Topic 2: Genetics of reproduction traits (important genes and heritability) Lecture

The topic of today's lecture is the Genetics of Animal Reproductive Traits. This lecture is part of Module 1: Animal Genetics, which is part of the ISAGREED project. This presentation was supported by an Erasmus+ KA2 - Partnerships for Cooperation grant: "Innovating the structure and content of curricula in the management of animal genetic and food resources using digitalisation".

Reproduction is essential for all organisms because it is the key to survival and evolution. Reproductive traits are important for the survival of species not only in natural populations, but also in livestock populations, where they have a major impact on the economic efficiency of breeders and farms. Reproductive traits are economically important for sustainable food production. Reproductive traits, especially those related to fertility, litter size and preweaning viability, are important components to reduce the cost of animal meat production. Therefore, great efforts are being made to identify ways of improving these traits. This improvement in reproductive traits is related to different areas, with management, nutrition and genetics having a major impact.

Fertility is a fundamental biological and utilitarian trait of livestock. It is a prerequisite for the maintenance of the species as well as the production of animal products. Fertility is generally understood as the ability to produce reasonably numerous and constitutionally fit offspring. Fertility is subject to considerable environmental influences and a distinction must therefore be made between potential and actual fertility. Potential fertility is to be understood as the ability of the female to release eggs capable of fertilisation regardless of what happens to them next, so it can largely be understood as hereditary fertility, which is difficult to detect without examination. By the action of various environmental factors, by which we mean not only the internal environment of both sexes but also the external environment, hereditary fertility in the sense of the above definition is considerably reduced, so that by actual fertility is to be understood the number of live-born offspring. Potential fertility can therefore be considered as an expression of the genotype and actual fertility as an expression of the fertility phenotype.

The most significant indicators of female fertility include: Length of gestation, number of pups, litter percentage, interperiod length, service period, and difficulty of delivery.

Male fertility indicators may include: sperm count, incidence of sperm malformations, and libo sexualis, which is related to sex hormone levels.

The importance of different reproductive traits is not the same and also varies between species. In cattle, low female fertility has a very negative impact on production, as it causes economic losses by increasing the need for additional inseminations, higher veterinary costs, significantly longer lactations and fewer calves. Also, calving ease is closely related to reproductive success and viability of mother and offspring. In meat sheep production, litter size and days to estrus are two of the most important traits, but their inclusion in selection schemes has been limited due to low heritability. In pig breeding, as in other multipair species such as rabbits, litter size is probably the most relevant and easily measurable reproductive trait that receives the most attention. Litter size is determined by the interaction of many physiological components, such as the number of ovulated eggs, which determines the maximum number of possible offspring, and the rate of prenatal survival. Age at first foaling is further considered an important trait in evaluating the reproductive performance of mares with results across the entire equine breeding system, as it indicates the beginning of the reproductive life of individuals. Age at last foal is a reproductive trait that is considered to be a measure of the cessation of reproductive activity of individuals, thus indicating the reproductive life of the population. However, equine reproductive parameters related to fertility are complex traits influenced by a wide range of genetic, environmental and management factors, making it difficult to identify the underlying factors directly related to the animals themselves.

Factors affecting reproductive traits. The reproductive traits of livestock are influenced by internal and external factors. Internal factors are related to the genotype of the animal, while external factors are related to its environment. Ovulation rate and litter size are important reproductive traits in livestock that have high economic value and are genetically controlled. Genetic improvement can be achieved by identifying polymorphisms in genes associated with reproductive traits. Non-genetic factors can be divided into those that have measurable effects, such as parity, maternal weight, and calving period, and those that do not have measurable effects, such as infections. Another external factor affecting reproductive traits is variation in photoperiods, which control melatonin secretion.

Quantitative genetics

Reproductive traits are conditioned by polygenes and their inheritance is dealt with in quantitative genetics. Quantitative genetics studies the inheritance of complex phenotypic traits. Its essence is the study of phenotypic variation between individuals, focusing primarily on traits that take on a continuous range of values. Any phenotypic expression that has different values in different individuals and does not follow a pattern of simple Mendelian inheritance can be referred to as a quantitative trait.

Quantitative traits:

- Are conditioned by a large number of small effect genes, otherwise called polygenes,
- environmental factors have a large influence on the variability of the traits in question,
- they have smooth, uninterrupted, continuous variability that can be objectively measured,

- in a sufficient number of individuals, they tend to have a normal distribution of phenotypic classes and are characterised by a Gaussian curve,

- their variability is expressed in units of measurement and evaluated by statistical methods. Quantitative traits are measurable, we express the phenotype numerically and their variability can be characterized using statistical quantities. With statistical quantities are important:

- the mean,
- the variance or covariance,
- the mean error,
- the standard deviation,
- the coefficient of variation.

Genetic parameters of quantitative traits

To study quantitative genetic variation, statistical methods introduced by Fisher and Wright are used to analyze the variance components of phenotypic variation and describe the similarity between related individuals, often without knowledge of the genes involved.

Genetic parameters of quantitative traits can be divided into two groups:

1. Primary genetic parameters, which we estimate based on biometric analysis of the population. They represent the individual components of the variance partitioned according to the source that caused the variance. These include:

- genetic variance,

- environmental variance,

- covariance - the joint variability of two given traits.

2. Secondary genetic parameters are calculated as functions of primary genetic parameters. These include:

- Heritability (heritability),

- repeatability,

- genetic, environmental and phenotypic correlations.

