Topic 3: Coat color genetics in farm animals

Practical examples

Following the theoretical lecture on coat color genetics in farm animals, we will now show several examples. The presentation is part of Module 2: Animal genetics, that is a part of the ISAGREED project. This presentation was supported by Erasmus+ KA2 Cooperation Partnerships Grant "Innovation of the content and structure of study programmes in the field of management of animal genetic and food resources using digitalization".

I chose a horse as a model mammal species. In horses, the basic coat color is determined by the genotype at the two loci listed here. This is the EXTENSION locus, where the gene for the melanocortin receptor (MC1R), is located, and the AGOUTI locus, where the gene for the agouti signaling protein (ASIP), is located. The resulting coat color is then determined by the interaction between these loci.

The recessive epistasis of the EXTENSION locus to the AGOUTI locus applies here. This means that in the case of a homozygous recessive genotype at the EXTENSION locus, the alleles of the hypostatic AGOUTI locus will not express in the phenotype; in other words, "it doesn't matter at all" what genotype the individual has at the AGOUTI locus. In an individual with a recessive genotype at the EXTENSION locus, only a pigment known as pheomelanin is produced and the resulting coat color is chestnut, which possibly have fur on the body in different shades of red and a mane and tail in the same color, possibly even lighter or darker, never but not black.

In the case of the presence of at least one completely dominant allele at the EXTENSION locus, the resulting coat color is determined by the genotype at the AGOUTI locus. A completely dominant allele of this locus restricts the distribution of the black pigment eumelanin only to the distal parts of the limbs, mane and tail, and the resulting coat color is bay. In the case of a recessive genotype at the AGOUTI locus, eumelanin is deposited evenly throughout the body, resulting in a black coat color.

You can see the possible genotypes and connected in this table, if a dot is used in the notation, it means that any allele can be at this position. In the last column, the basic colors are sorted from the most recessive chestnut to the dominant bay.

Now we will show a practical example.

Make a Punnett square for mating of two bay parents, both heterozygous at AGOUTI and EXTENSION loci. Calculate phenotypic segregation ratio in their offsping.

First, we write down the genotypes of both parents - bays. We already know that a bay horse must have at least one dominant allele from each locus in its genotype. In the assignment, it is stated that both parents are heterozygous individuals, so we write the genotype as follows.

To determine the possible offspring, it is first necessary to deduce what all possible gametic combinations the parents will form. As these are heterozygotes at the two loci considered, this number will be 2 squared, i.e. 4 different types of gametes. Since the EXTENSION and AGOUTI loci are located on different chromosomes, there is no genetic linkage between them and all the mentioned combinations will arise with equal probability.

Now we write the gametes of both parents in the Punnett square and we derive the corresponding phenotypes according to the genotype that results from their fusion. To simplify the work, it is better to write down the gametes of both parents in the same order - i.e. in the first row and in the first column.

From the completed square, we count the number of times individual phenotypes occur here and write them into the segregation ratio. We see here 9 bays, 3 blacks, and 4 chestnuts; the phenotypic segregation ratio is therefore 9 : 3 : 4, which is the segregation ratio corresponding to recessive epistasis between two loci, if in each of them, complete dominance between the alleles applies, which is exactly the case.

If you find this procedure time-consuming, it is possible to solve the case in a slightly different way, which would be applicable, especially if we are not interested in the exact segregation ratio, but, for example, only in what is the probability of a particular color in offspring.

For example, if we were wondering what is the probability that these two parents will produce a chestnut foal? We can find the answer either in this square, when we see that it will be 4 cases out of 16 possible, i.e. 4/16, which is ¼, i.e. 25%.

However, we can achieve the same result without having to fill in the entire square.

It is enough to write down a common genotype for chestnut, in this case it is the recessive genotype at the EXTENSION locus and since we know that this genotype is epistatic over the alleles of the AGOUTI locus, we just need to solve the result of these monohybrid crossing, which will result in a recessive combination with a probability of 25 %.

We would solve the other two colors in a similar way, but here we would have to consider the genotypes at both loci.

If we were interested in the probability of a black foal, we would write down its common genotype E. aa, we calculate the probability of the desired genotype occurring for each locus separately and then multiply the two probabilities because we need both of these phenomena to occur simultaneously.

It will be then ¾ for E locus multiply ¼ for A locus. Therefore, the probability of a black foal being born is 18.75%.

Well, the last possible case is the bay with common genotype as follows and then the probability will be ¾ multiply 3/4, so overal 56.25 %.

To check our results, we can add all three values together 25 % + 18,75 % + 56,25 % = 100 % and we are therefore sure that we have calculated correctly and at the same time that we included all possible phenotypes that can appear in foals.

GREY is another common coat color in horses. This coat color occurs in many breeds, in some such as Old Kladruber Horse or Lipizzaner, it is considered a breed characteristic.

A grey horse has a coat color characterized by progressive depigmentation of the colored hairs of the coat. Most grey horses have black skin and dark eyes. Grey horses may be born any base color, depending on other color genes present. White hairs begin to appear at or shortly after birth and become progressively more prevalent as the horse ages as white hairs become intermingled with hairs of other colors. Greying can occur at different rates—very quickly on one horse and very slowly on another. As adults, most grey horses eventually become completely white, though some retain intermixed light and dark hairs.

At the molecular level, the GREY gene was described relatively late (as late as 2008) compared to other color genes. It is the syntaxin 17 (STX17) gene.

Within the mentioned locus, the complete dominance of the allele for greying applies, i.e. the presence of one is enough to start greying, BUT it has been shown that greying in heterozygous individuals is slower and sometimes incomplete compared to dominant homozygotes. The allele for greying is epistatic to alleles of other pigmentation genes, i.e. it gradually overlaps their expression. Therefore, the carrier of this allele will turn grey regardless of the base color. In other words, we can say that the G locus is in dominant epistasis to other coat color loci.

This slide shows the possible result (phenotypic segregation ratio) of the mating of two greys, both heterozygous at the G locus and at the same time heterozygous at the AGOUTI and EXTENSION loci. Due to the fact that, from a genetic point of view, these are so-called trihybrids, each of the parents will create 8 different types of gametes. Since each of the considered loci is located on a different chromosome, all the mentioned combinations will arise with equal probability.

You can see that in this case the Punnett square is already quite large and it is time-consuming to complete it all. However, we can clearly see that with the greatest probability (75%), a grey foal will be born, but with a probability of 25%, a non-greying foal can appear. Well, due to the heterozygosity of the parents, this foal can theoretically have any of the basic colors.

However, if we would like to find out the probability of grey foal, it is enough to consider G locus. Since both parents are heterozygotes at this locus, the genotype segregation ratio in the offspring will be 1: 2 : 1, the phenotype segregation ratio is then 3 : 1. Given that the mare is uniparous, it is probably more appropriate to express the possible result as a probability, i.e. that we have a 75% probability that greying will occur in foals.

Example 3: What genotype at the G locus must a stallion have to obtain as many greys as possible after mating to Old Kladruber black mares?

Due to the dominant character of grey phenotype, every grey horse must have at least one grey parent. That is, we exclude a non-grey stallion (recessive homozygote). Only 50 % of the foals will turn grey if a heterozygote stallion is used. If a stallion is homozygous dominant at the G locus, he will pass the allele for greying to all his offspring and all his foals will turn grey, regardless of the color of the mares. The third option is, therefore, the best for our purpose.

At this moment I would like to thank you for your attention. If you have any questions, you can use the email listed here.