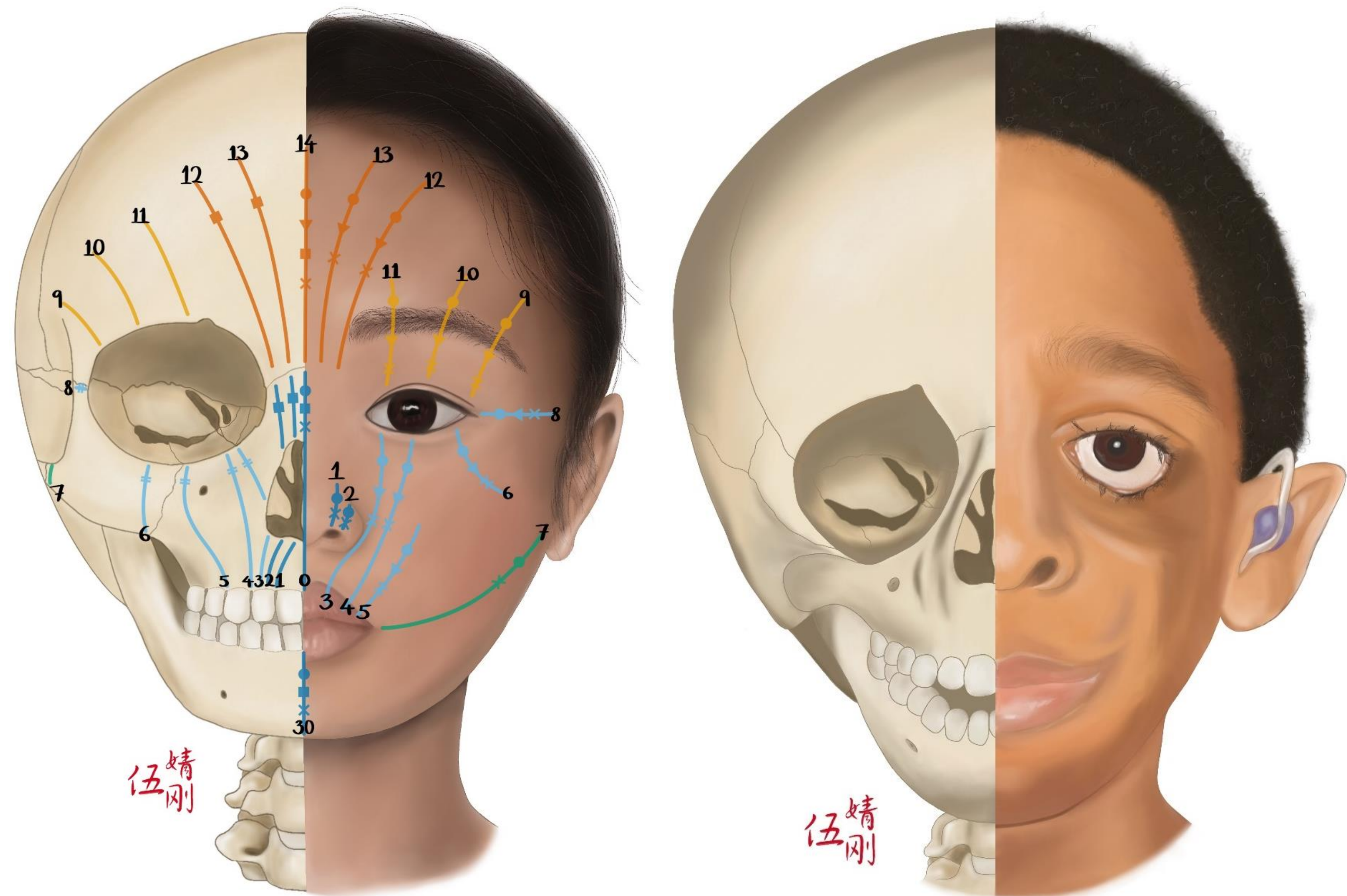


Periorbital Outcomes of Rare Craniofacial Clefts

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BACKGROUND

- Craniofacial clefts are exceedingly rare congenital anomalies with a kaleidoscopically diverse range of manifestations.
- They are distinct from common orofacial clefts in that they involve a defect of the nose, maxilla, mandible, periorbital region, or cranium.
- Depending on the location and severity, craniofacial clefts can have differential effects on facial units, and may have substantial functional concerns regarding the functioning of the eyes, eyelids, and nasolacrimal system



OBJECTIVES

We examined the periorbital outcomes of rare craniofacial clefts, with attention paid to ophthalmologic findings, surgical burden, and vision.

