



www.NGSSLifeScience.com

Topic: Human Body Systems Disorder Project

Summary: Students will do a BLAST and learn about how the mutated protein affects cells, organs, and organ systems.

Goals & Objectives: Students will learn how to do an effective Internet search to research sickle cell disorder. Students will learn how to compare DNA code of real genes.

NGSS Standards: *LS1-1, LS1-2, LS1-3*: Construct an explanation based on evidence for how the structure of DNA determines the structure of proteins which carry out the essential functions of life through systems of specialized cells.

Time Length: 2-3 days

Prerequisite Knowledge: Protein synthesis and that mutations in the DNA can cause a change in the protein that is made.

Materials:

- Laptops with Internet access
- Preferred (students have a google account where two students can type on the same report at the same time)
- Handout of the project

Procedures:

1. Have every student do part 1, the BLAST. Once they get to part 1 step 10, students can work in groups of two to share / split the work.
2. Parts 1-4 usually takes two days. Students are supposed to use all of the website links provide along with Google searches. Teacher students how to an effective Internet search.
3. Once students are done with part 4, you can have them write on a separate paper a Claim Evidence Reasoning paragraph(s) answering the following question “**How do scientists really know that DNA codes for proteins and those proteins are represented in our traits?**”.

Editable DOCX File and Answer Key:

Available at www.ngsslifescience.com

Name: _____ Row: _____

Date: _____ Period: _____

Human Body Systems Disorder Project

Background:

Many human disorders are caused by mutations. Your goal in this research project is to learn about one. Through your research, you should gain insight on how important DNA is and how it actually really affects your body.

Assignment:

Working in groups of two with a computer, your goal is the find the actual DNA code that codes for a protein found inside your blood cells. Once you have found that DNA code, you need to compare it to mutant DNA that cause diseases in humans. Next, you will research how that mutant DNA affects the blood cells and how those cells affect its body system along with other body systems. Last, you will research how those body systems are used to keep you alive.

Internet Research:

Typing into Google / Bing a question is not the best way to do Internet research. Figure out the most important word(s) in the question and type that in for the search query. Find the most important word, then the next most important word(s). For example: *How many proteins make up hemoglobin? What are the two types?*

Search Query: hemoglobin protein

Bringing It All Together:

On the last page of the project, you will summarize all of the evidence you have collect to answer the question “**How does DNA affect your body to help keep you alive?**”. This question will be answered on another day, but you get to use your evidence page when you go to answer this question. In order to answer this question, you need to make connections between DNA, proteins, cells, body systems, and homeostasis.

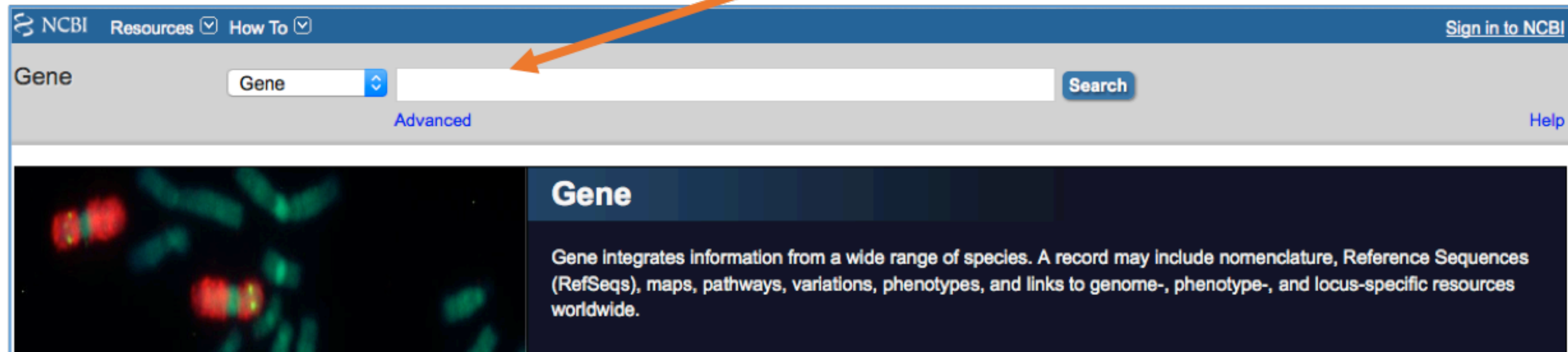
Part 1: Gene Look-up Direction:

