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Case Report

An unusual occurrence: Treacher Collin syndrome with nasal dermoid cyst

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ABSTRACT

Treacher-Collins syndrome (TCS), characterized by autosomal dominant inheritance, affects craniofacial development and presents with diverse clinical manifestations, including facial bone hypoplasia, ear malformations, and ocular abnormalities. The syndrome's association with nasal dermoid cysts is rare and poorly documented in the literature. We present a case report of a 10-year-old male with TCS who presented with a nasal dermoid cyst, a noteworthy and atypical finding within this patient population. The patient exhibited typical features of TCS, including facial asymmetry, malar and mandibular hypoplasia, and microtia. The diagnosis was confirmed through clinical evaluation and diagnostic imaging, which revealed additional anomalies such as conductive hearing loss and absence of the external auditory canal. Surgical excision of the nasal dermoid cyst was performed, utilizing the V-Y advancement glabellar flap technique for closure. This case underscores the importance of comprehensive multidisciplinary care in managing complex conditions like TCS, where treatment strategies must address both medical needs and cosmetic outcomes. Further exploration of rare associations like nasal dermoid cysts in TCS contributes to a broader understanding of the syndrome's phenotypic variability and informs optimal clinical management strategies.

Keywords: Treacher Collins syndrome, Nasal dermoid cyst, Craniofacial anomalies, Multidisciplinary care, Case report

INTRODUCTION

Treacher Collins syndrome (TCS), also called Treacher Collins-Franceschetti syndrome or mandibulofacial dys¬ostosis, is an autosomal dominant disorder affecting the development of structures derived from the first and second brachial arches during early embryonic development. The estimated incidence of TCS ranges from 1:40,000 to 1:70,000 of live births.[1] From a genetic standpoint, Treacher-Collins syndrome (TCS) exhibits a high degree of penetrance, meaning that individuals who carry the genetic mutation are very likely to display some syndrome features. However, there is considerable variability in how these features manifest, known as variable phenotypic expression. The clinical characteristics of TCS result from a loss-of-function mutation in the TCOF1 gene located on chromosome 5.[2] Treacher-Collins syndrome is characterized by deafness, hypoplasia of facial bones (mandible, maxilla, and cheekbone), antimongoloid slant of palpebral fissures, coloboma of the lower lid, and bilateral anomalies of the auricle. Hypoplasia of the facial bones may be the first indicator of the disorder.[3] Patients usually present with bilateral microtia or anotia of varying severity. The inner ear is usually morphologically normal; however, from a functional standpoint, the pathologic ossicular chain results in conductive hearing loss in about 40–50% of suffering individuals.[4] Patients with Treacher-Collins

syndrome (TCS) may sometimes have additional abnormalities beyond the typical features. These can include choanal atresia, complete or sub mucous cleft palate, absent parotid glands, cervical spine malformations, cryptorchidism, extremity malformations, renal anomalies, and congenital heart disease. However, these are not defining characteristics of TCS and do not consistently occur in every patient with the syndrome.[5] The nose is often described as "beaked"; however, anthropometric studies of TCS patients' noses show relatively normal nose measurements, and the hypoplasia of the surrounding tissue is the main contributor to abnormal facial balance.[6]

Patients with this condition do not typically experience developmental delays or neurologic diseases. However, they often encounter significant social challenges throughout their lives due to their physical appearance. To address their needs effectively, care must be provided by a multidisciplinary team starting from birth and continuing into adulthood. This comprehensive approach ensures that all aspects of the patient's well-being, including physical health, psychological well-being, and social integration, are carefully considered and addressed. Proper planning, thorough counselling, and advanced surgical techniques are crucial components for achieving the best possible outcomes for these patients.

In this case report, we detail the rare and atypical occurrence of a nasal dermoid cyst in a 10-year-old child who has been diagnosed with hemifacial microsomia, commonly referred to as Treacher-Collins syndrome. This presentation is unusual and noteworthy given the typical clinical manifestations associated with Treacher-Collins syndrome. By documenting this case, we seek to enhance the understanding of the potential variations and complexities in Treacher-Collins syndrome, emphasizing the importance of thorough evaluation and individualized care in managing these rare associations.

CASE REPORT

A 10-year-old male patient presented to our outpatient department with a complaint of a wound on the right side of his nose, which had been discharging mucopurulent fluid for the past three months, as shown in figure 1. This wound was initially preceded by swelling and pain at the same site, which eventually subsided once the discharge commenced.

