EVOLUTION

Unit Issue

TUBERCULOSIS (TB) IS A HUMAN disease caused by the bacterium *Mycobacterium tuberculosis* (*M. tuberculosis*). It affects the lungs and other parts of the body and can lead to severe illness and even death. As many as a third of the people on Earth are infected with this bacterium. In 2009, more people died from TB than in any previous time in history. Yet, 9 out of 10 people have developed immunity to this bacterium and do not get sick. If they were to take a TB test, the result would be positive, but they would not have the disease and could not pass it on to other people. Only 1 in 10 will develop *active TB*, which is transmissible to other people.

Based on recent genetic evidence, researchers now believe that *M. tuberculosis* has affected human populations for as long as 40,000 years. Researchers have evidence that the disease originated in Africa and stayed with humans as they migrated to Europe and Asia. Researchers think the original strain of the bacterium was a generalist strain, infecting both humans and livestock such as sheep and cattle; it then evolved to become two distinct specialized strains. One of these strains evolved to become even more successful at infecting people. This strain continues to evolve today, and human actions—such as not completing a full course of prescribed antibiotics—are likely the primary cause. A significant result of this evolution is antibiotic resistance, which you will learn more about in this unit.

Other diseases caused by pathogens—diphtheria, smallpox, AIDS, and influenza A, to name just a few—are also thought to have long evolutionary histories. In some cases, researchers think that the disease-causing pathogens may have originally infected animals

living with, or in close proximity to, humans. The pathogens then evolved to infect humans. For example, the bacterium that causes diphtheria is thought to have originated in domestic herbivores, such as cows. The bacterium causing smallpox is thought to have originated in camels. The virus causing AIDS is thought to have originated in chimpanzees, and the virus causing influenza A is thought to come from birds.

FIGURE A: *Some animals that were the original hosts for infectious diseases that now affect humans*

In addition to the pathogens that have infected humans for hundreds, thousands, and even tens of thousands of years, new diseases have emerged or re-emerged more recently. For example, in 2015–2016, the Zika virus became an epidemic. The geographic range of this virus is typically limited to a narrow strip along the equator, but a mutation occurred that allowed it to spread more widely. In 2019, a new mutation in the spike protein of a coronavirus emerged and caused a global pandemic. Some scientists think that human actions, including land-use practices (such as deforestation), are at least partly responsible for the growing rate of emerging and re-emerging diseases.

Figure B shows a map similar to the ones you observed in the very first activity of this course, "Changing Landscapes" in Sustainability: Changing Human Impact. This map shows areas experiencing

