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Two-Gene Hypothesis for Hairy Pinnae of the Ear

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SUMMARY

A new hypothesis involving two genes has been proposed to explain the nature of hairy pinnae of the ear. The insignificant values of the goodness of fit χ^2 seem to confirm the proposed hypothesis.

Introduction

A little more than six decades have passed since the discovery of the trait, hairy pinnae of the ear, as a genetic character in human populations. In spite of numerous attempts, the true nature of the underlying hypothesis does not seem to have been fully explored. However, a great contribution in this direction has already been made in that it is a Y-linked character. The literature available on this subject is, undoubtedly, very rich.

The object of this additional load to the already heavy literature is to suggest a concrete hypothesis involving two nonallelic genes, one of which is situated on the nonhomologous part and the other on the homologous part of the Y-chromosome (the homologous part of the X-chromosome also shares a corresponding allele).

Fig. 1 depicts the location of the two loci in sex chromosomes of a male, the loci being named after the segment of the chromosome (NH for nonhomologous and H for homologous).

Two-Gene Hypothesis

The alleles at the NH-locus are B and b, whereas they are A and a at the H-locus. This leads to two genotypes for the NH-locus and four genotypes for the H-locus. The following are the different types of X and Y chromosomes:

Y-chromosome: AB, aB, Ab, ab;
X-chromosome: A, a.

The genotypes of males and females are given by:

Male : A (AB), a (AB), a (aB), A (aB), A (Ab), a (Ab), A (ab), a (ab);
Female: AA, Aa, aa.

Rec. 18. VIII. 1969

Acta Genet. Med. Gemellol. (1970), 19: 448-453

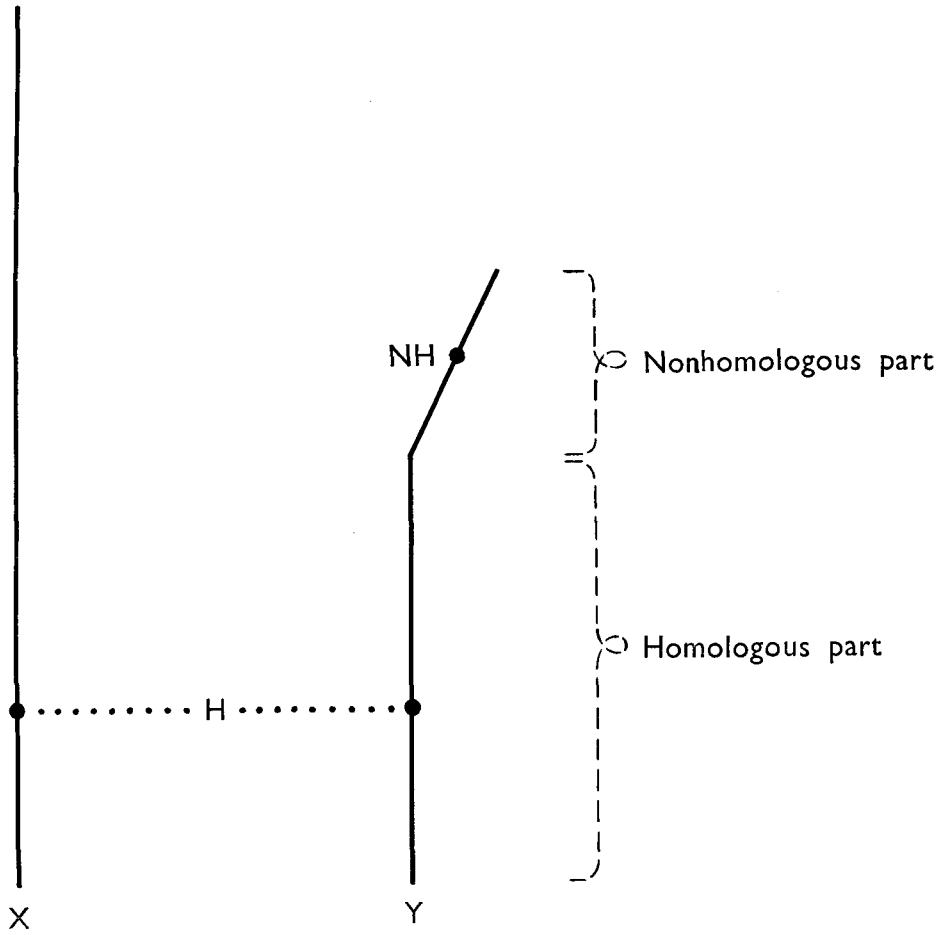

Fig. 1. The two sex chromosomes in man

Observe that, in case of males, the types of Y-chromosomes are written within parentheses. We propose the following:

1) The presence of B gene at the NH-locus determines the trait;

2) Given B gene at the NH-locus, the genotype for the H-locus (AA, Aa, aA, or aa) determines the intensity of the trait (this, beyond doubt and in accordance with the observations, affords to say that no female can either exhibit or transmit the trait to her offspring, and for this very reason we shall no more talk about females in this study);

3) All males can be classified into five phenotypes (to be identified by the intensity of the trait).

