Human Genetic Variation:
Hardy-Weinberg Equilibrium Worksheet

Hardy-Weinberg Equilibrium with an autosomal recessive allele: ΔCCR5 and HIV resistance example

"Given the impact of this mutation on the current HIV epidemic, we would like to know the frequency of this genotype."

[pause at 12:18]

From Martinson et al[1]., the following genotypic categories were quantified:

Genotype	*Phenotype*	*Martinson et al.*
CCR5/ CCR5	Normal HIV infection susceptibility	647
CCR5/ ΔCCR5	Delay in progression to AIDS after HIV infection	134
ΔCCR5/ ΔCCR5	Partial HIV resistance	7
Total		788

1. Given these numbers, calculate the frequency of each genotype.

2. How can we also use the data in the table to calculate the frequency of each allele?

[1] Martinson JJ, Chapman NH, Rees DC, Liu YT, and Clegg JB (1997) Global distribution of the CCR5 gene 32-basepair deletion. Nat Genet. 16(1):100-3.

Students should resume the video at 16:11 if answers match the given values.

[pause at 17:50]

3. Using Hardy-Weinberg equilibrium, calculate the expected frequencies of each genotype from the allelic frequencies.

Equations: Hardy-Weinberg equilibrium

$$p+q = 1$$
$$p^2 + 2pq + q^2 = 1$$

p = frequency of allele A
q = frequency of allele a
p^2 = frequency of genotype AA
q^2 = frequency of genotype aa
2pq = frequency of genotype Aa

4. Do these frequencies match those ascertained by Martinson et al.?

[Resume play at 22:26 – pause at 23:22]

Hardy-Weinberg Equilibrium with an autosomal dominant allele:
Marfan Syndrome example

5. Marfan's syndrome is caused by an autosomal dominant mutation in the fibrillin-1 gene. Given that the incidence of Marfan's syndrome in a particular population is 1 in 100,000 individuals, and that individuals homozygous for this dominant allele are, for all intents and purposes, non-existent in the population, what is the allelic frequency of mutated fibrillin-1 in this population? Does this allele frequency predict the observed absence of individuals with the homozygous genotype in the population?

[Resume play at 25:14 – pause at 25:50]

Hardy-Weinberg Equilibrium with an X-linked recessive allele:
Red-green color blindness example

Protanopia is one type of red-green color blindness inherited in an X-linked recessive fashion. In a certain population, the prevalence of protanopic males is 1 in 100.

6. What is the frequency of protanopic females?

[Resume play 27:08]