

Exome Sequencing for Healthcare Interpreters

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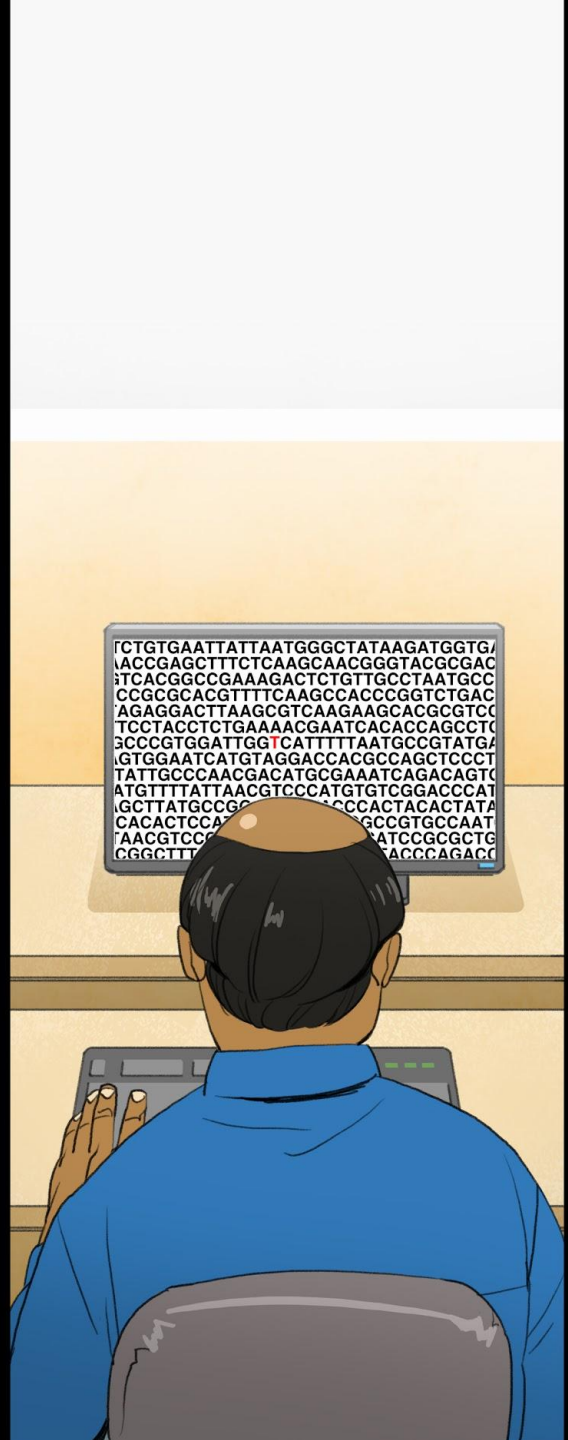
Illustrations Booster Shot Media