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For males under random mating, the genotypes, phenotypes (along with grades used for the intensity of the trait) and the phenotypic frequencies are shown in Tab. I; where p, r, q and s are the frequencies of the Y-chromosomes AB, aB, Ab and ab, while t and u stand for the X-chromosome frequencies of types A and a respectively ($p + r + q + s = 1 = t + u$).

Statistical Analysis

The phenotypes, expected (phenotypic) frequencies under random mating and observed frequencies in a sample of n unrelated males are shown in Tab. II.

The maximum likelihood estimates of p, r and t are given by:

$$\hat{r} = \frac{\sqrt{(n_0 + n_1 + n_2)^2 + 4\,(n_1 + n_2)^2} - (n_0 + n_1 + n_2)}{2\,(n_1 + n_2)};$$

$$\hat{p} = \frac{(n_3 + n_4)\,(1 - \hat{r})}{(n_0 + n_3 + n_4)};$$

$$\hat{t} = \frac{n_2 + n_4}{n - n_0}.$$

The proportion of individuals showing the trait (of non-zero grade) is simply the B-gene frequency, which is given by $P(B) = p + r$.

As a simple test of the proposed hypothesis, let us consider a few samples of unrelated males from Indian populations, which are abstracted from earlier publications.

Of the three samples considered below, the first one was published by Sarkar et al (1961), the second and third ones by Stern et al (1964). It is to be noted that the ear-

Tab. I. Genotypes, phenotypes, phenotypic frequencies and the grades for males under random mating

Genotype	Phenotype	Phenotypic frequency	Grade
A (AB)	A (AB)	pt	4 (very marked)
a (AB)	a (AB)	pu	3 (marked)
A (aB)	A (aB)	rt	2 (scanty)
a (aB)	a (aB)	ru	1 (very scanty)
A (Ab) a (Ab) A (ab) a (ab)	Āb	q+s	0 (nothing)

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Tab. II. Phenotypes, expected and observed frequencies of males under random mating

Phenotype	Grade	Expected frequency	Observed frequency
A (AB)	4	n p t	n_4
a (AB)	3	n p u	n_3
A (aB)	2	n r t	n_2
a (aB)	1	n r u	n_1
Ā b	0	n (q+s)	n_0
Totals		n	n

lier authors have used altogether six grades (0 to 5), whereas the present author recommends only five (0 to 4). In the present study, grades 4 and 5 of earlier authors have been merged into grade 4.

Sample 1 (from West Bengal, India)

A sample of 226 males is incorporated in Tab. III, in accordance with Tab. II, showing the detailed computations.

The estimates of p, r and t are:

$$\hat{p} = 0.061768;\ \hat{r} = 0.047000;\ \hat{t} = 0.291667.$$

Sample 2 (from Vellore, A.P., India)

The results are shown in Tab. IV.

For this sample, the estimates are:

$$\hat{p} = 0.031098;\ \hat{r} = 0.293957;\ \hat{t} = 0.190164.$$

Sample 3 (from West Bengal, India)

The results are shown in Tab. V.

Tab. III. West Bengal

Phenotype	Grade	Observed frequency (O)	Expected frequency (E)	$\frac{(O-E)^2}{E}$
A (AB)	4	2	4.0716	1.0541
a (AB)	3	12	9.8900	0.4502
A (aB)	2	5	3.0980	1.1677
a (aB)	1	5	7.5240	0.8467
Ā b	0	202	201.4164	0.0017
Totals		226	226.0000	3.5204

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Tab. IV. Vellore

Phenotype	Grade	Observed frequency (O)	Expected frequency (E)	$\frac{(O-E)^2}{E}$
A (AB)	4	4	5.2694	0.3058
a (AB)	3	23	22.4389	0.0140
A (aB)	2	54	49.8069	0.3530
a (aB)	1	224	212.1088	0.6666
$\bar{A}$ b	0	586	601.3760	0.3931
Totals		891	891.0000	1.7325

Tab. V. West Bengal

Phenotype	Grade	Observed frequency (O)	Expected frequency (E)	$\frac{(O-E)^2}{E}$
A (AB)	4	4	6.6193	1.0365
a (AB)	3	15	12.2931	0.5961
A (aB)	2	10	7.8010	0.6199
a (aB)	1	11	14.4874	0.8395
$\bar{A}$ b	0	261	259.7992	0.0056
Totals		301	301.0000	3.0976

Tab. VI. Summary of χ^2 values

Sample number	1	2	3
χ^2 value	3.52	1.73	3.10

For this sample, the estimates are:

$$\hat{p} = 0.062832;\ \hat{r} = 0.074048;\ \hat{t} = 0.350000.$$

The values of the goodness of fit χ^2 (each with 1 df) are summarized in Tab. VI.

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RIASSUNTO

Viene proposta un'ipotesi digenica per il carattere delle orecchie pelose. L'ipotesi è confermata dall'analisi del χ^2.

RÉSUMÉ

Une hypothèse digénique est proposée pour le caractère du poil aux oreilles. L'hypothèse est confirmée par l'analyse du χ^2.

ZUSAMMENFASSUNG

Eine Analyse des χ^2 bestätigt die Hypothese, dass das Merkmal der behaarten Ohre auf einer Dysgenie beruhe.

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