Further investigation revealed that the patient had a similar issue nine years earlier. During that episode, an abscess had formed at the same location. The abscess was treated with an incision and drainage procedure, followed by conservative management to alleviate the symptoms. The patient's past medical history did not reveal any significant findings. His birth history was unremarkable, and he had met all developmental milestones at the appropriate ages. Additionally, there was no consanguinity reported in the parents' marriage.

Examination

Upon further examination, several distinctive and striking facial features were observed in the patient. These included noticeable facial asymmetry on the right side, underdevelopment (hypoplasia) of the malar (cheek) and mandibular (jaw) prominences, and a depressed and widened nasal bridge. Additionally, the patient exhibited a down-slanting palpebral fissure on the left side.

There was also evidence of a preauricular epithelial tissue tag on the left side. Examination of the left external ear revealed significant deformity, which was classified as Marx grade II microtia according to the Marx classification system for grading microtia.[7] These findings are depicted in figure 2. This comprehensive assessment highlighted the complex craniofacial anomalies present in the patient and led us to provisionally form the diagnosis of hemifacial microsomia in this patient.

Figure 1: Clinical photograph of the patient.

Figure 2: Clinical examination findings of the patient.

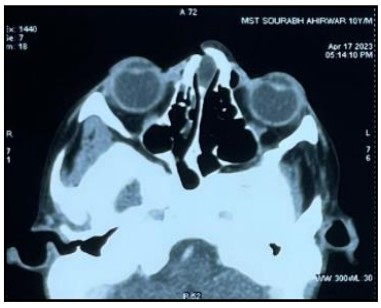

Figure 3: NCCT Head showing absence of left EAC.

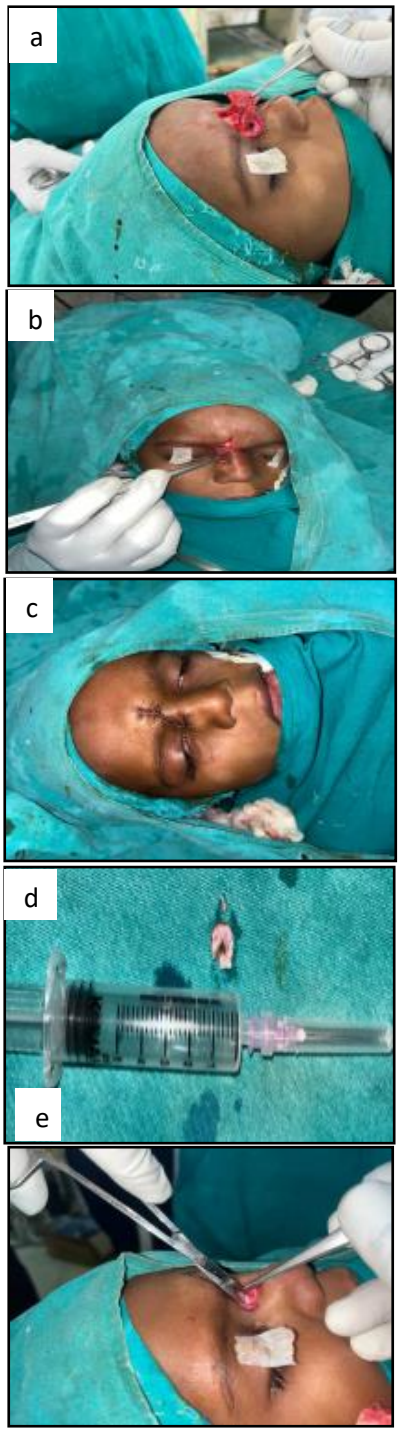

Figure 4: (a) Excision of nasal dermoid cyst; (b) defect reconstruction using glabellar flap; (c) defect reconstruction using glabellar flap; (d) immediate post-operative picture & (e) excised dermoid cyst.

Investigations and management

Following the interim diagnosis, a series of investigations were conducted to substantiate the suspicion of Treacher-Collins syndrome in the patient. Additionally, these investigations aimed to thoroughly assess the presenting complaint of a nasal dermoid cyst, ensuring a comprehensive and effective management plan for the patient's condition. The first diagnostic step was a pure tone audiogram, which indicated a conductive hearing loss of 48.6 dB on the left side. Subsequently, a non-contrast CT scan of the face was performed to examine the internal structures in detail. The CT scan findings revealed a complete absence of the external auditory canal on the left side as shown in figure 3. The results from both of these investigations further substantiated the suspicion of Treacher-Collins syndrome in the child.

