

Hardy-Weinberg Equilibrium and Chi-Squared Test

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Hardy-Weinberg Principle of Equilibrium

- Principle of equilibrium to describe a population's genetic makeup
- A population's allele frequencies are inherently stable unless an evolutionary force is acting upon the population
 - Assumes random mating and an infinite population size
- That is, without natural selection, genetic drift, gene flow, or mutation you do not expect allele frequencies to change across generations

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Hardy-Weinberg Principle of Equilibrium

- Typical equation for a single locus with two alleles (biallelic)
- Two alleles, with respective frequencies designated p and q
- Since there are only two alleles, $p + q = 1$
- With two alleles there are three possible genotypes
 - Homozygous allele1 (freq p^2)
 - Heterozygous (freq $2pq$)
 - Homozygous allele2 (freq q^2)
- $p^2 + 2pq + q^2 = 1$

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Practice: “spirit” black bear

- The white “Spirit” black bear differs from the ordinary black bear by a single amino acid change in the *melanocortin 1 receptor gene* (*MC1R*)
- In this population, the gene has two alleles: the “white” allele b and the “black” allele B
- The trait is recessive: white bears have two copies of the white allele of this gene (bb), whereas a bear is black if it has one or two copies of the black allele (Bb or BB)



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Practice: “spirit” black bear

- A study was conducted with 87 bears sampled and genotyped
 - BB : 42
 - Bb : 24
 - bb : 21
- What is the estimated frequency of B allele in this population?
 - Total B in sample = $(42 \times 2) + 24 = 108$
 - Total alleles in sample = $87 \times 2 = 174$
 - Frequency of $B = 108/174 \approx 0.62$
- What is the estimated frequency of b allele?
 - $B + b = 1$, so...
 - $\sim 0.62 + b = 1$, thus $b \approx 0.38$

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Practice: “spirit” black bear

- Observed vs expected genotype frequencies
- Observed:
 - BB : 42
 - Bb : 24
 - bb : 21
- Expected? Can use Hardy-Weinberg Equilibrium as a null
 - $p^2 + 2pq + q^2 = 1$

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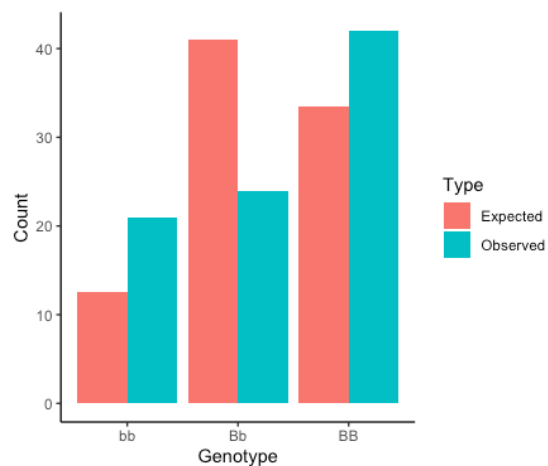
Practice: “spirit” black bear

- R script...
 - Establish observed counts and expected proportion
 - Run `chisq.test()`
 - Interpret P-value
 - If significant then which groups deviate the most from expected?

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Practice: “spirit” black bear

- P-value = 0.0005752
- Excess of homozygotes, deficiency of heterozygotes



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