The Hardy-Weinberg principle

G H Hardy to the editor of Science

"I am reluctant to intrude in a discussion concerning matters of which I have no expert knowledge, and I should have expected the very simple point which I wish to make to have been familiar to biologists" (Hardy 1908)[1].

[1] Hardy, G H (1908). Mendelian proportions in a mixed population. *Science*, 28:49-50. [Link]

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"In a word, there is not the slightest foundation for the idea that a dominant character should show a tendency to spread over a whole population, or that a recessive should tend to die out" (Hardy 1908)[1].

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G H Hardy to the editor of Science

- Hardy-Weinberg Equililbrium (HWE) specifies a null hypothesis as to what genotype frequencies should be in the absence of evolution.
- If we know what genotype frequencies *should be* in the absence of evolution, then we can compare to what genotype frequencies *actually are* to observe evolution.

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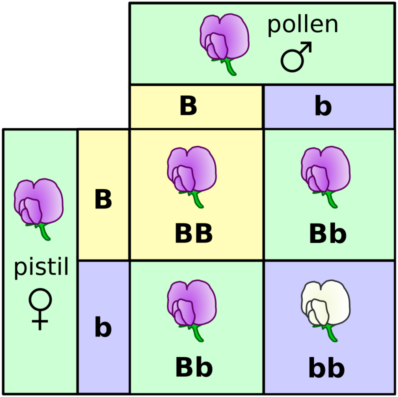
Predicting genotype and phenotype frequencies

- 2 alleles for flower colour
  - B (dominant)
  - b (recessive)
- 3 possible genotypes
  - BB
  - Bb
  - bb
- 2 possible phenotypes
  - Pink (BB, Bb)
  - White (bb)



[1]Image: Public Domain

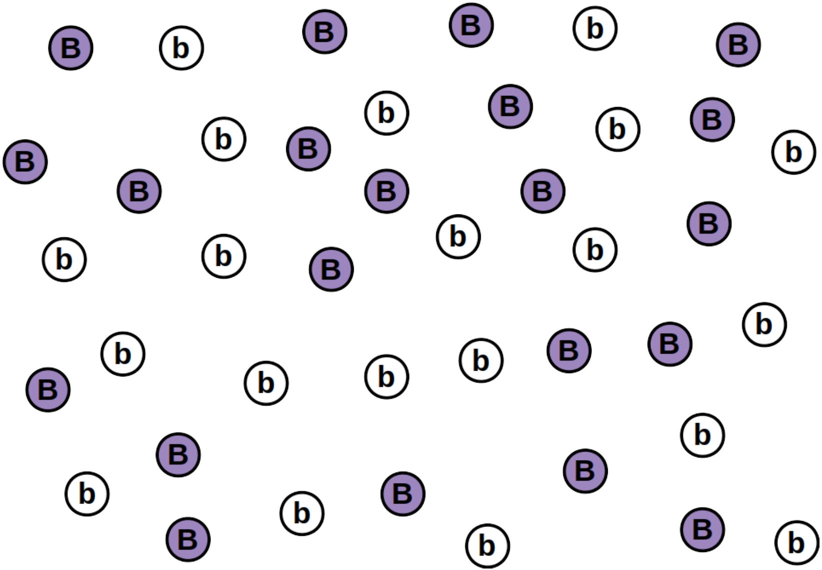
Predicting genotypes from allele frequencies

What about the whole population of pea plants?

Can we predict the frequency of each genotype (BB, Bb, and bb) if we know the frequency of each allele (B and b) in the population?

