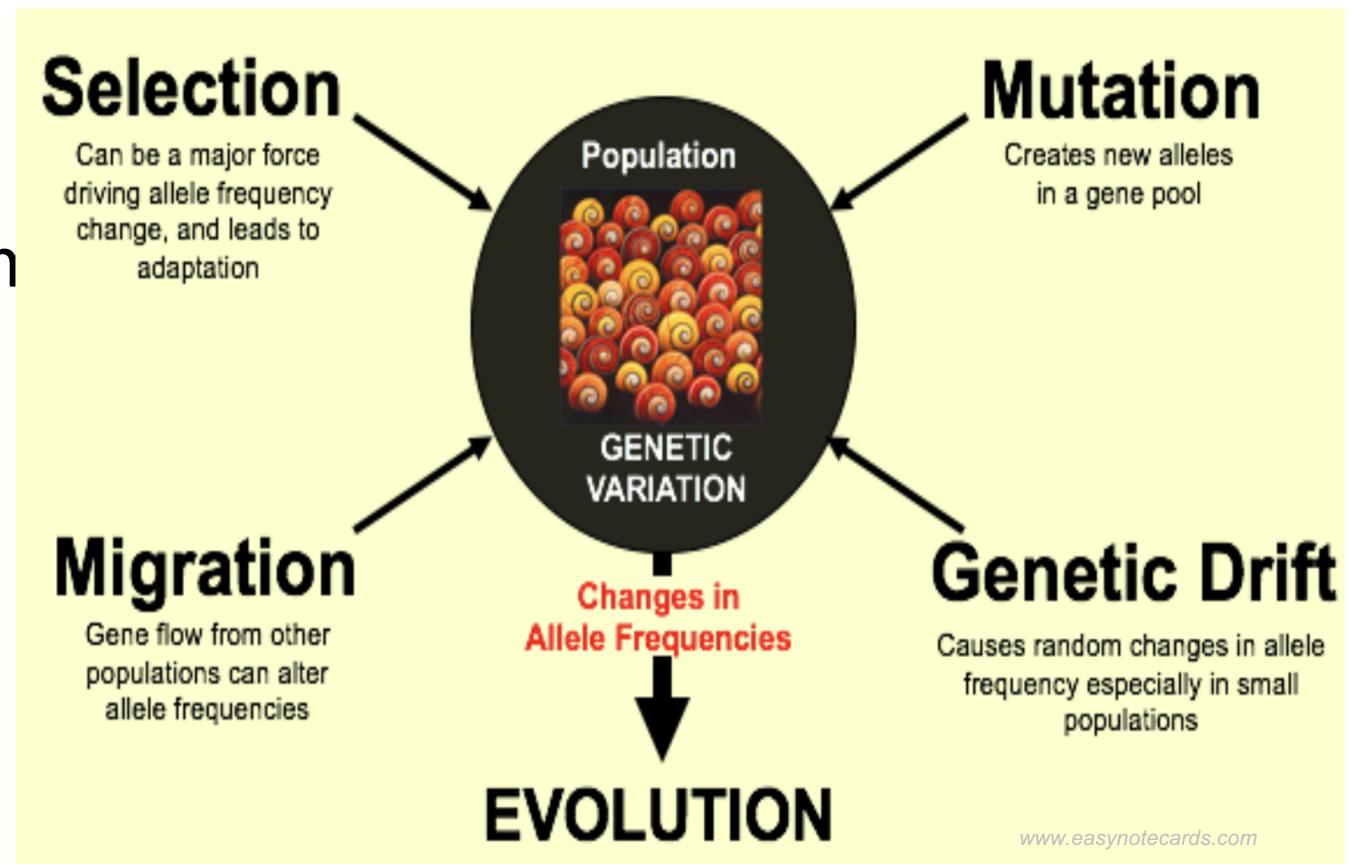


Population Genetics

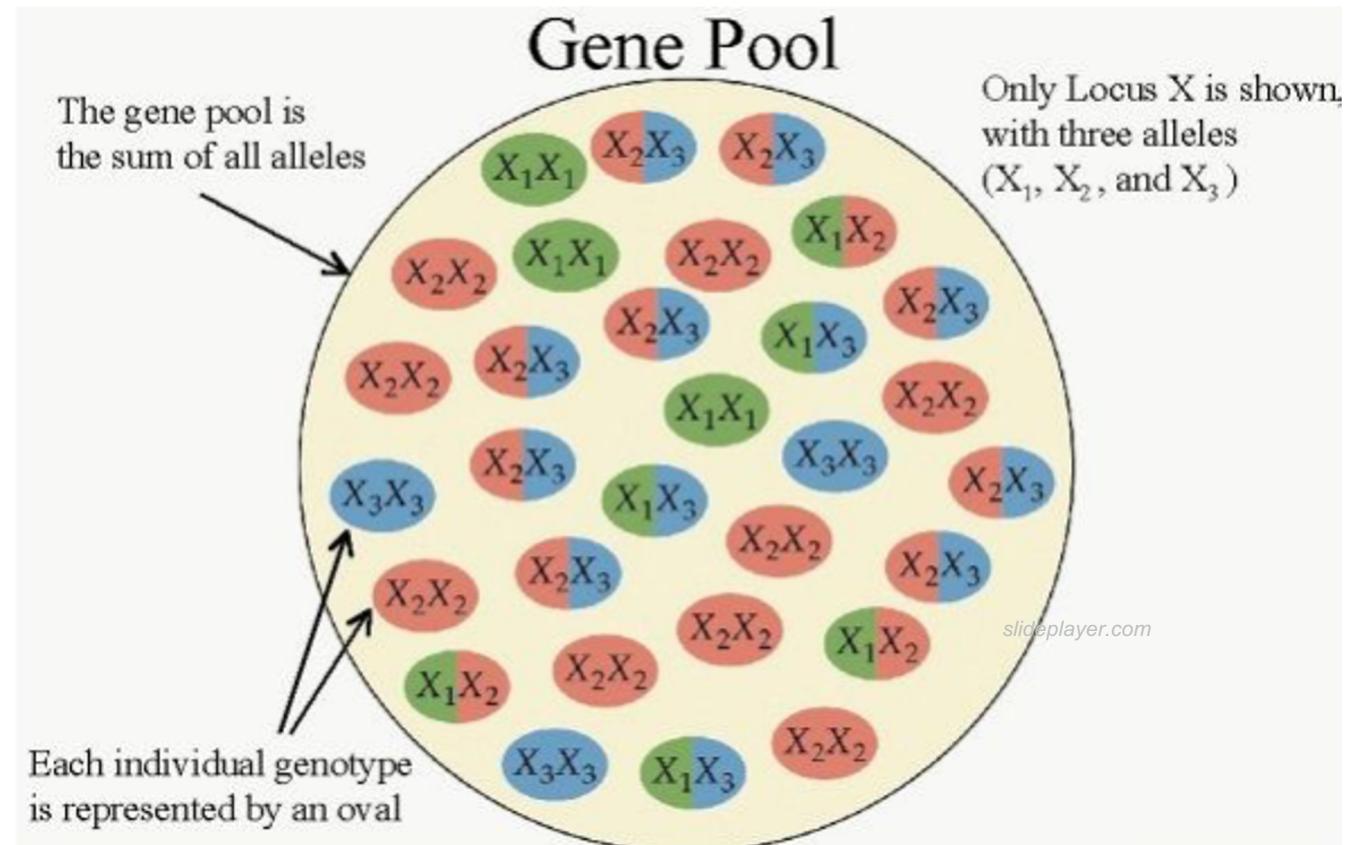
Population genetics

- Gene pool
- Allele Frequencies
- Hardy-Weinberg Equilibrium
- HW assumptions
- Testing for HW
- Chi-squared test
- Inbreeding



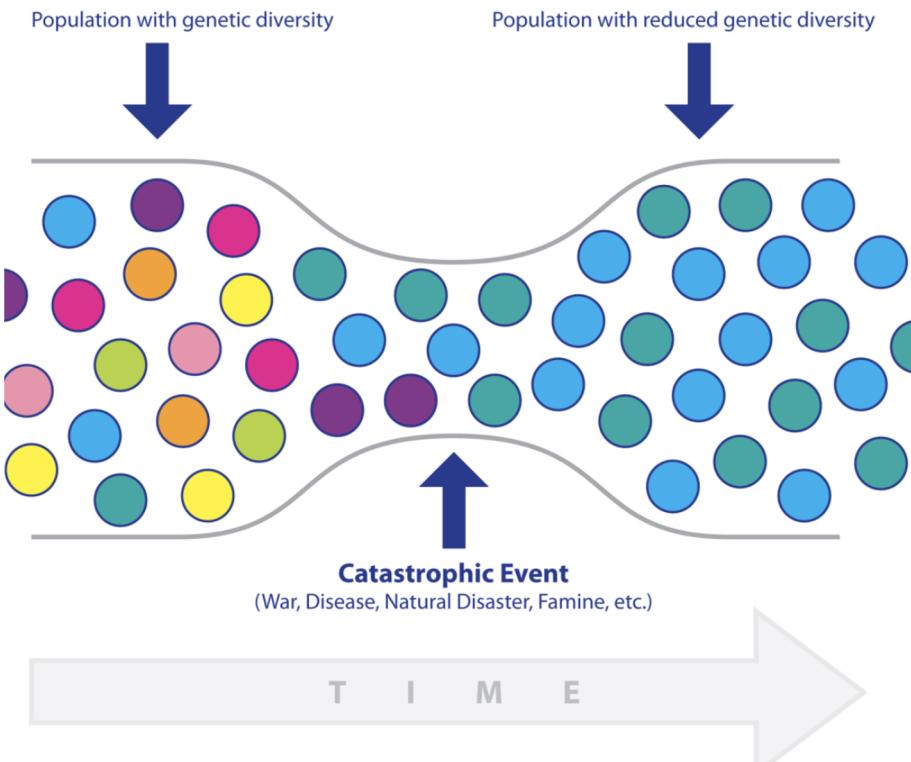
