2. Эволюционные факторы, влияющие на генетическую изменчивость в популяциях

Populations and species are affected by two sets of processes:

1) **Genetic** - mutation, recombination, independent assortment, transposition, meiotic drive

2) **Ecological** - changes in population size, dispersal, mating system, environmental variation

How do these processes affect population genetic variation?

Evolutionary factors and their interactions that affect genetic variation and changes in gene frequencies

- **Mutation**
- Molecular drive
- Recombination
- **Genetic drift** (= population size)
- Gene flow via migration and gene exchange
- Nonrandom mating
- **Natural selection**

Mutation

- **It is the ultimate source of variation and may be caused by:**
  - ✓errors in DNA replication
  - ✓errors in chromosome crossing over and segregation
  - ✓damage by radiation
  - ✓transposition and nonreciprocal recombination
- **Mutation increases diversity but, because spontaneous mutations are rare, the rate of change in gene frequency is very low**
- **Consequently, mutation alone does not drive the evolution of populations and species**

Molecular Drive

The process by which mutations are able to spread through a single-copy or multi-gene family and through a population as a consequence of a variety of mechanisms of non-reciprocal DNA transfer within and between chromosomes

Recombination

- It is the meiotic process whereby a sex cell generates new chromosomal combinations, alleles and haplotypes in gametes, compared with that original parental cell.
- It normally does not create new mutations but generates new combinations of the existing diversity and usually increase diversity.
- Genetic variance can occur very quickly through recombination, given that segregating alleles exist at different loci.

Genetic Drift

- Genetic drift is a random change in gene frequencies attributable to sampling error - loss of alleles by chance events.
- The effect of drift becomes increasingly important as population size becomes smaller.
- Genetic drift is random because the frequency of any given allele can both increase and decrease depending on the allele frequency.
- In small populations genetic drift results in repeated loss of variability and eventual fixation or loss of alleles.

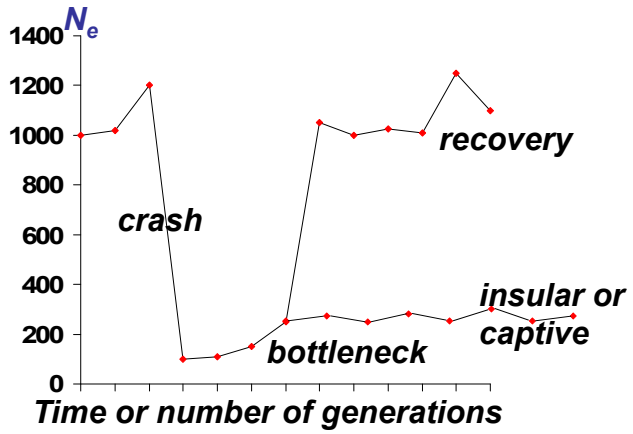
Effective population size (N_e)

- N_e is the number of parents responsible for the genetic composition of the next generation
- N_e is generally less than N because of:
  - variation of population size from generation to generation
  - unequal sex ratio
  - overlapping generations
  - geographic dispersion of populations

$$N_e = \frac{4\,N_m\,N_f}{N_m + N_f}$$

Population Bottleneck

An observable and dramatic decrease in population size (N_e)



With bottleneck and founder effects:

- Heterozygosity declines at the rate of:

$$H_1 = (1 - \frac{1}{2N_e})H_0$$

- Alleles are lost at the rate of:

$$P = p^{2Ne} + q^{2Ne}$$

Consequences of a decreasing population size

- favors genetic drift (increases random variation of allele frequencies)
- increases inbreeding
- increases homozygosity: fixation and loss of alleles
- promotes subpopulation differentiation
- a bottleneck effect develops when the population size drops sharply
- the founder effect occurs when a few individuals colonize and become established in a new environment

Gene flow or Migration

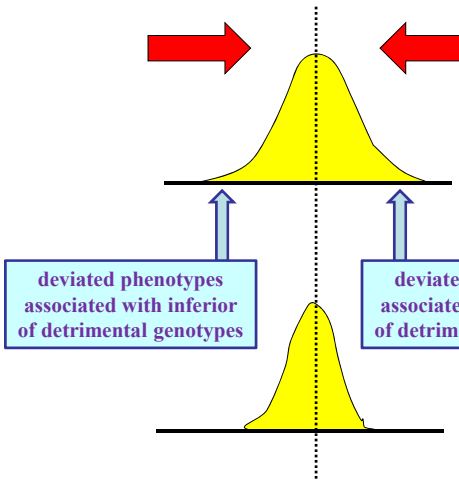
- It is the movement of individuals, gametes, pollen, seeds or any form of introduction of genes from one population to another
- Migration increases diversity and its rate can be large, causing significant changes in frequency
- The change in gene frequency is proportional to the difference in frequency between the recipient population and the average of the donor populations

