Facial image analysis to detect gestational alcohol exposure

Large-scale screening and surveillance would identify communities most at risk of fetal alcohol syndrome in South Africa.

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An objective, quantitative approach to identifying the facial dysmorphology associated with fetal alcohol syndrome (FAS) would assist screening and surveillance efforts. Such an approach is provided by digital imaging, which is an active area of research for the detection of facial dysmorphology. This paper reviews the use of digital imaging and shape analysis to assess the characteristic FAS facial appearance.

Fetal alcohol syndrome (FAS) is the most severe form of a broad range of structural anomalies and neurocognitive and behavioural disabilities, known as fetal alcohol spectrum disorder (FASD), resulting from the exposure of the developing fetus to alcohol. The highest prevalences of FAS and FASD worldwide have been reported among first-grade children in South Africa. In a wine-growing region in the Western Cape, FAS prevalence has grown from 65.2 - 74.2 per 1 000 children reported in a 2005 study[1] to 68.0 - 89.2 per 1 000 in a 2007 study.[2] A study conducted in the Northern Cape during the period 2001 - 2004 revealed rates of the most severe FASD categories (FAS and partial FAS) of 93.2 - 149.9 and 61.0 - 90.3 per 1 000 children in De Aar and Upington, respectively.[3]

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A simple, cost-effective method for large-scale FAS and FASD screening and surveillance would allow the identification of those communities most at risk and the implementation of suitable prevention and intervention programmes. Interventions may be aimed at the community to increase awareness of the dangers of alcohol use during pregnancy, or selectively at high-risk groups. Educational programmes may be introduced to address the learning needs of affected children. Improved language and literacy outcomes[4] as well as improved mathematics performance and behaviour[5] have been reported from early interventions for children with FAS. An understanding of the patterns of FASD occurrence may lead to understanding and prevention of the maternal risk factors that combine with alcohol abuse to play a role in the teratogenesis that leads to FASD.

Diagnosis of FASD depends on evidence of growth retardation, neurodevelopmental abnormalities and a characteristic facial dysmorphology.[6,7] The term dysmorphology refers to a set of human growth and structural defects; a recognisable pattern of dysmorphic signs constitutes a syndrome.[8]

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Studies aimed at objective identification of the facial dysmorphology associated with gestational alcohol exposure have primarily concentrated on children diagnosed with full-blown FAS, rather than the less severe categories of FASD. The facial phenotype is the only aspect of the syndrome that is considered specific to FAS;[6] short palpebral fissure length, smooth philtrum and midface hypoplasia are key components of the FAS diagnostic criteria. The palpebral fissure length measurement and an example of a smooth philtrum are shown in Fig. 1.

Traditionally, the FAS facial features were identified using the gestalt method, which considers the general appearance of the face, and can, when used by trained professionals, be sufficiently accurate and reproducible. However, diagnosis by medical professionals not trained in dysmorphology may lead to inaccuracies which would hamper screening and surveillance efforts.[6] Thus a more objective, quantitative approach to identifying the FAS facial phenotype has been the research goal of several groups over the past decade.[9] Digital imaging to detect facial dysmorphology is an active area of research, and has also been adopted in FAS studies.

Anthropometry

The term anthropometry refers to measurement of any aspect of the human form. Anthropometry provides an objective method of assessing facial shape, in order to diagnose genetic and acquired malformations, to plan and evaluate surgery, to study normal and abnormal growth and to differentiate between the results of treatment and normal growth.[10] Anthropometry for the assessment of facial features in dysmorphic patients was first used for facial clefts and now provides a quantitative platform for pattern recognition and syndrome

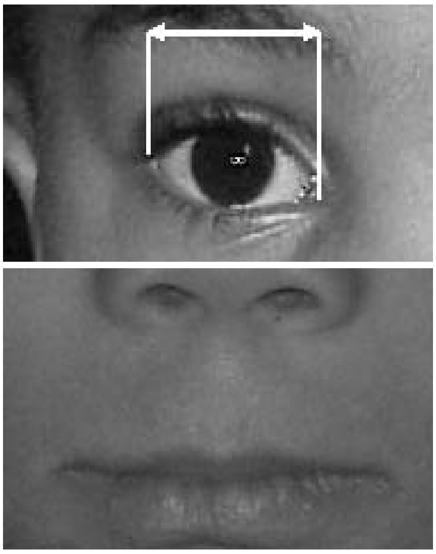

Fig. 1. Top: measurement of the palpebral fissure length. Bottom: example of a smooth philtrum.

