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Case Series

Spectrum of ocular manifestations in apert syndrome

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ABSTRACT

Apert syndrome is a congenital type 1 acrocephalosyndactyly characterized by craniosynostosis, dysmorphic facial features and symmetrical syndactyly. There is premature fusion of cranial sutures which leads to restriction of intracranial and orbital space expansion giving characteristic dysmorphic facial appearance. We are reporting two cases of Apert syndrome of different age groups from ophthalmic point of view, and with different sets of ocular manifestation. Of the two cases, the one who presented early had a better visual prognosis. Apert Syndrome has a social stigma and the patients often suffers social and psychological disturbances. Patient counselling, timely management by multidisciplinary approach and regular follow ups are an important aspect which can offer a better quality of life.

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1. Introduction

Apert syndrome was first mentioned by Baumgartner in 1842.[1] Eugene Apert in 1906 described the systemic presentation of the syndrome.[1] According to Cohen, the incidence of Apert syndrome is 15 out of 10 lakhs live birth.[1,2] It is an autosomal dominant rare genetic disorder characterized by craniosynostosis, dysmorphic facial features, severe symmetric syndactyly of the hands and feet, a variety of abnormalities of the skin, skeleton, brain, and visceral organs along with various ocular manifestations.[2] The condition results from a specific missense mutation in the gene encoding fibroblast growth factor receptor-2 (FGFR-2), on 10q26 chromosome.[1–3] The said anomaly results in premature fusion of cranial sutures, most commonly of the coronal suture and agenesis of sagittal and metopic sutures giving characteristics of turri-brachycephalic head shape. Antero-posterior shortening of the cranial base due to spheno-occipital and spheno-ethmoidal synchondrosis and early fusion of fronto-ethmoidal suture leads to acrocephaly or brachycephaly.[1,2] Virchow in 1851 reported the cessation of growth in a direction perpendicular to that of the affected suture while growth proceeds in a parallel direction.[4]

As a result of these early fusion of sutures, there is restriction of intracranial and orbital space expansion. The eyeball protrudes in relation to the cranial base and to the orbit resulting from marked protrusion of the lateral orbital wall. The dysmorphic growth and fusion of bony orbit results in hypertelorism, proptosis and antimongoloid slant.[2] Restriction of orbital space expansion leads to increased intra-orbital pressure. Premature fusion of sutures with continued brain growth can lead to raised intracranial pressure. It can compress optic nerve leading to papilledema, which leads to optic atrophy if not addressed on time. The corneal exposure keratopathy results due to ocular proptosis and inadequate lid closure, thus compromising vision. It can even lead to melting of cornea and loss of eye. There is no direct cause of decreased visual acuity. Cause of visual impairment includes compressive optic atrophy, strabismus, refractive

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error, and exposure keratopathy.[3] Structural alterations of the extraocular muscles have been associated with some cases leading to strabismus. The mechanism of strabismus could be due to absent muscle, anomalous insertion of muscle, excyclotorsion of muscles and instability of muscle pulleys.[3] If not addressed timely, it may progress to amblyopia.

The dysmorphic bony growth also results in a depressed nasal bridge with deviated nasal septum, hypoplastic narrow maxilla, high arched palate and severe dental crowding. Syndactyly is present due to ossification of interphalangeal joints.[2] Skeletal abnormalities like limitation of elbow and glenohumeral joint mobility, cardiovascular, gastrointestinal, genitourinary, mental retardation, hearing and speech defects have also been recorded.[2]

2. Case Series

Here we present two cases of Apert Syndrome of different age with different ocular manifestations.

2.1. Case 1

A nine months male infant with no family history of similar disorder was referred from pediatric department for ocular examination. The child fixes and follows light with both eyes. On external examination proptosis, hypertelorism and down slanting palpebral fissures were present (Figure 1a). Rest anterior segment examination was normal. Ocular movements apparently appeared to be normal. Fundus under mydriasis was within physiological limit. Infant's systemic manifestation were acrocephaly, hypoplastic mid-facies, symmetrical syndactyly (Figure 1b), high arched palate (Figure 1c), and urogenital hypoplasia.

