The 3D face of Smith-Magenis syndrome (SMS): a study using dense surface models

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The SMS facial phenotype has been studied previously both subjectively and objectively, the latter using anthropometry. Here, we build a 3D surface model of face shape using images of 30 individuals with SMS and 146 controls, all under 20 years. A dense correspondence of thousands of points on each of the faces is computed and their co-ordinates are subject to a principal component analysis (PCA). This reduces the number of variables from tens of thousands to just 44 PCA modes, giving a compact model that is computationally amenable to further analysis. A linear morph between the average face surfaces of each subgroup gives a dramatic 3D visualisation of dominant shape differences including many identified previously: broad and square face shape; heavy brow; close deep-set eyes; and major nose and upper lip differences. Notable are the upward and backward displacement of the pronasale and outward rotation of the philtrum giving the previously noted “tented appearance”. The PCA modes were analysed using pattern-recognition techniques to discriminate between SMS subjects and controls using 10-fold cross validation of training and unseen test sets. The best performing algorithm, nearest mean, gave an average specificity of 96%, a sensitivity of 80% and an overall accuracy of 93%. These results confirm the potential of 3D dense surface models to support both training and clinical practice in dysmorphology. When more SMS data is available, even better results are anticipated.