Natural Selection

- changes gene and genotype frequencies as a result of differential reproductive success
- the most important evolutionary force
- occurs due to the inherited difference in ability of organisms to survive and reproduce
- acts in such a manner that, with time, inferior genotypes decrease (stabilizing, purifying or negative selection) and superior genotypes respectively increase (directional positive selection) or maintain (balancing selection) their frequency in the population

Natural selection

1. Stabilizing selection (purifying or negative selection)

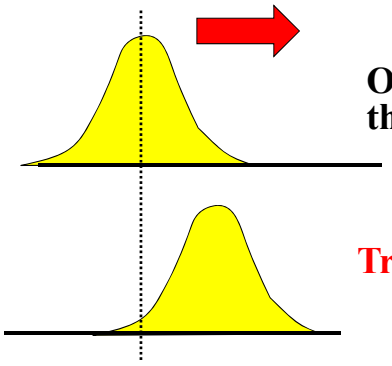


Favors mean over tails

It leads to a loss of variation or maintaining it at a particular and usually low level

Natural selection

2. Directional selection (positive selection)



One tail is favored over the other

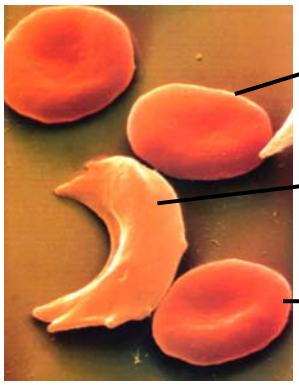
Trait is changing over time

Eventually leads to a fixation of favorable allele and loss of variation, although can increase variation at the early mutation stage

Natural selection

3. Balancing selection via heterozygote advantage - heterozygote individuals have higher fitness than either homozygote individuals. This is a common phenomenon in plant and animal breeding, but we still do not know how often it occurs in natural populations.

Example: Серповидноклеточная анемия - Red blood cells - *sickle cell disease* (an allelic substitution at the structural-gene locus for the *β* chain of hemoglobin that results in substitution of valine for the normal glutamic acid at chain position 6)



Homozygote *HbA/HbA* (normal cells): vulnerable to malaria

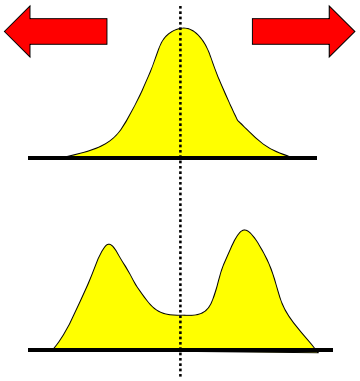
Homozygote *HbS/HbS* (sickled cells): have a severe anemia, and survivorship is low

Heterozygote *HbA/HbS*: have a mild anemia, but resistant to malaria

Variation is maintain at a particular level

Natural selection

4. Disruptive selection (diversifying selection)



Selection favors the two tails over the mean

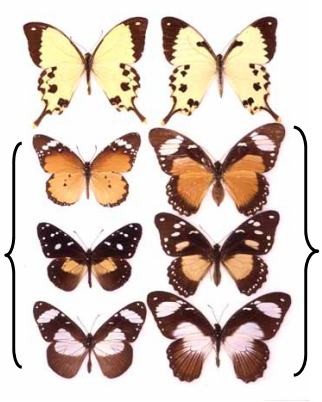
There are two forms

Variation is maintain at a particular level, but it increases differentiation within a population

Natural selection

5. Frequency-dependent selection - a mode of selection where a phenotype is only favored when it is either rare or common.

Example: Swallowtail butterfly (*Papillo dardanus*)



Males

Toxic species

Swallowtail females mimic these toxic species without being toxic themselves

It maintains variation at a particular, but very low level

Only works if the cheats are rare ☻

Factors affecting genetic variation in populations: Summary

Factor	Effect on variation
➢ Mutation	Increases
➢ Recombination	Increases (doesn't create new mutations but new combinations)
➢ Genetic drift	Reduces
➢ Gene flow via migration	Increases
➢ Inbreeding	Reduces
➢ Bottleneck and founder effect	Reduces
➢ Selection:	
- stabilising	Reduces
- directional	Increases at early but Reduces at late stages
- balancing (heterozygote superiority)	Balanced
- diversifying	Increases
- frequency-dependent	Balanced