BIOS256- Week 7 Case Study: Cystic Fibrosis

Genetics refers to the study of heredity, specifically which genes are passed on from each parent, and which genes are active. Our nuclear DNA is a combination of DNA from both the paternal and maternal parent, and the genes associated with this DNA can lead to a variety of traits. The term **genotype** refers to the actual combination of alleles, whereas the **phenotype** would be how the trait is being expressed. For most of the genes that will generate our physical characteristics, they will be two forms, the **dominant** form, and the **recessive** form. A **dominant** allele (gene) is one that will be expressed if there is at least one copy of the gene. For example, one or both parents could give a copy of this gene, and the expression pattern would be the same. A **recessive** allele is one that can only be expressed in the absence of a dominant gene, and typically, both copies of the gene must be recessive. In the case of **complete dominance inheritance**, if either of the genes is the dominant form, then the phenotype would be the dominant trait. If all the genes are in the recessive form, or there is no dominant allele, then the phenotype would be the recessive trait.

When discussing genetic disorders associated with any genes on chromosomes 1 – 22, we refer to them as autosomal conditions. **Autosomal dominant** disorders are those that will occur if either parent gives the offspring the dominant gene. Examples of this would include Marfan's syndrome, familial hypercholesterolemia, and hereditary long QT syndrome. **Autosomal recessive** disorders require both parents to give the offspring a copy of the recessive gene, regardless of if the parent expresses the dominant phenotype due to having another copy of the gene that is dominant. Examples of this would include sickle cell disease, cystic fibrosis, and Tay-Sachs disease. The parents in the case of an autosomal recessive disorder may be considered **carriers** if they have both a copy of the dominant gene and a copy of the recessive gene. Individuals with autosomal recessive disorders will pass on that recessive gene if they do produce offspring.

Cystic fibrosis is a genetic condition that predominantly affects Caucasian individuals. This condition affects the cellular membranes in the lungs and digestive systems, leading to a buildup of fluids such as mucus, sweat, and digestive juices. These fluids become viscous (thick) that can lead to blockages in passageways. These blockages can lead to malnutrition, poor growth, breathing abnormalities, and make the individual more prone to respiratory infections. Medications can be prescribed to thin mucus, widen airways, reduce inflammation, and treat chest infections; however, there is no cure for this condition. Sweat will have an excess amount of salt, and a common test for cystic fibrosis is to determine the relative levels of chloride, and if they are elevated, then it could indicate cystic fibrosis.

Background:

Tyler Turner, a 2-week-old infant, has an abnormality on his universal newborn screening lab test that indicates he may have cystic fibrosis. He is currently doing well. His parents report that in the hospital, he did not pass his first bowel movement until the third day of life, and the staff was concerned. His parents have noted that he "tastes salty" when they kiss him.

Desired Outcomes:

1. Differentiate between genotype and phenotype.
2. Educate the parents on how Tyler could have developed cystic fibrosis.