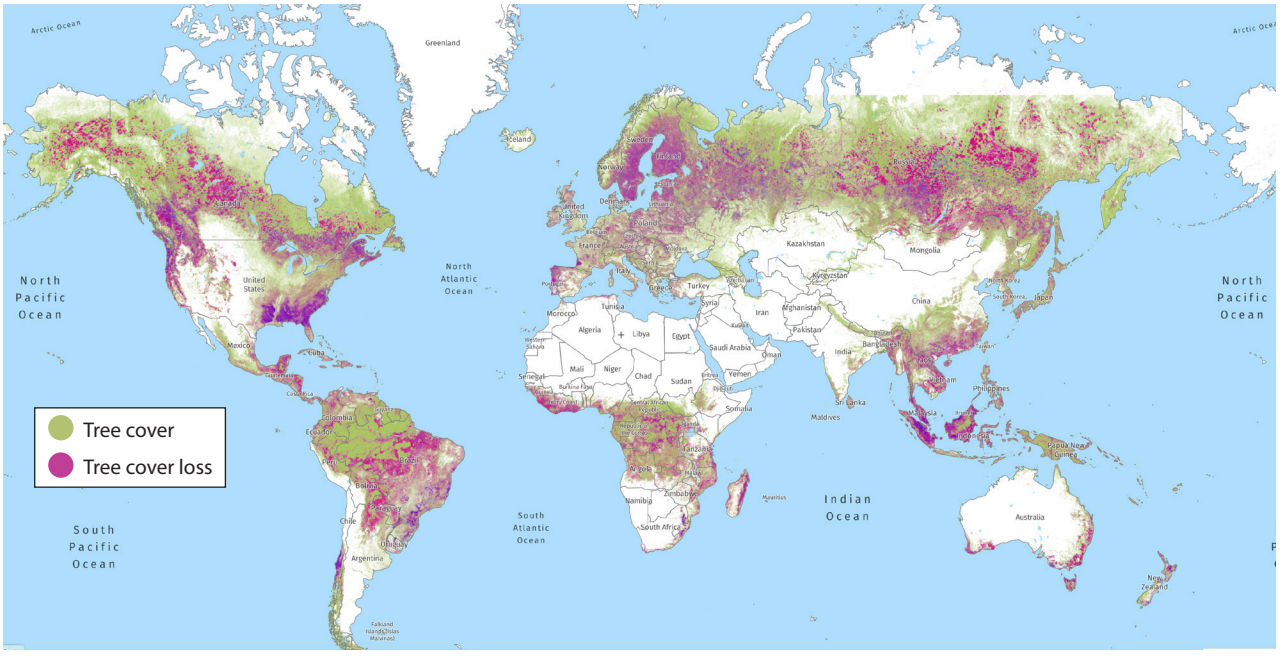

FIGURE B: *Map of global tree cover and tree cover loss*

deforestation and loss of biodiversity. Compare this map with the one in Figure C, which shows scientists' predictions about where emerging diseases are likely to be found—the lighter the color, the greater the probability. What patterns do you notice? What questions do you have?

In this unit, you will explore some of these questions as well as an overarching question: How do human activities affect the evolution of other species, and what are the consequences for both biodiversity and ourselves?

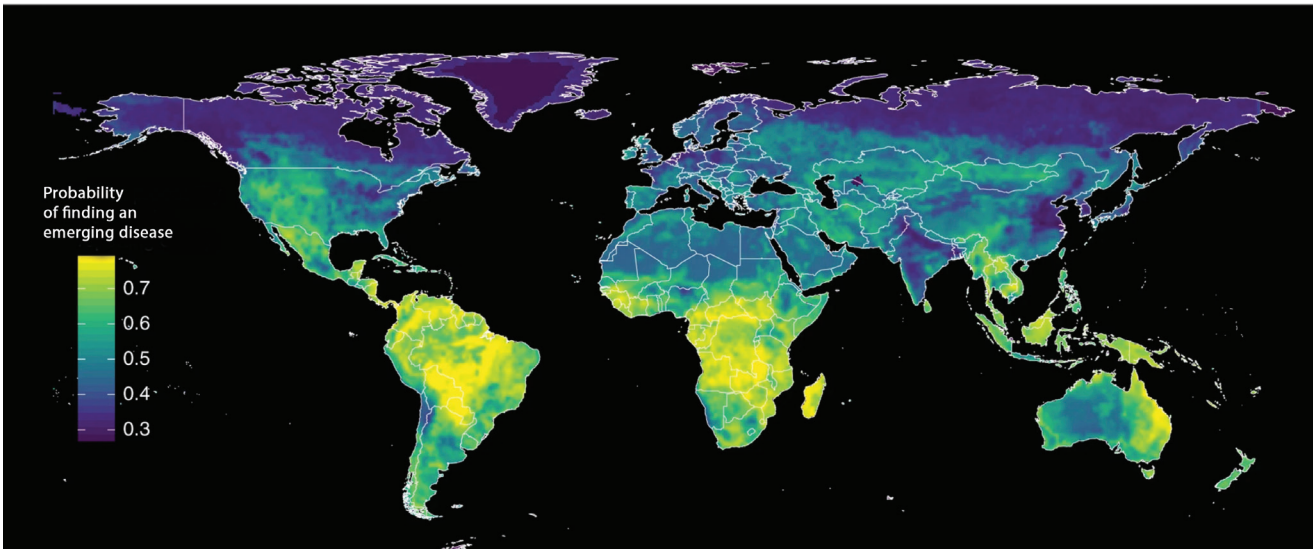

FIGURE C: *Map showing where emerging diseases are likely to be found. The lighter the color, the greater the probability of finding an emerging disease; the darker the color, the lower the probability.*

4 Genetic Variation and Change

IN THE PREVIOUS ACTIVITY, YOU explored how behavioral traits become more common, or evolve, in a population when the trait increases the chance that offspring or close kin (with many of the same genes) will survive and reproduce. Genes bring about traits in organisms and are inherited from one generation to the next. Small changes or mutations can happen when genes replicate. Some mutations, like those that influence body size in marine iguanas, have the potential to lead to evolutionary change in a population. In this activity, you will investigate the relationship between the genetic disease cystic fibrosis and the highly infectious disease TB.

