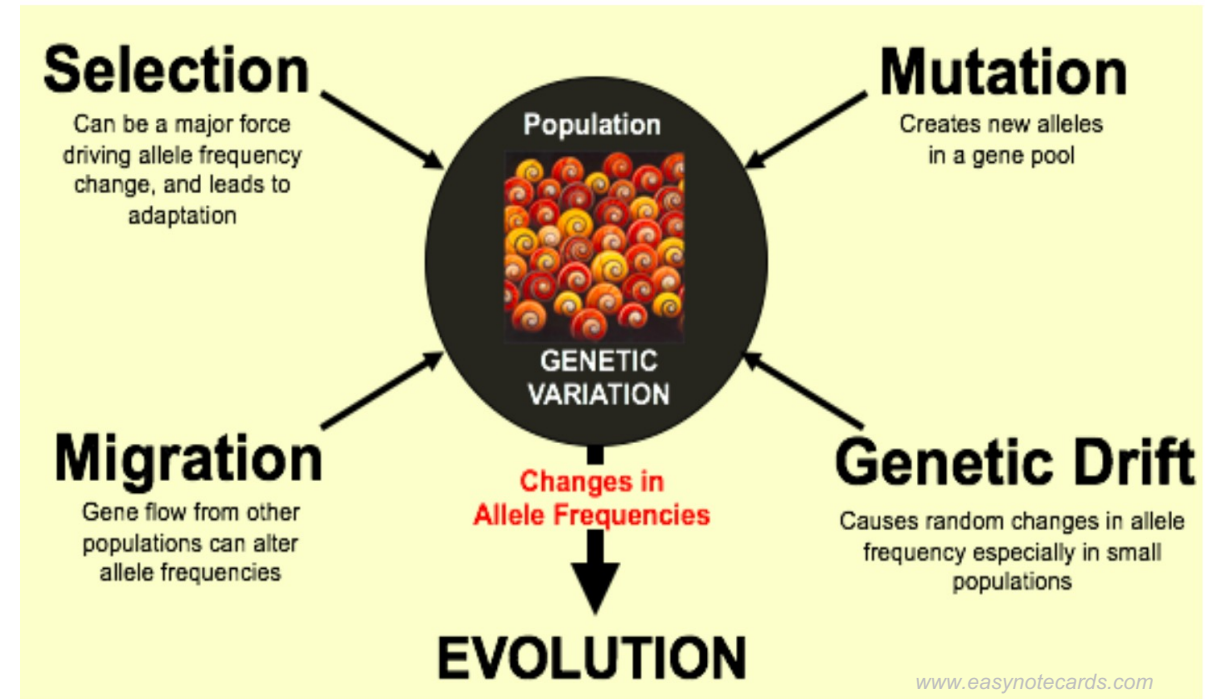


Population genetics

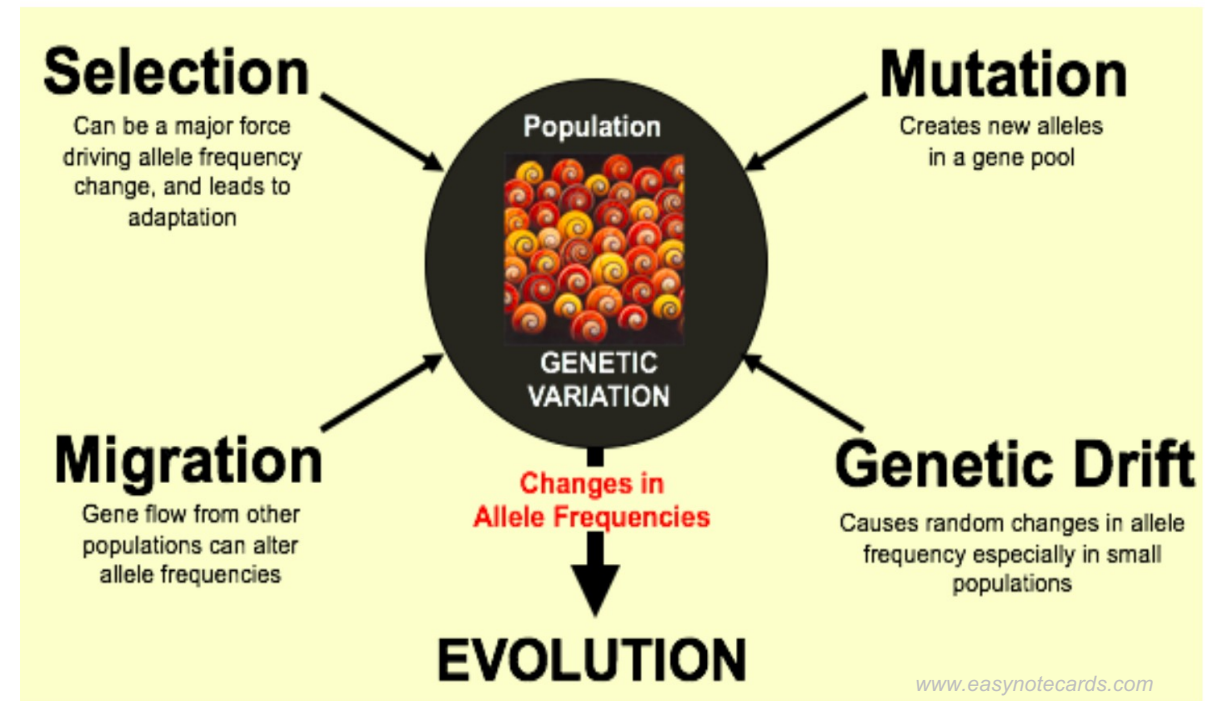
Population genetics

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



