Molecular Genetics

Section 6.3
Transcription and Translation

Pre-View 6.3

- **Anticodon** – the sequence of three nucleotides on transfer RNA that pairs with the codons of messenger RNA
- **Codon** – a sequence of three nucleotide bases that represents the code for one amino acid
- **Messenger RNA (mRNA)** – a type of RNA that transfers the code from DNA in the nucleus to the cytoplasm
- **Peptide bond** – the bond between each amino acid in a protein
- **Ribosomal RNA (rRNA)** – a type of RNA that "reads" the codons from messenger RNA
- **Ribonucleic acid (RNA)** – a single strand of nucleotides; different types are used to translate instructions from DNA into making proteins
- **RNA polymerase** – the enzyme responsible for creating RNA from a DNA template
- **Stop codon** – a sequence of three nucleotide bases that indicates the end of protein synthesis
- **Transcription** – the process occurring in the nucleus of a cell that copies the instructions from a part of DNA onto a strand of messenger RNA
- **Transfer RNA (tRNA)** – a type of RNA that carries an amino acid and transfers it to the protein chain being assembled in the ribosome
- **Translation** – the process occurring in the cytoplasm of a cell that builds proteins

How do the genes on a chromosome determine how proteins are made? The sequence of nucleotide bases on a strand of DNA is like a language. The only letters in the language are A, T, C, and G, which stand for the four nucleotide bases. Words in this language are made up of three letters. There are 64 possible "words" that can be made from the four letters. Genes are like sentences made up of these three letter words. Each three letter word in the DNA is re-written (or is *transcribed*) into messenger RNA and represents (or is *translated* into) an amino acid. There are 20 amino acids. Some of the 64 "words" represent the same amino acid, and other "words" are like a period at the end of the sentence and indicate a "stop." Amino acids bond together to form polypeptides, and polypeptides bond together to form proteins. Each "word" represents an amino acid, and the sequence of amino acids in the "sentence" determines the type of protein that is made. To help you visualize this relationship, study figure 6-7.

The DNA Sequence of a Gene is the "Code" for a Protein

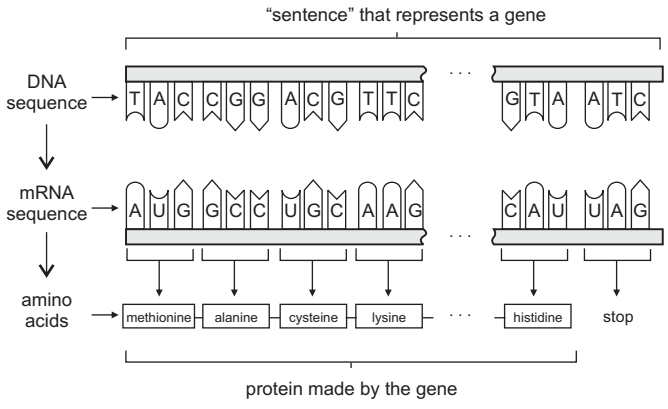


Fig. 6-7

Genetic Disorders Caused by Chromosome Mutations

Most chromosomal mutations are fatal. Some of the exceptions are nondisjuction errors in chromosome 21 and in the sex chromosomes. These types of chromosomal disorders are called trisomy disorders because there are three copies of one particular chromosome present in cells instead of the normal pair of two.

One of the most common examples of a chromosomal mutation is **Down syndrome**. The most common type of Down syndrome is trisomy 21, which means that the person affected has three copies of chromosome 21 instead of just two. Most of the time, Down syndrome can be traced to the mother where it is most often caused when homologous chromosomes fail to separate during meiosis I. Regardless of cause, the affected person has 47 chromosomes in each body cell instead of 46. A person with Down syndrome usually has mild to severe mental retardation and is more likely to have heart problems and other health issues.

Figure 7-5 shows an example of a karyotype of a female with Down syndrome.

Example 3: How can you tell from this karyotype that it is from a female and not a male?

