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Orbital volume and shape in Treacher Collins syndrome

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ABSTRACT

Orbito-palpebral reconstruction is a challenge in Treacher Collins syndrome (TCS). This study investigates orbital phenotypes in TCS using cephalometry and orbital shape analysis.

Eighteen TCS and 52 control patients were included in this study, using the Dr Warehouse database. Orbital cephalometry was based on 20 landmarks, 10 planes, 16 angles, and 22 distances. Orbits were segmented. Registration-based, age-specific mean models were generated using semi-automatic segmentation, and aligned and compared using color-coded distance maps – mean absolute distance (MAD), Hausdorff distance (HD), and Dice similarity coefficient (DSC). Symmetry was assessed by mirroring and DSC computing.

Central orbital depth (COD) and medial orbital depth (MOD) allowed 100% of orbits to be classified. COD and lateral orbital depth (LOD) were different from the controls. Average MAD between TCS and controls was $\leq$1.5 mm, while for HD it was >1.5 mm, and for DSC <1. TCS orbits were more asymmetrical than controls, and orbital volumes were smaller when age was considered as a confounding factor, and had a trend for normalization with age.

This report emphasizes the importance of combining different morphometric approaches in the phenotype characterization of non-trivial structures such as the orbit, and supports composite skeletal and soft-tissue strategies for the management of the peri-orbital region.

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1. Introduction

Treacher Collins syndrome (TCS) (Hennekam et al., 2010) is a mandibulofacial dysostosis affecting 1/50,000 to 1/35,000 births, that is, around 16 new cases per year in a large European country such as France. TCS is an autosomic dominant syndrome due to mutations in *TCOF1* (5q32–q33.1), a gene coding for a nucleolar protein called TREACLE.

TCS is generally referred to as a first-arch syndrome, which causes facial deformity, including bilateral hypoplasia of the midface (maxilla and zygoma) and the mandible, affecting both soft tissues and bones (Fig. 1). Patients with TCS also present with external ear anomalies and orbito-palbebral malformations (lower eyelid coloboma, ectropion, absence of eyelashes on the lower eyelid and downslanting palpebral fissures) (Plomp et al., 2013).

Lineage marking studies in the chick and the mouse have shown that all the bones forming the orbit are neural crest derived; furthermore, *TCOF1* plays a key role in the survival and the proliferation of neural crest cells (Dixon et al., 2006). Orbital malformations are thus a prime component of TCS malformations, and a

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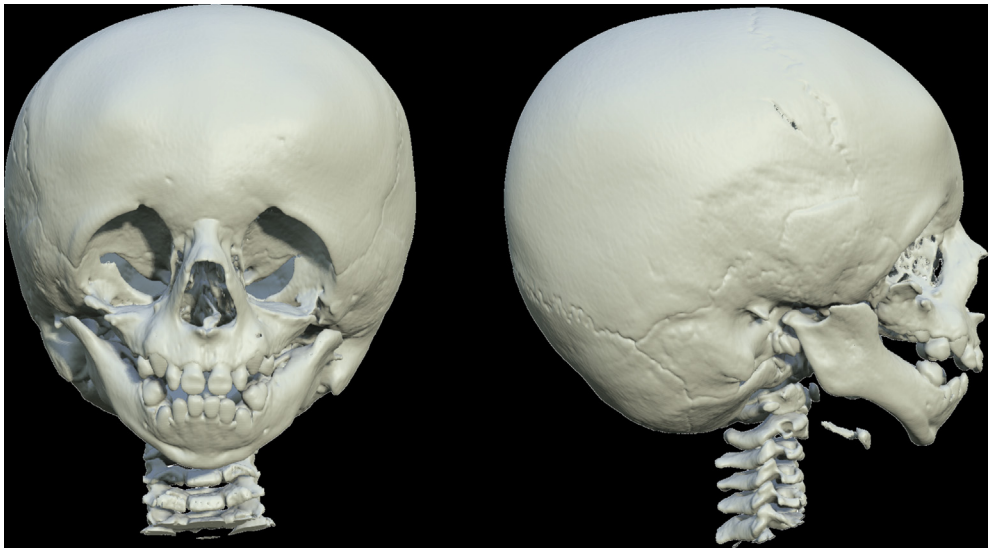

Fig. 1. 3D reconstruction of a typical case of Treacher Collins syndrome at 10 years of age, showing the bilateral orbito-zygomatic dysplasia.

major concern in the study and management of this condition, providing reasons for functional disadvantage and appearance differences, and a model for molecular developmental mechanisms. There is correlation between the range of deficiency in *TCOF*-mediated human skeletal growth, the severity of the soft tissue envelope phenotype, and the surgical burden that results (Ibrahim et al., 2016).

