

Anthropology 215

■ POPULATION GENETICS



"I know you miss the Waterweights, Bobby, but they were weak and stupid people — and that's why we have wolves and other large predators."



"Natural selection at work."

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Darwin

- characteristics that provide the best adaptation to environment have better chance of being represented in the following generation

Some Early Skeptics

- If a fitter variant arises, will it not, out of necessity, have to mate with a less fit individual, which eventually would then lead to a dilution of the new characteristic and hence a dilution of the beneficial characters?

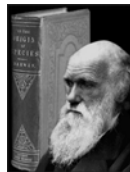
Mendel

- characteristics passed on as discrete units (genes) which do not lose their integrity Hardy-Weinberg law
- A blending process does not occur

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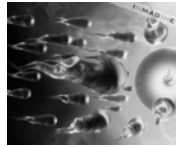
Fleming Jenkins

- shouldn't dominant phenotype swamp the recessive one?



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e.g., 2 alleles: A, a



paternal/maternal		
sperm\ova	A	a
A	AA	Aa
a	Aa	aa

p = frequency of **A** (dominant) allele
q = frequency of **a** (recessive) allele
p + q = 1.0 or 100%

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Freq. of alleles	p_A	q_a
p_A	p^2_{AA}	pq_{Aa}
q_a	pq_{Aa}	q^2_{aa}

p^2 = chance of **A** combining with **A** (**AA**)
 $2pq$ = chance of **A** combining with **a** (**Aa**)
 q^2 = chance of **a** combining with **a** (**aa**)



AA : **Aa** : **aa**
 p^2 + $2pq$ + q^2
 1 : 2 : 1

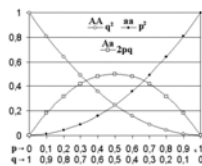
$p^2 + 2pq + q^2$

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e.g., $p = 0.9$ and $q = 0.1$

	$p_A = .9$	$q_a = .1$
$p_A = .9$	AA = .81	Aa = .09
$q_a = .1$	Aa = .09	aa = .01

81% AA **18% Aa** **1% aa**
 p^2 $2pq$ q^2
.81 **.18** **.01**



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Mating Combinations

AA x AA
AA x Aa
AA x aa
Aa x Aa
Aa x aa
aa x aa

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Frequency of the 9 mating types:

$p = AA$ $q = Aa$ $r = aa$

	$p_{AA}=.81$	$q_{Aa}=.18$	$r_{aa}=.01$
$p_{AA}=.81$.6561	.1458	.0081
$q_{Aa}=.18$.1458	.0324	.0018
$r_{aa}=.01$.0081	.0018	.0001

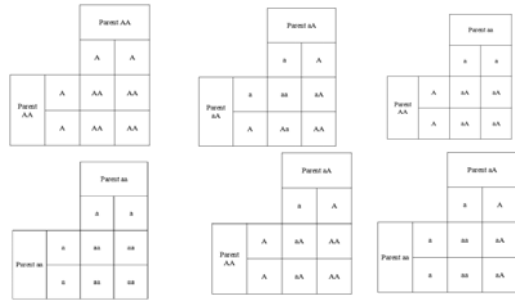
$(p+q+r)^2$ or $.0651 + 2(.1458) + 2(.0081) + .0324 + 2(.0018) + .0001$

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Mating	Frequency	AA	Aa	aa
AA x AA	.81 x .81	.6561	--	--
AA x Aa	2(.1458)	.1458	.1458	--
AA x aa	2(.0081)	--	.0162	--
Aa x Aa	.18 x .18	.0081	.0162	.0081
Aa x aa	2(.0018)	--	.0018	.0018
aa x aa	.01 x .01	--	--	.0001
		.81	.18	.01

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Mating Types & Expected Offspring



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Summary

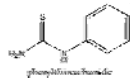
- No matter what the original values of p and q , in a randomly mating population, genotypes will remain in the ratio: $p^2 : 2pq + q^2$
- Hardy Weinberg Equilibrium
- Memorize Two Formulae:
 - $p^2 : 2pq + q^2$
 - $p + q = 1$



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Calculation of gene and genotype frequencies for autosomal dominant traits

- E.g., ability to taste PTC
- Tasters = dominant (TT or Tt)
- Non-tasters = recessive (tt)