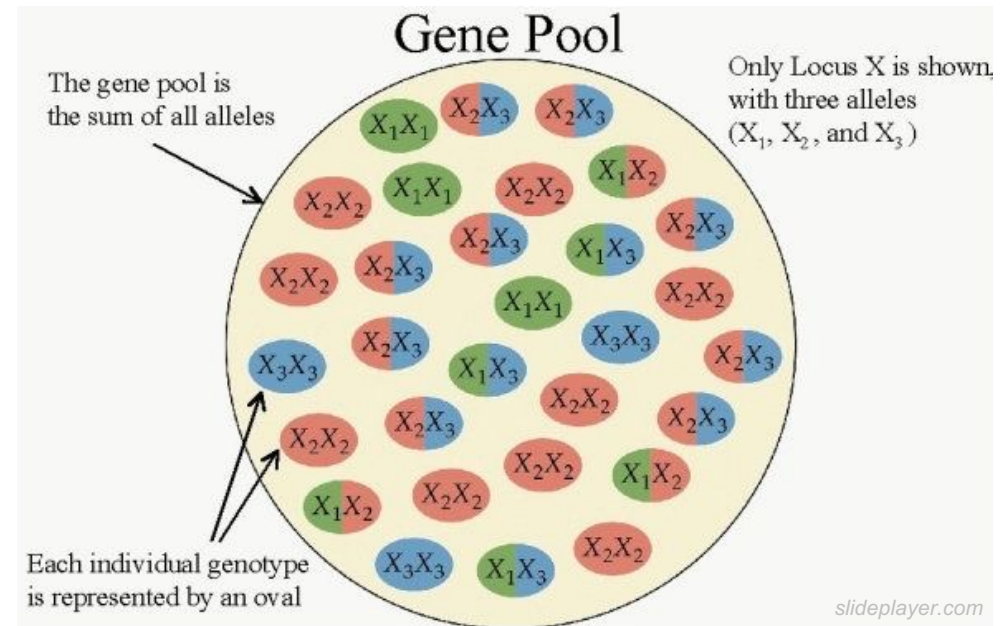
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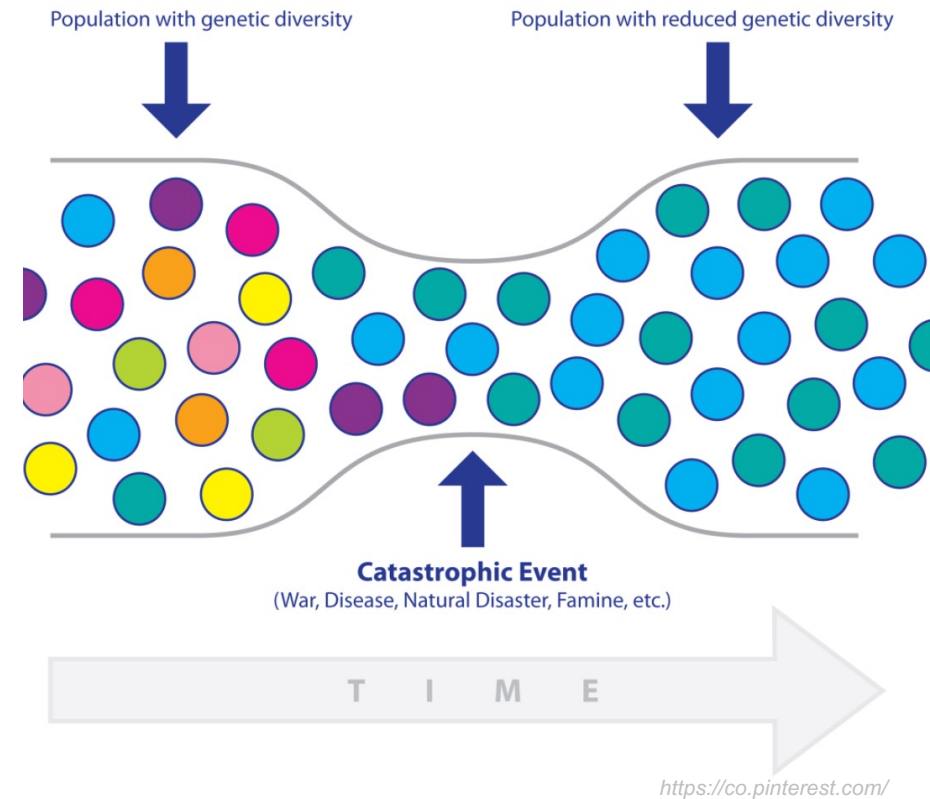
Gene pools

- 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.

