Facial Abnormality in a Neonate: What Is Your Diagnosis?

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A full-term neonate was brought to the emergency department on day 20 of life with an unusual facial abnormality, as depicted in Figure 1. Examination revealed facial asymmetry, macrostomia, and deficient perioral musculature without facial nerve palsy. The neonate had external ear deformities with preauricular skin tags, but palatal integrity was intact. No respiratory distress or other systemic anomalies were noted. Initial management focused on assisted feeding support, airway assessment, and a multidisciplinary evaluation to determine syndromic associations and plan for surgical correction.

Figure 1: A day 20 neonate with a deep lateral facial cleft extending from the oral commissure to the temporal region, showing an unfused mouth angle and a distal skin tag.

Question

1. **What is the clinical diagnosis?**

   a. Goldenhar Syndrome

   b. Treacher-Collins Syndrome

   c. Hemifacial Microsomia

   d. Isolated Tessier Cleft 7

Answer

d. Isolated Tessier Cleft 7

Discussion

Tessier clefts are a classification system for craniofacial clefts proposed by Paul Tessier in 1976, numbering clefts from 0 to 14 based on their anatomical location.[1] Tessier cleft 7, also known as lateral facial cleft or transverse facial cleft, is a rare congenital anomaly with an incidence of 1/80000-1/300000 live births.[2] It affects the oral commissure and extends toward the ear or temporal region. It results from a developmental failure of fusion between the maxillary and mandibular processes of the first branchial arch during the fifth week of intrauterine life [Figure 2]. The first and second pharyngeal arches contribute to the formation of the face, and improper merging of these structures can lead to macrostomia and associated anomalies. This defect is thought to be caused by genetic mutations, environmental teratogens, or amniotic band disruption during early fetal development.[3] Tessier cleft 7 varies from mild macrostomia (widened mouth opening) to a deep cleft with soft tissue and bony defects affecting facial symmetry.

While Tessier cleft 7 may present as an isolated anomaly, it is often associated with craniofacial syndromes, particularly those involving the first and second pharyngeal arches. The most common syndromic associations include:

- Goldenhar Syndrome (Oculo-Auriculo-Vertebral Spectrum, OAVS): Characterized by hemifacial microsomia, ear deformities, epibulbar dermoids, vertebral anomalies, and congenital heart defects.[4]
- Treacher-Collins Syndrome: Features zygomatic and mandibular hypoplasia, down-slanting palpebral fissures, ear anomalies, and dental malformations.[4]
- Hemifacial Microsomia: Unilateral underdevelopment of the mandible, maxilla, and soft tissues, often affecting facial symmetry.
- Other rare associations: Craniofacial microsomia, branchio-oto-renal syndrome, and oto-mandibular dysplasia.

Clinically, patients exhibit facial asymmetry, absent or hypoplastic perioral musculature, and external ear abnormalities. Although facial nerve function is typically preserved, severe cases may present with asymmetrical facial movement. A major concern in affected neonates is feeding difficulty due to an incomplete oral seal, drooling, and risk of aspiration, which can lead to poor weight gain and nutritional deficiencies. Early intervention with specialized cleft palate bottles, feeding therapy, and multidisciplinary evaluation is essential for optimal neonatal care.

Surgical correction of Tessier cleft 7 is typically performed between 3 to 6 months of age, depending on the severity and associated anomalies. The primary objectives of surgery are to restore oral competence, achieve facial symmetry, preserve facial nerve function, and minimize scarring.[5] Various surgical techniques, including Z-plasty, straight-line closure, vermilion square flap, and soft tissue rearrangement, are employed to reconstruct the perioral musculature and align the oral commissure. In cases with bony involvement, additional maxillofacial procedures may be necessary to correct mandibular or maxillary hypoplasia.[2,6] Postoperative management is crucial for long-term success and includes scar management, speech therapy, orthodontic evaluation, and regular follow-ups to monitor facial growth, dental alignment, and functional outcomes. Long-term multidisciplinary care ensures optimal aesthetic and functional rehabilitation, improving both feeding efficiency and speech development, ultimately enhancing the patient's quality of life and psychosocial well-being.

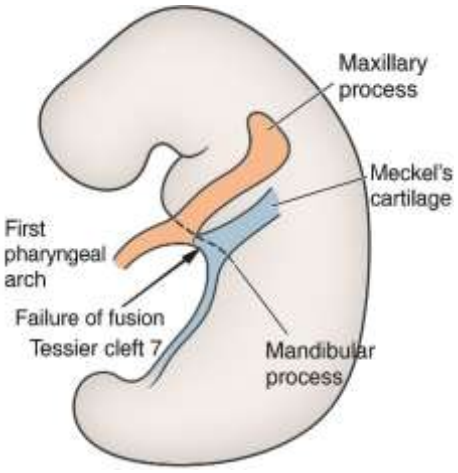

Figure 2: Schematic representation of the embryology of Tessier cleft 7, illustrating the failure of fusion between the maxillary and mandibular processes of the first pharyngeal arch, leading to a lateral facial cleft.

Conclusion

Tessier cleft 7 is a rare but significant craniofacial anomaly requiring early diagnosis, multidisciplinary intervention, and tailored surgical correction. Timely feeding support, surgical repair, and long-term follow-up are essential to ensure optimal functional, aesthetic, and psychosocial outcomes for affected individuals.

Disclosure

The authors declared no conflicts of interest. Written consent was obtained from the kin of the patient.

References

1. Tessier P. Anatomical classification facial, cranio-facial and latero-facial clefts. J Maxillofac Surg 1976 Jun;4(2):69-92.

2. Khorasani H, Boljanovic S, Knudsen MA, Jakobsen LP. Surgical management of the Tessier 7 cleft: A review and presentation of 5 cases. JPRAS Open 2019 Jul;22:9-18.

3. Coleman JR Jr, Sykes JM. The embryology, classification, epidemiology, and genetics of facial clefting. Facial Plast Surg Clin North Am 2001 Feb;9(1):1-13.

4. Kuriyama M, Udagawa A, Yoshimoto S, Ichinose M, Suzuki H. Tessier number 7 cleft with oblique clefts of bilateral soft palates and rare symmetric structure of zygomatic arch. J Plast Reconstr Aesthet Surg 2008;61(4):447-450.

5. Mannali AR. M P, Kumar S. Tessier 7 Cleft: Clinical Presentation and Surgical Correction in a Case Report. Cureus [Internet]. 2024 [cited 2025 Mar 31]; Available from: https://www.cureus.com/articles/295493-tessier-7-cleft-clinical-presentation-and-surgical-correction-in-a-case-report

6. Woods RH, Varma S, David DJ. Tessier no. 7 cleft: a new subclassification and management protocol. Plast Reconstr Surg 2008 Sep;122(3):898-905.