Proceedings of the Iowa Academy of Science

Volume 44 | Annual Issue Article 42

1937

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Recommended Citation
Norton, H. W. III (1937) "General Formulae for Homozygosis," *Proceedings of the Iowa Academy of Science, 44(1),* 139-143.
Available at: https://scholarworks.uni.edu/pias/vol44/iss1/42

GENERAL FORMULAE FOR HOMOZYGOSIS

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To find f(x) from the linear fractional relation

$$f(x+1) = \frac{a + bf(x)}{c + df(x)} \qquad (1)$$

let

$$f(x) = \frac{g}{\phi(x)} + e \qquad (2)$$

and by (1),

$$\phi(x+1) = \frac{dg + (c+de)\phi(x)}{b - de + \dfrac{a + (b-c)e - de^2}{g}\phi(x)} \qquad (3)$$

Choosing

$$e = \frac{b - c \pm \sqrt{(b-c)^2 + 4ad}}{2d} \qquad (4)$$

$$g = \frac{b}{d} - e \qquad (5)$$

and letting

$$h = \frac{c + de}{b - de} \qquad (6)$$

(3) becomes

$$\phi(x+1) = 1 + h\phi(x) \qquad (7)$$

By mathematical induction it may be shown that

$$\phi(x+n) = \sum_{i=0}^{n-1} h^i + h^n\phi(x) \qquad (8)$$

Letting x = 0, substituting by means of (2), and solving for f(n),

$$f(n) = \frac{g + e\sum_{i=0}^{n-1} h^i + \dfrac{eg}{f(0) - e}h^n}{\sum_{i=0}^{n-1} h^i + \dfrac{g}{f(0) - e}h^n} \qquad (9)$$

A more general solution includes in the right member of (9) any periodic function of period unity, but in this paper only integral values of n are of interest. In general h ± 1, and (9) may be written

$$f(n) = \frac{A + Bh^n}{C + Dh^n} \qquad (10)$$

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where
$$A = g + \frac{e}{1-h} \tag{10a}$$
$$B = e\left[\frac{g}{f(0)-e} - \frac{1}{1-h}\right] \tag{10b}$$
$$C = \frac{1}{1-h} \tag{10c}$$
$$D = \frac{g}{f(0)-e} - \frac{1}{1-h} \tag{10d}$$

The special case $h = 1$ will arise if
$$(b-c)^2 + 4ad = 0 \tag{11}$$
for if $h = 1$, by (4) and (6) respectively,
$$2de = b - c \pm \sqrt{(b-c)^2 + 4ad}$$
$$2de = b - c$$
so that
$$e = \frac{b-c}{2d}$$
$$g = \frac{b+c}{2d}$$
Substituting in (9), simplification yields
$$f(n) = \frac{\dfrac{(b+c)f(0)}{2df(0)-b+c} + \dfrac{b-c}{2d}n}{\dfrac{b+c}{2df(0)-b+c} + n} \tag{12}$$

Inspection of (1) reveals that $f(x)$ has asymptotes which may be found by setting $f(x+1)$ equal to $f(x)$. Solving for $f(x)$, these limiting values are
$$f(x) = \frac{b - c \pm \sqrt{(b-c)^2 + 4ad}}{2d}$$
This is e of (4), the suitable additive constant of the linear fractional transformation (2).

An application of this solution occurs in genetics. It is well established that genes, the hereditary units, occur in pairs. An individual is homozygous in a locus if the two genes in that locus are alike, heterozygous if the genes are unlike. If a gene and its allel be represented by A and a respectively, the three possible pairs are AA, Aa and aa. In the formation of gametes, all gene-pairs are divided and two gametes are formed, each containing one member of each of the parental gene-pairs. Union of two gametes produces a new individual having the same number of gene-pairs as each of the parents. It may happen that the effect of A completely masks the effect of a so that AA and Aa types are indistinguishable. In such a case, the probability of homozygosis of an A- individual is of interest.

If two A- individuals have probabilities p_s and p_d respectively of being homozygous, that is, AA, the probability of homozygosis of an A- individual among their progeny may be calculated. Using $q = 1 - p$, among the sire's gametes there will be $2p_s + q_s$ of A to q_s of a, and among the dam's gametes $2p_d + q_d$ of A to q_d of a. Among the progeny, the frequencies of the three possible types will then be

AA	$(2p_s + q_s)(2p_d + q_d)$	or	$(1 + p_s)(1 + p_d)$
Aa	$(2p_s + q_s)q_d + (2p_d + q_d)q_s$	or	$2(1 - p_sp_d)$
aa	q_sq_d	or	$(1 - p_s)(1 - p_d)$

