

6. The genetic significance of mitosis and meiosis

Hello. In this lecture, we will focus on the genetic significance of mitosis and meiosis. The lecture is part of Module 1, Animal Genetics. The creation of this presentation was supported by the ERASMUS+ KA2 grant within the project ISAGREED, Innovations in the Content and Structure of Study Programs in the Field of Animal Genetic and Food Resources Management Using Digitalization.

Gregor Mendel was the first to recognize that processes related to his elements during gamete formation are closely related to heredity. He knew nothing about chromosomes in his time; their existence was described only towards the end of his life by Flemming. It was not until after 1900 that the connection between Mendel's discrete units - elements and their location on chromosomes during cell division - was demonstrated. The chromosomal theory of heredity was formulated in the 20s and 30s of the 20th century. The linking of Mendel's discrete units - elements with their location on chromosomes became an integral part of scientists' understanding of heredity at the beginning of the 20th century.

To better understand, we will show how chromosomes behave and what happens to them during the cell cycle. Diploid organisms always have two copies of the same chromosome - one inherited from the mother and the other from the father. They form a homologous pair of chromosomes.

A cell that grows normally, undergoes metabolism, and has one DNA molecule in each chromosome is formed by a single chromatid. In the image on the left, we have a gene located on the homologous pair of chromosomes, in which it is heterozygous, Aa. Once the cell decides to divide, it enters the S phase (synthetic phase), where, from a genetic point of view, DNA replication and chromosome duplication take place. However, the number of chromosomes does not change, but within each chromosome, two chromatids appear, which are connected and form one chromosome. These chromatids are identical, carry the same genetic information, and are called sister chromatids. We can see them in the image on the right. Such two-chromatid chromosomes enter cell division, whether mitosis or meiosis, through the G2 phase. This is essential for understanding the connection between the inheritance of genes located on chromosomes and the behavior of chromosomes during subsequent divisions.

This schematic representation simplifies the process. In the G0 and G1 phase, the chromosomes are one-chromatid. Each chromosome is formed by one DNA. The total DNA content is $2c$ (c as content) and the number of chromosomes is $2n$ (diploid state). The DNA content is the same as the number of chromatids. In the S phase and G2 phase, after DNA replication, the number of chromosomes remains the same, $2n$, but the amount of DNA and therefore the number of chromatids is $4c$. These two-chromatid chromosomes subsequently enter cell division - mitosis or meiosis.

What is the genetic perspective on chromosome behavior during mitosis? We will observe two pairs of chromosomes when an individual is a dihybrid Aa Bb. Each gene is on a different chromosome pair - gene A is on the green chromosomes, gene B is on the red chromosomes. In the initial cell in the G1 phase, we see a state of $2n$ and $2c$ (in our case, a total of 4 chromosomes and 4 chromatids) because the chromosomes are one-chromatid. After chromosome duplication, DNA replication occurs, resulting in two-chromatid chromosomes and a state of

$2n$ and $4c$ (we have 4 chromosomes, but 8 chromatids). This cell then enters mitotic division. Mitosis consists of 4 phases. In prophase, chromosomes shorten because they condense and thicken to form a compact mass visible in the optical microscope. In metaphase, chromosomes align in the equatorial plane and attach to microtubules of the dividing spindle from both sides of the cell's poles. We still have a state of $4c$, $2n$. Because the microtubules of the dividing spindle are attached to the centromere of each specific chromosome from both poles, chromatid separation or segregation of the original chromosome occurs in anaphase, resulting in the formation of two chromosomes with only one chromatid from one chromosome. Thus, the situation is $2c$ and $2n$ to one pole of the cell and also $2c$ and $2n$ to the other pole. After telophase and cytokinesis, we can see two new cells that are genetically identical to each other and to the original mother cell. By evenly segregating chromatids, the genetic information is accurately reproduced from one generation of cells to the next. This is the main significance of mitosis. This is how our somatic cells divide.

Meiosis consists of two separate divisions. Before the first meiosis, DNA replication occurs in the S phase. Thus, the cell is again transformed from a state of $2n$ and $2c$ to a cell with $2n$ and $4c$. And this cell enters meiosis I. Meiosis I is genetically the most important because it involves the main processes related to Mendelian principles of inheritance. In prophase I, chromosome condensation occurs as in mitosis. The main difference between mitosis and meiosis I is that pairs of homologous chromosomes are close to each other and form bivalents (2 chromosomes together) or tetrads (4 chromatids together). And because they are close to each other, crossing over can occur. This process will be presented in the lecture on gene linkage. Importantly, they are close to each other, so in metaphase, I put them in the equatorial plane of the cell. When we observe two pairs of homologous chromosomes, then there are two possible ways of their arrangement, with a probability of 50 to 50%. In either the right or left cell, microtubules of the dividing spindle connect ONLY to each chromosome from one side. This is another fundamental difference between mitosis and meiosis. In our case, in metaphase I, two combinations of chromosome pair arrangements can occur. Either one or the other. This state explains Mendel's principle of independent assortment, which Mendel observed in the F₂ generation in a dihybrid cross. Further, either the right or left cell then enters the next phase of meiosis I, anaphase I. Here, segregation occurs, but due to the attachment of microtubules of the dividing spindle from each pole of the cell only to one chromosome from the pair, the entire chromosomes segregate. As a result, haploid cells $1n$ and $2c$ are formed at the end of meiosis I because they segregated entire two-chromatid chromosomes. This process also explains Mendel's principle of segregation, which he discovered in the analysis of a monohybrid cross. These cells are not yet mature gametes, so they must enter meiosis II.

Haploid cells, $1n$ and $2c$, formed by meiosis I, pass through prophase II and enter metaphase II, where the chromosomes again align in the equatorial plane. Microtubules of the dividing spindle extend from both sides towards each chromosome. In anaphase II, segregation of sister chromatids occurs. This process is the same as in mitosis. By segregating chromatids in anaphase II, we obtain two new cells at the end of meiosis II, which are still haploid $1n$ but each chromosome carries only 1 DNA in one chromatid, i.e., they are $1c$. This is the final product. Haploid cells, $1n$ and $1c$, when fused in fertilization, create a zygote with a $2n$ and $2c$ number of chromosomes and DNA. So what is the genetic significance of meiosis? Meiosis is a process that reduces genetic information by half, so that offspring formed through sexual reproduction have the same amount of genetic material as their parents. Additionally, due to the free combination of chromosomal pairs in metaphase I, diverse gametes are formed

in terms of gene combinations. Because these two genes are on different chromosomal pairs, they can freely combine and segregate independently during anaphase I. This creates variability in gametes, and in our case, when thousands of dihybrid Aa Bb cells enter meiosis, four types of gametes AB, Ab, aB, and ab are formed in a 1:1:1:1 ratio.

When we summarize both types of division, the genetic significance of mitosis and meiosis is clear. During mitosis, genetic information is transferred from one diploid cell to two new diploid cells, but they are genetically identical. This mainly concerns somatic cells of multicellular organisms. On the other hand, the genetic significance of meiosis lies in the fact that due to free combination and segregation, non-identical haploid cells, gametes, are formed, which have great genetic variability.

Thank you for your attention.