Genetic Disorders

*Features of an offspring that result from genes passed on by parents are called **inherited traits**.*

The length and quality of a human life is determined by many factors, including genes, gender, and interaction with the environment. Faulty genes can result in disease or dysfunction in body parts or systems. Poor environmental conditions, such as inadequate nutrition, medical care, housing, and sanitation can also cause disease or premature death. A third factor that influences the length and quality of life is personal health behavior. All of these factors can be categorized as either inherited or acquired traits.

Inherited Traits:

An organism's physical features are called *traits* or *characteristics*. **Inherited** traits are determined before birth and cannot be permanently changed. Eye color, hair color, skin color, freckles, and dimples are inherited characteristics of humans. Some diseases are inherited, or at least influenced by the inheritance of certain genes. The way that parents pass on genes to their offspring means that the offspring often look *similar* to their parents. However, this does not mean that children will look exactly like their parents. One reason is that some of the genes that are passed on to children stay hidden.

Acquired Traits:

Features that a person gets through choices or interactions with the environment are called **acquired** traits. Scars, tattoos, pierced ears, and dislikes of certain foods are acquired traits because they are not inherited. The clothing and hairstyle that people choose to wear are also acquired characteristics. In addition, anything that a person has learned is acquired, rather than inherited. For example, parents might pass on a love of music to their children. However, this is something that the children have learned and not something they inherited at birth. People and some animals acquire behaviors. For example, people can learn to brush their teeth after meals because it helps prevent tooth decay. Dogs can learn to play fetch. Some animals in changing environments can learn to find new food sources or move to a location where the food is more plentiful. The ability to acquire or learn behaviors can help determine an organism's likelihood of survival.

This girl inherited her hair color from her mother, but the girl will have to learn to play the guitar.

Inherited Genetic Disorders:

Genes are the building blocks of heredity. They are passed from parent to child. They hold DNA, the instructions for making proteins. Proteins do most of the work in cells. They move molecules from one place to another, build structures, break down toxins, and do many other maintenance jobs.

Sometimes there is a mutation, which is a change in a gene or genes. The mutation changes the gene's instructions for making a protein, so the protein does not work properly or is missing entirely. This can cause a medical condition called a genetic disorder.

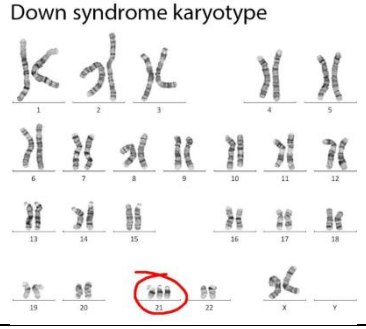
You can inherit a gene mutation from one or both parents. A mutation can also happen during your lifetime. Genetic tests on blood and other tissue can identify genetic disorders.

There are three types of genetic disorders:

- Single-gene disorders, where a mutation affects one gene. Sickle cell anemia is an example.
- Chromosomal disorders, where chromosomes (or parts of chromosomes) are missing or changed. Chromosomes are the structures that hold our genes. Down syndrome is a chromosomal disorder.
- Complex disorders, where there are mutations in two or more genes. Often your lifestyle and environment also play a role. Colon cancer is an example.

Disorders: Choose whether each disease is **inherited** (a result of the genes passed down from your parents) or **acquired** (a result of choices you make in your life and things you are exposed to in your environment). Identify the cause of each disorder. (Hint: some diseases could be inherited AND acquired.)