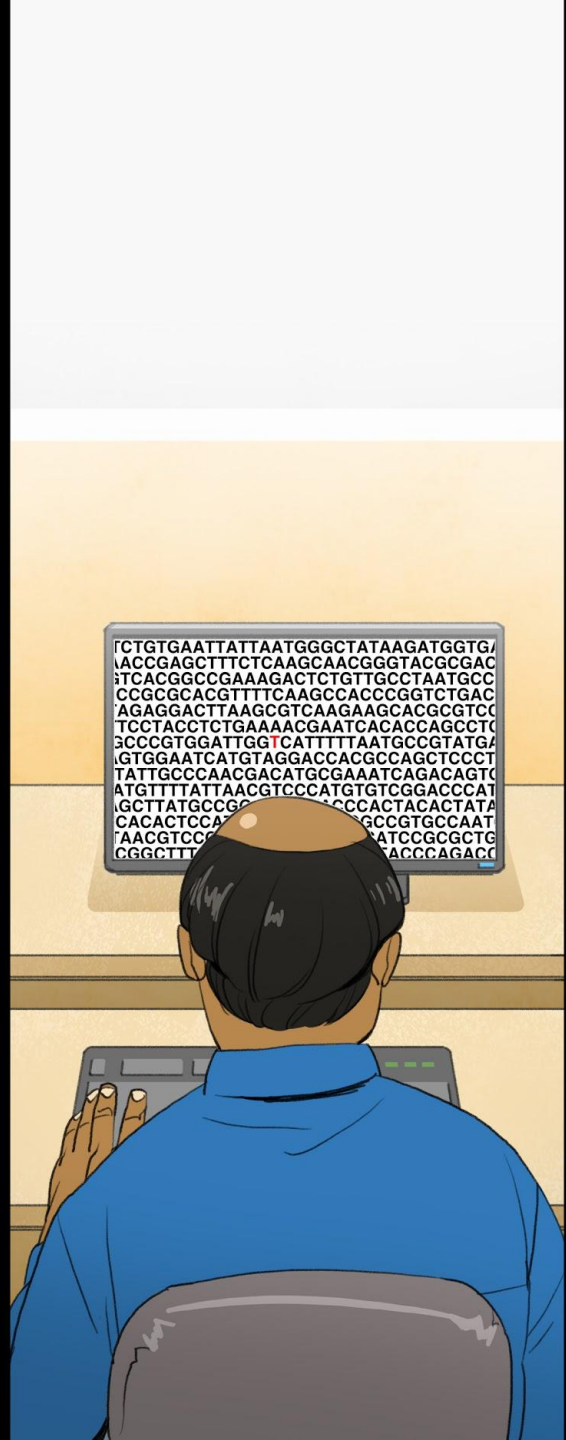
What to expect

1. Online class (today)
2. Vocabulary exercises (on your own)
3. Bilingual glossary of terms
