

HUMAN ANATOMY & PHYSIOLOGY-II- BP201T

UNIT: 5 (REPRODUCTIVE SYSTEM)

CLASS: 8

TOPIC: Protein synthesis

Proteins are composed of a long chain of amino acids linked together one after another. There are 20 different amino acids that can be used in protein synthesis—some must come from the diet (essential amino acids), and some are made by enzymes in the body. As a chain of amino acids is put together, it folds upon itself to create a complex three-dimensional structure. It is the shape of the folded structure that determines its function in the body. Because the folding is determined by the precise sequence of amino acids, each different sequence results in a different protein. Some proteins (such as hemoglobin) contain several different folded chains. Instructions for synthesizing proteins are coded within the DNA.

Coding

Information is coded within DNA by the sequence in which the bases (A, T, G, and C) are arranged. The code is written in triplets. That is, the bases are arranged in groups of three. Particular sequences of three bases in DNA code for specific instructions, such as the addition of one amino acid to a chain. For example, GCT (guanine, cytosine, thymine) codes for the addition of the amino acid alanine, and GTT (guanine, thymine, thymine) codes for the addition of the amino acid valine. Thus, the sequence of amino acids in a protein is determined by the order of triplet base pairs in the gene for that protein on the DNA molecule. The process of turning coded genetic information into a protein involves transcription and translation.

Transcription and translation

Transcription is the process in which information coded in DNA is transferred (transcribed) to ribonucleic acid (RNA). RNA is a long chain of bases just like a strand of DNA, except that the base uracil (U) replaces the base thymine (T). Thus, RNA contains triplet-coded information just like DNA.

When transcription is initiated, part of the DNA double helix opens and unwinds. One of the unwound strands of DNA acts as a template against which a complementary strand of RNA forms. The complementary strand of RNA is called messenger RNA (mRNA). The mRNA separates from the DNA, leaves the nucleus, and travels into the cell cytoplasm (the part of the cell outside the nucleus). There, the mRNA attaches to a ribosome, which is a tiny structure in the cell where protein synthesis occurs.

With **translation**, the mRNA code (from the DNA) tells the ribosome the order and type of amino acids to link together. The amino acids are brought to the ribosome by a much smaller type of RNA called transfer RNA (tRNA). Each molecule of tRNA brings one amino acid to be incorporated into the growing chain of protein, which is folded into a complex three-dimensional structure under the influence of nearby molecules called chaperone molecules.

Genetic pattern of inheritance

Genetics is the study of how genes bring about characteristics, or traits, in living things and how those characteristics are inherited. Genes are specific sequences of nucleotides that code for particular proteins. Through the processes of meiosis and sexual reproduction, genes are transmitted from one generation to the next. Because the principles established by Mendel form the basis for genetics, the science is often referred to as *Mendelian genetics*. It is also called *classical genetics* to distinguish it from another branch of biology known as molecular genetics. Mendel believed that factors pass from parents to their offspring, but he did not know of the existence of DNA. Modern scientists accept that **genes** are composed of segments of DNA molecules that control discrete hereditary characteristics. Most complex organisms have cells that are diploid.

Diploid cells have a double set of chromosomes, one from each parent. For example, human cells have a double set of chromosomes consisting of 23 pairs, or a total of 46 chromosomes. Among these is one pair of chromosomes, called the sex chromosomes, that determines the sex of the individual (XX in females, XY in males). The remaining 22 chromosome pairs are called autosomal chromosomes.

The offspring of sexual reproduction obtain one gene of each type from each parent. The different forms of a gene are called **alleles**. In humans, for instance, there are two alleles for earlobe construction. One allele is for earlobes that are attached, while the other allele is for earlobes that hang free. The type of earlobe a person has is determined by the alleles inherited from the parents. The gene composition of a living organism is its **genotype**. The expression of the genes is referred to as the **phenotype** of a living thing.

