Geometric Morphometric Analyses of Facial Shape in Twins

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Citation

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Abstract

Twin pairs represent a unique window into the role of heredity in the determination of any human trait. A primary characteristic by which humans present themselves to the external world is via the anatomy of their facial shape. Thus, facial images are becoming increasingly significant in biometrics and anthropological research. In this study, the method of geometric morphometrics such as superimposition and Procrustes fitting were used to generate facial shape variables in the form of relative warps of landmarks derived from perceived identical twins. Sixty pairs of twins participated in the study. A digital camera was used to photograph the subjects in neutral mode facing front. The shapes of the face were then summarized using a total of 39 landmark points from the face. Heritability was measured as concordance in the shapes of the face of each member of pair of twin. Specifically, correlation between the relative warp scores of the pairs of twins was used to test for heritability. Results showed that facial shapes of the twins had a moderate to highly positive correlation indicating that the landmark points established on one twin can also be observed in the other twin. Furthermore, the probability values of each correlation coefficient revealed that there is a significant linear relationship of the landmarks between twin 1 and twin 2 indicating a high degree of heritability of facial shapes. The results of the study underscore the utility of geometric morphometrics in the study of inheritance and variability of traits in organisms.

INTRODUCTION

Anthropology has long played a central role in the development and application of new methods in quantitative biology, in general, and morphometrics, in particular. Questions concerning patterns of variation, association, causation and inheritance in human populations are always the main focus in anthropological research. Many studies on the human face including that of cephalo-facial measurements to study human groups are based on identified biometric features[1-13], which are genetic properties of a human being. Facial data is commonly obtained by direct anthropometric measurements[14]. While these traditional approaches to morphometric analysis usually involves the application of multivariate statistical procedures to collections of distances, angles, or distance ratios[14-17], the last couple of decades have shown the growth and development of new conceptual models and analytical tools. With the introduction of computer-assisted techniques, morphometrics has evolved towards a synthetic approach which is based on multivariate analysis of landmark coordinates[18-19]. Capturing geometry by way of landmark data has become very common to genetic, evolutionary, ecological and anthropological studies. The method of 'geometric morphometrics' (GM), an adaptation of multivariate statistics and graphics to the study of phenotypic variation, detects form changes thus shape variations in living organisms can be detected. This method identifies the relative locations of a set of individually identified points or "landmarks" and the 'shape coordinates' are used as biometric variables that are regressed one by one on the factors that cause them or the features of the systems they are presumed to affect thus is used to study anthropometric variations and the heritability of face shapes in twins. For over 100 years, twin studies served as a basic tool in evaluating the relative contribution of genetic and environmental factors[20]. It is done mostly to study whether heritability of a trait or traits is genetic or environmentally influenced. The heritability of human craniofacial morphology for example has been thoroughly investigated in twins and families. A genetic component has been reported for 60-90% of craniofacial traits including facial height, position of the lower jaw and cranial base dimensions[21]. Twin pairs represent a unique window into the role of heredity in the determination of any human trait. A primary characteristic by which humans present themselves to the external world is via the anatomy of their facial shape[22].

Thus, facial images are becoming increasingly significant in biometrics and anthropological research.