Females have two X chromosomes as shown on the karyotype. Notice that the karyotype shown in figure 7-4 has one X and one Y chromosome. Do you see the difference?

Example 4: How does this karyotype indicate Down syndrome?

In a normal karyotype, a person has two of each chromosome. In this karyotype, the person has three copies of chromosome 21. A person with three copies of chromosome 21 has the type of Down syndrome called trisomy 21.

Karyotype of Female with Down Syndrome

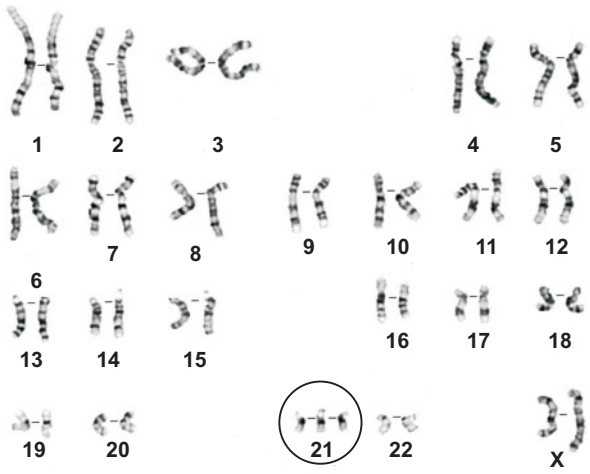


Fig. 7-5

Turner syndrome is a chromosomal mutation resulting from nondisjunction during either meiosis I or meiosis II in forming either sperm or egg cells. This syndrome affects the sex chromosomes (also known as the 23rd pair). In the case of Turner syndrome, there is a missing or incomplete X chromosome, so instead of having two X chromosomes, the female has only one (XO). Turner syndrome is also known as Monosomy X. The female possesses only 45 chromosomes. Turner syndrome affects the genes involved in growth and sexual development. Turner syndrome females have abnormal sexual characteristics that include infertility. They also typically have a short, stocky build. These females may also have heart and kidney defects, high blood pressure, and problems with swelling of the hands and feet.

Klinefelter syndrome is also a nondisjunction chromosomal mutation that occurs during either meiosis I or meiosis II in forming either sperm or egg cells. It affects the sex chromosomes and results in males that possess an additional X chromosome (XXY). Males with Klinefelter syndrome have a total of 47 chromosomes instead of the normal 46 chromosomes. Klinefelter syndrome affects the genes involved in testes development and the production of testosterone hormone. These males are usually tall and often lack typical secondary sex characteristics. Coincidentally, this disorder is usually not identified until puberty. The onset of puberty in Klinefelter males may include the development of breast tissue, very little facial or body hair, and diminished muscle definition. These males may also be infertile due to their inability to produce sperm.

Results of the Human Genome Project
The Human Genome Project revolutionized biotechnology innovations around the world, and the project was able to surpass its original goals. According to the National Institutes of Health, the Human Genome Project had the following notable accomplishments:

Notable Accomplishments of the Human Genome Project

- It sequenced 99% of the gene-containing regions of human DNA with a 99.99% accuracy.
- It mapped 3.7 million variations in the human DNA sequence that may account for difference in inherited traits or that may indicate genetic disorders.
- It enabled the discovery of over 1,800 disease genes.
- It enabled the development of over 2,000 genetic tests for human genetic conditions, which give patients the ability to know disease risks and allow healthcare professions to better diagnose diseases.
- In addition to sequencing human DNA, it also sequenced the DNA of animals important in research. These include the genomes of the mouse and rat, as well as several others.

Ethical Concerns of the Human Genome Project
The knowledge and discoveries sought by the Human Genome Project raised significant concerns from its beginning, so part of the project also funded the Ethical, Legal, and Social Implications (ELSI) program to anticipate and address these concerns. Many of these concerns centered around an individual's right to privacy and the risk of discrimination. Think about some of the questions that arose. How should genetic information of an individual be kept private? Could genetic information from an individual be obtained without consent? Could genetic information indicating a disease risk be used to deny employment or to deny health insurance coverage? What are the implications for family planning? Could knowing a fetus's genetic profile lead to an increase in the abortion rate? These are only a few of the questions raised by increasing knowledge of the human genome.

