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A Family Study of Dermatoglyphic Traits in India: Segregation Analysis of Accessory Palmar Triradii and the atd Angle

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ABSTRACT Accessory triradii and the atd angle were examined via complex segregation analysis in order to evaluate possible genetic effects on these dermatoglyphic traits, measured in an endogamous Brahmin caste of peninsular India. The phenotypes considered included: presence of accessory palmar triradii a' and d', associated with the interdigital areas II and IV, respectively; presence of an accessory axial triradius tt' associated with the proximal margin of the palm; and an arctanh-transformation of the atd angle measurement. For all accessory triradii considered in the present investigation familial resemblance was evident. The most parsimonious model which could account for the observed resemblance was a multifactorial model that includes polygenic effects as well as transmissible environmental effects that are inherited in the same pattern as polygenes. Evidence of familial resemblance was also found for the arctanh-transformed atd angle, which could be attributed, initially, to both a major effect and a multifactorial component. Tests of transmission of a putative major gene were performed which yielded results consistent with Mendelian transmission, although an alternative test of no transmission of the major effect also fit the data. In light of these contrasting results we are precluded from accepting with confidence the notion of a major gene influence on the atd angle. We have concluded that the accessory triradii a', d', and tt', and the atd angle are influenced by multifactorial effects, including additive polygenes and possible environmental factors, such as intrauterine effects.

Four digital triradii are found characteristically in the distal palmar region in man. These triradii are situated in proximal relation to the base of digits II, III, IV, and V and are designated, in radio-ulnar sequence, a, b, c, and d, respectively. In the interdigital spaces II and IV additional triradii, termed accessory triradii, may occur occasionally. These accessory triradii are most related to triradii a and d and are designated a' and d'. Triradii a' and d' occur usually alone on a single palm; very rarely they may occur on the same palm.

The genetics of accessory triradii a' and d' were investigated by Bansal and Rife (1962), who studied 53 families of Punjabi Khatris, in which one or more family members exhibited at least one accessory triradius or vestige in interdigital area II. Bansal and Rife concluded that the presence or absence of

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accessory triradii was under the control of a single major locus with two alleles, in which the presence of accessory triradii was dominant. It is noted here that no further investigations were made to examine the repeatability of the genetic results obtained by Bansal and Rife, although Loesch (1971) did not confirm their association between interdigital loops II and IV.

Accessory triradii in another palmar area have also been observed. Axial triradii (t) are located in the interval between the thenar and hypothenar areas, corresponding anatomically to the narrow field aligned with the axis of the fourth digit (Cummins and Midlo, 1976). Axial triradii are located most frequently at or near the proximal margin of the palm, although they may occur as far distal as the center of the palm. On most palms only a single triradius t occurs. On the remaining palms more than one axial triradius occurs; these additional triradii are designated accessory axial triradii (tt') (Cummins and Midlo, 1976; Malhotra et al., 1981). Incidence rates of accessory axial triradii range widely depending on the population surveyed. Low frequencies of the trait have been observed in Oriental and Amerindian populations (2–5%), intermediate rates in American Black, Caucasian, and Asian Indian samples (5–18%), and higher rates in Australasians (3–28%) (Plato et al., 1975a,b; Steinberg et al., 1975).

Cummins (1939) demonstrated for the first time a strong association between centrally located axial triradii and Mongoloidism (Down's Syndrome). The patterns in the palmar hypothenar region are also intimately related to the number and location of axial triradii on the palm (Malhotra et al., 1981). As yet, it is unknown whether or not the occurrence of accessory axial triradii, irrespective of palmar location, is under genetic control.