Inherited	Acquired	Disorder	Cause
		Sickle-Cell Anemia – red blood cells become half-moon, or sickle-shaped; because of this unusual shape, the red blood cells cannot carry as much oxygen (leading to fatigue) and can block blood vessels (which can lead to lung and heart damage and stroke). Sickle-cell anemia is caused by a co-dominant allele. A person with 2 sickle-cell alleles will have the disease, but a person with 1 sickle-cell allele with produce both normal and sickle-shaped blood cells. *The picture shows a normal red blood cell (top) and sickle-cell (bottom).* 	
		Diabetes – the body does not produce enough insulin or the body's cells do not respond to the insulin that is produced, resulting in high blood sugar levels; this can lead to increased thirst, and frequent urination. Excess weight and inactivity can contribute to the development of diabetes. If not controlled, complications such as amputation of legs, blindness, and increased risk of heart attack/stroke may occur.	
		Obesity – obesity is having a body mass index (BMI) over 30; to calculate your BMI: weight in pounds X 705 ÷ height in inches X height in inches. Weight gain is the result of eating more calories than the body can use. By eating foods with lots of sugars and fats and limiting exercise, the body cannot burn all of the calories, so the excess is stored as fat in the body. Being overweight increases your risk for high blood pressure, diabetes, stroke, some cancers, and sleep apnea/respiratory problems.	
		Breast Cancer – uncontrolled cell growth, in which cells divide and reproduce uncontrollably, leading to tumors which may invade other parts of the body. The exact cause of breast cancer is not known, but having certain risk factors increases the likelihood of getting the disease. Risk factors include use of alcohol, obesity, and having the BRCA1 or BRCA2 gene. A person can be tested for the breast cancer gene, but just because you have the gene does not guarantee you will develop breast cancer, it just makes it more likely.	
		Lung Cancer – uncontrolled cell growth, in which cells divide and reproduce uncontrollably, leading to tumors which may invade other parts of the body. Lung cancer can cause symptoms such as coughing, weight loss, shortness of breath, and chest pains. Smoking and exposure to second hand smoke (breathing in the fumes from other people's cigarettes) has been determined to cause the majority of cases of lung cancer.	

		Hemophilia – caused by a recessive allele on the X chromosome. Because males only have 1 X chromosome, males are more likely to have this disorder. A person's blood clots very slowly or not at all; without treatment, they can bleed to death from internal bleeding due to common bumps and bruises.	
		Colorblindness – the inability or decreased ability to perceive differences in color, most often red and green look identical. This is a sex-linked trait caused by a recessive allele on the X chromosome, therefore males are more likely to be colorblind than females. 7-10% of all males have some type of colorblindness.	
		Cystic Fibrosis – a recessive genetic disorder in which the body produces abnormally thick mucus in the lungs and intestines; this makes it difficult for the person to breathe and absorb the proper nutrients from food, which can lead to poor growth.	
		Down Syndrome – a person with Down Syndrome has an extra copy of chromosome 21, so instead of a total of 46 chromosomes in each cell, each cell contains 47 chromosomes. A person with this disorder may have some degree of mental retardation, upward slant to the eyes, decreased muscle tone. *A karyotype is a picture of all the chromosomes in a cell and can be used to diagnose Down Syndrome.* 	
		Emphysema – is a result of long-term tobacco smoking. It can also be caused by exposure to air pollutants, factory fumes, and coal dust. Alveoli and lung tissue are destroyed so oxygen cannot get into the blood. This causes difficulty breathing, shortness of breath, coughing, wheezing, and tiredness. Emphysema cannot be reversed or cured.	
		Tay-Sachs – caused by a mutation on the 15th chromosome, Tay-Sachs causes a chemical to accumulate in the nerve cells of the brain. This results in a weakening of mental and physical abilities, and eventually causes death. Blood tests can determine if a person carries the Tay-Sachs allele.	
		Heart Disease – plaque (a fatty substance) builds up on artery walls causing them to thicken and stiffen which slows the flow of blood. This plague build up is caused by an unhealthy diet, lack of exercise, being overweight, and smoking.	
		Phenylketonuria (PKU) – a defective gene prevents people who suffer from this disease from producing an enzyme their bodies need to use a certain amino acid. This can cause seizures and developmental disorders (they will not grow correctly physically and/or mentally). This disease can be completely controlled as long as the person avoids eating the amino acid, which is found in most foods that contain protein (meat, yogurt, beans).	