Ophthalmic Considerations in Cleft Lip and Palate Patients

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Abstract Cleft lip and palate (CL and P) represents the second most frequently occurring congenital deformity after clubfoot deformity. CL and P could be associated with many other structural abnormalities of the adjacent vital structures of the face. In this study, an attempt was made to identify the abnormalities of the ocular structures seen in patients with isolated CL and P as well as in those with syndromic CL and P. Of the 322 patients with cleft lip and palate screened, 27 (8.3%) had ocular abnormalities. Totally 47 ocular defects were identified in 27 patients. Abnormalities of the eyelid were the commonest accounting for 22% of the total defects (11/47), which includes symblepharon, ectropion, lid colobomas, euryblepharon and ptosis. Second commonest abnormality was squint 8/47 (17%) followed by orbital defects 8/47 (17%) (Telecanthus and Hypertelorism). Abnormalities of the nasolacrimal apparatus 3/47 (6%), refractive errors 7/47 (15%), dermoids 4/47 (6%), cataract 2/47 (4%) and retinal colobomas 2/47 (4%) constituted the rest. Thus, children with cleft lip and palate should be assessed as soon as possible after birth by a multidisciplinary team involving the

Pediatrician, Ophthalmologist and specialists from Maxillofacial, ENT and Plastic surgery. The medical problems in this group of children are global and therefore, should not be looked at in isolation.

Keywords Congenital deformities · Ocular abnormalities · Cleft lip and palate

Introduction

Cleft lip and palate (CL and P) represents the second most frequently occurring congenital deformity after clubfoot deformity. CL and P could be associated with many other structural abnormalities of the adjacent vital structures of the face like the ears, eyes, nose, teeth and brain. CL and P are inherently known to produce functional problems affecting the oropharynx (feeding and breathing), hearing, vision and speech; in addition, they have a negative cosmetic effect.

Almost all skeletal and soft tissue components of the craniofacial area are unbelievably derived from the neural crest cells [1]. Because eyes originate as an extension of the forebrain, malformations involving ocular structures invariably accompany those of the face and brain and vice versa. Disturbances in normal migration of eye fields from the lateral to frontal regions of the embryo's face during the fifth through eighth weeks have been suggested as a possible cause of facial clefting and ocular hypotelorism and hypertelorism [2].

As in any other organ or system in the body, a family of genes regulates the development of craniofacial structures [3]. Teratogens and environmental factors are known to adversely influence the development of the eye and craniofacial structures [4–7].

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Ophthalmologists have a unique role in the early identification of many ocular problems associated with this condition. In this hospital based study, an attempt was made to identify the ocular abnormalities seen in patients with CL and P, so that early treatment can be provided to these patients for the same.

Materials and Methods

Patients with cleft lip and palate attending the three departments of K.S. Hegde Charitable Hospital, namely, Maxillofacial, Ophthalmology and Pediatrics were studied. All patients with cleft lip and palate were screened for ocular abnormalities in eye clinic and those with abnormalities were further studied.

The patients were grouped into (i) 'Syndromic' if they had an identifiable syndrome and (ii) 'Non-Syndromic' if they had no features of known syndromes or other associated systemic disorders.

A careful history was taken, which included maternal administration of drugs, alcohol consumption, radiation and viral infection during pregnancy. Information on consanguineous marriage and family history of craniofacial disorders were also collected. A detailed physical examination of the eyelids, palpebral fissures, anterior segment, pupil and fundi were carried out in all patients. Ocular motility, refraction and ocular pressure were also recorded in all cases.

