Does Race Exist?

Lab 9

Traces of a Distant Past

Lab 9 Does Race Exist I Pre-lab Reading

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Look around on the streets of any major city, and you will see a sampling of the outward variety of humanity: skin tones ranging from milk-white to dark brown; hair textures running the gamut from fine and stick-straight to thick and wiry. People often use physical characteristics such as these–along with area of geographic origin and shared culture–to group themselves and others into "races." But how valid is the concept of race from a biological standpoint? Do physical features reliably say anything informative about a person's genetic makeup beyond indicating that the individual has genes for blue eyes or curly hair?

The problem is hard in part because the implicit definition of what makes a person a member of a particular race differs from region to region across the globe. Someone classified as "black" in the U.S., for instance, might be considered "white" in Brazil and "colored" (a category distinguished from both "black" and "white") in South Africa.

Yet common definitions of race do sometimes work well to divide groups according to genetically determined propensities for certain diseases. Sickle-cell disease is usually found among people of largely African or Mediterranean descent, for instance, whereas cystic fibrosis is far more common among those of European ancestry. In addition, although the results have been controversial, a handful of studies have suggested that African-Americans are more likely to respond poorly to some drugs for cardiac disease that are members of other groups.

Over the past few years, scientists have collected data about the genetic constitution of populations around the world in an effort to probe the link between ancestry and patterns of disease. These data are now providing answers to several highly emotional and contentious questions: Can genetic information be used to distinguish human groups having a common heritage and to assign individuals to particular ones? Do such groups correspond well to predefined descriptions now widely used to specify race? And, more practically, does dividing people by familiar racial definitions or by genetic similarities say anything useful about how members of those groups experience disease or respond to drug treatment?

In general, we would answer the first question yes, the second question no, and offer a qualified yes to the third. Our answers rest on several generalizations about race and genetics. Some groups do differ genetically from others, but how groups are divided depends on which genes are examined; simplistically put, you might fit into one group based on your skin-color genes, but another based on a different characteristic. Many studies have demonstrated that roughly 90% of human genetic variation occurs within a population living on a given continent, whereas about 10% of variation distinguishes continental populations. In other words, individuals from different populations are, on

average, just slightly more different from one another than are individuals from the same population. Human populations are very similar, but they often can be distinguished.

Classifying Humans

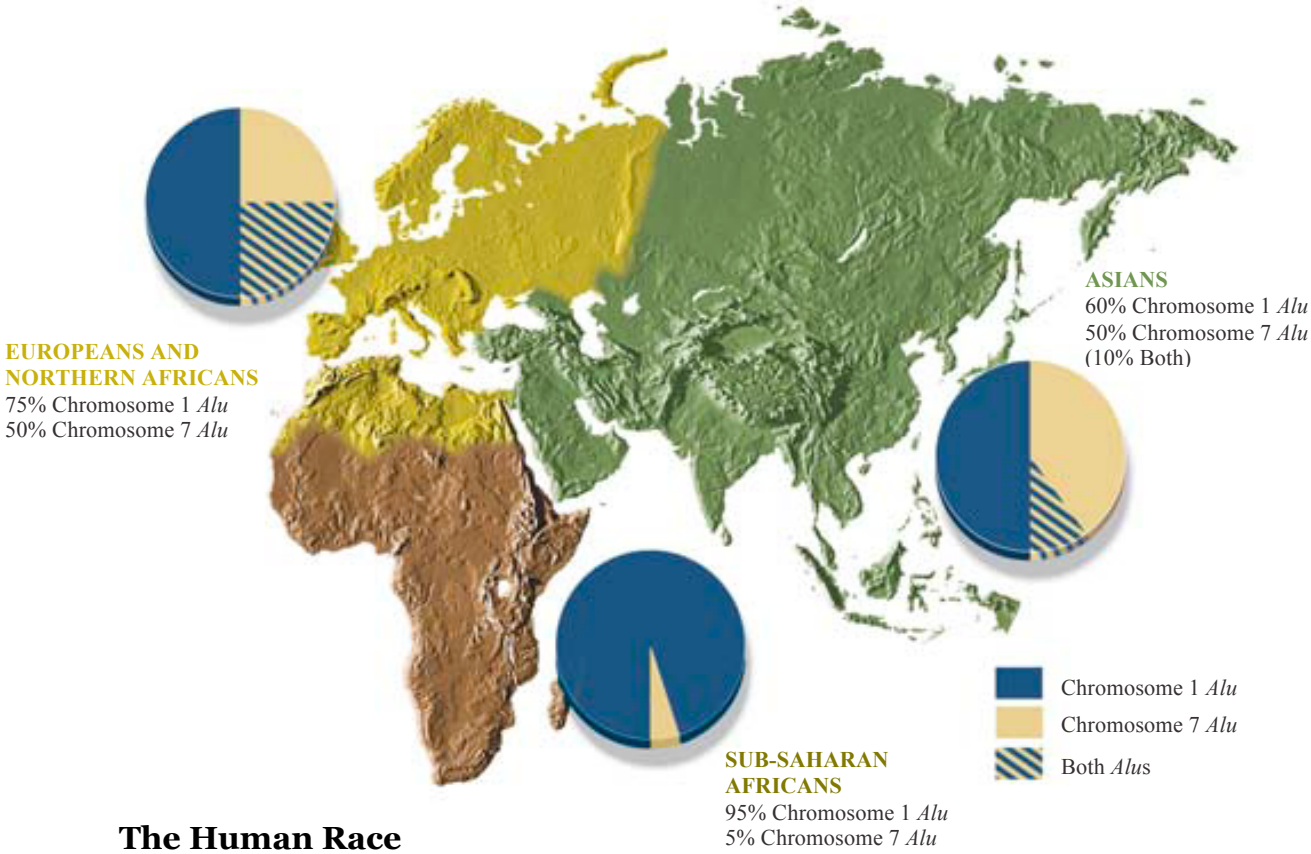
As a first step to identifying links between social definitions of race and genetic heritage, scientists need a way to divide groups reliably according to their ancestry. Over the past 100,000 years or so, anatomically modern humans have migrated from Africa to other parts of the world, and members of our species have increased dramatically in number. This spread has left a distinct signature in our DNA.

