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

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Background

Smith-Magenis syndrome (SMS) is caused by an interstitial deletion of chromosome 17p11.2 resulting in a characteristic craniofacial appearance and other physical, developmental and behavioural features. The incidence of SMS is at least 1 in 25,000 births and over 300 cases have been identified worldwide. Although diagnostic tests are available, delayed diagnosis is not uncommon.

Aim

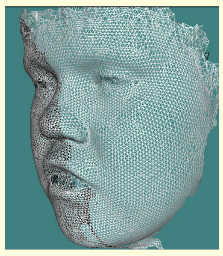
(i) To use 3D dense surface models of face shape to delineate the characteristic appearance of individuals with SMS;

(ii) to evaluate the application of pattern recognition techniques in discriminating between controls and individuals with SMS.

Data Capture

3D scans of 145 controls and 30 individuals with SMS aged between 1 and 19 years were captured using two different photogrammetric face scanners.

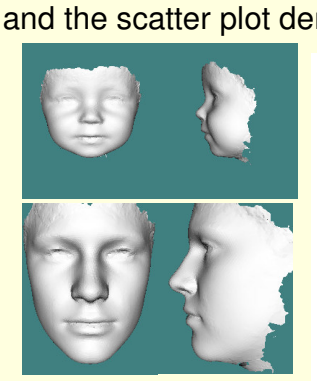
Example of face surface showing some of the 15 hand-placed landmarks used