Results

Of the 322 patients with cleft lip and palate screened, 27 (8.3%) had ocular abnormalities. Of the 27 patients (Table 4), 13 (48.14%) patients had bilateral cleft lip and palate, 5 (18.5%) patients had unilateral cleft lip and palate, 2 (7.4%) patients had facial clefts, 2 (7.4%) patients had clefts with alveolus and remaining 5 (18.5%) patients had recognizable syndromes. The sex distribution was 1:1. 55% patients were below the age of 5 years, 29% were below the age of 10, 11% were below the age of 20 and 3% were above the age of 20 years (Table 1). Of the 27 patients

Table 1 Showing syndromic and non-syndromic cleft lip & palate

Non-syndromic	Syndromic	Total
Total 22 (82%)	Treacher Collins-2	27
	Goldenhar-1	
	Blepharochelidonotic-1	
	Ocular hypertelorism-1	
	Total 5 (18%)	

Table 2 Showing various ocular abnormalities found in 25 cases

Type of abnormality	Total number of defects (%)
Lid abnormality	11 (23.4%)
Squint	8 (17%)
Orbital	8 (17%)
Dacrocystitis	3 (6.9%)
Dermoid	3 (6.9%)
Microphthalmia	3 (6.9%)
Cataract	2 (4.2%)
Coloboma of iris & retina	2 (4.2%)
Refractive errors	7 (14%)
Total	47

studied 82% (22) were non-syndromic and 18% (5) had identifiable Syndromes (Table 2).

The various ocular abnormalities found in the study are listed in Table 3. Totally 47 ocular defects were identified in 27 patients. Abnormalities of the eyelid were the commonest accounting for 22% of the total defects (11/47), which includes symblepharon, ectropion, lid colobomas, euryblepharon and ptosis. Second commonest abnormality was squint 8/47 (17%) followed by orbital defects 8/47 (17%) (Telecanthus and Hypertelorism). Abnormalities of the nasolacrimal apparatus 3/47 (6%), refractive errors 7/47 (15%), dermoids 4/47 (6%), cataract 2/47 (4%) and retinal colobomas 2/47 (4%) constituted the rest.

Discussion

Lid Abnormalities

The different types of eyelid defects found were; epicanthus, coloboma, euryblepharon, ectropion (Fig. 1), Ptosis (Fig. 2) and symblepharon. Embryologically the fusion of maxillary and palatine processes and development of eyelid occur almost simultaneously (sixth to eighth week of the intrauterine life) and therefore a developmental defect of the palate could lead to an ocular defect [8].

The isolated eyelid coloboma (Fig. 3) could result from pressure from the amniotic band or by localized failure of fusion of the lid folds. Congenital Entropion is caused by vertical deficiency of the posterior lamella. Euryblepharon is characterized by primary symmetric enlargement of horizontal palpebral aperture due to an abnormal separation of the lid fissure and abnormal displacement of lateral canthus and hypoplasia of orbicularis oculi or the tarsal plate. Symblepharon is an adhesion between the palpebral and bulbar conjunctiva. Only one patient had congenital ptosis of the eyelid which is therefore an uncommon finding in present study of patients with CL and P.

Table 3 Ocular abnormalities in relation to type of cleft lip and palate

Bilateral cleft lip & palate—13 (48.14%) cases	
Squint	4
Coloboma lid	1
Microphthalmia	1
Coloboma iris	1
Symblepharon	2
Dacryocystitis	2
Limbal dermoid	2
Cataract	1
Myopia, astigmatism	4
Ptosis	1
Telecanthus	4
Unilateral cleft lip & palate—5 (18.5%) cases	
Telecanthus	2
Ptosis	1
Squint	2
Euryblepharon	1
Congenital nystagmus	1
Hypermetropia	2
Limbal dermoid	1
Facial clefts—2 (7.4%) cases	
Microphthalmia	1
Dacryocystitis	1
Epicanthus	1
Symblepharon	1
Coloboma lid	1
Syndromic—5 (18.5%) cases	
Microphthalmia	1
Coloboma lid	3
Limbal dermoid	1
Nystagmus	1
Hypermetropia	
Cataract	1
Squint	1
Symblepharon	1
Telecathus	1
Hypertelorism	1

Orbital Defects

The main orbital defects seen in patients with CL and P in the present study were hypertelorism and telecanthus. The defective development of palatine and maxillary process in CL and P result in defective medial eye ball migration thus causing hypertelorism

Squint

Disturbance in ocular movement or squint is not unusual in CL and P. In our study 20% of patients had non-paralytic

Fig. 1 Euryblepharon and ectropion **Fig. 2** Ptosis

squint. This is attributed to the abnormal shape and position of the orbital cavity and abnormal insertion of extra ocular muscles or both [9].

