10. Genetic variation in populations (allele and genotype frequencies), HWE

Hello everyone, I welcome you to another lecture from the Animal Genetics module, the topic of which is Genetic variability in populations and Hardy-Weinberg equilibrium.
In the lecture, we will introduce the background of genetic variability and the already mentioned Hardy-Weinberg equilibrium. We understand genetic variability as the representation of individual genotypes and genes (alleles) in the population and their changes in the sequence of generations.
The basis of genetic variability is the representation of individual genes at the individual level. We can imagine genetic variability at the individual level using the following example. Consider that individual phenotypes in a population are based on a gene series. It means that a more significant number of alleles, in our case four, will occur in the population for the given trait. Individual alleles are located on one locus and show complete dominance among themselves. In our case, the wild allele is completely dominant over all other alleles, and the allele for albinotic colouring is recessive for all other alleles. Since the genotype for a given trait consists of only two alleles, an individual can exhibit only one of four phenotypic colourss. From the point of view of genetics, we are mainly interested in the genetic variability within the population, which is based precisely on the individual's genetic variability.
Genetic variability at the population level monitors the population's representation of individual genotypes and genes and its changes between subsequent generations. For example, let's assume a population of 10 individuals with the indicated phenotypes: 5 wild colourd, three chinchillas and two Himalayan. Since each individual carries two alleles for a given colour, there are 20 alleles in a given population (generation). We will further predict the following representation of alleles. It means that in a given generation (population) there are seven alleles for wild colour, five alleles for chinchilla colour, four alleles for Himalayan colour and four alleles for albino colour. We can find the proportion of single alleles shown here based on the individual ratios. Furthermore, we were to consider that we are only interested in the frequency of the allele for wild colouring and the alleles of the others. In that case, that means the sum of all other alleles, we get an allele frequency of 0.35 for the allele for wild colouring and 0.65 for the other forms of colours.
Populations can be divided into populations with multiallelic polymorphic loci, meaning there are different genotypes in the population, and more forms of alleles of one gene are represented. Furthermore, the population with monomorous loci means that there is only one type of allele in the population, and the population with polymorous loci are byallelic, which means that only two forms of alleles of one gene occur in the population.
If we consider a population with biallelic loci and incomplete dominance, three forms of phenotypes based on three different of genotypes will occur in the population. Dominant homozygotes, heterozygotes and recessive homozygotes. We denote the absolute number of dominant homozygotes as capital D, the absolute number of heterozygotes as capital H and the absolute number of recessive homozygotes as capital R. The total number of individuals in the population is denoted as capital N. The frequencies of individual genotypes are obtained as the ratio of individual of absolute numbers of genotypes to the total number of individuals in the population or generation. We mark the relative number of dominant homozygotes with a small "d", the relative number of homozygotes with a small "h" and the relative number of recessive homozygotes with a small "r".
Based on the relative frequencies of every genotypes, we are also able to determine the relative number of alleles in the population. When we denote the relative number of dominant alleles

as a small "p" and get it as the sum of the relative frequencies of dominant homozygotes and half the relative frequency of heterozygotes. Since a dominant homozygote carries both dominant alleles and a heterozygote only one dominant allele, this means half of its genotype, therefore only half the frequency of heterozygotes. We mark the relative frequency of the recessive allele in the population with a small "q". The equivalence relation can be used to obtain the relative frequency of the recessive allele, where it is the sum of the relative frequency of recessive homozygotes and half the relative frequency of heterozygotes.

If we know the frequencies of individual alleles participating in meting process, we can estimate the relative genotypic representation of the next population. A homozygous dominance in the next generation will result from the combination of the dominant allele from the sire and the dominant allele from the dam, that is, the frequency of the dominant allele in the subpopulation of male and the frequency of the dominant allele in the subpopulation of females. Since we expect that the frequencies of dominant alleles in the subpopulation of male and female are the same, and mathematically, these frequencies are multiplied, it follows that the frequency of dominant homozygotes in the next generation, we get the as square of frequency of dominant alleles. We proceed similarly with the frequency of recessive homozygotes in the next generation, when we obtain as square of the frequency of recessive alleles. The genotype of the heterozygote in the next generation can be obtained in two ways. The first way is when the dominant allele is passed on by the sire and the recessive allele by the dam, or vice versa; the individual gets the recessive allele from the sire, and the dominant allele gets it from the dam. Therefore the expected frequency of heterozygotes in the next generation can be twice the frequency of dominant times recessive alleles in the parent population.

Generally, the markings, small “d”, small “h”, and small “r”, are given as the frequency of genotypes in the base populations and p2, 2pq and q2 as the frequency of genotypes in the next population.

Another concept that we will deal with in the lecture is the Hardy-Weinberg equilibrium. The principle of Hardy-Weinberg equilibrium is that the frequency of alleles and genotypes in a population remains constant, i.e. unchanged. If we talk about the population as being in the Hardy-Weinberg equilibrium, the frequency of genotypes and genes remains constant from generation to generation. It is true if there are no interfering howls.

Disturbing influences mentioned include Selection, Mutation, Migration, Non-Random Maiting and Genetic Drift. It is important to realize that outside of laboratory conditions, at least one or more of these "interfering" influences are always present.

Hardy-Weinberg equilibrium is impossible. Genetic equilibrium is an ideal state that provides a basis for measuring genetic change in a population.

Examples of how the mentioned phenomena disrupt the Hardy-Weinberg equilibrium are given in the following pictures. If we select only a specific genotype from the base population as the parent of the next population, we will change the genetic background in the next population.

Similarly, the mutation also changes the genetic beckground of the population because if one allele for wild colour is mutated to an allele for chinchilla colour, or a mutation occurs from an allele for colour of a Himalayan colour to an allele for chinchilla colour, again, there will be a change in the frequencies of genotypes in the population.

During the immigration of genotypes from foreign populations or the emigration of genotypes to other populations, there must again be a change in the ratio of genotypes in the population of individuals.

We can take non-random mating as equivalent to selection and, therefore, also change the representation of genotypes in the next population.

Another phenomenon that disrupts the Hardy-Weinberg equilibrium is genetic drift. It is generally assumed that if a population is in Hardy-Weinberg equilibrium, it is in infinite size. However, this fact does not apply to the general population. If we have two populations with the same frequency of individual genotypes, the given populations differ in absolute size. Thus, even with the exclusion of other disturbing phenomena in the subsequent generation, other characteristics of the genotypes can be expected. Because in smaller populations the rule that each individual has the same probability of becoming the parent of individuals in the subsequent population is violated.

Let's again consider a population with biallelic loci and incomplete dominance and consider that the population is in Hardy-Weinberg equilibrium so that no disturbing phenomena such as selection, mutation, migration, genetic drift or non-random mating will be present. The number of alleles and individual genotypes will be constant, meaning that in any subsequent generation, the relative frequency of individual genotypes will be the same as the relative frequency in the starting population.

It follows from the nature of the Hardy-Weinberg equilibrium that the highest frequency of heterozygotes in the population, when the Hardy-Wienberg equilibrium is maintained, is equal to 50%. Therefore, if a population's relative frequency of heterozygotes is higher than 50%, the given population cannot be in Hardy-Weinberg equilibrium.

In this lecture, we introduced the basic of genetic variability and the principles of Hardy-Weinberg equilibrium. Thank you for your attention, and I look forward to meeting you at the following lecture.