4. List of additional resources

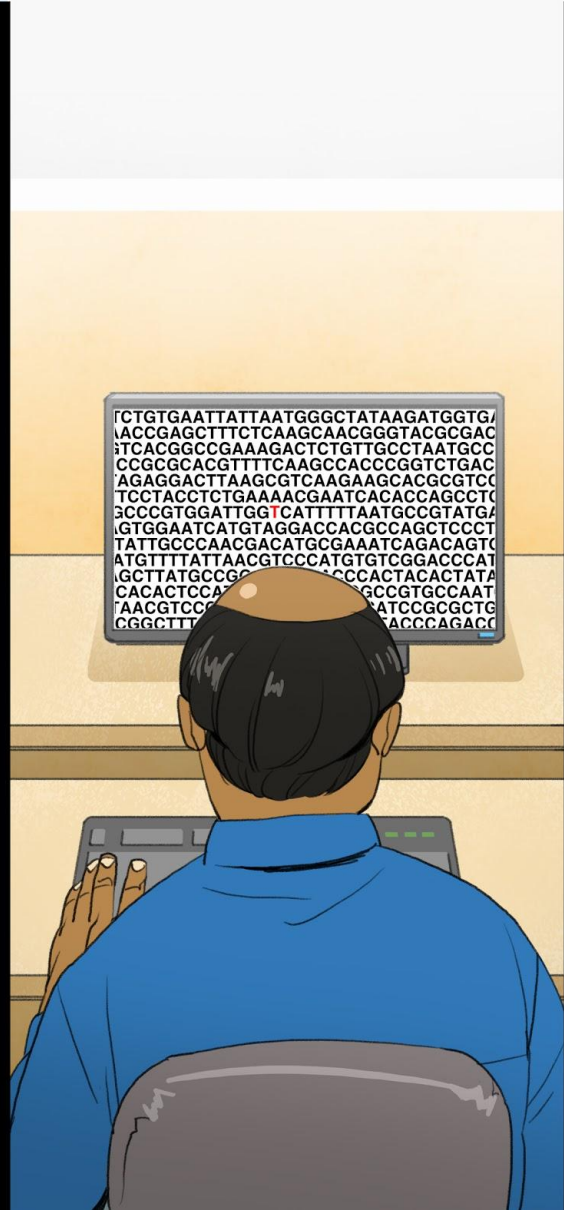
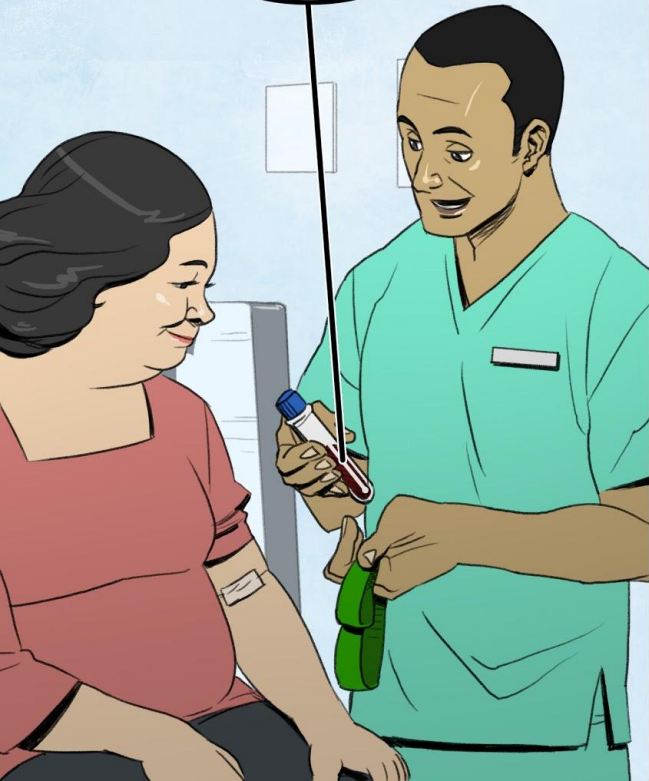
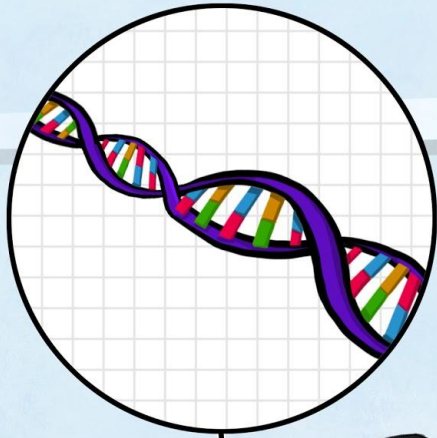




