

DISTAL HYPEREXTENSIBILITY OF THE THUMBS

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Among anatomical oddities, none would seem to be much more amusing or less consequential than doublejointedness at the terminal joint of the thumb (Fig. 1). This trait, technically termed distal hyperextensibility of the thumb, might more colloquially be called "hitchhiker's thumb." Although it has been previously mentioned in genetic literature as probably hereditary, no thorough investigation of the trait appears to have been made. *L. F. Whitney* [1932], in discussing doublejointedness at the proximal joint of the thumb, stated that he had also collected some data on the distal type of doublejointedness, and added: "This characteristic also runs in families, and it is interesting that it is inherited independently of the doublejointedness in the second joint." The projected paper, however, has never appeared. *D. D. Whitney*, in his book entitled *Family Treasures*, figures the condition in a mother and son (p. 157), and remarks that it is rather common and probably inherited (*D. D. Whitney* [1942], p. 163).

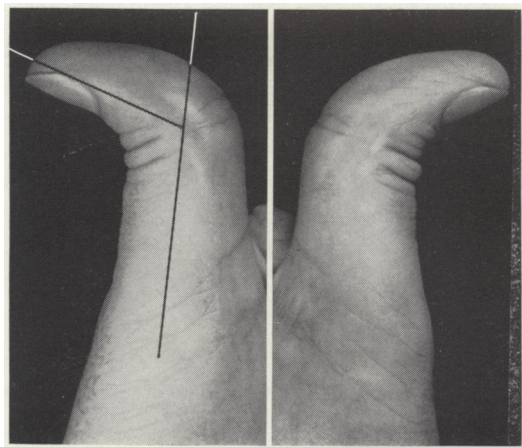


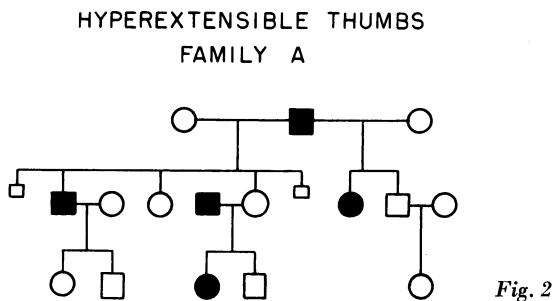
Fig. 1 A

Fig. 1 B

Distal hyperextensibility of the thumbs does not appear to be affected by age or sex. The senior author, whose own thumbs are hyperextensible at the distal joints (see Fig. 1), can remember wondering about the peculiarity from his earliest childhood. In his father, at 77 years of age, it is equally marked. In spite of the commonness of the trait, however, most possessors of such thumbs suppose them rare, and are even surprised to find that other members of their own families are similarly characterized. Except in rare instances, persons with distally hyperextensible thumbs show no other type of doublejointedness. They are not in general loose-jointed.

X-ray photographs of normal thumbs were compared with those of distally hyperextensible thumbs from two individuals, one of whom had both thumbs and the other only a single thumb doublejointed in this characteristic way. It appeared that the distal end of the proximal phalanx is rather flatter and bears a sharper angle on the dorsal side, whereas the corresponding face of the proximal phalanx in a distally hyperextensible thumb exhibits a more rounded angle. More cases will have to be studied to substantiate such an interpretation, but it seems that the trait may have a skeletal basis and may not be due simply to longer ligaments.

The senior author's study of the trait in his own family (Fig. 2)



indicated that distal hyperextensibility of the thumbs is hereditary. Its unbroken transmission might be taken for that of a simple autosomal dominant. However, its reintroduction into one branch of the family by marriage might imply that it is quite common. Examined from that point of view, it will be seen that there is no evidence from this pedigree to rule out the possibility that the trait is a simple autosomal recessive. In that case, a large proportion of the population

should be heterozygous, and the trait itself extremely common. A survey of several samples of the population was therefore undertaken.

The method used was the following. A transparent plastic protractor was reddened along the straight edge and two red lines were drawn parallel to this edge and respectively 0.5 and 1.0 cm from it. These assisted in aligning the straight edge of the protractor parallel to the posterior (dorsal) surface of the thumb, the hand being held palm upwards, and the midpoint of the protractor being placed on the end of the palmar skin folds, at the center of the distal joint of the thumb. The angle was then read along the line from this point to the corner or most laterally projecting part of the thumb-nail (Fig. 1). All persons who had either thumb extensible to a 50° angle or greater were classified as possessors of the trait, all others as non-affected.

