BiteScis | STUDENT

Mitochondrial Inheritance

Genes That (Almost) Always Come Only from Mom

Introduction

Unless you are an identical twin, no one else in the world shares your exact combination of genes. Genetic information is **inherited,** passed from parents to their offspring, in the form of nucleic acids. Usually, when we talk about genetic inheritance, we are referring to the DNA in the nucleus of our cells. Offspring get one copy of every gene from each of their parents, for a total of two copies per gene. **Mendelian traits** are traits that result from the action of a single gene and are inherited in predictable patterns. If you've ever done a Punett square, you've dealt with Mendelian traits. **Pedigrees** are visual tools used in tracking patterns of inheritance. In this lesson, we will use pedigrees to track both Mendelian traits as well as funky **"non-Mendelian traits"** that don't follow the rules!

What To Do

Read "Medical Mystery!" below. Then, answer the analysis questions, reading the Science Bite when instructed.

Medical Mystery!

A few years ago, a doctor in Cincinnati who specializes in mitochondrial diseases stumbled upon an unusual patient. A mother brought her four-year-old son into the clinic because she was worried he had a disease due to a mutation in his mitochondrial DNA. To diagnose the boy, the doctor took a sample of his blood and ran it through a DNA sequencer to "read" his mitochondrial genome. Typically, humans have one mitochondrial genome that they inherit from their mothers. That's why the doctors were so confused when they looked at the boy's blood sample. It looked like the boy's cells contained *multiple* mitochondrial genomes—as if mitochondria from multiple people had taken up residence inside of them! **Figure 1** is a representation of what they found.

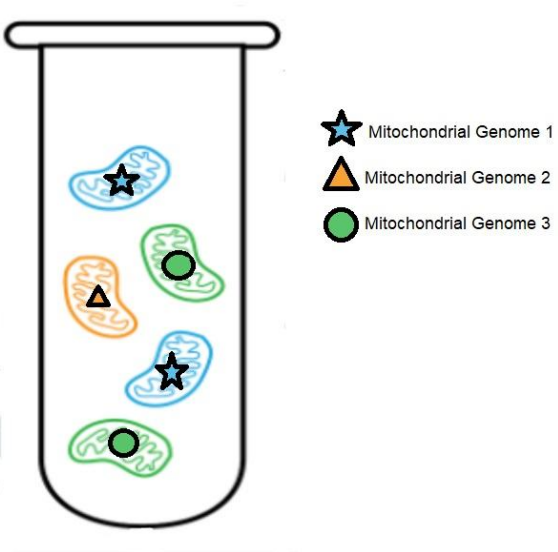

Figure 1. The Boy's Mitochondrial Genome. For illustrative purposes, this is a schematic of what the scientist's test tube looked like. The sample he took from his patient looked like it contained genes from many different mitochondria. *Source: Modified from Luo et al. 2018.*

The doctor collected fresh blood samples and ran the test again. And again. And again… each time, with the same results. Confused, he collected more blood and sent the samples to several other labs, all of which confirmed the results.The doctor had to accept the impossible. A central rule in biology had been broken: Children are supposed to inherit their entire mitochondrial genome from their mother, but this boy seemed to have inherited multiple mitochondrial genomes!

Analysis Questions

1. Based on what mitochondria do, what do you think the symptoms of a disease that affects mitochondria might be? Explain your reasoning.

2. Compare what's going on with the both to both traditional Mendelian inheritance and traditional mitochondrial inheritance. Identify similarities and differences.

3. The boy's doctor reran the blood tests to make sure that the results weren't from a lab error. What kind of error were they most likely thinking had occurred?

The doctor decided he had to get to the bottom of the mystery, and decided to sequence the mitochondrial genomes of more members of the boy's immediate family, which included his two sisters, his mother, and his father. The boy's sisters' mitochondrial genomes looked identical to his, with a mix. **Figure 2** is a model representing what they found.

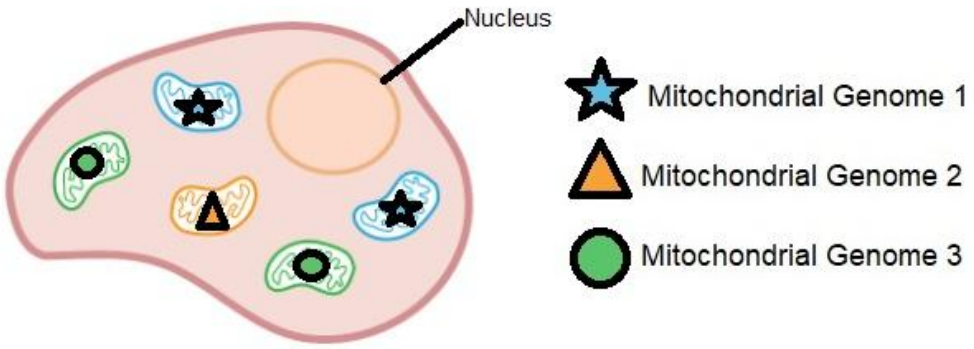

Figure 2. The Children's Mitochondria. Schematic of what the siblings' cells looked like. They all contained multiple types of mitochondria, each with their own genome. *Source: Modified from Luo et al. 2018.*