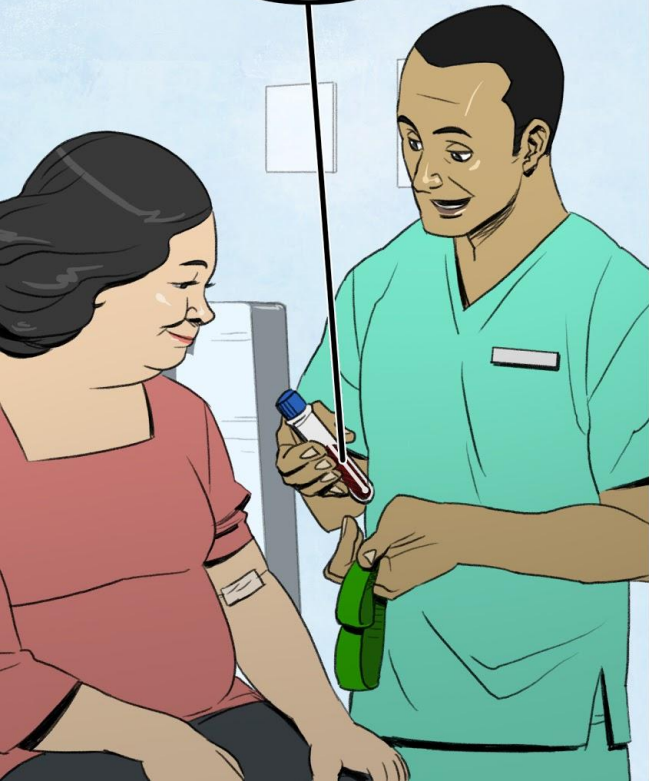
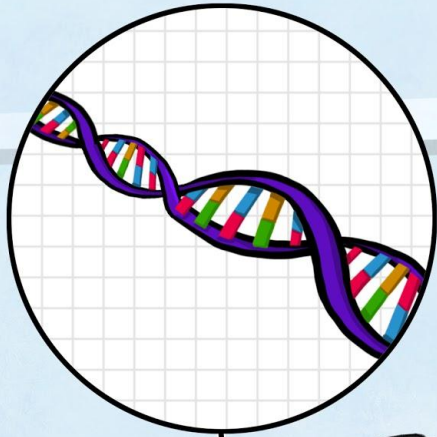