Gene pools

- A gene pool is the collection of different *genes* within a population.
- Gene pool is the sum of all the *alleles* at all of the genes of a population.



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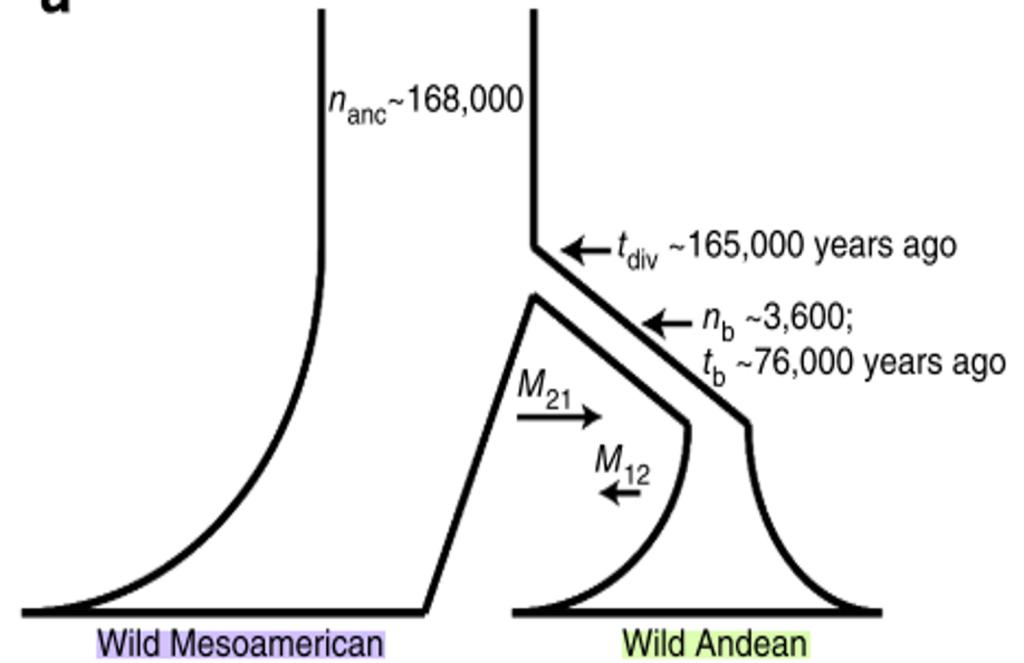