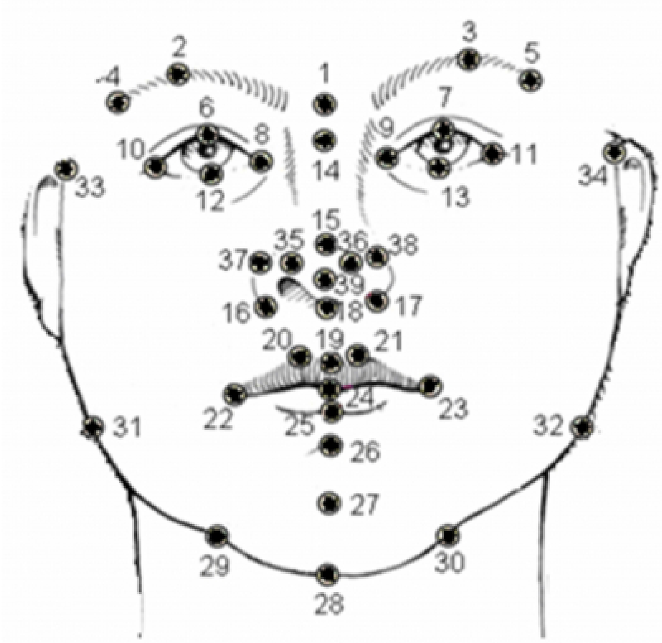
MATERIALS AND METHODS

Since the human cranium is the result of a composite functional system, several factors are involved in its morphology[23]. Many polygenic craniofacial traits however, are susceptible to environmental modification and can be difficult to study with conventional methods. Two dimensional photographs are most commonly used to facilitate visualization, assessment, treatment of facial abnormalities and morphometric studies. In this study we use geometric morphometric methods such as superimposition and Procrustes fitting to yield relative warps of landmarks derived from perceived identical twins to study heritability in craniofacial traits. This study will establish whether the landmark characteristics of the face between identical twins are heritable.

There were 60 twin pairs who participated in the study. A digital camera was used to photograph the subjects in neutral mode facing front. The 39 landmark points used in this research was based on a study on a three-dimensional morphometric study of craniofacial shape in Schizophrenia[24] (Figure 1). Landmarks were generated using tps software[25]. The generation of landmarks involve the following basic steps (1) building of TPS file format from jpeg format, (2) image acquisition and (3) digitization. Landmarks generated per subject were replicated 3x using the digitization tools of tpsDig2 program.

Figure 1

Figure 1. Landmark points of the face used in the study (adapted and modified from Buckley et. al. 2005). 1-Glabella, 2,3-Superciliare, 4,5-Frontotemporale, 6,7-Palpebrale Superius, 8,9Endocanthion, 10,11-Exocanthion, 12,13Palpebrale inferious, 14-Sellion, 15-Pronasale, 16,17-Alare, 18-Subnasale, 19-Labiale superious, 20,21-Labiale superious (left & right), 22,23-Cheilion, 24-Stomion, 25-Labiale inferious, 26-Sublabiale, 27-Pogonion, 28-Gnathion, 29,30 added landmark, 31,32-Gonion, 33,34-Tragion, 35,36-Lateral pronasale, 37,38-Superior alare, 39-Infrapronasale.



Prior to analysis of shapes, links and slider files were created. These files were used simultaneous with the input of data for the relative warp analysis. The landmark configuration of each specimen generated from digitization of the images[26] was normalized for translation and rotation using the concept of Principal Component Analysis (PCA) as embedded in the relative warp analysis tpsRelw program[27]. The purpose of doing this was to scale the specimens to a common size so that the remaining differences between individuals or mean shapes are differences in mean shape only which is achieved by Procrustes superimposition[23,28-30]. Once the landmark configurations are Procrustes aligned, extraction of the shape was done through tpsRelw[27]. The variation in shape of the face was then used as inputs to multivariate analyses and the observed variation and mean differences were viewed using tpsRelw program.

To determine heritability, the relative partial warp scores were used as shape variables and correlation analysis was done to determine whether the landmark points that served as shape variables are heritable in twins. Heritability was estimated using the PAST (version 1.92) software[31]. Looking at the scatter plot with its corresponding correlation coefficient value and its probability value, positive correlation coefficient values (r) indicate heritability and probability value (p value) less than 0.05 indicate that r is significant.

RESULTS AND DISCUSSION

Figure 2 shows the consensus configuration of the face among the twins. The resulting Procrustes coordinates captured shape information only and was used for subsequent multivariate statistical analyses[32]. The deviation of landmark points is highly variable in the area of the gonion and tragion.