Bone reconstruction in TCS allows us to obtain a satisfactory projection of the lower orbital rim and of the zygoma (Nikkhah et al., 2013; Herlin et al., 2013). Nevertheless, the management of peri-orbital soft tissues is still a challenge (Plomp et al., 2013; Franchi et al., 2015). A better understanding of the orbital phenotype in this syndrome can therefore help the understanding of the mechanisms of this disease, and improve its management.

Here we investigate the orbital phenotype in TCS using cephalometric methods involving the description of the orbital bones and 3D shape analyses focused on the inner mold of the orbital cavity, based on a previously reported framework (Khonsari et al., 2013, 2016).

2. Materials and methods

2.1. Clinical data

This retrospective study was performed according to the local ethical regulations. The French national database for rare craniofacial disorders (CEMARA database, Centre des Maladies Rares) and the Dr Warehouse database (Garcelon et al., 2017) contained data from 56 patients with Treacher Collins syndrome (TCS). The most prevalent terms within the patient files were listed according to their frequency and to TF-IDF (term frequency–inverse document frequency), in order to quantify the occurrences of orbito-palpebral-related terms in the files of the 56 TCS patients. Amongst these 56 patients, 18 patients (36 orbits, mean age 7.85 ± 7.56) had positive *TCOF1* mutation screening and pre-operative head and neck CT-scans. For the cephalometric analysis, 17 controls were selected (34 orbits, mean age 6.01 ± 2.21) with no craniofacial anomalies (patients admitted for minor craniofacial trauma or headaches with normal CT-scans). For the 3D shape analysis and volume measurements, we considered a subgroup of 13 patients from the previous TCS group (26 orbits, mean age 6.85 ± 6.91) divided into two age groups (Aspert et al., 2002): 0–5 years, 6 patients (12 orbits, mean age 1.32 ± 1.0); and (Chong et al., 2008) 5–19 years, 7 patients (14 orbits, mean age 13.5 ± 5.8). We compared these 26 orbits with 49 control patients (98 orbits, mean age 2.43 ± 3.19) with no craniofacial anomalies (patients admitted for minor craniofacial trauma or headaches with normal CT-scans) subdivided into similar age groups (Aspert et al., 2002): 0–5 years, 40 patients (80 orbits, mean age 1.24 ± 1.46); and (Chong et al., 2008) 5–19 years, 9 patients (18 orbits, mean age 8.7 ± 3.0).

2.2. Cephalometric analysis

The landmarking and measurements were performed using Maxilim (Medicim, Leuwen, Belgium) (Olszewski et al., 2008, 2010; Lee et al., 2014). CT-scans were segmented and reconstructed in 3D by thresholding, using the built-in tools of the Maxilim software. We defined 16 3D anatomical landmarks and four computed landmarks in the orbital area, the upper airways, the dental arches, and the skull base, based on previous studies (Khonsari et al., 2013, 2016), and used these landmarks in order to define 10 planes, 16 angles, and 22 distances corresponding to anatomically relevant orbital parameters (Fig. 2). Further details about the landmarks and the parameters used in the cephalometric analysis are given in Supplementary Tables 1–4.

2.3. Shape analysis

We segmented all orbits using a semi-automatic segmentation technique in which the operator 'paints' or 'sculpts' the soft-tissue content of the orbit with a volumetric brush tool (Nysjö, 2016). The mean orbital size and shape models for TCS and controls were automatically generated from the segmented orbits through registration-based landmarking (Frangi et al., 2002). The method has been described in detail in a previous study (Khonsari et al., 2016). The orbital volume was estimated based on individual orbital segmentations (Fig. 3).

We compared the mean and individual TCS orbits versus mean control orbits by aligning them and then computing and visualizing the surface-to-surface distance and spatial overlap. The surface-to-surface distance and spatial overlaps between the aligned orbits were visualized as color-coded distance maps and semi-transparent overlays. Quantification of shape discrepancy was obtained by computing the mean absolute distance (MAD), the

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