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Computer-assisted midface reconstruction in Treacher Collins syndrome part 1: Skeletal reconstruction

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ABSTRACT

Introduction: Treacher Collins syndrome (TCS) is a severe and complex craniofacial malformation affecting the facial skeleton and soft tissues. The palate as well as the external and middle ear are also affected, but his prognosis is mainly related to neonatal airway management. Methods of zygomatico-orbital reconstruction are numerous and currently use primarily autologous bone, lyophilized cartilage, alloplastic implants, or even free flaps. This work developed a reliable "customized" method of zygomatico-orbital bony reconstruction using a generic reference model tailored to each patient.
Methods: From a standard computed tomography (CT) acquisition, we studied qualitatively and quantitatively the skeleton of four individuals with TCS whose age was between 6 and 20 years. In parallel, we studied 40 controls at the same age to obtain a morphometric database of reference. Surgical simulation was carried out using validated software used in craniofacial surgery.
Results: The zygomatic hypoplasia was very important quantitatively and morphologically in all TCS individuals. Orbital involvement was mainly morphological, with volumes comparable to the controls of the same age. The control database was used to create three-dimensional computer models to be used in the manufacture of cutting guides for autologous cranial bone grafts or alloplastic implants perfectly adapted to each patient's morphology. Presurgical simulation was also used to fabricate custom positioning guides permitting a simple and reliable surgical procedure.
Conclusions: The use of a virtual database allowed us to design a reliable and reproducible skeletal reconstruction method for this rare and complex syndrome. The use of presurgical simulation tools seem essential in this type of craniofacial malformation to increase the reliability of these uncommon and complex surgical procedures, and to ensure stable results over time.

1. Introduction

Treacher Collins syndrome (TCS) is a rare and severe craniofacial malformation without limb deformity (OMIM #154500). Its annual incidence varies between 1/35,000 to 1/50,000 live births (Fazen et al., 1967; Gorlin, 1990). TCS is the result of a mutation in the TCOF1 gene (5q32-q33.1), which encodes a nucleolar phosphoprotein called Treacle. It has autosomal dominant transmission with a penetrance nearly 90% with viable expressivity, even in affected patients of the same family (Dixon et al., 2007; Trainor et al., 2009). Numerous other genes, including a recessive one, have also been identified as being responsible for TCS, reopening the debate about its expressivity (Dauwerse et al., 2011).

In TCS zygomatico-orbital bony involvement is present in about 90% of the cases. It is always bilateral, and usually symmetrical (Rogers, 1964; Teber et al., 2004). Principally manifesting itself as hypoplasia or aplasia of the zygomas (Fuente del Campo et al., 1994; Gundlach and Pfeifer, 1991; Bigorre, 2001).

The skeletal reconstruction of the TCS patient continues to be a subject of debate in the literature. Even with multiple techniques available, most surgeons have unsatisfactory results and significant donor site morbidity (Gillies and Millard, 1957; Obwegeser and Bertschinger, 1974). The bony reconstruction forms the foundation for appropriate soft tissue reconstruction. This is especially true in

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cases with significant lateral canthal dystopia. It is thus interesting to determine which part of the skeletal or soft tissue reconstruction affects the final result.

During the first part of our work, we sought a reliable and accurate reconstruction technique for the zygomatic bones, without an in vivo model of symmetry as there are in unilateral anomalies. From the tomodensitometric study of four typical TCS subjects, a computer-aided custom technique was developed to reconstruct the zygomas in ideal proportions and position. This method is applicable for the two principal techniques actually used: the calvarial bone graft, which remains the method of choice for most authors (Tessier et al., 2005a,b; Neovius and Engstrand, 2010), and the alloplastic implant, in which interest has been revived due to recent advances in tissue engineering (Sailer and Farmand, 1991; Wei et al., 2004; Yaremchuk, 2008; Taylor, 2010).

2. Materials and methods

2.1. Skeletal volumetric analysis and realization of a volumetric database

From 12 patients with TCS 4 were chosen for a tomodensitometric study. The patient's age at the time of the study was of 6, 10, 14 and 20 years respectively (average 12.5 years). Patient selection was based on the type and severity of their deformity and their age, which covered the ages used by different teams to perform corrective surgery of the zygomatico-orbital skeleton.

To draw conclusions on the data acquired from computed tomography (CT) and realize a volumetric database, 10 controls per age category were studied. The data came from 40 patients (80 "healthy" control sides) who received a craniofacial CT for problems not related to the facial skeleton.

