Part 1: SNP Frequencies

1. **Check one box** in each row under the ancestral population you believe the corresponding face on the left is depicting.

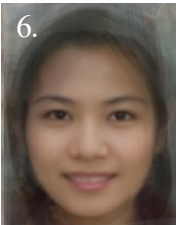**TABLE 1**
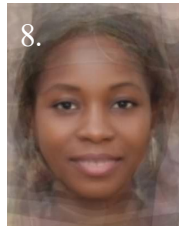
Picture	African	American	East Asian	European	South Asian
1	☐	☐	☐	☐	☐
2	☐	☐	☐	☐	☐
3	☐	☐	☐	☐	☐
4	☐	☐	☐	☐	☐
5	☐	☐	☐	☐	☐
6	☐	☐	☐	☐	☐
7	☐	☐	☐	☐	☐
8	☐	☐	☐	☐	☐
9	☐	☐	☐	☐	☐
10	☐	☐	☐	☐	☐
11	☐	☐	☐	☐	☐

*All populations around the world are grouped into these 5 ancestral populations. For example, American consists of North America and South America.

TABLE 2 contains a list of known SNPs in the human genome. Each letter along the top is a representative of one of the five major populations from the previous page. For example, B & J might be representative of the Asian population.

TABLE 2: Skin Pigmentation SNP Chart

Gene	Variant	A	B	C	D	E	F	G	H	I	J	K
IRF4	rs12203592	C	C	C	C	C	C	C	C	C	C	C
OCA2	rs12913832	A	A	A	A	A	A	G	A	G	A	A
SLC45A2	rs16891982	C	C	G	C	C	C	G	G	G	C	C
HERC2	rs1667392	G	G	G	G	G	G	C	C	C	G	G
MC1R	rs885479	G	G	G	A	G	G	G	G	G	G	G
GRM5	rs7118677	T	G	G	T	T	G	G	G	G	T	T
ATP8B4	rs8033655	G	G	A	G	G	G	G	G	G	A	A

1. Pick a SNP column (A-K) from **TABLE 2**. Write the letter of the SNP set of your choice into the blank at the top of column 3 of **TABLE 3**.

    A. Go to ensembl.org. Try grch37.ensembl.org if other link does not work.

    B. Type the variant name into the search bar and click enter or search.
       Ex. rs1426654

    D. Click on the result that has variant name your entered and (Human Variant)

    E. Under "Explore this variant" click on "Population genetics."

    F. The pie charts have the allele frequencies for all and then broken down by major populations.

    G. Record the allele frequencies **FOR YOUR ALLELE** (based on the SNP Set you chose) for each population in the tables for your allele in TABLE 3.

TABLE 3: Population Frequency

GENE	Variant	SNP Set	African	American	East Asian	European	South Asian
IRF4	rs12203592	C	99%	93%	100%	88%	99%
OCA2	rs12913832						
SLC45A2	rs16891982						
HERC2	rs1667392						
MC1R	rs885479						
GRM5	rs7118677						
ATP8B4	rs8033655						

2. Using the frequencies given in **TABLE 3**, rank from 1 to 5 the likelihood that your SNP Set is part of that population. Using 1 for the most likely population (closest to 100%) and 5 for the least likely population (closest to 0%). When 2 populations have the same frequencies, give them the same ranking (see example).

TABLE 4

GENE	Variant	SNP Set	African	American	East Asian	European	South Asian
IRF4	rs12203592	C	2	3	1	4	2
OCA2	rs12913832						
SLC45A2	rs16891982						
HERC2	rs1667392						
MC1R	rs885479						
GRM5	rs7118677						
ATP8B4	rs8033655						
		Total					

3. Total each Population column in Table 4. The column with LOWEST number represents your SNP set's most probable population based on the data given.

4.   What population is mostly likely the ancestral population for the SNP Sets you chose?

SNP Set _____

Population_____

5.   Do you feel that you have enough information to accurately determine the population and face for your SNP sets? Why or Why not. Refer to other groups and their conclusions as well.

6.   How many SNPs do you think would give you enough information to accurately determine the population and face for your SNP sets?

7. What features did you use to identify the ancestral populations of the faces in question 1?

8. See your instructor for the key. Were you able to correctly identify the ancestral population of your average faces based on the 8 SNPs? If not, why the discrepancy?

9. The concept of race is sometimes used to categorize people, for example on the US census. All of these genes and variants are part of complex system that codes for skin color. Do you think the genes underlying skin color can be used to categorize people by race? Explain.

10. Can these same genes be used to determine one's ethnicity? Explain.

11. Can these same genes be used to determine one's ancestry? Explain.

12. What is the difference between race, ancestry and ethnicity?

13. What are some potential issues about assuming someone's ancestry based on how they look?

[Lack of] Human Diversity
Using SNPs to help Determine Ancestry

Part 2: Phylogenetic Tree

Tree #1

Phylogenetic trees group together those that are most similar. Now that you know the ancestral population of the faces from question 1, sketch a tree below using those 11 faces to show your best guess at who is more closely related to whom. Label your tree with both face number and ancestral population from the key.

If you cannot upload a pic of your tree to this box, attach a file containing the picture of it with your submission in Canvas.

[Lack of] Human Diversity
Using SNPs to help Determine Ancestry

Tree #2

The SNPs from part 1 have been converted into fasta format for you on the right. You will be using these SNPs to create a phylogenetic tree. The SNPs that vary the least, will be closer to each other in the tree. The SNPs that vary the most, will be further away from each other.

A. Copy the sequences to the right.
B. Open www.phylogeny.fr in a browser.
   DO NOT CLICK ON LINK IF USING A WEB BROWSER!!!
C. Under Phylogeny Analysis, select "One Click" from the homepage.
D. Paste sequences into the box.
E. Click "submit"
F. Save the image of the tree to your computer and then upload it to the box below.

If you cannot upload a pic of your tree to this box, attach a file containing the picture of it with your submission in Canvas.

SNPs in fasta format

>African 1
CACCCGGCGCTCAGT

>African 2
CACCCGGCGCGTAGT

>American
AAGCCGGCGTGCGAT

>East Asian 1
CACCCGACGCTCAGT

>East Asian 2
CACCCGGCGTGCGG T

>European 1
CACCCGCGATGCAGT

>European 2
CGGCCCGCATGCAGT

>European 3
CGGCCCGCATGCAGT

>European 4
AGGCCCGCATGCAGT

>South Asian 1
CACCCGGCGTTCAAT

>South Asian 2
CACCCGGCGTTCAAT

1. Compare Tree #1 to Tree #2. Are they the same?

2. What surprises you most when comparing the trees?

3. Which tree do you think is more accurate? Explain.

4. How many SNPs do you think it would take to determine ancestry?

5. Can a phylogenetic tree be used to determine someone's race or ethnicity? Explain.