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Case Report

CROUZON SYNDROME: CASE REPORT AND REVIEW OF LITERATURE

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ABSTRACT
Crouzon syndrome is a rare genetic disorder with autosomal dominant inheritance with the prevalence of 1 in 25,000 live births, and it constitutes 4.8% of all craniosynostosis. Maxillary hypoplasia, craniosynostosis, shallow orbits, ocular proptosis and hypertelorism are the characteristic features of Crouzon syndrome. This report describes a patient with crouzon syndrome showing characteristic dentofacial deformities, along with review of literature.

Keywords: *Craniosynostosis, Crouzon Syndrome, Dentofacial Anomalies*

INTRODUCTION
Craniofacial abnormalities are usually present at birth and may progress with time. These cranial malformations are not very common, but it compromises not only the function but also the mental well-being of the person. Crouzon syndrome, is an autosomal dominant disorder caused by a mutation in the Fibroblast Growth Factor Receptor 2 gene (FGFR2) (Maloth *et al.*, 2010). It is characterized by craniosynostosis, shallow orbits, ocular proptosis, midface hypoplasia, and a curved, beaklike nose. It accounts for approximately 4.8% of all cases of craniosynostosis making it the most common syndrome within the craniosynostosis group. The worldwide prevalence rate approximately accounts for 1 per 25,000 live births (Maloth *et al.*, 2010; Singer *et al.*, 1997), indicating the rarity of the syndrome. In this article, we present a case of 46-year-old female patient with crouzon syndrome showing characteristic features.

CASE REPORT
A 46 year old mentally retarded unmarried female patient reported our clinic with a complaint of bulging of eyes [Figure1]. Detailed family and medical history revealed, mother had normal labor and delivery. There were no anomalies in any siblings or near relatives.

Figure 1: Clinical photograph at presentation

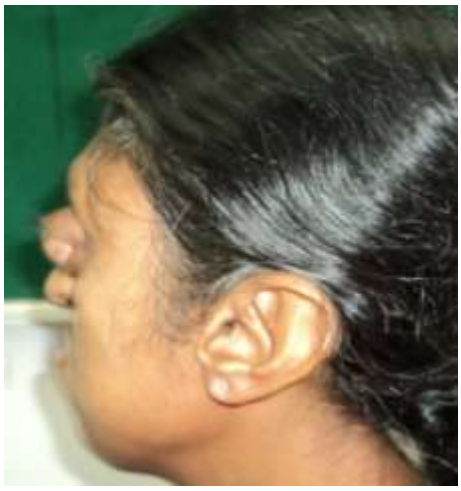

Figure 2: Lateral profile of showing maxillary deficiency with relative mandibular prognathism

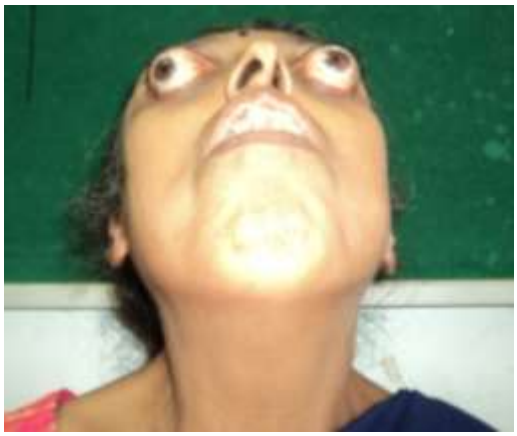

Figure 3: Clinical photograph showing wide nasal bridge, elliptical-shaped head, shallow orbits and presence of prominent eyeballs

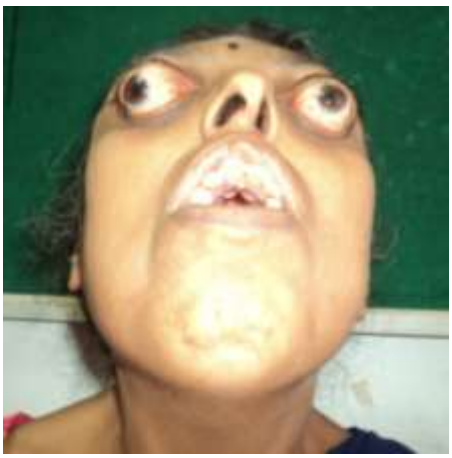

Figure 4: Clinical photograph showing high arch palate, hypoplastic maxilla

On general examination and extra-oral examination, the patient was of short stature, had a wide nasal bridge, elliptical-shaped head, concave profile, maxillary retrognathism [Figure 2], malar deficiency, shallow orbits, ocular proptosis and hypertelorism The presence of prominent eyeballs [Figure 3], which is the characteristics of the Crouzon disease triad, can be observed. The enlarged size of the head and

bulging of eyes was noticed by the mother when she was at a younger age and the severity had gradually increased.

