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Treacher Collins Syndrome: Diagnosis and Management of a Case

Dr. Zeine El Abidine Baba El Hassene[1*], Dr. Oussalem Amine[1], Pr. Dani Bouchra[1], Pr. Boulaadas Malik[1]

[1]Maxillo-Facial Surgery and Stomotology Department, University Hospital of Ibn Sina – Rabat – Morocco

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***Corresponding author:** Dr. Zeine El Abidine Baba El Hassene
Maxillo-Facial Surgery and Stomotology Department, University Hospital of Ibn Sina – Rabat – Morocco

Abstract | **Case Report**

Treacher Collins syndrome (TCS) is a genetic disorder resulting in a congenital craniofacial malformation. Patients typically present with oblique palpebral fissures, inferior palpebral colobomas, microtia, and malar and mandibular hypoplasia. This autosomal dominant disorder has variable phenotypic expression, and patients do not exhibit associated developmental delays or neurological disease. Management of these patients requires a multidisciplinary team from birth to adulthood. Appropriate planning, support, and surgical techniques are essential to optimize patient outcomes. We report a case of TCS treated in the maxillo-facial surgery departement of the university hospital of Ibn Sina of Rabat.

Keywords: Treacher Collins Syndrome, Facial Reconstruction, Canthopexy, Malar Augmentation, Iliac Bone Graft, Craniofacial Surgery, Zygomatic Hypoplasia.

INTRODUCTION

Treacher-Collins syndrome (TCS), also known as mandibulofacial dysostosis, is a complex malformation of craniofacial morphogenesis. It is an autosomal dominant hereditary disorder affecting 1 in 50,000 live births. [1, 2].

In most cases, it is caused by a mutation in the TCOF1 gene at loci 5q31.3, which encodes a nucleolar phosphoprotein essential for the development of the first and second branchial arches [3, 4].

Diagnosis is primarily clinical. Clinical manifestations include hypoplasia of the facial bones, particularly the mandible and zygomatic complex, palpebral fissures, coloboma of the lower eyelid, and cleft palate [5].

Phenotypic expressiveness and severity of manifestations vary from one individual to another, requiring multidisciplinary management [6].

Conductive deafness is present in 50% of cases, which is due to malformations of the external and internal ear. [7, 8]. This work aims to shed light on the screening and diagnosis of this relatively rare pathology, as well as its management, particularly surgical treatment.

CASE PRESENTATION

An 11-year-old male patient from a consanguineous marriage presented with a facial malformation since birth. Examination revealed downward tilt of the eyes, depressed zygomatic arches, sunken cheekbones, micrognathism, and coloboma of the lower eyelids. (Fig.1) A CT scan was performed, which revealed hypoplasia with discordant facial development.

Figure 1: Pre-operative aspect of TC syndrome

The patient underwent an iliac bone graft for cheekbone and orbital rim reconstruction with bilateral external canthopexy. (Fig.2)

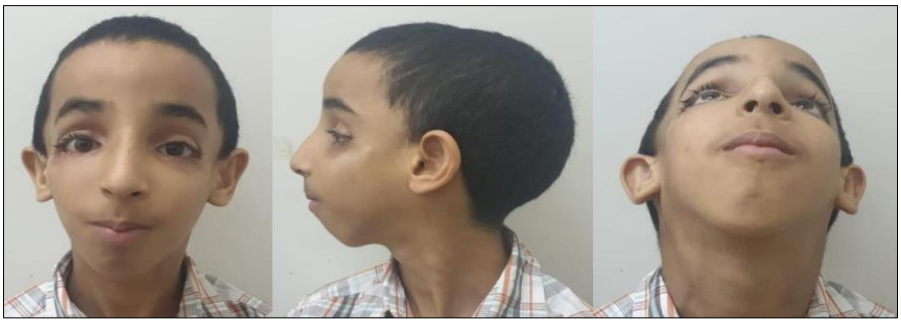

Figure 2: 1 month after surgery

DISCUSSION

Treacher Collins syndrome is a congenital disorder of craniofacial development. Clinical features include hypoplasia of facial bones, particularly the mandible (78%) and the zygomatic complex (81%), associated with ophthalmic anomalies (89%), which was observed in our case.

Franceschetti-Klein syndrome or TC syndrome or dysostosis-mandibulofacial is an autosomal dominant genetic syndrome characterized by the existence of an anomaly of craniofacial development [9, 10]. In only 40% of cases, the family history is positive and 60% of cases correspond to a de novo mutation [11]. The responsible gene is located on chromosome 5 at q31, 3q32 and was identified in 1996 [12]. This gene, TCOF, codes for the nucleolar phosphoprotein "Treacle" which is involved in the transcription of ribosomal genes [13, 14], whose function appears to be essential for the survival of cephalic neural crest cells [15]. It is transmitted in an autosomal dominant manner with a penetrance of 90% and variable expressivity, even in affected patients within the same family. More than 130 mutations have been identified to date, affecting different regions of the gene, without any correlation between the type of mutation and its phenotypic expression having been found [16]. These mutations can be insertions, deletions or nonsense mutations creating a premature stop codon [17]. Genotypic analysis of this family found a duplication in exon 25 of the TCOF1 gene not listed in the literature to date. It is clinically characterized by hypoplasia of the facial bones (malar and zygoma 81% and mandible 78% and temporal bone) associated with ear anomalies (hypoplasia of the auricles 77%, atresia of the external auditory canals 36%, anomaly of the ossicular chain) responsible for conductive deafness in 40% of cases.

It is associated with coloboma of the lower eyelids in 89% of cases, a downward and lateral obliquity of the palpebral fissures as well as a cleft palate in 28% of cases [18]. Facial malformations are bilateral and asymmetrical. In our case, the patient had hypoplasia of the facial bones, particularly of the mandible and zygomatic complex, associated with ophthalmic anomalies.

Respiratory difficulties can manifest early due to the narrowness of the upper airways [19], and require early placement of a tracheostomy [20, 21]. Intelligence is generally preserved. In addition to questioning and the search for evidence of a familial pathology, imaging, and in particular two-dimensional ultrasound, remains the most effective means of screening and diagnosis [19-26]. A comprehensive search for associated malformations must be systematic and allowed to find 20% of associated cardiac malformations in the Hsieh study [27]. The management is mainly functional and aesthetic, depending on the malformations present. In our case, the patient benefited from a harvest of the iliac bone and graft for the reconstruction of the cheekbones and the orbital rim with a bilateral external canthopexy.

CONCLUSION

Treacher Collins syndrome is a malformation complex of craniofacial morphogenesis of autosomal dominant transmission in 40% of cases, whose phenotypic expressivity and severity of manifestations vary from one person to another, requiring multidisciplinary management.

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