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Treacher-Collins Syndrome: A case study

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Abstract

The combination of bilateral choanal atresia and Treacher Collins syndrome is a life-threatening neonatal emergency. We report the case of a female newborn with typical facial dysmorphia and immediate respiratory distress due to airway obstruction. Although urgent surgical correction was performed, the outcome was brain death. This case highlights the serious prognosis of severe neonatal forms and emphasises the critical importance of antenatal diagnosis and multidisciplinary care planned from the delivery room onwards.

Keywords: Polymalformative syndrome, Choanal atresia, Mandibular hypoplasia, Oto-mandibular dysostosis

Introduction

Treacher Collins syndrome (TCS) is a rare congenital anomaly that specifically affects the development of facial structures. Also known as mandibulo-facial dysostosis, this syndrome manifests as a set of malformations mainly affecting the bones and soft tissues of the craniofacial region.

From a genetic point of view, this condition is hereditary, following an autosomal dominant pattern. The prevalence of this syndrome in the general population is estimated at approximately one newborn in 50,000 live births, which classifies it as a rare disease [1,2].

We report a case of a newborn with Treacher Collins syndrome in order to shed light on the screening, diagnosis and management of this relatively rare condition.

Observation

This is a female newborn, born at term, admitted at 12 hours of life for further management of a polymalformative syndrome diagnosed in the delivery room.

The newborn was born to non-consanguineous parents. Delivery was vaginal, with poor adaptation to extrauterine life marked by immediate severe neonatal respiratory distress.

No antenatal diagnosis of a polymalformative syndrome was noted, nor were there any similar cases in the family. There was also no evidence of exposure to drugs or toxins during pregnancy.

The clinical examination performed on admission revealed marked facial dysmorphia, combining bilateral and symmetrical hypoplasia of the malar bones and mandible, complete atresia of the external auditory canals and poorly shaped auricles, folded in on themselves. The mouth opening was significantly limited. (Fig. 1)

Examination of the upper airways revealed bilateral nasal obstruction, confirmed by the inability to pass a probe, suggesting choanal atresia.

The general physical examination did not reveal any abnormalities of the limbs. The hips were stable, the anus was well positioned, and the vulvar cleft was normal in appearance. However, a small tuft of hair was noted on the sacrum, which could suggest a neural tube closure defect (Fig. 2).

In terms of management, the patient was immediately stabilised with appropriate ventilatory and haemodynamic support, while a thorough assessment for malformations was carried out. This confirmed the presence of bilateral choanal atresia and complete agenesis of the outer and middle ear on both sides. No significant abnormalities were detected in imaging of other organs, including the heart, kidneys and central nervous system. (Fig. 3) A standard karyotype was performed and revealed no detectable chromosomal abnormalities.

Faced with a respiratory emergency due to nasal obstruction, emergency surgery was decided upon to correct the choanal atresia. The operation proceeded without any notable intraoperative incidents, but the immediate aftermath was complicated by persistent respiratory instability despite optimal resuscitation treatment.

Given the persistence of episodes of hypoventilation and desaturation, a tracheotomy was performed to ensure effective and prolonged ventilation.

The clinical outcome was unfavourable. In the days following the operation, the patient's neurological condition progressively deteriorated. Brain stem reflexes were gradually lost, and the electroencephalogram performed in this context confirmed the total absence of electrical activity in the brain, indicating brain death.

Fig. 1: Image of the newborn showing the appearance of the craniofacial region

Fig. 2: Image of the newborn in the supine position illustrating the general appearance

