

## Genetic Variation in Human Populations Student Material

### Introduction

Humans get malaria from the bite of a malaria-infected mosquito. When a mosquito bites an infected person, it ingests microscopic malaria parasites (*Plasmodium*) found in the person's blood. The parasites cause mosquitoes to regurgitate saliva into the human host, which transmits sporozoites that travel to the liver. During this time when the parasites are in the liver, the person has not yet felt sick. The parasites leave the liver and enter red blood cells; this may take as little as 8 days or as many as several months. Once inside the red blood cells, the parasites grow and multiply. As the parasites over populate a red blood cell (RBC), they break the wall and infect other RBC's. It is during the breaking of the RBC's walls that a person infected feels chills. Toxins (poisonous substances) from the parasite are also released into the blood, making the person feel sick. If a mosquito bites this person while the parasites are in his or her blood, it will take in the tiny parasites. After a week or more, the mosquito can infect another person.

*About 10-20% of the world's cases of Plasmodium vivax malaria occur in Africa, south of the Sahara. In eastern and southern Africa, P. Vivax represents around 10% of malaria cases but in less than 1% of cases in western and central Africa. Plasmodium vivax malaria was eliminated in Australia in the 1980s but a very small number (probably less than 10) have occurred there recently.*

Malaria infects about 300 million people annually worldwide and kills more than 3 million people. Each year in the United States, a few cases of malaria result from blood transfusions, are passed from mother to fetus during pregnancy, or are transmitted by locally infected mosquitoes.

Study the following three maps to answer the following questions:

1. What is the range of frequencies for each allele shown?
2. Which allele varies most in frequency?
3. Which allele varies least in frequency?
4. Study the 4<sup>th</sup> map that shows the patterns in the occurrence of *Plasmodium vivax* malaria. Compare the incidence of *Plasmodium vivax* malaria with the distribution of the three different alleles. Is there any

correlation or relationship between any of the alleles and the incidence of *Plasmodium vivax malaria*?

5. Review the background information that describes how malaria spreads and the description of the function of each of three alleles. Propose one or more hypotheses to explain the distribution of one or more of these alleles.

Description of the function of each of the three alleles:

- The GC gene, which has two major alleles, 1 and 2, codes for a blood protein that attaches to vitamin D and regulates its distribution within the body.
- The HP gene codes for another blood protein (haptoglobin). This protein attaches to the hemoglobin released by blood cells when they decay at the end of their natural life or when they are destroyed by a disease, such as malaria.
- The FY gene codes for a protein that is normally found on the surface of red blood cells. The protein makes it easier for a particular malarial parasite, *Plasmodium vivax*, to get into the red blood cell. Once in the red blood cell, *P. vivax*, like all malarial parasites, multiplies. The FY-O allele results in the absence of the protein.

The data on maps 1-3 show that the distribution of three different alleles varies in human populations worldwide. Even though the FY-O allele is most frequent in a large section of Africa, the other alleles would be present. Thus, there is a wide range in the frequency of alleles in that region. This is not unusual. These and other alleles also vary within smaller populations. Study table 1, which shows the percent variation in 6 different gene segments.

6. What is the pattern of the data on Table 1, which describes the percent variation in gene regions? You do not need to understand the function of each of the genes. However, information about the gene segments is provided on the bottom of this page.

7. The table that shows the percentage of variation at two levels of population structure is from a study of worldwide genetic variation in 255 individuals (72 Africans, 63 Asians, and 120 Europeans). On the basis of the distribution of the genetic variation shown in the two tables and the three maps, what can you conclude about the genetic differences in humans? Based on these data, could you draw boundaries that would separate human populations clearly on the basis of genetic differences?
8. A spiral-shaped bacteria called *Helicobacter pylori* infect the stomachs of more than half the world's population, causing gastritis in many people and ulcers and stomach cancer in some. These bacteria adhere, or bind, to the stomach by sticking to the same complex sugars that define blood types A, B, and O. People with blood type O tend to have a higher frequency of ulcers and stomach cancer than individuals with other blood types. Europeans, Japanese, and native Alaskans, whose populations include individuals of all blood types, are infected by *Helicobacter pylori* that bind equally well to the A, B, and O sugars that define these blood types. "Bugs" from South American Indians, however, bound only the group O antigen.

