

Name: \_\_\_\_\_

# The Mystery of the Crooked Cell

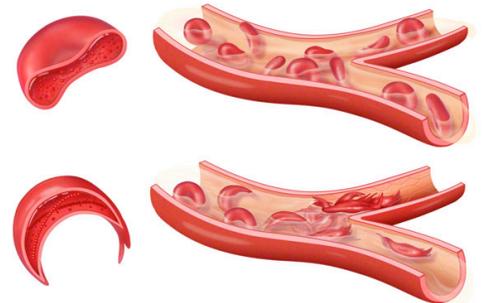
## Investigating the Heredity of a Blood Disorder

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A local high school football player was rushed to the hospital after collapsing on the field. The player reported to the emergency room physician, Dr. James Herrick, that he had experienced similar symptoms multiple times during his childhood. These symptoms include extreme fatigue after non-strenuous exercise, being unusually short of breath, extreme pain in his joints and muscles, a yellowish tint in the whites of his eyes, and tenderness in his abdomen. Consequently, the patient would often have to spend time in bed after a “crisis”.

A look at his immediate family history reveals that neither of his parents have had similar symptoms. However, upon further investigation, it is revealed that the patient’s paternal uncle and grandmother shared the same symptoms under the same circumstances. Based on this medical history, Dr. Herrick believes the patient may have sickle cell anemia.



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Sickle cell anemia is a hereditary disease that is passed down to an individual from their parents. The disease causes red blood cells to form into an abnormal crescent shape which prevents the cells from efficiently carrying oxygen to the body’s tissues. This crescent, or sickle, shape is due to a mutation in the gene that codes for a protein in red blood cells called hemoglobin, the component of red blood cells which binds to oxygen. In particular, the mutation leads to a change in one of the amino acids in the hemoglobin protein.

Dr. Herrick has asked for your help in confirming his diagnosis. To do so, you will be testing the patient’s hemoglobin protein using gel electrophoresis. Based on your results, you can then confirm the patient’s diagnosis.

### What is heredity?

## MATERIALS

Patient's hemoglobin  
Normal hemoglobin  
Sickle hemoglobin

Micropipette and tips  
Electrophoresis equipment  
Microcentrifuge

Practice dye

### PART I - Sample preparation

1. Locate the following samples in your colored tube rack, and, along with the instructor, fill in the following chart:

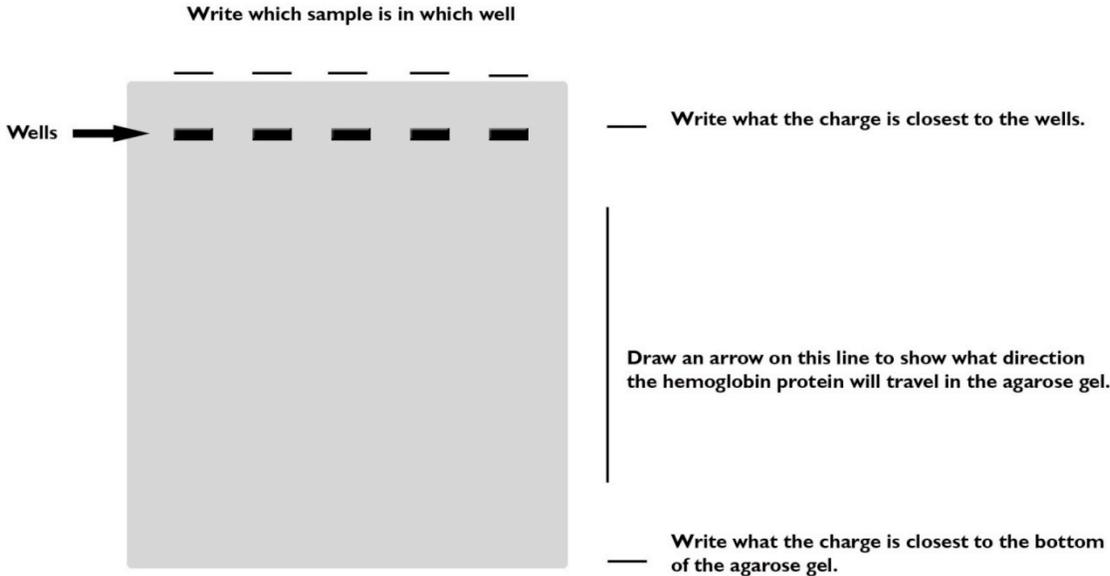
| Sample             | What is the label on the tube? | Well number |
|--------------------|--------------------------------|-------------|
| Normal Hemoglobin  |                                |             |
| Sickle Hemoglobin  |                                |             |
| Patient Hemoglobin |                                |             |