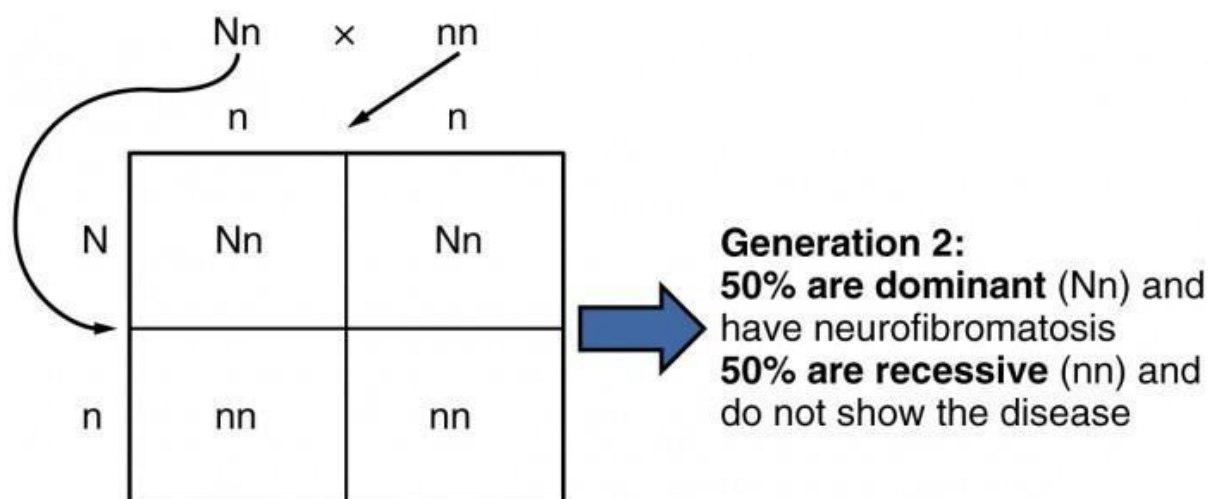
The two paired alleles in an organism's genotype may be identical, or they may be different. An organism's condition is said to be **homozygous** when two identical alleles are present for

a particular characteristic. In contrast, the condition is said to be **heterozygous** when two different alleles are present for a particular characteristic. In a homozygous individual, the alleles express themselves. In a heterozygous individual, the alleles may interact with one another, and in many cases, only one allele is expressed.

When one allele expresses itself and the other does not, the one expressing itself is the **dominant** allele. The “overshadowed” allele is the **recessive** allele. Dominant alleles always express themselves, while recessive alleles express themselves only when two recessive alleles exist together in an individual.

Autosomal Dominant Inheritance

In the case of cystic fibrosis, the disorder is recessive to the normal phenotype. However, a genetic abnormality may be dominant to the normal phenotype. When the dominant allele is located on one of the 22 pairs of autosomes (non-sex chromosomes), we refer to its inheritance pattern as autosomal dominant. An example of an autosomal dominant disorder is neurofibromatosis type I, a disease that induces tumor formation within the nervous system that leads to skin and skeletal deformities. Consider a couple in which one parent is heterozygous for this disorder (and who therefore has neurofibromatosis), Nn , and one parent is homozygous for the normal gene, nn . The heterozygous parent would have a 50 percent chance of passing the dominant allele for this disorder to his or her offspring, and the homozygous parent would always pass the normal allele. Therefore, four possible offspring genotypes are equally likely to occur: Nn , Nn , nn , and nn . That is, every child of this couple would have a 50 percent chance of inheriting neurofibromatosis. This inheritance pattern is shown in the table below, in a form called a Punnett square, named after its creator, the British geneticist Reginald Punnett.

