

Crouzon Syndrome: A case reportMalleswari D¹, Rama Kishore AV²

1- Associate Professor, 2- Assistant Professor, Department of Pediatrics, Government Medical college, Anantapuramu. A.P

Corresponding Address: Dr. D. Malleswari, Associate Professor and in charge Head, Department of Pediatrics, Government Medical college, Anantapuramu. 515 001, Andhra Pradesh.**Abstract:**

Crouzon's syndrome is an autosomal dominant genetic disorder with complete penetrance and variable expressivity. Crouzon's syndrome is a rare genetic disorder which is a resultant of mutation in the fibroblast growth factor receptor 2 (FGFR2) gene. The syndrome manifests with earlier closer of skull sutures and tamper the normal growth of the brain. The syndrome usually results in premature synostosis of coronal and sagittal sutures which begins in the first year of life and the subjects are likely to suffer with mental retardation. The current case is about a 5 year old case of Crouzon's syndrome without mental retardation.

Key words: Crouzon's Syndrome, Craniosynostosis, Craniofacial dysostosis, turricephaly

CASE REPORT:

A 5 year old girl child presented to the outpatient department of Department of pediatrics, Government General Hospital, Anantapuramu with a history of fever and watery diarrhea of 5 days duration. The child also presented with history of gradually progressing distension of abdomen since birth and recurrent abdominal pain. Occasionally dull abdominal ache not associated with vomiting or micturational disturbances. No other significant symptoms were presented. Clinical examination revealed signs like conical shaped head (turricephaly), prominent eye balls, concave upper half of face, high forehead, wide beaked nose with narrow anterior nares, long philtrum, microstomia and micrognathia. Examination of oral cavity revealed crowding of teeth with conical shaped incisors and canines in lower jaw and high arched palate. Other features included midfacial hypoplasia, low set ears and acanthosis nigricans around the mouth (figure-1).

Vital signs were within normal limits. Anthropometric measurements revealed microcephaly, Craniosynostosis, stunted growth and failure to thrive. Cardiovascular, respiratory system findings were within normal limits. Abdominal examination revealed normal status except for abdominal distension and divarication of rectus

**Figure 1: Crouzon's Syndrome-features**

abdominis muscle (Fig 2 & 3). Ophthalmic examination showed temporal pallor, dull foveal reflex and visual acuity was 6/12 in both eyes.

CT scan demonstrated obliteration of all skull sutures except sagittal suture. Blood investigations indicated microcytic normochromic anemia along with mild degree of hypo chromic anisopoikilocytosis with and presence of tear drop cells, elliptocytes and polychromasia. White blood cell count was 12,200 cells/cu.mm, Differential count was within normal limits and platelets were adequate. X-rays of long bones, wrist and chest were

normal. However, plain X-ray of skull revealed silver beaten appearance (Figure.4).



Figure 2: Abdominal distension



Figure 3: Divarication of rectus muscle



Figure 4: Plain X-ray of skull (A.P and Lat view): Silver beaten appearance.

Discussion

Crouzon's syndrome is a genetic disorder and is also known as brachial arch syndrome. This syndrome affects the first brachial arch in the embryonic phase which is precursor of the maxilla and mandible. This syndrome is named after French

neurologist Octave Crouzon, who first described this disorder in 1912 in mother and son implying its genetic basis. In the initial stages this syndrome was referred as "craniofacial dysostosis" as this disorder is characterized by a triad of features like Craniosynostosis, midface hypoplasia and proptosis [1,2]. Premature closure of cranial sutures results in abnormal skull growth, affecting the growth and development of the orbits and maxillary complex.

The mental capacity of subjects of Crouzon's syndrome will usually be within the normal range or there may be diminished mental function as apparent in 12 % of patients. The subjects with this syndrome may also manifest with visual disturbances and hearing loss owing to recurrent ear infections, ear canal stenosis/atresia. The prevalence of this syndrome in the US is 1 per 60,000 live births and as such Crouzon's syndrome accounts for approximately 4.8 % of all cases of Craniosynostosis [2].

The molecular analysis of Craniosynostosis syndromes identifies mutations in the fibroblast growth factor receptor-2 gene which is mapped to chromosome locus 10q25q26. In more than 50% of cases with Crouzon's syndrome mutations are identified in the fibroblast growth factor receptor-2 gene, several mutations are in the Ig three domain of fibroblast growth factor receptor 2 and a novel mutation Tyr 281 cys substitution at the exon three a of fibroblast growth factor receptor 2 were observed. Based on the specific mutations of fibroblast growth factor receptor-2, other mutations of this and other still unidentified genes, it can be concluded that the Crouzon's syndrome phenotype shows heterogeneity while molecular analysis of the fibroblast growth factor receptor 2 gene provide useful information and help with confirming the diagnosis and performing prenatal diagnosis [1,3,4]. The Crouzon's syndrome has to be differentiated from Apert syndrome, Pfeiffer syndrome, Carpenter syndrome, Seatre-Chatzen syndrome and Jackson Weiss syndrome which also present with similar features [5,6].

Treatment involves a multidisciplinary approach and aims at staging reconstruction to coincide with facial growth patterns, visceral function and psychosocial development of the patients. Satisfactory results can be reached by plastic surgery as Crouzon's syndrome is one of the few syndromes where in the cosmetic results of the surgery can be strikingly effective. Surgery is typically aimed at prevention of closure of the sutures of the skull from hindering the brain's development [7]. These patients tend to have

involvement of multiple sutures; most specifically bilateral coronal sutures and hence either open vault surgery or strip craniectomy (if child is under 6 months of age) can be performed so as to achieve targeted result. In the latter stages, the patient is advised to wear a helmet for several months following surgery. Once treated for the cranial vault symptoms these patients generally go on to live a normal span of life [8].

Conclusion:

Early detection, patient relative education and timely intervention form the important steps in the management of this syndrome. Initiation of early multidisciplinary approach ensures for better survival of the child with Crouzon's syndrome and also helps the subjects to fight out of the social stigma as the life span of the subjects with Crouzon's syndrome is almost normal.

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