Original article

Patients with ocular manifestations in congenital craniofacial anomalies at Chiang Mai University Hospital

Sakarin Ausayakhun,[1] Kamolrit Nimitkul,[1] and Krit Khwanngern[2]

[1]Department of Ophthalmology, [2]Department of Surgery, Faculty of Medicine, Chiang Mai University

Objective To report on patients with ocular manifestations in congenital craniofacial anomalies at Chiang Mai University Hospital.

Material and methods Patients were examine and diagnosed with congenital craniofacial anomalies retrospectively at Chiang Mai University Hospital between 1st June 2013 and 28th February 2014.

Results Of 55 patients with congenital craniofacial anomalies, 18 (32.7%) had ocular abnormalities, 1 (1.82%) had craniosynostosis in Crouzon's syndrome, 54 (98.2%) had clefting syndrome, 36 (65.45%) had cleft lip and palate, 15 (27.28%) had mandibulofacial deformitiesand, and 3 (5.45%) had oblique facial clefts, as shown in Table 1. The mean age of the patients was 5.689 years (range 1 month-19 years), of which 38.2% (n=21) were female and 61.8% (n=34) male. Fifty five percent of the patients were below the age of 5 years and 23.6% below the age of 10. The various ocular abnormalities found in this study are listed in Table 2. A total of 21 ocular defects were identified in 18 patients. Abnormalities of the eyelid were the most common, accounting for 28.5% of the total defects (6/21), which included ectropion, lid colobomas, euryblepharon and lagopthalmos. The second most common abnormality was equally strabismus and refractive errors, 4/21(19.0%), followed by nasolacrimal duct obstruction, 3/21(14.3%), limbal dermoid, 1/21(4.8%), preauricular skin tag, 1/21(4.8%), enlarged cupping, 1/21(4.8%), and retinal atrophy, 1/21(4.8%).

Conclusion Common ocular manifestations in patients with congenital craniofacial anomalies included eyelid abnormalities at Chiang Mai University Hospital**. Chiang Mai Medical Journal 2015;54(4):157-61.**

Keywords: ocular manifestations, congenital craniofacial anomalies, craniosysnostosis, cleft lip, cleft palate

Introduction

Patients with congenital craniofacial anomalies are best treated by a multidisciplinary team that applies plastic surgery, neurosurgery, ophthalmology, otolaryngology, oromaxillofacial surgery, orthodontics, anesthesia and genetics, and has the ability to work together in providing overall wellbeing for the patient. To assist in evaluating and treating patients,

Address correspondence to: Sakarin Ausayakhun, M.D., Department of Ophthalmology, Faculty of Medicine,Chiang Mai University, Chiang Mai 50200 Thailand. E-mail: sakarin_a@cmu.ac.th
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the ophthalmologist must process and understand the craniofacial syndrome as well as the necessary medical and surgical interventions required to solve ocular and adnexal problems[1]. Cleft lip and palate represent the second most frequently occurring congenital deformity after clubfoot, and could be associated with many other structural abnormalities in adjacent vital facial structures such as the ears, eyes, nose, teeth and brain. Lip abnormality is most common in patients with clefting syndrome[2]. Syndromic craniosynostoses is rare; nevertheless, there are over 100 syndromic synostoses described, with the most common being Pfeiffer's, Crouzon's, Apert's, and Saethre-Chotzen's syndrome[3,4]. Seventy four point six percent of ophthalmic craniosynostosis manifestations are in the form of strabismus, refractive errors, exophthalmos, and optic atrophy[5]. The most common cause of visual impairment is amblyopia[6]. The purpose of this study, therefore, was to determine ophthalmic findings in patients with congenital craniofacial anomalies at Chiang Mai University Hospital between 1st June 2013 and 28th February 2014 (8 months).

Materials and methods

This study retrospectively reviewed the medical records of patients diagnosed with congenital craniofacial anomalies and examined ocular problems in the outpatients' Department of the Ophthalmology Department at Chiang Mai University Hospital, Chiang Mai, Thailand, between 1st June 2013 and 28th February 2014. The patients were divided into 2 groups: 1) craniosynostosis, including Apert's, Crouzon's and Pfeiffer's syndrome as well as other isolated suture abnormalities, and 2) clefting syndrome including cleft lip and palate, oblique facial clefts and mandibulofacial deformities such as Goldenhar's syndrome, Treacher Collins syndrome and facial microsomia.