To evaluate the patient's current concern, a Sinogram was conducted with the use of contrast dye and fluoroscopy. This procedure successfully identified a 2 cm tract terminating in a blind sac containing a collection.

The nasal dermoid cyst was treated with early surgical excision, aiming for complete removal while preserving the cyst wall integrity. The procedure also included excising the associated sinus tract. Incomplete or partial excision of the cyst wall leads to 20% recurrence rates.[8] Closure of the resulting defect utilized a V-Y advancement glabellar flap technique, ensuring effective coverage and promoting optimal healing. The steps of the procedure, along with the V-Y advancement glabellar flap technique for closure, are illustrated in figure 4. This approach is standard for managing nasal dermoid cysts, emphasizing complete excision and meticulous closure for favourable cosmetic and functional outcomes.

DISCUSSION

Treacher Collins syndrome characterized by autosomal dominant inheritance, manifests with distinctive clinical features stemming from abnormalities in structures originating from the first and second brachial arches.

Treacher-Collins syndrome (TCS) was first described in 1889 by George Andreas Berry. He identified it as a congenital neonatal deformity characterized by colobomata of the lower eyelids. Berry's initial description marked the beginning of the medical understanding of this condition.[9] In the year 1900, Edward Treacher Collins, an ophthalmologist, made a significant contribution to medical literature by presenting the cases of two patients at a meeting in London. These patients exhibited specific ocular and perioral complications.

Following his presentation, Collins went on to publish one of the earliest detailed case reports. In this report, he meticulously described the findings and characteristics associated with these conditions, laying the groundwork for future research and understanding in this area.[10] In the 1940s, Adolphe Franceschetti of Switzerland, along with his colleague David Klein, conducted extensive research on the disorder. They provided a more detailed characterization of its facial features and introduced the term "mandibulofacial dysostosis" to describe it. Their work significantly advanced the understanding of the

disorder.[11] The most frequent clinical manifestations, among a great variety of alterations, include antimongoloid slanting of palpebral fissures (89%), malar hypoplasia (81%), mandibular hypoplasia (78%), auricular pinna malformations (77%), and lower palpebral coloboma (69%).[12] Treacher-Collins syndrome shares differential diagnoses with several clinical entities, such as Goldenhar syndrome (vertebral oculoauricular dysplasia), Nager's acrofacial dysostosis, and Miller's syndrome. A distinguishing feature of Treacher-Collins syndrome is its characteristic symmetrical and bilateral involvement of craniofacial structures.[13] The correlation between Treacher Collins syndrome and nasal dermoid cysts is rare and has received limited exploration in the literature.

A dermoid cyst is a benign cutaneous developmental anomaly that arises from the entrapment of ectodermal elements along the lines of embryonic closure.[14] They can develop anywhere on the body, but dermoid cysts are most often found in the periorbital lateral eyebrow area (in the head and neck area). For the first time in medical history in 1937, New and Erich categorized congenital inclusion dermoid cysts in the head and neck area into four groups: the periorbital region (group 1), the nose (group 2), the submental region (group 3), and the midventral and mid-dorsal fusion area of the suprasternal, thyroidal, and suboccipital region (group 4).[15]

Nasal dermoids account for 5-10% of dermoid lesions and have traditionally been described as light, flesh-coloured, pearly, or erythematous nodules or masses in the literature.[16] The objective of the case report is to explore the relationship between Treacher Collins syndrome, a genetic disorder affecting craniofacial development, and the occurrence of nasal dermoid cysts.

Following the establishment of a TCS diagnosis using procedures such as molecular genetic testing and, in certain cases, clinical examination, and adequate multidisciplinary craniofacial care should be carried out. Performing even routine restorative procedures can be particularly challenging in this patient population due to the presence of concurrent medical conditions. These conditions include congenital heart defects, compromised oropharyngeal airways, hearing impairment, and significant treatment-related anxiety. As a result, these patients typically require a comprehensive and interdisciplinary treatment approach. This approach involves collaboration among specialists in audiology, speech and language pathology, otorhinolaryngology, general dentistry, orthodontics, oral and maxillofacial surgery, as well as plastic and reconstructive surgery. The goal of this collaborative effort is not only to address health needs but also to enhance facial aesthetics and overall quality of life.

