



Fundamentals of genetic variability assessment in AnGR.

Modul no. 2: Conservation and Sustainable Use of Animal Genetic Resources

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Genetic variability

- Genetic variability is the variability of alleles and genotypes occurring in the studied population.
- Genetic polymorphism constitutes a significant part of intrapopulation genetic variability. → Genetic variability is conditioned by the existence of genetic polymorphisms.
- Genetic polymorphisms change the evolutionary potential of populations due to their ability to respond to short-term selection pressures



Factors affecting genetic variability

- Historic and current effective population size
- Bottle Neck

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- Breeding program
- Natural selection

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Factors affecting genetic variability

- Historic and current effective population size
- Bottle Neck
- Breeding program
- Natural selection
- Different intensity of mutations
- Immigration or Emigration between populations
- Interaction between the mentioned factors



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Genetic variability

Origin and sources of genetic variability

- Genetic variability – the existence of multiple alleles of many genes
- Source of genetic diversity - gene polymorphism
- Creation of genetic variability - mutation x maintenance in the population = selection
- The advantage of polymorphic genes – they give rise to a large number of variants of different genotypes



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- Genetic variability – the existence of multiple alleles of many genes
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- Creation of genetic variability - mutation x maintenance in the population = selection
- The advantage of polymorphic genes – they give rise to a large number of variants of different genotypes
- Genetic variability is primarily due to the **large amount of genetic information** encoded in DNA molecules (deoxyribonucleic acid) present in the nuclei of cells in the form of chromosomes.



Methods of assessment of genetic diversity

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➤ Pedigree analysis

- Pedigree completeness
- Probability of the identity by descent
- Probability of gene origin

➤ Molecular genetic methods

- Polymorphic information index
- Observed and expected heterozygosity
- Wright's fixation indexes



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Methods of assessment of genetic diversity

- Basic indicators of genetic diversity assessment include:
 - Inbreeding coefficient
 - Relatedness coefficient
 - Effective population size
- These indicators can be used both in pedigree and molecular genetic methods



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Pedigree analysis

➤ Pedigree completeness

- Basic parameter for the study of genetic variability.
- Level of completeness of the pedigree determines the accuracy of the estimated following parameters.
- Statement:
 - Percentage representation of known ancestors in individual generations
 - The higher the values in most generations bring higher the accuracy of the analyzed coefficients.



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Pedigree analysis

- Probability of the identity by descent
 - Individual inbreeding coefficient (F_x)
 - Relationship coefficient (R_{xy})
 - Effective population size (N_e)

Inbreeding coefficient

Inbreeding coefficient (F_x) is the quantitative measure of inbreeding and it is defined as:

Malécot G (1948), as a probability that two homologous genes in an individual are identical by descent (autozygous).

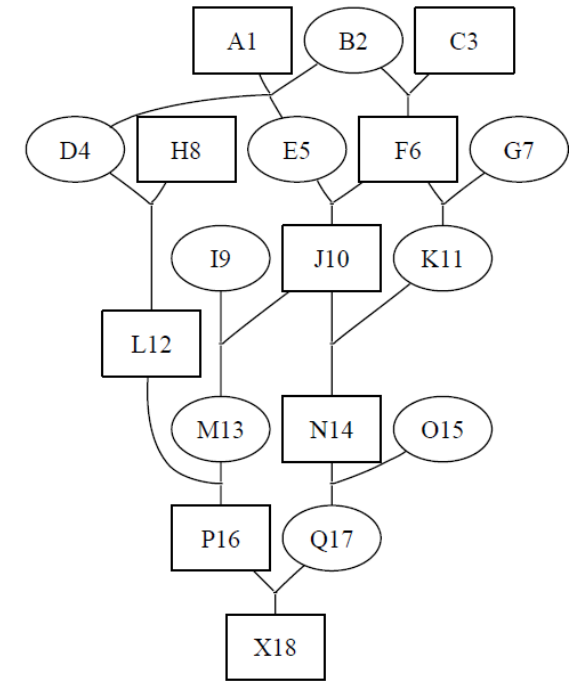
Autozygosity is the state of the genotype where two alleles are (IBD).

Identity by status

A_1A_2

Identity by descents

A_3A_3



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Relationship coefficient (R_{XY} ; Wright, 1922)

R_{XY} , after Wright, is the correlation between genetic (additive) values of two individuals (correlation between breeding values of two individuals).

$$R_{XY} = \frac{\sum [(1/2^n)(1+F_A)]}{\sqrt{(1+F_X)(1+F_Y)}}$$

$$R_{XY} = 2 f_{XY} / \boxed{[(1+F_X)(1+F_Y)]^{0.5}}$$



Effektive population size

Effective population size – is the size of ideal (Wright-Fisher) population that will result in the same amount of genetic drift as in the actual population considered.

$$N_e = \frac{1}{2\Delta F} = \frac{4 N_m + N_f}{N_m \times N_f}$$

N_m – number of mating sire

N_f – number of mating dam

ΔF – rate of inbreeding in one generations



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Pedigree analysis

➤ Probability of gene origin

➤ Effective number of founders (f_e)

- The number of equally contributing founders that would be expected to produce the same genetic diversity as in the populations under study.

➤ Effective number of ancestors (f_a)

- The minimum number of ancestors explaining the genetic diversity in population under study.

➤ Founder genome equivalent (f_g)

- The effective number of founders with the non-random loss of founder alleles that would be expected to produce the same genetic diversity as in population under study



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Genetic diversity and founders

- Maximizing the preservation of genetic variability within a closed population is possible when an identical or larger number of offspring from each basic ancestor are mated.
- A large number of offspring minimizes the random loss of genetic variability that occurred in the generation of the basic ancestors.
- The equal representation of basic ancestors in offspring generations reinforces the genetic variability found in each basic ancestor that has not been eliminated from the progeny population if additional alleles of the basic ancestors are present in multiple individuals.



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Molecular genetic methods

Polymorphic information content

- It is presented as a criterion of variability (informability) of the analyzed loci
- It is mainly used in linkage disequilibrium analysis

Observed and expected heterozygosity

- study intra-population variability and F_x status in the population
- based on the ratio of these two quantities, the loss/increase of variability in the population is evaluated

Wright's fixation indexes

F_{IS} – reduction in heterozygosity of an individual due to non-random mating within its subpopulation

F_{ST} – Fixation index, reduction in heterozygosity of a sub-population due to random genetic drift

F_{IT} – reduction in heterozygosity of an individual due to non-random mating & population subdivision relative to total population, overall inbreeding coefficient



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