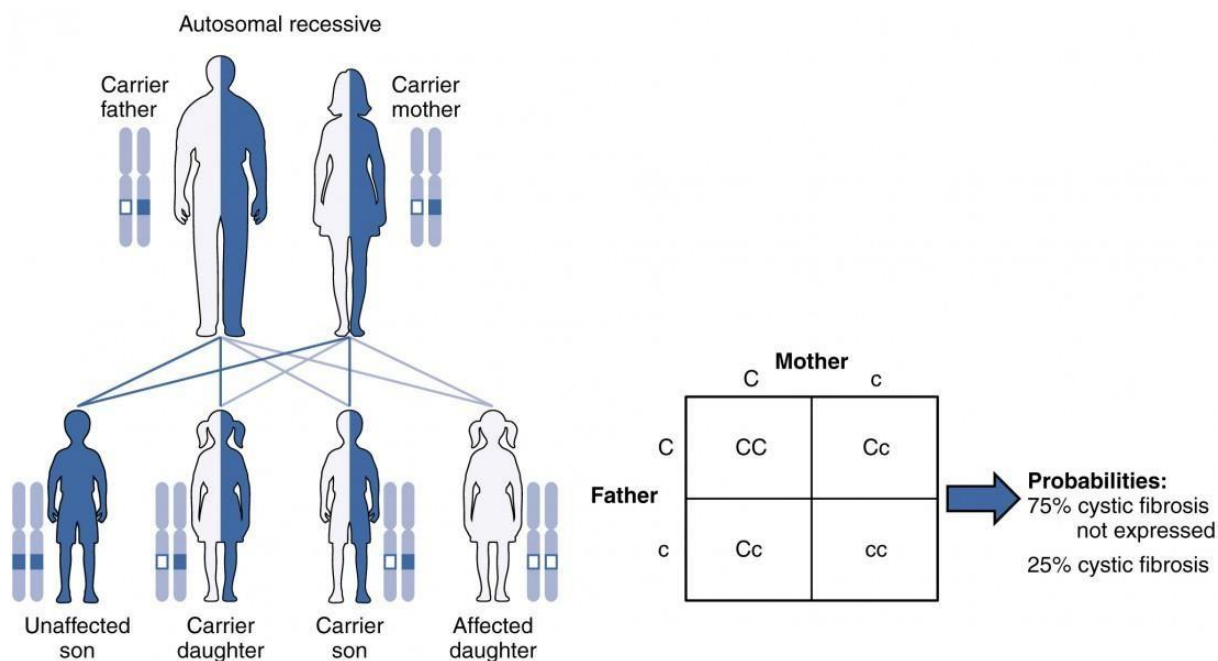


Autosomal Recessive Inheritance

When a genetic disorder is inherited in an autosomal recessive pattern, the disorder corresponds to the recessive phenotype. Heterozygous individuals will not display symptoms of this disorder, because their unaffected gene will compensate. Such an individual is called a carrier. Carriers for an autosomal recessive disorder may never know their genotype unless they have a child with the disorder.

An example of an autosomal recessive disorder is cystic fibrosis (CF), which we introduced earlier. CF is characterized by the chronic accumulation of a thick, tenacious mucus in the lungs and digestive tract. Decades ago, children with CF rarely lived to adulthood. With advances in medical technology, the average lifespan in developed countries has increased into middle adulthood. CF is a relatively common disorder that occurs in approximately 1 in 2000 Caucasians. A child born to two CF carriers would have a 25 percent chance of inheriting the disease. This is the same 3:1 dominant:recessive ratio that Mendel observed in his pea plants would apply here. The pattern is shown in the image below, using a diagram that tracks the likely incidence of an autosomal recessive disorder on the basis of parental genotypes.

On the other hand, a child born to a CF carrier and someone with two unaffected alleles would have a 0 percent probability of inheriting CF, but would have a 50 percent chance of being a carrier. Other examples of autosomal recessive genetic illnesses include the blood disorder sickle-cell anemia, the fatal neurological disorder Tay–Sachs disease, and the metabolic disorder phenylketonuria.