The doctor next sequenced the mother and father's mitochondrial genomes, modeled in **Figure 3** and **Figure 4.**

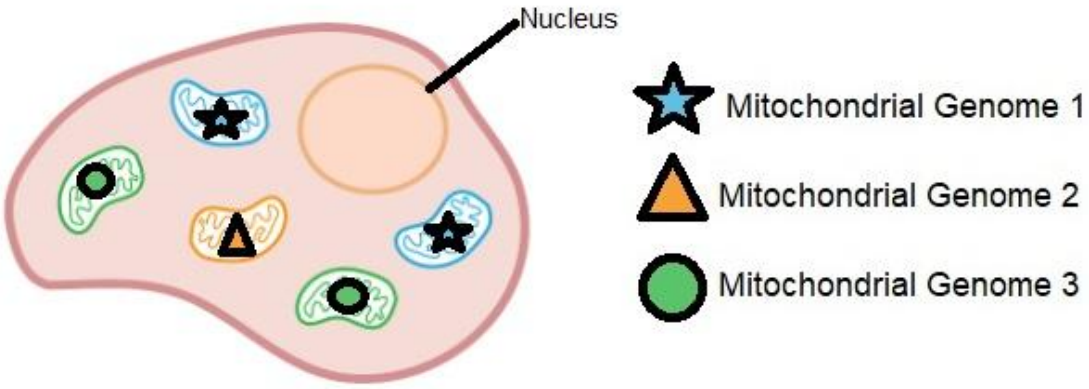

Figure 3. The Mother's Mitochondria. Schematic of what the mother's cells looked like. They all contained multiple types of mitochondria, each with their own genome and each found within her children. *Source: Modified from Luo et al. 2018.*

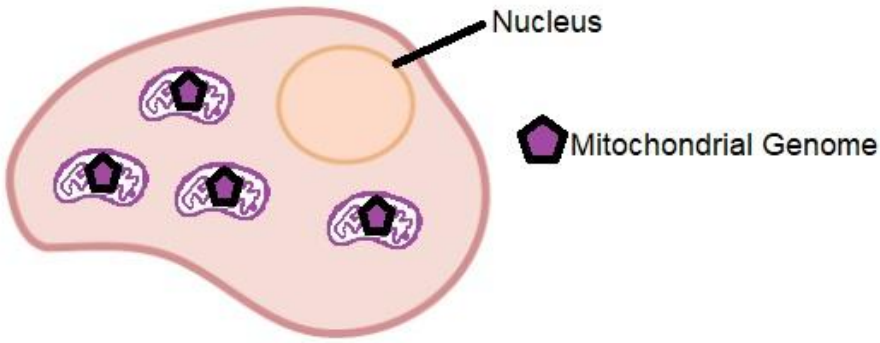

Figure 4. The Father's Mitochondria. Schematic of what the father's cells looked like. They all contained one type of mitochondria, with its own genome; this genome was not found within his children. *Source: Modified from Luo et al. 2018.*

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4. Who did the boy and his sisters inherit their mtDNA from? Is this an expected, or unexpected result? Explain.

The doctor next sequenced the mitochondrial genomes of the mother's parents (the boy's maternal grandparents). The results are modeled in **Figure 5.** You can see a pedigree representing the whole family in **Figure 6.**

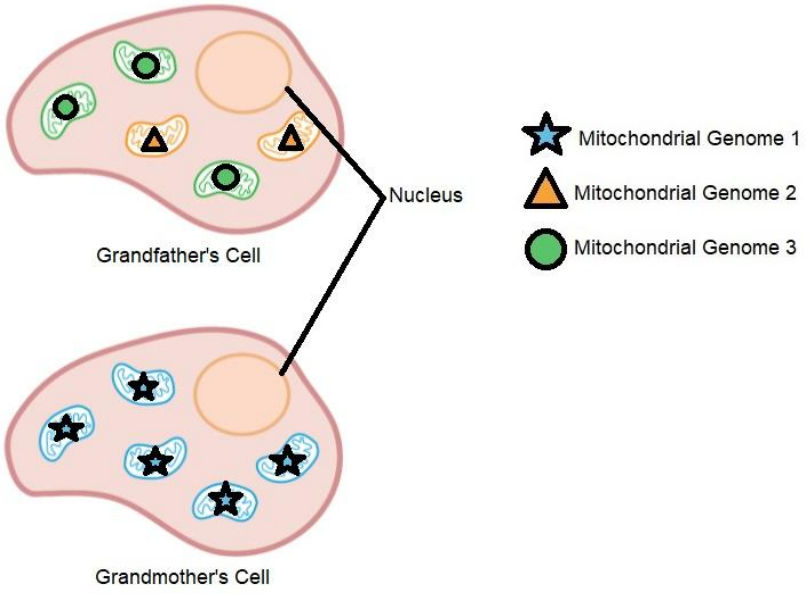


Figure 5. The Grandparents' Mitochondria. Top: the grandfather's mitochondria. Bottom: the grandmother's mitochondria. *Source: Modified from Luo et al. 2018.*