diagnosis.[11] Anthropometric studies are based on biological homology, i.e. spatial correspondence between distinct points or landmarks on anatomical structures across individuals, and geometric variation in the relative location or pattern of these points.[12] The extent of deviation of an individual's pattern from the norm is determined by quantitative comparison to reference values collected from normal individuals. Syndrome diagnosis relies on the definition of characteristic abnormal patterns associated with a given syndrome. Typically for FAS, linear facial distances such as the palpebral fissure length are measured for comparison to reference values. Qualitative determinations are made for some features, for example, in assessing the smoothness of the philtrum. Multivariate statistical analysis has been used to determine those features that best describe the FAS facial phenotype. For example, Astley and Clarren[13] reported reduced palpebral fissure length/inner canthal distance ratio, smooth philtrum, and smooth upper lip, as features that distinguish individuals with FAS from controls with 100% accuracy. However, the clusters of facial features with which FAS is most accurately classified differs across ethnicities.[14]

Photogrammetry

Photogrammetry is the process of obtaining measurements by means of photographs. Indirect anthropometric methods such as photogrammetry have several advantages over direct anthropometry.[10] Contact of the anthropometric instruments during direct measurement may deform the facial surface and lead to inaccuracies. The period of interaction with the patient is potentially shorter for indirect measurements, since measurement takes place after data acquisition. Indirect methods are consequently less dependent on patient behaviour and the need for the patient to keep still for long periods. Some direct measurements, such as those around the eyes, pose a risk of discomfort or injury to the patient.

Facial measurement from images as an alternative to direct measurement in anthropometry, which usually relies on instruments such as rulers or callipers has been introduced in both 2D and 3D applications. Measurement from single 2D photographs may be inadequate for a 3D object such as the face. Stereo-photogrammetry makes use of multiple (or stereo) images obtained of an object within a calibrated space to produce 3D information on the object. The 3D information may take the form of landmarks or facial surfaces (landmark-based and surface-based stereo-photogrammetry, respectively). Another 3D facial surface imaging modality commonly used in facial anthropometry is laser scanning, which typically sweeps a narrow laser beam across the subject's face. Digital cameras capture the sweeping, and depth information is obtained from the captured locations of the reflected laser beam.

Facial shape analysis

The standard use of multivariate statistical procedures to assess the facial dysmorphology of FAS based on directly measured inter-landmark distances, has been extended to distances derived from both landmark-based and surface-based stereo-photogrammetry. Such studies, however, fail to capture the complete spatial configuration of a set of landmarks.[9]

Geometric morphometric or statistical shape analysis methods retain the geometric arrangement of landmarks and thus are able to provide a comprehensive description of facial shape. Facial shapes, defined by landmarks or surfaces, are aligned to remove the effects of rotation, translation and scaling, so that the distances between corresponding landmarks are minimised. Average shapes for groups can then be derived and compared, e.g. the average FAS facial shape can be compared with the average control facial shape. Fig. 2 shows landmarks defining features typically affected in individuals with FAS.[15] Landmark-based shape analysis has been used to assess the FAS facial dysmorphology in South African cohorts, broadly confirming the FAS gestalt reported in the literature, and also confirming the notion the FAS facial phenotype diminishes with age.[15,16] Mutsvangwa *et al.*[15] were able to classify facial shapes of FAS and control subjects with an accuracy of 95% for 5-year-olds and 80% for 12-year-olds, indicating potential for the use of facial shape alone in a screening tool for FAS (Fig. 3).