Genetics

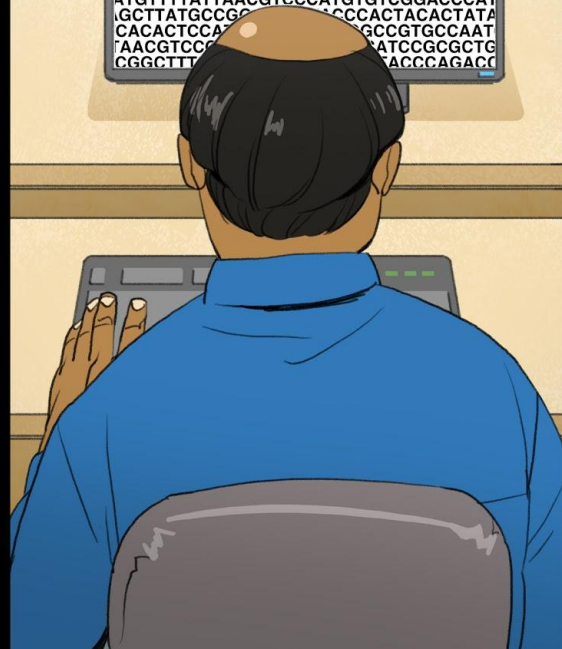


Basic concepts



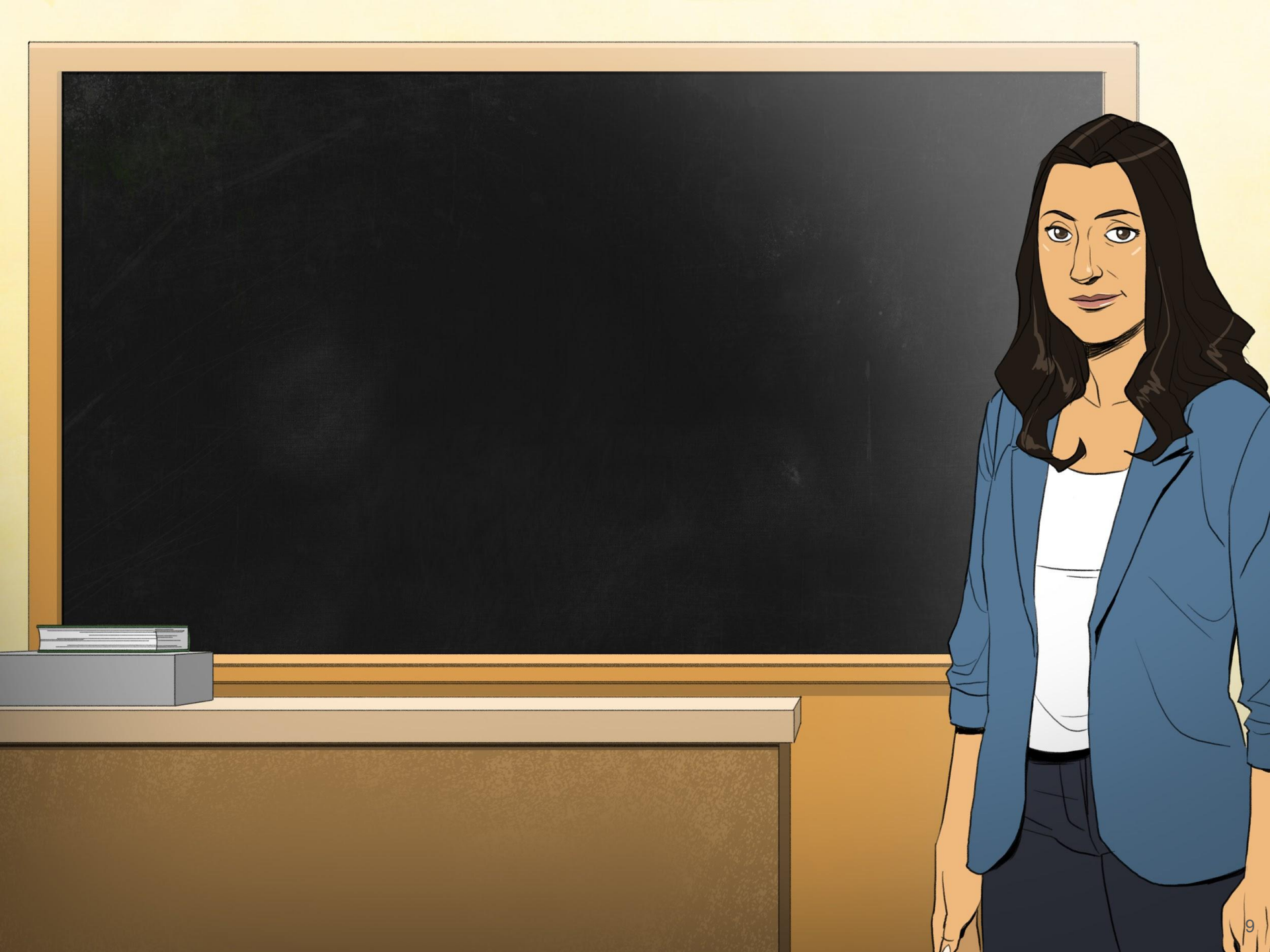
Exome sequencing

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GTGGAATCATGTAGGACCACGCCAGCTCCCT  