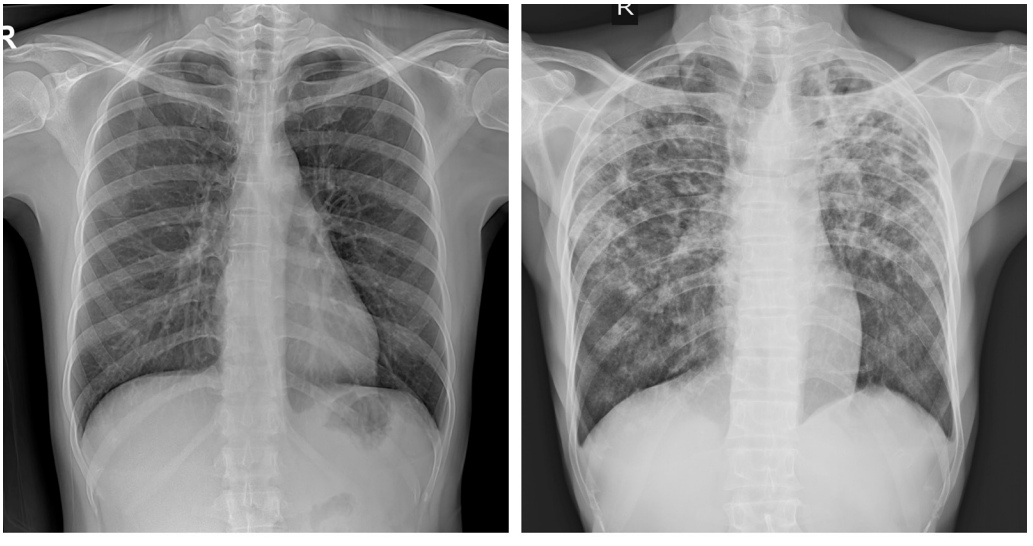

FIGURE 4.1: *Normal and TB-infected lungs*

Guiding Question

What is the role of genetic variation in evolution?

Materials

FOR EACH PAIR OF STUDENTS

computer with Internet access

Procedure

Part A: Exploring the Link

1. Read "Cystic Fibrosis and Tuberculosis" for some background information on each disease.

Cystic Fibrosis and Tuberculosis

Cystic fibrosis (CF) is a recessive genetic disease that affects about 1 in 2,500 people in the United States; about 1 in 20 people are carriers of the genetic mutation that causes this disease. Because CF is recessive, in order to have the disease, offspring must inherit two copies of the mutated gene from each parent. Those who inherit only one copy of the mutated gene—*carriers*—do not develop symptoms but can pass on the gene to their offspring. For those with CF, a mutation in a channel protein prevents the normal flow of salts out of cells of the gut and lungs. This leads to the formation of thick mucus that can obstruct the intestine and block breathing in the lungs.

Historically, without treatment, CF patients died before they reached childbearing age. Today—with modern medications, therapies, and surgeries—patients can live beyond 40 years old.

It is curious that such a potentially lethal disease has persisted in human populations at such a high level. Normally, such mutations fade out of a population over time, as those with the disease die before they can reproduce. Scientists studying CF theorized that carriers may be protected from other diseases, which could explain why CF persists.

As you read in the Unit Issue, TB is a disease caused by the bacterium *Mycobacterium tuberculosis* and mainly affects the lungs. Between 1600 and 1900 in Europe, the TB pandemic caused 20% of all deaths. Because both TB and CF affect the lungs, scientists suspected a connection between the two diseases. They have hypothesized that having one copy of the CF mutation and one normal gene provides protection against certain diseases, including TB.

2. With a partner, discuss the following questions:
   - What is the difference between a genetic disease and an infectious disease?
   - How do you think a genetic disease could provide protection against an infectious disease?
   - If there were a continuous high chance of contracting TB among a population, would you expect the number of CF cases to increase, decrease, or remain the same over several generations? Explain your thinking.
   - How might people's access to health care affect the scenario in the previous question?
3. Follow your teacher's instructions to share your thinking with the class.

Part B: Simulating the Hypothesis

4. Prepare for the simulation by reading "The Cystic Fibrosis and Tuberculosis Simulation."

The Cystic Fibrosis and Tuberculosis Simulation

You will use a simulation to analyze data about the relationship between CF and TB among adults in a population. This simulation is based on real-world data in Europe, where the CF mutation is thought to have originated. The simulation allows you to choose the probability of getting TB (0%, 40%, or 80%) and to vary the average health-care level for people with CF, based on the era (pre-1950, 1950–1990, and post-1990). Each era provided unique health-care options to CF patients. Prior to the 1950s, only routine comfort care was available. Between 1950 and 1990, medications became available to treat patients' symptoms, and after 1990 some patients had access to surgeries (such as lung transplants), enzyme therapies, and specialized antibiotics.

The simulation also allows you to analyze the data by wealth distribution. Specifically, you will be able to determine the relative rates of genotypes among adults for the top 20th and the bottom 20th percentile in wealth.

5. With your partner, visit the *SEPUP SGI Third Edition* page of the SEPUP website at **www.sepuplhs.org/high/sgi-third-edition**. Follow your teacher's instructions to access the simulation.