The facial shapes in Fig. 3 show a scaled version of the shape differences between the

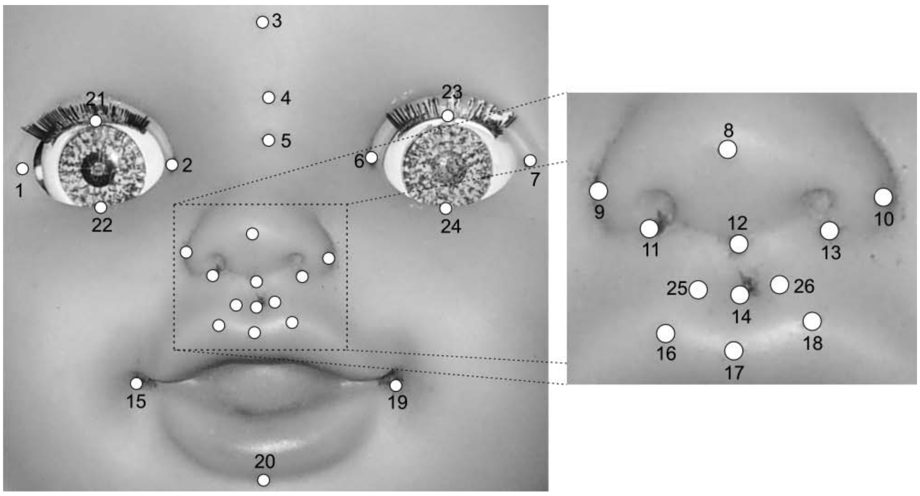

Fig. 2. Landmarks defining features affected in FAS[15]: 1 = right exocanthion, 2 = right endocanthion, 3 = glabella, 4 = n-nasion, 5 = ellion, 6 = left endocanthion, 7 = left exocanthion, 8 = pronasale, 9 = right alare, 10 = left alare, 11 = right subalare, 12 = subnasale, 13 = left subalare, 14 = centre of philtrum furrow, 15 = right cheilion, 16 = right crista philtri, 17 = labiale superius, 18 = left crista philtri, 19 = left cheilion, 20 = labiale inferius, 21 = right upper mid eye ridge, 22 = right lower mid eye ridge, 23 = left upper mid eye ridge, 24 = left lower mid eye ridge, 25 = mid right philtrum ridge and 26 = mid left philtrum ridge.

normal mean shape in the sample and the FAS mean shape. For 5-year-olds, the coronal view shows smaller palpebral fissure lengths compared with the control mean. The glabella is more superiorly positioned in the FAS group. The sagittal plane shows substantial ventro-dorsal retraction of the pronasale and glabella in the FAS mean shape. In the coronal view, the upper vermillion border of the FAS group is thinner and the philtrum is longer. The FAS lower lip is thinner than the control lower lip.

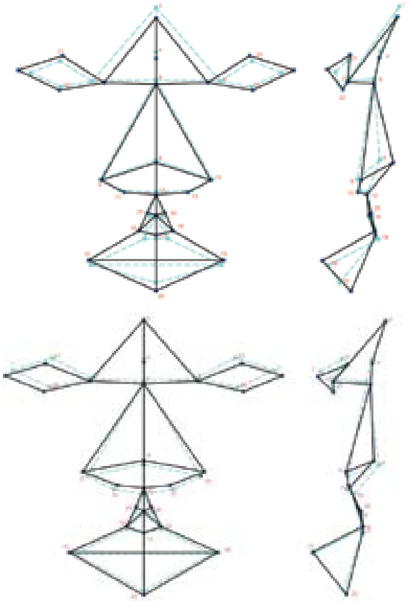

Fig. 3. Shape differences between FAS and normal children, defined by the landmarks shown in Fig. 2.[15] 5-year-olds (top) and 12-year-olds (bottom). The solid line is the normal mean shape, and the dashed line is the FAS mean shape. The diagrams on the left show the coronal view, while those on the right show the sagittal view.

Reduced palpebral fissure length is also evident in the 12-year-old FAS group. The eyes slant upwards and the upper lip is slightly thinner in the FAS group. In the sagittal plane, the nose is upturned for control individuals, in contrast with the results for 5-year-olds.

Klingenberg *et al.*[17] have used-surface based facial shape analysis to study directional asymmetry, defined as systematic differences between the left and right sides of the body, associated with prenatal alcohol exposure. While subtle facial directional asymmetry is present in healthy individuals, the faces of individuals with FAS from both Finland and South Africa were found to display greater directional asymmetry than those of control individuals. The asymmetry was dominated by a shift of the midline landmarks to the right and of the eyes to the left of the face, as shown in Fig. 4. Images were obtained using a laser scanner. Considering the subtlety of differences between FAS and control groups, the authors concluded that facial asymmetry was unlikely to become a practical diagnostic tool for FAS, but suggested that it might provide information that could complement other measures.