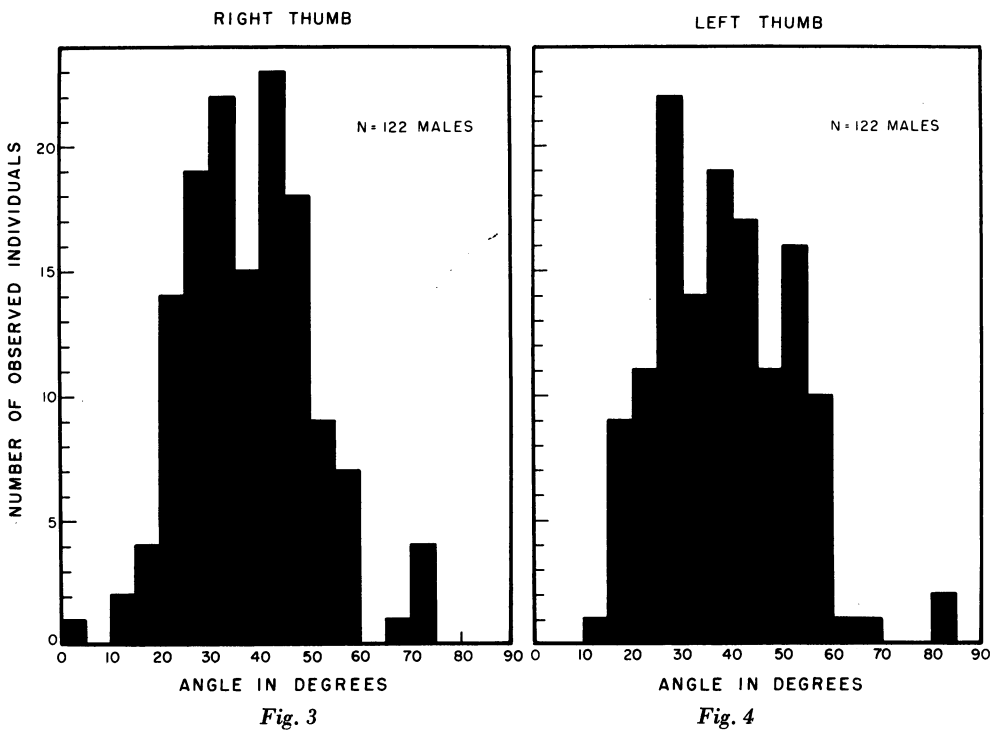
The first sample consisted of students at the Johns Hopkins University, who represented quite an extensive geographic area but came preponderantly from the Middle Atlantic States, with a large local contingent. This sample was compared with a more strictly local sample comprising mainly male students at a large high school in Baltimore, but including some adults of both sexes. Sibs and other close relatives were excluded. The third sample consisted of both male and female students at Morgan College in Baltimore. These represented the American Negro population. It soon became apparent that the trait is far commoner than had been supposed, and also, that some persons have only a single distally hyperextensible thumb, which may be either the right or the left. All persons with either thumb, or both thumbs, distally hyperextensible were considered in the enumeration to possess the trait. The frequencies found in the three samples of the population, and the penetrance, calculated on the assumption that non-expression of the condition in the right and left hands is independent, are given in Table 1. Approximately one-fourth of the white population and one-third of the Negro population examined consist of persons with distally hyperextensible thumbs. The penetrance is high. Only 3.5 per cent of those who carry the genotype for distally hyperextensible thumbs fail to manifest the trait on at least one hand or the other. The difference between the samples of the white population is not significant, but the frequency of the trait is significantly higher in the Negroes than in the Whites.

Following the original preparation of this paper and its presentation at the meeting of the Human Genetics Society in Minneapolis,

in September 1951, the paper of *Harris and Joseph* [1949] on the incidence of the same trait in 133 male and 100 female Europeans, 31 male Indians, and 30 male West Africans came to our attention. *Harris and Joseph* carried out their study by means of measurements on radiographs, and used the angle between the posterior surfaces of the proximal and distal phalanges as a standard measurement. They said that "sometimes the proximal phalanx does not show a well-defined straight line posteriorly, but in such cases a line may be drawn parallel to the greatest thickness of the cortical bone. The axis for the curved distal phalanx was more difficult to determine, since the phalanx is always curved in a posterior direction. However, there is nearly always a straight line of cortical bone about one millimetre wide on the posterior aspect, and a line drawn on the posterior surface of the phalanx parallel to this was chosen as the axis. It might be argued that the greater the curvature of the distal phalanx the greater should be the angle between it and the middle phalanx. It was found, however, that the curvature did not vary enough to affect the results materially." The angle was then measured to the nearest half degree. For the male Europeans the mean angle for the interphalangeal angle of the right thumb was 29.16° ($\sigma = 11.65^\circ$), and for the left thumb it was 32.20° ($\sigma = 11.09^\circ$). Graphs of the separate distributions for the two thumbs show fairly symmetrical continuous distributions, with a slight indication of a skewed or bimodal type of distribution.

