The Crouzon Syndrome-A Case Report

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ABSTRACT
The Crouzon syndrome is a genetic disorder which is known as the brachial arch syndrome. It is an autosomal dominant disorder which is one of a rare group of syndromes which is characterized by cranio synostosis or a premature closing of the cranial sutures. The major features are brachiocephaly, ocular proptosis, an underdeveloped maxilla, mid face hypoplasia, a rare cleft lip or palate, hypodontia (some teeth missing) and crowding of teeth. Due to the maxillary hypoplasia, the Crouzon syndrome patients generally have a considerable permanent underbite and they subsequently cannot chew by using their incisors. We have presented in this article, a case of the Crouzon syndrome which was seen in a girl who was aged six years, with similar symptoms and the multidisciplinary approach which has to be followed in managing the case.

INTRODUCTION
The Crouzon syndrome is named after the French neurologist, Octave Crouzon, who described this disorder [1-3] which includes a triad of skull deformities, facial anomalies, and an exophthalmus [4, 5]. Many bones which form the skull are separated by sutures which allow the skull to expand and develop in synchrony with the growth of the brain. Premature synostosis commonly involves the sagittal and the coronal sutures. Occasionally, the lambdoid sutures are involved. The order or rate of the suture fusion determines the degrees of deformity and disability [1, 2]. A premature sutureal fusion may occur alone or together with other anomalies, thus causing various syndromes [6]. The craniofacial abnormalities are often present at birth and they may progress with time. A decreased mental function is present in approximately 12% of the patients. The family history may reveal individuals with a mild Crouzon syndrome. The headaches and a failing vision are attributable to an elevated intracranial pressure.

In this article, we are presenting a case of the Crouzon syndrome which is one of the rare syndromes and is associated with synostosis of the cranial sutures, which is seen in 1 in 60,000 persons. The differential diagnosis of the Crouzon syndrome includes simple synostosis as well as the Apert, Pfeiffer and the Saethre-chotzen syndromes.

CASE REPORT
A six years old female child who was born by a full term normal delivery, with a normal appearance, developed an impaired vision with an excessive prominence of the eyeballs, an abnormal shape of the skull which was characterized by a premature fusion of the coronal sagittal and the bilateral parieto-occipital sutures, a maxillary complex (craniosynostosis), an exophthalmos, Table/Fig-1 a beaked nose [Table/Fig-1], a short upper lip [Table/Fig-1], a hypoplastic maxilla, and a relative mandibular prognathism [Table/Fig-2], crowding of the teeth, frontal bossing, a depressed nasal bridge, a broad nasal bone, low set ears, [Table/Fig-2] a high arched palate, a long philtrum and webbing of the neck [Table/Fig-3]. The ophthalmic features were, an extremely shallow orbit, an apparent proptosis, an inferior sclera which was caused by the maxillary hypoplasia, upper lid ptosis, nystagmus central (rotatory) exotropia and left eye myopia. The intercanthal distance was 28mm. There was a history of a spontaneous prolapse of the right eye ball, 2 to 3 years ago, due to rubbing of the eye. There was no recurrence. No digital abnormalities were present.

Table/Fig-1: Picture showing exophthalmus

Table/Fig-2: Picture showing mandibular prognathism, frontal bossing, low set ears
DISCUSSION

The phenotypic features of the Crouzon syndrome may be absent at birth and they may evolve gradually during the first few years of life [7, 8] as seen in the figures [Table/Fig-1-4]. It is commonly inherited as an autosomal dominant trait, with complete penetrance and a variable expressivity, but about one third of the cases do arise spontaneously. The male to female preponderance is 3:1 [2, 9]. With the advent of molecular technologies, the gene for the Crouzon syndrome could be localized to the Fibroblast Growth Factor Receptor 2nd gene (FGFR2), at the chromosomal locus 10q25.3-q26, and more than 30 different mutations within the gene have been documented in separate families [10]. The cranial synostosis initiates changes in the brain and the adjoining structures such as an increased intracranial pressure, a reduced orbital volume, occlusal derangements and exophthalmoses [11]. A prenatal diagnosis of the exophthalmus has been reported by5 ultra sonography. So, an early recognition is essential, to guide the growth and development of the face and the cranium [7]. In 30% of the cases, the hydrocephalus is in progress, as per the literature. There was a mild dilatation of the ventricles in our patient too, which was suggestive of a hydrocephalus, which is one of the rare signs. A multidisciplinary approach was required to manage this case with treatment measures, to minimize the intracranial pressure and the secondary calvarias deformities. The innovations in craniofacial surgery enabled the patient to achieve their full potential, by maximizing their opportunities for an intellectual growth, a physical competence and a social acceptance. So, the prognosis depends on the severity of the malformations. The patients usually lead normal lives.

The Crouzon syndrome requires major surgery and there can be major complications such as death or blindness, but in the hands of an experienced craniofacial team, this is rare. Other complications such as a collection of blood, an infection, bony irregularities, an electrolyte imbalance and a haemodynamical instability can occur. It should be remembered that these conditions are problems in the growth and that subsequent “re-corrective surgeries” may be required.

REFERENCES

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