DERMATOGLYPHIC INVESTIGATIONS IN TWINS AND SIBLINGS

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The objective of this study is to evaluate the dermatoglyphic differences in twin and sib pairs and to estimate the effect of genetic and intrauterine environmental factors upon dermatoglyphic development. Three sets of data were collected.

1. *Monozygotic twins*, diagnosed as such by a battery of blood-group, anthropometric, and perinatal tests. These twins have identical genetic structure and share intrauterine and gestational environmental experiences. This sample is composed of 125 male pairs and 148 female pairs.

2. *Dizygotic twins*, diagnosed by the use of the same criteria as above. These twins have a 50% genetic relationship and are for the most part under similar intrauterine and external environmental pressures. This sample involves 50 male pairs, 47 female pairs and 68 pairs of unlike sex.

3. *Siblings*. The sibs like the dizygotic twins have a 50% genetic relationship but unlike twins, sibs do not share similar intrauterine and external gestational environmental pressures. This sample involves 75 male pairs, 75 female pairs, and 75 pairs of unlike sex.

Recently, we proposed classifications of the C-line terminations (modal types of the C line) and subclassifications of the interdigital patterns. It has been shown that these features have considerable polymorphism, both between populations as well as among disease entities. The present twin and sib samples add further support to the usefulness of the newly proposed classifications. In addition, other digital and palmar dermatoglyphic features were studied.

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NORMAL AND ABERRANT PALMAR CREASES IN TWINS AND SIBLINGS

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Normal palms usually have three primary flexion creases: distal, proximal, and thenar. These creases represent important developmental landmarks. The relationship between the simian crease and Down's syndrome as well as other anomalies has been known for some time. Recently, a form of proximal crease (Sydney crease) was shown to be associated with leukemia as well as Down's syndrome. Both simian and Sydney creases tend to aggregate in families.

The present study involving MZ and DZ twin pairs, as well as paired normal sibs, was undertaken to investigate further the genetic involvement in the formation of normal palmar crease patterns, as well as simian and Sydney patterns.

In addition to the simian and Sydney creases we studied the distal and thenar creases. Two variations of normal creases were investigated: (*a*) the thenar type R (thenar crease originating at the radial border of the palm independently of the proximal crease), and (*b*) the distal type I (distal crease terminating in the interdigital area). The R and I types cluster in families. The R is found to be more frequent in the females. Both male and female MZ twins are more concordant for the R as well as the I type than their DZ counterparts or sibs. Sib studies according to parental mating type, as well as the twin data, suggest strong genetic involvement in the development of both the R and I crease types. Parents with simian and/or Sydney creases have more children with these traits than offspring of parents without these traits; however, concordance rate comparisons of MZ or DZ twins and sibs do not suggest strong genetic involvement for simian a

creases as in the case of the R and I crease types.

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PENETRANCE OF GENE FOR ABSENT C-TRIRADIUS FROM MZ TWINS

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Among 59 sets of MZ male twins and 48 sets of MZ female twins from Italy, 7 male sets (11.9%) and 7 female sets (14.6%) had absent *c*-triradius on one or more palms; among 214 individuals who had a MZ twin, 19 (8.9%) had absent *c*-triradius in one or both palms. Due to small numbers these percentages are comparable to the occurrence of absent *c*-triradius found in 8.2% of 3946 Caucasians.
When one member of a set of MZ twins showed the trait in one or both palms, 35.7% (5/14) of the other members also showed the trait in one or both palms. When an individual showed the trait in one palm, 26.3% (5/19) of the other palms also showed the trait. These two estimates of penetrance combined give an average value of 30.3% (10/33) which is comparable to an estimate of 30% found in a study of 478 family units in which transmission was compatible with an autosomal dominant gene.

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HERITABILITY STUDY ON SIZE OF THE PHYSIOLOGIC CUP OF THE OPTIC NERVE HEAD

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Cupping of the optic nerve head is regarded as one of a triad of diagnostic signs associated with glaucoma. This study was undertaken to assess the role of genetic factors in determining size of the physiologic cup of the normal optic nerve head, as measured by a horizontal cup/disc ratio. Understanding the determinants of normal variation in size of the optic cup is important to our ultimate understanding of the determinants of pathologic change.
A sample of 37 pairs of MZ and 26 pairs of like-sex DZ twins, of age 15 years and older, was studied. Horizontal cup/disc ratio was estimated clinically by stereoscopic examination of the nerve head as obtained using the Allen-Thorpe contact lens at the Haag-Streit biomicroscope. Twin zygosity was determined by blood serotyping.
Differences between MZ and DZ samples with respect to intrapair variance and intraclass correlation coefficient were highly significant and were consistent with a major genetic influence on size of the normal physiologic cup. This finding of high heritability for measurements of cup diameter is in contrast to low heritability estimates found in a companion investigation on heritability of the effect of corticosteroids on intraocular pressure. Findings of the completed study on corticosteroid hypertension will also be cited. The protocol of the latter study was originally described at the First International Symposium on Twin Studies, at which time the study was still in progress.

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