The designation of presence or absence of accessory palmar triradii represents qualitative phenotypes. The relative locations of triradii may be quantified by measurement of the atd angle (Penrose, 1949, 1954). The atd angle is defined by Penrose (1954) as the angle subtended at the most distal t triradius by the most medial d triradius and the most lateral a triradius on each hand. The size of the atd angle has been shown to be related closely to the presence or absence of the hypothenar pattern, which has been found to be a familial trait (Weninger, 1947). Penrose (1954) remarked that the atd angle measurement, if used as the only means of diagnosis of Mongoloidism, would misclassify only 12% of the affected individuals. Penrose (1954) conducted an investigation of the atd angle on a control population of 2,046 individuals and on 235 Mongoloid probands and members of their families. He concluded that one or more genes which influence the atd angle also affect the susceptibility to Mongoloidism.

The purpose of this investigation is to examine the inheritance of the occurrence of accessory triradii a', d', and tt' and of the atd angle. Complex segregation analysis was performed on family data derived from an endogamous Brahmin caste of peninsular India.

MATERIALS AND METHODS

Data and variables

The data have been described elsewhere in considerable detail (Borecki et al., 1985). To summarize, dermatoglyphic prints were taken on 625 related individuals from an endogamous population. Dermatoglyphic prints were taken by the same investigators, which minimized possible variation in measurements. The family series consists of 125 nuclear families with 375 offspring, sampled from the Telaganya Brahmin caste residing in Waltair, Andra Pradesh.

Dermatoglyphic traits considered for analysis here include accessory palmar triradii a' and d', associated with the interdigital spaces II and IV, respectively, the accessory axial triradius tt', and the atd angle, averaged over both palms. The correlation between angle measurements of the right and left palms was 0.70. With regard to the accessory triradii, the phenotypes considered are qualitative; an individual is considered affected if an accessory triradius is present *on at least one palm*. The incidence values for the various phenotypes determined for the accessory triradii in the T. Brahmin population and in the Punjabi Khatris (Bansal and Rife, 1962) studied here are presented in Table 1.

TABLE 1. Incidences of accessory palmar triradii

Trait	Incidence	
	T. Brahmin population	Punjabi Khatris
a'	0.15	0.17
d'	0.34	0.38
a' and/or d'	0.39	0.44
tt'	0.21	—[1]

[1]Not studied by Bansal and Rife (1962).

The atd angle, measured in degrees, is not a linear measurement, and therefore was transformed to linear values prior to data analysis: $z = \text{arctanh}\ (4\theta/\pi - 1)$ where θ = atd angle in radians.

Five individuals with missing values were excluded from analysis, leaving 620 valid observations. The arctanh-transformed atd angle, standardized to zero mean and unit variance prior to analysis, demonstrated significant skewness and kurtosis.

Commingling analysis

Commingling analysis is used to distinguish between spurious skewness and mixtures of distributions. MacLean et al. (1976) suggested the use of a power transformation to remove the effects of skewness, since spurious skewness may be interpreted as a mixture of distribution due to a major locus. Thus, the use of a power transform provides a more conservative test of the major gene hypothesis.

For a standardized score x, the power transformed score y is given by

$$y = \begin{cases} r/p\,[(x/r + 1)^p - 1], \text{ if } p \neq 0 \\ r \ln (x/r + 1), \text{ if } p = 0 \end{cases}$$

where the constant r is chosen such that every $(x/r + 1)$ is positive. The computer program SKUMIX estimates the value of p under the assumption of one, or a mixture of two or three distributions.

Parameters of the model are estimated by the method of maximum likelihood, and tests of hypotheses are carried out using the likelihood ratio test. The test criterion, given by twice the difference between the log-likelihoods of a pair of compared hypotheses is distributed asymptotically as a chi-square whose degrees of freedom are equal to the difference in the number of parameters estimated in the two models.

For the average, arctanh-transformed atd angle, a power transformation value was estimated after the variable was standardized to mean zero and unit variance under the assumption of a single distribution.