Practice 1

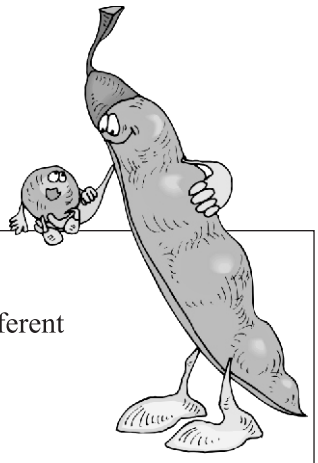
Match each vocabulary word with its definition. Write the letter that corresponds to the correct definition in each blank.

_____ 1. gel electrophoresis
_____ 2. genome
_____ 3. stem cells
_____ 4. transgenic organism
_____ 5. DNA fingerprint
_____ 6. plasmid
_____ 7. recombinant DNA
_____ 8. Human Genome Project
_____ 9. cloning

A. a unique pattern created from DNA segments that can be used to identify an individual or to identify family relationships
B. a laboratory technique that runs electrical current through a gel to separate DNA fragments by their size
C. all of an organism's hereditary information
D. DNA that is formed by joining a short piece of DNA from one organism to the DNA of another organism
E. a process of producing genetically identical copies of genes, tissues, or an entire organism
F. an international effort that successfully sequenced human DNA
G. a circular piece of DNA found in bacterial cells
H. undifferentiated cells that have the ability to become different types of specialized cells
I. an organism that contains genes from a different species

Mendelian Genetics and Inheritance

Section 8.1
Introduction to Mendelian Genetics



Pre-View 8.1

- **Alleles** – different variations of the same gene; for example, blue and brown are different alleles for eye color
- **Dominant** – a trait that is expressed over another trait
- **Gene** – a section of DNA that determines a specific trait, such as eye color
- **Genetics** – the study of heredity
- **Genotype** – the combination of alleles for a particular trait (homozygous or heterozygous)
- **Heredity** – the passing of traits from one generation to the next
- **Heterozygous (**or **hybrid)** – having two different alleles for a trait
- **Homozygous (**or **pure)** – having two of the same alleles for a trait
- **Homozygous dominant** – having two dominant alleles for a trait
- **Homozygous recessive** – having two recessive alleles for a trait
- **Law of dominance** – a natural law stating that a dominant allele will always mask a recessive allele
- **Mendel**, **Gregor** – an Austrian monk whose study of garden peas earned him the title Father of Genetics
- **Phenotype** – the physical characteristics of an organism that show how genes are expressed
- **Recessive** – a trait that can be hidden by another trait
- **Traits** – characteristics; often physical qualities such as color, height, etc.
- **True-breeding** – homozygous organism that always produces offspring with identical traits

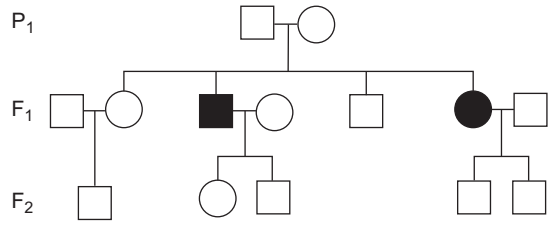
In the 1800s, an Austrian monk named **Gregor Mendel** studied garden peas. He studied a LOT of garden peas — thousands of them. (Just think about it. He was living in a monastery with no TV, no radio, no telephone, no computer, no internet, and no video games, so garden peas were pretty interesting!) He started writing his observations, and he noticed that, over time, certain patterns appeared in the plants. For many of the plants' characteristics, or **traits**, the peas would have two contrasting forms. Flowers would be purple or white, plant height would be tall or short, the seeds would be wrinkled or smooth, etc. He also noticed that some of the plants were **true-breeding** for certain traits — that is, they always produced offspring that had traits identical to the parent plants. Then he began experimenting with the plants. Through his experiments, he was able to discover some of the basic concepts of genetics and heredity. Because of his work, Mendel is known as the Father of Genetics.