This puzzling fact, in the light of the indication that the inheritance of the trait might be simple, led us to make a study (a) of the distribution of the interphalangeal angles from external measurements, and (b) of the comparative agreement between such measurements made independently by three observers on the same group of subjects. It was our hope that sufficient agreement and precision might be found in taking external measurements to obviate the need for measurements based on roentgenograms. Clearly, if the latter were essential, the trait would not be very useful to physical anthropologists and geneticists in field studies of human populations.

Measurements were made upon 122 male individuals, not the same persons as those in the earlier study. The method was the same as already described. The distributions obtained for right and left thumbs separately are shown in Figs. 3 and 4. The mean for the left thumb was $41.5^\circ \pm 0.85^\circ$ ($\sigma = 13.9^\circ$); and for the right thumb,



$43.5^\circ \pm 0.825^\circ$ ($\sigma = 13.55^\circ$). It is interesting that although the median and range for the left thumb are higher than the median and range for the right, as in the data of *Harris* and *Joseph*, nevertheless the mean for the left thumb is lower than that for the right, contrary to the finding of *Harris* and *Joseph*. This is due to the asymmetry of the distributions. The differences between the means in the present series is not significant; and it is therefore to be presumed that the difference found by *Harris* and *Joseph* has no real meaning, either.

The difference between the means of the British workers' series and ours is 11.82° , owing to the different nature of the measurement. The ranges show the same general difference:

<i>H.</i> and <i>J.</i>	left	$5^\circ - 70^\circ$	right	$0^\circ - 65^\circ$
<i>G.</i> and <i>K.</i>		$10^\circ - 85^\circ$		$0^\circ - 75^\circ$

The 50° angle taken as the criterion of distal hyperextensibility of the thumb would thus become about a 38° angle according to the standard measurement used by *Harris* and *Joseph*.

In our earlier study we classified each individual who had one

or both thumbs distally hyperextensible as a possessor of the trait, in contrast to persons who had neither thumb distally hyperextensible. It is unfortunately impossible from the manner in which *Harris* and *Joseph* have presented their data to make a similar classification of their subjects; although they did report a high correlation between right and left joints ($r = 0.66$ in males and 0.81 in females). When our new series of 122 male subjects was classified in the same way as the first, 30 (24.6 per cent) were found to have one thumb exceeding 50° in extension. This is in excellent agreement with the result obtained in the first series of 221 male subjects (see Table 1).

Table 1. A. Frequency of Persons with Hyperextensible Thumbs (dht) in Three Samples of the United States Population.

	dht	non-dht	Total	Percentage dht $\pm$ S.E.
White: total	221	674	895	24.7 ± 1.44
J.H.U.	85	294	379	22.4
B.C.	136	380	516	26.3
Negro: M.C.	56	101	157	35.6 ± 3.8
χ^2 between two white samples, 1.82. P, .20 — .10				
χ^2 between total white and negro, 8.36. P < .01				

B. Penetrance of Hyperextensible Thumb (J.H.U. sample)

	Both	R. only	L. only	Total
Frequency	51	23	11	85
	.60	.27	.13	
Coincidence of non-expression	$.27 \times .13 = .035$			
Penetrance	$100.0 - 3.5 = 96.5\%$			

The three observers (*Ray A. Wilson*, *Lowell R. King*, and *Louis A. Mucelli*), to whom the authors acknowledge their indebtedness for the measurements on the second series of 122 male individuals, supplied a total of 726 deviations of measurements (3 observers $\times$ 122 individuals $\times$ 2 thumbs per individual — 6 for thumbs missing). The modal deviation was 1° , and the median deviation fell at 4° . Ninety-five per cent of the deviations were 12° or less, but it is evident that the accuracy of measurement is by no means fine. This is due to several factors. Subjective errors may be made in aligning the straight edge of the protractor with the back side of the proximal joint of the thumb, or in the exact placement of the center of the

protractor. If the subject tries hard to bend the thumb back as far as possible, fatigue rapidly sets in and the reading may decrease a few degrees. The subject will also, when asked to extend the thumb to the maximum, vary from time to time in the amount of extension. Subjects who are double-jointed at the base of the thumb (metacarpophalangeal joint) are particularly difficult to measure, because the degree of distal hyperextensibility is markedly affected by the position of the proximal joint at the time. This was also reported by *Harris* and *Joseph* in their study.

