Management of Severe Proptosis from Pfeiffer Syndrome

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Abstract: Pfeiffer Syndrome is a genetic disorder in which premature fusion of skull occurs causing features such as extreme proptosis. Therefore, conventional glasses are not ideal because they can cause severe corneal damage.
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- **Case History**
  - 7-year-old African American male
  - Chief complaint: misplaced his glasses and his corneas touch the lenses on regular glasses
  - Ocular history
    - Corneal scarring
    - Severe proptosis
    - Exposure keratopathy
    - Exotropia
    - Photophobia
    - Myopia
    - Surgical history of tarsorrhaphy in both eyes to reduce globe exposure
  - Medical history
    - Pfeiffer syndrome
    - Hearing loss
    - Speech impairments
    - Cranial deformities
    - Airway anomalies
  - Medication
    - Lacrilube ointment 1 stip QID OU
    - Azopt 1% 1 gtt BID OU
    - Erythromycin 0.5% 1 strip QHS OU
    - Fluromethalone 0.1% 1 gtt BID OU

- **Pertinent Findings**
  - Clinical
    - Eye muscles: Full range of muscles
    - Fix and follow: Yes, in all gazes
    - Cover test: Intermittent left exotropia, no complaints of diplopia
    - Peripheral vision: Restricted 360 OU due to eyelid structure
    - BCVA: 20/180 using Teller Acuity Cards
  - Physical
    - Lids: Tarsorrhaphy sutures are present on both nasal and temporal lids
    - Lashes: Matted with artificial tears ointment in both eyes
    - Conjunctiva and sclera: Trace injection in both eyes
    - Corneas: Diffuse scarring 360, edematous OU
    - Iris: Flat
    - Anterior chamber: Deep and quiet
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- Dilated exam result obtained from ophthalmologist

**Differential Diagnosis**
- Primary diagnosis:
  - Pfeiffer Syndrome causing proptosis and exposure keratopathy
- Others:
  - Rhabdomyosarcoma
  - Retinoblastoma
  - Orbital cellulitis

**Diagnosis and Discussion**
- Pfeiffer syndrome is characterized by
  - Craniosynostosis - premature closure of cranial structures
  - Syndactyly - a webbing of fingers or toes
- Discussion:
  - Pfeiffer syndrome has an estimated incidence of one in 100,000
  - Pfeiffer syndrome has high incidence of hearing, vision, airway issues, and acquired Chiari malformations
  - Mild variants of Pfeiffer syndrome tend to be hereditary and carry a good prognosis
  - Ocular signs are some of the most prominent characteristics and proper management of the patient symptoms is important in preventing and/or reducing vision loss
  - Vision loss with this syndrome occurs from at least two different mechanisms
    - The first mechanism includes optic atrophy created by sustained elevation in intracranial pressure
    - The second mechanism includes exposure keratopathy and corneal infections from extreme proptosis due to shallow orbits
    - Permanent tarsorrhaphies are one of the first operative procedures undertaken on severely affected infants to prevent development of chemosis and further scarring
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- **Treatment and Management**
  - Treatment includes generous lubrication for his ocular surface
  - Correct any significant refractive errors with glasses
    - Therefore, frame selection is crucial when facial dysmorphisms and severe proptosis exists.
    - Since the protrusion of his globe will cause the cornea to rub against the lens in a typical fitted frame, a custom designed frame needs to be produce to accommodate his proptosis, facial structure, and hearing aid.
    - The custom designed frame was made closely with the optician.
      - 1st frame front without lenses and temples is glued to a 2nd frame front with the patient’s actual prescription and temples (design to be shown via photos)
  - Transitions lenses to be made due to patient’s photophobia

- **Response to treatment:**
  - Able to adapt to new glasses and able to wear glasses full time without the lenses touching his corneas.

- **Management includes continue with follow up visits with current ophthalmologist for management of exposure keratopathy and elevated intraocular pressure**

- **Future recommendations:**
  - Large print reading used at school
  - Continue services with visually impaired teacher
  - Recommend orientation and mobility training
  - Consideration of monocular telescope use in the near future
  - Consideration of conventional or electronic magnifiers in the near future

- **Bibliography:**

- **Conclusion**
  - Pfeiffer syndrome is a developmentally detrimental diagnosis and mainly affects the craniofacial areas. As eye care professionals, we are essential in the role of managing the patient’s symptoms and providing solutions to allow the patient to successfully wear glasses.
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- Conventional glasses’ frames are not very effective in patient’s with extreme proptosis. Rather, innovative ways such as the one prescribed to this patient may be the only way to help this patient population to maximize visual potential.