Name: **Date:** **Class:**

Information Gathering Worksheet Answer Key
Sickle Cell Anemia

Which genetic disorder are you researching? Sickle Cell Anemia
Be sure to cite your sources and use reliable sources throughout.

What are the symptoms of this disorder?
Symptoms of sickle cell anemia usually appear around 6 months of age. They vary from person to person and may change over time. Symptoms can include: **Anemia.** Sickle cells break apart easily and die. Typical red blood cells usually live for about 120 days before they need to be replaced. But sickle cells usually die in 10 to 20 days, leaving a shortage of red blood cells. This is known as anemia. Without enough red blood cells, the body can't get enough oxygen. This causes fatigue. **Episodes of pain.** Periodic episodes of extreme pain, called pain crises, are a major symptom of sickle cell anemia. Pain develops when sickle-shaped red blood cells block blood flow through tiny blood vessels to the chest, abdomen, and joints. The pain varies in intensity and can last for a few hours to a few days. Some people have only a few pain crises a year. Others have a dozen or more a year. A severe pain crisis requires a hospital stay. Some people with sickle cell anemia also have chronic pain from bone and joint damage, ulcers, and other causes. **Swelling of hands and feet.** Sickle-shaped red blood cells block blood circulation in the hands and feet, which can cause them to swell. **Frequent infections.** The spleen is important for protecting against infections. Sickle cells can damage the spleen, raising the risk of developing infections. Babies and children with sickle cell anemia commonly receive vaccinations and antibiotics to prevent potentially life-threatening infections such as pneumonia. **Delayed growth or puberty.** Red blood cells provide the body with the oxygen and nutrients needed for growth. A shortage of healthy red blood cells can slow growth in babies and children and delay puberty in teenagers. **Vision problems.** Tiny blood vessels that supply blood to the eyes can become plugged with sickle cells. This can damage the portion of the eye that processes visual images, called the retina, and lead to vision problems.
Source: https://www.mayoclinic.org/diseases-conditions/sickle-cell-anemia/symptoms-causes/syc-20355876

Name: **Date:** **Class:**

How common is this disorder?
Approximately 70,000 to 100,000 Americans have sickle cell disease. Sickle cell disease is more common in certain ethnic groups, including: • People of African descent, including African-Americans (among whom 1 in 12 carries a sickle cell gene). • Hispanic-Americans from Central and South America. • People of Middle Eastern, Asian, Indian, and Mediterranean descent.
Source: https://www.hematology.org/education/patients/anemia/sickle-cell-disease#:~:text=Approximately%2070%2C000%20to%20100%2C000%20Americans,of%20an%20inherited%20blood%20disorder
Which specific cells, tissues, and organs are affected by this disorder?
Sickle cell anemia is one of a group of inherited disorders known as sickle cell disease. It affects the shape of red blood cells, which carry oxygen to all parts of the body. Red blood cells are usually round and flexible, so they move easily through blood vessels. In sickle cell anemia, some red blood cells are shaped like sickles or crescent moons. These sickle cells also become rigid and sticky, which can slow or block blood flow.
Source: https://www.mayoclinic.org/diseases-conditions/sickle-cell-anemia/symptoms-causes/syc-20355876
If applicable, what are the current treatments? How successful are they?
There are no standard treatments that cure sickle cell disease. However, there are treatments that help people manage and live with the disease. Treatment relieves pain, prevents infections, minimizes organ damage, and controls complications and can include medications, such as pain relievers and hydroxyurea (Hydrea), at times blood transfusions, and other options as needed. Bone marrow transplants can cure SCD, but appropriate donors are hard to come by. The procedure also carries risks of dangerous side effects.
Source: https://www.hematology.org/education/patients/anemia/sickle-cell-disease#:~:text=Approximately%2070%2C000%20to%20100%2C000%20Americans,of%20an%20inherited%20blood%20disorder https://www.nih.gov/news-events/nih-research-matters/fixing-sickle-cell-disease-gene#:~:text=Sickle%20cell%20disease%20(SCD)%20is,sickle%2Dshaped%20red%20blood%20cells

Name: **Date:** **Class:**

What specific gene is mutated in people with this disorder?
Sickle cell disease (SCD) is a genetic disorder caused by a mutation in both copies of a person's *HBB* gene. This gene encodes a component of hemoglobin, the oxygen-carrying protein in red blood cells. The mutation causes hemoglobin molecules to stick together, creating sickle-shaped red blood cells.
Source: https://www.nih.gov/news-events/nih-research-matters/fixing-sickle-cell-disease-gene#:~:text=Sickle%20cell%20disease%20(SCD)%20is,sickle%2Dshaped%20red%20blood%20cells
Is this mutation dominant or recessive?
Recessive; people with one copy of the mutated gene have sickle cell trait, while people with two copies of the mutated gene have sickle cell anemia.
Source: https://www.hematology.org/education/patients/anemia/sickle-cell-disease#:~:text=Approximately%2070%2C000%20to%20100%2C000%20Americans,of%20an%20inherited%20blood%20disorder
What specific mutation causes this disorder? List the healthy and mutated DNA sequence below. Include at least 15 nucleotides before and after the mutation.
SCA is an autosomal recessive disease caused by a point mutation in the *hemoglobin beta gene* (*HBB*) found on chromosome 11p15.5. Healthy Sequence 5' CAGTAACGGCAGACTTCTCC**T**CAGGAGTCAGATGCACCAT 3' Mutated Sequence 5' CAGTAACGGCAGACTTCTCC**A**CAGGAGTCAGATGCACCAT 3'
Source: https://www.ncbi.nlm.nih.gov/books/NBK22238/#:~:text=SCA%20is%20an%20autosomal%20recessive,are%20somewhat%20protected%20against%20malaria https://genome.ucsc.edu/cgi-bin/hgTracks?db=hg38&lastVirtModeType=default&lastVirtModeExtraState=&virtModeType=default&virtMode=0&nonVirtPosition=&position=chr11%3A5226941%2D5227063&hgsid=2219623990_2hmB3Gs4FglF0xDA7GYhWnSueKgC