1. Go to the website below to look up a gene.

<http://www.ncbi.nlm.nih.gov/gene/>

2. In the search box, type hemoglobin.

Hemoglobin



3. In the search results, click on the HBB link.

Results: 1 to 20 of 2881 << First < Prev Page 1 of 145 Next > Last >>

[See also 1060 discontinued or replaced items.](#)

Name/Gene ID	Description	Location	Aliases	MIM
<input type="checkbox"/> HB1 ID: 816103	non-symbiotic hemoglobin 1 [<i>Arabidopsis thaliana</i> (thale cress)]	Chromosome 2, NC_000962.4 (6982531..6983624, complement)	T2G16060, AHB1, ARATH GLB1, ATGLB1, CLASS HEMOGLOBIN, F7H1.8, F7H1_8, GLB1, HEMOGLOBIN, NSHB1, hemoglobin 1	
<input type="checkbox"/> HB2 ID: 820216	non-symbiotic hemoglobin 2 [<i>Arabidopsis thaliana</i> (thale cress)]	Chromosome 3, NC_003074.8 (3276226..3277860, complement)	AT3G10520, AHB2, ARABIDOPSIS HEMOGLOBIN 2, ARATH GLB2, ATGLB2, GLB2, HEMOGLOBIN, HEMOGLOBIN 2, NON-SYMBIOTIC HAEMOGLOBIN 2, NSHB2, haemoglobin 2	
<input type="checkbox"/> HBB ID: 3043	hemoglobin, beta [<i>Homo sapiens</i> (human)]	Chromosome 11, NC_000011.10 (5225466..5227071, complement)	CD113t-C, beta-globin	141900
<input type="checkbox"/> HBG1 ID: 3047	hemoglobin, gamma A [<i>Homo sapiens</i> (human)]	Chromosome 11, NC_000011.10 (5248272..5249857, complement)	HBG-T2, HBGA, HBGR, HSGGL1, PRO2979	142200
<input type="checkbox"/> HBG2 ID: 3048	hemoglobin, gamma G [<i>Homo sapiens</i> (human)]	Chromosome 11, NC_000011.10 (5253191..5254781, complement)	HBG-T1, TNCY	142250
<input type="checkbox"/> HBA1 ID: 3039	hemoglobin, alpha 1 [<i>Homo sapiens</i> (human)]	Chromosome 16, NC_000016.10 (176651..177522)	HBA-T3, HBH	141800
<input type="checkbox"/> HBA2 ID: 3040	hemoglobin, alpha 2 [<i>Homo sapiens</i> (human)]	Chromosome 16, NC_000016.10 (172847..173710)	HBA-T2, HBH	141850
<input type="checkbox"/> HBD ID: 3045	hemoglobin, delta [<i>Homo sapiens</i> (human)]	Chromosome 11, NC_000011.10 (5232829..5234628, complement)		142000
<input type="checkbox"/> HBE1 ID: 3046	hemoglobin, epsilon 1 [<i>Homo sapiens</i> (human)]	Chromosome 11, NC_000011.10 (5268345..5281945, complement)	HBE	142100
<input type="checkbox"/> Hbb ID: 24440	hemoglobin, beta [<i>Rattus norvegicus</i> (Norway rat)]	Chromosome 1, NC_005100.4 (168971269..168972680, complement)		

- On the HBB hemoglobin beta [*Homo sapiens* (human)] page, you will see a link in the third section down “Genomic regions, transcripts, products”. Click on the FASTA link.

HBB hemoglobin, beta [*Homo sapiens* (human)]
 Gene ID: 3043, updated on 16-Aug-2015

Summary

Official Symbol HBB provided by [HGNC](#)
Official Full Name hemoglobin, beta provided by [HGNC](#)
Primary source [HGNC:HGNC:4827](#)
See related [Ensembl:ENSG00000244734](#); [HPRD:00786](#); [MIM:141900](#); [Vega:OTTHUMG00000066678](#)
Gene type protein coding
RefSeq status REVIEWED
Organism [Homo sapiens](#)
Lineage Eukaryota; Metazoa; Chordata; Craniata; Vertebrata; Euteleostomi; Mammalia; Eutheria; Euarchontoglires; Primates; Haplorrhini; Catarrhini; Hominidae; Homo
Also known as CD113t-C; beta-globin
Summary The alpha (HBA) and beta (HBB) loci determine the structure of the 2 types of polypeptide chains in adult hemoglobin, Hb A. The normal adult hemoglobin tetramer consists of two alpha chains and two beta chains. Mutant beta globin causes sickle cell anemia. Absence of beta chain causes beta-zero-thalassemia. Reduced amounts of detectable beta globin causes beta-plus-thalassemia. The order of the genes in the beta-globin cluster is 5'-epsilon -- gamma-G -- gamma-A -- delta -- beta--3'. [provided by RefSeq, Jul 2008]
Orthologs [mouse](#) [all](#)

