RARE CYSTIC CHANGES IN SKULL ASSOCIATED WITH CROUZON SYNDROME

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ABSTRACT

Crouzon syndrome is a rare birth defect which in most instances is inherited as an autosomal dominant trait without complete penetrance. Though the typical features of Crouzon syndrome have been reported by various authors, to our knowledge, cystic changes in skull associated with Crouzon syndrome have never been reported in the literature. Herein, we report a rare case of Crouzon syndrome in a seven-month-old boy with remarkable multiple cystic changes in the skull.

Key words: Crouzon syndrome, cystic change, skull, craniosynostosis

INTRODUCTION

Crouzon syndrome is one of the most common craniofacial syndromes and is seen 1 in 60,000 persons. In most instances it is inherited as autosomal dominant with variable expression. Though the typical features of Crouzon syndrome have been reported by various authors, to our knowledge, cystic changes in skull associated with Crouzon syndrome have never been reported in the literature.

CASE REPORT

A seven-month-old baby born to non-consanguineous parents was admitted to our department with the chief complaint of malformation skull since his birth. He had outstanding oxycephaly which increased progressively with age. His father was normal but his mother and grand-mother showed a very slight degree of the same craniofacial
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abnormalities. On examination, He had asymmetrical enlargement of the skull, irregularly shaped vault with left side of the head enlarged vertically, involving the sagittal and coronal sutures. Other features like exophthalmos, a slight prognathous lower jaw and the maxillary hypoplasia were also seen (Figure 1). The limbs were normal. No mental or neurological defects were noticed.

Figure 1. The patient with outstanding oxycephaly, exophthalmos and a slight prognathous lower jaw.

The most remarkable Computer Tomography scan findings were multiple cystic changes and characteristic digital impressions in the parietal and occipital bones. Other findings included a mild degree of hydrocephalus, craniosynostosis with partial premature closure of sutures, shallow orbits, hypoplastic maxilla and zygoma, deviation of nasal septum (Figure 2 and Figure 3).
Chromosome analysis was normal in G-band karyotype. However, deoxyribonucleic acid studies showed a common mutation point of Cys278Phe in exon IIIa of the fibroblast growth factor receptor 2 (FGFR2) gene. The mutation was also present in his mother and grand-mother.

All of the above features were consistent with Crouzon syndrome. Cranial reconstruction surgery was recommended by neurosurgeons, but his parents declined for various reasons.

Figure 2. Computer Tomography scan showing multiple cystic changes and digital impressions in the skull and hydrocephalus.
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DISCUSSION

Crouzon syndrome is a genetic disorder, characterized by premature fusion of the cranial sutures, resulting in craniosynostosis, which gives rise to typical symptoms, such as increase in intracranial pressure, reduced orbital volume, exophthalmos, maxillary hypoplasia and occlusal derangement\textsuperscript{1-10}. Since the original description by Crouzon, other features and complications have been added; these include glaucoma, extraocular muscle anomaly, hearing loss, dental

Figure 3. 3D Computer Tomography scan showing early partial fusion of the cranial sutures, hypoplastic maxilla and zygoma, characteristic digital impressions in the skull.
problems, pituitary dysfunction, acanthosis nigricans, chronic upper airway obstruction, increasing intracranial pressure giving rise to headaches, seizures, mental retardation, etc. However, cystic changes in skull associated with Crouzon syndrome as in our case have never been reported before. Though the exact cause of cystic changes in the present case could not be ascertained, high intracranial pressure, poor circulation, bone degeneration and abnormal calcium metabolism could be the probable aetiological factor. In some instances, Crouzon syndrome is inherited as an autosomal dominant trait, and most patients have been shown to carry mutations of the FGFR2 gene. In other cases, affected people have no family history of the disease, in which Crouzon syndrome may result from new genetic mutations that occur randomly for unknown reasons.

The diagnosis of Crouzon syndrome is based on clinical findings and radiological examination. Crouzon syndrome must be differentiated from simple craniosynostosis and other syndromic craniosynostosis, such as Apert, Pfeiffer, Jackson-Weiss, Carpenter and Saethre-Chotzen syndromes. Crouzon syndrome is distinguishable from other craniosynostosis syndromes by absence of limb abnormalities.

The management of Crouzon syndrome requires a multidisciplinary team approach. Treatment includes measures to minimize intracranial pressure, secondary craniofacial abnormalities and other complications.

In conclusion, we report a rare case of Crouzon syndrome in a seven-month-old baby with remarkable multiple cystic changes in the skull. To our knowledge, this is the first case of this disease in which cystic changes in skull have been observed.

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CONSENT

The patient has consented to the publication of this case.

COMPETING INTERESTS

The authors declare that they have no competing interests.

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