To determine the degree of relatedness among groups, geneticists rely on tiny variations, or polymorphisms, in the DNA – specifically in the sequence of base pairs, the building blocks of DNA. Most of these polymorphisms do not occur within genes, the stretches of DNA that encode the information for making proteins (the molecules that constitute much of our bodies and carry out the chemical reactions of life). Accordingly, these common variations are neutral, in that they do not directly affect a particular trait. Some polymorphisms do occur in genes, however; these can contribute to individual variation in traits and genetic diseases.

As scientists have sequenced the human genome (the full set of nuclear DNA), they have also identified millions of polymorphisms. The distribution of these polymorphisms across populations reflects the history of those populations and the effects of natural selection. To distinguish among groups, the ideal genetic polymorphism would be one that is present in all the members of one group and absent in the members of all other groups. But the major human groups have separated from one another too recently and have mixed too much for such differences to exist.

Polymorphisms that occur at different frequencies around the world can, however, be used to sort people roughly into groups. One useful class of polymorphisms consists of the *Alus*, short pieces of DNA that are similar in sequence to one another. *Alus* replicate occasionally, and the resulting copy splices itself at random into a new position on the original chromosome or on another chromosome, usually in a location that has no effect on the functioning of nearby genes. Each insertion is a unique event. Once an *Alu* sequence inserts itself, it can remain in place for eons, getting passed from one person to his or her descendants. Therefore, if two people have the same *Alu* sequence at the same spot in their genome, they must be descended from a common ancestor who gave them that specific segment of DNA.

RESEARCHERS OFTEN USE SHORT PIECES of DNA called *Alu* polymorphisms to determine whether various populations are related to one another. *Alu*s have no known function, yet they copy and insert themselves at random throughout a person's genome. Because previously inserted *Alu*s do not excise themselves, *Alu* patterns can be used as yardsticks to estimate how close two people—and, on average, two populations—are genetically. For example, an *Alu* polymorphism on chromosome 1 occurs in roughly 95 percent of sub-Saharan Africans who have been sampled, 75 percent of Europeans and northern Africans, and 60 percent of Asians, whereas a different *Alu* polymorphism on chromosome 7 is carried by approximately 5 percent of sub-Saharan Africans, 50 percent of Europeans and northern Africans, and 50 percent of Asians. Some individuals carry both polymorphisms. No single polymorphism can, by itself, distinguish all the members of one major human group from all the members of another group, but by analyzing hundreds of these polymorphisms, scientists can group individuals sampled from different locations on the basis of their genetic profiles. *—M.J.B. and S.E.O.*



The Human Race

Given that people can be sorted broadly into groups using genetic data, do common notions of race correspond to underlying genetic differences among populations? In some cases they do, but often they do not. For instance, skin color or facial features--traits influenced by natural selection--are routinely used to divide people into races. But groups with similar physical characteristics as a result of selection can be quite different genetically. Individuals from sub-Saharan Africa and Australian Aborigines might have similar skin pigmentation (because of adapting to strong sun), but genetically they are quite dissimilar.

Another example of how difficult it is to categorize people involves populations in the U.S. Most people who describe themselves as African-Americans have relatively recent ancestors from West Africa, and West Africans generally have polymorphism frequencies that can be distinguished from those of Europeans, Asians, and Native Americans. The fraction of gene variations that African-Americans share with West Africans, however, is far from uniform, because over the centuries African-Americans have mixed extensively with groups originating from elsewhere in Africa and beyond.

Over the past several years, Mark D. Shriver of Pennsylvania State University and Rick A. Kittles of Howard University have defined a set of polymorphisms that they have used to estimate the fraction of a person's genes originating from each continental region. They found that the West African contribution to the genes of individual African-Americans averages about 80%, although it ranges from 20 to 100%. Mixing of groups is also apparent in many individuals who believe they have only European ancestors. According to Shriver's analyses, approximately 30% of Americans who consider themselves "white" have less than 90% European ancestry. Thus, self-reported ancestry is not necessarily a good predictor of the genetic composition of a large number of Americans. Accordingly, common notions of race do not always reflect a person's genetic background.

Membership Has Its Privileges

Understanding the relation between race and genetic variation has important practical implications. Several of the polymorphisms that differ in frequency from group to group have specific effects on health. The mutations responsible for sickle cell disease and some cases of cystic fibrosis, for instance, result from genetic changes that appear to have risen in frequency because they were protective against diseases prevalent in Africa and Europe, respectively. People who inherit one copy of the sickle cell polymorphism show some resistance to malaria; those with one copy of the cystic fibrosis trait may be less prone to the dehydration resulting from cholera. The symptoms of these diseases arise only in the unfortunate individuals who inherit two copies of the mutations.