Segregation analysis

Segregation analysis was performed using the unified mixed model, implemented in the computer program POINTER (Lalouel et al., 1983; Lalouel and Morton, 1981; Morton and MacLean, 1974). This model assumes that a trait y results from one or more of three sources: 1) a major transmissible effect, g, (which may or may not be a Mendelian gene); 2) a multifactorial transmissible component, c; and 3) a random, nontransmitted environment, e, with $y = g + c + e$. These three factors are assumed to be independently distributed. Factors c and e are assumed to be normally distributed, N(0,C) and N(0,E), respectively. The major effect is modeled as a single locus with two alleles A and a, leading to three genotypic classes. The prior probability of allele a in the reference population is q. The distance between the two homozygous means on the scale of y is called t, the displacement. The position of the heterozygous mean relative to both homozygous classes is represented by d, the dominance parameter. The expected value of g is u, the overall mean. The variance of y is denoted by V such that $V = G + C + E$, where G is the variance due to the major effect. Multifactorial heritability is defined by $H = C/V$. Thus, in the mixed model, as used here, there are six parameters under the assumption of Mendelian transmission of the major effect: V, u,d, t, q, H. In the analysis of qualitative traits V and u are fixed, arbitrarily to 1 and 0, respectively. However, specification of the unified mixed model also requires the definition of transmission rules from parent to offspring. Random mating is assumed. The transmission probabilities τ_1, τ_2, and τ_3 denote, respectively, the probabilities of the genotypes AA, Aa, and aa transmitting the A allele. Under Mendelian transmission, $\tau_1 = 1$, $\tau_2 = 1/2$, and $\tau_3 = 0$. These transmission probabilities are also estimable.

Parameters are estimated by the method of maximum likelihood, and null hypotheses are tested using the likelihood ratio criterion. Setting all parameters but the mean and variance to zero provides a test of no familial transmission. The hypothesis of no major effect corresponds to $d = t = q = 0$; H is fixed at zero for the test of no multifactorial heritability. If a major effect is inferred, it is important to test for Mendelian transmission before concluding that the major effect is a major gene. This test may be performed by iterating τ_2, or more generally, by iterating all three transmission probabilities, in addition to the other relevant parameters of the mixed model. A test of no transmission of a major effect may also be conducted by constraining the three transmission probabilities to be equal.

RESULTS

Accessory triradii

Sex differences in the incidence of accessory triradii were observed for the accessory a' triradius (right palm) and the accessory axial triradius (left palm). Over both sexes, significant differences were observed between the left and right palms for the accessory a' and d' triradii. Segregation analysis was performed using sex-specific incidences or on each palm separately, if sex or palm differences in incidences warranted special analyses. In order to present results in a concise and consistent manner, analyses of the phenotype defined as presence on at least one palm, without regard to possible sex differences, are presented, since the results of both analyses were qualitatively comparable.

The results of segregation analysis of the accessory triradius a' are presented in Table 2. The data are best fit by a multifactorial model (d = t = q = 0). No significant improvement in the likelihood could be achieved by the addition of the parameters which describe a major effect.

For the accessory triradius d' although a dominant model has the best likelihood (Table 2), a more parsimonious model, with only a multifactorial component fits the data equally well.

When affection status is considered as presence of the a' and/or d' accessory triradii (Table 2), the significant evidence for familial resemblance is explained most parsimoniously by a multifactorial component.

Segregation analysis of the accessory axial triradius tt' yields evidence for familial resemblance, again accountable by a multifactorial component. The major gene model leads to no significant improvement in the likelihood. A converged solution for the mixed model was unobtainable for these data.

In their analysis of 53 nuclear families, Bansal and Rife (1962) had concluded that presence of accessory palmar triradii located in the second and fourth interdigital areas (a' and d') was influenced by a single dominant gene with incomplete penetrance. Because of the discrepancies in the results of analysis of our data and of those reported by Bansal and Rife (1962), segregation analysis of the data studied by Bansal and Rife was undertaken employing the unified mixed model, and results are presented in Table 3. For the accessory triradius a' the data best fit a recessive model, although the most parsimonious model which accounts for the observed resemblance is a model with only a multifactorial component. Significant evidence is found for familial resemblance. For the presence of a' and/or d' accessory triradii, the hypothesis of no familial resemblance cannot be rejected.

