Syndromic Orbital Malformations

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Congenital craniofacial abnormalities may present as isolated disorders of orbit or in association with multisystem syndromes. Syndromic malformations may be as Craniofacial clefts (CFCs) & clefting syndrome or Craniosynostosis and craniostenosis. Most complex and disturbing malformation is oblique facial clefts accounting for 0.22% of all cases.[1] As orbit is the central point of all oblique clefts, it induces anophthalmia, microphthalmia and canthal deformities. These congenital soft tissues and/or skeletal gaps on the face and/or cranium are mainly due to failure of fusion of facial processes or mesodermal migration. Saadi et al demonstrated the molecular and genetic basis of the CFCs. He reported that mutations in critical SPECC1L gene that controls facial morphogenesis is responsible for CFCs.[2] The CFCs are usually associated with true hypertelorism. In severe cases binocular vision is impaired and patient may develop strabismus and amblyopia in the nonfixating eye. The management of clefting syndrome is essentially surgical. Multistage surgery includes correction of hypertelorism by orbital box osteotomy or midface advancement.

Craniosynostoses/cranistenosis is characterized by premature fusion of one or more cranial sutures resulting into deformation of the cranial vault. Craniosynostosis may be nonsyndromic or syndromic. The most frequent craniofacial malformation with orbital involvement are the craniofacial dysostoses syndrome including Crouzen's syndrome, Klippel-Feil anomaly and Kleeblattschadel's anomaly[3] First syndromic craniosynostosis was described by Eugene Apert in 1896 in Paris, characterized by abnormal cranial shape and syndactly of both hands and feet. In 1912 Octave Crouzon described new hereditary syndrome characterized by proptosis, divergent strabismus, maxillary retrusion and abnormal cranial shape.

Apert's and Crouzon's syndromes are associated with mutations on the gene FGFR2 (fibroblast growth factor). Pfeiffer syndrome is associated with mutation on FGFR1 gene. Orbit is a transitional zone between face and skull which gets deformed in craniosynostosis. Ocular morbidity is quite high in all the three major syndromes. 35-40% of the patients have vision loss at least in one eye.[4] Depending on severity and number of sutural closure, orbits are extremely shallow leading to severe degree of proptosis and exposure keratopathy with spontaneous globe subluxation. Such malformations can be corrected by anterior cranial base and orbital surgeries including craniotomies and facial osteotomies.

References

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