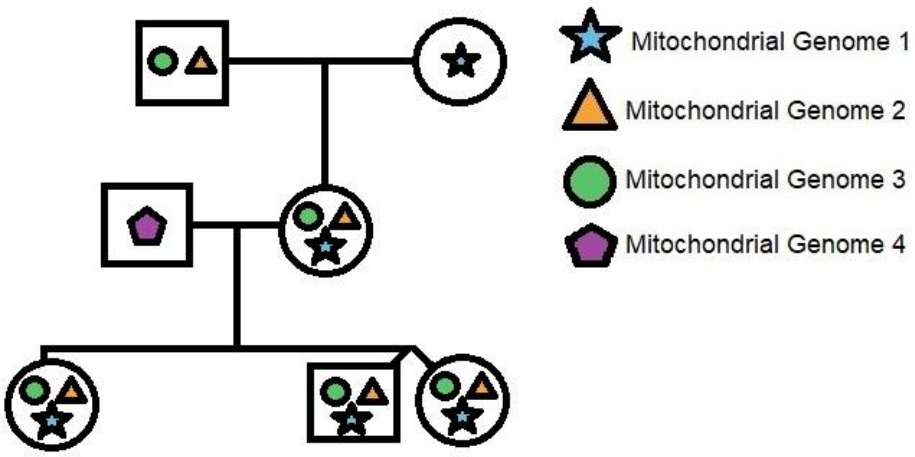
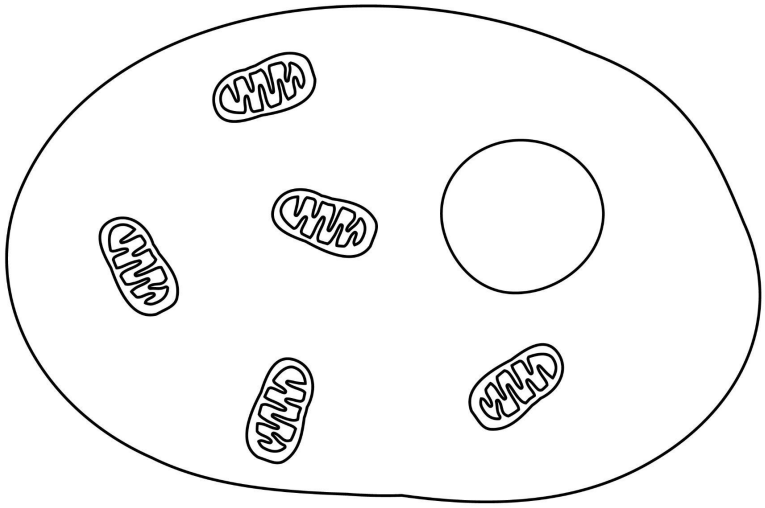


Figure 6. The Family Pedigree. A pedigree representing the inheritance of mitochondrial DNA in this family. Members tracked (top) the boy's grandparents, (middle) the boy's parents and (bottom) the boy and his two sisters. *Source: Modified from Luo et al. 2018.*

5. Who did the boy's mother (**Figure 3)** inherit her mtDNA from? Is this an expected, or unexpected result? Explain.

6. The children's father's mitochondrial genome was *not* seen in his children, yet their mother's side grandfather's was. How could this have occurred? Explain your reasoning.

7. What would you expect if you looked at the mitochondrial genomes of the grandfather's parents (the boy's great grandparents)? Draw the mitochondria below in the space provided, using the appropriate symbols or colors (provide a key if necessary). ***Hint!*** There are at least two possible answers!



8. After reading about the research, a classmate claims that the trait that causes fathers to give their mitochondria to their offspring follows a dominant inheritance pattern. Do you agree with this claim? Justify your answer by citing evidence from the pedigree in the Bite.

9. In your opinion, is inheriting mitochondria from both parents potentially a good thing? Why or why not?

10. Think about the original patient, the four-year-old boy. Make a prediction about what will happen if he has biological children of his own. Is there more than one possibility? What might that tell us about his mom?

11. **Connect to the Big Question.** In the YouTube video "A Biologist's Mother's Day Song," the artist claims that "slightly more than half of everything I am is thanks to you." The "you" in the claim is the artist's mom. Do you support this claim? Provide evidence and reason for whether or not you agree with this statement.