GENETIC PENETRANCE FROM MZ TWINS OF ABSENT *C*-TRIRADIUS, A PALMAR DERMATOGLYPHIC TRAIT

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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.*

When ridges flow past a common point in three directions, their junction is called a triradius. Digital triradii typically occur at the base of each finger and are designated *a* at the base of the index finger, *b* at the base of the middle finger, *c* at the base of the ring finger, and *d* at the base of the little finger. In a study of 3946 Caucasians, triradius *c* was found to be missing, on the left hand only, in about 1 in 27 persons; on the right hand only, in 1 of 48 individuals; and it was found to be missing bilaterally in 1 of 42 persons. It was missing on one or both palms in about 1 in 12 individuals. The Figure represents the right palm of the senior author showing absence of this triradius; the trait occurred bilaterally in one of four siblings, and unilaterally in one of them. At the 2nd International Congress of Human Genetics in 1961, there was a presentation (Kloepfer and Cummins 1963) on norms and heritability of palmar dermatoglyphic features based on computer analysis of 1281 German males and 768 German females. In this study, absence of *c*-triradius was thought to be under genetic control, but MZ twin material was not available for use in proposing a model for the pattern of inheritance involved.

An alternative method, proposed to estimate penetrance, was to consider the two palms as simulating a set of MZ twins, since one hand has the same genotype as the other hand just as one member of a pair of MZ twins has the same genotype as the other. Penetrance is determined by the percentage of expression of a trait among individuals who share the same genotype for that trait. When one member of a set of MZ twins shows a trait, the percentage of other members who also show the same trait is a classic method to determine an estimate of penetrance. When absence of *c*-triradius occurred in one palm in the cited study, it was absent also in 28.7% of the other palms. There was speculation whether a " twin method " applied to the two hands of individuals gives an estimate of penetrance similar to one based on MZ twin material or family studies.

Recently, when prints collected from families over a period of 20 years were reviewed, it was found that, by the family method, the material was sufficient not only to verify the estimate of penetrance published in 1963, but also to propose a possible genetic model for the inheritance of absent *c*.

The material included 1823 Caucasians from Louisiana distributed in 460 family units. Prints from

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Figure. Right palm showing absent *c*-triradius and main lines traced from triradii *a*, *b*, and *d*.

Table 1. *Occurrence of absent* c-*triradius in MZ twins* (*N* = *59 male and 48 female pairs*)

Twin A		Twin B		Male pairs	Female pairs
L	R	L	R		
0	+	+	+	4	3
0	+	0	+	1	
0	+	0	0		2
+	0	+	+	1	
+	0	+	0		1
0	0	+	+	1	
0	0	0	0		1
+	+	+	+	52	41

L = left hand, R = right hand; 0 = absence of triradius *c*, + = presence of triradius *c*.

74 additional persons distributed in 18 family units were from the files of Dr. Cummins, making prints available from 1897 Caucasians in 478 family units. All prints were formulated by the methods of Cummins and Midlo (1961). Care was taken in the formations not to misinterpret an accessory *d*-triradius for a *c*.

A genetic model based on an autosomal dominant gene with 31% penetrance was compatible with the observations of occurrence of the trait within families, whereas genetic models were incompatible when based either on an autosomal recessive gene or a sex-linked recessive gene. The estimate of penetrance by the family method was obtained by dividing the number of individuals observed to have the trait within a family by the number expected to have the heterozygous genotype for the trait under the assumption that the trait was caused by an autosomal dominant gene.

Data were compatible from four sources of family information: (1) parents of family units ascertained through an offspring with the trait; (2) offspring of family units ascertained through a parent with the trait; (3) siblings born subsequent to the first occurrence of the trait in a sibling when the trait was absent in both parents; and (4) siblings born subsequent to the first occurrence of the trait in a sibling when no information was available about the trait in one or both parents.