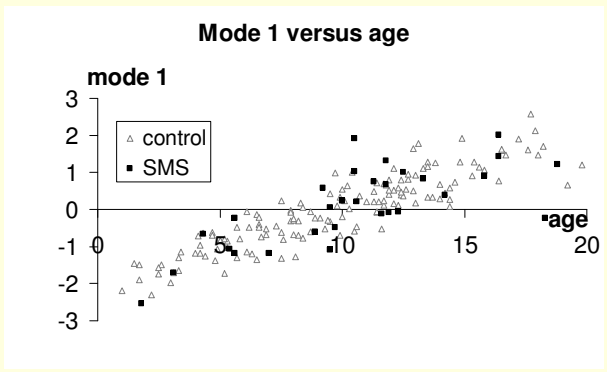
Underlying mesh of about 8,000 points

Data Preparation

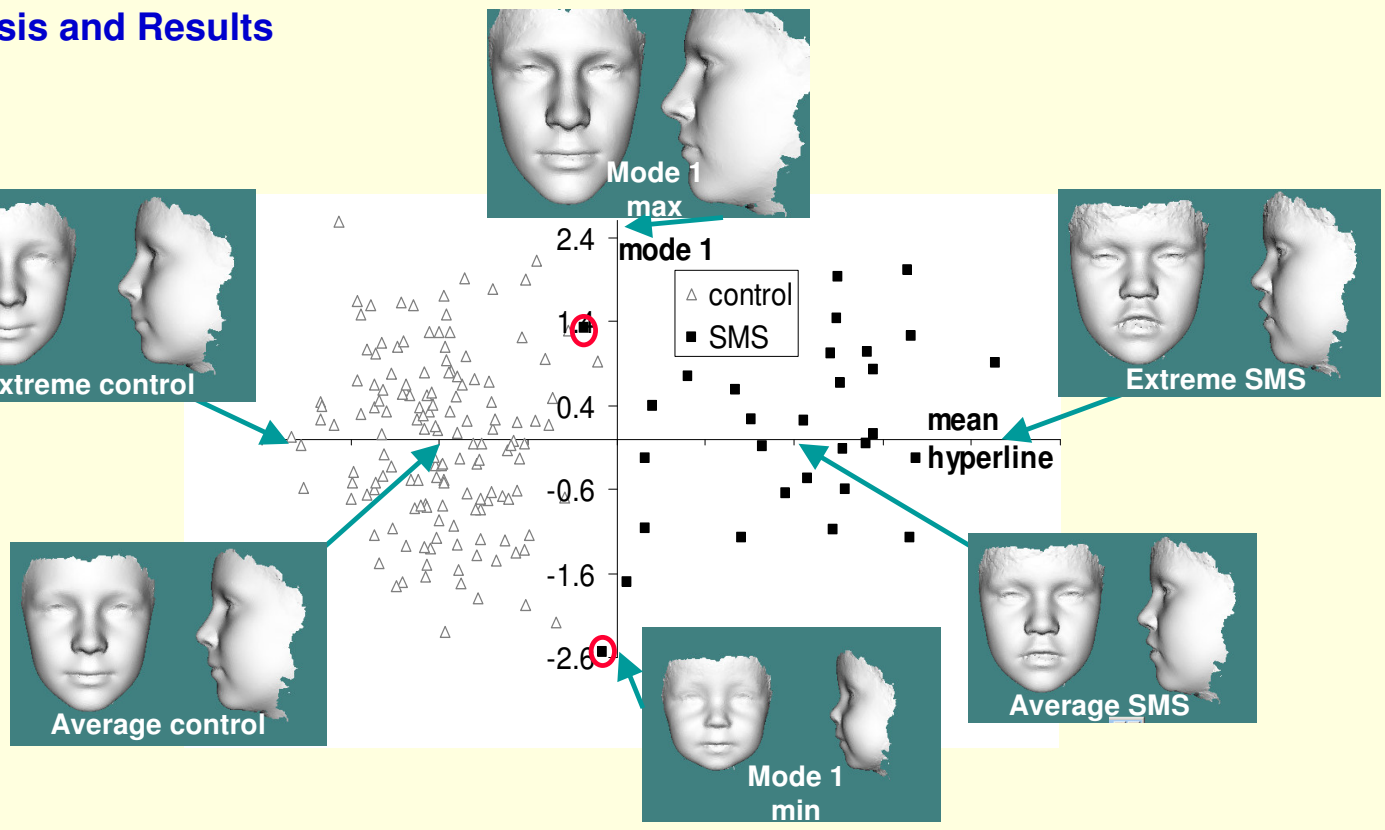
A set of faces is closely aligned using the landmarks and TPS warping [2]. Points on faces are densely corresponded and the surfaces are unwarped, trimmed and aligned.The average face is computed and deviations of each face from the average are subject to a principal component analysis (PCA). The extremes of PCA mode 1 and the scatter plot demonstrate its strong correlation with age.



Extremes of PCA mode 1



Analysis and Results



The scatter plot left shows all SMS and control faces.The vertical axis is PCA mode 1 and the horizontal axis measures the projection of a face onto a line joining the average SMS and control faces.

The plot also illustrates how proximity to nearest mean can be used to classify each face as falling in the control or SMS subgroups. Here, two individuals with SMS are misclassified as controls (encircled in red).

The PCA modes were used to train five pattern recognition algorithms (nearest mean; decision tree induction; logistic regression; neural networks; support vector machines). A clinical diagnosis of Smith-Magenis syndrome was used as a gold standard.

For the 10-fold cross validation, training examples were used to compute the mean hyperline and unseen examples for testing. The average discriminating ability of each algorithm in terms of sensitivity and specificity was computed with nearest mean producing the best results at 80% and 96% respectively.

Conclusions

The PCA modes and dynamic morph between average SMS and control faces provide an excellent 3D visualisation of dominant face shape differences, including many identified previously [1]: 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 nearest mean algorithm gives good discriminating ability at **80% sensitivity** and **96% specificity** considering only 30 individuals with SMS were available for the analysis.

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References

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[2] Hutton TJ, Buxton BF & Hammond P. (2001) Dense Surface Point Distribution Models of the Human Face In: Proc. IEEE Workshop on Mathematical Methods in Biomedical Image Analysis, Kauai, Hawaii. pp. 153-160.