

Crouzon's Syndrome: A Case Report from Rural Medical College with Review of literature.

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Abstract: Crouzon's syndrome (CS) is a rare autosomal dominant condition with multiple mutations of the fibroblast growth factor receptor (FGFR2) gene. (2) CS accounts for 4.8% of all cases of craniosynostosis. The characteristic features are premature closure of cranial sutures, cranial deformities, midface hypoplasia, relative mandibular prognathism, hypertelorism, proptosis, strabismus and short upper lip, crowding of teeth, pseudocleft or sometimes cleft palate and other associated abnormalities. Here, we report a case of this rare entity. The patient presented with plagiocephaly, maxillary hypoplasia, exophthalmos, mandibular prognathism, along with dental and orbital abnormalities with delayed menarche.

INTRODUCTION

French neurologist, Octave Crouzon (1874-1938) first described in 1912 a hereditary syndrome of craniofacial dysostosis in a mother and her daughter which included a triad-cranial deformities, facial anomalies and exophthalmos¹. CS one of the varieties of craniosynostosis caused by premature obliteration and ossification of two or more sutures. It may be transmitted as an autosomal dominant inheritance but 25% of cases represent fresh mutations. Over 100 syndromes with craniosynostosis have been described of which Apert and Crouzon's syndromes are well known. CS presents similar craniosynostosis as in the Apert, Pfeiffer and Saethre-Chotzen syndromes except with no digital abnormalities⁴. The appearance of CS can vary in severity from a mild to severe forms with multiple fused cranial sutures and marked mid-face and ocular defects. MR is not a feature of CS.

We report one such case of CS showing all the classical features.

CASE REPORT

16 year old girl presented to our dept for abd-pelvic usg which revealed small size uterus & ovary for her age. On exam pt had craniofacial abnormalities plagiocephaly exophthalmos. Depressed nose high arched palate, hypertelorism. Radiological study showed thinned calvaria with silver beaten appearance, maxillary hypoplasia, proptosis, hydrocephalus and chiary malformation(tonsillar herniation with syrinx). Spine and hands are normal.



Figure 1: Patient photograph shows (a) maxilla prognathism of mandible and exophthalmos. **Figure 2:** Patient photograph shows hypoplastic maxilla, hypertelorism (white arrow), parrot beak nose

DISCUSSION

In 1912, a French neurologist, Octave Crouzon¹ first described the hereditary syndrome of craniofacial synostosis. CS is an autosomal dominant disorder caused by mutations in the fibroblast growth factor receptor-2 (FGFR2) gene². CS has no racial or sex predilection. When the craniosynostosis is of sagittal or metopic types, the predominance increases in boys, while coronal craniosynostosis is more common in girls^{3,4}. Premature closure of cranial sutures most



Figure 3: Patient photograph of Intra oral view shows high arch palate and malocclusion



Figure 4: Lateral skull projection reveals mandibular prognathism, maxillary hypoplasia, copper beaten appearance. Hand x-ray reveals mild clinodactyly

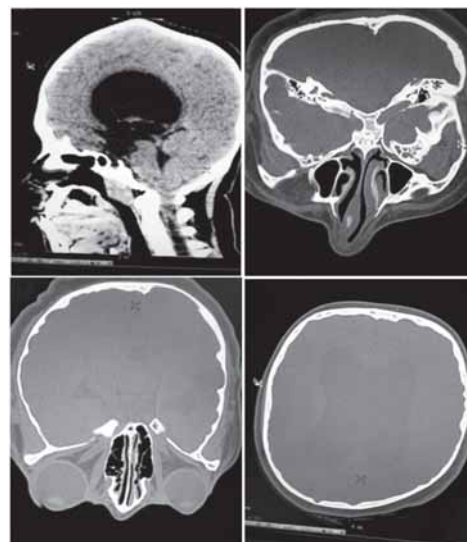


Figure 5: NECT Brain showing hydrocephalus, plagiocephaly, maxillary hypoplasia, proptosis and chiary 1 malformation.