Genetic variation also plays a role in individual susceptibility to one of the worst scourges of our age: AIDS. Some people have a small deletion in both their copies of a gene that encodes a particular cell-surface receptor called a chemokine receptor 5 (CCR5). As a result, these individuals fail to produce CCR5 receptors on the surface of their cells. Most strains of HIV-1, the virus that causes AIDS, bind to the CCR5 receptor to gain entry to cells, so people who lack CCR5 receptors are resistant to HIV-1 infection. This polymorphism in the CCR5 receptor gene is found almost exclusively in groups from northeastern Europe.

Several polymorphisms in CCR5 do not prevent infection but instead influence the rate at which HIV-1 infection leads to AIDS and death. Some of these polymorphisms have similar effects in different populations; others only alter the speed of disease progression in selected groups. One polymorphism, for example, is associated with delayed disease

progression in European-Americans but accelerated disease in African-Americans. Researchers can only study such population-specific effects--and use that knowledge to direct therapy--if they can sort people into groups.

In these examples--and others like them--a polymorphism has a relatively large effect in a given disease. If genetic screening were inexpensive and efficient, all individuals could be screened for all such disease-related gene variants. But genetic testing remains costly. Perhaps more significantly, genetic screening raises concerns about privacy and consent: some people might not want to know about genetic factors that could increase their risk of developing a particular disease. Until these issues are resolved further, self-reported ancestry will continue to be a potentially useful diagnostic tool for physicians.

Ancestry may also be relevant for some diseases that are widespread in particular populations. Most common diseases such as hypertension and diabetes are the cumulative results of polymorphisms in several genes, each of which has a small influence on its own. Recent research suggests that polymorphisms that have a particular effect on one group may have a different effect on another group. This kind of complexity would make it much more difficult to use detected polymorphisms as a guide to therapy. Until further studies are done on the genetic and environmental contributions to complex diseases, physicians may have to rely on information about an individual's ancestry to know how best to treat some diseases.

Race and Medicine

But the importance of group membership as it relates to health care has been especially controversial in recent years. Last January the U.S. Food and Drug Administration issues guidelines advocating the collection of race and ethnicity data in all clinical trials. Some investigators contend that the differences are so small and the historical abuses associated with categorizing people by race so extreme that group membership should play little if any role in genetic and medical studies. They assert that the FDA should abandon its recommendation and instead ask researchers conducting clinical trials to collect genomic data on each individual. Others suggest that only by using group memberships, including common definitions of race based on skin color, can we understand how genetic and environmental differences among groups contribute to disease. This debate will be settled only by further research on the validity of race as a scientific variable.

A set of articles in the March 20 issue of the New England Journal of Medicine debate both sides of the medical implications of race. The authors of one article – Richard S. Cooper of the Loyola Stritch School of Medicine, Jay S. Kaufman of the University of North Carolina at Chapel Hill, and Ryk Ward of the University of Oxford – argued that race is not an adequate criterion for physicians to use in choosing a particular drug for a given patient. They pointed out two findings of racial differences that are both now considered questionable: that a combination of certain blood vessel-dilating drugs was more effective in treating heart failure in people of African ancestry and that specific enzyme inhibitors (angiotensin converting enzyme, or ACE, inhibitors) have little efficacy in such individuals. In the second article, a group led by Neil Risch of Stanford

University countered that racial or ethnic groups can differ from one another genetically and that the differences can have medical importance. They cited a study showing the rate of complications from type 2 diabetes varies according to race, even after adjusting for such factors as disparity in education and income.

The intensity of these arguments reflects both scientific and social factors. Many biomedical studies have not rigorously defined group membership, relying instead on inferred relationships based on racial categories. The dispute over the importance of group membership also illustrates how strongly the perception of race is shaped by different social and political perspectives.

In cases where membership in a geographically or culturally defined group has been correlated with health-related genetic traits, knowing something about an individual's group membership could be important for a physician. And to the extent that human groups live in different environments or have different experiences that affect health, group membership could also reflect non-genetic factors that are medically relevant.

Regardless of the medical implications of the genetics of race, the research findings are inherently exciting. For hundreds of years, people have wondered where various human groups came from and how those groups are related to one another. They have speculated about why human populations have different physical appearances and about whether the biological differences between groups are more than skin deep. New genetic data and new methods of analysis are finally allowing us to approach these questions.
The result will be a much deeper understanding of both our biological nature and our human interconnectedness.

Lab 9: Does Race Exist I Pre-Lab Homework Questions #1-3

1. **Context**:
   a. Imagine you are at a party and someone pipes up, "But of course, <insert ethnic group> are < choose characteristic, smarter than, more athletic, more thrifty, more hard-working> than <insert other ethnic group>; it's in their genes." Using what you read in the article, how would you explain what is known about the utility of social definition of race, such as skin color or other physical features, and the underlying DNA evidence of how closely related two human beings are. (**1 point**)

   b. According to the authors, why would it be useful to define race from a biological standpoint? (**1 point**)

   Give one example used in the article where knowledge of racial differences has been used to benefit humans. (**1 point**)

   c. In the article, what are the reasons given to continue using self-reported ancestry as a diagnostic tool for physicians rather than using genetic testing? (**1 point**)

d. What is the function of *Alus* in the human genome, and what characteristic of transposons makes them perfect tracers for genealogy? (**1 point**)

e. Of the total genetic variability within the human species, approximately what percentage of variation can be found within a population, say in Europe? How much variation exists between populations, say Europeans as compared to Asians? (**1 point**)

2. **Method:** In your own words, define a polymorphism. Use two made-up DNA sequences to help support your definition. (**1 point**)

Explain how researchers could analyze polymorphisms such as *Alus* to determine ancestry. (**1 point**)

3. **Results:** Give two examples of how researchers have documented polymorphisms that result in effects on health. (**2 points**)

In-Lab Activity (Remainder of Pre-Lab):

The authors of the pre-lab reading make the point that there are racial differences in both susceptibility to diseases as well as in the effects of some therapies. A recent debate has ignited over an FDA (Food and Drug Administration) recommendation that researchers collect survey information about a test patient's race and ethnic identity. Some argue that outward signs of race are not adequate for distinguishing a person's genetic characteristics. Others suggest that we need to know this information to help understand how genetic and environmental differences among groups contribute to disease. During class today, your group will choose a position on the debate, and each person in the group will explain, in his/her own words, examples of evidence that supports your group's position.