Intra-oral examination revealed high arch palate, hypoplastic maxilla causing prognathism of the mandible [Figure 4], and class III malocclusion. Ophthalmic evaluation was also done.

DISCUSSION

Crouzon syndrome was first described by Octave Crouzon in 1912 as one of the varieties of craniofacial dysostosis caused by premature obliteration and ossification of two or more sutures, most often coronal and sagittal (Shaffer *et al.,* 2006) and suggested the term "dysostose craniofaciale hereditare" (Gorlin *et al.,* 1976). It is a rare genetic disorder. The syndrome is also referred to as craniofacial dysostosis, hereditary craniofacial dysostosis, dysostosis craniofacialis (Gorlin *et al.,* 1976), syndromic craniosynostosis and premature craniosynostosis (Davis and Lauritzen, 2008).

It was initially described as hereditary syndrome of craniofacial synostosis, this includes a triad of skull deformities, facial anomalies, and exophthalmos (Maloth *et al.,* 2010; Stephaine *et al.,* 2008). Similar characteristic features were seen in our patient.

It's an autosomal dominant disorder with complete penetrance and variable expressivity (Maloth *et al.,* 2010; Stephaine *et al.,* 2008). Its incidence is estimated at 1 in 25,000 births (Singer *et al.,* 1997; Pharaoh, 2005). Of these cases, 67% are familial (Gorlin *et al.,* 2001), whereas 33–56% may arise as a result of spontaneous mutations (Pharaoh, 2005).

According to study conducted by Kriberg, new mutations due to increased parental age played a significant role in the etiology of this syndrome (Gorlin *et al.,* 2001). It is caused by mutations in the fibroblast growth factor receptor-2 (FGFR2) gene which is mapped to chromosome locus 10q25 - 10q26, but exhibits locus heterogeneity with causal mutations in FGFR2 (CS) and FGFR3 (CS with acanthosis nigricans) in different affected individuals (Maloth *et al.,* 2010; Stephaine *et al.,* 2008; Pournima *et al.,* 2011; Haroop *et al.,* 2006).

There has been no reported racial or sex predilection (Padmanabham *et al.,* 2011). But increased predominance is seen in boys when the craniosynostosis is of sagittal or metopic types, while coronal craniosynostosis is more common in girls (Maloth *et al.,* 2010; Pournima *et al.,* 2011). The condition is usually noticed during in the first year of life (Maloth *et al.,* 2010). However, in the congenital premature forms the synostosis begins inside the uterus and is evident at birth with facial deformities (Singer *et al.,* 1997).

Simple craniosynostosis and other syndromic craniosynostosis must be differentiated from crouzon syndrome. Differential diagnosis includes Apert syndrome and other problems including Carpenter syndrome, Pfeiffer syndrome, Seatre-Chotzen syndrome, and Jackson Weiss syndrome. Also acanthosis nigricans associated condition can have FGFR3 mutation (Gorlin *et al.,* 2001).

Clinical feature of Crouzon syndrome include skull deformity which may be brachycephaly, oxycephaly or trigonocephaly. Hydrocephalus and mental retardation may be present because of premature fusion of cranial sutures (Noetzel *et al.,* 1985). Once the sutures close; growth potential of those sutures is restricted. Multiple sutural synostoses frequently results in premature fusion of skull base causing midface hypoplasia, shallow orbit, maxillary hypoplasia, and occasional upper airway obstruction (Padmanabham *et al.,* 2011). Other clinical features include hypertelorism, exophthalmos, strabismus, beaked nose, short upper lip, maxillary hypoplasia, and relative mandibular prognathism with no digital abnormalities (Haroop *et al.,* 2006; Babic and Babic, 2009; Rani *et al.,* 2012). The condition can be differentiated from other craniosynostosis syndromes by lack of abnormalities in hand or foot or both (Pournima *et al.,* 2011).

Various innovations in field of craniofacial surgery have enabled patients to achieve their full potential by maximizing their opportunities for intellectual growth, physical competence, and social interaction (Stephaine *et al.,* 2008; Gorlin *et al.,* 2001). Crouzon syndrome in adults, as seen in our case which presented with marked midface hypoplasia and exorbitism, can be corrected by orbital decompression and zygomaticomaxillary advancement (Arathi *et al.,* 2007; Taglialatela *et al.,* 2008), but the prognosis depends on the severity of malformations and the timing of intervention.

Conclusion

Craniofacial abnormalities diagnosis and management has always been challenge in clinical practise, an understanding of these abnormalities is necessary to ensure that the patient receives the best available care. Early diagnosis and management of crouzon syndrome is essential, because as age advance it results in impaired facial appearance and other complications like mental retardation, airway obstruction, and decreased visual acuity. With proper and timely management, these patients can be provided with better quality of life.

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