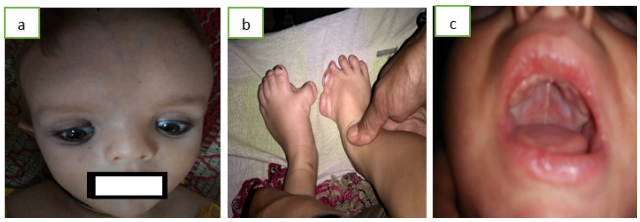

Fig. 1: a: Infant with hypertelorism proptosis, frontal bossing, depressed nasal bridge; **b:** Syndactyly of feet, **c:** High arched palate.

2.2. Case 2

A five years male child with no family history of similar disorder was referred from plastic surgery department for forward protrusion of both eyes, incomplete closure of lids since birth and whitening of the center of black part of left eye since a year. The child followed light with right eye but not with left eye. There was no perception of light in left eye. On external examination, classical features of apert such as bilateral proptosis, down slanting palpebral fissure and hypertelorism were noted. On anterior segment examination there was central leucomatous corneal opacity of about 3*3 mm with stromal thinning and exotropia in left eye (Figure 2a). Fundus examination revealed a pale disc with ill-defined margins and temporal pallor in both eyes. Infant had systemic manifestations like acrocephaly, hypoplastic mid-facies, dental crowding, syndactyly (Figure 2b) and high arched palate (Figure 2c).

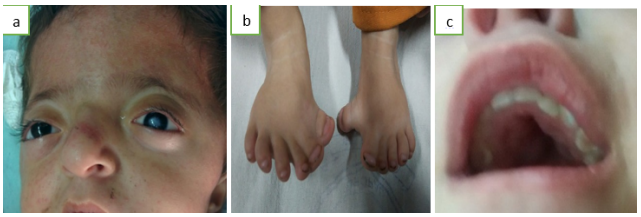

Fig. 2: a: left eye corneal opacity, **b:** Syndactyly of feet, **c:** High arched palate.

3. Discussion

Generally clinical signs and symptoms are sufficient to confirm the diagnosis of 'Apert Syndrome'. Characteristic radiographic finding of hand, foot and AP view of skull further confirms clinical diagnosis.

The patients of apert syndrome have normal life expectancy. These children are faced with psychological and social challenges due to physical appearance.[3] Family requires counseling to handle the situation. Definitive treatment is not available.[3] Corrective surgery for anatomical deformities improves cosmesis and can provide functional cure.[3] Timely management and regular follow-up by the ophthalmologist for children with apert syndrome are essential. Of the two cases who presented to us, it is clearly evident that delay in diagnosis and intervention in Apert syndrome can result in grave visual prognosis and if diagnosed and managed early in life can aid normal neurological development and prevent ocular complications including visual function. It highlights the importance of timely medical and surgical intervention by multiple medical specialities, hence offering a better quality of life.[2,3]

The treatment of apert syndrome begins at birth and a multidisciplinary approach, including neurosurgeons, neurologists, craniofacial surgeons, ophthalmologists, ENT surgeon, orthodontists and pediatrician is essential for a successful planning and treatment.[2,3,5] Cranial surgery to remove synostotic sutures and reshaping the calvaria allow normal cranial development to proceed with respect to shape, volume, and bone quality and relieves increased intracranial pressure. It is often performed during six months of life.[2,6] Shunting procedure also

reduces intracranial pressure. A recent advancement called endoscopic strip craniectomy which is a camera assisted operation is also being performed.[7]

Correction of syndactyly is done in first year of life. Cosmetic correction for midface deformities is done at 4-6 years of life.[2] Orthodontic and orthognathic surgery is performed after eruption of permanent dentition and completion of growth.[2,3]

A new technique of craniofacial disjunction followed by gradual bone distraction (Ilizarov's procedure), has been reported to produce complete correction of exophthalmos.[5]

The role of ophthalmologists in monitoring the visual development in children with apert syndrome is extremely important. Correction of ocular proptosis by normalization of midface and expansion of the inferior orbit. Surgical options can also be considered for complex ocular misalignments. Protection of the cornea by instilling lubricating agent, lateral or medial tarsorrhaphy are measures to prevent exposure keratopathy.[3] Thus, with the timely intervention we can save the vision of the child, emphasizing the importance of early surgical intervention for preserving visual function.

4. Conflict of Interest

None.

5. Source of Funding

None.

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