Although you should discuss this as a group, your written work should be your own and will be graded as such by your GLA. Be sure to address the following four issues and respond to each with a 2-3 sentence response: (**5 points**)

Issues:

- How important are genetic differences in susceptibility to disease?

- What research findings support racial differences in susceptibility to disease or in response to medication?

- How could historical abuses associated with categorizing people by race effect your argument?

- What is the problem with using common definitions of race based on skin color when trying to infer a person's genetic heritage?

Objective 1 Experimental Design

Carry out your experiment and take notes below describing what you did and how your experiments helped you achieve Objective 1. Your descriptions should be clear and complete enough so that someone else can follow your logic and repeat your tests if necessary. **Although you should discuss this as a group, your written work should be your own and will be graded as such by your GLA.**

When you are done, please place used swabs in the containers provided. You may dump your tubes into the trash can.

Experimental Design (5 points)

☐ ***Objective***: What are your experiments designed to find?

☐ ***Prediction***: What sort of results do you expect to see?

☐ ***Procedure***: Instructions for extracting DNA are provided on the next page. Briefly indicate the purpose of each step so that you remember what you did for your final reports.

Rinse:

Swab:

Spin:

NaOH base:

Heat:

Add Tris:

Extracting Human Genomic DNA

1. Rinse your mouth out well with water from the fountain in the hallway.

2. Use a sharpie pen to label three 1.5 ml tubes with the numbers 1, 2, and 3. Also write your initials on the top cap as well as on the side of each tube. Mark tube #3 with: "1/10 dilution". Give a smaller 0.5 ml tube with your initials to your GLA.

3. Observe your GLA's lesson in using pipettors. Once you have mastered the technique, move to the next step.

4. Pipette 500 µl of water into the first tube using the **blue-topped pipettor** and a **blue tip**. Discard the tip in the container provided.

5. Using the swabs your GLA gives you, gently rub 20 times on each inner cheek.

6. Put swab in your first tube with the 500 µl of water. Twirl the swab vigorously. Remove swab and discard in beaker of bleach. Snap the tube shut.

7. Flick tube with finger. The solution should be cloudy.

8. Place tube in centrifuge with a partner's tube on the opposite side to balance. When all student tubes are in the centrifuge, your GLA will set the machine to spin for 2 min. When you pull out your tube, you will see a 'pellet' of cells at the bottom and the liquid 'supernatant' on top.

9. Pour the liquid supernatant from your tube into your second fresh tube. Recap your first tube with its pellet of cellular debris. Once the lab is complete, you will discard it into the bleach bucket along with your swab.

10. Bring your #2 tube to your GLA so s/he can add 200 µl of 50mM NaOH. Your GLA will carefully replace the cap tightly for you, as this is a strong base (strong bases can burn your skin and eyes.)

11. Place your tubes for 5 min in the 95˚C heat block.

12. While you are waiting for your DNA to be heated, take your third tube (the one that has your initials and says 1/10 dilution) and using the **yellow pipettor** and **yellow tips**, place 180µl of water into it. Discard the tip into the trash.

13. Bring your #2 tube from the 95°C bath carefully over to your GLA along with your #3 "1/10 dilution" tube with your initials on it. Your GLA will add 20 µl of 1 M Tris (Tris is a buffer that will neutralize the NaOH) to your #2 tube, cap and invert to mix, and then pipette 20µl from tube #2 to tube #3 "1/10 dilution".

14. Give your GLA your tube #3, 1/10 dilution to add to the PCR mix.

Does Race Exist?
II

Lab 10

Variations in Populations

Lab 10 Does Race Exist II
Pre-Lab Homework Questions #1-3

You learned in last week's lab that *Alu* transposable elements are useful as DNA markers for human population studies. In fact, there are over 5000-7000 new *Alu* insertions into the human genome since our ancestors split from a common ancestor with chimpanzees. Therefore, some humans may not have a specific insertion. In other cases, some of the new insertions may have accumulated mutations that result in polymorphisms that can be used as markers to identify ancestry. Researchers David H. Kass, Nicole Jamison, Melanie M. Mayberry, and Eillen Tecle (2007) identified a single *Alu* element that exists in human populations as 4 different polymorphisms. These polymorphisms are changes in the DNA sequence that must have occurred accidentally in different human populations since the time they migrated to different global regions. They sampled populations from Africa, South America, Europe, and Asia for these polymorphisms and uncovered variations in the presence of the different alleles that could be used to elucidate ancestry. The PCR primers that you used last week amplified those *Alu* transposons. This week your restriction enzyme digestion of these *Alu* elements will identify which you inherited.

