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Oculo-orbital developmental disorders

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Developmental disorders of orbit or its contents may occur occasionally as an isolated anomaly or more often gates associated with other nervous system malformations or may occur as part of multisystem syndrome. It includes craniofacial cleft, craniosynostoses and defective development of eyeball. Such developmental disorders are often associated with irreversible congenital blindness.(1) Oculo-orbital developmental disorders represent a challenging management scenario requiring complex, multiple surgeries at different intervals and sometimes suboptimal results may be obtained. Although craniofacial congenital defects are rare but their psychological impact on patients and their families is enormous and lifelong.

Development of eye occurs in early embryonic life by means of anterolateral outgrowth of neural tube. It develops from optic vesicles on either side of developing forebrain and surface ectoderm. The embryonic development of orbit is a complex process, entire contents of orbit and bony orbit formed by Cranial neural crest cells (CNCCs), considered by some authors as fourth germ layer (Ectomesenchyma).The degeneration or failure of development of the entire neural tube leads to Primary Anophthalmia and degeneration of optic vesicles after initial development results in Secondary Anophthalmia. In UK study revealed incidence of Anophthalmos of 0.6 per 100,000 births and microphthalmos 2.5.(2) Several genetic mutations have been linked to eyeball malformation. The first gene to be identified was PAX6 gene, although SOX2 is the major causative gene, other reported genetic associations include RAX and CHX10 gene.(3-5)

Anophthalmia and microphthalmia may lead to hypoplasia of bony orbit midface and periocular region. Early recognition and intervention is recommended because 90% of orbital growth occurs within 5 years thus main objective is to enlarge bony orbit. Orbital volume expansion may be achieved by using serial enlarging confirmers, an acrylic or polymethyl methacrylate (PMMA) implant, dermis fat graft (DFG), hydrogel tissue expanders etc.

Craniosynostoses and craniostenoses are characterized by premature fusion of one or more cranial sutures resulting into alteration in the shape and size of cranium /orbit. The most frequent craniofacial malformation with orbital involvement are the craniofacial dysostoses syndromes including Crouzen's syndrome, Klippel-Feil anomaly and Kleeblattschadel's anomaly.(6) Such malformations can be corrected by anterior cranial base and orbital surgeries including craniotomies and facial osteomies.

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