

3D Digital Stereo Photogrammetric Analysis of Face Shape in Noonan Syndrome



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Background

In Noonan Syndrome (NS), the facial features include hypertelorism, ptosis, downward sloping palpebral fissures and low-set posteriorly rotated ears. Previous facial studies of NS have used anthropometry [1] and 2D photogrammetry [2]. There is evidence that some of the facial features soften in adulthood.

Aim

To evaluate the use of 3D face surfaces and different pattern recognition techniques for discriminating between controls and individuals with Noonan syndrome for four age bands (<10, <20, <30, <60) using pairs of training and unseen test sets in a 10-fold cross validation.

Data Capture



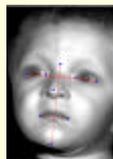
3D scans of 225 controls and 92 patients with NS were captured with a DSP400 face scanner



The face surface captured for a 3 year old child with Noonan syndrome



Underlying mesh for same child with hand-placed landmarks (in blue)



For each face scan, measurements were automatically derived using the landmarks: mid face height, inner canthal distance, palpebral fissure width, palpebral fissure slant, interalar distance, mouth width, and philtrum length. Some are shown left in red.

Data Preparation



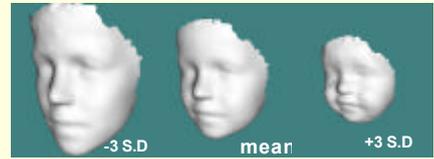
The training set of faces for an age band were closely aligned using the landmarks and Thin-Plate Spline warping (each face is shown in a different colour) [3]



Points on the face scans are densely corresponded using a single base mesh; surfaces are then unwarped, trimmed and aligned.



A mean for the set is computed and deviations of set members from the mean are subject to a principal component analysis (PCA). Examples of PCA modes are shown right.



1st PCA mode for all under 10 data (40 controls; 30 NS)



2nd PCA mode for all under 10 data (40 controls; 30 NS)

Analysis and Results

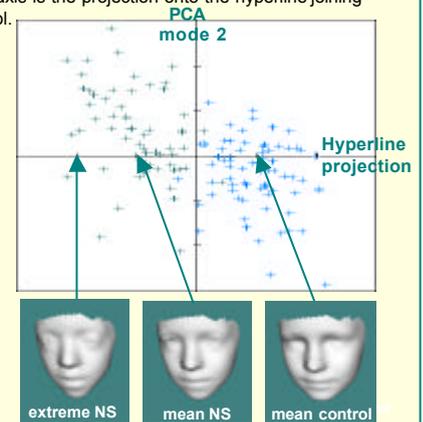
The derived facial measurements and PCA modes were used to train four pattern recognition algorithms (nearest mean; decision tree induction; logistic regression; neural networks). A clinical diagnosis of Noonan syndrome was used as a gold standard.

Unseen sets of faces, for each age band, were used to test the trained algorithms. A 10-fold cross validation, equivalent to drop 10% out testing, was used and the average performance calculated.

For the derived facial measurements, the discriminating ability of each algorithm was inferior to that for PCA mode values with accuracy for the four age bands of 83, 73, 65 and 76 per cent respectively. The measurements used were restricted by the landmarks which were chosen for reproducibility. Thus, better results may be obtained with other landmarks.

PCA MODE RESULTS	Sens	Spec	Acc	PCA MODE RESULTS	Sens	Spec	Acc
UNDER 10 (30 NS; 40 control)				UNDER 30 (80 NS; 110 control)			
Nearest mean	77	90	84	Nearest mean	83	93	89
C5.0 Trees	80	88	84	C5.0 Trees	60	77	70
Neural Networks	67	85	77	Neural Networks	76	89	84
Logistic Regression	83	88	86	Logistic Regression	79	86	83
UNDER 20 (60 NS; 70 control)				UNDER 60 (92 NS; 225 control)			
Nearest mean	85	91	89	Nearest mean	87	91	90
C5.0 Trees	63	81	73	C5.0 Trees	52	84	75
Neural Networks	82	83	82	Neural Networks	74	94	88
Logistic Regression	83	93	89	Logistic Regression	81	91	88

The scatter plot below shows all Noonan and control faces for the under 20 age band with excellent discrimination between control and NS groups. The vertical axis is PCA mode 2 and the horizontal axis is the projection onto the hyperline joining the mean Noonan and mean control.



Conclusions

For the PCA modes, logistic regression and nearest mean were the most powerful discriminators with % **sensitivity** and % **specificity** ranges of **83-87** and **90-93** respectively. Somewhat surprisingly, the simple nearest mean classification does consistently well. For children under 10, the induced decision trees needed only 3 PCA modes to give a **sensitivity** of **80%** and a specificity of **88%**. Since the abstract submission, the same data has been analysed using support vector machines with even better results.

A very recent 50% increase in the number of face scans for the Noonan syndrome group will provide further training and test examples. In addition, the recent introduction of a new scanner with finer resolution and increased surface coverage (consistently capturing both ears in the scan) can only improve on the results described here. Syndromes such as Angelman, Rett, Rubinstein-Taybi, Smith-Magenis, VCFS and Williams are being studied in a similar fashion.

References

- [1] Allanson JE, Hall JG, Hughes M, Preus M & Witt RD. (1985) Noonan Syndrome: The Changing Phenotype. *Am J Med Gen* **21** 507-514.
- [2] Sharland M, Morgan M & Patton MA. (1993) Photoanthropometric Study of Facial Growth in Noonan Syndrome. *Am J Med Gen* **45** 430-436.
- [3] 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.