Genetics is the study of heredity, and **heredity** is the passing on of traits from one generation to the next. To study genetics, Mendel started with true-breeding parent plants. We'll label them *P* for parental. The parental plants had contrasting forms of a trait. For example, one parental plant would have white flowers, and the other parental plant would have purple flowers. When he crossed these plants, the offspring (we'll call them F1 for *first filial*) were identical to each other and to one of the parent plants. In the case of one parent having white flowers and the other parent having purple flowers, the offspring all had purple flowers. From this, Mendel reached several conclusions, and these conclusions later became known as Mendel's Laws. It's pretty amazing that Mendel was able to come up with these laws a long time before people knew anything about DNA, genes, and chromosomes!

Pedigrees can be used to show different modes of inheritance: autosomal recessive, autosomal dominant, sex-linked recessive (X-linked), and sex-linked (Y-linked). Y-linked disorders are few and rare, so you will not commonly see a pedigree for a sex-linked trait that is carried on the Y chromosome.

As you may have noticed, sometimes pedigrees show carriers and sometimes they do not. When carriers are not identified, it can be a little more difficult to determine what kind of trait is being traced. To identify the type of inheritance pattern shown in a pedigree, consider the following tips and trends.

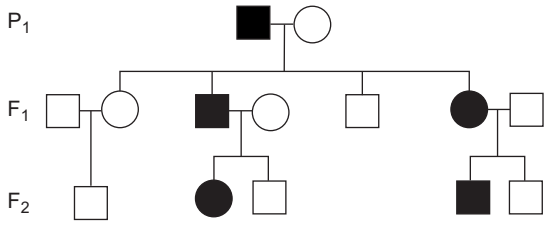
Autosomal Recessive



Autosomal recessive traits and disorders show up in either male or female offspring of unaffected parents. Although not indicated, both P_1 parents must be carriers.

The F_2 generation will often be carriers of the trait but not express it. Notice the skip of generations having the trait.

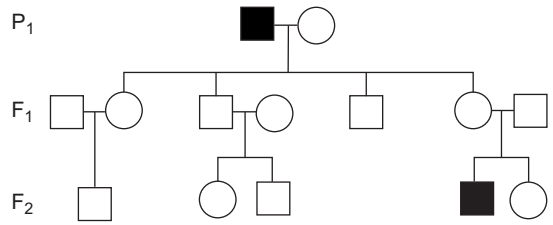
Autosomal Dominant



Autosomal dominant traits will often show up in every generation instead of skipping generations. Both male and female offspring are equally affected.

If one parent has the trait but is heterozygous for the trait, about half the offspring will have the trait. The P_1 male above is heterozygous. If one parent has the trait and is homozygous, every offspring will also have the trait (not shown).

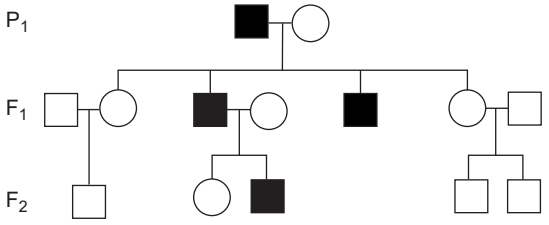
Sex-Linked Recessive (X-Linked)



Sex-linked traits carried on the X chromosome are similar to autosomal recessive traits, except they show up most often in male offspring.

Remember, females carry these traits that are passed on to their male children. These pedigrees will follow a pattern of female carriers having affected male offspring. ALL daughters of an affected male parent will be carriers.

Sex-Linked (Y-Linked)



Sex-linked traits carried on the Y-chromosome are rare but easy to see on a pedigree. Every male offspring of an affected male will have the trait in every generation. Females are unaffected because they never carry a Y chromosome.

Note: If a pedigree *does* indicate carriers, the trait MUST be either autosomal recessive or sex-linked recessive.