Genomic context

Location: 11p15.5 [See HBB in Epigenomics, MapViewer](#)
Exon count: 3

Annotation release	Status	Assembly	Chr	Location
107	current	GRCh38.p2 (GCF_000001405.28)	11	NC_000011.10 (5225466..5227071, complement)
105	previous assembly	GRCh37.p13 (GCF_000001405.25)	11	NC_000011.9 (5246696..5248301, complement)

Genomic regions, transcripts, and products

Genomic Sequence: [NC_000011.10 Chromosome 11 Reference GRCh38.p2 Primary Assembly](#)

Go to nucleotide: [Graphics](#) [FASTA](#) [GenBank](#)

- On this page, you will see the entire gene and all of its nitrogen bases (letters). Click on the link titled “Run BLAST”. On the next page, click the BLAST button at the bottom of the screen

Display Settings: FASTA

Homo sapiens chromosome 11, GRCh38.p2 Primary Assembly
 NCBI Reference Sequence: NC_000011.10
[GenBank](#) [Graphics](#)

>gi|568815587:c5227071-5225466 Homo sapiens chromosome 11, GRCh38.p2 Primary Assembly

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ACATTTGCTTCTGACACAAGTGTCTCACTAGCAACCTCAAACAGACACCATGGTGCATCTGACTCCTGA
GGAGAAGTCTGCCGTTACTGCCCTGTGGGCAAGGTGAACCTGGATGAAGTTGGTGGTGGAGCCCTGGGC
AGGTTGGTATCAAGGTTACAAGACAGGTTTAAGGAGACCAATAGAACTGGGCATGTGGAGACAGAGAAG
ACTCTGGGTTTCTGATAGGCACTGACTCTCTGCTATTTGGTCTATTTCCACCCTTAGGCTGCTGG
TGGCTTACCCTTGGACCCAGAGTCTTTGAGTCTTTGGGGATCTGTCCACTCCTGATGCTTATGGG
CAACCCTAAGGTGAAGGCTCATGGCAAGAAAGTCTCGGTGCTTTAGTGTATGGCTGGCTACCTGGAC
AACCTCAAGGGCACCTTGGCACACTGAGTGAAGCTGCACTGTGACAAGCTGCACGTGGATCCTGAGAACT
TCAGGGTGAATCTTGGGACGCTTGAATGTTTCTTCCCTTCTTTCTATGGTTAAGTTCATGTCATAG
GAAGGGATAAGTAACAGGGTACAGTTAGAATGGGAAACAGACGAATGATGCATCAGTGTGAAGTCT
CAGGATCGTTTATGTTTCTTTATTTGCTGTTCAACAATGTTTCTTTGTTAATCTTCTGCTTCT
TTTTTTTCTCTCCCAATTTTACTATATATACTTAATGCCTTAACATTTGTGTATAAAGAAATA
TCTCTGAGATACATTAAGTAACCTTAAAAAATTTTACACAGTCTGCTAGTACATTAATTTGGAAT
ATATGTGCTTATTTGCATATTCATAATCTCCCTACTTATTTCTTTTAAATTTAATGATACATAAT
CATATACATATTTAAGGTTAAGTGAATGTTTAAATATGTACACATATTGACCAATCAGGGTAA
TTTTGCATTTGTAATTTAAAAAATGCTTCTTTTAAATATACTTTTGTATCTTATTTCTAATA
CTTCCCTAATCTCTTTCTTTCAGGGCAATAATGATAAATGATCATGCTCTTTCGACCAATCTAAG
AATAACAGTGAATTTCTGGGTTAAGGCAATAGCAATATCTCTGCATATAAATTTCTGCATATAAT
TGTAACATGATGAAGAGGTTTCTATATGCTAATAGCAGCTACAATCCAGCTACCATTCTGCTTTATTTT
ATGGTTGGGATAAGGCTGGATTTCTGAGTCCAAGCTAGGCCCTTTGCTAATCATGTTCCATACCTCTT
ATCTTCCCTCCACAGCTCTGGGCAACGTGCTGGTCTGTGCTGGCCCACTTTGGCAAGAATTCAC
CCCCACAGTGCAGGCTGCTATCAGAAAGTGGTGGTGGTGGCTAAAGCCCTGGCCACAAAGTATCA
CTAAGCTCGCTTCTGCTGCTCAATTTCTATTAAGGTTCTTTGTTCCCTAAGTCCAACCTACTAACT
GGGGATATTAAGAGGCTTGGACATCTGGATCTGCTCAATAAAAAACATTTATTTTCATTGTC
  