Results

Of 55 patients with congenital craniofacial anomalies, 18 (32.7%) had ocular abnormalities, 1 (1.82%) had craniosynostosis in Crouzon's syndrome, 54 (98.2%) had clefting syndrome, 36 (65.45%) had cleft lip and palate, 15 (27.28%) had mandibulofacial deformities and 3 (5.45%) had oblique facial clefts, as shown in Table 1.

The mean age of the patients was 5.689 years (range 1 month-19 years), of which 38.2% (n=21) were female and 61.8% (n=34) male. Fifty five percent of the patients were below the age of 5 years, and 23.6% below the age of[10]. The various ocular abnormalities found in this study are listed in Table 2. A total of 21 ocular defects was identified in 18 patients. Abnormalities of the eyelid were the most common, accounting for 28.5% of the total defects (6/21), which included ectropion, eyelid colobomas, euryblepharon and lagopthalmos. The second most common abnormality was equally strabismus and refractive errors, 4/21(19.0%), followed by nasolacrimal duct obstruction, 3/21(14.3%), limbal dermoid, [1/21 (4.8%)], preauricular skin tag, [1/21 (4.8%)], enlarged cupping, [1/21 (4.8%)], and retinal atrophy, 1/21(4.8%).

Table 1. Patients with congenital craniofacial anomalies

Craniosynostosis	Clefting syndrome	Total patients
Crouzon's syndrome – 1 (1.82%) patien	Cleft lip and palate – 36 (65.45%) patients CLCP-29, CL-5,CP-2 Mandibulofacial deformities – 15 (27.28%) patients Goldenhar's syndrome – 5 patients Treacher Collins syndrome – 2 patients Facial microsomia – 8 patients Oblique facial clefts - 3 (5.45%) patients	55

*CLCP= cleft lip and cleft palate, CL= cleft lip, CP= cleft palate

Table 2. Various ocular abnormalities found from 55 cases

Type of abnormality	Total number of defects (%)
Eyelid abnormality	6 (28.5%)
Strabismus	4 (19.0%)
Refractive errors	4 (19.0%)
Nasolacrimal duct obstruction	3 (14.3%)
Limbal dermoid	1 (4.8%)
Preauricular skin tag	1 (4.8%)
Enlarged cupping	1 (4.8%)
Retinal atrophy	1 (4.8%)
Total	21

Table 3. Ocular abnormalities in relation to type of clefting syndrome

Cleft lip and palate – 36 (65.45%) patients	
Cleft lip and cleft palate-29 patients	
Strabismus	1
Refractive errors	1
Nasolacrimal duct obstruction	1
Enlarge cupping	1
Lagopthalmos	1
Cleft lip – 5 patients	-
Cleft palate – 2 patients	
Strabismus	1
Mandibulofacial deformities – 15 (27.28%) patients	
Goldenhar's syndrome – 5 patients	1
Refractive errors	1
Limbal dermoid	1
Preauricular skin tag	
Treacher Collins syndrome – 2 patients	1
Refractive errors	1
Ectropion	1
Lagopthalmos	1
Euryblepharon	1
Coloboma lid	
Facial microsomia – 8 patients	1
Refractive errors	1
Nasolacrimal duct obstruction	1
Strabismus	
Oblique facial clefts – 3 (5.45%) patients	
Nasolacrimal duct obstruction	1
Lagopthalmos	1
Retinal atrophy	1

Discussion

This retrospective descriptive study showed that the majority of craniofacial anomalies was clefting syndrome at 98.1% (n=55/56), and one patient with craniosynostosis in Crouzon's syndrome had V-pattern exophoria. Abnormalities of the eyelid were the most common, accounting for 28.5% of all the defects.

Lid abnormalities

Embryological fusion of the maxillary and palatine processes, and development of the eyelid occur almost simultaneously (in the sixth to eighth week of intrauterine life), thus a developmental defect of the palate could lead to an ocular defect[7]. An isolated eyelid coloboma could be the result of pressure from the amniotic band or due to localized failure of lid fold fusion. Euryblepharon is characterized by primary symmetric enlargement of the horizontal palpebral aperture, due to abnormal separation of lid fissure and abnormal displacement of the lateral canthus and hypoplasia of the orbicularis oculi or tarsal plate. The eye problems in Treacher Collins syndrome vary from coloboma of the lower eyelids and aplasia of lid lashes to short, down-slanting palpebral fissures and missing eyelashes[8]. This study found different types of eyelid defects such as lagophthalmos, coloboma, euryblepharon and ectropion, as seen in one of two patients with Treacher Collins syndrome. Previous study showed lid abnormality as the most common anomaly in patients with clefting syndrome[2]. In this study, lagophthalmos also was found in cleft lip and palate and facial microsomia cases, as shown in Table 3.

