

**Hardy-Weinberg Law**

$f(A)$  = frequency of allele A,  $f(AA)$  = frequency of genotype AA, etc.

Call  $f(A) = p$ ;  $f(a) = q$ . Then  $p + q = 1$  since every allele is either A or a. At equilibrium:

$f(AA) = p^2$ ;  $f(aa) = q^2$ , and  $f(Aa) = 2pq$ , and  $p^2 + 2pq + q^2 = 1$ , since every individual is either AA, aa, or Aa.

**Genotype of a Population.**

The genotype of a population (aka "genetic structure of population") is described if you know  $f(A)$ ,  $f(a)$  and  $f(AA)$ , etc. = allele frequencies and genotype frequencies.

**When does H-W Law Apply?**

The H-W law applies ONLY if the population is in equilibrium.

If population is NOT in equilibrium, you can still define the genotype of the population if you know  $f(A)$ ,  $f(AA)$  etc. However you can NOT calculate the proportions of genotypes from the allele frequencies (or vice versa) using the H-W law. You have to use the "seat-of-the-pants" method

**Some examples of use of H-W***1. How to use frequencies, assuming equilibrium*

Cystic Fibrosis = CF = most common genetic disease among whites. It's recessive. Person with disease is aa.

Know CF = 1/2000 live births. How many carriers (heterozygotes without symptoms) are there?

$f(a)^2 = f(aa) = 1/2000 = 5 \times 10^{-4}$ ; therefore  $f(a) = \sqrt{5 \times 10^{-4}} = \text{about } 2 \times 10^{-2}$ .

$2pq = 2 \times (1 - 2 \times 10^{-2}) \times 2 \times 10^{-2} = \text{about } 4 \times 10^{-2} = 4/100 = 1/25$ . That's a lot of people.

Over 300 million people in USA, so over  $10^7$  (over 10 million) are carriers; therefore screening is worth it.

*2. How to use the H-W law to test for equilibrium. 2 examples with MN. (M & N are surface proteins found on blood cells. M and N are coded for by two co-dominant alleles of the same gene. See problem 9-2.)*

a. The method. You don't need to check each generation separately to see if we have reached equilibrium. Only need to check proportions of genotypes and alleles of the total population and see if it all fits the H-W distribution.

b. Here are two cases:

Case 1 (real situation in US)	Case 2 (fake)
MM = 30%	MM = 25 %
MN = 50 %	MN = 60 %
NN = 20 %	NN = 15 %
$f(M) = \sqrt{.3} = .55 = p$	if plug in, $f(M) = \sqrt{.25} = .5$
$f(N) = \sqrt{.2} = .45 = q$	$f(N) = \sqrt{.15} = .4$
$p + q = 1$ , ok.	$p + q \text{ not } = 1$ , not ok.
This population is in genetic equilibrium.	This population is not in genetic equilibrium.