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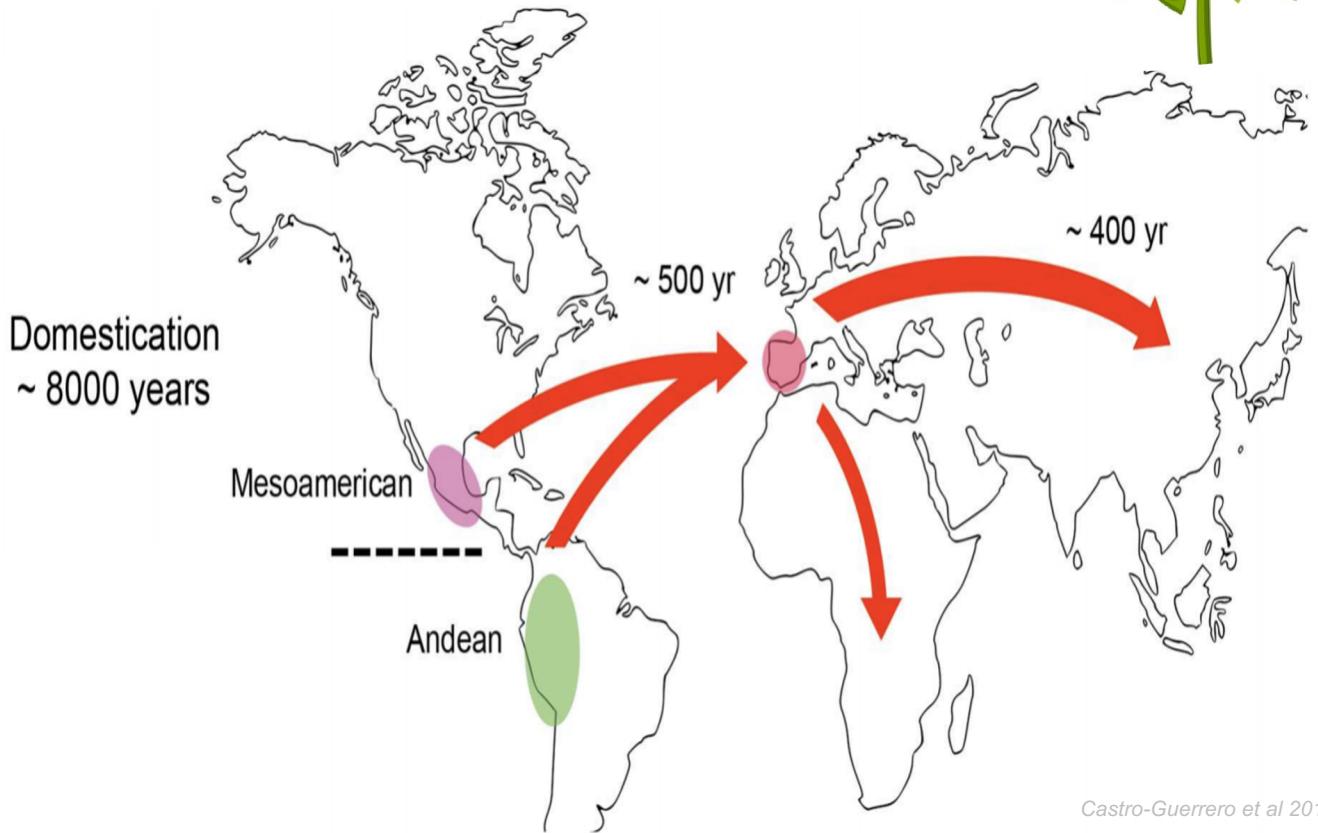
Phaseolus vulgaris Gene pools



a



Schmutz et al 2014



Castro-Guerrero et al 2016.

Allele Frequencies

- Consider a locus with two possible alleles (**A** and **a**)

$$A = p, a = q$$

- We can estimate the **frequency** of AA genotype by dividing the number of AA individuals by total number of individuals in the population.
- Allele Frequencies:

$$AA + Aa + aa = 1$$

$$p = f(AA) + \frac{1}{2} f(Aa) = \text{frequency of } A \qquad \qquad p + q = 1$$

$$q = f(aa) + \frac{1}{2} f(Aa) = p - 1 \text{ frequency of } a$$

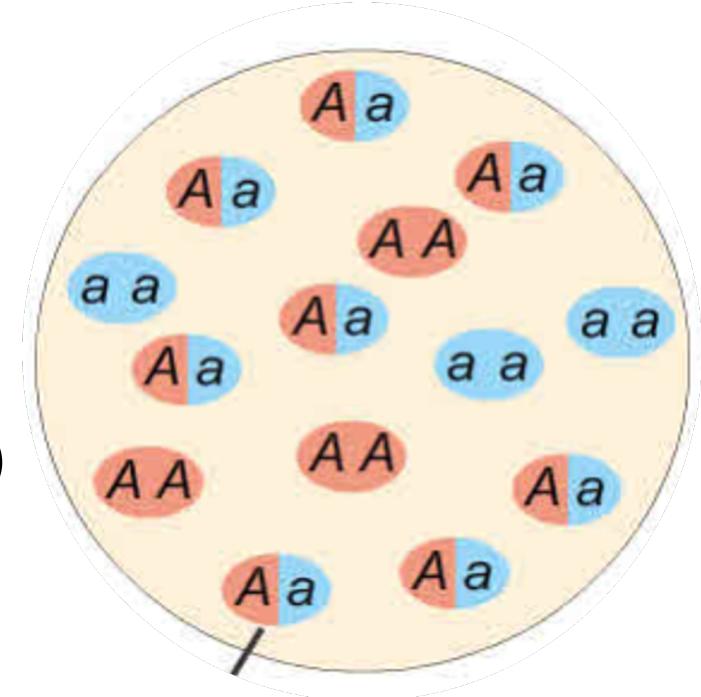
Hardy-Weinberg Equilibrium

Meiosis:

- Aa produces gametes A and a (in equal frequency)
- The homozygous AA only produce one gamete: A
- Likewise aa only produce one gamete: a

Random sampling of gametes (random sampling of parents + no mutation)

$$p(A)=p \quad p(a) = q = (1-p)$$



Expected genotype frequencies (random mating)

