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Case Report

Crouzon Syndrome: A Case Report

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ABSTRACT

Introduction: Crouzon syndrome is a rare genetic disorder with autosomal dominant inheritance. The underlying pathological process is premature synostosis of the coronal, sagittal and occasionally lambdoid sutures beginning in the first year of life and completed by 2-3 years of life. Mutation of the gene for fibroblast growth factor receptor 2 (FGFR2) is responsible for Crouzon syndrome.

Case Report: An 8 year old boy was brought to our hospital with complaints of proptosis & a solitary swelling over forehead with a history of some surgical procedure done at another hospital. Patient had extensive proptosis, hypoplastic maxilla and a realatively mandibular prognathism, crowding of teeth, hypertelorism & proptosis. Blood investigations were within normal limits. Xray skull was suggestive of chronic raised ICT, a bony defect in the frontal bone in the midline with herniation of brain parenchyma with CSF. Depending on history, clinical findings & investigations a diagnosis of Crouzon syndrome was made.

Discussion: The main clinical feature of Crouzon syndrome is brachycephaly, hydrocephalus and mental retardation, usually because of premature fusion of cranial sutures. There is maxillary hypoplasia with relative mandibular prognathism and dental malocclusion, parrot beaked nose, bilateral proptosis, spontaneous luxation of the globes, strabismus and hypertelorism. In radiological examination, `Paw marking' of the skull is seen due to raised intracranial pressure. Cephalometric studies measure the dimensions of some functional spaces like orbits, rhinopharynx and nasal cavities. Management of a patient includes the release of prematurely fused sutures which is mainly carried out early after 3-6 months and a Craniofacial reconstructive surgery later.

Keywords: Craniofacial synostosis, FGFR2 gene mutation, raised intracranial pressure

INTRODUCTION

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. In 25% of cases, it may also occur sporadically because of a fresh mutation. The underlying pathological process is premature synostosis of the coronal, sagittal and occasionally lambdoid sutures beginning in the first year of life and completed by 2-3 years of life. This fusion does not allow the bones to grow normally, affecting the shape of the head, appearance of the face and the relationship of the teeth. The diagnosis is based on clinical findings

and radiological examination. This syndrome was described by Crouzon in 1912 who described a patient with a characteristic group of deformities which were then observed in other individuals. Crouzon syndrome is caused by malformations of the mesenchyme and ectoderm. Mutation of the gene for fibroblast growth factor receptor 2 (FGFR2) is responsible for Crouzon syndrome and this gene has been mapped to the long arm of chromosome 10 and mutations in exon B of FGFR2 gene have been described. [1]

CASE REPORT

An 8 year old boy was brought to our hospital with complaints of proptosis since 6-7 years & a solitary swelling over forehead since 3 years. He had no complaints of vision, mentation or any auditory disability. He had a history of some surgical procedure done at another hospital 3 years back following which the forehead swelling appeared. The swelling was solitary, initially small, progressively increased to 4X4 cms in size, non-reducible, firm and nontender.

Patient had extensive proptosis, craniosynostosis, beaked nose, short upper lip, hypoplastic maxilla and a relatively mandibular prognathism. There was crowding of teeth, depressed nasal bridge, broad nasal bone, low set ears, high arched palate and a long philtrum. The ophthalmic features were shallow orbits, hypertelorism & proptosis. Scars of previous surgery were seen on scalp.

Blood investigations were within normal limits. X ray skull showed craniofacial synostosis with moth beaten appearance in the cranial bones suggestive of chronic raised ICT, mandibular prognathism, crowding of teeth. MRI was showing dilated ventricles, brachycephaly & thinning of corpus callosum, suggestive of raised intracranial pressure. There was a bony defect in the frontal bone in the midline which might be due to previous surgery. Herniation of brain parenchyma with CSF was noted through the bony defect in the swelling suggestive of encephalocoele.

The patient had a positive family history with the father suffering a similar ailment. Depending on history, clinical findings & investigations a diagnosis of Crouzon syndrome was made.

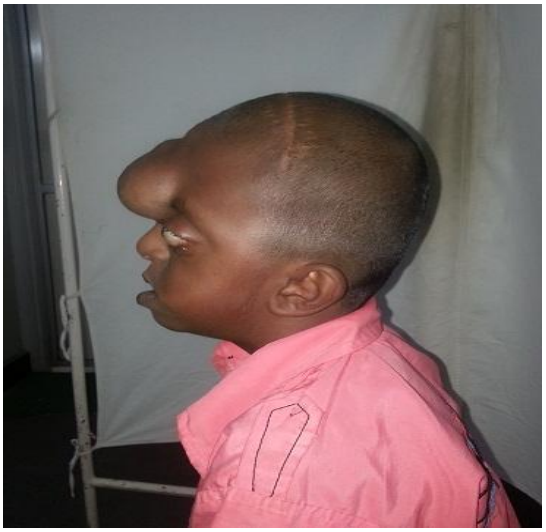

Fig 1: Patient showing proptosis, maxillary hypoplasia with relative mandibular prognathism, low set ears and a solitary frontal swelling.

