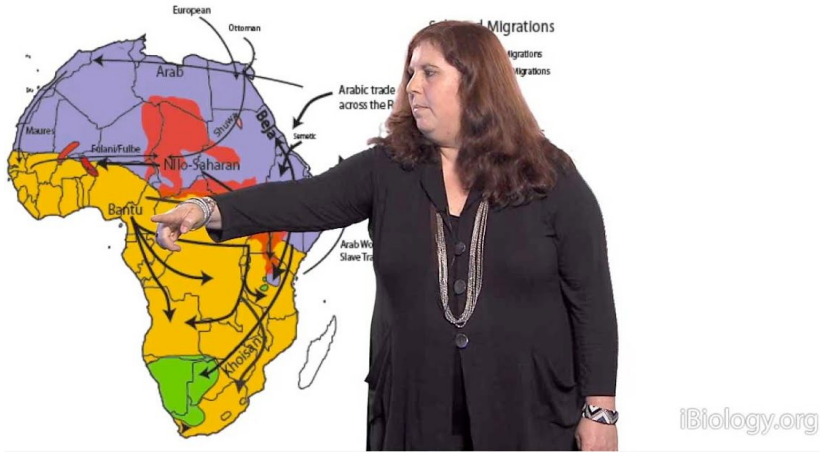
Session 8: Human Evolution

Overview:
Sarah Tishkoff studies the evolution and migration of human populations. She describes work from her lab looking at genotypic and phenotypic variation among populations speaking different African languages. Her results provide genetic insights into the history of African ethnic groups and African Americans. Tishkoff's last talk focuses on the importance of natural selection in human evolution. For instance, mutations that are associated with disease in modern African populations may have been advantageous to the same populations at some point in their past.

First video (Review from Session 2):
Title: African Genomics: Human Evolution
Speaker: Sarah Tishkoff

Second video:
Title: African Genomics: African Population History
Speaker: Sarah Tishkoff



Questions for Part 2:

1. Select all the answers that apply. DNA polymorphisms among African populations correlate with
    a. skin color.
    b. geography.
    c. weather patterns.
    d. spoken language.
    e. None of the above.

2. Which of the following observations could be explained by a recent migration event?
    a. Populations that are genetically distinct.
    b. Populations that are geographically close but genetically distinct.
    c. Populations that are geographically far and genetically distinct.
    d. Populations that are geographically close but genetically identical.
    e. None of the above.

For questions 3-5, determine if the statement is true or false. If the statement is false, change one word to make it true.

3. Some populations from Eastern Africa are genetically similar to populations in Western Africa due to migration events.

4. DNA analysis has shown that Europeans have the highest number of polymorphism in a population.

5. Vitamin D production was a possible driver for the generation of skin color polymorphisms in African populations.

6. Karen is a doctor in a region of the U.S. with a high population of African Americans and she is trying to decide if a recently approved drug would benefit her population of patients.
    a. Which factors should Karen consider before administering this drug to her population of patients? Briefly explain.

b. Studies have shown that the African Americans can have African and European polymorphisms at different regions within the same chromosome. How might that affect drug therapy?

7. Why do non-African populations exhibit less genetic diversity than African populations? Briefly explain.

8. (Think, Pair and Share) In this video, Dr. Tishkoff shows some of the physical, logistical and ethical challenges that scientists face when they perform their experiments in places like Africa. Pair with one of your classmates and discuss these challenges. Are all of these challenges overcome when scientists perform research in the U.S.? Briefly explain.

Third video:
Title: African Genomics: Natural Selection
Speaker: Sarah Tishkoff

Questions for Part 3:

1. Which of the following phenotypes would increase fitness of individuals in a population of nocturnal moths that live in a dark forest?
    a. Light-wing color.
    b. Luminescent-wing color.
    c. Dark-wing color.
    d. None of the above.

2. Choose all of the answers that apply. The persistence of lactase expression in humans is an example of
    a. Natural selection.
    b. Genetic drift.
    c. Convergent evolution.
    d. Speciation.
    e. Bottleneck effect.

3. Which of the following traits would not improve fitness in the following populations?
    a. Retaining iodine in an environment with relatively low levels of iodine.
    b. Light-skin color in areas with low sun exposure.
    c. Digestion of algae in a society that primarily eats sushi.
    d. Retention of water in the desert.
    e. None of the above.

For questions 4-6, determine if the statement is true or false. If the statement is false, change one word to make it true.

4. A phenotype could be a consequence of strong selective pressures acting on distant genes.

5. Exposure to disease and short life span could have lead to short stature in Pygmies populations.

6. Through DNA analysis scientists can find evidence of natural selection and genetic drift in the human population.

7. Scientists use DNA analysis in order to study the evolution of different traits in humans.
    a. Scientists have identified polymorphisms on specific genes that have been selected through evolution. Provide an example of a polymorphism that has been selected through evolution? What was the selective pressure driving selection of the trait?