Strabismus

In a previous study, children with craniosynostosis syndromes commonly had strabismus[9]. In this study, 4 (19.0%) of the total number of defects had V-pattern exophoria in Crouzon's syndrome, alternate esotropia in Pierre Rubin syndrome, right hypotropia in cleft lip and palate, and alternate exotropia in facial microsomia.

Refractive errors

This study had 4 (19.0%) refractive errors that were mostly myopia and astigmatism.

Three (14.3%) nasolacrimal duct obstructions were found in cleft lip and palate, facial microsomia and oblique facial cleft, with most of them having a scar contracture that created nasolacrimal duct obstruction. One of five Goldenhar's syndrome cases in this study had limbal dermoid and preauricular skin tag, which were not found in the other four.

The limitations of this study were missing data, cases of craniosysnostosis that were rare during the study period, with only one found in Crouzon's syndrome, and most of the patients were not of Thai ethnicity, which produced problems of communication during examinations and follow up.

Conclusion

This retrospective descriptive review showed that eyelid abnormality was the most common ocular anomaly in patients with congenital craniofacial anomalies at Chiang Mai University Hospital. The second most common abnormality was equally strabismus and refractive errors.

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ความผิดปกติทางตาที่ตรวจพบในผู้ป่วยที่มีความพิการบนใบหน้าและศีรษะแต่กำเนิด ในโรงพยาบาลมหาราชนครเชียงใหม่

ศักรินทร์ อัษญคุณ,[1] กมลฤทธิ์ นิมิตรกุล[1] และ กฤษณ์ ขวัญเงิน[2]
[1]ภาควิชาจักษุวิทยา, [2]ภาควิชาศัลยศาสตร์ คณะแพทยศาสตร์ มหาวิทยาลัยเชียงใหม่

วัตถุประสงค์ เพื่อรายงานความผิดปกติทางตาที่ตรวจพบในผู้ป่วยที่มีความพิการบนใบหน้าและศีรษะ ในโรงพยาบาลมหาราชนครเชียงใหม่

วัสดุและวิธีการ การศึกษาย้อนหลังเวชระเบียนในผู้ป่วยที่ได้รับการวินิจฉัยว่ามีความพิการบนใบหน้าและศีรษะ ในช่วงเวลาตั้งแต่ มิถุนายน พ.ศ. 2556 ถึงกุมภาพันธ์ พ.ศ. 2557 ในโรงพยาบาลมหาราชนครเชียงใหม่

ผลการศึกษา ผู้ป่วยจำนวน 55 ราย พบว่าผู้ป่วยมีอายุโดยเฉลี่ย 5.6±5.4 ปี (ช่วงอายุตั้งแต่ 1 เดือน ถึง 19 ปี) แบ่งเป็นเพศหญิงร้อยละ 38.2 (21 ราย) ตรวจพบความผิดปกติทางตาจำนวนร้อยละ 32.7 (18 ราย) โดยความผิดปกติที่พบมากที่สุดคือความผิดปกติที่เปลือกตา ร้อยละ 28.5 รองลงมาคือตาเข ร้อยละ 19.0 สายตาผิดปกติ ร้อยละ 19.0 และท่อน้ำตาอุดตันร้อยละ 14.3 ตามลำดับ ความผิดปกติอื่น ๆ ที่ตรวจพบได้แก่ dermoid, preauricular skin tag, ขั้วประสาทตาใหญ่ และจอตาฝ่อ

สรุป ความผิดปกติทางตาที่ตรวจพบได้มากที่สุดในผู้ป่วยที่มีความพิการบนใบหน้าและศีรษะ ในโรงพยาบาลมหาราชนครเชียงใหม่ คือความผิดปกติที่เปลือกตา **เชียงใหม่เวชสาร 2558;54(4):157-61.**

คำสำคัญ: ความผิดปกติทางตา ความพิการบนใบหน้าและศีรษะ ความผิดปกติที่เปลือกตา