$$p(AA)=p^2, \quad p(aa)=q^2, \quad p(Aa)=2pq$$

Hardy-Weinberg Equilibrium

$$N = 14$$

$$AA = 3$$

$$Aa = 8$$

$$aa = 3$$

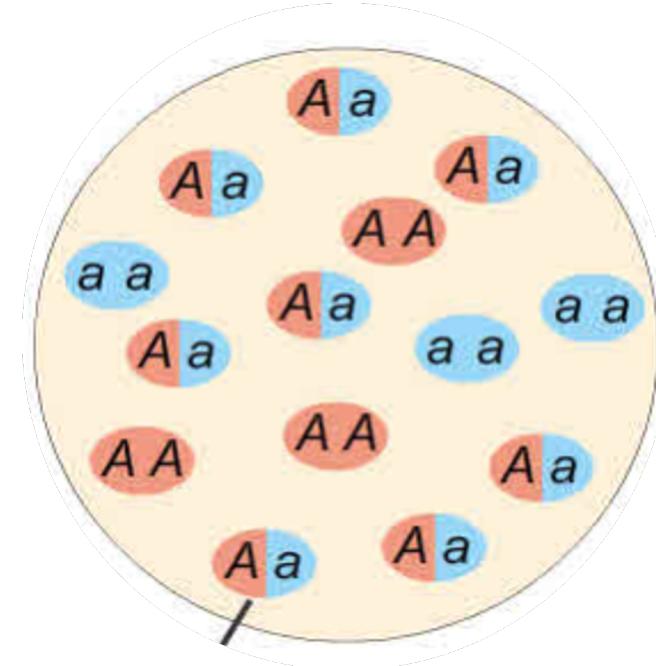
$$p(A) = p \quad p(a) = q = (1-p)$$

$$p = f(AA) + \frac{1}{2} f(Aa) = \text{frequency of } A$$

$$p = (3/14) + \frac{1}{2} (8/14) = 0.5$$

$$q = (1-p)$$

$$q = 1 - 0.5 = 0.5$$



Hardy-Weinberg Equilibrium

$N = 14$, AA = 3 , Aa = 8, aa = 3

$$p = 0.5$$

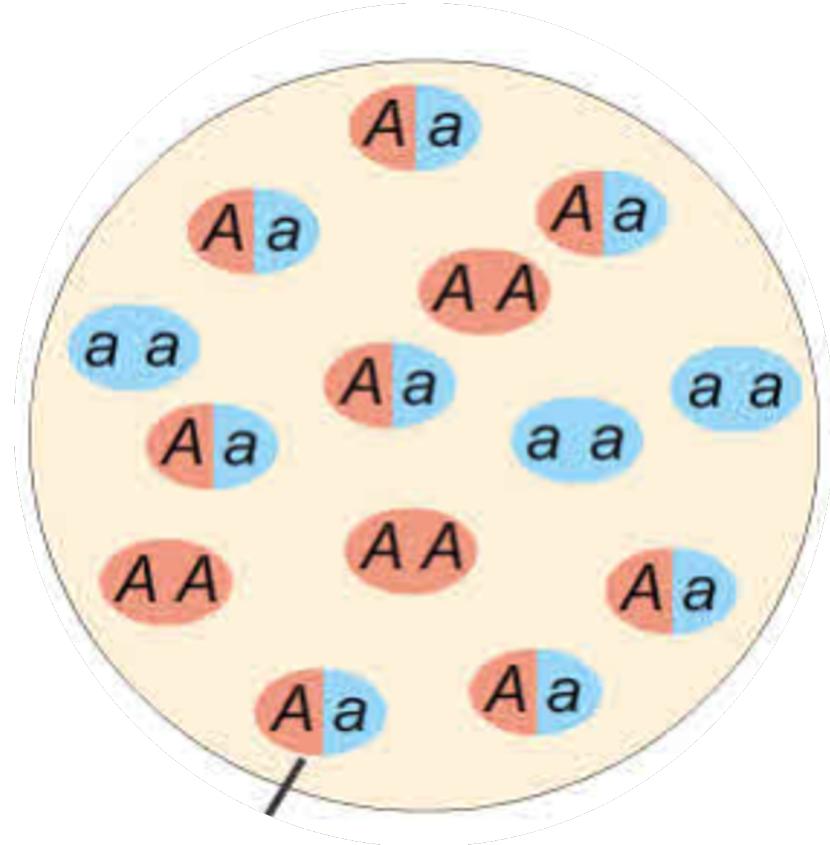
$$q = 0.5$$

Expected genotype frequencies :

$$p(AA) = p^2, p(Aa) = 2pq, p(aa) = q^2$$

$$p^2 + 2pq + q^2$$

$$0.5^2 + 2(0.5 \cdot 0.5) + 0.5^2 = 0.25, 0.5, 0.25$$

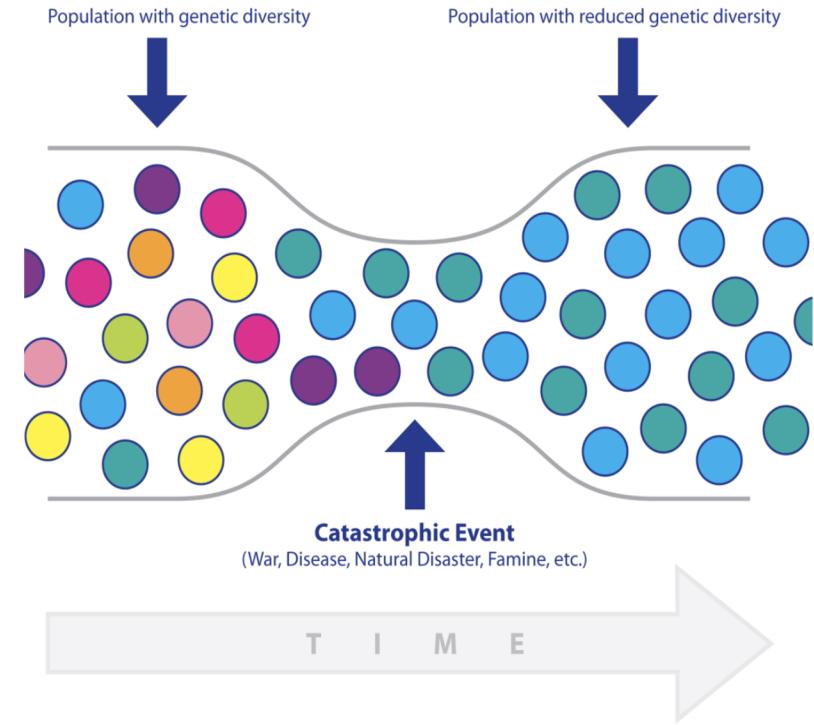


Hardy-Weinberg assumptions

- Random mating
- Genotypes have same viability
- Population must not be divided into subpopulations
- Apply only to large populations
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- No drift

If any of these assumptions is not true, the population could depart from HWE.

These departures in most cases manifest as an excess of homozygosity relative to HWE.



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Hardy-Weinberg equilibrium

Generation	Genotype frequencies			Gene frequencies	
	A/A	A/a	a/a	A	a
t_0	0.64	0.32	0.04	0.8	0.2
t_1	0.64	0.32	0.04	0.8	0.2
\vdots	\vdots	\vdots	\vdots	\vdots	\vdots
t_n	0.64	0.32	0.04	0.8	0.2

In a large population, genetic variation is neither created nor destroyed

Testing for HW Proportions

- Genes not fitting expected proportions indicate that at least one of the HW assumptions is not true.