Given that 16 % of population are non-tasters (tt) and 84% are tasters (TT, Tt):

1. What is the frequency of T and t alleles?
2. What is the frequency of TT and Tt genotypes?

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Hardy-Weinberg predicts that the 3 genotypes are in the proportion:

$$p^2 : 2pq : q^2$$

$$TT \quad Tt \quad tt$$

Solution

Can solve for q (freq. of recessive allele) since non-tasters have only 1 genotype (tt) which = q^2

$$q^2 = .16$$

$$q = \sqrt{.16}$$

$$q = .4$$

Since $p + q = 1$
 $p + .4 = 1$
 $p = 1 - .4$
 $p = .6$

Substitute values of p and q back into H-W to calculate frequencies of TT and Tt:

$$TT = p^2 = (.6 \times .6) = .36$$

or 36 %

$$Tt = 2pq = 2(.6)(.4) = .48$$

or 48%

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Calculation of gene frequencies when dominance and recessiveness is lacking (i.e. co-dominant traits)

Genotype	Alleles			Total
	n	M	N	
MM	197	394	-	394
MN	131	131	131	262
NN	22	0	44	44
	350	525	175	700

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Gene frequencies:

- frequency of M = $525/700 = .75$
- frequency of N = $175/700 = .25$

Genotype frequencies:

- frequency of MM = $197/350 = 0.563$
- MN = $131/350 = 0.374$
- NN = $22/350 = 0.063$

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Using the formula, $p^2 + 2pq + q^2$, fill in the blanks

- p = frequency of the _____ allele in the population
- q = frequency of the _____ allele in the population
- p^2 = percentage of _____ individuals
- q^2 = percentage of _____ individuals
- $2pq$ = percentage of _____ individuals

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Question 1

In a population with two alleles for a particular locus, B and b , the allele frequency of B is 0.7. What would be the frequency of heterozygotes if the population is in Hardy-Weinberg equilibrium?

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Answer

- Given that $B = .7 = p$
- Because $p + q = 1$
- $.7 + q = 1$
- $q (b) = 1 - .7$, or $.3$
- Heterozygotes = $2pq = 2 (.7) (.3) = .42$ or 42%

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Question 2

If the percentage of the homozygous recessive genotype (aa) is 36% in a population, calculate the following:

- The frequency of the "aa" genotype.
- The frequency of the "a" allele.
- The frequency of the "A" allele.
- The frequencies of the genotypes "AA" and "Aa."
- The frequencies of the two possible phenotypes if "A" is completely dominant over "a."

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Answers-1

- The frequency of the "aa" genotype. Answer: 36%, as given in the problem itself.
- The frequency of the "a" allele is = .6. How?: The frequency of aa is 36%, which means that $q^2 = 0.36$. If $q^2 = 0.36$, then $q = 0.6$ because the square root of .36 is .6.

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Answers-2

- The frequency of the "A" allele is .4 or 40%.
- Since $q = 0.6$, and $p + q = 1$, then $p = 0.4$; the frequency of A is by definition equal to p.
- The frequencies of the genotypes "AA" and "Aa." Answer: The frequency of AA is equal to p^2 , and the frequency of Aa is equal to $2pq$. So, using the information above, the frequency of AA is 16% (i.e. p^2 is $0.4 \times 0.4 = 0.16$) and Aa is 48% ($2pq = 2 \times 0.4 \times 0.6 = 0.48$).

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Answers-3

- The frequencies of the two possible phenotypes if "A" is completely dominant over "a."
- Because "A" is totally dominate over "a", the dominant phenotype will show if either the homozygous "AA" or heterozygous "Aa" genotypes occur. Therefore, the frequency of the dominant phenotype equals the sum of the frequencies of AA (p^2) and Aa ($2pq$), .64 or 64%.
- The recessive phenotype is controlled by the homozygous aa genotype so the frequency of the recessive phenotype is simply the frequency of aa, .36% which was given initially.

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Question 3

In a population that is in Hardy-Weinberg equilibrium, 16% of the individuals show the recessive trait. What is the frequency of the dominant allele in the population?

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Answer

- $q^2 = .16$
- $q = .4$
- $p + q = 1.0$
- $p = 1 - .4 = .6$

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