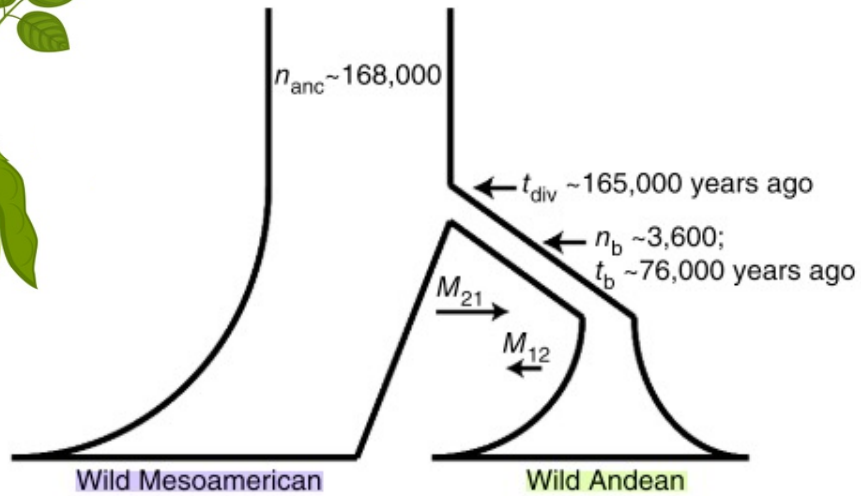


Gene pools

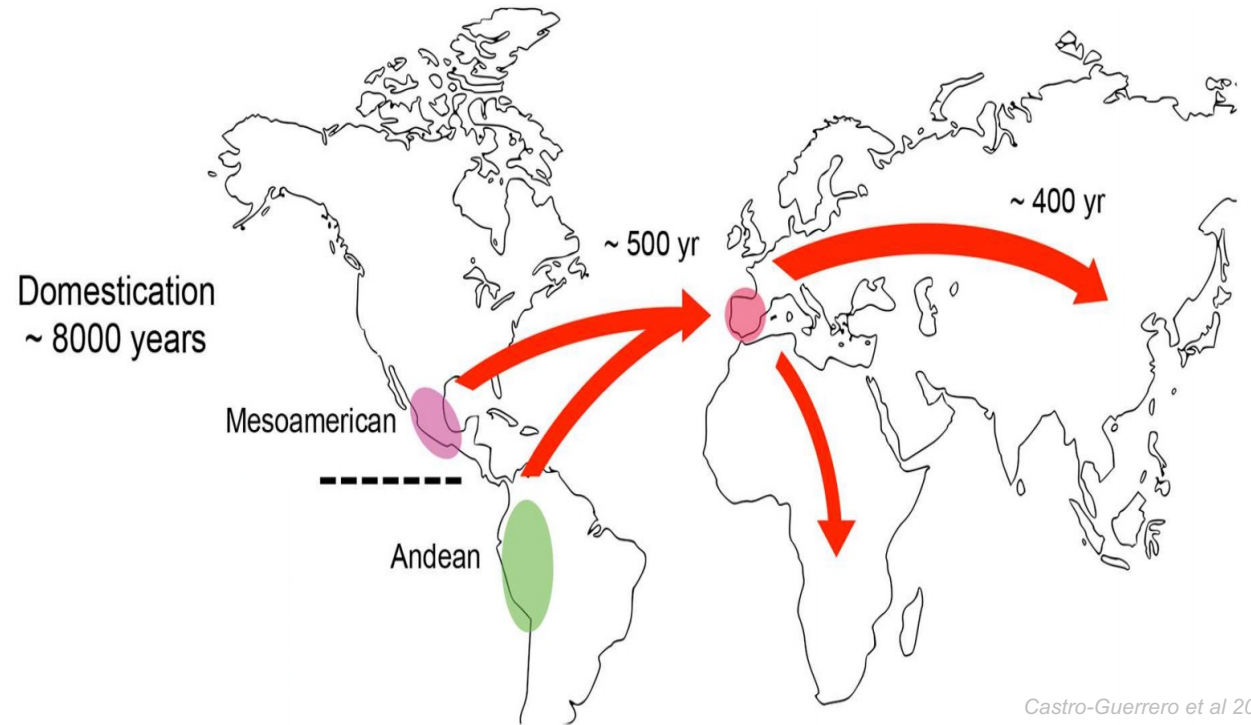
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Gene pools



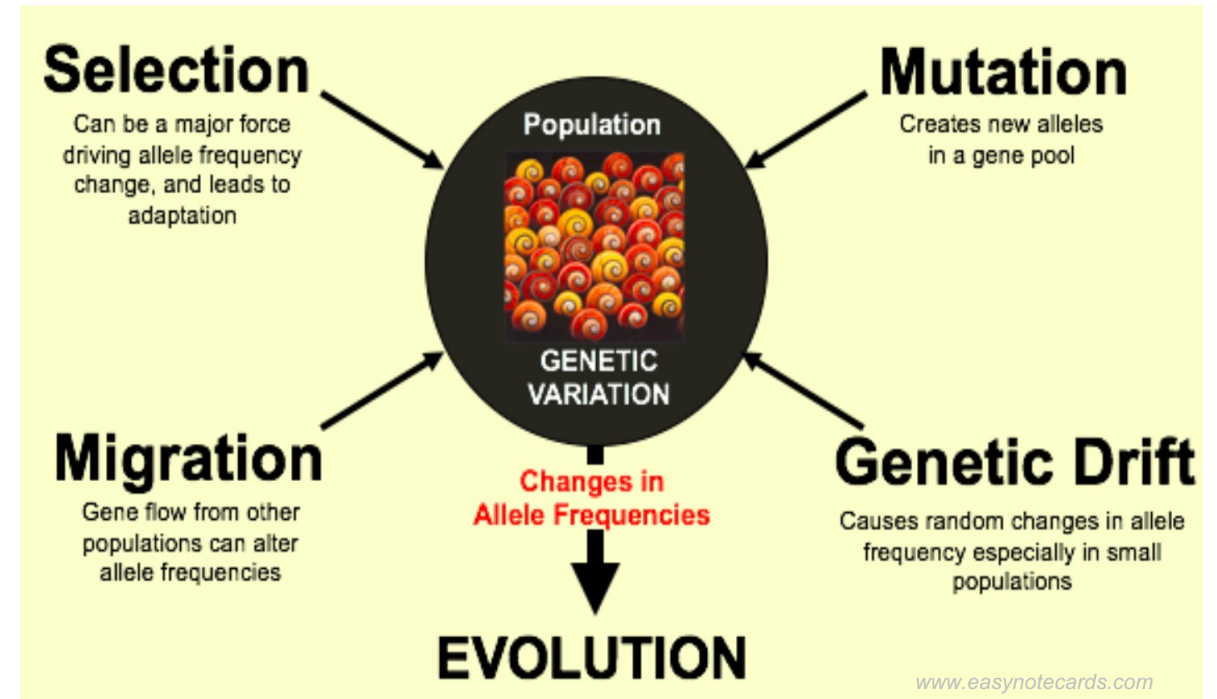
Schmutz et al 2014



Castro-Guerrero et al 2016.