A single examiner performed the data collection and analysis. All CT's were realized by the same scanner (General Electric® Inc. – LightSpeedVCT, 64 b, 140 kV, 800 mA). The image transfer was done on an Advantage Window®. Console. The slice thickness was of 625 μm with a cut interval of 200 μm. Acquisition parameters (kV, mA, X-ray exposure times, tube rotation speed, table advancement, number of detectors) were monitored and recorded.

Post-image processing was performed with MYRIAN® expert v. 1.5.2 software (Intrasense SAS, Montpellier, France). The first step of bony segmentation was carried out automatically. After manual confirmation, the listed geometric parameters were determined: the zygomatic bone volume and the orbital volume, in both TCS patients and control patients (Fig. 1). Bone volume was calculated from the size of a pixel, deduced from the acquisition parameters (field of view, matrix), and from the slice thickness itself. With reliability close to 95%, this approach is superior to the immersion method (Canovas et al., 2004; Bonnel et al., 2007; Largey et al., 2007).

2.2. Presurgical simulation and computer-assisted surgery

The simulation was performed using the Surgicase CMF® software (Materialise®, NV Leuven, Belgium).

Using the CT scan of a patient with a severe form of TCS, complete custom surgical planning was performed.

The simulation consisted of four steps, which are illustrated in the results section:

1. Extraction of a three-dimensional (3D) image of the patient.
2. Positioning of the "ideal" zygoma on the patient's skeleton.
3. Transformation of the "ideal" zygoma into a 3D phantom image to create an adaptable cutting guide or an implant using rapid prototyping.
4. Bone graft or custom implant placement using a positioning guide.

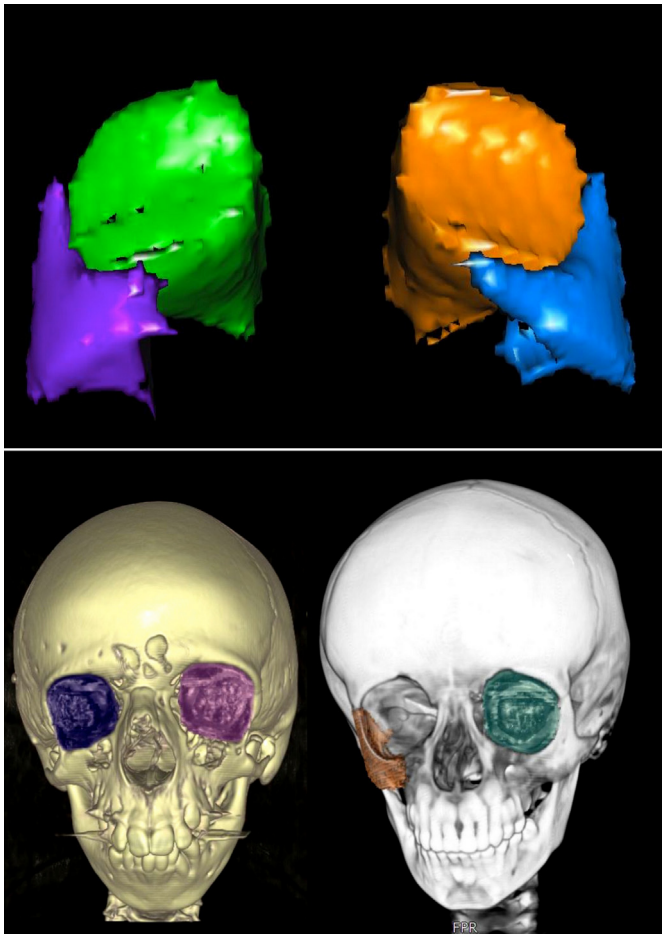

Fig. 1. Volumetric representation of the studied volumes on CT in TCS patients and control patients. Orbits and zygomatic bones.

2.3. Statistics

All statistics were computed with the SPSS Statistics 17.0 software (version 17.0.0, SPSS Inc, Chicago, Il). Appropriate bivariate and multivariate statistics were calculated, and the level of statistical significance was set at $P < 0.05$.

3. Results

3.1. Skeletal volumetric analysis and realization of a volumetric database

Orbital volumes are shown in Table 1. Excellent correlation was found between the right and left orbital volumes (from 93 to 99% depending on the age). The volumes were slightly higher in control patients (Fig. 2). Volumes of control patients were somewhat lower but comparable to those reported by Bentley et al. (2002) and by Chau et al. (2004).

Zygomatic bone volumes are shown in Table 2. A lesser correlation was found between the right and left volumes (from 83 to 98% depending on the age). TCS patients had significantly lower zygomatic bone volumes compared to the controls (Fig. 3, all $P < 0.001$). Zygomatic growth had a linear progression in controls. Significant hypoplasia was found in TCS patients, with volumes averaging 0.2cm^3.