**QUICK CHECK:** Compare the hemoglobin samples to each other. Can you tell a difference between them?

2. Locate the practice gel at your station. Practice loading 15 $\mu$ L of practice dye into three different wells.
3. After practicing, load 15 $\mu$ L of each hemoglobin sample into the well that it was assigned to in the chart above.
4. Locate the electrophoresis box, and place your sample into one of the six gel slots. The gel should be completely covered by the electrophoresis buffer. Place the lid on the box, ensuring that the black wire is on top.
5. Run the gel at approximately 240 volts for around 10 minutes.
6. Please go back to your station.

**PART II - Hemoglobin Electrophoresis**

- 7. Hemoglobin is a protein that has a red color. You will be able to see how far your hemoglobin samples have migrated through the agarose gel. Fill in the example below with your MdBioLab instructor.

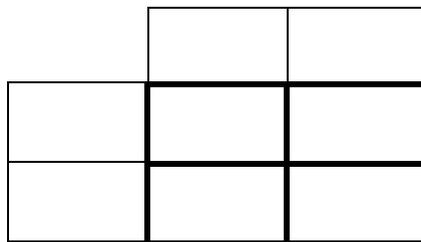


During electrophoresis, why does normal hemoglobin move at a different speed than sickle hemoglobin?

How will you use your patient's results to determine if they have sickle cell disease?

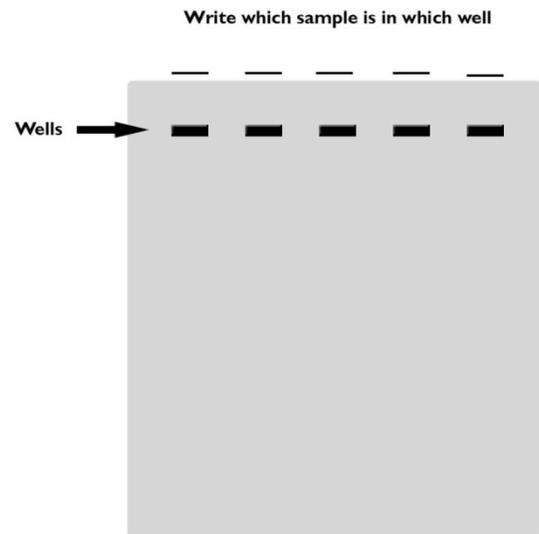
Since neither of the patient's parents have sickle cell disease, is it possible that the patient could have it?

Support your theory with a Punnett square which depicts the patient's possible genotypes:



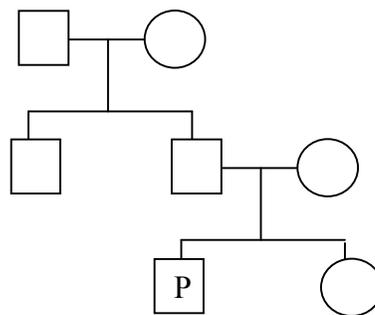
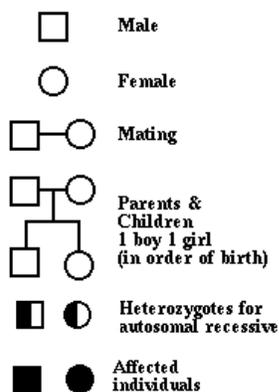
- 8. Draw your results in the diagram, then fill in the patient's genotype and phenotype in the chart below.

| Genotype | Phenotype |
|----------|-----------|
|          |           |



### PART III – Data analysis

Genetic counselors use pedigree charts to show the occurrence and appearance of genotypes and phenotypes within a family. Recall the patient's family history as described on page 1 and his genotype as determined from the analysis of his hemoglobin protein (you may assume that family members exhibiting symptoms had the disease); using the pedigree below, attempt to trace the occurrence of sickle cell disease within his family.



### PART IV - Conclusion

It's known that the patient's sister does not have sickle cell anemia, though her genotype is unknown. Considering the patient's sister is a serious athlete, do you think she should be tested for the sickle-cell trait?



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