Glossary

codominance: in a heterozygote, complete and simultaneous expression of both alleles for the same characteristic

complete dominance: in a heterozygote the dominant allele masks the effect of the recessive allele

incomplete dominance: in a heterozygote, expression of two contrasting alleles such that the individual displays an intermediate phenotype

pleiotropy: describes when one gene controls two or more different characteristics

polygenic inheritance: describes when each gene that an individual inherits has a small additive effect on the overall phenotype

9.4 Chromosomal Basis of Inheritance

Learning objectives

By the end of this section, you will be able to:

- *Discuss the Chromosomal Theory of Inheritance*
- *Explain the effect of linkage and recombination on gamete genotypes*
- *When multiple alleles exist for a gene, know the difference between the wild type and variants*
- *Be able to define and explain all bolded terms*

Long before scientists visualized chromosomes under a microscope, the father of modern genetics, Gregor Mendel, began studying heredity in 1843. With improved microscopic techniques during the late 1800s, cell biologists could stain and visualize subcellular structures with dyes and observe their actions during cell division and meiosis. With each cell division, chromosomes replicated, condensed, and migrated to separate cellular poles. These advancements allowed scientists to connect inheritance with the physiological events of cell division.

Chromosomal Theory of Inheritance

The speculation that chromosomes might be the key to understanding heredity led several scientists to examine Mendel's publications and reevaluate his model in terms of chromosome behavior during mitosis and meiosis. In 1902, Theodor Boveri observed that sea urchin embryonic development does not occur unless chromosomes are present. That same year, Walter Sutton observed chromosome separation into daughter cells during meiosis. Together, these observations led to the **Chromosomal Theory of Inheritance**. The Chromosomal Theory of Inheritance states that genes are found at specific locations on chromosomes and that it is the chromosomes that independently assort and segregate during metaphase I and anaphase I of meiosis I (Figure 9.17).

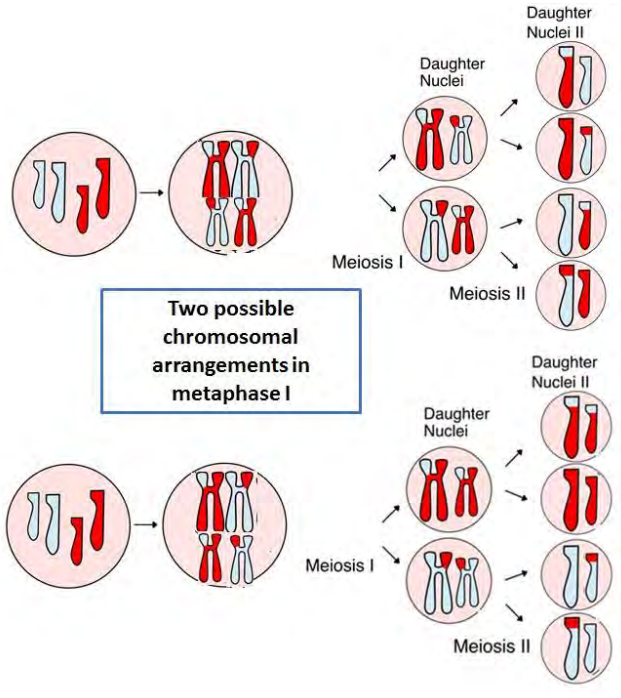

Figure 9.17: Shows how chromosomes are separated, or segregated, during meiosis. (credit: Modified by Elizabeth O'Grady original work of Rdbickel / Wikimedia commons)

The Chromosomal Theory of Inheritance was consistent with Mendel's laws. The following observations supported the connection between the two:

- During meiosis, homologous chromosome pairs migrate as discrete structures that are independent of other chromosome pairs.
- Chromosome sorting from each homologous pair into gametes appears to be random.
- Each parent synthesizes gametes that contain only half their chromosomal number.
- Even though male and female gametes, sperm and egg, differ in size and shape, they have the same number of chromosomes, suggesting equal genetic contributions from each parent.
- The chromosomes found in each gamete come together during fertilization to produce offspring with the same chromosome number as their parents.

Scientists proposed the Chromosomal Theory of Inheritance long before there was any direct evidence that chromosomes carried traits. Critics pointed out that individuals had far more independently segregating traits than they had chromosomes. It was only after several years of carrying out cross fertilizations with the fruit fly, *Drosophila melanogaster*, that Thomas Hunt Morgan provided experimental evidence to support the Chromosomal Theory of Inheritance.

Linked Genes Violate the Law of Independent Assortment

Although all of Mendel's pea characteristics behaved according to the law of independent assortment (Figure 9.3), we now know that some allele combinations are not inherited independently of each other. Genes that are located on separate non-homologous chromosomes will always sort independently. However, each chromosome contains hundreds or thousands of genes, organized linearly on chromosomes like beads on a string. The segregation of alleles into gametes can be influenced by **linkage**, in which genes that are located physically close to each other on the same chromosome are more likely to be inherited as a pair. However, because of the process of recombination, or "crossover," it is possible for two genes on the same chromosome to behave independently, or as if they are not linked. To understand this, let's consider the biological basis of gene linkage and recombination.

Homologous chromosomes possess the same genes in the same linear order. The alleles may differ on homologous chromosome pairs, but the genes to which they correspond do not. In preparation for the first division of meiosis, homologous chromosomes replicate and form tetrads. Genes on the homologous pairs align with each other. At this stage, segments of homologous chromosomes exchange linear segments of genetic material (Figure 9.18). This process, called *recombination* or crossing over, is a common genetic process. Because the genes are aligned during recombination, the gene order is not altered. Instead, the result of recombination is that maternal and paternal alleles are combined onto the same chromosome. Across a given chromosome, several recombination events may occur, causing extensive shuffling of alleles.