[1] Image: Public Domain

Hardy-Weinberg Equilibrium (HWE)



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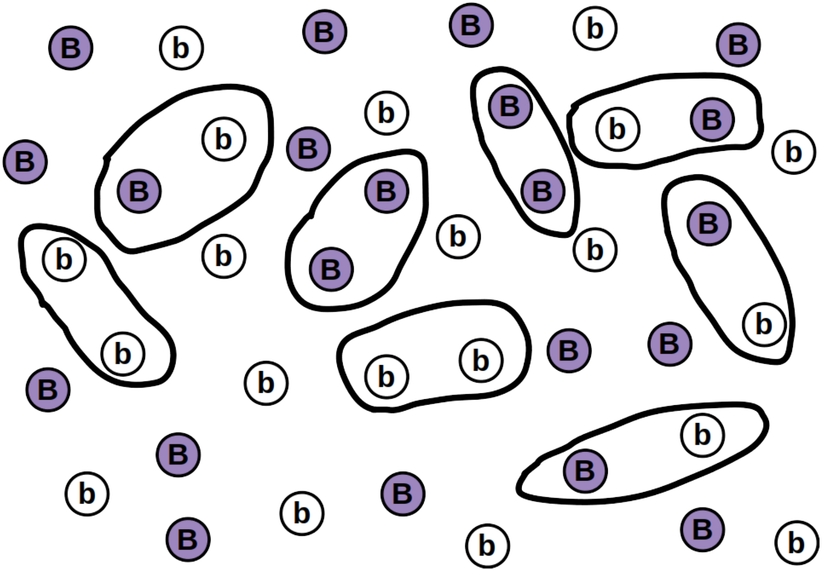

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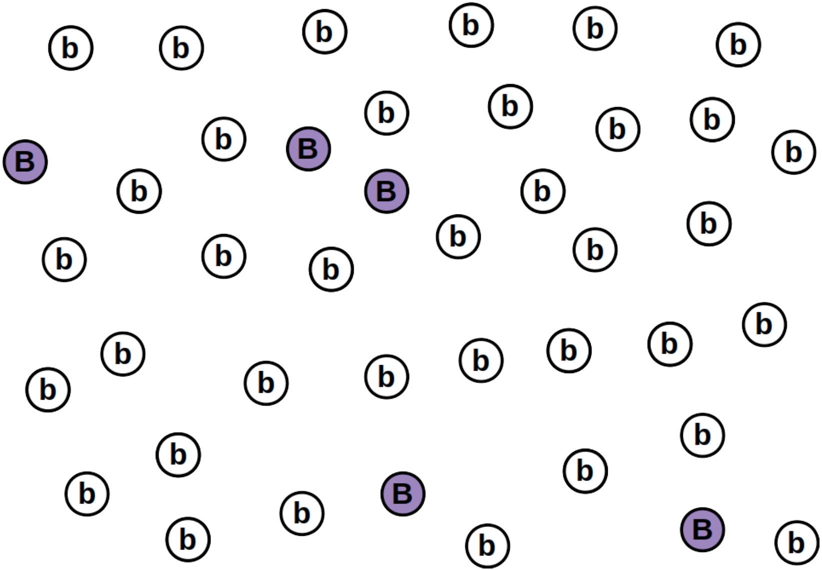

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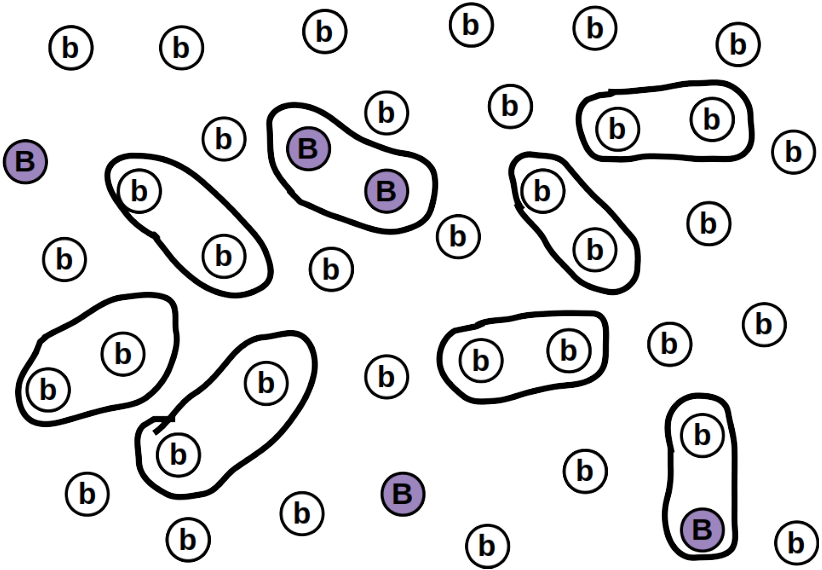