Population genetics

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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.

$$AA + Aa + aa = 1$$

- Allele Frequencies:

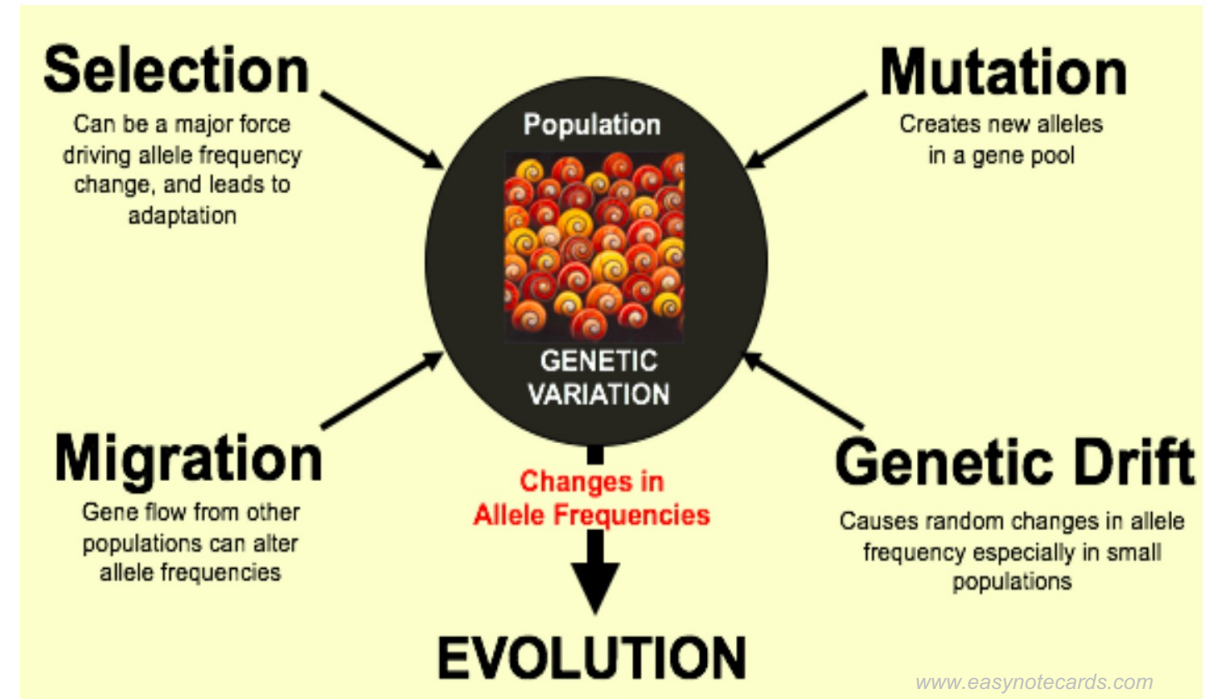
$$p = f(AA) + \frac{1}{2} f(Aa) = \text{frequency of } A$$

$$p + q = 1$$

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

Population genetics

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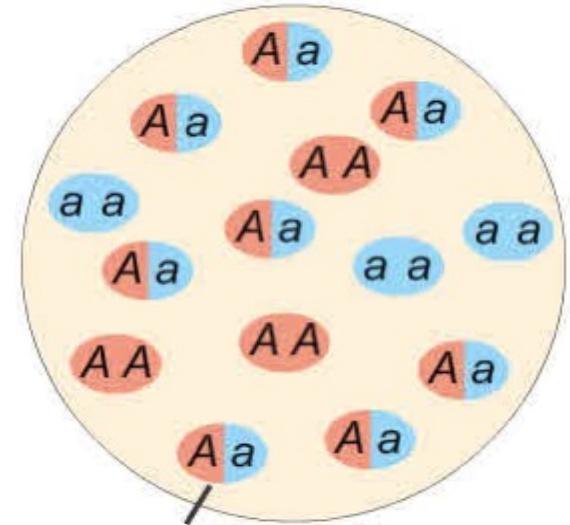
Hardy-Weinberg Equilibrium

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

p^2 = dominant homozygous frequency (AA)

$2pq$ = heterozygous frequency (Aa)

q^2 = recessive homozygous frequency (aa)



Expected genotype frequencies (**random mating**)