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Genetic specialists



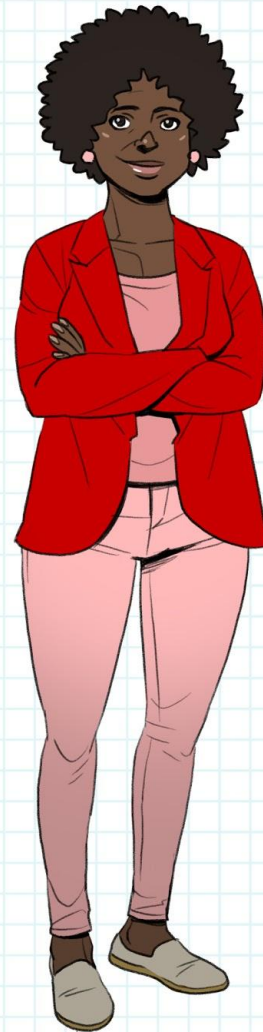


What is genetics?



What is genetics?

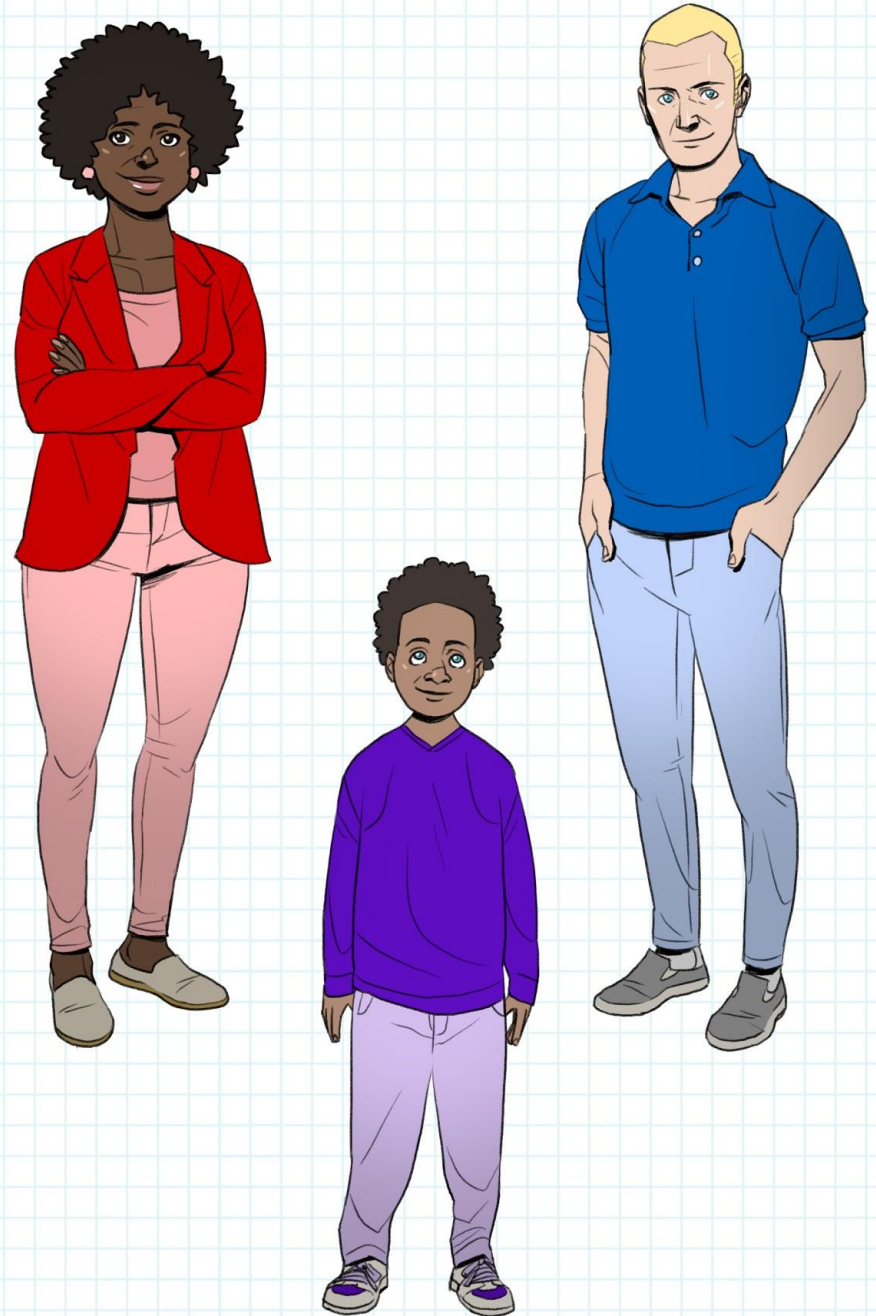
The field of science and medicine that studies the biologic basis of heredity...