The most important course of action for a dermoid cyst is early resection with full surgical excision, but only after adequate pre-operative radiological studies to rule out any intracranial or intraspinal extension or any unusual presentation, as was done in this case report. The progressive expansion of a nasal dermoid can result in soft tissue and skeletal deformities, local infection, meningitis, and brain abscess. As a result, timely detection is critical, and surgical excision is the sole treatment option. The prognosis is generally favorable. Awareness of their diverse clinical presentations, thorough radiological assessment, and appropriate surgical intervention are essential in ensuring positive patient outcomes in cases of dermoid cysts.

CONCLUSION

In conclusion, the association between Treacher Collins syndrome (TCS) and nasal dermoid cysts represents a rare and unusual occurrence, as highlighted in this case report. TCS, an autosomal dominant disorder affecting craniofacial development, typically manifests with distinctive clinical features such as facial bone hypoplasia, ear malformations, and ocular abnormalities. The presence of a nasal dermoid cyst in a patient with TCS adds complexity to the clinical presentation, underscoring the variability in phenotypic expression associated with this genetic condition.

Effective management of TCS and associated conditions like nasal dermoid cysts necessitates a multidisciplinary approach involving specialists in audiology, speech pathology, otorhinolaryngology, dentistry, orthodontics, oral and maxillofacial surgery, and plastic and reconstructive surgery. This collaborative effort aims not only to address medical needs but also to improve facial aesthetics and overall quality of life for affected individuals. In this case, timely diagnosis and intervention were crucial in managing the nasal dermoid cyst, which posed risks such as local infection and potential intracranial complications. The use of advanced diagnostic imaging and surgical techniques, including the V-Y advancement glabellar flap for closure, exemplifies the comprehensive approach required for optimal outcomes in treating such complex cases. Further research and documentation of similar cases can contribute to a deeper understanding of the intersection between genetic syndromes like TCS and dermatological anomalies such as nasal dermoid cysts, ultimately enhancing clinical management strategies and patient care.

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REFERENCES

1. Farooqui A, Munde A, Mishra S, Farooqui M. Treacher collins syndrome. IP Int J Maxillofac Imaging. 2021;7(1):25-7.
2. Dixon J. Treacher Collins syndrome. J Med Genet. 1995;32(10):806–8.

3. Shetty B, Thomas A, Pidamale R. Treacher Collins Syndrome: A Case Report and a Brief Review on Diagnostic Aids. Int J Clin Pediatr Dentist. 2011;4(3):235–9.
4. Fernandez O, Ronis L. The Treacher-Collins Syndrome. Arch Otolaryngol 1964;80:505–20.
5. Chang C, Steinbacher M. Treacher collins syndrome. Seminars Plastic Surg. 2012;26(2):83–90.
6. Farkas G, Posnick C. Detailed morphometry of the nose in patients with Treacher Collins syndrome. Ann Plast Surg. 1989;22(3):211–9.
7. Andrews J, Kopacz AA, Hohman MH. Ear Microtia. In: StatPearls. StatPearls Publishing, Treasure Island (FL); 2023.
8. Wardinsky D, Pagon A, Kropp J, Hayden W, Clarren K. Nasal dermoid sinus cysts: Association with intracranial extension and multiple malformations; cleft palate craniofacial. 1991;28:87-95.
9. Scully C, Langdon J, Evans J. Marathon of eponyms: 20 Treacher Collins syndrome. Oral Dis. 2011;17(6):619–20.
10. Dixon MJ. Treacher Collins syndrome, Human Molecular Genetics. 1996;5(1):1391–3.
11. Franceschetti A, Klein D. The mandibulofacial dysostosis; a new hereditary syndrome. Acta Ophthalmol (Copenh). 1949;27(2):143–224.
12. Aguiar RS, Santos S. Síndrome de Treacher Collins. Rev Port Estomatol Cir Maxilofac. 1989;30(2):137–42.
13. Posnick C, Ruiz L. Treacher Collins syndrome: current evaluation, treatment and future directions. Cleft Palate Craniofac J. 2000;37(5):434.
14. Julapalli R, Cohen A, Hollier H, Metry W. Congenital, ill-defined, yellowish plaque: the nasal dermoid. Pediatr Dermatol. 2006;23(6):556-9.
15. New B, Erich B. Dermoid cysts of the head and neck. Surg Gynecol Obstet. 1937;65:48–55.
16. Nakajima K, Korekawa A, Nakano H, Sawamura D. Subcutaneous dermoid cysts on the eyebrow and neck. Pediatr Dermatol. 2019;36(6):999-1001.

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