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

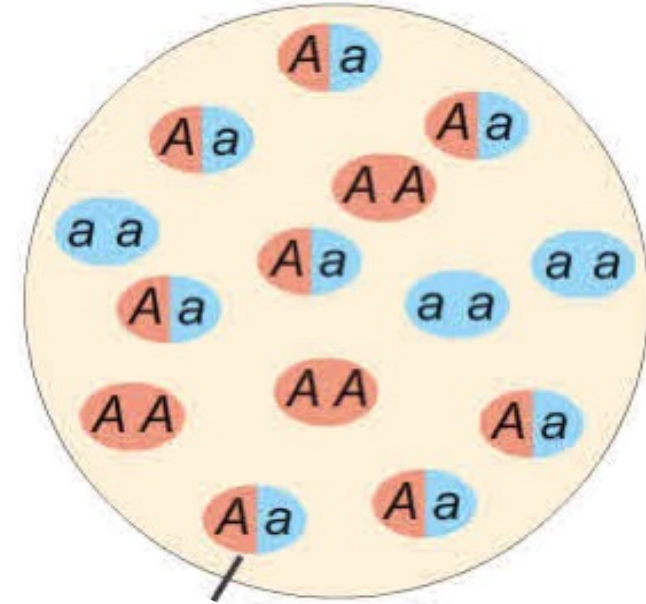
Hardy-Weinberg Equilibrium

$$N = 14$$

$$AA = 3$$

$$Aa = 8$$

$$aa = 3$$



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

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

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

$$q = (1-p)$$

$$q = 1 - 0.5 = 0.5$$

Hardy-Weinberg Equilibrium

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

$p = 0.5$

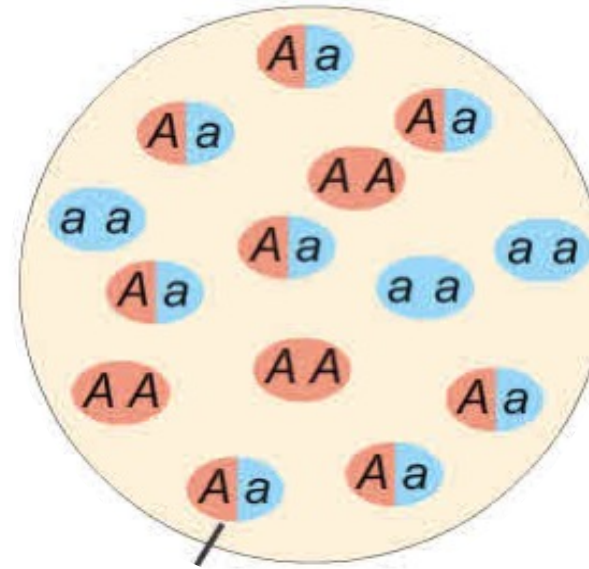
$q = 0.5$

Expected genotype frequencies :

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

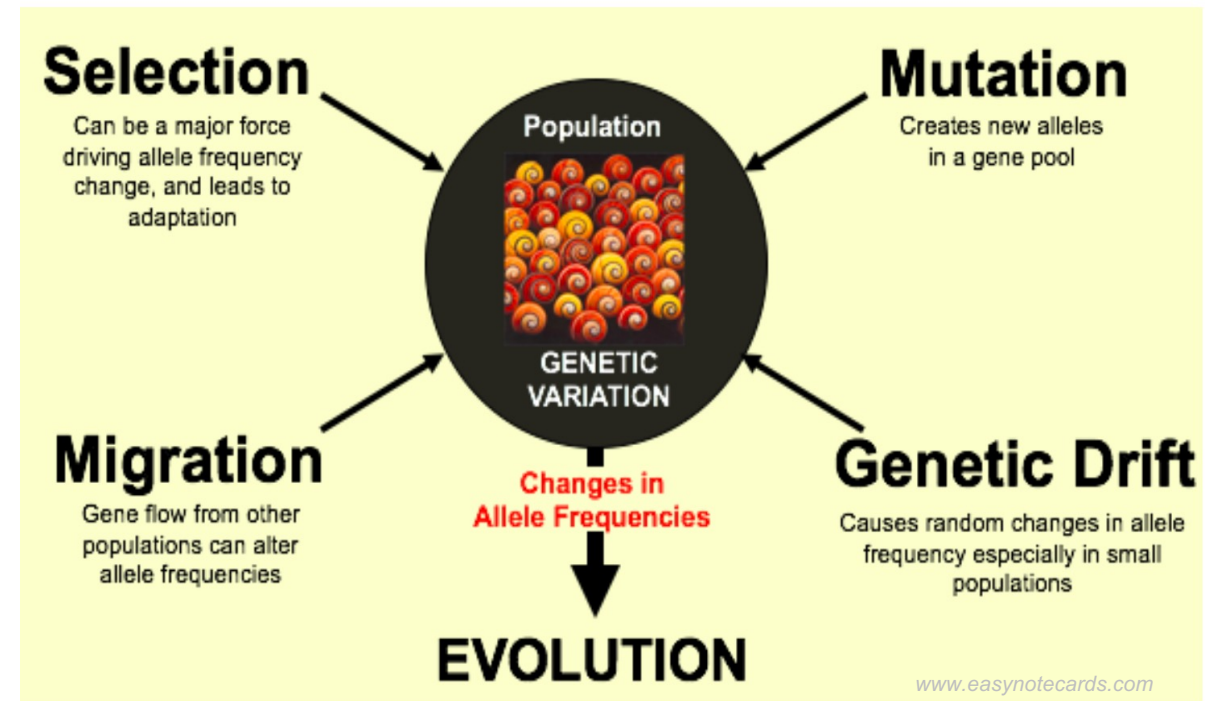
$p^2 + 2pq + q^2$

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



Population genetics

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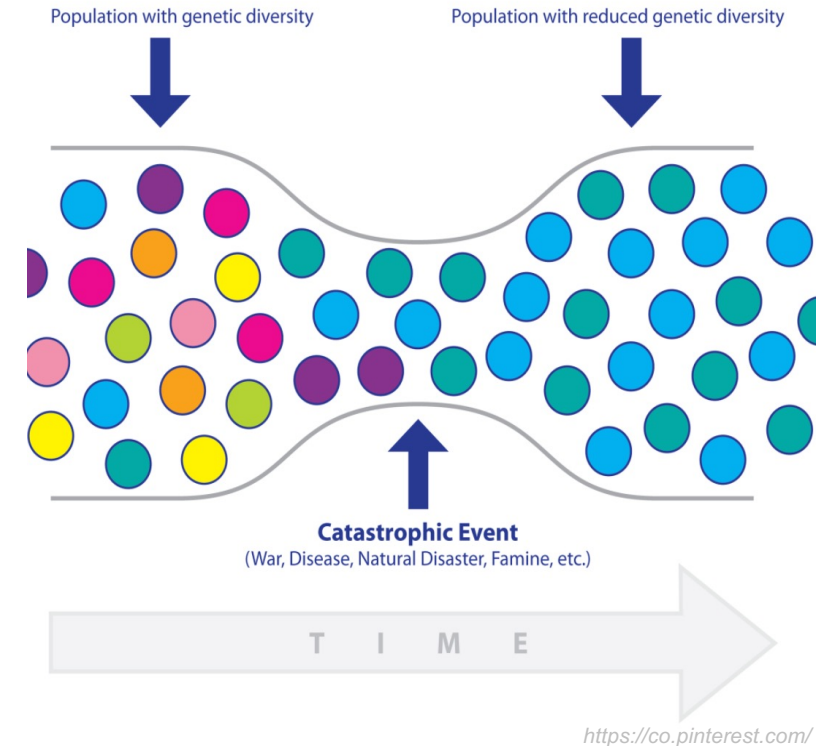