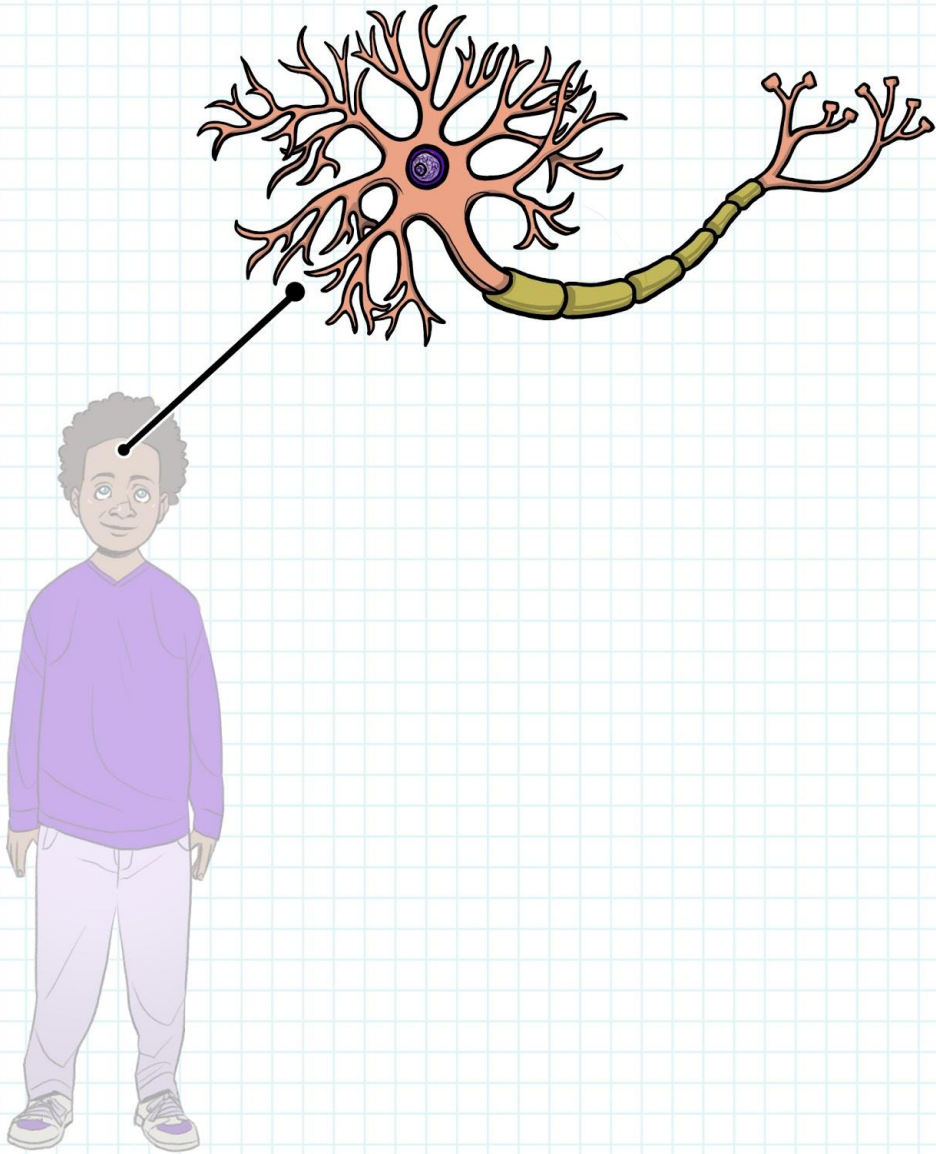
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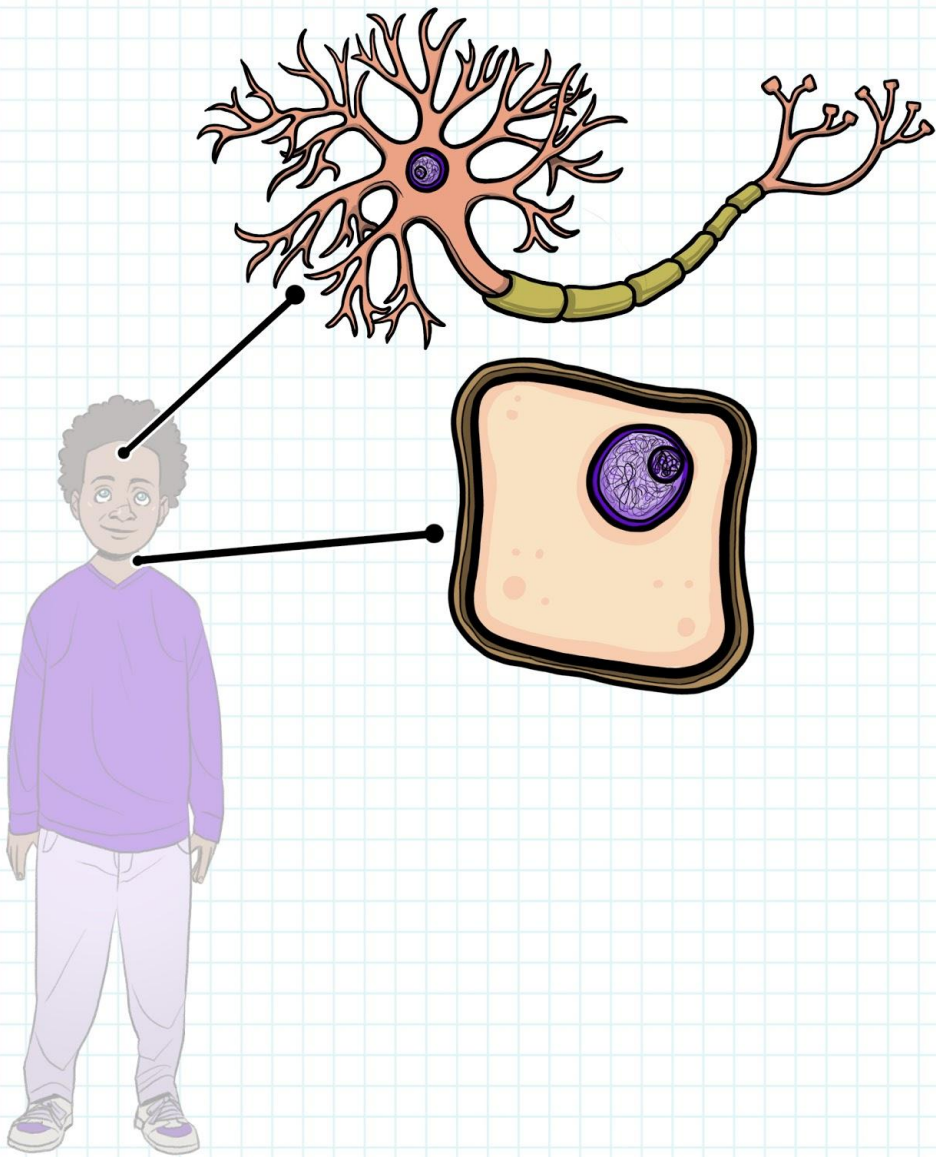
The field of science and medicine that studies the biologic basis of heredity...