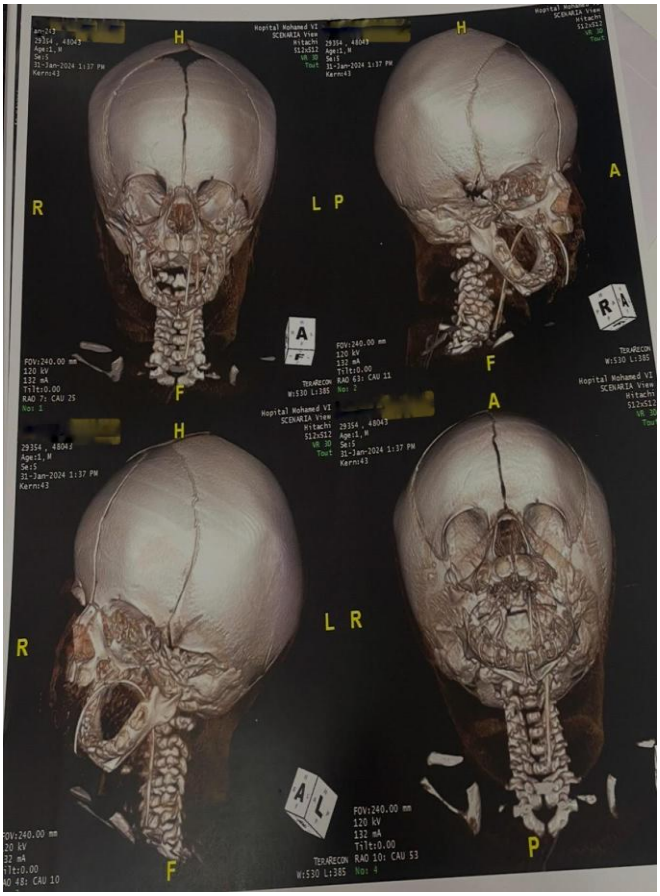

Fig. 3: Cranial CT scan (3D) showing the cranial vault and craniofacial structure in anterior, posterior and lateral views. The images show **premature fusion of the cranial sutures**, causing **deformation of the cranial vault** and associated **craniofacial asymmetry**, with no obvious bone abnormalities of the skull base on these reconstructions.

Discussion

Polymalformative syndromes constitute a large group of congenital disorders resulting from abnormalities in embryonic development, often of genetic, environmental or multifactorial origin. They reflect an early disruption of the morphogenetic process, most often occurring between the fourth and eighth week of gestation, a critical period of organogenesis [3,4]. Early recognition of these syndromic mains a major challenge, both for therapeutic management and for genetic counselling and prevention of familial recurrence [5].

In the case we report, the clinical presentation combines bilateral and symmetrical hypoplasia of the malar and mandibular bones, complete atresia of the external auditory canals, agenesis of the middle ear and bilateral atresia of the choanae. This cluster of abnormalities points to a disorder in the development of the first and second branchial arches, which play an essential role in the formation of the orofacial and auricular structures [6]. Any damage to these arches results in craniofacial malformations of varying severity, sometimes isolated, sometimes part of a broader polymalformative picture [7].

The differential diagnoses to consider mainly include syndromes belonging to the spectrum of oto-mandibular dysostoses, such as Treacher-Collins syndrome, Goldenhar syndrome (or hemifacial microsomia), and Nager syndrome. CHARGE syndrome could also be considered due to its association with choanal atresia, although it is usually accompanied by ocular (coloboma), cardiac, genital and internal ear abnormalities, which are absent in our observation [8,9].

The absence of major systemic abnormalities and the normal karyotype suggest a probable sporadic form, possibly linked to a de novo gene mutation affecting a craniofacial development gene. Studies have highlighted the involvement of the TCOF1 gene in Treacher-Collins syndrome, as well as POLR1C, POLR1D and EFTUD2, mutations of which cause similar phenotypes by altering ribosome biogenesis and neural crest cell proliferation [10].

Clinically, the combination of bilateral choanal atresia and middle ear agenesis has major functional consequences. In newborns, breathing is almost exclusively nasal; thus, bilateral choanal obstruction causes immediate respiratory distress at birth, requiring urgent intervention. Surgery in the first hours of life is often unavoidable, although it is associated with a high rate of respiratory and infectious complications. Difficult ventilation, tissue fragility and the coexistence of other anatomical abnormalities can considerably complicate postoperative management [11,12].

Despite early surgical correction, our patient's prognosis was poor, marked by persistent respiratory instability and a fatal outcome secondary to brain death. This outcome illustrates the serious prognosis of severe neonatal forms of oto-mandibular dysostosis, where anatomical abnormalities compromise ventilation, feeding and survival. Several studies have shown that neonatal mortality is closely correlated with the extent of upper airway malformations and the presence of associated brain or heart damage [13,14].

In terms of prognosis, survival depends on early diagnosis, the nature of the associated abnormalities and the resuscitation resources available. Moderate forms may benefit from a progressive surgical strategy and multidisciplinary care involving maxillofacial surgeons, ENT specialists, paediatricians, anaesthetists and geneticists. In severe forms,

such as the one described, the prognosis remains extremely poor despite recent advances in neonatal surgery and assisted ventilation [15].