METHODS

- Retrospective cohort study of patients diagnosed with a rare craniofacial cleft and who underwent evaluation by the Division of Plastic, Reconstructive and Oral Surgery, and the Division of Ophthalmology at our institution
- Patients were divided into two cohorts: Treacher Collins syndrome, and all other craniofacial clefts: clefts were categorized into superior periorbital (Tessier 9 to 11), inferior periorbital (3 to 6, 8), superior medial (12 to 14), inferior medial (0 to 2, 30), and lateral (7)
- Primary outcomes were periorbital findings, surgical procedures, and visual acuity at latest ophthalmologic examination.

Category	Fixation	Teller	Optotype
Good	CSM at age <6 months	Normal range for age	Better than 20/80
Fair	CSM at age >6 months	Just under normal range for age	20/200 to -20/80
Poor	CSuM or worse	Far below normal range for age	Worse than 20/200

CSM, central, steady, maintained; CSuM, central, steady, unmaintained; Teller, Teller preferential looking testing. Source: Areaux RG, Jr., Orlin SE, Zaidman GW, et al. Anatomic and visual outcomes of corneal transplantation during infancy. J AAPOS. Jun 2020;24(3):134 e1-134 e6. doi:10.1016/j.jaapos.2020.02.003

RESULTS: CRANIOFACIAL CLEFT COHORT

- 104 patients were included in the craniofacial cohort. Mean follow-up was 9 years.
- Approximately 80% of the cohort had one or more periorbital abnormalities, the most common being ocular surface pathology, strabismus, and nasolacrimal duct obstruction (NLDO). Almost 40% had poor vision in one or more eyes.
- Superior periorbital clefts were more likely to have ocular surface pathology, lagophthalmos, eyelid retraction. Inferior periorbital clefts were more likely to have eyelid retraction, ectropion.
- Superior medial clefts were associated with hypertelorism and entropion. Inferior medial clefts were more likely to present with hypertelorism.
- Among 25 cases of NLDO, 18 were congenital and 5 were acquired. Those who underwent hypertelorism correction were more likely to develop NLDO.
- Mean age at most recent visual acuity measurement was 9 years. Among 202 eyes, vision was “Good” in 129 (63.9%), “Fair” in 27 (13.4%), and “Poor” in 46 (22.8%). Most common etiologies of poor vision were microphthalmos or anophthalmos, exposure keratopathy; optic nerve hypoplasia or atrophy; intraocular coloboma.

RESULTS: TREACHER COLLINS COHORT

- In this subset, we had 50 total patients with a mean follow-up of 10 years. The most common periorbital abnormalities were downslanting palpebral fissures, eyelash deficiency, and eyelid coloboma.
- 20 patients underwent 86 periorbital procedures.
- Lateral canthopexies accompanied by malar fat grafting less likely to require reoperation (0% versus 72%, p=0.004).
- Eyelid coloboma more likely to have exposure keratopathy (30% versus 4%, p=0.020).
- Visual acuity in 78 eyes of 39 patients: Good, 90%; Fair, 5%; Poor, 6%
- Poor vision etiology: Intraocular coloboma (n=2 eyes); Orbital apex syndrome (n=2); Anisometropic amblyopia (n=1)

Risk of Vision Loss	Associated Ophthalmologic Findings	Percentage of Study Cohort (%)	Periorbital Management Considerations
Low	- Downslanting palpebral fissures - Eyelash deficiency - Eyelid coloboma - Trichiasis/distichiasis	58	- Perinatal ophthalmology evaluation. - Unlikely to require ophthalmology follow-up.
Moderate	- Entropion - Ectropion - Eyelid retraction - Nasolacrimal duct obstruction - Lagophthalmos	18	- Perinatal ophthalmology evaluation. - Requires ophthalmology follow-up. - May require surgical intervention during childhood.
High	- Amblyogenic conditions, such as refractive error, strabismus, and deprivation (including cataract, ptosis, and corneal scar). - Eyelid coloboma causing high-risk corneal exposure. - Other structural eye disease, including optic nerve or retinal coloboma.	24	- Perinatal ophthalmology evaluation. - Requires ophthalmology follow-up. - May require surgical intervention in infancy.

- In collaboration with pediatric ophthalmologists and oculoplastic surgeons, we created a vision risk scale. Among patients with a visual acuity measurement at age 6 or older (n=29), those with “High” vision risk were more likely to have “Poor” vision in any eye (13%), those with “Moderate” risk were more likely to have “Fair” vision in any eye (33%), and those with “Low” risk were more likely to have “Good” vision in both eyes (100%) (p=0.050).

CONCLUSION

There was a high prevalence of periorbital abnormalities and significant surgical burden. All individuals with craniofacial clefts are at risk for vision loss. Additionally, acquired nasolacrimal duct obstruction associated with hypertelorism correction. Among those with Treacher Collins syndrome, most subjects had good vision bilaterally with appropriate management. Early ophthalmologist evaluation and follow-up is recommended.

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