These data were combined to obtain an estimate of 0.312 since 54 persons presented absence of *c*-triradius on one or both palms from a total of 173 who would have been expected to have the genotype based on family relationships. The estimate of penetrance based on all available Caucasians by the "twin method" (obtained by dividing the number of individuals with the trait on both hands by the number who had the trait on either or both palms) was 95/322 or 0.295. This estimate was very similar to 0.312, and two estimates gave a weighted average of 0.301.

The purpose of the present investigation was to learn whether the estimate of penetrance based on twin material from Italy is compatible with estimates of penetrance based on Louisiana family material and an estimate of penetrance based on the two hands of individuals as simulating a set of MZ twins from the German material. A material ascertained and already studied with respect to other dermatoglyphic traits by Parisi and Bacco (1968) and Parisi et al. (1969) has been used.

Table 1 shows the frequency of presence and absence of *c* in 59 sets of male MZ twins and 48 sets

of female MZ twins. In spite of the small size of the twin material, both the percentage occurrence of absent *c*-triradius in the sample and the estimate of penetrance based on the sample are compatible with values that have been obtained in previous studies.

Table 2 shows the percentage of absent *c* in twins compared to non-twins. Since such absence in twins may be ascertained in either or two individuals and in only one individual in single births,

Table 2. *Occurrence of absent* c-*triradius in MZ twins vs. non-twins*

	Twins				Non-twins			
	Male pairs (N = 59)		Female pairs (N = 48)		Male (N = 2201)		Female (N = 1745)	
	n	%	n	%	n	%	n	%
Absence of *c*-triradius	7	11.9	7	14.6	155	7.0	168	9.6
Presence of *c*-triradius	52	88.1	41	85.4	2046	93.0	1577	90.4

it is to be expected that the trait would be observed in a higher frequency in the sets of MZ twins that in the single births.

Table 3 shows the percentage occurrence of absent *c* in the left palm only, right palm only, both palms, or neither palm, of 214 individuals who had a MZ cotwin compared to 3946 non-twin individuals. In spite of small sample size, the occurrence of absent *c*-triradius in MZ twins was comparable to the occurrence in non-twins.

Table 3. *Occurrence of absent* c-*triradius in MZ twin individuals vs. non-twins*

c-Triradius		Twin individuals (N = 214)				Non-twins (N = 3946)			
		Male (N = 118)		Female (N = 96)		Male (N = 2201)		Female (N = 1745)	
L	R	n	%	n	%	n	%	n	%
0	+	6	5.1	5	5.2	70	3.2	77	4.4
+	0	1	0.85	2	2.1	37	1.7	43	2.5
0	0	1	0.85	4	4.2	48	2.2	48	2.8
+	+	110	93.2	85	88.5	2046	93.0	1577	90.4

L = left hand, R = right hand; 0 = absence of *c*-triradius, + = presence of *c*-triradius.

When one member of 14 sets of MZ twin pairs had absent *c*, the other member also had absent *c* in 35.3% of the pairs. When absent *c* occurred in one palm of 19 individuals who were members of a twin pair, *c* was absent in 26.3% of the other palms of the same individual. These two estimates of penetrance may be combined to give a value of 30.3% which is compatible with the estimate of 30.1% found in family studies and in studies of the two palms of non-twins.

It is common practice for investigators with a background in plant or animal genetics to illustrate inheritance in man by the selection of an atypical gene which is 100% penetrant. Since the dominant gene accounting for the occurrence of absent *c* is about 30% and not 100% penetrant, this gene is more typical of genes of concern to man, which typically also are not 100% penetrant.

Although the proposed model for the inheritance of absent *c* best fits all available data, experience from the study of genes which affect the health of man indicates that pathological conditions typically are not determined solely by a single chromosomal locus. There is no a priori reason to believe that all individuals with absent *c* have the same genotype.

This effort to estimate the penetrance of an autosomal dominant gene for absent *c*-triradius from MZ twin material may be summarized as follows. If dermatoglyphic findings based on the present small sample are confirmed by similar findings from a larger MZ twin sample, it may be concluded that the estimates of penetrance obtained from family studies give values similar to estimates based on MZ twin material.

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