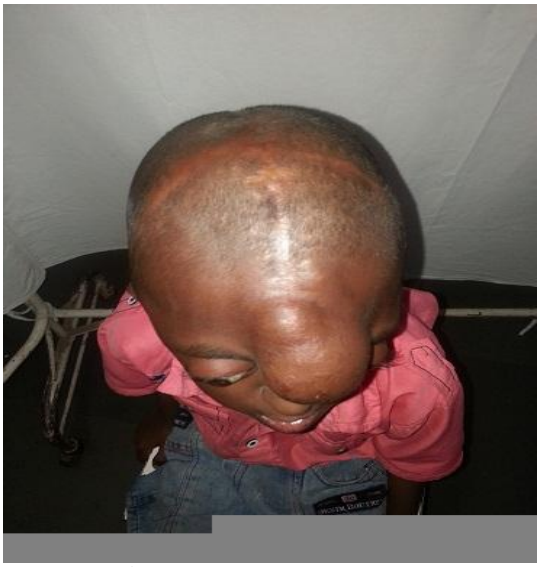

Fig 2: Scars of some previous surgical procedure done at another hospital.

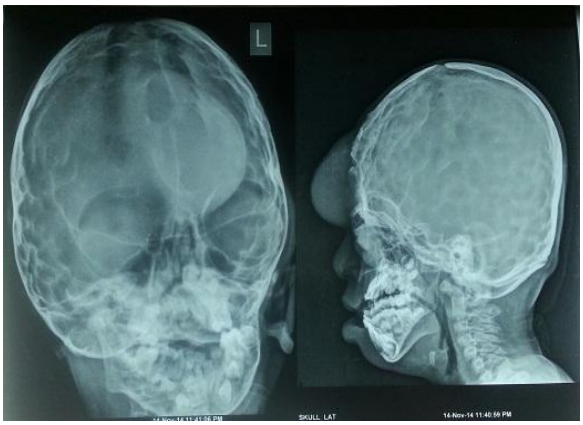

Fig 3: X ray skull showing paw marking of the vault, bony loss of the frontal bone, maxillary hypoplasia and the crowding of the teeth.

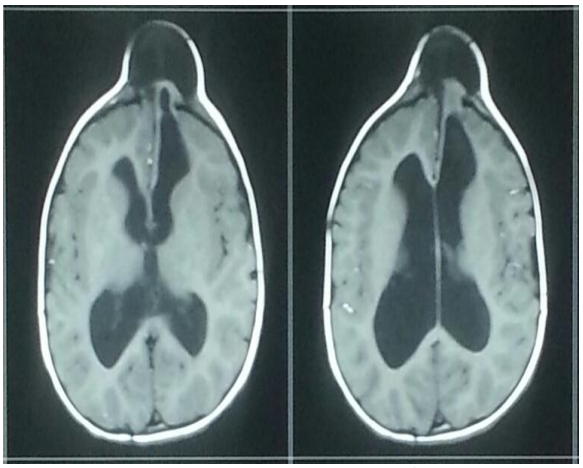

Fig 4: T1 MRI image showing protrusion of the CSF and the brain parenchyma through the bony defect in the frontal bone s/o encephalocoele.

Fig 5: The father of the patient showing similar facial features of proptosis, maxillary hypoplasia with a relative mandibular prognathism.

DISCUSSION

The main clinical feature of Crouzon syndrome is a skull deformity which may be brachycephaly, oxycephaly or trigonocephaly. Hydrocephalus and mental retardation is usually associated because of premature fusion of cranial sutures. [2] There is maxillary hypoplasia with relative mandibular prognathism and dental malocclusion. There can be deviation of the nasal septum, narrowed or obliterated anterior nares, wide parrot beaked nose and rhinolalia. Midfacial anomalies like cleft lip and cleft palate might be present. Hearing loss may be there which is usually conductive. Orbital defects include bilateral proptosis because of shallowing of orbits secondary to arrested growth of the maxilla and zygoma, and anterior positioning of the greater wing of the sphenoid. There might be spontaneous luxation of the globes also. There is strabismus (esotropia or exotropia) and hypertelorism. There might be vision threatening complications like exposure keratopathy and optic atrophy due to compression at the optic foramen. Acanthosis nigricans can also be associated with Crouzon syndrome. Patients of Crouzon syndrome are usually dark complexioned. In radiological examination, anteroposterior, lateral and cephalometric views of the skull are taken. It shows premature suture closure and provides information about maxillomandibular relation. `Paw marking' of the skull is seen due to raised intracranial pressure. [3] CT scan confirms the standard radiographical findings and provides information on ventricular size. Suture closure can be graphically displayed by 3-dimensional CT scans. Cephalometric studies measure the dimensions of some functional spaces like orbits, rhinopharynx and nasal cavities. [4] Management of a patient of Crouzon syndrome has two aspects. First is the release of prematurely fused sutures based

on evidence of raised intracranial pressure. Surgery is mainly carried out early after 3-6 months. [5] The principle is the release of bony ankylosis by exposure of fused sutures via a coronal flap. Second, the Craniofacial reconstructive surgery including advancement of the maxilla and frontonasal complex; and other surgeries depending upon the deformities in the patient like rhinoplasty, oculoplasty and cleft lip and cleft palate repair can be done. Early and accurate diagnosis of a patient of Crouzon syndrome is essential. Genetic counselling plays an important role. The need, extent and timing of treatment depend upon the severity of the disease and age of the patient. For complete evaluation, optimum treatment planning and comprehensive services, a multidisciplinary approach to the management of a patient of Crouzon syndrome is needed.

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