

Delineation and Visualisation of Congenital Abnormality using 3D Facial Images

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INTRODUCTION

One in fifty children is born with significant congenital abnormality [1]. Some have multiple anomalies constituting a specific dysmorphic syndrome. Of 5,000 or so dysmorphic syndromes, over 700 involve dental, oral or craniofacial differences. Dysmorphic craniofacial features include extended distance between eyes (telecanthus) or pupils (hypertelorism); inner/outer eye corners at different levels (sloping palpebral fissures); unusual nose width (interalar distance), bridge of nose, skull shape or position/orientation of ears; occurrence of skin folds; and, drooping eyelids (ptosis).

Syndrome diagnosis is based on clinical observation of abnormal body parts and proportions, and unusual appearance. Early diagnosis is important if it is to guide clinical investigation and treatment, and genetic counseling of parents. The condition focused on here, Noonan syndrome [2], involves short stature (72%), mild to moderate learning disability (61%), congenital heart disease, low set ears, hypertelorism, neck webbing, sloping palpebral fissures and ptosis. Noonan syndrome affects approximately 1 child in 2000 and on average a diagnosis is made by four years of age. Figure 1 illustrates characteristic facial features of Noonan syndrome.

Visual clues are important in dysmorphology. The age of individuals being assessed is also significant, since some features are present at birth and others evolve with time [3]. Recognition of facial characteristics of particular syndromes lags behind somatic and behavioural characteristics and may delay diagnosis [4]. Characteristic features may not develop simultaneously but become apparent as a child develops, and may even become less

obvious in later life [5]. In Noonan syndrome, physical manifestations in adults may be subtle and some without a known heart defect or other medically significant problem may have gone unrecognised [6].

Rarity of syndrome occurrence is a problem for clinicians with infrequent exposure to congenital disorders. Thus, an analysis of craniofacial features and their change over time is important in the delineation of dysmorphic syndromes and training in their detection. For this reason, more than 10,000 2D photographs already form an important component of the London Dysmorphology Database (LDDDB), which catalogues some 3,500 dysmorphic syndromes [7]. Although 2D photographs can depict typical craniofacial features of syndromes, there are problems with pose that are eliminated by employing 3D images. Once captured, 3D images can be rotated and viewed from any angle to support clinical assessment, whereas 2D photographs are fixed in the same pose forever.

We report on 6 months' use of a photogrammetric scanner to capture 3D facial shape and appearance and give an analysis of data gathered from 62 children, of whom 22 have Noonan syndrome. The objective is to evaluate the use of 3D images to visualise and delineate facial features in dysmorphic syndromes. The next section introduces craniofacial assessment [8], summarises our data collection and describes problems encountered. The penultimate section is a preliminary analysis of the data collected. The final section contains an interpretation of the results and a summary of future plans. All image manipulations, other than capture, are provided by *ShapeFind*, a shape analysis system developed in-house.



Figure 1: facial shape/appearance using 2D photos of a control group and individuals with Noonan syndrome
In 1b, note eye spacing, downward slope of eyes, lower upper eyelids, broader nasal base and lower set ears

DATA COLLECTION AND PREPARATION

For over 45 years, 2D photographs have been used as a quickly obtained, permanent record of a patient's appearance. Facial measurements can be taken directly from photographs and do not require the physical co-operation of subjects, often not forthcoming from young children. However, fewer measurements can be taken from a fixed photograph, palpation of hard tissue landmarks is impossible, and, of course, there are errors due to pose and lighting. Ratios and angles, rather than raw distances, can often reduce errors of pose. Anthropometry, direct manual measurement using specialised but simple measuring devices, has several advantages: it is simple, non-invasive, cheap and based on direct measurement. Major disadvantages are lack of permanent records other than measurements obtained and variation in accuracy in relation to training differences and idiosyncracies of individual anthropometrists.

2D data collection

A collection of 2D photographs of children aged between 5 and 11 was available from an earlier study [9]. None of the children was known to suffer from a condition affecting craniofacial growth. The photographs, simultaneous front and lateral portraits, were taken while using a restraint device to control pose. A shape template of 120 landmarks, representing the combined shape of the lower jaw, lips, nose, eyes, eyebrows and ears, was overlaid on 124 frontal portraits (fig 2). The templates were registered using the Procrustes algorithm [10] to remove rotational, translational and scalar variation, and to enable a mean shape to be computed as in figures 1a and 1b. An average appearance was computed by warping shape templates to the mean shape and averaging greyscale intensities (figs 1c & 1d).

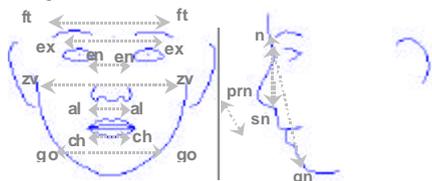


Figure 2: 10 facial measurements used in our manual craniofacial assessment - shown with respect to shape template

A group of 51 2D photographs of individuals with Noonan syndrome was also collected from clinical records and the LDDDB image collection. Most of these photographs had been taken with no attempt to standardise pose or lighting, and they included both children and adults. The mean shape and appearance (see figures 1b and 1d) reconfirm well established Noonan syndrome features of hypertelorism, downward sloping palpebral fissures and ptosis. The observation of low set ears in figure 1b and the effect of pose is discussed in more detail later.