Or access sim here

6. Decide with your partner how you would like to set the variables of the simulation. You can choose the likelihood of getting TB in your population and the average health-care level by era.
7. Press "Run Simulation." The line graph shows the percentage of each trait in the population for each generation over time.
8. Record your results and any additional data or observations in your science notebook. Be sure to note any questions you have that you would like to test.
9. Below the first graph, set the variables again to repeat the simulation. Keep one variable the same, and change the other. For example, if you initially set the TB rate to 80%, keep this variable at 80% and choose another health-care variable.
10. Look over the second line graph, and compare it to your initial line graph. Record the new data and any differences you observed in your science notebook.
11. Above the second line graph, press "See Differential Outcomes Based on Wealth Disparity." Two line graphs show the percentage of each trait in the population for each generation over time among the top 20th and the bottom 20th percentile in wealth.
12. Record your results in your science notebook. Be sure to note any questions you have that you would like to test.
13. Continue to run the simulation and change the variables.

    Hint: Keep changing just one variable at a time until you can explain the relationship between CF and TB. Be sure to record your variables and results for each test that you run.

Build Understanding

1. What is the evidence for a cause-and-effect relationship between the probability of contracting TB and the frequency of the CF mutation?
2. CF is caused by a single mutation. Explain how the environment affects whether this mutation is beneficial, harmful, or neutral for a person.
3. Explain how environmental changes affect the CF trait over time in your population. Use evidence, including mathematical representations, from your investigation to support your explanation.
4. **Issue connection:** How might people be changing the environment, and therefore how are they affected by evolution?

KEY SCIENTIFIC TERMS

carrier
evolution
gene
genetic variation
mutation

Extension: Engineering Connections

Scientists are using genetic engineering to treat genetic diseases, like CF and sickle cell anemia. A gene-editing technique called CRISPR has already been used to correct the CF mutation in human stem cells, and the same technique is currently being used in humans to reverse the symptoms of sickle cell anemia. To learn more about these very recent efforts, visit the *SEPUP SGI Third Edition* page of the SEPUP website at **www.sepuplhs.org/high/sgi-third-edition**.

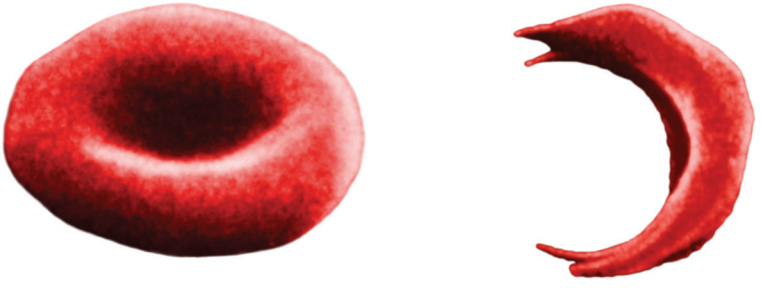

FIGURE 4.2: *Normal red blood cell (left) and a sickle cell (right)*

STUDENT SHEET 4.1

CYSTIC FIBROSIS AND TUBERCULOSIS SIMULATION

For Runs 1–3, keep the average health-care level for people with cystic fibrosis the same. For example, if you initially set the average health-care level to pre-1950, keep this variable at pre-1950 and choose a different chance of getting tuberculosis.

Run 1 Results

Chance of getting tuberculosis	Average health-care level for people with cystic fibrosis (CF)	Relative % of normal genotype after 30 generations	Relative % of CF carrier genotype after 30 generations	Relative % of CF genotype after 30 generations

Run 2 Results

Chance of getting tuberculosis	Average health-care level for people with cystic fibrosis (CF)	Relative % of normal genotype after 30 generations	Relative % of CF carrier genotype after 30 generations	Relative % of CF genotype after 30 generations

Run 3 Results

Chance of getting tuberculosis	Average health-care level for people with cystic fibrosis (CF)	Relative % of normal genotype after 30 generations	Relative % of CF carrier genotype after 30 generations	Relative % of CF genotype after 30 generations

What differences do you notice between the graphs?

What questions do you have about the data?

Name_____ Date_____

STUDENT SHEET 4.1 *(continued)*

CYSTIC FIBROSIS AND TUBERCULOSIS SIMULATION

For Runs 4–6, keep the chance of getting tuberculosis the same. For example, if you initially set the tuberculosis rate to 80%, keep this variable at 80% and choose a different health-care variable.

Run 4 Results

Chance of getting tuberculosis	Average health-care level for people with cystic fibrosis (CF)	Relative % of normal genotype after 30 generations	Relative % of CF carrier genotype after 30 generations	Relative % of CF genotype after 30 generations

Run 5 Results

Chance of getting tuberculosis	Average health-care level for people with cystic fibrosis (CF)	Relative % of normal genotype after 30 generations	Relative % of CF carrier genotype after 30 generations	Relative % of CF genotype after 30 generations

Run 6 Results

Chance of getting tuberculosis	Average health-care level for people with cystic fibrosis (CF)	Relative % of normal genotype after 30 generations	Relative % of CF carrier genotype after 30 generations	Relative % of CF genotype after 30 generations

What differences do you notice between the graphs?

What questions do you have about the data?