Develop a hypothesis based on these data.

#### **Definitions for question 6:**

**STRP:** autosomal short-tandem-repeat-polymorphisms. These are variable segments of DNA that are 3, 4, or 5 bases long, repeated over and over.

**RSP:** autosomal restriction-site polymorphism. DNA sequence variations that occur at a restriction enzyme recognition sequences that result in variations (polymorphisms) in the length of DNA fragments obtained by cutting DNA with a particular restriction enzyme.

**ALU:** Alu-insertion polymorphism. Alu sequences are repeat sequences that are about 300 bases long. There are many thousands of Alu repeats in the genome and they appear within genes and between genes. They have no known function.

**HVSI and HVS2:** hypvariable sequences 1 and 2. This is DNA from mitochondria taken from regions that have a lot of differences in DNA sequences.

**YSTRP:** Y chromosome short-tandem-repeat polymorphisms.

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## Genetic Variation in Human Populations

### Teacher material

#### Overview

Data showing genetic variation in human populations are analyzed in this activity to answer a series of questions about variation within and between human populations.

#### TEKS-Biology

- 7 knows the theory of evolution
- 7A identify evidence of change in species using fossils, DNA sequences, anatomical similarities, & embryology
- 7B illustrate the results of natural selection in speciation, diversity, phylogeny, adaptation, behavior & extinction.
- 12C compare variations, tolerances, & adaptations of plants & animals in different biomes
- C3E evaluate models according to their adequacy in representing biological objects or events

#### Engagement

How do we ordinarily identify a person as a member of a particular racial or ethnic group?

What problems emerge from using external characteristics such as skin color, hair texture, or facial structures as a basis for identifying individuals by race or ethnic group?

Are there other biological indicators are useful in providing knowledge about the similarities and differences in and among groups? (focus of this lesson)

#### Exploration

##### *Introduction:*

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Malaria infects about 300 million people annually worldwide and kills more than 3 million people. Each year in the United States, a few cases of malaria result from blood transfusions, are passed from mother to fetus during pregnancy, or are transmitted by locally infected mosquitoes.

Study the following three maps to answer the following questions:

1. What is the range of frequencies for each allele shown? *GC-1: The range is from a low of 74% to a high of 88%; HP1: The range is from a low of 17% to a high of 60%; FY-O: The range is from a low of 0% to a high of 96%.*
2. Which allele varies most in frequency? *FYO*
3. Which allele varies least in frequency? *Gc-1*
4. Study the 4<sup>th</sup> map that shows the patterns in the occurrence of *Plasmodium vivax* malaria. Compare the incidence of *Plasmodium vivox* malaria with the distribution of the three different alleles. Is there any correlation or relationship between any of the alleles and the incidence of *Plasmodium vivox* malaria? *The incidence of malaria is the highest in Africa and the lowest in Australia and South America. The incidence, or presence, of Plasmodium vivox malaria is the highest in Africa where the distribution of the FY-O allele is 96% and absent in Australia where the FY-O allele is not present.*
5. Review the background information that describes how malaria spreads and the description of the function of each of three alleles. Propose one or more hypotheses to explain the distribution of one or more of these alleles.

*Hypothesis 1: The absence of a protein normally found on the surface of red blood cells makes it more difficult for Plasmodium vivox malaria parasites to get into red blood cells.*

*Hypothesis 2: The FY-O gene provides some protection against Plasmodium vivox malaria.*

*Other hypotheses could be proposed. Evidence supports hypotheses 1 & 2. The presence of FY-O genes would increase the survival rate of humans in an area where Plasmodium vivox malaria is common. In areas where Plasmodium vivox malaria is not common, the presence of FY-O would not confer any survival value to a human population.*

Description of the function of each of the three alleles:

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The data on maps 1-3 show that the distribution of three different alleles varies in human populations worldwide. Even though the FY-O allele is most frequent in a large section of Africa, the other alleles would be present. Thus, there is a wide range in the frequency of alleles in that region. This is not unusual. These and other alleles also vary within smaller populations. Study table 1, which shows the percent variation in 6 different gene segments.