	Genotypes			Sum
	A/A	A/G	G/G	
Observed number	17	55	12	84
Observed frequency	0.202	0.655	0.143	1
Expected frequency	0.281	0.498	0.221	1
Expected number	23.574	41.851	18.574	84
(Observed - expected) ² /expected	1.833	4.131	2.327	8.29

Source: International HapMap Project (www.hapmap.org).

$$\chi^2 = \sum \frac{(O-E)^2}{E}$$

Observed frequency

$$p^2 = 17/84 = 0.202$$

$$2pq = 55/84 = 0.655$$

$$p = f(AA) + \frac{1}{2}f(AG) = 0.202 + \frac{1}{2}0.655 = 0.53$$

$$q = 1-p = 0.47$$

Expected frequency

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

$$0.53^2 + 2(0.53 * 0.47) + 0.47^2$$

$$0.281 + 0.498 + 0.221 = 1$$

Testing for HW Proportions

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H_0 : HWE
 H_A : not in HWE

We use 1 df

Degree of Freedom	Probability of Exceeding the Critical Value								
	0.99	0.95	0.90	0.75	0.50	0.25	0.10	0.05	0.01
1	0.000	0.004	0.016	0.102	0.455	1.32	2.71	3.84	6.63
2	0.020	0.103	0.211	0.575	1.386	2.77	4.61	5.99	9.21
3	0.115	0.352	0.584	1.212	2.366	4.11	6.25	7.81	11.34
4	0.297	0.711	1.064	1.923	3.357	5.39	7.78	9.49	13.28
5	0.554	1.145	1.610	2.675	4.351	6.63	9.24	11.07	15.09
6	0.872	1.635	2.204	3.455	5.348	7.84	10.64	12.59	16.81
7	1.239	2.167	2.833	4.255	6.346	9.04	12.02	14.07	18.48
8	1.647	2.733	3.490	5.071	7.344	10.22	13.36	15.51	20.09
9	2.088	3.325	4.168	5.899	8.343	11.39	14.68	16.92	21.67
10	2.558	3.940	4.865	6.737	9.342	12.55	15.99	18.31	23.21

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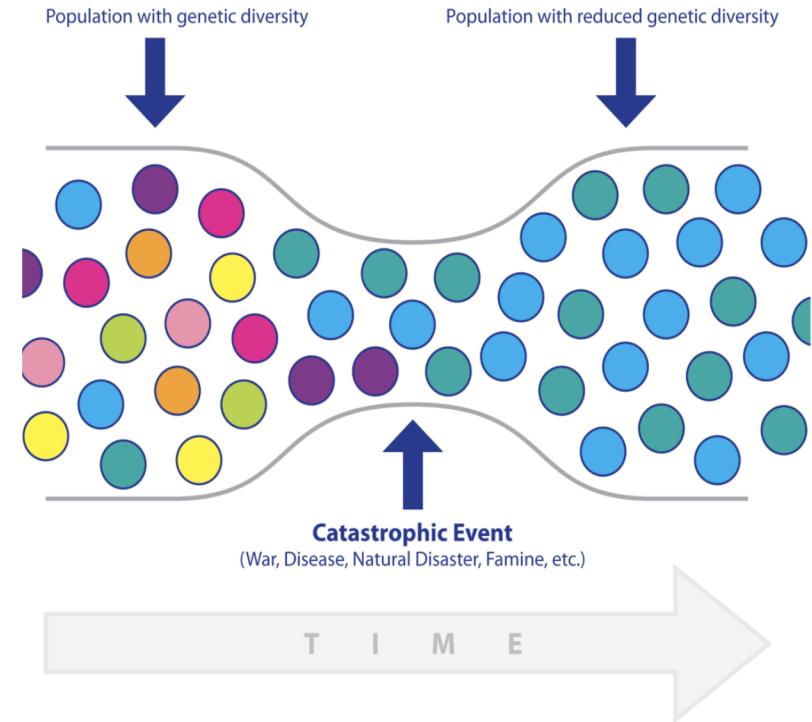
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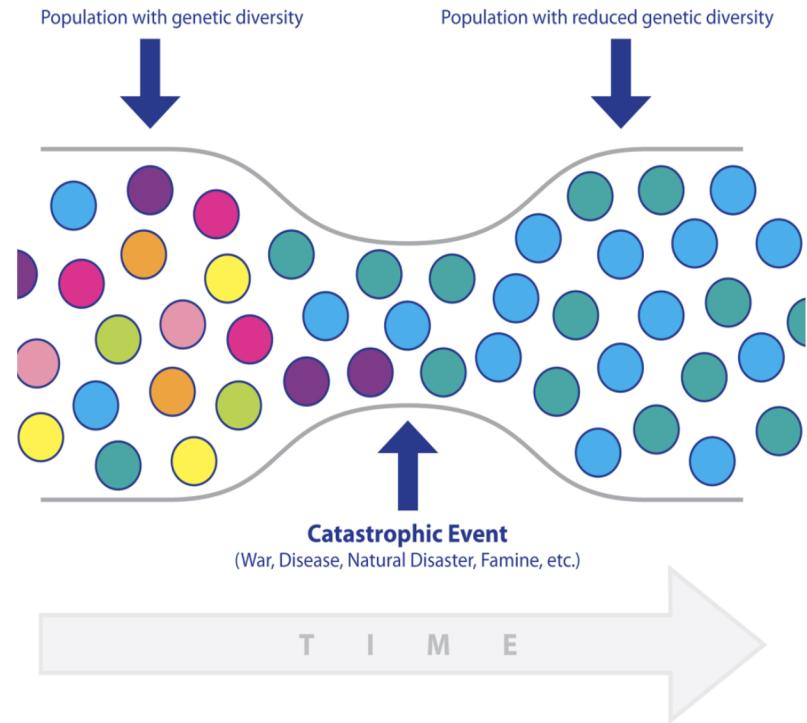
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<https://co.pinterest.com/>