1. **Context**:

The nucleotide sequence of the *Alu* element that you amplified last week in lab can be found in human populations with one of the following four polymorphisms (YB8, S1, S2, and L):

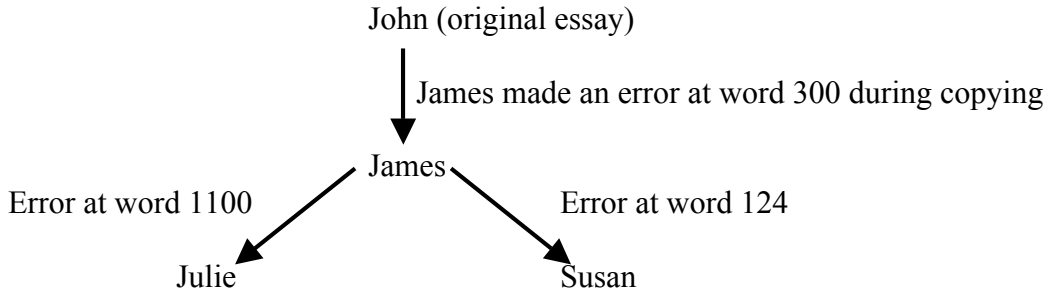
```
     62         70     73
YB8 _GGCCGGGCGCGGTGGCTCACGCCTGTAATCCCAGCACTTTGGGAGGCCGAGGCGGGTGGATCATGAGGTCA
S1   T . . . . .GGCGCC. . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . .
S2   T . . . . ..T . . . . A . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . .
L    T . . . . ..T . . . . A . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . .

                                                             172
YB8 GGAGATCGAGACCATCCTGGCTAACAAGGTGAAACCCCGTCTCTACTAAAAATACAAAAAATTAGCCGGGCGC
S1   . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . A. . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . .
S2   . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . .
L    . . . . . . . . . . .  . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . .

                   217                      236-237             250       257
YB8 GGTGGCGGGCGCCTGTAGTCCCAGCTACTCGGGAGGCTGAGGCAGGAGAATGGCGTGAACCCGGGAAGCGG
S1   . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . TA . . . . . . . . . . . . .G . . . . . . .G . . . . . . . . . . . . . . . . . . . . . .
S2   . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . TA. . . . . . . . . . . . . G . . . . . . .G. . . . . . . . . . . . . . . . . . . . . .
L    . . . . . . . . . . . A . . . . . . . . . . . . . . . . . . . . .TA. . . . . . . . . . . . . G . . . . . . .G. . . . . . . . . . . . . . . . . . . . .

                                298                                         338
YB8 AGCTTGCAGTGAGCCGAGATTGCGCCACTGCAGTCCGCAGTCCGGCCTGGGCGACAGAGCGAGACTCCGTCTC
S1   . . . . . . . . . . . . . . . . . . . . . C. . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . .
S2   . . . . . . . . . . . . . . . . . . . . . C. . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . .  A . . . . . . . . . . . .
L    . . . . . . . . . . .  . . . . . . . . . .C . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . . A . . . . . . . . . . . .
```

a. Assuming that these polymorphisms occurred following random genetic changes (mutations to the original sequence (YB8). How many changes occurred in each?

Polymorphism	# nucleotide changes	(3 points)
YB8 » S1		
YB8 » S2		

YB8 » L

b. The changes in the *Alu* element above can be thought of with a plagiarism analogy: Imagine a professor gets 4 nearly identical responses to an in-class writing assignment, and he wishes to figure out what evidence in the writing may uncover the story of what occurred. One student, John, submits a nearly perfect paper. The second student, James, submitted a nearly identical paper to John's but he has a missed word, number 300, that renders the text meaningless at that point. The third student, Julie, submitted a paper identical to James' with the same mistake at word 300, but she also has a new mistake at word 1100. Finally, the fourth student, Susan, has the same identical error at word 300, but she has a new mistake at word 124. In this scenario, the professor could try to determine the order of the plagiarism. Obviously, the papers were all copied in some way, and one would have to assume that the introduction of errors occurred during the copying. So, the professor can start to chart out (as shown below) what might have occurred before he confronts the students:



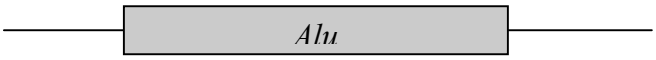
The professor could then see if this pattern was supported by their seating arrangements.

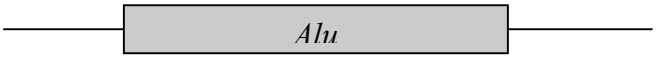
This is exactly how scientists compare DNA sequence changes in these polymorphisms. Random errors must have occurred in the copying mechanism during DNA replication, and the descendents of those humans would have inherited the errors. One can then use the errors like the professor did to chart the most logical series of steps. Using the YB8, S1, S2, and L sequences construct a map like the one above that charts the changes in the evolution of the original YB8 element in human populations. **Be sure to mark your chart with each of the four elements and describe each mutational change. (4 points)**

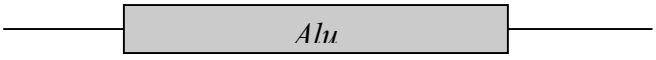
2. **Method:** The nucleotide changes in the four different polymorphisms for this *Alu* allele can be distinguished by DNA sequencing. A fast but expensive way to determine which two alleles you have is to send your PCR reactions off to be sequenced. However, a cheaper and just as effective way to determine which polymorphism you have uses two other techniques, restriction enzyme digestion and gel electrophoresis. These can be done right here in your laboratory. Remember that restriction enzymes are able to cut DNA into pieces at very specific sequences. For example, the restriction enzyme *KasI* recognizes the sequence GGCGCC underlined in the sequence from homework question #1. Using the sequence from question #1, fill in the following table to indicate how many times the enzyme would cut each polymorphism allele YB8, S1, S2, and L, and how many pieces would be generated. (**4 points**)

