Commentary Facial variation: from visual assessment to three-dimensional quantification

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SUMMARY The extensive study presented by Toma and coworkers is one of the first presenting normative three-dimensional soft tissue face data derived from a large group of 15-year-old Caucasians using laser surface scanning. This data is not only important to quantify facial variation in a normal population but can help to analyse facial dysmorphology. The study methodology and statistical handling of the data provide a good basis for future studies into facial variation.

Introduction

There is a long history of collecting two-dimensional data that describe craniofacial morphology and growth. Those data have been collected over a period of nearly 80 years since the method of obtaining standardized cephalometric head films was introduced, which turned out to be an unprecedented step forwards in the assessment of growth of the head and face. However, there is a still raging discussion whether such a two-dimensional representation of a three-dimensional (3D) object like the face gives us the true picture of what we need to know. The rapidly developing 3D imaging techniques seem to give the clinician and the researcher a more accurate representation of facial morphology than is possible with visual inspection only.

Recent developments

The soft tissues of the face (skin, connective tissues, fat, and muscles), the facial skeleton, and the dentition are the three aspects of the face that need to be addressed when attempting to quantify the face. Three-dimensional imaging techniques that are available now include multislice computed tomography, cone-beam computed tomography, magnetic resonance imaging, 3D stereophotogrammetry, laser surface scanning, and digital dental models. Most of these techniques image only one of the three aspects of the face optimally. Therefore, image fusion utilizing more than one 3D imaging data set has the potential to provide an accurate virtual model of a patient's head in the near future (Plooij *et al.*, 2011). As we have not arrived at this point yet, the single use of a non-invasive high-quality 3D imaging system such as laser surface scanning as used in the study

of Toma and coworkers (2012) is to be preferred. It enables the creation of large cross-sectional data sets for normative populations, the quantitative assessment of longitudinal facial changes, and clinical audit of orthodontic and surgical treatment outcomes in the head and neck region.

Laser surface scanners belong to the earlier devices on the market and have proven to be an accurate and reliable technique to visualize and quantify soft tissue facial morphology in the different planes of space. The latest systems on the market are safe for the eyes as well. Another, nowadays, widely spread technique to reproduce the surface geometry of the face is 3D stereophotogrammetry (Heike *et al.*, 2010). The advantage of the latter technique over laser surface scanning is that it is possible to map realistic colour and texture data onto the geometric shape, which results in a realistic life-like 3D rendered photograph. Another advantage, especially in young children, is the short acquisition time, which reduces the risk of movement artifacts. It is to be expected that laser surface scanning of the human face will be gradually superseded by 3D stereophotogrammetry.

Why is the topic important?

Three-dimensional imaging has the advantage to enable facial evaluation from any view point. For medical purposes, the measurement and characterization of the human face in three dimensions is fundamental to the objective analysis of facial variation and facial deformity (Toma *et al.*, 2008). So far, much research in the medical field using 3D imaging has focused on the latter aiming at phenotyping individuals based on dysmorphic facial features (Hammond, 2012). Although this seems obvious

for major visible malformations such as cleft lip and palate, it has to be realized that many syndromes and malformations are not that straight forward to recognize, even for experienced clinical geneticists. Slight variations in craniofacial morphology or a combination of minor distinct features in the face are far more difficult to detect but may be decisive in a syndrome diagnosis. Here comes the importance of large collections of 3D facial images into the picture to quantify the similarity between a subject and an average of a certain group, either a selected dysmorphic group or a population-based sample. To this purpose, statistical tools need to be developed to quantify the degree of similarity, which should also take into consideration the normal variation in a population. In their study, Toma et al. (2012) made clever use of the existing technique of principal component analysis to identify key components of facial variation. Of course, this study is limited to a certain population group; in this case, a British Caucasian population of 2514 girls and 2233 boys of 15.5 year old, but this cohort was recruited from a very well-designed longitudinal study, which provided a unique opportunity to perform this study into facial variation.

Data collection

Nowadays, descriptive studies are not the sexiest to perform. Editors assign scientific priority to novel original research reports rather than largely cross-sectional or longitudinal studies. According to present medical ethical standards regarding research involving human subjects, the earlier studies using roentgen cephalometry will never be repeated and longitudinal studies using cone-beam computer tomography will never be performed. In contrast, large databases of non-invasive 3D records could be set up. Hammond and Suttie (2012) give an overview of known databases with 3D facial imaging data sets on Caucasians and non-Caucasian populations. Two large-scale initiatives are the Facabase Consortium and the ALSPAC study.

The FaceBase Consortium (www.facebase.org) that consisted originally of 11 projects in the domain of craniofacial genetic research has established a web-based resource that serves as a central repository for data and bioinformatics tools generated by the FaceBase Consortium, as well as data offered for inclusion by other investigators working on similar projects. Both the FaceBase database and the consortium are funded by the National Institute of Dental and Craniofacial Research of the National Institutes of Health of the United States. As a result of one of the FaceBase projects, a 3D human facial norms database has been constructed with DNA samples of 3500 healthy Caucasian individuals between 3 and 40 years.

The UK-based large-scale ALSPAC study (Avon Longitudinal Study of Parents and Children) also called 'Children of the 90s' (http://www.bristol.ac.uk/alspac/) of which the data were derived for the current study comprises comprehensive data of approximately 14,541 pregnancies and their outcome. Expecting mothers were enrolled during their pregnancy in 1991 and 1992, and their children have been followed since then. The study includes an assessment of laser scans of the faces of 4747 British Caucasian school children at the age of 15.5, which can serve as a normative database.

It is to be expected that more collections of different populations will follow within due time. Collaborative efforts will focus on accessibility of those data for research purposes.

Conclusion

The data presented by Toma and coworkers is not only important to quantify facial variation in a normal population but can help to analyse facial dysmorphology and understand genotype/phenotype associations. This seems to be just the beginning of a meaningful use of data derived from novel non-invasive imaging techniques. The study methodology and statistical handling of the data provide a good basis for future studies into facial variation.

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