Harris and *Joseph* also checked the reliability of measurements by remeasuring 10 subjects on new radiographs made after an interval. The deviations ranged up to 10° , although most were under 6° . Therefore it seems that the accuracy of measurement is slightly better than in the case of our external measurements, but not enough so to warrant the extra trouble required. Two observers also independently measured the same ten radiographs three successive times. In general, as in our observations, the same observer repeated a given measurement closely, but several times there were deviations of 3° or 4° ; and the two observers differed as much as 6.5° in one instance. It does appear that the considerable error involved in the external measurements makes classification of individuals uncertain unless measurements are checked by repeated trial. In a sufficiently large sample, however, the error becomes statistically unimportant. It is also worth mention that when persons rather than individual thumbs are the basis of classification, there is less error, since misclassification of a person would usually occur only when there was a coincidence of errors in measuring both the right and the left thumb. In the data of *Harris* and *Joseph* [1949] for 30 male West Africans, the mean angle of distal extensibility of the right and left thumbs was 7.35° and 10.13° greater than in the sample of male Europeans. Although the individual data are not given, there would consequently be in their sample too a greater percentage of Negroes than of Whites classified according to our criteria as having distally hyperextensible thumbs. The 31 male Indians they measured also exceeded the male Europeans, by 7.54° in the mean for the right thumb and 7.56° for the left.

Tests for Mode of Inheritance.

Data were collected on the proportions of offspring with and without distally hyperextensible thumbs (dht), in the three possible types of mating: dht $\times$ dht; dht $\times$ non-dht; and non-dht $\times$ non-dht.

These data were then tested (Table 2) by the formulae derived by Snyder [1934] from the *Hardy-Weinberg* principle. These formulae give the proportions, for a population breeding at random, to be expected for a single autosomal recessive gene in the matings of (1) dominant $\times$ dominant and (2) dominant $\times$ recessive. If dht is a simple recessive, then the mating dht $\times$ dht should produce only dht offspring. Actually, one exception occurred among 24 offspring in such families; but precisely such a frequency of apparent exceptions is to be expected for hyperextensibility of the thumbs, on account of its incomplete penetrance. For families in which the parents were dht $\times$ non-dht, the expected proportion of recessives is $q/(1+q)$, where q is the gene frequency of the recessive allele. The value of q may be derived from the frequency of the trait in the population, for if the trait is recessive, its frequency according to the *Hardy-Weinberg* principle will be q^2 . Hence the frequency of a recessive gene is the square root of the frequency of possessors of the trait in the population. For families in which both parents are dominant, the expected frequency of recessive type offspring is $\left(\frac{q}{1+q}\right)^2$. The agreement found between the calculated values and those actually observed is excellent.

Table 2. Test for Mode of Inheritance by Sibship Formulae.

Type of parents	Number of families	dht	Offspring non-dht	Proportions dht	among offspring non-dht
dht $\times$ dht	11	30	1	.968	.032
dht $\times$ non-dht	48	37	71	.343	.657
non-dht $\times$ non-dht	132	32	281	.102	.898

A. Test for Simple Recessive Inheritance

	$q^2 = .247$		$q = .496$	Exceptions: 1/24	
$\frac{q}{1+q}$	Observed 37 71		$\left(\frac{q}{1+q}\right)^2$	Observed 32	281
	Expected 35.9 72.1			Expected 35	278
	$\chi^2 = .05$ n = 1			$\chi^2 = .29$ n = 1	
	P = .90—.80			P = .70—.50	

B. Test for Simple Dominant Inheritance.

(If dht is dominant, then $q^2 = \text{non-dht} = .753$)

	$q^2 = .753$		$q = .867$	Exceptions: 32/313	
$\frac{q}{1+q}$	Observed 37 71		$\left(\frac{q}{1+q}\right)^2$	Observed 1	23
	Expected 58 50			Expected 5	19
	$\chi^2 = 16.4$ n = 1			χ^2 not reliable	
	P < .01				

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On the other hand, if *dht* is assumed to be a dominant gene, then the frequency of the recessive, non-*dht* type is 1.0-.247, or .753, and $q = .867$. The cross of non-*dht* $\times$ non-*dht* would in this case be expected to yield only non-*dht* offspring. Actually, there were 10 per cent of exceptions among 313 children of non-*dht* parents, a frequency too high to explain on the basis of the calculated incomplete penetrance of distal hyperextensibility of the thumbs. The results of the mating *dht* $\times$ non-*dht* also do not fit the expectancy. For the remaining type of mating, the data are too few to permit a reliable statistical test. Clearly, the hypothesis that *dht* is a dominant gene is ruled out anyway.