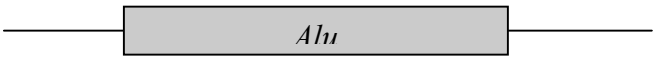
Polymorphism	# Times Enzyme would cut	# Pieces Made
YB8		
S1		
S2		
L		

3. **Results:** Using the following figures of the YB8, S1, S2, and L *Alu* polymorphisms that were amplified by PCR last week, mark the location of the KasI sites with a horizontal line and indicate the nucleotide number, so you can visualize your answers from above. (**4 points**)

YB8 

S1 

S2 

L 

Restriction Enzyme Experimental Design

You will carry out your restriction digest with help from your GLAs. Your objective for today is NOT to come up with the experiment but rather to determine a **prediction** about what your results will show next week. You will make this prediction based on Table 1 from the Kass *et al*. 2007 paper (below). **Although you should discuss the table with your group, your written work should be your own and will be graded as such by your GLA.**

Experimental Design (5 points)

☐ ***Objective***: What are your experiments designed to find?

Table 1

Polymorphism *Alu* allele frequencies among various analyzed human populations

Group	- *Alu*	+ *Alu* YB8	+ *Alu* S1	+ *Alu* S2	+ *Alu* L
African Americans	17.6%	82.4%	41.2%	23.5%	17.6%
Asian	73.7%	26.3%	10.5%	0	15.8%
European Caucasian	55.9%	44.1%	0	26.5%	17.6%
South American	52.9%	47.1%	0	14.7%	32.4%
Indo-Pakistani	50.0%	50.0%	11.1%	0	38.9%
Kenyan	15.4%	84.6%	65.4%	11.5%	7.7%
Chinese	58.3%	41.7%	16.7%	0	25.0%
Druze	100%	0	0	0	0
Nigerian	20.0%	80.0%	40.0%	20.0%	20.0%
Melanesian	75%	25%	25%	0	0

☐ ***Prediction***: You can see from Table 1 that certain polymorphisms are more predominant depending on what part of the world the population was sampled. Using what you know about your family's ancestry, what prediction would you make about which polymorphism you probably inherited. (If you don't know your family's ancestry, choose one group, and describe which polymorphisms would be most likely and least likely to be found in the sample.)

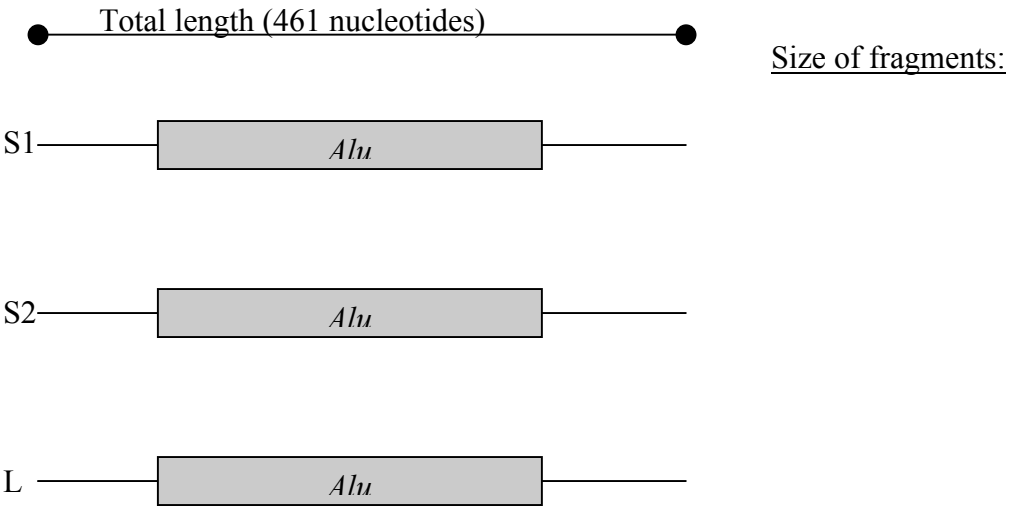
Can you reconcile what you mapped out as an answer to pre-lab question 1b with the data from Table 1?

Every human is diploid, so you have two homologous copies of every chromosome in your body. Thus, you have two copies of the *Alu* allele.

Remember the four *Alu* alleles are as follows: - (no *Alu* allele), S1, S2, or L.

List all the possible genotypes that could exist in a population:

Go back to your answers to question 3 from the pre-lab, and for each allele (S1, S2, and L), indicate the sizes of the DNA fragments that would be generated from restriction enzyme digestion of each allele.



Using these lengths and considering a person with the L allele on one chromosome and the S1 allele on the other chromosome (L/S1 genotype), indicate the number and size of all fragments created by *KasI* digestion of their PCR sample:

Procedure: ***KasI Restriction Enzyme Digestion of amplified DNA sample***

1. Use a sharpie pen to label one 1.5 ml tube with your initials and "Digest," and give the tube to your GLA (who will place it into an ice bucket.)

2. Work on your experimental designs for this week while your GLA makes up enough restriction digestion mix for the whole class.

3. When your GLA is ready, s/he will announce that groups from each table should come to the front of the room to get your 0.5µl PCR reaction tube from the rack.

