

## 2.2 Hardy-Weinberg equilibrium and statistical tests

**Definition of H-W equilibrium.** In a large population with random mating H-W equilibrium will occur after one generation provided that the same gene frequencies occur in both sexes. Hardy-Weinberg equilibrium implies that gene and genotype frequencies are constant from generation to generation. If disequilibrium occurs, equilibrium will be reestablished after one generation of random mating. The H-W conditions also imply that when the gene frequencies are  $p$  and  $q$ , the genotype frequencies will be respectively  $p^2$ ,  $2pq$  and  $q^2$  for the dominant, the heterozygotes and the recessive in a two allele system. This can be inferred by the arguments given for the recessive type under dominant inheritance

### Statistical test for H-W equilibrium

Exemplified by data from the albumin types in Danish German Shepherd population, shown in section 2.1. The following observed numbers of the three genotypes (obs), and the calculated expected numbers under Hardy Weinberg equilibrium (exp) with frequencies  $p(S)=0.56$  and  $q(F)=0.44$  was found in the population.

|                |        |        |        |           |
|----------------|--------|--------|--------|-----------|
| -----          |        |        |        |           |
| Genotype       | SS     | SF     | FF     | Total     |
| Number, obs    | 36     | 47     | 23     | = 106 = N |
| Frequency, exp | $p^2$  | $2pq$  | $q^2$  | = 1,00    |
| Number, exp    | $p^2N$ | $2pqN$ | $q^2N$ | = N       |
| Number, exp    | 33.2   | 52.3   | 20.5   | = 106     |
| Deviation      | 2.8    | -5.3   | 2.5    |           |
| Chi-square     | 0.24   | 0.54   | 0.31   | = 1.09    |
| -----          |        |        |        |           |

The expected numbers for Hardy Weinberg equilibrium (exp) are calculated using the multiplication rule for probabilities.

For the SS genotype an S gene from a father and an S gene from a mother has to be drawn, the same repeated 106 times. Therefore the probability for an SS genotype is  $p \cdot p \cdot 106$ . The corresponding arguments for the other two genotypes lead to the results shown in the table above.

Now a Chi-square test for H-W equilibrium can be calculated as the sum of squared deviations, each divided by the expected number.

The test has 1 degree of freedom as there are three classes, and two free parameters given by the material,  $p$  and  $N$ , has to be applied to calculate the expected numbers. The last used parameter ( $q$ ) is not free, as it can be calculated as  $(1 - p)$ .

By use of the chi-square table (chapter 13), with  $DF=1$  the value 3.84 equals the 5% limits for maintaining the  $H_0$  hypothesis, that the data has H-W proportions. Therefore the found deviation between observed and expected numbers has a probability which is larger than 5%. Conclusion: There is no statistical deviation from H-W equilibrium.