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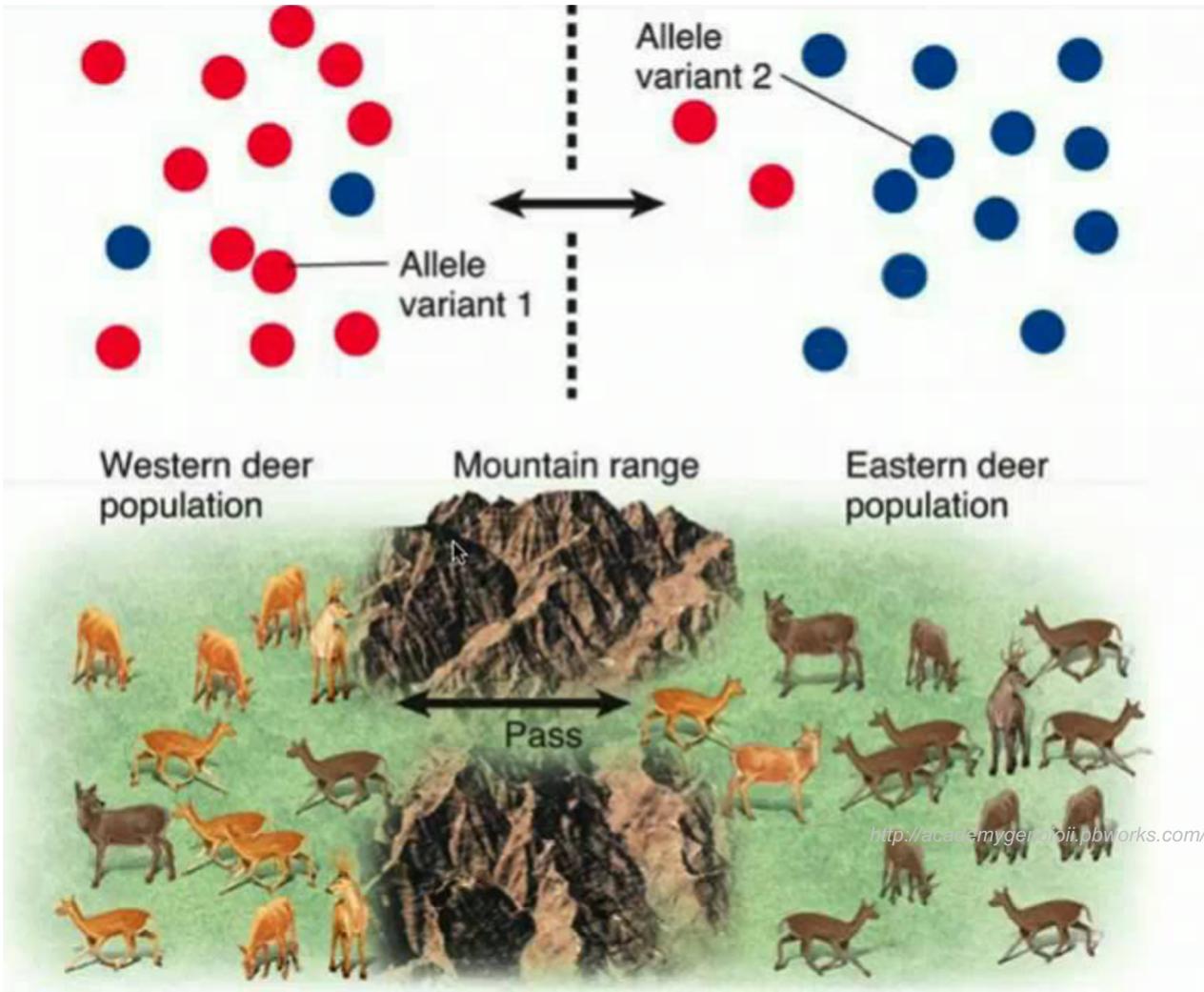


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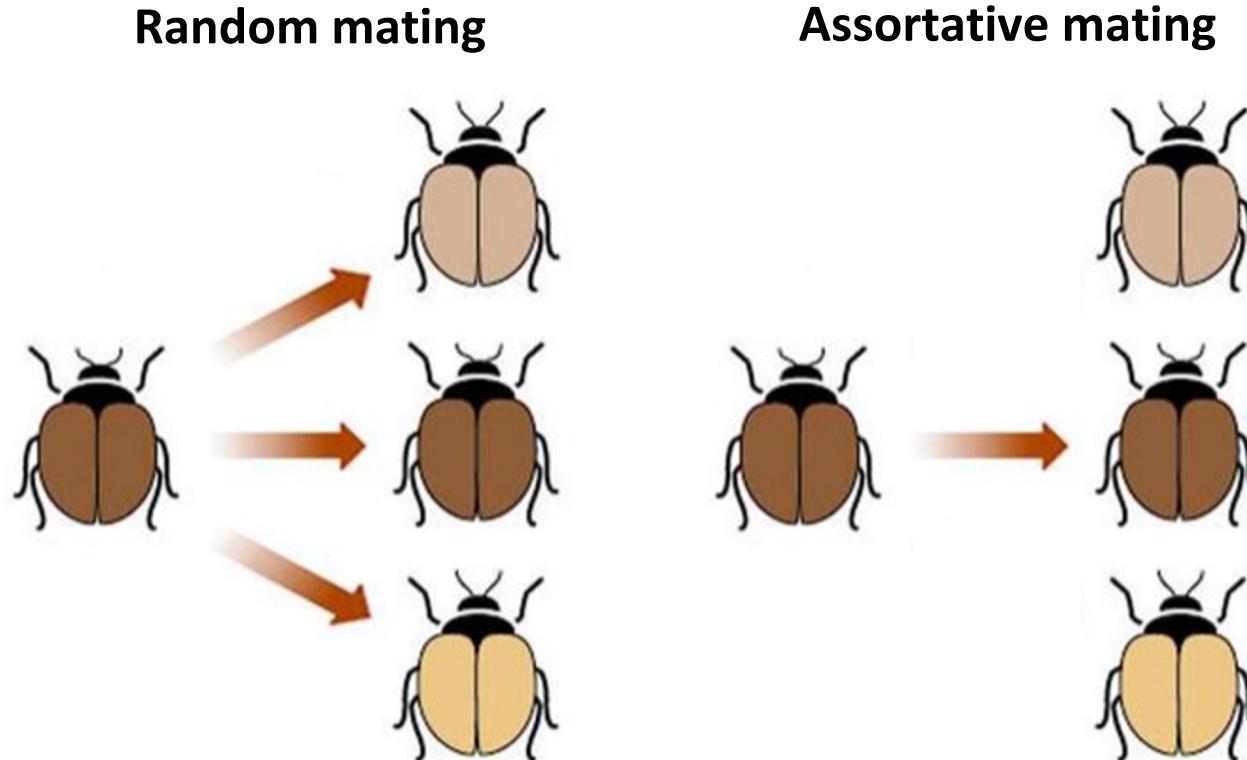
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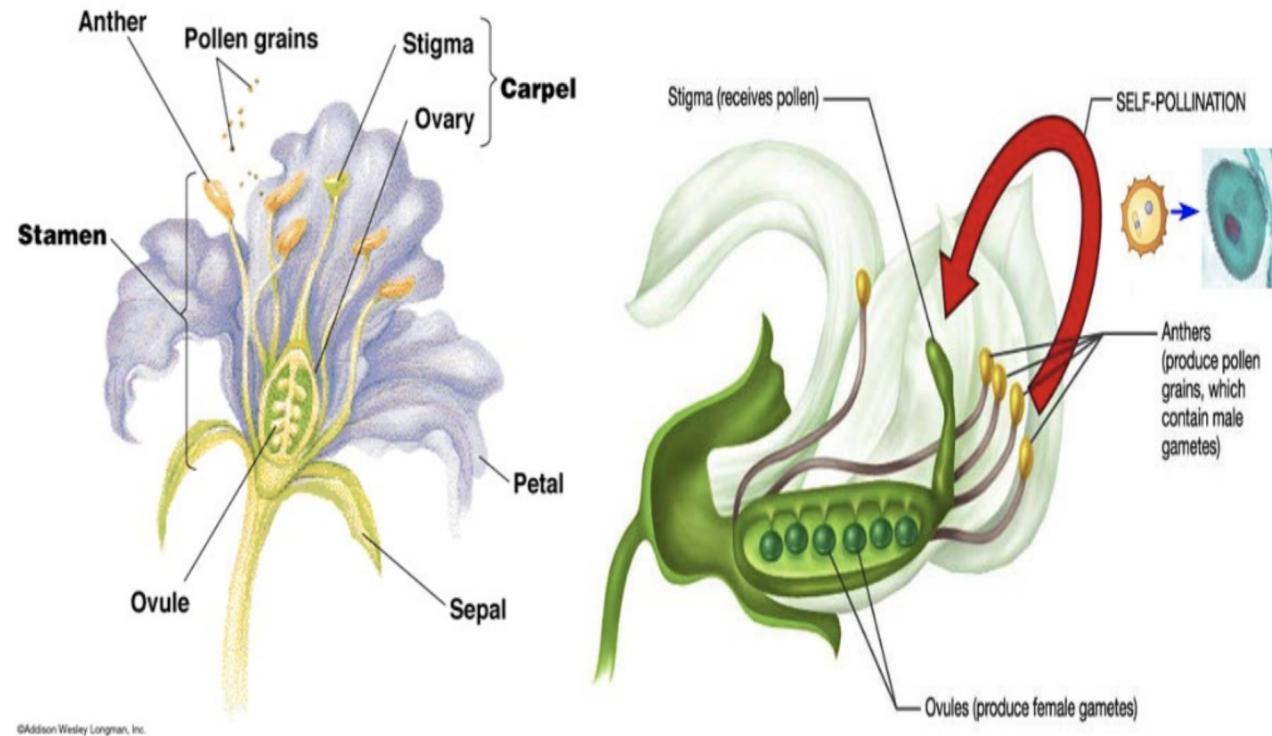
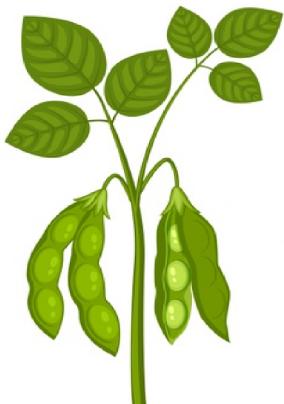
Bias in mate choice: isolation by distance