Anthropometric and 3D data collection

An annual meeting for families with members affected by Noonan syndrome provided an opportunity to carry out a craniofacial assessment of 21 children and 11 adults using standard manual, anthropometric techniques. Figure 2 shows the facial measurements that were recorded. Of

these 32 individuals, 22 had previously been diagnosed with Noonan syndrome, 5 did not have the syndrome and for 5 adults a diagnosis was not recorded. On the same occasion, a TCTi DSP400 scanner was available to record 3D images of both facial geometry and appearance. The instantaneous nature of a photographic scan is essential when capturing images of young children who do not remain sufficiently still for alternative laser-based capture. The surface geometry, typically involving between 4,000 and 9,000 vertices, and appearance can be viewed separately or in combination. The simultaneous capture of surface geometry and appearance aids visualization and subsequent landmarking.

In general, the DSP400 scanner produces very good images. Shiny patches due to skin oils or moisture can cause strong light reflections and affect the surface geometry obtained. A major drawback for this particular application, given the involvement of irregular ear position in Noonan syndrome, is the inability to capture both ears reliably in a single scan. Moreover, for many subjects even landmarks such as gonion (see *go* fig 2) were not captured on both sides of the face.

The machine-based measurements for some landmarks, such as those involving the eyes and mouth, are likely to be more accurate, especially for less co-operative children. Inevitably, there are difficulties in placing landmarks on images without being able to palpate the soft tissue and feel underlying bony landmarks. Consequently, there are differences in the electronically derived measurements (see table 1). The relatively large error in *sn-prn* is due to light reflection from shiny nose tips affecting the surface geometry captured. These differences would be significant if they were the only measurements available from the electronic image. It should be emphasised, however, that the mesh has thousands of points and so lack of precision on a few measurements could well be compensated for by extensive coverage of the subtleties of variation in facial geometry.

Abs diff	mf-mf	zy-zysn	zn-prn	n-gn	n-sn	al-al	ch-ch	en-en	ex-ex
mean	1.04	2.53	2.37	1.48	1.84	0.78	1.42	1.50	2.25
st dev	0.88	2.31	1.64	1.51	1.74	0.90	1.97	1.33	1.62

Table 1: differences (mms) in manual & machine based measurement

DELINEATION/VISUALIZATION USING 2D DATA

A principal component analysis (PCA) of the landmark coordinates on the photographs produces a set of modes and for each mode a percentage coverage of the overall template shape variation. For the Noonan subgroup, the largest variation (47.7%) by far is vertical pose (Table 2). Although low ear position appears to be involved in this mode, it could easily be confused with variation in vertical pose. Horizontal pose (12.8%) plays a smaller, but significant, role in the second mode and the third mode reflects variation (11.6%) in the ratio between vertical and horizontal facial dimensions. Mode 3 is likely to be highly correlated with age, but as ages of the subjects were not available this could not be confirmed. The top 3 modes for a PCA of the control subgroup were similar but with differences in coverage.

When the Control and Noonan subjects are combined in a single dataset, a PCA of the shape templates gives three top modes similar to those for the separate groups – vertical pose, horizontal pose and aspect ratio of the face – in all, covering about 70% of the shape variation. The next three modes display variations of hypertelorism, ptosis, interalar distance and slope of palpebral fissures.

MODE	NOONAN (N=51)		
	- 3 SD	MEAN	+ 3 SD
1 47.7%			

Table 2: first mode of PCA for 2D shape data

One way to visualise the effect of multiple mode changes simultaneously is to construct an orthogonal projection of all dataset examples onto a hyperline in the multi-dimensional shape space of the modes. For example, a line constructed between the two means of the separate control and Noonan subsets gives a good separation between the two subgroups – a desirable feature if we are to consider syndrome detection in the future. The *ShapeFind* software provides a tool to visualise the effect of traversing the hyperline and simultaneously to view the corresponding facial shape and associated mode value changes. For the 2D dataset, mode 1 is one of the modes that alters significantly. Therefore, for 2D images it is not straightforward to separate Noonan facial feature variation from variation in pose. This emphasises the need to use 3D images where pose variation can be corrected for.

DELINEATION/VISUALISATION OF 3D DATA

The DSP400 image capture software exports surface and appearance data separately. The 2D appearance is used to place landmarks which are then projected onto the 3D surface. Once again, the Procrustes algorithm is used to compute mean landmarks. Thin-Plate Spline warping is used to warp the surface meshes onto the mean set of landmarks and after further manipulation the mesh vertices themselves can be used as input to build a new 3D PCA model of shape variation.