Average atd Angle

Commingling analysis performed under the assumption of a single distribution yields significant evidence of skewness ($\chi_1^2 = 10.60$). The power transform value estimated under

TABLE 2. Complex segregation analysis of accessory palmar triradii in T. Brahmin population

	Trait									
	a'					d'				
Hypothesis	−2lnL + C	d	t	q	H	−2lnL + C	d	t	q	H
Mixed model	1.96	0.78	2.04	0.25	0.13	0.00	1.00	1.41	0.28	0.01
No familial resemblance	27.73	(0)[1]	(0)	(0)	(0)	34.45	(0)	(0)	(0)	(0)
No major effect	0.00	(0)	(0)	(0)	0.86	3.54	(0)	(0)	(0)	0.63
No multifactorial component	2.07	0.76	2.35	0.21	(0)	0.09	1.00	1.42	0.28	(0)
	a' and/or d'					tt'				
Mixed model	0.00	1.00	1.49	0.26	0.01	—[2]				
No familial resemblance	51.62	(0)	(0)	(0)	(0)	11.28	(0)	(0)	(0)	(0)
No major effect	1.21	(0)	(0)	(0)	0.80	0.32	(0)	(0)	(0)	0.44
No multifactorial component	0.08	0.93	1.58	0.27	(0)	0.00	0.84	1.99	0.36	(0)

[1]Parameters set in parenthesis were fixed at value shown.
[2]Unable to obtain converged solution.

TABLE 3. Complex segregation analysis of accessory palmar triradii in Punjabi Khatri population (data from Bansal and Rife, 1962)

	Trait									
	a'					a' and/or d'				
Hypothesis	−2lnL + C	d	t	q	H	−2lnL + C	d	t	q	H
Mixed model, d = 0	0.00	(0)[1]	3.06	0.96	0.26	0.00	(0)	8.14	0.12	0.00
Mixed model, d = 1	2.64	(1)	2.93	1.00	0.68	3.40	(1)	1.17	0.37	0.01
No familial resemblance	19.31	(0)	(0)	(0)	(0)	5.96	(0)	(0)	(0)	(0)
No major effect	1.80	(0)	(0)	(0)	0.86	3.26	(0)	(0)	(0)	0.31
No multifactorial component	1.49	(0)	1.49	0.49	(0)	2.76	(0)	2.47	0.39	(0)

[1]Parameters set in parentheses were fixed at value shown.

TABLE 4. Complex segregation analysis of arctanh-transformed average atd angle

Hypothesis	−2lnL + C	V	u	d	t	q	H	τ_1	τ_2	τ_3
Mendelian mixed model	0.00	1.48	0.46	0.00	6.87	0.11	0.42	(1)[1]	(1/2)	(0)
No familial transmission	140.66	1.17	0.08	(0)	(0)	(0)	(0)	—	—	—
No major effect	80.87	1.38	0.74	(0)	(0)	(0)	0.71	—	—	—
No multifactorial component	60.61	2.57	0.41	0.15	6.97	0.20	(0)	(1)	(1/2)	(0)
$\tau_3 = 1/2$	−1.64	1.82	0.51	0.00	6.75	0.13	0.38	(1)	0.69	(0)
Generalized transmission	−1.65	1.62	0.47	0.00	6.62	0.12	0.41	0.91	1.00	1.00
No Mendelian transmission is constrained to be equal	−0.22[2]	1.55	0.47	0.00	6.78	0.11	0.41	0.93	0.93	0.93

[1]Parameters set in parentheses were fixed at value shown.
[2]Note: This model is compared with the model in which all three transmission probabilities are estimated.

the assumption of a single distribution is 0.79. The value is used to remove skewness in the average atd angle prior to segregation analysis; thus, more conservative tests of genetic hypotheses are achieved.