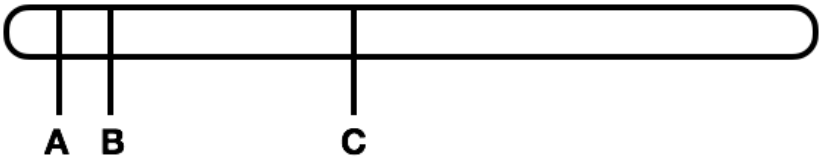
    b. Is it easier or harder to study the evolution of polygenic or monogenic traits? Why? Include examples of polygenic and monogenic phenotypes.

    c. Blue eye color is a polygenic trait in humans. Individuals with blue eyes tend to be more sensitive to sun exposure and more likely to experience sun-related damage to the retina than individuals with dark colored eyes. Provide an explanation for how the blue eye color may have been evolutionarily selected.

8. In the magic forest there are two population of unicorns, shorthorn and longhorn unicorns. Surprisingly the longhorn unicorns are faster runners than shorthorn unicorns. The faster runner unicorns have better fitness as they are better at escaping from predators. After a few generations, all of the unicorns evolved to have longhorns and to be fast runners. What scientific evidence would you gather to investigate if these two genes co-evolved? Briefly explain.

9. Why do some scientists think that the high incidence of type 2 diabetes in modern humans may be due to pressures that selected for genes that may have been advantageous in the past? Briefly explain.

10. Shown below is the diagram of a representative human chromosome, with polymorphisms in genes A, B, and C. Polymorphism A enhances the immune system, polymorphism B results in big feet, and polymorphism C results in baldness. (Assume these traits are each due to a single gene.)



    a. If there is a strong selective pressure towards variant A, which polymorphism (B or C) is most likely to have co-evolved with A? Briefly explain.

    b. Given the information shown above, can you predict the phenotype of this population after several generations? Briefly explain.

Answers for Session 8:

Questions for Part 2:

1. Select all the answers that apply. DNA polymorphisms among African populations correlate with
    - **a. skin color.**
    - **b. geography.**
    - c. weather patterns.
    - **d. spoken language.**
    - e. None of the above.

2. Which of the following observations could be explained by a recent migration event?
    - a. Populations that are genetically distinct.
    - **b. Populations that are geographically close but genetically distinct.**
    - c. Populations that are geographically far and genetically distinct.
    - d. Populations that are geographically close but genetically identical.
    - e. None of the above.

For questions 3-5, determine if the statement is true or false. If the statement is false, change one word to make it true.

3. Some populations from Eastern Africa are genetically similar to populations in Western Africa due to migration events.
   True.

4. DNA analysis has shown that Europeans have the highest number of polymorphism in a population.
   False; ~~Europeans~~ Africans

5. Vitamin D production was a possible driver for the generation of skin color polymorphisms in African populations.
   True.

6. Karen is a doctor in a region of the U.S. with a high population of African Americans and she is trying to decide if a recently approved drug would benefit her population of patients.
    a. Which factors should Karen consider before administering this drug to her population of patients? Briefly explain.
    Yes and no, it would depend how the clinical trial was conducted. Some clinical trials do not have enough representation from non-white ethnicities. Given the genetic variation among human populations, a drug that works well for most white individuals may not be effective in most non-white individuals.

    b. Studies have shown that the African Americans can have African and European polymorphisms at different regions within the same chromosome. How might that affect drug therapy?
    If a drug is known to have different effects in individuals of African or European ancestry, ethnic self-identification should not be the only determinant in deciding if a drug should be used in a patient. It would be necessary to look at the specific polymorphism of the gene involved in drug metabolism (or the region of the chromosome containing that gene) to determine if the drug would be beneficial for a particular patient. This is an example of personalized medicine.

7. Why do non-African populations exhibit less genetic diversity than African populations? Briefly explain.
    a. **Humans originated in Africa. A subset of the total human population migrated out of Africa, resulting in a genetic bottleneck and a decrease in the genetic diversity of the population that left.**
    b. Founder effect. As humans moved further away from Africa, each new population was founded by a small group of humans. All subsequent members of the population would be descendants of the founders.

8. (Think, Pair and Share) In this video, Dr. Tishkoff shows some of the physical, logistical and ethical challenges that scientists face when they perform their experiments in places like Africa. Pair with one of your classmates and discuss these challenges. Are all of these challenges overcome when scientists perform research in the U.S.? Briefly explain.
    Challenges in Africa:
    - **Lack of scientific infrastructure (reliable electricity, equipment, etc).**
    - **Lack of transportation infrastructure (roads, bridges, etc).**
    - **Need to explain their science and experiments to the population (non-scientists) and likely a need for translators.**
    - **Need to provide information about the benefits of their research.**
    - **Need to provide information about the results.**

Challenges in USA:

- **Infrastructure is better in the US, however, experiments and results still need to be explained to non-scientifically trained populations**

Questions for Part 3:

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    d. Retention of water in the desert.
    e. None of the above.

