A Rare Case of Crouzen Syndrome-Diagnosis and Detailed Evaluation

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Abstract: Crouzen syndrome (Craniofacial dysostosis) is rare genetic disorder of autosomal dominance, caused due to mutation in fibroblast growth factor receptor-2 gene. It characterized with premature closure of cranial sutures-craniosynostosis, mid facial hypoplasia and orbital features-Hypertelorism, Strabismus, Exophthalmos, optic Atrophy. A 56 yr/M present with diminution of vision Bulging of eyes and deviation of eyeball and facial deformity. Examination of eyes revealed RE exotropia on hirschberg test, exophthalmos after exophthalmometer finding. Fundus evaluation with indirect ophthalmoscope B/L optic atrophy revealed. Therefore it becomes important to evaluate such patients thoroughly. MRI findings of craniosynostosis and B/L optic atrophy confirms diagnosis. It is also helpful to diagnose such cases earlier to avoid severe visual impairment and refer them to concerned specialist for further evaluation and timely intervention and psychosocial support to entire family is important.

Keywords: Crouzen syndrome, Autosomal dominance, craniosynostosis, Exophthalmos, Optic atrophy, mid facial hypoplasia

1. Introduction

Crouzen syndrome also known as Craniofacial Dysostosis / Craniofacial Dysostosis type I /Craniosenosis-Crouzen type. It is an Autosomal dominant disorder. Incidence 1: 25, 000 live birth. (¹) It is most common among all craniosyntosis disorders. (¹) Both genders affected equally. Etiology includes Sporadic, Spontaneous genetic mutation in egg or sperm cell and has Associated with FGFR-2 gene in both sporadic and inherited cases. (²)

In 44.67% cases positive family history present. The diagnosis is based on clinical findings and radiological examination.

2. Case Report

A 56 year male present with Bulging of eyes and deviation of eyeball and facial deformity since birth & diminution of vision BE since 20 years.

Which was gradually progressive painless in nature. No history of similar complaints in family. Patient was slow learner at school.

On Ocular Examination-
Vision-RE FC 1 Mtr
LE FC 3 mtr
Near vision-N-36
Colour vision-Defective.
B/L Exophthalmos (after exophthalmometer finding)
- Right eye exotropia on hirschberg test.
- B/L pupil- normal size ill-sustained
- Rest anterior segment normal.

Photograph of case

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Investigations:
1) **On Audiogram**-B/L mild to moderate sensory neural hearing loss.
2) **On MRI Brain + Orbit:**

Brachycephaly with bilateral optic nerve atrophy along with compression of pituitary fossa sideside to side with mickels cave.

- **Facial**-Parrot like nose, maxillary hypoplasia, mandibular prognathism
- **Ear**-Low set ear, B/L atresia of auditory canal, Sensory neural hearing loss.
- **Eye**-Shallow orbits, exophthalmos, hypertelorism, Strabismus (Divergent), ptosis, downsllanting Palpbral fissure, optic atrophy.
- **Oral**-High arched & narrow palate, cleft palate, class III malocclusion with maxillary crowding.
- **Neurological**-Headache, mild-mod. Mental retardation, seizures.
- **Musculoskeletal**-Scoliosis.
- **Respiratory**-Breathing difficulty, Sleep apnea.
- **Cardiac** (rarely)-Patent ductus arteriosus, Coarctation of aorta, VSD. Co-existence with VSD is not accessible in literature. (4)

Exophthalmos is universal feature of crouzen syndrome.

Class III malocclusion with maxillary crowding seen in 75% patients occurred due to retrusive and short maxilla. (5)

Five autosomal dominant craniosynostosis syndromes

(Apert, Crouzon, Pfeiffer, Jackson-Weiss and Crouzon syndrome with acanthosis nigricans) result from mutations in FGFR genes. (6)

4. Conclusion

The clinical diagnosis of this case is crouzon syndrome which is confirmed by MRI brain + orbit findings.

For definitive diagnosis Moecular genetic testing needed.

Lubricating eye drop given to avoid exposure Keratitis.

It is also helpful to diagnose such cases earlier to avoid severe visual impairment and refer them to concerned specialist for further evaluation and timely intervention and psychosocial support to entire family and to patient is important.

Multidisciplinary treatment approach is needed if systemic involvement is there. and long term follow up is required.

The case has been presented to increase the awareness of this entity and its systemic association in general public.

References