Nasolacrimal Apparatus Abnormalities

Patients with CL and P may have defects of the nasolacrimal apparatus from lacrimal punctum to the opening of nasolacrimal duct in the inferior meatus of the nose like;

Fig. 3 Coloboma of the left lower eyelid

agenesis or stenosis of the canaliculi, lacrimal sac, nasolacrimal duct [10]. Three of our patients had congenital dacryocystitis.

Dermoids

Four patients had limbal dermoids, due to entrapment of epidermal cells along the embryonic fusion lines.

Cataract

Two patients had congenital cataract. The interference in the development of lens occurs between fifth and eighth weeks of intrauterine life results in abnormal primary lens fibers.

Microphthalmos

Pure microphthalmos (Nanophthalmos) is a rare condition where the size of the eyeball is smaller than normal (2/3rd of normal size). It is due to arrest in the development of eyeball in all dimensions after the closure of optic fissure.

Coloboma of Iris and Retina

Closure of the embryonic fissure begins mid-way in the fissure and then extends antero-posteriorly, completing the process by the end of the seventh week of gestation. Incomplete closure of the fissure leads to the formation of colobomas of the choroids and retina.

Goldenhar Syndrome or “Oculo-Auriculo-Vertebral (OAV) Syndrome”

Its commonly associated eye abnormalities include narrowing of palpebral fissure, epibulbar dermoid, anophthalmia and colobomas of upper eye lid. Other findings include mandibular hypoplasia, maxillary hypoplasia, macrostomia/lateral cleft lip, preauricular tags/pits, deformity of pinna and external auditory meatus, deformity of middle ear and ossicles causing conductive hearing loss and hypoplasia of temporal bone.

Treacher Collin’s Syndrome (Figs. 4 and 5)

It is characterized by bilateral symmetrical abnormalities of the first and second branchial arches. The main characteristics of Treacher Collin’s Syndrome are downward slanting eyes, small lower jaw, and malformed or missing ears.

Blepharocheinodontic Syndrome (Figs. 6 and 7)

This is a rare autosomal dominant condition of congenital facial clefting. The eyelid anomalies associated with patients with this syndrome are lagophthalmos, eyelid retraction, euryblepharon with cleft lip and palate.

Ocular Hypertelorism (Fig. 8)

It is a condition where there is increased separation between the bony orbits. Characteristic features of the syndrome are median cleft nose, lip and palate. The most common ocular abnormality is non-paretic form of exotropia.

Fig. 4 Treacher Collin Syndrome with operated cleft lip and palate

Fig. 5 Treacher Collin Syndrome with operated cleft lip and palate

Fig. 6 Blepharocheinoontic syndrome with cleft lip and palate

Conclusion

An attempt was made to study the various ocular abnormalities associated with isolated CL and P and in those with syndromes. These abnormalities may be limited to the adnexa and the ocular structures or it could involve both.

Children with cleft lip and palate should be assessed as soon as possible after birth by a multidisciplinary team involving the Pediatrician, the Ophthalmologist as well as specialists from Maxillofacial, ENT and Plastic surgery.

Fig. 7 Blepharocheinoontic syndrome with cleft lip and palate

Fig. 8 Ocular hypertelorism

Appropriate follow-up plans should be drawn to identify and to assess abnormalities involving feeding, hearing, vision, speech, growth and developmental abnormalities. Geneticists and Dysmorphologists have a special role to play in the assessment and counseling.

If an honest and concentrated effort is made, in due course, enough skill and means to support these children and their families can be developed. Early detection of congenital defects is the ultimate aim of medical knowledge and if possible prevention of its occurrence.

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