Bias in mate choice: assortative mating



Bias in mate choice: inbreeding

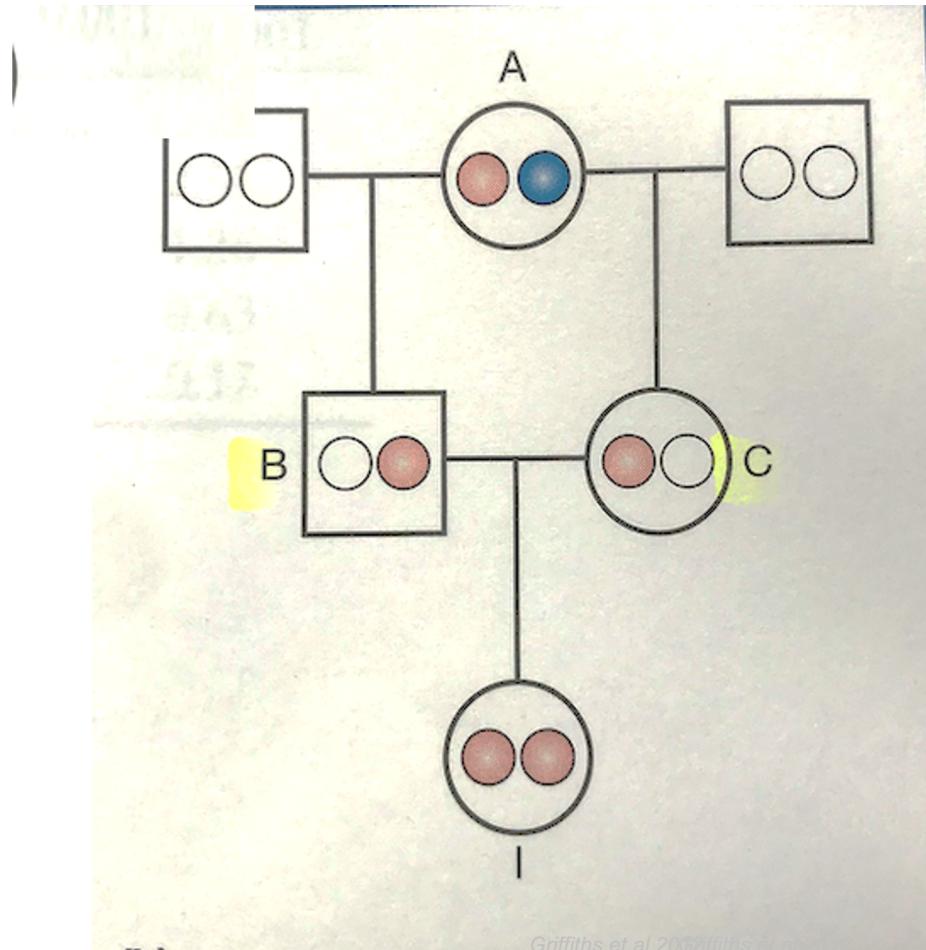


Inbreeding or mating between relatives!