Hardy-Weinberg assumptions

- Random mating
- Population must not be divided into subpopulations
- Apply only to large populations
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If any of these assumptions is not true, the population could departure from HWE.

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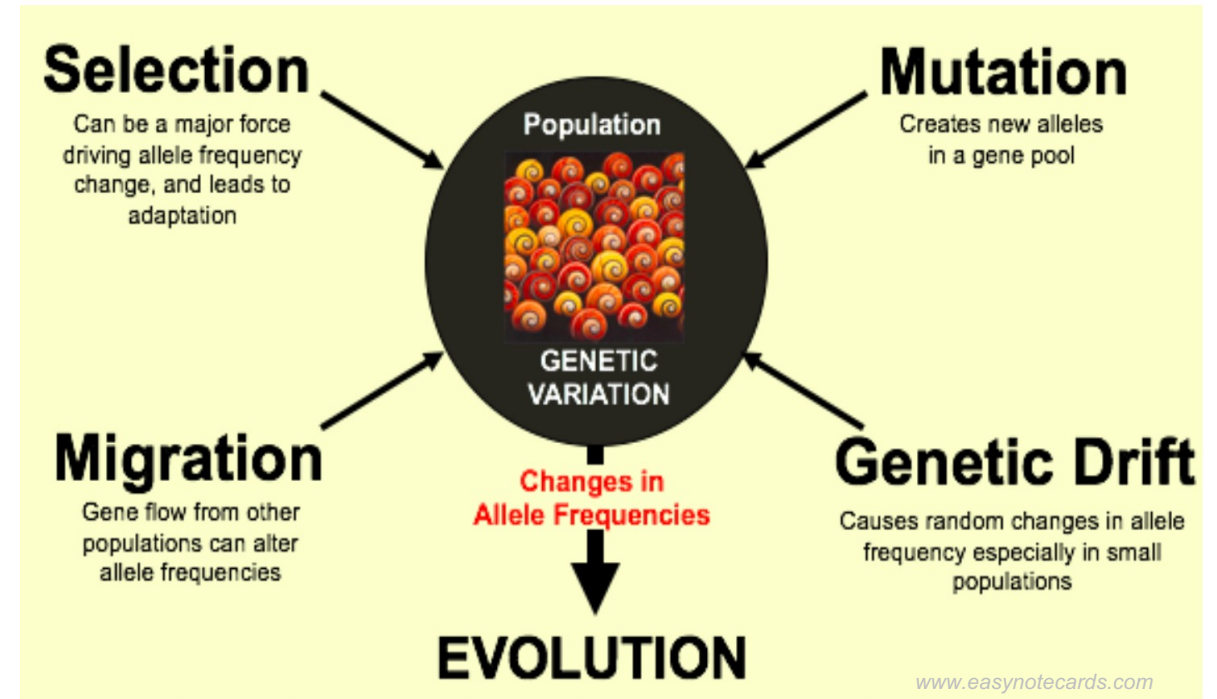
Hardy-Weinberg equilibrium

Generation	Genotype frequencies			Gene frequencies	
	<i>A/A</i>	<i>A/a</i>	<i>a/a</i>	<i>A</i>	<i>a</i>
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

Genetic variation is neither created nor destroyed

Population genetics

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Testing for HW Proportions

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

	<i>Genotypes</i>			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

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

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

$$q^2 = 12/84 = 0.143$$

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

$$q = 1-p = 0.47$$

Expected frequency

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

$$0.53^2 + 2(0.53 \cdot 0.47) + 0.47^2$$

$$0.281 + 0.498 + 0.221 = 1$$

Testing for HW Proportions

	<i>Genotypes</i>			Sum
	A/A	A/G	G/G	
Observed number	17	55	12	84
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Testing for HW Proportions