Figure 2

Figure 2. Consensus configuration of the face

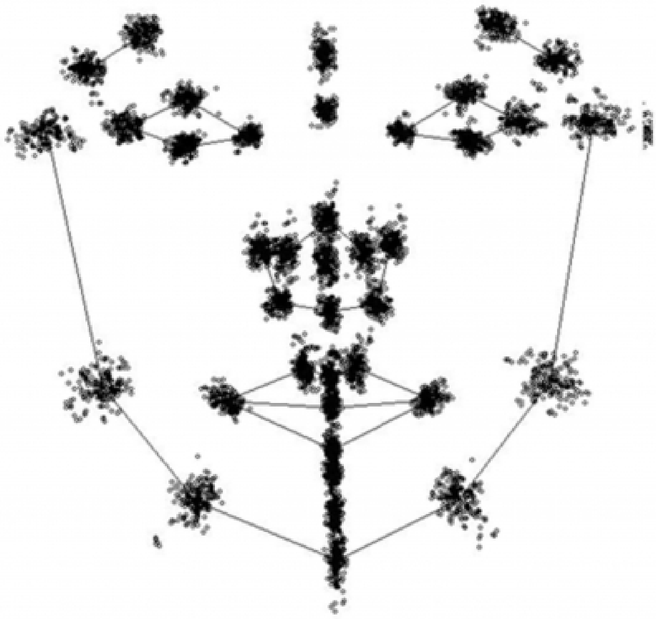
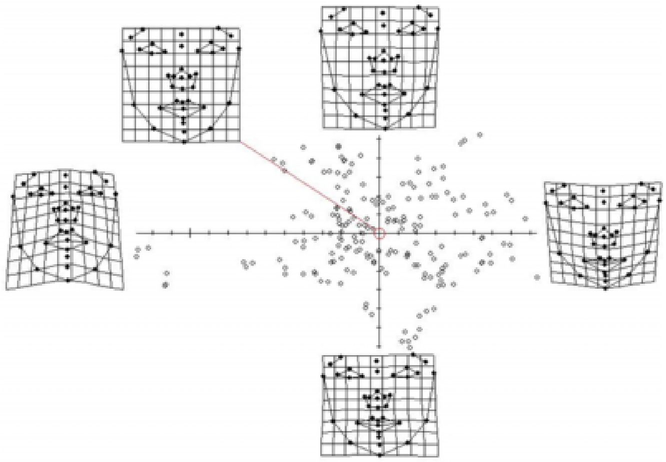


Figure 3 show the thin plate spline visualization grids of the facial shape of the twins. The leftmost side of axis 1 (horizontal) revealed a face with a wider jaw, broader chin, pouting lips and slightly small eyes. In contrast the rightmost side shows a face with a narrower jaw, pointed chin, thin upper lip and wider eyes. The uppermost part of axis 2 (vertical) denotes more on the outline of the face wherein it is broadest in the part of the tragion and slowly narrow into the jaw and the chin. In contrast the lowermost part of axis 2 denotes broader facial shape in the tragion, jaw and chin.

Figure 3

Figure 3. Thin plate spline visualization grids of facial shape derived from relative warp axes 1 and 2.



Thin plate spline visualization grids of the face of the twins by the first four relative warp axes are shown in Figure 4. The shape differences depicted by the thin-plate spline transformation grids revealed that facial shape can be warped into different sizes and shape based on the Procrustes fitting. The facial shape when warped can show differences in the size and shape of the eyes, nose, lips and facial outline.

Relative warp 1 described a face with the head which is narrow while the jaw and chin are broad; both the upper and lower lips are thick; and the eyes are narrower (-sd). Facial shape in +sd show a broader portion of the head, narrow jaw, pointed chin, thick lower lip and wider eyes. Relative warp described the facial shape which is broad, the nose more pointed, the upper lip is thinner and the entire shape outline of the lip is wider (-sd). In contrast, the +sd show a straighter facial shape with pointed chin, thick upper lip, shape outline is narrow and nose has a wider base. Relative warp 3 described the orientation of the face. In –sd, the facial shape is stretched toward the right side of the grids while in the +sd, the face is more compressed at the middle of the grid. The very obvious difference is that in the +sd, the chin is more pointed than in the –sd. Relative warp 4 described a face showing deformations of the eyes and lips. In the –sd, the eyes are smaller and the upper lips are thinner as compared to the +sd.

