

The Genotype

So far, we have not yet explained how individual characters can be inherited and modified. In 1865 Mendel arrived at the conclusion that individuals reproduce by transmitting specific particles, now known as genetic material, to their own offspring. Recent progress in genetics (the discipline studying the structure and behavior of genes) and in functional genomics (the discipline studying the role of genes in organisms) has provided several clues to the molecular mechanisms and processes that support inheritance and variation. Although Darwin was probably not aware of Mendel's conclusions when he formulated the theory of evolution, genetics has become an integral part of modern evolutionary theories.

GENOTYPE The genetic material of an individual is known as the *genotype*, whereas its
PHENOTYPE manifestation as an organism is known as the *phenotype*. Natural selection operates solely on the phenotype, but the genotype is the ultimate vehicle of inheritance. The extent to which we are determined by our genotype or phenotype and the relationship between these two aspects of our individuality is a complex and much debated issue (S.J. Gould 1977; West-Eberhard 2003).

In what follows, we will introduce genes, adopting the rather conventional framework described in most textbooks. We will then point to recent results that, at the time of writing, are changing our perspectives on the role of genes in the development and evolution of organisms.

PROTEINS The conventional story involves three types of molecules and goes as follows. Cells contain a class of molecules, known as *proteins*, whose shape, concentration, and behavior determine the properties of the cell. For example hair cells and muscle cells are different because they are composed of different proteins. The definition of specific proteins depends on another
DNA molecule, known as *DNA* (deoxyribonucleic acid), which in turn relies on

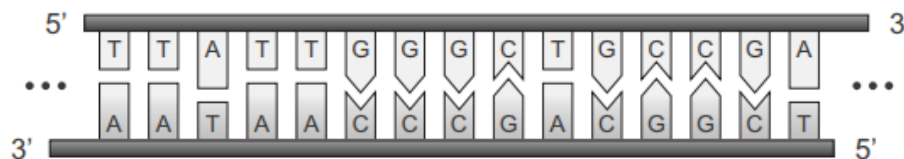


Figure 1.1 Structure of a piece of DNA molecule showing the two strands with matching nucleotides. The numbers 5 and 3 refer to the atomic structure of the molecule and affect the way in which the molecule sequence is translated into a protein. The order of translation always proceeds in the direction from 5' to 3'.

RNA proteins to become operative and on the mediation of a third type of molecule, known as *RNA* (ribonucleic acid), which is structurally similar to the DNA molecule.

NUCLEOTIDES The DNA is the genetic material that is transmitted over generations. It is often enclosed within the nucleus of the cell and all cells in the organism have the same genetic material. DNA molecules (figure 1.1) are long chains of complementary strands composed of four types of chemical units (*nucleotides* or bases): adenine (A), cytosine (C), guanine (G), and thymine (T). The two strands stick together because nucleotides can lock to each other: Adenine binds to thymine and cytosine binds to guanine. This specific binding means that the two DNA strands are perfectly complementary. If we find the sequence ACA on one strand, we know that the corresponding part of the complementary strand will display the sequence TGT (although some mismatch may occur very rarely).

CHROMOSOMES The genetic material is organized in several separated DNA molecules, called *chromosomes*. Furthermore, in several organisms chromosomes occur in pairs (also known as diploid organisms in contrast to haploid organisms). The two chromosomes in a pair are approximately homologous in the sense that corresponding areas produce proteins with a similar functionality in similar cells. The number of chromosome pairs and total length of the DNA molecules vary from species to species. For example, humans have 23 pairs of chromosomes totaling several hundreds of millions of nucleotides (International Human Genome Sequencing Consortium 2001). The redundant structure of the genetic material (two chromosomes, two strands) allows replication of DNA molecules during cell replication.

MITOSIS There are two types of cell replication: mitosis and meiosis (figure 1.2). *Mitosis* occurs during growth of the organism when a cell divides by producing a copy with the same number of chromosomes (23 times 2 in humans). During mitosis, the two strands of the 46 DNA molecules are separated and each

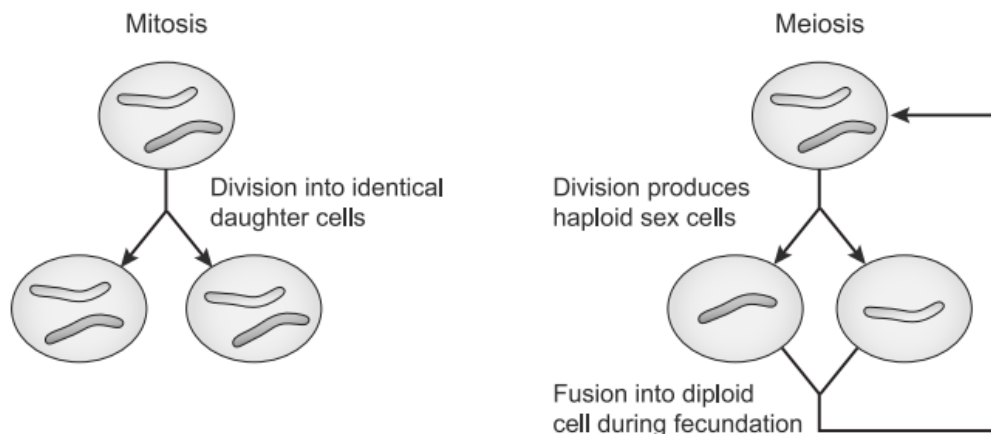


Figure 1.2 Cell replication during mitosis and meiosis. For the sake of simplicity, only a pair of homologous chromosomes are shown.

MEIOSIS

strand goes to one cell. Each strand then rebuilds the missing strand by recruiting the complementary nucleotides. The process ends with two exact copies of the double-stranded DNA molecule, one for each cell. *Meiosis* occurs during the production of sex cells (sperm and eggs). Sex cells receive only one chromosome for each pair. In diploid organisms the pairs of chromosomes are recombined during fecundation of the egg cell (containing the set of chromosomes from the mother) by the sperm cell (containing the set of chromosomes from the father). Although the chromosomes from the mother and father sex cells are homologous, their sequences may be slightly different and produce different proteins for the same functionality. This may result in the expression of features that belong either to the mother or to the father.