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H₀: 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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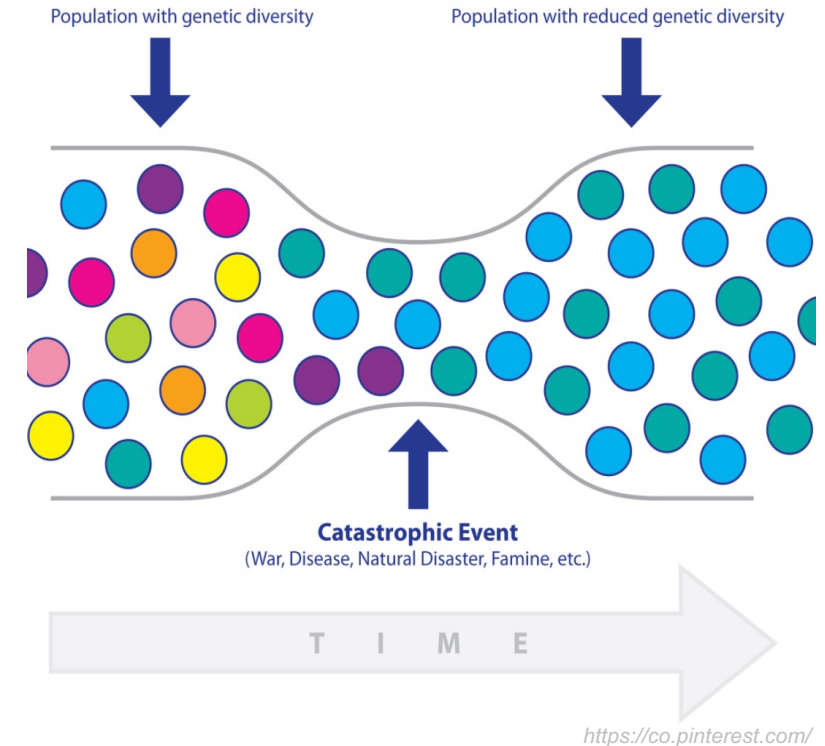
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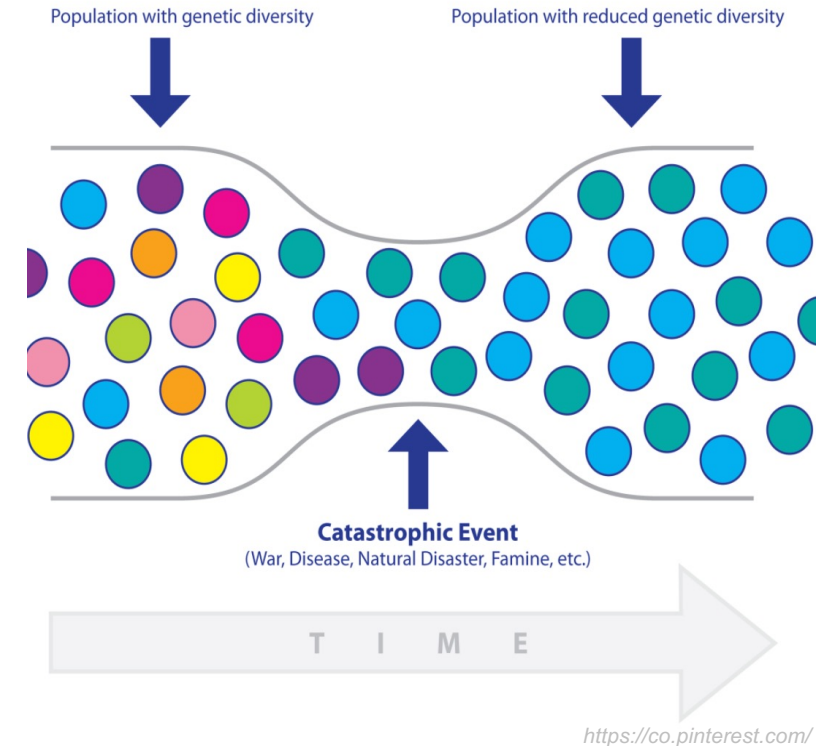


Hardy-Weinberg assumptions

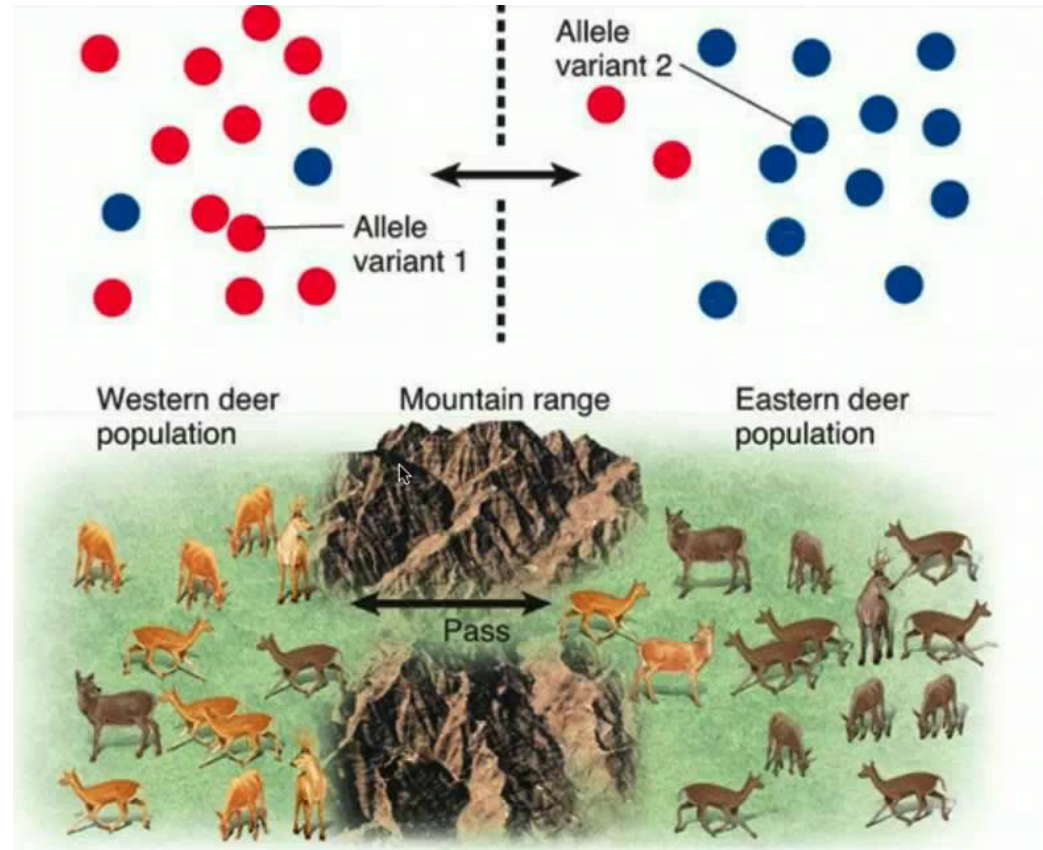
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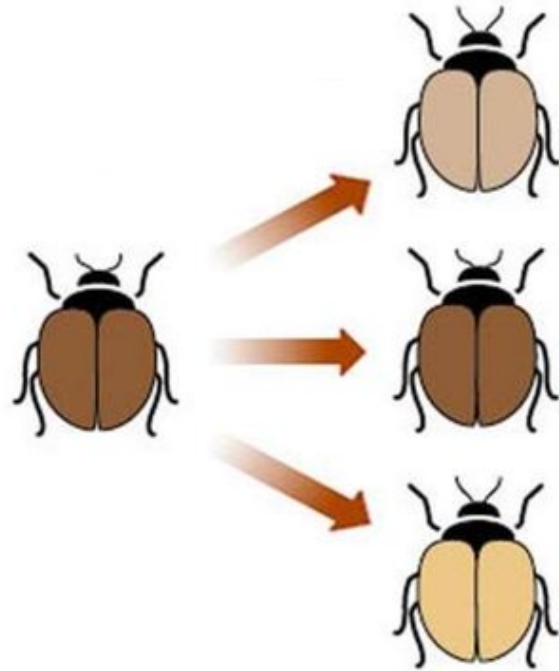