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commonly the coronal and sagittal ones results in abnormal skull growth and affects the growth and development of the orbits and maxillary region. The skull shape can vary from brachycephaly (most commonly observed) to scaphocephaly (boat-shaped head), oxycephaly, plagiocephaly, trigonocephaly (triangle-shaped head) or in severe disease cloverleaf skull (kleeblattschädel) like deformity complex⁵. Other clinical features include hypertelorism, exophthalmos, strabismus, beaked nose, short upper lip, maxillary hypoplasia, and relative mandibular prognathism with no digital abnormalities⁶. The facial and oral malformations consist of hypoplastic maxilla and zygoma, pointed nose (psittichorhina/ parrotbeak-like nose) due to the short and narrow maxilla, narrow high-arched palate.

Thorough clinical and radiological analyses are required for early recognition and diagnosis of CS.

X-Ray skull

Reveal obliterated sutures, hypoplastic maxilla with shallow orbits, shortened cranial fossa, enlarged hypophyseal cavity, and small paranasal sinuses. Prominent cranial markings of the inner surface of cranial vault may be seen as multiple radiolucencies appearing as depressions resulting in hammered silver/metal/copper beaten appearance (due to an increase in intracranial pressure, as a result of premature cranial suture fusions)⁵. Spine and hand were unremarkable.

Differential Diagnosis

Is made with other syndromes associated with features of craniosynostosis such as Pfeiffer's syndrome, Apert syndrome, Saethre-Chotzen syndrome, Carpenter syndrome, and Jackson-Weiss syndrome⁵. All these involve craniofacial abnormalities, as well as other abnormalities including the hands or feet.

Complications of Crouzon's syndrome may include conjunctivitis or keratitis, luxation of the eye globes, exotropia, poor vision due to optic atrophy and corneal injury, blindness. Frequent headaches, seizures, mental deficiency, increasing hydrocephaly, conductive hearing deficit, upper airway obstruction develop secondary to septal

deviation, midnasal abnormalities, choanal abnormalities and nasopharyngeal narrowing. Others include nystagmus, iris coloboma, aniridia, anisocoria, corectopia, microcornea, megalocornea, keratoconus, cataract, ectopia lentis, blue sclera and glaucoma⁶.

Prognosis : depends on the severity of malformations and the timing of intervention. Innovations in craniofacial surgery have enabled patients to achieve their full potential by maximizing their opportunities for intellectual growth, physical competence, and social interaction.

CONCLUSION

Crouzon syndrome should be diagnosed managed as early as possible as it results in impaired facial appearance and other complications like, airway obstruction, and decreased visual acuity as the patient gets older. With proper treatment by multimodality, these patients can be productive and active members of the main stream of society.

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LITERATURE REVIEW

OUTCOMES OF KIDNEY TRANSPLANTATION RECIPIENTS WITH HEPATITIS IN THE ANTIVIRAL THERAPY ERA: A SINGLE-CENTER EXPERIENCE.

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The use of organs from hepatitis B surface antigen (HBsAg)+ donors could increase the donor pool substantially. However, fulminant hepatic failure requiring urgent liver transplantation or resulting in death has been reported in recipients of HBsAg+ renal transplantation (KT) in pre-nucleos(t)ide analog (NA) era. With effective antiviral therapies such as NAs, it seems feasible to transplant such recipients more safely. To address this issue, we conducted a retrospective, cohort study to evaluate the safety and long-term risks of HBsAg+ KT recipients in the NA era. From December 2006 to January 2013, 112 patients undergoing KT were followed at our institute. We analyzed patient and graft outcomes, hepatitis status (HBsAg status, hepatitis B virus [HBV] DNA level, liver function tests, presence of hepatitis C virus [HCV] co-infection), and graft source (domestic or transplant tourism). Ninety-two KT recipients were still alive. Nine patients were died of nonhepatic factors. Among 112 patients, there were 19 of 92 recipients alive who were HBsAg+, including 6 patients with HBV and HCV dual infections. Two of 19 patients experienced symptomatic hepatitis, one de novo and the other re-activation. Liver functions of these 2 recipients recovered progressively after introduction of NAs. No recipients in our study had experienced graft loss at the time of analysis. In terms of patient survival and quality of life, KT seems be a safe and feasible therapy of choice for HBsAg+ patients with end-stage renal disease. Infection is easier to prevent than to treat. KT recipients at high risk for HBV reactivation and for complications of HBV, with or without HCV co-infection, may benefit from longer prophylaxis. However, the optimal duration of prophylaxis remains unclear. Furthermore, several issues needed to be solved for clinical concerns, such as frequency and intensity of adverse effects, high costs, increased pill burden, drug-drug interactions, and the emergence of viral resistance variants.

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