Figure 4

Figure 4. Thin plate spline visualization grids of the first four relative warp axes

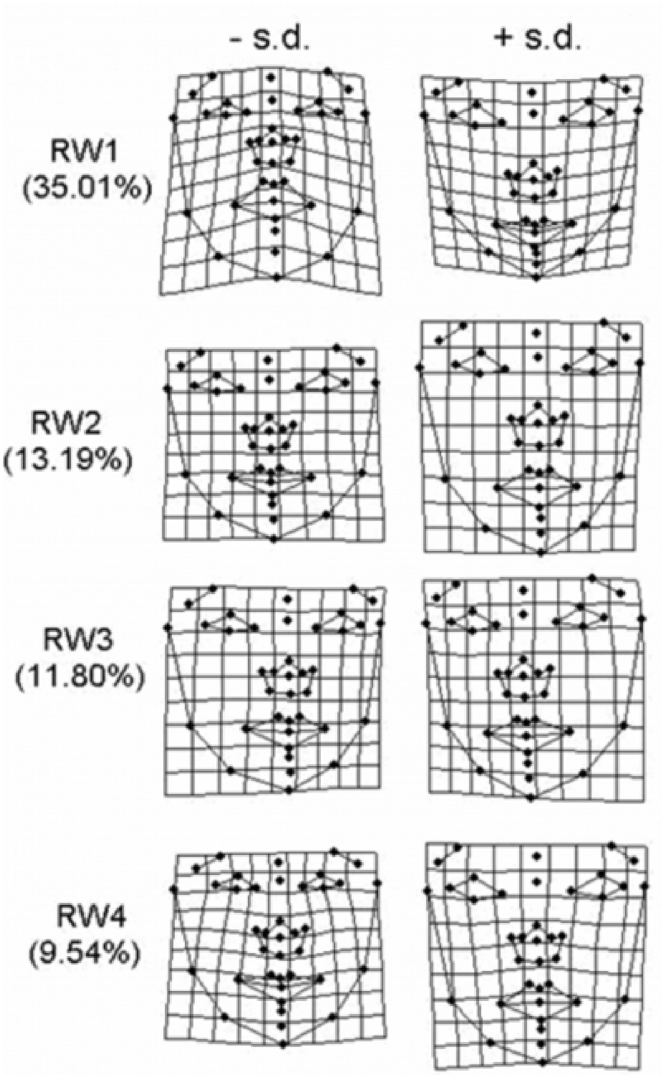
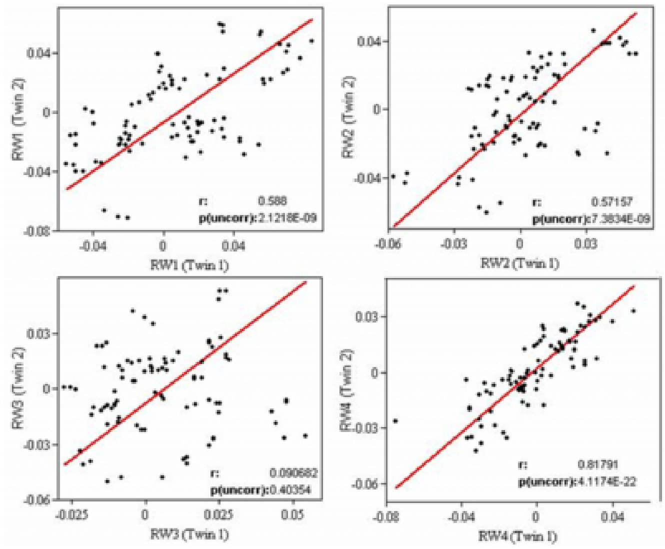


Figure 5 show the r values and p values of the first four relative warp axes. The linear regression line shown in the scatter plot diagrams show a positive linear correlation coefficient. All the relative warp axes showed positive correlation coefficient values (r) indicating that the landmark points established on one of the twins can also be observed in the other twin. Furthermore, the probability values of each correlation coefficient revealed that there is a significant linear relationship of the landmarks between twin 1 and twin 2. This proves that the r values are not due to chance but instead is due to heritability of the landmark areas. The results of this study therefore suggest that the landmark points being used are significantly heritable between twin pairs. The results of this study complements the research done by several authors[20,22,32,33].

Figure 5

Figure 5. Heritability of facial landmarks between twins.



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