caucasian cases[9,27-29]. The Chinese family and 1 Caucasian family with CS have a dominant inherited amino acid 342 mutation [9,27]. The Japanese case and the German patients with CS were sporadic[28,29]. Since the fraternal twin sister and the parents did not present gene mutations and similar presentations, our patient's mutation was considered to be a de novo mutation. In 11 of the 21 families with CS who underwent tracing to determine the origin of the mutation by analyzing parents and the other family members, *FGFR2* mutations arose de novo, representing a high mutation rate for *FGFR2*[29].

This case report describes a patient with CS, who presented with a high forehead, ocular proptosis, shallow eye sockets, hypertelorism, palpebral ptosis, and a skeletal Class III relationship. The carious teeth were fully restored, and oral hygiene instruction was performed in the clinic under behavior management and guidance. The child was advised to follow-up regularly with preventive care for teeth. The boy was satisfied with his good oral hygiene. Interceptive orthodontics was contemplated. Midfacial advancement with a Le Fort III, Le Fort II plus zygomatic repositioning, monobloc, or facial bipartition is required for midface retrusion[30]. The appropriate time of surgery is deemed at 8 years of age[31]. He has been referred to the Department of Oral and Maxillofacial Surgery for orthognathic surgical procedures in the next few years. He has also been referred to the Departments of Neurosurgery, Ophthalmology, Otolaryngology, and Psychology to prevent complications.

CONCLUSION

In summary, CS could occur in a fraternal twin caused by a de novo mutation of the *FGFR2* gene and is characterized by craniosynostosis, a prominent forehead, midface hypoplasia, and proptosis. Oral hygiene instruction and preventive programs on oral hygiene should be performed regularly. A

multidisciplinary approach involving oral and maxillofacial surgeons and orthodontists was necessary for the treatment of midface hypoplasia.

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