In Table 3 an additional test, also based on the *Hardy-Weinberg* principle, is applied. This test, described by *Dahlberg* and *Hultkrantz* [1927] and independently developed and used by *Allan* [1928, 1933] in his studies of migraine and diabetes, in certain respects has advantages over the test developed by *Snyder*. Like the latter, it requires data over two generations, but it does not require complete sibships, the index cases and their parents being sufficient. It is, of course, essential that the index cases be obtained by a true random sampling of the population. This is most easily achieved by including only one index case per family. Only if the sample is so large that sibs might be expected to occur within it on a random basis would it be permissible to include sibs, and the same might be said for other degrees of close genetic relationship. The method consists simply in comparing the *ratio* between the index cases that come, respectively, from the three types of mating: affected by affected; affected by unaffected;

Table 3. Test for Recessive Inheritance by Proportion of Total Possessors of Trait Coming from Crosses of Different Types.

	Frequency of mating	Frequency of total recessives	
non-dht (Aa) × non-dht (Aa) . .	4p ² q ²	p ² q ²	
non-dht (Aa) × dht	2(2pq ³)	2pq ³	
dht × dht	q ⁴	q ⁴	
p ² q ² :2pq ³ :q ⁴ = p ² :2pq:q ² = .254+.499+.247			
	Expected	Exp. Corr. Observed	
non-dht × non-dht	23.9	25.6	26
non-dht × dht	46.9	46.8	53
dht × dht	23.2	21.6	15
	91		
χ ² = 3.87 n = 2			
P = .20—.10			

and unaffected by unaffected. It has often been shown that the frequency of those matings which can produce recessives is $4p^2q^2:4pq^3:q^4$ (e.g., see *Boyd* [1950], pp. 435–438; *Li* [1948], pp. 17–21; *Stern* [1949], p. 167). The first type of mating produces only one-fourth offspring of the recessive type, on the average; the second, one-half; the third, all. Hence the ratio of recessives from the three classes of matings is $p^2q^2:2pq^3:q^4$, obviously reducible to $p^2:2pq:q^2$. In other words, the proportions of recessives coming from the three types of matings in any panmictic population at equilibrium are exactly the same as the ratio in the population between the three classes, dominant homozygotes, heterozygotes, and recessive homozygotes for the character studied.

In the present case, the ratio $p^2:2pq:q^2$ has already been determined. In a total of 91 index cases forming a random sample and for whom the type of parental mating was ascertained, the expected fit to the ratio was observed ($\chi^2 = 3.87$; d.f. = 2; $P = .20 - .10$). This fit is not quite as good as that obtained in the first test, however, the deviation from expectation in the single class produced from the *dht* $\times$ *dht* mating being rather large ($\chi^2 = 2.9$). This is probably attributable to two factors: first, chance, because the expected size of this class is small enough to be affected considerably by sampling error; and second, the fact that this particular class will be specially affected by the incomplete penetrance of the trait. For any given *dht* person derived from the mating of two genotypically *dht* parents, there is a probability of $2 \times 0.035 = 0.07$, or 7 per cent, that one or the other parent will fail to manifest the trait on either hand. A correction of 1.62 should therefore be deducted from the expected size of the *dht* $\times$ *dht* class, and be added to the non-*dht* $\times$ *dht* class. A similar correction (amounting to 3.5 per cent) could then be applied to decrease the size of the non-*dht* $\times$ *dht* class and correspondingly increase the size of the non-*dht* $\times$ non-*dht* class. These corrections are small, but the net effect is to increase the expected size of the non-*dht* $\times$ non-*dht* class, to reduce that of the *dht* $\times$ *dht* class, and to leave the class non-*dht* $\times$ *dht* virtually unaffected. χ^2 is thus reduced to 2.84 with 2 degrees of freedom, and P is .30 to .20. The deviations are therefore attributable to chance.