For questions 4-6, determine if the statement is true or false. If the statement is false, change one word to make it true.

4. A phenotype could be a consequence of strong selective pressures acting on distant genes.
    False; ~~distant~~ adjacent

5. Exposure to disease and short life span could have lead to short stature in Pygmies populations.
    True.

6. Through DNA analysis scientists can find evidence of natural selection and genetic drift in the human population.
    True.

7. Scientists use DNA analysis in order to study the evolution of different traits in humans.
    a. Scientists have identified polymorphisms on specific genes that have been selected through evolution. Provide an example of a polymorphism that has been selected through evolution? What was the selective pressure driving selection of the trait?
        i. **MC1R polymorphisms lead to light skin (or coat) color. For individuals living in regions with low sun exposure, light skin allows for the absorption of more light and production of more vitamin D. Thus the ability to produce vitamin D could have driven selection for light skin. For animals living in an area where light coat color provides better camouflage from predators, this could have been the driver for the light coat MC1R polymorphism.**
        ii. **Persistence of lactase activity in humans. A pastoralist or cattle-based economy in which milk (lactose) was a food staple could have been the driver for selection of this trait.**

    b. Is it easier or harder to study the evolution of polygenic or monogenic traits? Why? Include examples of polygenic and monogenic phenotypes.
    It's harder to study the evolution of polygenic traits (e.g. stature, intelligence, coat color, skin color, etc) than monogenic traits (e.g. lactase persistence). For polygenic traits, natural selection needs to act on multiple genes to confer a specific phenotype.

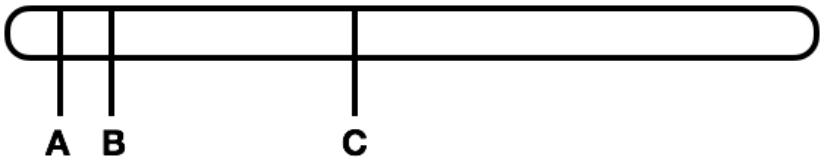
    c. Blue eye color is a polygenic trait in humans. Individuals with blue eyes tend to be more sensitive to sun exposure and more likely to experience sun-related damage to the retina than individuals with dark colored eyes. Provide an explanation for how the blue eye color may have been evolutionarily selected.
    Natural selection must have acted to select for an advantage provided by blue eye color. Sometimes, genes are involved in multiple pathways and it is possible that genes involved in determining blue eye color also impact another phenotype that provides an advantage. Alternatively, blue eye color could be determined by an allele that is closely linked to the gene encoding another trait that provided an evolutionary advantage and blue eye color co-evolved with that trait.

8. In the magic forest there are two population of unicorns, shorthorn and longhorn unicorns. Surprisingly the longhorn unicorns are faster runners than shorthorn unicorns. The faster runner unicorns have better fitness as they are better at escaping from predators. After a few generations, all of the unicorns evolved to have longhorns and to be fast runners. What scientific evidence would you gather to investigate if these two genes co-evolved? Briefly explain.
    Genes are more likely to co-evolve if they are in close proximity. To investigate whether or not the fast runner and long horned genes co-evolved, identify the

location of the genes for horn length and running speed. Identify a polymorphism that determines the longhorn phenotype and a polymorphism that determines the ability to run fast. Determine if these phenotypes are monogenic. If these traits are monogenic, and they are close together, it is probable that the pressure that selected for survival of unicorns with the gene for fast running also swept along the gene for long horns resulting in unicorns with both traits.

9. Why do some scientists think that the high incidence of type 2 diabetes in modern humans may be due to pressures that selected for genes that may have been advantageous in the past? Briefly explain.
   In the past, humans led active lives and had to expend a lot of energy to gather, hunt, or farm food. Under these conditions, genes that helped to store fat and survive periods of famine were advantageous. Modern humans live a very sedentary life and have easy access to much more food. Thus the same genes that once were advantageous may now lead to increased obesity and diabetes.

10. Shown below is the diagram of a representative human chromosome, with polymorphisms in genes A, B, and C. Polymorphism A enhances the immune system, polymorphism B results in big feet, and polymorphism C results in baldness. (Assume these traits are each due to a single gene.)



    a. If there is a strong selective pressure towards variant A, which polymorphism (B or C) is most likely to have co-evolved with A? Briefly explain.
       Gene B. Because it is closer to A on the chromosome and therefore more likely to sort with A.

    b. Given the information shown above, can you predict the phenotype of this population after several generations? Briefly explain.
       Yes. The population is likely to have big feet as well as a better immune system.