4. When it is your turn, watch your GLA extract 30µl of your PCR reaction and add it to your 1.5µl tube with your initials and "Digest" on it.

5. Flick tube with finger to mix the contents. Add your tube to the 37°C heating block.

6. After one hour, remove your tube.

7. Place your tube in the rack at the front of the room.

8. Your GLA will add 7µl of 6X Loading Dye to your restriction digest and will place the tubes back into the refrigerator for next week.

Kass, D. H., Jamison, N., Mayberry, M. M., & Tecle, E. (2007). Identification of a unique Alu-based polymorphism and its use in human population studies. *Gene, 390*(1-2), 146-152.

Does Race Exist?
III

Lab 11

Building Trees

Lab 11 Does Race Exist III
Gel Electrophoresis

During the past two labs, you extracted your own DNA and performed restriction enzyme digestion of your YB8 *Alu* elements. This week you will use gel electrophoresis technique to separate your restriction fragment pieces by size and then visualize them using a DNA dye. Your objectives for today are to understand your results and to prepare to write an explanatory document for your family.

In the predictions from the experimental design from last week's lab, you should have:

1) Used your knowledge of your ancestry and Table 1 on page 10-5 to make a prediction about whether or not you would have the YB8 *Alu* allele, and if you did, which of the 3 polymorphisms (S1, S2, or L) you would most likely have.

2) Created a list of all the possible genotype combinations of these 4 alleles (-, S1, S2, and L).

3) Determined the sizes of the restriction fragment pieces that would be generated for each allele (S1, S2, and L).

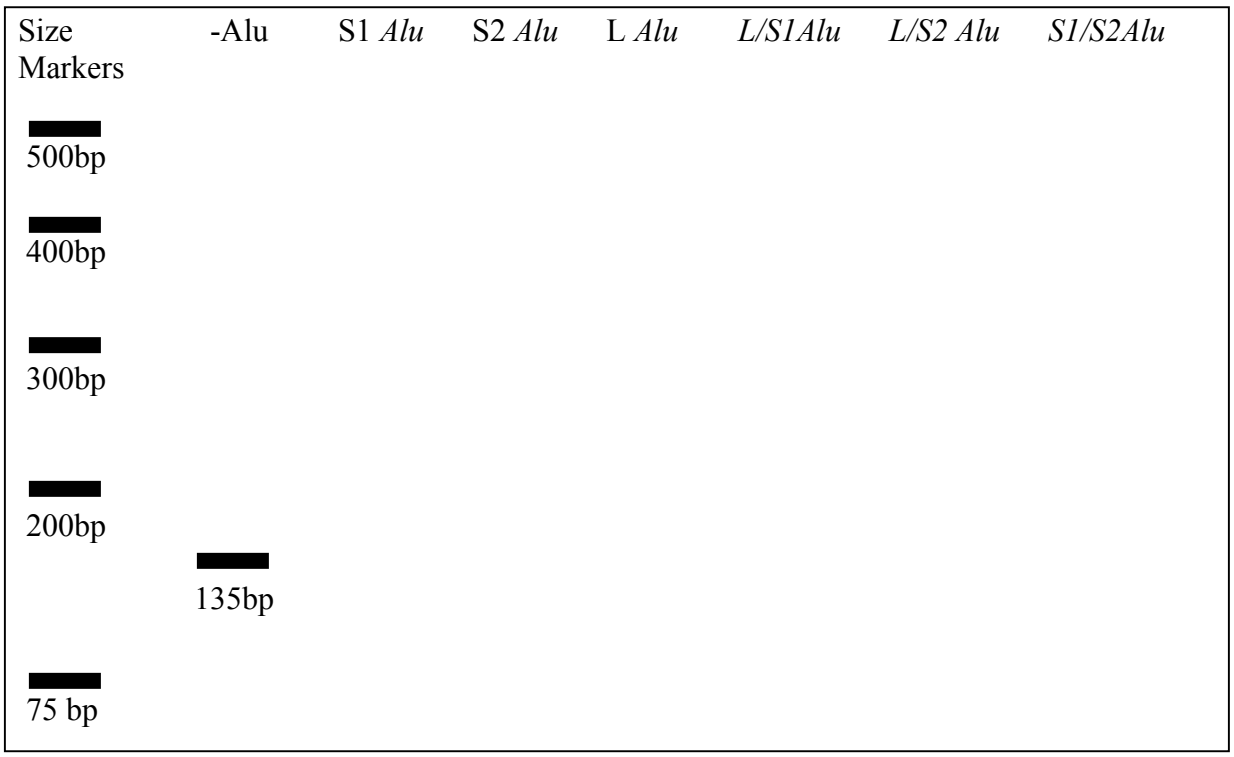
Watch your GLA load your samples onto an agarose gel. While the gel is running for an hour, you will complete the drawing on the next page with your group. You can that use that drawing to help interpret the results of the gel electrophoresis.

Gel Electrophoresis (*electro* as in electricity + *phoresy*, movement) is the movement of charged molecules in an electric field. In this case, your DNA fragments are large molecules that will be loaded into small wells cut into a sponge-like agarose gel. An electric current will be applied, and the negatively-charged DNA will move through the spongy matrix of the gel to get to the positive charge on the opposite end of the gel. Large molecules move slower than small ones so that molecules of different sizes appear as separate bands; the smaller DNA fragments move farther through the gel toward the opposite positive end.

Gel Staining: Several different dyes can be used to bind to the DNA in the gel so that the gel can be visualized after it is photographed. Your GLA will demonstrate this for you and will photograph your gel.