Bias in mate choice: isolation by distance

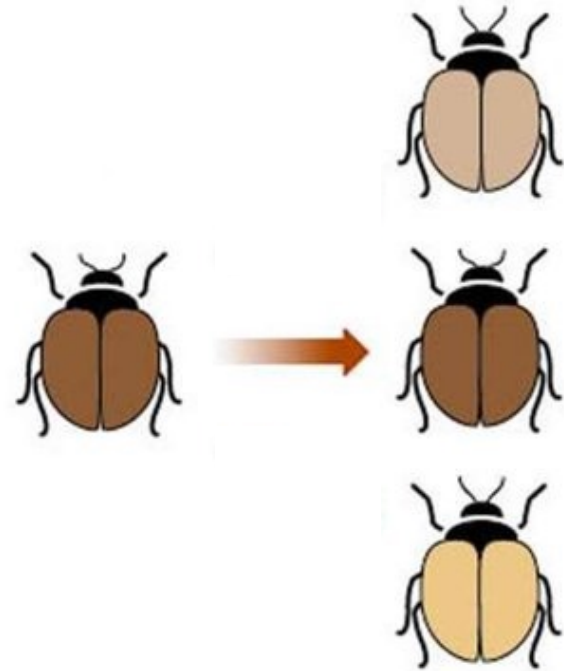


Bias in mate choice: assortative mating

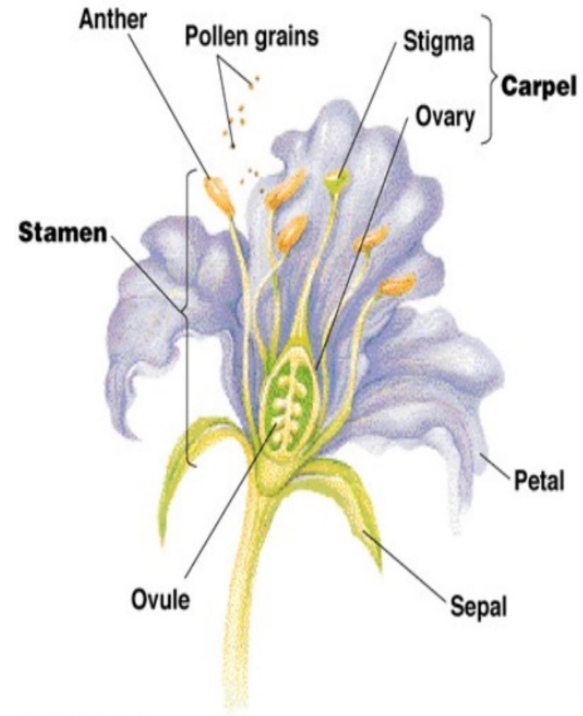
Random mating



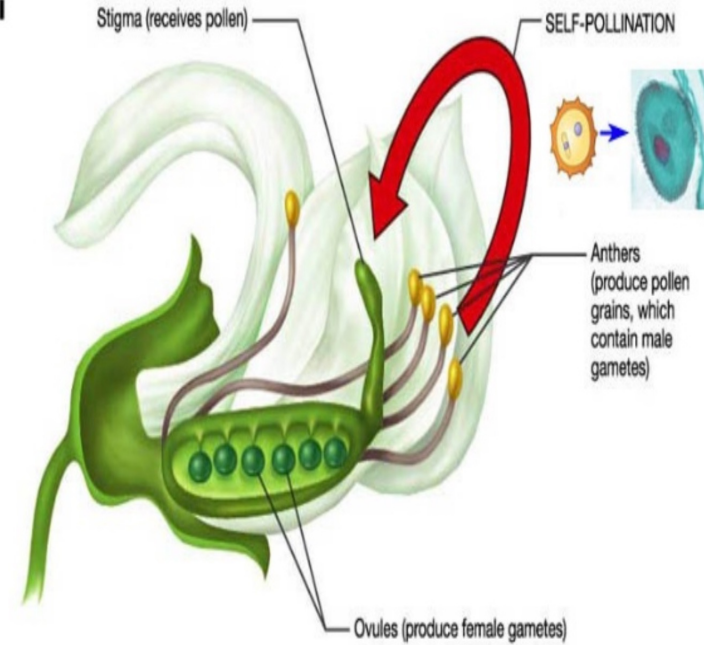
Assortative mating



Bias in mate choice: inbreeding



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Inbreeding or mating between relatives!

Review

- Gene pool
- Allele Frequencies
- Hardy-Weinberg Equilibrium
- HW assumptions

Graphical Representation of the Hardy-Weinberg Principle