Decomposition of phenotypic variance

When assessing the heritability of a quantitative trait, it is essential to determine to what extent the trait is genetically determined and to what extent it is environmentally determined. The phenotype as an external manifestation of a trait is the result of the action of complex relationships between the genotype and the environment. The phenotypic value P of an individual is the sum of its genotypic value G and its environmental value E.

P=G+E

The variance of a phenotype, which is defined as G+E, is characterized by the following relationships:

$$V_P = V_G + V_E + 2cov_{GE}$$
 or $\sigma_P^2 = \sigma_G^2 + \sigma_E^2 + 2cov_{GE}$

The value VP represents the variance of phenotypic values, VG represents the variance of genotypic values, VE represents the variance of environmental variance, and covGE represents the covariance between genotype and environment.

Heredity - Heritability

The most common summary statistic in quantitative genetics is the calculation of heritability. Heritability is a measure of the contribution of the genotype to the phenotypic realization of a trait. It determines the proportion of the heritable component in the final phenotypic expression of a trait. Heritability refers only to the population and environment in which they were measured. Thus, the same trait measured in different populations may have different heritabilities.

The mathematical expression of heritability is the coefficient of heritability, h2, which is the ratio of genetic variation expressed by the variance value to the total phenotypic variation also expressed by the variance value.

Heritability coefficient can be expressed in two ways viz:

Heritability coefficient in the broad sense:

$$h^2 = \frac{\sigma_G^2}{\sigma_P^2}$$
 or $h^2 = \frac{V_G}{V_P}$

includes all components of genotypic variance

expresses the extent to which the phenotype of an individual is determined by the genotype

Heritability coefficient in the narrow sense:

$$h^2 = rac{\sigma_A^2}{\sigma_P^2}$$
 or $h^2 = rac{V_A}{V_P}$

includes the component of genotypic variance caused by the additive effect of alleles

expresses the extent to which the phenotype of individuals is determined by two independent sets of genes that they acquired from their parents - i.e. it determines the degree of similarity between relatives

The $\Box 2A / \Box 2P$ proportion thus determines the degree of similarity between related individuals, corresponding to the general breeding value; the proportions due to dominance (D) and interactions (I) indicate only how much certain trait combinations increase or decrease the value of the additive effect. The coefficient of heritability is the most important genetic parameter for populations of quantitative traits; it expresses the extent of variability over which a population can be influenced. It is a measure of the efficiency of selection.

The values of the heritability coefficient

Heritability coefficient values vary from 0 to 1 or from 0 to 100%. What is important, however, is how the heritability coefficient value is practically understood in a particular case and what it is then used for in the selection technique. A heritability coefficient value close to 1 indicates that most of the observed variation is due to variation in the average effects of different alleles. Conversely, while low heritability means that VA is small, it tells us little about VG because the genetic effects may be largely non-additive (D and I). This means that even a trait with h2 = 0 may still have considerable genetic variation at loci, contributing to the observed trait variability. In general, the higher the value of the heritability coefficient, i.e., the more heritability contributes to the overall variability, the simpler the selection methods we can use. Conversely, the lower the value of the heritability coefficient, i.e. the more environmental factors contribute to the variability of the trait, the more complicated the selection methods we

have to choose. In practice, this means that in the case of traits with high heritability, we can expect the offspring to have roughly the same development as the parents for that trait.

The resulting value of the heritability coefficient (h2) is influenced by:

- the degree of variability of the trait under study,

- the degree of diversity of genotypes in the population of interest,
- and the degree of diversity of breeding conditions.

Repeatability

When observing quantitative traits and characteristics, it is typical that many of them can be observed and measured repeatedly in the same individual throughout his or her lifetime. Repeatability refers to the degree of similarity in the values of repeated traits or characteristics in a given population. Repeatability of production traits in organisms is divided into four groups for selection and breeding purposes:

- repeatability over time - expresses the extent to which the same trait is repeated at different ages in the life of an individual,

- partial repeatability - expresses the extent to which the same performance is repeated at different periods and over the whole period of production.

- spatial (topographic) repeatability - determines the extent to which the same trait is repeated on different parts of the animal's body,

- paratype repeatability (environmental) - expresses the extent to which the same performance is repeated in different environments.

Repeatability coefficient - rop

The measure of repeatability is expressed numerically by the repeatability coefficient - rop. The coefficient of repeatability indicates the proportion of variability caused by genetic differences out of the total phenotypic variability. It represents the maximum level of heritability, highlights the additive component of the genotype and expresses the extent to which the same performance (level of performance) is repeated. The higher the value of rop, the higher the probability of the same performance, so the trait is genetically determined to a high degree. It includes the whole component of genotypic variability, but also variance conditioned by the effect of the permanent environment and variance conditioned by the interaction between the permanent environment and the genotype:

$$r_{OP} = (\sigma_G^2 + \sigma_{Ep}^2 + \sigma_{GEp}^2)/\sigma_P^2$$

Heritability of reproductive traits

Reproductive performance in males and females is the basis of profitable livestock production systems.

Some of the variation in reproductive traits is under genetic control, which means that reproductive traits are heritable and can help to extend genetics within a population by improving the intensity of selection and shortening the generation interval. The heritability of reproductive traits is low to moderate, so an extensive data record is needed to identify specific selectable traits. Male reproductive traits - including ejaculate volume and progressive sperm motility - are moderately heritable and could be used in selection programs. In contrast, female reproductive traits are low heritability and are currently the limiting factor for genetic improvement. Litter size and parturition interval are the main constraints to increasing the intensity of selection and shortening the generation interval in a breeding programme.