The small number of scanned individuals with Noonan syndrome (almost exclusively children) forces any analysis reported here to be restricted to 62 children aged 4 months to 16 yrs, and including 22 individuals with Noonan syndrome. This collection is reasonably balanced in terms of age. The first mode of the PCA for shape (row 1; Table 3) is strongly correlated with age. The -3SD image demonstrates the characteristic round face of infancy. The nose is small with a depressed root, and overall configuration is relatively concave. With age (+3SD image), the vertical dimensions increase with particular lengthening of the nose and chin, and protrusion of nasal root and tip leading to increased facial convexity. From visual inspection, mode 2 (row 2; Table 3) has a strong Noonan to control variation. The -3SD image illustrates features typical of Noonan syndrome: widespread eyes, shallow orbits, reduced growth of the upper jaw, and small retro-placed chin. It suggests low facial muscle tone which is certainly apparent clinically.

MODE	-3 SD	MEAN	+3SD
1 61.5%			
2 8.9%			

Table 3: first two modes of PCA for 3D surface data

As in the 2D image case, multiple modes can be varied simultaneously using the *ShapeFind* facility to traverse the mean hyperline projection and simultaneously to identify which modes undergo major variation as focus moves between the two subgroup means. For example, mode 1 undergoes a negligible change which is consistent with the reasonable age balance of the dataset. On the other hand, other modes alter by almost two standard deviations as the extended hyperline is traversed. These individual modes can then be inspected to see if particular feature variation occurs. Table 4 (row 1) illustrates some of the faces obtained by varying all of the modes in a linear fashion to focus on five points along the mean hyperline.

The *ShapeFind* software can also generate a PCA model for variation in appearance using the greyscale values for the pixels in 2D images once they have been warped to give a shape free appearance. For this, we can use the 2D appearance exported from the 3D scanner. This model can then be combined with the surface geometry model of the previous section to give a combined model of 3D shape and appearance variation. The first mode is strongly correlated with age once again. Mode 2 shows some features of Noonan syndrome. The faces in Table 4 (row 2) correspond to those generated by the combined model at five positions along the mean hyperline projection.

In terms of clinical interpretation, the first synthesised face in row2 is typical of young children with Noonan syndrome. The face goes beyond that of the one above by illustrating drooping eyelids, a short broad nose and an inverted triangular face. Interestingly, the mean Noonan face generated (column 2, row 2) is unremarkable clinically speaking. This may be explained by the age range of the subjects which spans periods of dramatic change in facial appearance up to the age of 5 and during the pubertal growth spurt.

CONCLUSIONS AND FUTURE PLANS

The advantages of 3D photogrammetry over 2D photography have been well established, even in these early stages of the project. Problems with pose are avoided and landmarking is made easier by being able to view both surface and appearance from any desired angle. The problem of the completeness of the captured facial surface

Point beyond mean Noonan by half distance between Means	Point corresponding to mean Noonan	Mid point between means	Point corresponding to mean control	Point beyond mean control by half distance between means
				
				

Table 4: synthesized faces corresponding to selected points along the mean hyperline projection

is being discussed with the scanner manufacturer. The strength of lighting and appropriate diffusion and filtering, for a full range of skin tones, are being improved with advice from a photographer. Such improvements in hardware and lighting will inevitably improve the accuracy of derived measurements and the appearance variation in scanned and hence in synthesised faces.

Computer-based analysis of 2D images of dysmorphic faces has been proposed before [13], but the study terminated prematurely. 3D photogrammetry has also been used recently to record facial growth [12] but only for manually marked-up landmarks without computing full facial geometry. The use of PCA to study facial shape and appearance is not new [11] but its use to build combined 3D surface and appearance models in the study of dysmorphic syndromes is novel. The decomposition of the subtleties of the face into separate individual modes can be very enlightening and will certainly be of benefit in training in medical genetics. But, it is the composite variation of multiple modes, as with the variation between subgroup means along a linking hyperline, that is most impressive. This could prove to be a powerful visualisation tool showing continuous transition between syndromic and non-syndromic faces while simultaneously highlighting which modes are most influential during the transition. The ability to view both scanned and synthesised faces from any desired angle is a valuable bonus.

The intuitive approach to pattern recognition is usually "gestalt" or big picture. Anthropometry is unidimensional but very helpful in picking out features most different from the average, and thus those features which best discriminate between syndromic and non-syndromic faces. 3D photogrammetry and the visualisation tools provided in *ShapeFind* both discriminate and train the eye but in a 3D gestalt mode.

Plans are already in place to expand both the Noonan and control image collections of 3D scans. Once the collections reach a reasonable size, work can begin on detection using pattern recognition techniques. Age related variation will be more reliable and convincing once the dataset is better populated. More precise study of age-related changes will then be feasible. Related dysmorphic syndromes such as Cardio-facial-cutaneous, Costello and Williams will also be the focus of further work.

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