The pedigrees of families with the trait clearly eliminate any possibility that it is sex-linked. On the other hand, *Li* (in press) and *Steinberg* [1952] have shown respectively that neither the *Snyder* formulae nor the $p^2:2pq:q^2$ relationship can distinguish between a

monofactorial recessive type of inheritance and a double recessive condition. Other more complicated hypotheses, such as complementary dominant genes or duplicate recessive genes, perhaps might also fit the population formulae, if the frequencies of the alleles at the interacting loci were just right. However, a simple multifactorial inheritance, dependent upon two loci without dominance and acting cumulatively (as often postulated for human skin color in crosses of caucasoid by negroid) can be ruled out. As Rife [1951] has shown, in such a situation the frequencies of the genotypes are as follows:

Genotype	Grade	Proportion
<i>AABB</i>	4	p^2u^2
<i>AaBB, AABb</i>	3	$2pu(qu+pv)$
<i>AAbb, AaBb, aaBB</i>	2	$p^2v^2+4pquv+q^2u^2$
<i>Aabb, aaBb</i>	1	$2qv(pv+qu)$
<i>aabb</i>	0	q^2v^2

where p and q are the frequencies of A and a respectively; and u and v are the frequencies of B and b . It follows that the ratio

$$\frac{[4]}{[0]} \text{ is the square of } \frac{[3]}{[1]}$$

This test was applied to our sample of 122 male persons, using equal ranges for the grades 0 to 4. It proved quite impossible to break the distribution into grades so that the tested relationship would fit. Instead, the ratio of the extremes was twice as great, for both right and left hands, as the ratio of classes 3 and 1, squared. This is what would be expected if the total distribution was actually bimodal but with overlapping major and minor distributions.

After this study had been completed, the trait was selected, among others, for use in comparing the gene frequencies in a religious isolate in southern Pennsylvania with those characterizing the general white population (see Glass, Sacks, Jahn, and Hess [1952]). In this small, long-isolated and highly inbred community, the frequency of distally hyperextensible thumbs is significantly lower than 24.7 per cent. However, one rather large family was found in this isolate in which 10 out of 29 members belonging to three generations have distally hyperextensible thumbs. Both parents in generation P_1 of the family are dht, and of their seven children, all were likewise dht (Fig. 5). The inheritance of the trait in this family, being clearly that of a typical recessive, confirms the conclusion previously derived regarding the mode of its inheritance.

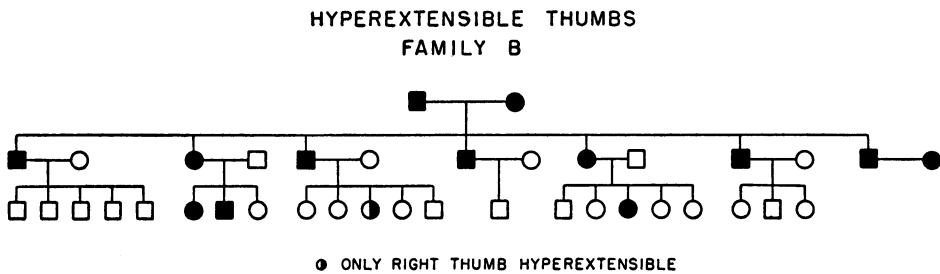


Fig. 5

Discussion.

Boyd [1950], in his excellent discussion of the need in physical anthropology for the use of genetic traits and the application of gene frequency analysis to the racial differentiation of the human species, has stated the criteria of a useful anthropological character as follows: It must be inherited according to a known mechanism, preferably simple. It must be accurately, and preferably easily, classifiable. It should be relatively stable and non-adaptive. It must be relatively common, not rare. It must vary in frequency in different populations. If it is morphological rather than physiological, and if it is a feature of parts that endure after the death of the individual, i.e., if it is skeletal, so much the better. *Boyd* comes to the conclusion that scarcely any human traits satisfy these criteria to a useable extent, except for the blood group systems, taste reaction to phenylthiocarbamide, the secretor gene which determines the presence or absence of blood group substances in the saliva and body fluids, and mid-digital hair. A few more traits might be added (see *Spuhler* [1950]), but the total of satisfactory ones is indeed few. One may agree emphatically with the conclusion that there is a very great need for the genetic analysis of more characters that meet these criteria and are suitable for the analysis of the genetic differences of human populations, not only for purposes of static description of present differences, but also to elucidate the dynamics of population change in genetic terms.

Hyperextensibility of the thumbs may now be added to the small group of suitable characters for this type of study, since it clearly satisfies all, or virtually all, of the criteria which have been set forth.