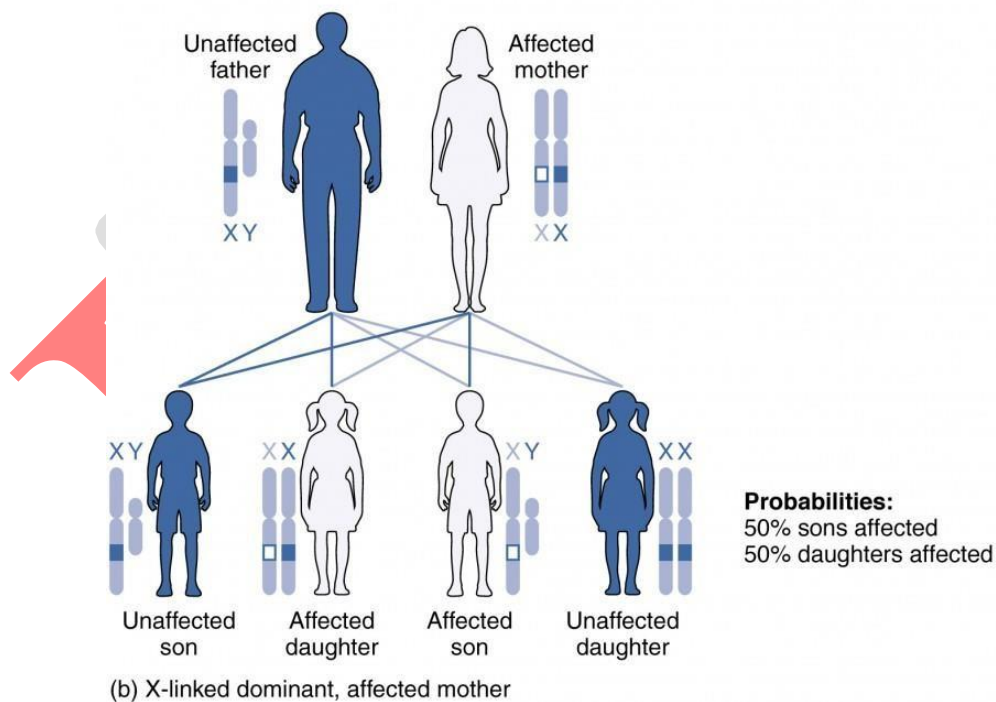
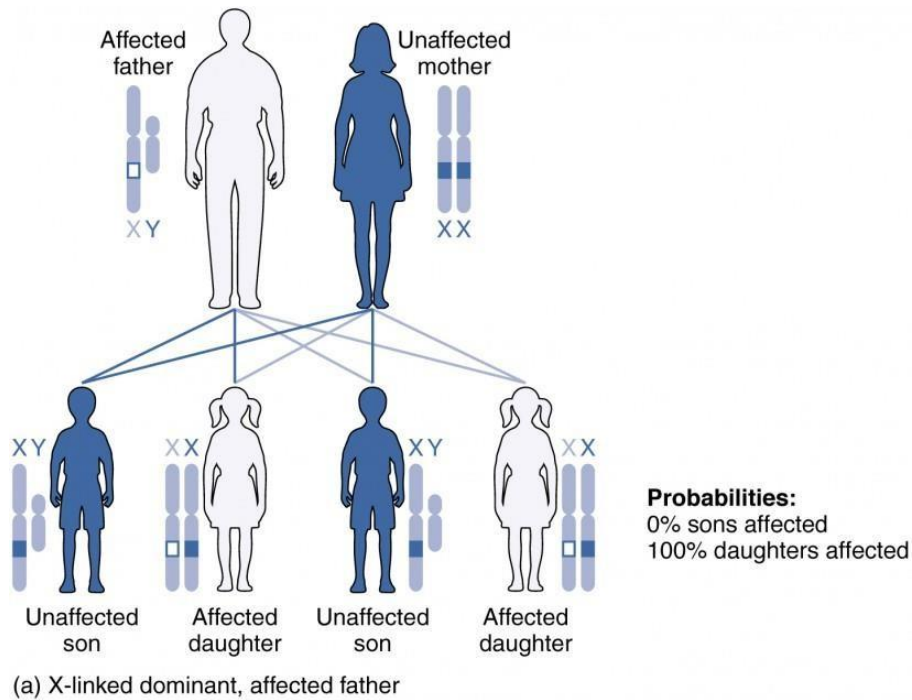


X-linked Dominant or Recessive Inheritance

An X-linked transmission pattern involves genes located on the X chromosome of the 23rd pair. Recall that a male has one X and one Y chromosome. When a father transmits a Y chromosome, the child is male, and when he transmits an X chromosome, the child is female. A mother can transmit only an X chromosome, as both her sex chromosomes are X chromosomes.

X-linked Dominant Inheritance

When an abnormal allele for a gene that occurs on the X chromosome is dominant over the normal allele, the pattern is described as X-linked dominant. This is the case with vitamin D-resistant rickets: an affected father would pass the disease gene to all of his daughters, but none of his sons, because he donates only the Y chromosome to his sons. If it is the mother who is affected, all of her children—male or female—would have a 50 percent chance of inheriting the disorder because she can only pass an X chromosome on to her children. For an affected female, the inheritance pattern would be identical to that of an autosomal dominant inheritance pattern in which one parent is heterozygous and the other is homozygous for the normal gene.



X-linked Recessive Inheritance

X-linked recessive inheritance is much more common because females can be carriers of the disease yet still have a normal phenotype. Diseases transmitted by X-linked recessive inheritance include color blindness, the blood-clotting disorder hemophilia, and some forms of muscular dystrophy. For an example of X-linked recessive inheritance, consider parents in which the mother is an unaffected carrier and the father is normal. None of the daughters

would have the disease because they receive a normal gene from their father. However, they have a 50 percent chance of receiving the disease gene from their mother and becoming a carrier. In contrast, 50 percent of the sons would be affected.

Incomplete Dominance

Not all genetic disorders are inherited in a dominant–recessive pattern. In incomplete dominance, the offspring express a heterozygous phenotype that is intermediate between one parent's homozygous dominant trait and the other parent's homozygous recessive trait.

Codominance is characterized by the equal, distinct, and simultaneous expression of both parents' different alleles. This pattern differs from the intermediate, blended features seen in incomplete dominance.