Glossary

Chromosomal Theory of Inheritance: a theory proposing that chromosomes are the genes' vehicles and that their behavior during meiosis is the physical basis of the inheritance patterns that Mendel observed

linkage: a phenomenon in which alleles that are located in close proximity to each other on the same chromosome are more likely to be inherited together

variants: genotypes or phenotype that deviate from the wild type

wild type: the most commonly occurring genotype or phenotype for a given characteristic found in a population

9.5 Patterns of Inheritance

Learning objectives

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

- *Name and describe examples of the most common human genetic diseases for each type of inheritance—autosomal or sex-linked, dominant or recessive*
- *Be able to perform Punnett squares to predict the possible outcomes of different patterns of inheritance*
- *Be able to define and explain all bolded terms*

Chromosomes carry genes necessary to maintain homeostasis. Thanks to the work of many scientists, it is now understood that chromosomes, not just individual genes, are the heritable units that are passed on from one generation to the next. Recall from chapter eight that both human males and females have twenty-two pairs of homologous chromosomes called autosomes. **Autosomes** are chromosome pairs one through twenty-two and do not determine a person's sex. The twenty-third pair of chromosomes are referred to as the **allosomes** and determine whether an individual will physiologically develop as a male or female.

Human Genetic Disorders

Some human disorders are genetically inherited. These conditions are caused by faulty genes located on chromosomes that often code for non-functional proteins. Genetic disorders can be classified based on whether the gene is located on autosomes or the allosomes. Disorders can be further classified as dominant or recessive, depending on whether a dominant or recessive allele causes the disorder. Most genetic disorders fall into one of three inheritance patterns: autosomal dominant, autosomal recessive, or X (sex) -linked disorders.

Autosomal Dominant Disorders

Autosomal dominant disorders occur when an individual inherits a mutated or faulty dominant allele on an autosome. The person may have one faulty dominant allele and one functional recessive allele (Aa) or two defective dominant alleles (AA). Regardless of whether the individual is homozygous dominant or heterozygous, they will have the genetic condition. Only individuals that are homozygous recessive (aa) will not be affected.

An example of an autosomal dominant disorder is neurofibromatosis type I, a disease that induces tumor formation within the nervous system and leads to skin and skeletal deformities. Consider a couple in which one parent is heterozygous (*Nn)* and has the disorder neurofibromatosis, and the other person (*nn)* is healthy and does not have the disorder. 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 on the normal/functional allele. Therefore, four possible offspring genotypes are equally likely to occur: *Nn*, *Nn*, *nn*, and *nn*. Every child of this couple would have a 50 percent chance of inheriting neurofibromatosis. This inheritance pattern is shown in Figure 9.20.

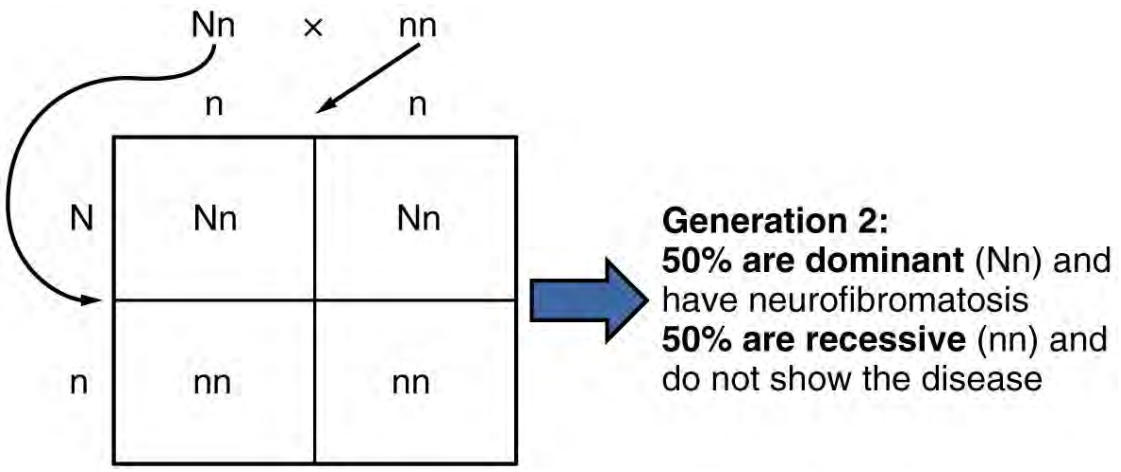

Figure 9.20 Autosomal Dominant Inheritance pattern of an autosomal dominant disorder, such as neurofibromatosis, is shown in a Punnett square. (credit: Betts et al. / Anatomy and Physiology OpenStax)

Other genetic diseases that are inherited in this pattern are achondroplasty dwarfism, Fibrillin – 1 syndrome, and Huntington disease. Because autosomal dominant disorders are expressed by the presence of just one allele, parents that do not have the autosomal dominant condition cannot pass the faulty allele on to their offspring. However, if an offspring has the condition, then at least one of their parents must have at least one defective allele and therefore also has the condition.

Autosomal Recessive Disorders

Autosomal recessive disorders occur when the gene is located on an autosome and the faulty allele which causes the disorder is recessive. When a genetic disorder is inherited in an autosomal recessive inheritance pattern, the condition corresponds to the homozygous recessive genotype. Heterozygous individuals will not display symptoms of this disorder because their functional dominant allele will be expressed. Heterozygous individuals are called "**carriers**." Carriers for an autosomal recessive disorder do not themselves have signs or symptoms of the condition, but they can pass the faulty allele on to their offspring. The carrier may never know they have a defective allele unless they have a child with the condition, or they have their genome sequenced. Only recessive disorders can have carriers since heterozygous individuals with autosomal dominant disorders will always show the disease.

An example of an autosomal recessive disorder is cystic fibrosis (CF). CF is characterized by the chronic accumulation of 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. Figure 9.21 shows what the probability is of having an offspring with an autosomal recessive condition if two carriers mate.