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Change region shown
 Whole sequence
 Selected region
 from: 5225466 to: 5227071 [Update View](#)

Customize view
Display options
 Show reverse complement [Update View](#)

Analyze this sequence
[Run BLAST](#)
[Pick Primers](#)
[Highlight Sequence Features](#)
[Find in this Sequence](#)

LinkOut to external resources
[Ensembl](#) [Ensembl]
[Ensembl](#) [Ensembl]
[Ensembl](#) [Ensembl]

- Under Choose Search Set, on the organism text box, type “Homo sapiens”.
- Scrolls down to the bottom and click + Algorithm parameters. Select “Max target sequences” to equal 1000. Then click the “BLAST” button at the bottom of the page.

The screenshot shows the NCBI BLAST search interface. The 'Database' is set to 'Nucleotide collection (nr/nt)'. The 'Organism' is 'Homo sapiens (taxid:9606)'. The 'Program Selection' section shows 'Highly similar sequences (megablast)' selected. The 'Algorithm parameters' section shows 'Max target sequences' set to 1000. A 'BLAST' button is visible at the bottom left of the search area.

- It may take a few seconds to run the DNA comparison search, but once you get to the results page, scroll down to the descriptions section. Listed here are hemoglobin genes that have been sequenced. Scroll down to near the bottom of the list until you get to **Homo sapiens beta-globin (HBB) gene, with t to c mutation L114P resulting in dominant beta-thalassemia intermedia**
- What you will notice is that there are several different types of mutations that will cause beta-thalassemia. Beta thalassemia is a type of sickle cell disease. Click the link below. **Homo sapiens beta-globin (HBB) gene, with t to c mutation L114P resulting in dominant beta-thalassemia intermedia**
- Look at line Query 5225884 for a mutation with subject 1352. A missing line represents a change in nitrogen base (change in the letter). G is changed into an A

Query	5225884	TTATATGCAGAAATATTTATATGCAGAGATATTGCTATTGCCTTAACCCAGAAATTATCA	52259
Sbjct	1354	TTATATGCAGAAATATTTATATGCAGAAATATTGCTATTGCCTTAACCCAGAAATTATCA	1295

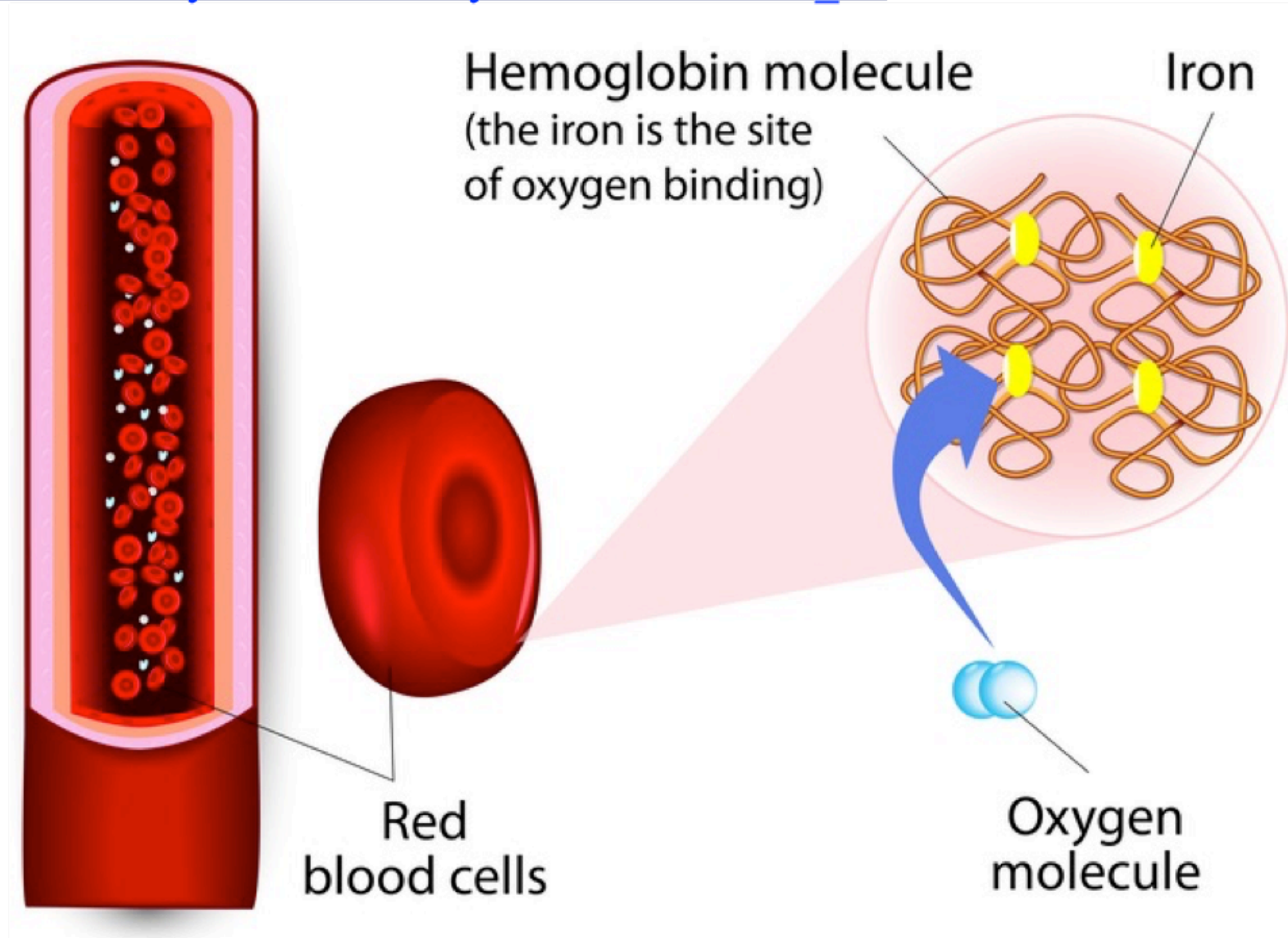
11. Try to find other mutations in the gene comparison and record the mutation in the data table below. Find three more mutations and enter in the location and which letter was changed.