While your gel is running, you will use the results of your predictions from above to fill out the following gel with the different sized fragments you would expect to see for different genotypes. The PCR fragment generated using just the ends of the sequence

without the *Alu* element is 135 bp as is shown below. Fill in the expected sizes for the remaining elements.



☐ ***Evidence***: Describe the results of your experiments. How many bands did you find and which pattern do they match?

☐ ***Explanation***: Explain why you think you saw these results. Are they consistent with your prediction based on the evidence from Table 1 on ancestry?

Rubric for FIRST draft of *Does Race Exist* summary essay

Assignment:
This is the first draft version of your *Does Race Exist* summary essay (**worth up to 10 points**). When commenting on this draft, your GLA will focus on **large-scale** issues related to your summary. This feedback and a subsequent peer discussion will allow you to revise your draft into a well-written final version.

Format of essay: Your paper should be approximately 3-5 pages in length, including figures and tables. Each figure and table should not take up more than half a page in length. **It does not need to follow a standard format for lab reports; you may write it as you would a general essay.**

Your draft summary paper must be **typed and double-spaced, font size 12 and font style Times New Roman**. **Hand-written assignments will not be accepted or graded.**

Keep in mind this paper will be longer than the 2-page Carb Cutter article you wrote earlier in the semester.

Imagine you are writing to your family to inform them about what your results indicate about your ancestry. Your paper should contain the components listed below.

- ✓ Include a title page with your name, lab section (lab – time, day), GLA name, names of group mates, and the academic term. **Up to 0.5 point**

- ✓ Important background information keeping in mind that this paper should be interesting and relevant to a non-scientist audience. Provide background information to your family member so that s/he understands why your study is important. For instance, what information about transposons and polymorphisms does your family need in order to understand and be interested in your experiment? Aside from ancestry, what is the benefit to mankind of defining race? You will need to balance scientific terminology with text that does not read like a list of glossary-defined definitions. **Up to 2 points**

- ✓ Introduce your prediction about which polymorphism you probably inherited by revising what you wrote for the pre-lab assignments about the underlying DNA evidence of how closely related two human beings are. How useful is the YB8 *Alu* element for predicting ancestry? What was your prediction for your genotype? Explain your reasoning. **Up to 2 points**

- ✓ How did the techniques you used (DNA extraction, PCR, gel electrophoresis) allow you compare *Alu* polymorphisms? **Up to 1 points**

- ✓ What were the results of your experiment? A photograph of your gel must be included and should have clear labels. Explain how you can infer your genotype from the gel. How did your genotype compared to predicted results? **Up to 2 points**

- ✓ Describe what you can conclude from the data you collected. Was your prediction supported? How much confidence can you put in this method? Where does the data indicate your family is from? **Up to 2 points**

- ✓ Essay follows correct conventions for grammar and spelling. Ideas are organized in a logical format. Paper is clear and concise. **Up to 0.5 points**

Rubric for FINAL draft of *Does Race Exist* summary essay

Assignment:
You have designed and completed an experiment that determined your genotype for the YB8 *Alu* transposable element. Now write an informative, scientific essay of your journey through this lab. You can earn up to **30 points** on this assignment. Remember, if you successfully completed a rough draft and received feedback from your GLA, be sure to hand that draft in along with your final version. **Failure to resubmit this graded rough draft will result in a 10-point deduction from your final grade on the summary. Also, if you do resubmit the draft but fail to make any revisions, a 10-point deduction from your final grade on the summary will be given.**

Format of essay: Your paper should be approximately 3-5 pages in length, including figures and tables. Each figure and table should not take up more than half a page in length. **It does not need to follow a standard format for lab reports; you may write it as you would a general essay.**

Your final summary paper must be **typed and double-spaced, font size 12 and font style Times New Roman**. **Hand-written assignments will not be accepted or graded.**

Keep in mind this paper will be longer than the 2-page Carb Cutter article you wrote earlier in the semester.

Imagine you are writing to your family to inform them about what your results indicate about your ancestry. Your paper should contain the components listed below.

- ✓ Include a title page with your name, lab section (lab – time, day), GLA name, names of group mates, and the academic term. **Up to 1 point**

- ✓ Keeping in mind that the reader of this essay is a family member without scientific background, provide background information to introduce why your essay is important enough to write about. For instance, what information about transposons and polymorphisms does your family need in order to understand and be interested in your experiment? Aside from ancestry, what is the benefit to mankind of defining race? What other information is necessary for your family to understand the importance of your study? You will need to balance the introduction of scientific terminology with text that does not read like a list of glossary-defined definitions. **Up to 4 points**

- ✓ Introduce your prediction about which polymorphism you probably inherited by revising what you wrote for the pre-lab assignments about the underlying DNA evidence of how closely related two human beings are. How useful is the YB8 *Alu* element for predicting ancestry? What was your prediction for your genotype? Explain your reasoning. **Up to 6 points**

- ✓ How did the techniques you used (DNA extraction, PCR, gel electrophoresis) allow you compare *Alu* polymorphisms? **Up to 4 points**

- ✓ What were the results of your experiment? A photograph of your gel must be included and should have clear labels. Explain how you can infer your genotype from the gel. How did your genotype compared to predicted results? **Up to 6 points**

- ✓ Describe what you can conclude from the data you collected. Was your prediction supported? How much confidence can you put in this method? Where does the data indicate your family is from? **Up to 6 points**

- ✓ Essay follows correct conventions for grammar and spelling. Ideas are organized in a logical format. Paper is clear and concise. **Up to 3 points**