A Cephalometric Study Of Craniofacial Variation In Adult Twins

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INTRODUCTION

The lack of available genetic data on human skeletal characteristics has had a limiting, if sometimes unrecognized, effect on progress toward the solution of some fundamental problems in anthropology and orthodontics. Although head form and facial proportions appear to be heritable, quantitative analysis of hereditary variability in cranial and facial dimensions in related individuals have been few. Consequently, attempts to establish morphological criteria for racial comparisons, or for orthodontic diagnostic purposes have been hampered by a need for more information relative to genetic variation in the face and skull.

The cephalometric roentgenographic method for obtaining skull measurements on the living has greatly facilitated research in growth and development of the head, and it is surprising that so few investigators have employed the technique in investigating hereditary variation. Wylie (1944) was the first to compare the craniofacial patterns of related individuals by means of the cephalogram, reporting on fifteen families of which thirteen had like-sexed twin pairs. Curtner (1953) reported on five families, three of which included twins. By superimposing tracings of various pairings in his study sample (mother-father, mother-daughter, father-daughter etc.) he determined "conformity" in various craniofacial outlines. Stein, Kelley, and Wood (1956) used the cephalometric x-ray in studying a large group of female college students, their siblings and parents. Four twin pairs were included in their investigation. The most recent reports are those of Noyes (1958), who presented cephalogram tracings of three generations of a family and a pair of twins as well, and Kraus, et al (1959), who studied six sets of triplets. Lundstrom's cephalometric study (1954, 1955) reported findings on the largest twin sample yet assembled, fifty pairs each of adolescent monozygotic and dizygotic twins.

In the present investigation the twin study method is employed in order to further explore genetic influences on variations in several craniofacial dimensions of a group of post-adolescent subjects in whom skull growth is essentially complete.

METHODS AND MATERIALS

A study which has been in progress for many years at the Columbia-Presbyterian Medical Center has drawn twin subjects, unselected as to sex and zygosity, from a variety of sources in New York City. The cephalographic study sample consisted of fifty-six pairs of like-sexed adult twins (thirty-five pairs monozygotic, twenty-one pairs dizygotic), having a median age of 24 years, with a range from 18-55 years. Diagnosis of zygosity was by the "proven dizygotic method," based upon blood group factors and reliable morphological characters. The method is discussed at length elsewhere. (Os-

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borne and DeGeorge)

Roentgenograms of the head in norma lateralis were obtained with each subject fixed in a Margolis cephalostat, employing a target mid-sagittal plane distance of sixty inches. Tracings of the headplates were made on tracolene paper, and second tracings of twenty of the headplates selected at random were used to determine measurement errors.

The following roentgenographic landmarks were used: nasion, sella turcica, Bolton point, articulare (Bjork), gonion, gnathion, point B, and the constructed points *x* and Ans′ (Fig. 1). Point *x* is determined by dropping a perpendicular from point B (Downs, 1948) to the mandibular plane and Ans′ is obtained by constructing a perpendicular from the anterior nasal spine to the line NGn.

Linear measurements only are reported in this study. Each dimension was measured to the nearest 0.5 mm and then the intrapair differences calculated. Comparisons were carried out between the average intrapair differences of monozygotic and dizygotic twins, expressed as variances. Fisher's F test was employed to test the significance of the observed differences.

Results

The intrapair variances obtained for each of the dimensions studied are presented in Table I. Three diameters, ramus height (Ar-Go), chin length, and the anterior concavity of the mandible (Meredith, 1957) showed sex differences upon comparison of the male-female intrapair differences of monozygotic twins and for that reason will be reported in a separate communication. No significant sex differences are apparent in the other dimensions studied. Therefore, the observations on male and female twins presented here have been combined for analysis.

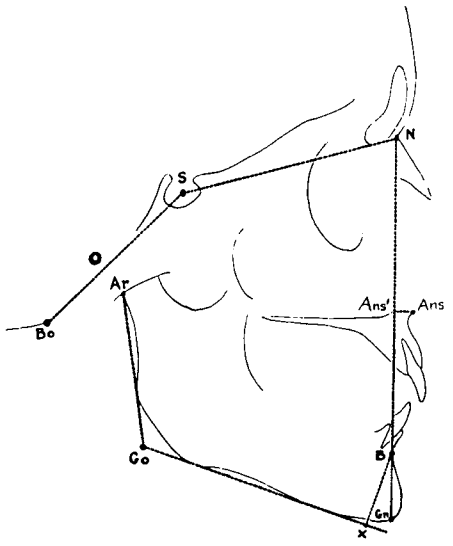

Fig. 1. Reference points used and linear dimensions recorded. (see text)

In dizygotic twins the variance found for the anterior cranial base (N-S) is about six times that calculated for monozygotic twin pairs, yielding an F ratio of 5.82 ($P < .001$). Thus, a strong hereditary component of variability is measured in this portion of the skull.

Although the dizygotic variance obtained for the posterior cranial base dimension (S-Bo) exceeds that of the monozygotic twin pairs, the difference is not statistically significant. Furthermore, as difficulty was encountered in accurately locating point Bo on some of the cephalograms, the measurement error variance (10.65) actually exceeds, though not significantly, the value obtained for monzygotic twin pairs. Bolton point has been similarly reported as an unreliable landmark by Bjork (1947), who considered it "inferior" to points such as Ar or N. It seems clear that further study of hereditary factors in the length of the posterior cranial base will require a more reliable posterior landmark than the Bolton point.