Conclusion

Facial anthropometry has evolved along a trajectory that has been strongly influenced by the development of computing and imaging technologies: direct manual facial measurements, measurements from 2D photographs, 3D measurements from stereo photographs, measurement from 3D surfaces, and finally the analysis of 3D shape data.

A major limitation of these approaches is that the FAS subjects studied are diagnosed using, among other criteria, the facial features described in the literature. Confirmation of the expected facial anomalies is therefore not surprising, although the approaches have contributed to defining the FAS gestalt, quantifying the deviation from the norm and refining the diagnostic criteria for FAS.[9]

Morphometric methods applied to image data enable quantitative analysis of facial shapes and have the potential to reveal as yet unidentified aspects of the FAS dysmorphology not represented in individual linear distances or their combination. They hold promise for delineation of more subtle facial anomalies that may be associated with less severe manifestations of FASD.

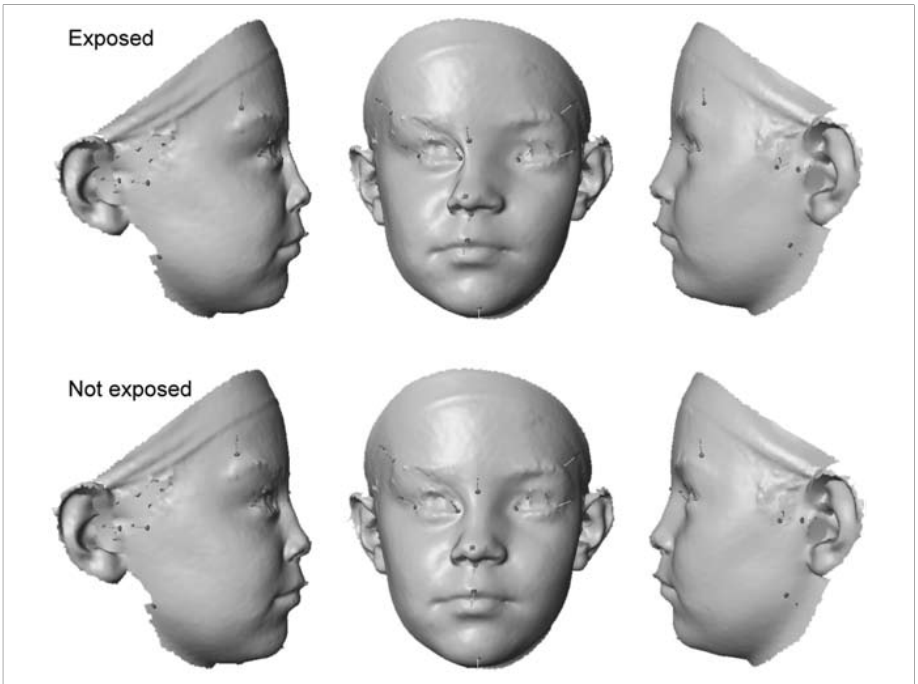

Fig. 4. Directional asymmetry in alcohol-exposed and control subjects from a South African coloured population; for better visualisation, the observed asymmetry has been exaggerated 5-fold. The facial surfaces were generated by warping a single facial image according to the landmark positions (shown on the images) for the mean asymmetries for each group. Reprinted from Klingenberg et al.,[17] with permission from Elsevier.

References available at www.cmej.org.za

In a nutshell

- Fetal alcohol spectrum disorder (FASD) refers to a broad range of structural anomalies and neurocognitive and behavioural disabilities.
- Fetal alcohol syndrome (FAS) is the most severe form of FASD.
- Diagnosis of FASD and FAS depends on evidence of growth retardation, neurodevelopmental abnormalities and a characteristic facial dysmorphology.
- FAS is characterised by short palpebral fissure length, smooth philtrum and midface hypoplasia.
- Digital imaging to detect facial dysmorphology has been used in FAS studies.
- Statistical shape analysis methods retain the geometric configuration of landmarks.
- Shape analysis holds promise for delineation of subtle facial anomalies that may be associated with less severe manifestations of FASD.