This observation highlights the crucial importance of antenatal diagnosis, which would have made it possible to anticipate respiratory distress and organise appropriate care in the delivery room. Morphological ultrasound, supplemented by foetal MRI when facial abnormalities are suspected, can provide valuable diagnostic information. However, in many contexts, particularly in countries with limited resources, restricted access to these screening techniques is a major obstacle to early detection [16].

She highlights the complexity of craniofacial embryonic development and the diversity of syndromes that can result from it, emphasising the need for an integrated approach combining clinical, imaging, genetic and neonatal resuscitation techniques in order to improve the survival and quality of life of patients with these rare anomalies. It also emphasises the value of case studies in the medical literature, which, although limited by their isolated nature, contribute to enriching our understanding of atypical phenotypes and guiding future genetic investigations [10,17].

Conclusion

This case illustrates the extreme fragility of newborns with major craniofacial malformations. It serves as a reminder that, even with rapid and appropriate care, the prognosis can remain poor due to the severity of the structural damage and the difficulty of ensuring effective ventilation in the first few hours of life. The future development of more accurate prenatal diagnostic strategies and wider access to genetic sequencing techniques could improve the detection, management and genetic counselling of these rare syndromes [10,16,18].

References

1. Sadler TW. Langman’s Medical Embryology. 14th ed. Philadelphia: Wolters Kluwer; 2019.

2. Jones KL. Smith's Recognisable Patterns of Human Malformation. 7th ed. Philadelphia: Elsevier; 2013.

3. Trainor PA. Craniofacial birth defects: the role of neural crest cells in the aetiology and pathogenesis of Treacher Collins syndrome and the potential for prevention. Am J Med Genet A. 2010;152A (12):2984-2994.

4. Hunter AGW. Mandibulofacial dysostosis (Treacher Collins syndrome). In: Stevenson RE, Hall JG, editors. Human Malformations and Related Anomalies. 2nd ed. Oxford University Press; 2006.

5. Issekutz KA, Graham JM Jr, Prasad C, et al. CHARGE association: an update and review for the primary paediatrician. Clin Pediatr (Phila). 2005;44(7):549-560.

6. Maggiolini F, Bonati MT, Magnani C, et al. Phenotypic variability in Goldenhar syndrome: a case series. Eur J Med Genet. 2019;62(3):103573.

7. Trainor PA, Dixon J. Genetic and epigenetic mechanisms in craniofacial malformations. Nat Rev Genet. 2020;21(9):599-614.

8. Cohen MM Jr. Craniofacial malformations: problems of diagnosis and classification. Clin Plast Surg. 1983;10(3):421–434.

9. Vermaak WJH, van der Merwe W. Genetic mutations leading to vitamin B12 deficiency: A comprehensive review. Nutrients. 2017;9(12):1350. [https://doi.org/10.3390/nu9121350] (https://doi.org/10.3390/nu9121350)

10. Goyal S, Swain SK. Choanal atresia: a review of current concepts. Indian J Otolaryngol Head Neck Surg. 2017;69(2):185–193.

11. MacArthur CJ, Deschler DG. Choanal atresia repair in neonates: surgical techniques and outcomes. Otolaryngol Clin North Am. 2012;45(1):141-153.

12. Jones MC, et al. Neonatal mortality in craniofacial syndromes with airway obstruction: a retrospective cohort study. J Craniofac Surg. 2018;29(3):645-651.

13. Stoler JM, McDonald-McGinn DM, Zackai EH, et al. Prenatal diagnosis and management of craniofacial anomalies. Semin Perinatol. 2013;37(1):55-64.

14. Chitayat D, et al. Advances in prenatal diagnosis of facial anomalies. Prenat Diagn. 2017;37(11):1071-1083.

15. Mulliken JB, Taub PJ. Craniofacial malformations and syndromes. Plast Reconstr Surg. 2014;134(5):1227e-1247e.

16. Hunter AGW. Challenges in prenatal diagnosis of facial dysostoses. Prenat Diagn. 2015;35(10):987-995.

17. Trainor PA. Molecular mechanisms of Treacher Collins syndrome. Am J Med Genet C Semin Med Genet. 2010;154C (4):343–353.

18. Dixon J, Trainor P. Genetics of craniofacial development. Nat Rev Genet. 2020;21(9):599-614.