The proportion, p_x, of AA individuals among those of the progeny that are A- is then

$$P_x = \frac{(1 + p_s)(1 + p_d)}{(1 + p_s)(1 + p_d) + 2(1 - p_sp_d)}$$

$$P_x = \frac{1 + p_s + p_d + p_sp_d}{3 + p_s + p_d - p_sp_d} \tag{13}$$

Two special cases are of interest. If one of the parental probabilities is a constant k, and if the other be represented by p, (13) becomes

$$P_x = \frac{(1 + k) + (1 + k)p}{(3 + k) + (1 - k)p} \tag{14}$$

In the other special case $p_s = p_d = p$ so that

$$P_x = \frac{1 + p}{3 - p} \tag{15}$$

These formulae are useful for passing from one generation to the next, but if a system of mating consisting of one of these special cases has been followed for many generations, repeated application of the appropriate formula becomes tedious. Since the probabilities for intervening individuals may be of no interest, (14) and (15) will be generalized by the method previously developed. Formula (14) may be written

$$f(n + 1) = \frac{(1 + k) + (1 + k)f(n)}{(3 + k) + (1 - k)f(n)}$$

where n is the generation number and f(n) is the probability that an nth generation A- individual is homozygous. Obviously, a =

$b = 1 + k$, $c = 3 + k$ and $d = 1 - k$. Substitution in (11) shows that the special case $h = 1$ arises if $k = \pm\sqrt{2}$ which cannot occur because k is a probability, demanding that $0 \leq k \leq 1$. Substituting in the appropriate formulae, the generalization of (14) is

$$f(n) = \frac{\frac{(1+k)(\sqrt{2-k^2} \pm k)}{2(1-k)\sqrt{2-k^2}} + \left[\frac{-1 \pm \sqrt{2-k^2}}{1-k}\right] \cdot \left[\frac{2+k \mp \sqrt{2-k^2}}{(1-k)f(0) + 1 \mp \sqrt{2-k^2}} - \frac{\sqrt{2-k^2} \mp (2+k)}{2\sqrt{2-k^2}}\right] \left[\frac{2+k \pm \sqrt{2-k^2}}{2+k \mp \sqrt{2-k^2}}\right]^n}{\frac{\sqrt{2-k^2} \mp (2+k)}{2\sqrt{2-k^2}} + \left[\frac{2+k \mp \sqrt{2-k^2}}{(1-k)f(0) + 1 \mp \sqrt{2-k^2}} - \frac{\sqrt{2-k^2} \mp (2+k)}{2\sqrt{2-k^2}}\right]\left[\frac{2+k \pm \sqrt{2-k^2}}{2+k \mp \sqrt{2-k^2}}\right]^n} \quad (16)$$

If $k = 0$, (16) simplifies to

$$f(n) = \frac{1 + \left[3 \mp 2\sqrt{2} - \frac{8 \mp 6\sqrt{2}}{f(0) + 1 \mp \sqrt{2}}\right]\left[3 \pm 2\sqrt{2}\right]^n}{1 \mp \sqrt{2} - \left[1 \mp \sqrt{2} - \frac{4 \mp 2\sqrt{2}}{f(0) + 1 \mp \sqrt{2}}\right]\left[3 \pm 2\sqrt{2}\right]^n} \quad (17)$$

and if $f(0) = 0$,

$$f(n) = \frac{1 - (3 \pm 2\sqrt{2})^n}{1 \mp \sqrt{2} - (1 \pm \sqrt{2})(3 \pm 2\sqrt{2})^n} \quad (18)$$

Formula (18) gives the probability, f(n), that an A- individual, resulting from n generations of a system in which one parent and both original parents were heterozygous, is homozygous. In formula (16) a special case arises if $k = 1$, in which case (14) may be written

$$f(n+1) = \tfrac{1}{2} + \tfrac{1}{2} f(n)$$

and by analogy with (7) et sqq.,

$$f(n) = 1 - \frac{1 - f(0)}{2^n} \quad (19)$$

If the parents are equally likely to be homozygous,

$$f(n+1) = \frac{1 + f(n)}{3 - f(n)}$$

may be written for (15) so that $a = b = -d = 1$ and $c = 3$. Substitution in (11) shows that this is the special case $h = 1$. Substituting in (12) therefore, and simplifying,

$$f(n) = \frac{\dfrac{2f(0)}{1 - f(0)} + n}{\dfrac{2}{1 - f(0)} + n} \qquad (20)$$

If $f(0) = 0$, this is simply

$$f(n) = \frac{n}{n + 2} \qquad (21)$$

These formulae are adapted only to the study of pedigrees. They are not prognostic because they ignore aa individuals which may result from Aa $\times$ Aa matings.

As an example of the use of these formulae, consider a system in which one parent and both original parents were heterozygous, and let the probability of homozygosis of an A- second generation individual be desired. In this system $k = 0$ and $f(0) = 0$ so that (18) is applicable. Putting $n = 2$, (18) gives

$$f(2) = \frac{1 - (3 \pm 2\sqrt{2})^2}{1 \mp \sqrt{2} - (1 \pm \sqrt{2})(3 \pm 2\sqrt{2})^2}$$

which simplifies to

$$f(2) = 2/5$$

This may be checked by substitution in (13), using $p_s = p_d = 0$, giving

$$p_1 = 1/3$$

and in turn

$$p_2 = \frac{1 + 1/3}{3 + 1/3}$$

or

$$p_2 = 2/5$$

checking the result obtained by use of (18).

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