Query: <u>5225884</u> Letter: <u>G</u>	Query: _____ Letter: ____
Sbjct: <u>1354</u> Letter: <u>A</u>	Sbjct: _____ Letter: ____
Query: _____ Letter: ____	Query: _____ Letter: ____
Sbjct: _____ Letter: ____	Sbjct: _____ Letter: ____

Part 2: Research Hemoglobin Proteins and Red Blood Cell

The red blood cell has proteins inside of it called hemoglobin. You need to research to answer the following questions. You can use an Internet search or the following website. You MAY NOT copy straight from the Internet, you need to write in your own words to answer the questions.

http://evolution.berkeley.edu/evolibrary/article/mutations_06



1. There are 4 polypeptide chains (alpha and beta) that make up hemoglobin. Each of the polypeptide chains are held together by iron. What is another function of iron for hemoglobin?
2. How is the normal hemoglobin and the hemoglobin in sickled red blood cells different?
Explain
3. What is a negative phenotype of sickled red blood cell? Explain

4. What is a positive phenotype of sickled red blood cell? Explain

5. What body system is the red blood cell part of? What is the main job of that body system?

6. Draw or provide a picture of a normal red blood cell and a sickle red blood cell.

Part 3: Research Sickle Cell Disease

You need to research more information on sickle cell disease, specifically beta-thalassemia. In your research make sure to answer the following questions. You can use an Internet search or the following website. You MAY NOT copy straight from the Internet, you need to write in your own words to answer the questions.

<http://ghr.nlm.nih.gov/condition/sickle-cell-disease>

7. What are the signs and symptoms of sickle cell disease?

8. What are other body organs / systems affected by sickle cell? Provide a list of organs / systems.

9. Select one system / organ from above that is NOT part of the circulatory system (ex. heart). Explain in detail how another body system / its main organ is affected.

Part 4: Summarize your evidence

Your goal is to now summarize all of the research you have done in this project to answer the following question: **How does DNA affect your body to help keep you alive?** The key thing to write is the evidence from your research that supports each of the ideas.

10. DNA codes for Proteins

11. Proteins provide an essential biological function

12. Cells make up tissues, organs, and body systems

Name: _____ Row: _____

Date: _____ Period: _____

Part 5: DNA to Proteins CER

Question: How do scientists really know that DNA codes for proteins and those proteins are represented in our traits? Write a Claim Evidence Reason paragraph to answer this question.

Your evidence needs to be based on real life information you found in this project. Your reasoning needs to include scientific concepts learned in class.
