

11. Genetics of quantitative traits - qualitative and quantitative traits, decomposition of phenotypic variance, heritability?

Hello everyone, I welcome you to another lecture from the Animal Genetics module, the topic of which is: Genetics of quantitative traits. In the lecture, we will introduce qualitative and quantitative traits and the decomposition of phenotypic variance.

If we are talking about the performance of an individual, we are talking about the so-called phenotype. The phenotype is a set of traits observed on individual and is a function of genotype and environment. It is therefore a set of all the traits of an individual that are of interest to us from the point of view of animal genetics.

We expertly divide these observed traits into qualitative or qualitative traits. When we talk about qualitative traits, we are talking about traits with a clear distinction between phenotypes, i.e., clearly determined categories (e.g., colour of individual, presence of horns, ...). These traits are influenced by a few genes with large effects so call major genes or oligogenes. The influence of the external environment on the given traits is negligible. Because if we consider, for example, an individual's colour, whether the individual is in a comfortable environment or a non-comfortable environment, it will always have the same colour. Qualitative traits show so-called alternative variability: animals are either black or white, nothing in between - for example, in the qualitative traits we can include: animal colour, genetic diseases, and more. Quantitative traits are traits with a continuous distribution of performance. Many genes of small effect or minor genes or polygenes influence quantitative traits. The influence of the external environment contributes to the individual's performance in different proportions for different traits. It means that if the animal is in a comfortable environment, it will perform differently than in an uncomfortable environment. In the quantitative traits we can include for example dairy performance, daily gain, and meat productions. Quantitative traits show normal distributions, including dairy performance, daily gain, and meat performance.

We will present the relationship between genotype and phenotype for qualitative and quantitative traits. For qualitative traits, the genotype is directly reflected in the phenotype. On the contrary, in the case of quantitative traits, the genotype with developmental factors, which may include, for example, the upbringing of the individual, the disease experienced, create so-called potential ability, which is further influenced by environmental conditions and only then manifests itself in the phenotype.

In animal genetics, variability is the most important parameter. Based on variability, we can choose and select individuals. If there was no variability in the population, it is impossible to select the best individual as the parent of the following population. In this picture, we will imagine the essence of genetic variability and the transition between quantitative and qualitative traits. Suppose we were to consider that the colour of an individual is controlled by only one gene pair, and it would be an incomplete dominance. In that case, the population will have three possible colour combinations - brown, orange and yellow, as shown in the picture on the left above. If the colour were affected by two genes at two loci, there would already be five colour combinations, as shown in the left below figure. If we were to consider genes on three loci, the population would have seven colour variants, as in the picture at the top right. But assuming many genes, we will already reach the so-called continuous variable, as shown on the lower

right. If we consider only one gene, we are talking about the so-called "Mendelian genetics"; if we think of many genes, we are already talking about the so-called "Biometric genetics". Which we subsequently describe using population statistical parameters.

Basic population parameters include arithmetic mean and variance. The arithmetic mean indicates the mean or average value of a given population. For example, the average milk productivity value of Holstein cattle in the Czech Republic in 2022 was 10,440 kg. The formula for obtaining the arithmetic mean is shown in the upper left image. Other, and from the point of view of animal genetics, more important parameters are the variance and standard deviation, which are used to evaluate the variability of the population. The standard deviation represents the average deviation in the performance of all individuals in the population from the mean of the given population. For example, the standard deviation for milk productivity of Holstein cattle in the Czech Republic in 2022 is 600 kg of milk. It means that, on average, each dairy cow deviates by 600 kg of milk from the average value of the population. Some dairy cows deviate higher and some lower, but they all vary by 600 kilograms on average. We cannot calculate the standard deviation directly because we always get a value of 0 if we add all the deviations from the population mean. Therefore, we get the standard deviation as the square root of the variance. The variance represents the mean square of the deviations from the mean. And as already follows from the description, the square of the deviations is always a positive number that mostly deviates from zero.

If we use the basic definition of the phenotype. The phenotype is always defined as a function of the genotype and the environment. We can define the phenotype as the sum of the level of the genotype, the environment and the relationship or interaction between the genotype and the environment.

If we convert this relationship into phenotypic variance, then phenotypic variance is influenced by genetic variance and environmental variance. Genetic variability can be divided into variability influenced by the additive component of the genotype, variability influenced by the dominance component and variability influenced by the component of gene interactions or epistasis. Environmental variability can be divided into variability influenced by the permanent or temporary environment.

This slide will explain the terms additivity, dominance and interaction. The concept of additivity can be explained as follows. Each gene has some effect. It is generally assumed that the dominant allele shows a higher performance value (e.g. 5 kg on average) than the recessive allele (e.g. 2 kg on average). The genetic value of the given individual for which we are considering the given genotype, affected only by the additivity effect, is 38 Kg. We obtained this value by summing the individual effects of individual genes. Conversely, dominance represents the relationship of two genes at one locus. For example: If it exists, let's call it over-dominance. That is, if the alleles at one locus are heterozygous, there is an increase in productivity by, for example, 10 kg. The genetic value of the given genotype affected only by the dominance effect (D) is, therefore, 20 kg since it contains only two gene pairs in the heterozygous state. And interaction, or epistasis, represents the relationship between two genes at different loci. Suppose there is a relationship between the dominant allele A and the dominant allele B, and this relationship increases performance by 10 kg. Therefore, in the genotype we are considering, the interaction effect will increase productivity by 20 kg because the genotype contains one dominant allele A and two dominant alleles B. By summing the effects of

additivity, and dominance of the interaction, we get the total genetic value, which is 78 kg for the genotype we are considering.

We can divide the effect of the environment into systematic or permanent effects and non-systematic or temporary effects. Systematic effects affect the whole group of animals in the same direction and magnitude, which allows them to be corrected. And that by both the calculation procedure and the standardization of the environment. These effects include, for example, age, litter frequency, economy, year, season, and others. On the contrary, non-systematic effects can affect only one individual in an unknown size and direction. Because we do not know the size or direction of the effect of genes, it is impossible to eliminate these effects. It includes, for example, the so-called residual error, which introduces inaccuracies into most genetic estimates.

In this lecture, we introduced the fundamental difference between qualitative and quantitative traits and restructured the phenotype and phenotypic variability concept. Thank you for your attention, and I look forward to meeting you at the following lecture.