Segregation analysis was performed on both untransformed and power transformed data and on each palm separately. Only the results of the analysis of the power transformed data averaged over both palms is presented here because both sets of analyses led to qualitatively similar conclusions. Parameter estimates and model likelihoods are presented in Table 4.

Significant evidence is demonstrated for familial resemblance ($\chi_4^2 = 140.66$), for a major effect ($\chi_3^2 = 80.87$) and for a multifactorial component ($\chi_1^2 = 60.61$). Tests of transmission were performed to evaluate whether or not the major effect behaved as a major locus. The test of the transmission probability, τ_2, the probability that an Aa heterozygote transmits the A allele, yields no significant difference from Mendelian expectations ($\chi_1^2 = 1.64$; $\tau_2 = 0.687$). Simultaneous estimation of all three transmission probabilities also gave results consistent with Mendelian transmission ($\chi_3^2 = 1.65$), although the estimates of the transmission parameters appeared to be curiously non-Mendelian ($\tau_1 = 0.91$, $\tau_2 = 1.00$, $\tau_3 = 1.00$). An additional hypothesis of no transmission of the major effect was also carried out by constraining the three transmission probabilities to be equal. Hypothesis testing is achieved by comparing the likelihood of this model with that of the model where all three transmission probabilities are estimated. The hypothesis of no transmission could not be rejected ($\chi_1^2 = 0.22$). The common transmis-

sion parameter was estimated at 0.93. Thus, we cannot conclusively demonstrate that a single major locus influences the atd angle measurement.

DISCUSSION

Various investigators have examined the inheritance of accessory triradii and the atd angle (Bansal and Rife, 1962; Penrose, 1949, 1954; Weinand, 1937; Weininger, 1935). All studies suggested that genetic factors influence the expression of these phenotypes. However, as Loesch (1971) points out, the conclusions put forth by the series of studies are inconsistent. Some studies suggest major gene effects, with varying degrees of penetrance, while others support polygenic inheritance.

The purpose of the present investigation is to examine the inheritance of accessory triradii and the atd angle via complex segregation analysis. We incorporate several tests of Mendelian segregation of putative major loci, thereby challenging detectable major effects to comply with known biological rules of transmission.

Analysis of the accessory triradius a' demonstrates that the observed familial resemblance may be attributable to a multifactorial component. Significant evidence for a multifactorial component is found also for the accessory triradius d'. Segregation analysis of the accessory axial triradius tt' yields significant evidence for familial resemblance, also attributable to a multifactorial component.

When the phenotype is considered as the presence of either an accessory triradius a' and/or d', significant evidence for familial resemblance is obtained, which is best accounted for by a multifactorial component. Bansal and Rife (1962) concluded, however, that the presence of these accessory triradii was determined by a dominant allele with 88% penetrance, a conclusion that we could not replicate with their own data. This discrepancy may be attributable to the fact that the sample of Bansal and Rife (1962) was ascertained if at least one family member exhibited an accessory triradius in the interdigital area II (a'). In their segregation analysis of the data, however, ascertainment of the sample was not taken into account.

Segregation analysis has been shown to be sensitive to skewness in quantitative traits, in that spurious evidence for a major gene may result. In addition, simulation studies performed by Demenais et al. (1985) emphasize the utility of the tests of transmission to guard against false inference of a major locus. Therefore, we elected to employ the power transformation of MacLean et al. (1976) to remove the significant skewness detected in the commingling analysis of the arctanh-transformed average atd angle. Significant evidence was found for both a polygenic component and for a major effect. It is emphasized here that even simultaneous estimation of all three transmission probabilities lent evidence consistent with Mendelian transmission. However, non-Mendelian transmission was also compatible with the data. Thus, we cannot confidently accept the hypothesis of a major gene.