6. What is the pattern of the data on Table 1, which describes the percent variation in gene regions? You do not need to understand the function of each of the genes. However, information about the gene segments is provided on the bottom of this page. *The percentage variation in a gene region is higher within a population but very low within populations on a continent. Thus, there could a high percentage of variation for a particular trait in a population clustered in Chicago or Atlanta, but the overall variation for the trait among all the population in the United States would be low.*
7. The table that shows the percentage of variation at two levels of population structure is from a study of worldwide genetic variation in 255 individuals (72 Africans, 63 Asians, and 120 Europeans). On the basis of the distribution of the genetic variation shown in the three maps and two tables, what can you conclude about the genetic differences in humans? Based on these data, could you draw boundaries that would separate human populations clearly on the basis of genetic differences?

**Major concept:** *The study of genetic variations in Homo sapiens shows that there is more genetic variation within populations than between populations as shown in these two tables. This means that two random individuals from any one group are almost as different as any two random individuals from the entire world. For example, all three of the alleles shown on the 3 maps were found in Africa even though the FY-O gene was the most frequent. Thus, it would be a mistake to use the presence of the FY-O gene as a marker or characteristic used to identify or classify*

someone of African descent.

*Although it may be easy to observe distinct external differences between groups of people, it is more difficult to distinguish such groups genetically, since most genetic variation is found within all groups. As a result, it is difficult to draw boundaries that separate humans on the basis of genetic differences.*

*F. Collins, Director, of the National Center for Human Genomic Research & National Institutes of Health in the presentations “Consequences of the Human Genome Project for Medicine & Society” during the 2001 Annual Meeting of the American Association for the Advancement of Science stressed the following:*

- *Humans are 99.9% identical genetically*
  - *Most of our genetic differences are shared among all ethnicities and races.*
  - *There is no scientific basis for precise racial categories.*
  - *Social and cultural factors define race. Science does not.*
  - *Founder group in Africa 100,000 years ago.*
7. To infect the stomach, ulcer-causing bacteria must first stick to the stomach wall, and some strains do it better than others. A recent study of various types reveals why: Infective bugs alter their tactics to adapt to different people's stomachs. The results help explain why South American Indians suffer so much from ulcer bugs, and it could help researchers develop a therapeutic vaccine that prevents ulcers and stomach cancer.

A spiral-shaped bacteria called *Helicobacter pylori* infect the stomachs of more than half the world's population, causing gastritis in many people and ulcers and stomach cancer in some. These bacteria adhere, or bind, to the stomach by sticking to the same complex sugars that define blood types A, B, and O. People with blood type O tend to have a higher frequency of ulcers and stomach cancer than individuals with other blood types. Europeans, Japanese, and native Alaskans, whose populations include individuals of all blood types, are infected by *Helicobacter pylori* that bind equally well to the A, B, and O sugars that define these blood types. “Bugs” from South American Indians, however, bound only the group O antigen.

Develop a hypothesis based on these data.

*Hypothesis: There is a specialized form of Helicobacter pylori that sticks to the sugar that define blood type O.*

Researchers have found that the gene for a protein called BabA, which tightly binds the blood-group sugars, has a slightly different sequence in specialist and generalist bacteria. Swapping a small stretch of DNA turned a specialist bacterium back into a generalist. That means that *Helicobacter* relies on BabA to bind the stomach lining. This is the first study that shows ulcer bugs adapt to different human populations. (reference: "Bug to gut: you're just my type" *Science Now*, July 22, 2004.  
<http://sciencenow.sciencemag.org/cgi/content/full/2004/722/3?etoc>)

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**YSTRP:** Y chromosome short-tandem-repeat polymorphisms.

### **Reference:**

The reference for the maps, chart, and some of the background information is unknown.

Developed by  
Gerald Skoog  
Texas Tech University  
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