Hardy-Weinberg Equilibrium (HWE) assumptions

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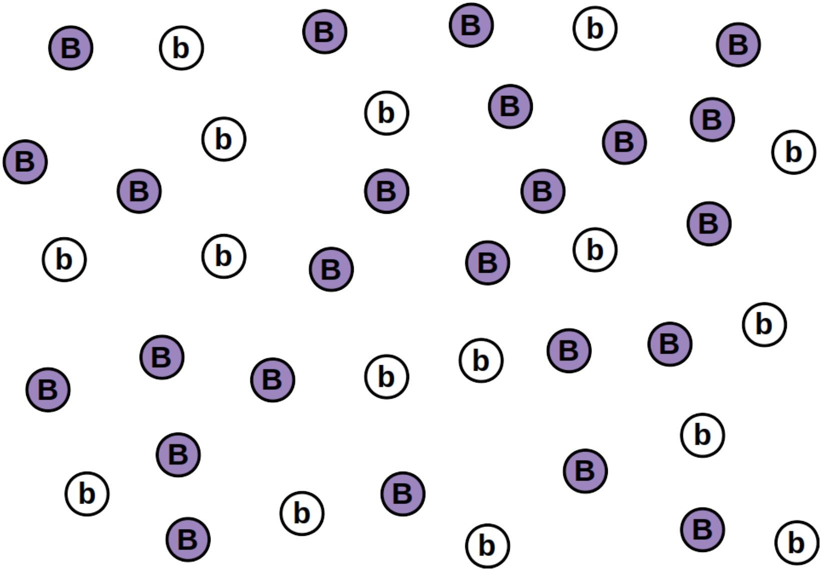
- In other words, if we know the gene or genotype frequencies in one generation, then *in the absence of evolution* the genotype frequencies in the next generation can easily be predicted.
- Define evolution as change in allele frequencies

Hardy-Weinberg Equilibrium (HWE) assumptions

- No natural selection
- No mutation
- No migration (no gene flow)
- Infinite population size
- Mating is random
- Non-overlapping generations

Although we know the above will never be entirely true, in practice, many genes behave as if they are in HWE.

Frequencies of the B and b alleles (40 total)



Allele frequencies

$$\begin{aligned} \text{24 B alleles: } \Pr(\text{B}) &= \text{p} = 24/40 = 0.6 \\ \text{16 b alleles: } \Pr(\text{b}) &= \text{q} = 16/40 = 0.4 \end{aligned}$$

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Genotype combinations

Allele 1	B	B	b	b
Allele 2	B	b	B	b

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Genotype probabilities

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	$\times$	$\times$	$\times$	$\times$
Allele 2	p	q	p	q
Freq.	pp	pq	qp	qq

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

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Hardy-Weinberg Equilibrium (HWE)

	Sperm gametic pool	
Egg gametic pool	Pr(B) = p	Pr(b) = q
Pr(B) = p	0.36	0.24
Pr(b) = q	0.24	0.16

Four conclusions of HW model

1. HW genotype frequencies are determined solely by gene frequencies and not by the frequencies of genotypes in the preceding generation.

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3. When a population's genotype frequencies are related to its gene frequencies in the ratios $Fr(AA) = p^2$, $Fr(Aa) = 2pq$, and $Fr(aa) = q^2$, then the population is said to be "in HW equilibrium".

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4. HWE represents a null hypothesis against which we can identify and estimate the forces that cause deviations from that equilibrium – the forces of evolution.