We conclude that the etiology of accessory triradii are influenced by additive polygenes with heritabilities in the range of 44–86%. Estimates of heritability are obtained in segregation analysis under the assumption of no major effect ($d = t = q = 0$). Possibly individual-specific environmental factors, such as intrauterine effects, influence these phenotypes as well. It is not apparent, though, that major genes are involved significantly in the genetic etiology of these traits.

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LITERATURE CITED

Bansal, P, and Rife, DC (1962) The inheritance of accessory triradii on palmar interdigital areas II and IV. Acta Gen. Et. Gemell. *11*:29–37.

Borecki, IB, Malhotra, KC, Mathew, S, Vijayakumar, M, Poosha, DVR, and Rao, DC (1985) A family study of dermatoglyphic traits in India: Resolution of genetic and uterine environmental effects of palmar pattern ridge counts. Am. J. Phys. Anthropol. *68*:417–424.

Cummins, H (1939) Dermatoglyphic stigmata in mongoloid imbeciles. Anat. Res. *73*:407–415.

Cummins, H, and Midlo, C (1976) Finger Prints, Palms, and Soles. An Introduction to Dermatoglyphics. 3rd Ed. Research Publishing Co., Inc., Mass.

Demenais, F, Lathrop, M, and Lalouel, JM (1986) Robustness and power of the unified mixed model in the analysis of quantitative measurements. Am. J. Hum. Genet. *38*:228–234.

Lalouel, JM, and Morton, NE (1981) Complex segregation analysis with pointers. Hum. Hered. *31*:312–321.

Lalouel, JM, Rao, DC, Morton, NE, and Elston, RC (1983) A unified mixed model for complex segregation analysis. Am. J. Hum. Gen. *35*:816–826.

Loesch, D (1971) Genetics of dermatoglyphic patterns on palms. Ann. Hum. Gen. *34*:277–293.

MacLean, CJ, Morton, NE, and Lew, R (1976) Analysis of family resemblance. IV. Operational characteristics of segregation analysis. Am. J. Hum. Gen. *27*:365–384.

Malhotra, KC, Reddy, BM, Karmakar, B, and Vijayakumar, M (1981) Relationship between types of axial triradii and the true hypothenar patterns. Dermatoglyptics *9*:32–55.

Morton, NE, and MacLean, CJ (1974) Analysis of family resemblance. III. Complex segregation analysis of quantitative traits. Am. J. Hum. Gen. *24*:489–503.

Penrose, LS (1949) Familial studies on palmar patterns in relation to mongolism. Proc. 8th Int. Conf. Genet., Hereditas, London (Suppl. Vol), p 412.

Penrose, LS (1954) The distal triradius t on the hands of parents and sibs of mongol imbeciles. Ann. Hum. Gen. *19*:10–38.

Plato, CC, Brown, HA, and Gajdusek, DC (1975a) The dermatoglyphics of the Elema people from the gulf district of Papua New Guinea. Am. J. Phys. Anthropol. *42*:241–250.

Plato, CC, Cereghino, JJ, and Steinberg, FS (1975b) The dermatoglyphics of American Caucasians. Am. J. Phys. Anthropol. *42*:195–210.

Steinberg, FS, Cereghino, JJ, and Plato, CC (1975) The dermatoglyphics of American Negroes. Am. J. Phys. Anthropol. *42*:183–194.

Weinand, H (1937) Familienuntersuchungen über der Hautleistenverlauf der Handflache. Ztsch. F. Morph. U. Anthropol. *36*:418–442.

Weninger, M (1935) Familienuntersuchungen über der Hautleistenverlauf am Thenar und am ersten Interdigitalballen der Palma. Mitt. Anthropol. Ges. Wien. *55*:182–193.

Weninger, M (1947) Zur Vererbung der Hautleistenmuster am Hypothenar des menschlichen Hand. Mitt. Anthropol. Ges. Wien. *73*:55–77.