Inbreeding

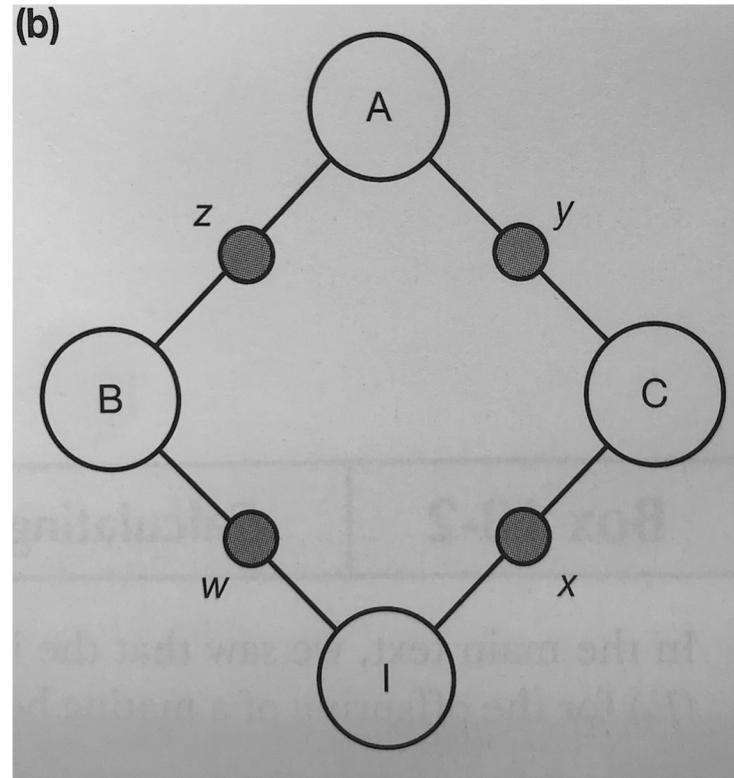
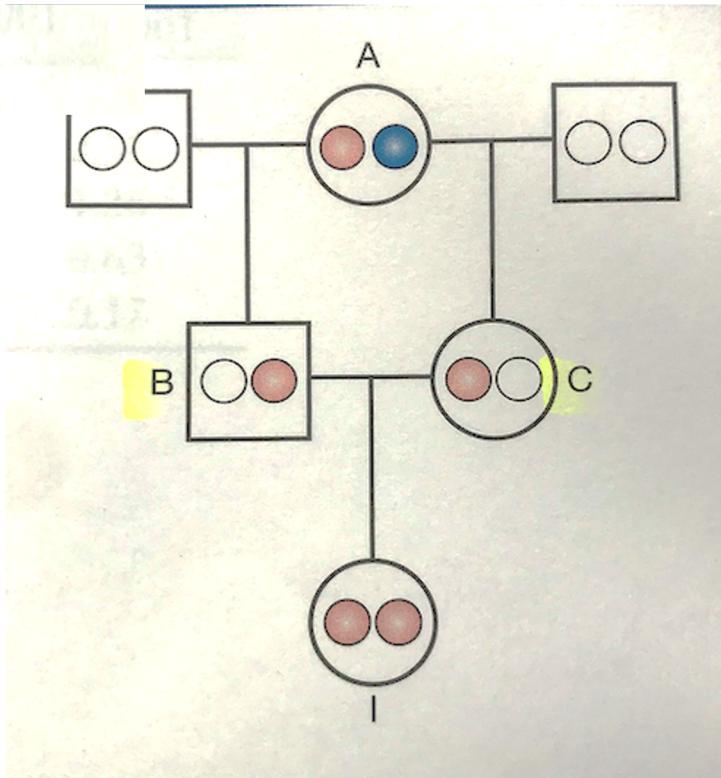
Inbreeding coefficient (f):

The probability that two alleles at a given locus are identical by descent.



Griffiths et al 2008

Inbreeding coefficient



$$F_i = P(\text{IBD}) = \left(\frac{1}{2}\right)^{Q-1}$$

$$F_i = \left(\frac{1}{2}\right)^{4-1} = 0.125$$

What if A is inbred?

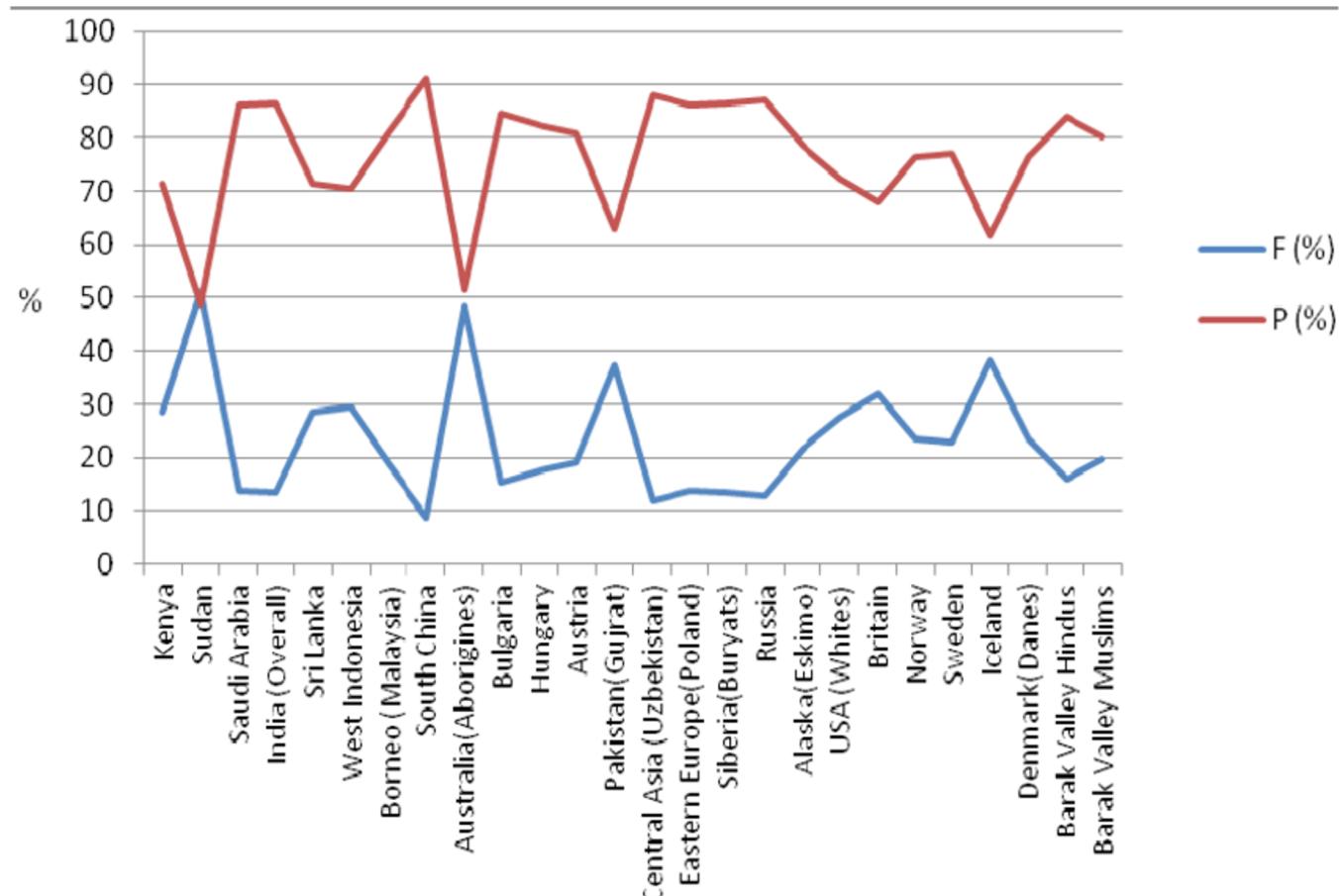
$$F_i = \left(\frac{1}{2}\right)^{Q-1} (1+F_A)$$

Panmictic Index: A measure of the relative heterozygosity

$$P = (1 - F) = H/H_0$$

H: Observed prop. of heterozygous

H_0 : 2pq HWE proportion



Review

- Gene pool
- Allele Frequencies
- Hardy-Weinberg Equilibrium
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- Testing for HW
- Chi-squared test
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Graphical Representation of the Hardy-Weinberg Principle