It has been shown that hyperextensibility of the thumbs at the distal joint is most probably inherited as a simple Mendelian recessive character, that if so, the dominant and recessive alleles are practic-

ally equal in frequency in the white population studied, and that the frequency of the trait varies in different ethnic groups. In spite of the slightly incomplete penetrance of the recessive trait, these properties would seem to rank it as having outstanding usefulness for anthropological studies. In the accuracy with which a population may be typed for the trait it approaches the accuracy of determination of the blood types, and the ease of typing far exceeds blood group determinations and can scarcely be surpassed by any trait whatsoever. The high frequency of the trait in the two populations studied is sufficient indication that it is not subject to adverse selection, nor does it seem likely that it is subject to positive selection either. It is a fortunate triviality, a boon of Nature to the genetical anthropologist. The senior author is mildly pleased that he has thus at length realized his childhood's desire to find some kind of usefulness for his exceptional thumbs.

Summary.

Hyperextensibility of the thumb at the distal joint (dht) can be classified almost as accurately by simple external measurement as from roentgenograms. In two different samples of the U.S. white population the frequency of dht was found to be 24.7 per cent. 40 per cent of affected persons (classified as having thumbs extensible to 50° or more) have only one thumb hyperextensible. From the coincidence the penetrance may be calculated as 96.5 per cent. The trait is not noticeably affected by age or sex. It is significantly higher in North American negroes (35.6 %) than in the white population.

Inheritance of the trait appears to be that of a simple Mendelian recessive, in spite of the fact that the frequency of distribution for the observed dht angles is of the continuous type. This conclusion is supported (a) by 11 pedigrees in which dht × dht matings yielded 30 dht children and only one exception, which would be expected on the basis of the calculated penetrance; (b) by a good fit to the expected ratios of affected siblings in dht × non-dht and non-dht × non-dht matings; (c) by a good fit to the expected distribution among types of matings of dht persons in a random sample of the population; and (d) by exclusion of dominant inheritance and of simple multifactorial inheritance. Inheritance through complementary or duplicate recessive genes is not excluded.

Assuming a simple recessive mode of inheritance, the frequencies of the alleles in the U.S. white population are estimated to be: *DHT*, .504; *dht*, .496. The value of a common, readily classified trait such as this in studies of human population, genetics and physical anthropology is pointed out.

Résumé.

L'hyperextensibilité de la dernière articulation du pouce (dht, c'est-à-dire « distal hyperextensibility of the thumb ») peut être déterminée à peu près avec la même exactitude en prenant des mesures sur des personnes qu'en les prenant sur des radiographies. Pour deux échantillons différents, pris dans la population blanche de l'Amérique du Nord, la fréquence de dht était de 24,7 %. 40 % des personnes atteintes (ayant des pouces extensibles jusqu'à un angle de 50° ou plus) n'avaient

que l'un des pouces hyperextensible. En se basant sur la fréquence observée on peut calculer que le taux de pénétrance est de 96,5 %. Le caractère n'est pas sensiblement influencé par l'âge ou le sexe. La fréquence plus élevée de ce caractère dans la population noire de l'Amérique du Nord (35,6 %) que dans la population blanche est significative du point de vue statistique.

Le mode de transmission du caractère semble être celui d'un caractère récessif mendélien simple, bien que la distribution de fréquence des angles de dht soit d'un type continu. Cette conclusion se base *a)* sur 11 arbres généalogiques dans lesquels des unions dht $\times$ dht donnaient 30 enfants atteints de dht et seulement une exception, comme on pouvait s'y attendre par suite de la pénétrance calculée; *b)* sur une concordance satisfaisante avec les taux prévus des descendants atteints dans des unions entre des personnes dht-non dht, et entre des personnes non dht-non dht; *c)* sur une concordance satisfaisante avec la distribution prévue d'enfants atteints provenant d'unions, prises au hasard dans la population, entre des personnes dont l'une est atteinte de dht; et *d)* sur l'exclusion de transmission dominante et de transmission plurifactorielle simple. L'hérédité par « complementary or duplicate recessive genes » ne peut pas être exclue.

Si l'on admet que le mode de transmission est simple récessif, la fréquence des allèles dans la population blanche de l'Amérique du Nord peut être évaluée à: *DHT*, .504; *dht*, .496. Du point de vue génétique et anthropologique on souligne la valeur d'un tel caractère, à la fois fréquent et facile à classer, pour des études de populations humaines.

Zusammenfassung.

