9. Fundamentals of genetic variability assessment in AnGR

Hello everyone, I welcome you to the following lecture from the Conservation module on Sustainable Use of Animal genetics, the topic of which is Fundamentals of Genetic Variability Assessment in Animal genetic resources.

In general, we can say that genetic variability is the basis of all breeding programs and procedures. By the term genetic variability, we mean the variability of alleles and genotypes that manifests itself in the monitored population.

The essence of genetic variability is genetic polymorphism. Since the basis of genetic polymorphism is the variability of alleles at one locus, the genetic variability of the population is conditioned precisely by the existence of genetic polymorphisms.

Due to their variability, genetic polymorphisms change the evolutionary potential of populations because these populations can respond to short-term selection pressures.

Among the effects affecting the genetic variability, we include:

Historical and current population size. Population size significantly affects genetic variability. There is a significant loss of genetic variability in small, often closed populations.

Another effect is the Bottleneck Effect. It is because, with a substantial reduction in the number of individuals in the population in the past, there is a reduction in the size of genetic variability, which, without the migration of new genetic variability nor an increase in the number of individuals in the population, will not increase genetic variability.

Also, breeding programs, through the selection of parental pairs, limit the amount of genetic variability. During these reasons, natural selection reduces the value of genetic variability.

Furthermore, as already mentioned, mutation and migration between populations increase the amount of genetic variability.

Last but not minor, genetic variability is influenced by the interaction between all the mentioned factors.

Multiple allelic sets of alleles of many genes and genetic polymorphism ensure genetic variability in the population. Genetic variability in populations arises with the help of positive mutations maintained and strengthened in the population by means of natural or artificial selection.

Genetic variability is primarily due to a large amount of genetic information (genetic polymorphism) encoded in DNA molecules, which is present in cell nuclei in the form of chromosomes.

The value of genetic variability can be determined using pedigree or molecular genetic methods. Among the pedigree analyses, we include indicators of completeness of pedigrees, Indicators derived from a common ancestor and Indicators of the probability of origin of genes. Among the molecular genetic methods describing the level of genetic variability, we include the polymorphic information index, Expected, and observed heterozygotes and Wright's fixation indices.

However, the most basic indicators of genetic variability include the inbreeding coefficient, the relatedness coefficient, and the effective population size. These indicators can be used both in pedigree and molecular genetic methods.

One of the important pedigree parameters is the so-called completeness of pedigree. It is a basic parameter for the study of genetic variability because the level of completeness of pedigree records determines the accuracy of the estimated following parameters.

It is expressed as a percentage representation of known ancestors in individual generations. The higher the values of the percentage drowning of ancestors in most generations, the higher the accuracy of the other analysed coefficients.

Among the indicators derived from the common ancestor are the coefficient of inbreeding, coefficient of relatedness and effective population size.

The inbreeding coefficient quantifies the value of inbreeding as the probability that two alleles on a chromosome locus are identical by descent (IBD, or autozygous). Autozygosity is defined as the state of one locus where two alleles are identical by descent. That is, they come from the same ancestor.

Furthermore, we also recognize alleles that are identical according to status, that means that the genotype is, for example, homozygous dominant, but the alleles were obtained from different

individuals. Here, in the given example, one from individual 1 and the other from individual 2. In the second example, we also have a homozygote, but he obtained both alleles from one individual – individual 3, so these are autozygous alleles, or alleles identical by descent.
Another parameter is the relationship coefficient, which is the correlation between the genetic value (additive) between two individuals (correlation between the genetic foundation of two individuals). We can obtain the kinship coefficient based on the above relationship, where n represents the number of paths from individual X and Y to a common ancestor. In the connection, the inbreeding coefficient of the common ancestor of individuals X and Y is also considered.
Another coefficient that can be used to evaluate genetic variability in a population is the effective size of the population. This coefficient estimates the number of unrelated animals that in an ideal population (panmictic population) would lead to the same loss of genetic variability – that is, an increase to the same increase in the coefficient of inbreeding from generation to generation as recorded in the analysed population. For example, according to the FAO, if the effective population size is lower than 50 individuals, it is a threatened population, even if the given population may have thousands of individuals.
Indicators of the probability of origin of genes include Effective Number of Founders, Effective Number of Ancestors and Effective Number of Founder Genomes.
Under the term effective number of founders, we can imagine the number of equally contributing founders - individuals with a unknown parents they can create the same genetic variability as the monitored population.
Under the term effective number of ancestors, we can imagine the number of equally contributing ancestors (not founders) who can explain the same genetic variability as a given population. This parameter is explained by the possible recent decreasing of individuals in pedigree, or so call the bottleneck effect, and partially explains the loss of genetic diversity in the monitored population.
The indicators shown on the previous slide use the principles by which it is possible to maintain a sufficient level of genetic variability. Among these principles is maximising the conservation of genetic variability within a closed population by mating an equal number of offspring from each basic ancestor. Furthermore, many pods minimise the random loss of genetic variability. The equal representation of basic ancestors in offspring generations reinforces the genetic variability found in each basic ancestor that has yet to be eliminated from the progeny population if additional alleles of the basic ancestors are present in multiple individuals.
Among the molecular genetic methods of assessing genetic variability are the polymorphic information index, which is stated as a criterion of variability (or informability) of the analysed loci and which is mainly used in studies dealing with linkage disequilibrium. A better method is to compare expected and observed heterozygosity. This method studies intra-population variability and the state, or level, of the inbreeding coefficient in the population. Based on the ratio of these two quantities, the loss/increase of variability in the population is calculated. Another method is the so-called Wright's fixation coefficients or indices. These parameters compare the loss of genetic variability at the individual, subpopulation, and whole population levels. This includes the loss of genetic variability of an individual relative to a subpopulation, the loss of genetic diversity of an individual relative to the entire population, and the differentiation of gene similarity between subpopulations, which in the narrow sense of the word is taken as a fixation index and quantifies the degree of genetic difference between populations. If the value of Fst is lower than 0.05, the two subpopulations are genetically identical, and above the value of 0.125, the populations are genetically different.
This presentation introduced the basic principles of assessing genetic variability based on pedigree and molecular genetic data. Thank you for your attention, and I look forward to meeting you at the following lecture.