Mandibular body length was measur-

Table 1

MEAN INTRAPAIR DIFFERENCES IN LINEAR CRANIOFACIAL DIMENSIONS OF MONOZYGOTIC AND DIZYGOTIC TWIN PAIRS

Linear Measurement	N (Pairs)	Variance	F Ratio	P
N-S				
Monozygotic	35	1.650		
Dizygotic	21	9.607	5.82	<.001
meas. error	20	.363		
S-Bo				
Monozygotic	35	10.193		
Dizygotic	21	15.738	1.54	.10-.25
meas. error	20	10.65		
N-Gn				
Monozygotic	35	5.346		
Dizygotic	21	26.774	5.01	<.001
meas. error	20	.550		
N-Ans'				
Monozygotic	35	3.664		
Dizygotic	21	3.101	1.18	>.75
meas. error	20	.794		
Ans'-Gn				
Monozygotic	35	3.589		
Dizygotic	21	21.232	5.92	<.001
meas. error	20	.319		
Go-x (mandibular body length)				
Monozygotic	35	4.17		
Dizygotic	21	19.80	4.75	<.001
meas. error	20	.59		

ed from Go to x. Comparison of the variances of monozygotic and dizygotic twin pairs indicate a high component of genetic variability in this dimension ($F = 4.75$, $P < .001$).

Average intrapair differences between monozygotic and dizygotic twins in total face height (N-Gn) also demonstrate a relatively large hereditary component of variability ($F = 5.01$, $P < .001$). A similar, but less marked, difference was found by Lundstrom in his adolescent twin data.

When face height is analyzed in terms of the respective upper and lower components, however, it is apparent that these areas do not contribute equally to hereditary variation in total face height. The lower face (Ans'-Gn) shows high genetic variability, while the upper facial component demonstrates relatively little.

Discussion

The anterior cranial base region is of importance to orthodontists as it is an area common to both the cranial skeleton and facial skeleton (Scott, 1955). The relationship of the N-S dimension to facial prognathism has been discussed by Bjork (1947), and its correlation with "sturdiness" in body build proposed by Lindegaard (1953). Anteroposterior enlargement of the cartilaginous cranial base occurs in the fetal period by growth at the sphenoethmoid and spheno-occipital synchondroses and, while this area continues to grow postnatally, it does so slowly relative to the rapid growth

of the facial skeleton (Applebaum, 1953). Brodie (1941) and Ortiz and Brodie (1949) have analyzed postnatal growth in this region by means of serial cephalograms.

In his study of adolescent twin subjects, Lundstrom found twice as much intrapair N-S variation in dizygotic twins as in monozygotic pairs. This difference is much more apparent and confirmed with a high degree of significance in the present study of adult twin pairs in whom growth in this area is essentially complete. On the other hand, Kraus, Wise and Frei (1959), found "no greater similarity" in the diameter of N-S in monovular twins than in diovular twins, and question the utility of diameters and angles in recognizing the inheritance factor. Their method of superimposition of paired tracings in order to assess concordance in "total morphologic configuration" of individual bones appears itself to be open to question owing to its fundamentally subjective nature.

To the authors' knowledge no prior cephalometric data on hereditary variability in jaw length have been reported. The dimension utilized here (Go-x) measures what is essentially the tooth bearing (or "effective") length of the mandible which is of importance in orthodontic analysis. The chin portion of the jaw thus may be thought of as a "process" of the mandibular body. As mentioned above, the chin dimension, which is "bigger and juts more strongly forward in the masculine sex" (Hooton, 1947) will be discussed more fully in another paper.

Although little is known about the inheritance of individual variations in face form, anthropologists have felt that, apart from functional and age considerations, "inheritance does seem to manifest itself as a generally controlling factor" (Hooton). Longitudinal studies of the relation of nasal, or upper face height, and subnasal (lower) face height (Brodie, 1941) (Meredith, et al 1958) are in some disagreement as to the constancy of the relationship of these two components to one another during growth and development.

Comparison of the intrapair differences between monozygotic and dizygotic twins for upper and lower face height reveal a marked difference in the variability of these two components. Lower face height provides a significant measure of genetic variation, while upper face height does not. It is apparent that it is the lower facial component which is primarily responsible for the hereditary variation observed in total face height in these data. This finding appears to confirm the conclusion of Krogman and Sassouni (1957) that "the midfacial area seems more stable than the lower facial area." These variations have been measured in this study as *within-family* differences. Other studies which would permit a comparison of interfamily differences might demonstrate the existence of *between-family* genetic variation in either or both components of facial height.

One possible explanation for the unequal contributions of the upper and lower components to measurable genetic variation in face height may be found in the growth patterns of these two areas. Lower facial development, although it continues over an extended period of time, occurs at relatively few growth sites. The ultimate form and size of the upper facial region, however, is dependent upon numerous elements which interact with one another in a complex way. (Scott, 1958) Since there are fewer growth sites within the lower face than in the upper facial complex, this would mean, presumably, that fewer genetic factors are brought to bear on the growth pattern of the lower facial component. With fewer genetic factors operating, greater *within-family* seg-

regation would occur resulting in the relatively larger dizygotic intrapair difference in lower face height observed here.

Summary

1. The relative genetic variability of various dimensions of the craniofacial skeleton has been studied by means of roentgenographic cephalometric tracings of fifty-six pairs of like-sexed twins in whom skull growth was essentially complete.

2. Highly significant hereditary variations occur in anterior cranial base, mandibular body length, total face height, and lower face height ($P < .001$).

3. These data indicate that upper face height is the more stable element in the facial profile as it does not contribute greatly to the genetic variability of the face as a whole. Lower face height demonstrates a large degree of hereditary variability.

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