Distale Hyperextensibilität des Daumens (dht = distal hyperextensibility of the thumb) kann nahezu gleich exakt durch einfache externe Messung wie auch durch Röntgenogramm klassifiziert werden. In zwei verschiedenen Auswahlen unter der weißen U.S.-Bevölkerung wurde für die dht Frequenz der Wert von 24,7 % gefunden. 40 % der behafteten Personen (mit einer Daumenextensibilität von 50° und mehr) haben dht nur bei einem Daumen. Aus der Koinzidenz kann man eine Penetranz mit 96,5 % annehmen. Das Merkmal ist nicht wesentlich durch Alter und Geschlecht beeinflusst. Es kommt sichtlich häufiger bei nord-amerikanischen Negern (35,6 %) als bei der weißen Bevölkerung vor.

Die Vererbbarkeit des Merkmals scheint einfach rezessiv zu sein, trotz der Tatsache, daß die Verteilungskurve der beobachteten dht Winkel kontinuierlich ist. Diese Feststellung wird unterstrichen durch: *a)* 11 Vererbungstafeln, in den dht $\times$ dht Kreuzungen 30 dht Kinder ergaben mit nur einer Ausnahme, die man auf Grund der einberechneten Penetranz erwarten durfte; *b)* eine gute Übereinstimmung mit der erwarteten Proportion behafteter Geschwister in dht $\times$ nicht-dht und nicht-dht $\times$ nicht-dht Kreuzungen; *c)* eine gute Übereinstimmung mit der erwarteten Verteilung unter den Typen bei Kreuzungen von dht-Personen in einer zufälligen Auswahl unter der Bevölkerung und *d)* durch den Ausschluß dominanter Vererbung und einfacher Polymerie. Vererbung durch « complementary or duplicate recessive genes » ist nicht ausgeschlossen.

In der Annahme eines einfachen rezessiven Erbganges schätzt man die Allelfrequenzen in der weißen U.S.-Bevölkerung auf *DHT* 0.504; *dht* 0.496. Der Wert eines gewöhnlichen, bereits klassifizierten Erbmerkmals wie dieses in Studien menschlicher Populationen aus genetischen und physisch-anthropologischen Gesichtspunkten wird herausgestellt.

REFERENCES

- Allan, Wm.*: Arch. intern. Med. 42, 590-599, 1928; 6, 1272-1274, 1932. – *Boyd, W. C.*: Genetics and the Races of Man. Little, Brown and Co., Boston 1950. – *Dahlberg, G. and J. V. Hultkrantz*: Arch. Rass. Ges. Biol. 19, 129-165, 1927. – *Glass, B., M. S. Sacks, E. F. Jahn and C. Hess*: Amer. Nat. 86, 145-159, 1952. – *Harris, H. and J. Joseph*: J. Bone Jt. Surg. 31B, 547-559, 1949. – *Li, C. C.*: An Introduction to Population Genetics, National Peking University Press, Peiping 1948; Amer. J. hum. Genet. (in press). – *Rife, D. C.*: J. Hered. 42, 162, 1951. – *Snyder, L. H.*: Genetics 19, 1-17, 1934. – *Spuhler, J. N.*: Cold Spr. Harb. Symp. quant. Biol. 15, 175-189, 1950. – *Steinberg, A. G.*: Amer. J. hum. Genet. 4, 373-375, 1952. – *Stern, C.*: Principles of Human Genetics, W. H. Freeman, San Francisco 1949. – *Whitney, D. D.*: Family Treasures, J. Cattell Press, Lancaster 1942. – *Whitney, L. F.*: J. Hered. 23, 425-426, 1932.

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ON THE THEORETICAL EFFECT OF MUTATION

By NILS von HOFSTEN

The effect of mutation proceeding at a steady rate is easily calculated, but an analysis of the process is perhaps not superfluous; it gives results which, though rather self-evident, merit attention or, at any rate, have a theoretical interest.

If a character is due to a single gene, the increase of the character-carriers through mutation is, in cases of typical dominance-recessivity, a simple function of that of the gene. What I intend to discuss especially is the effect of mutation affecting two different genes, above all in cases of dimerism where a character is due to two genes.

It is presumed in all following calculations and deductions that mutation is proceeding undisturbed by selection, back-mutation and mutation of the mutated genes. When a large number of generations is considered, this presumption is very unnatural. This does not affect the principles which are of importance for the present purpose, and the comparison between different cases is facilitated if such disturbing factors are left out of account. In science it is often essential to select one single factor from several involved in a process. A purely theoretical investigation has the advantage that this is always possible. Even when applied to the point of absurdity, this method may give valuable insight into a process.