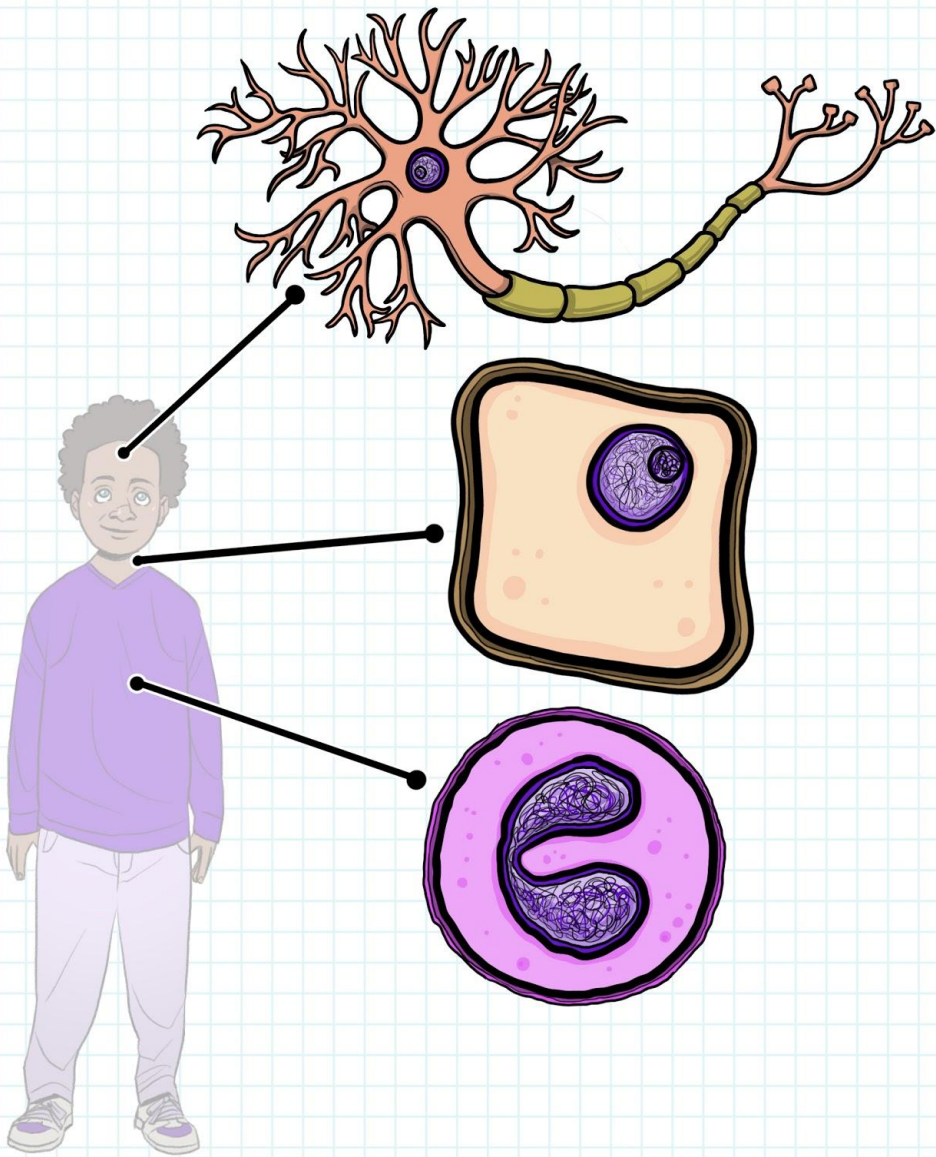
...and how the instructions for life are used by all living organisms.

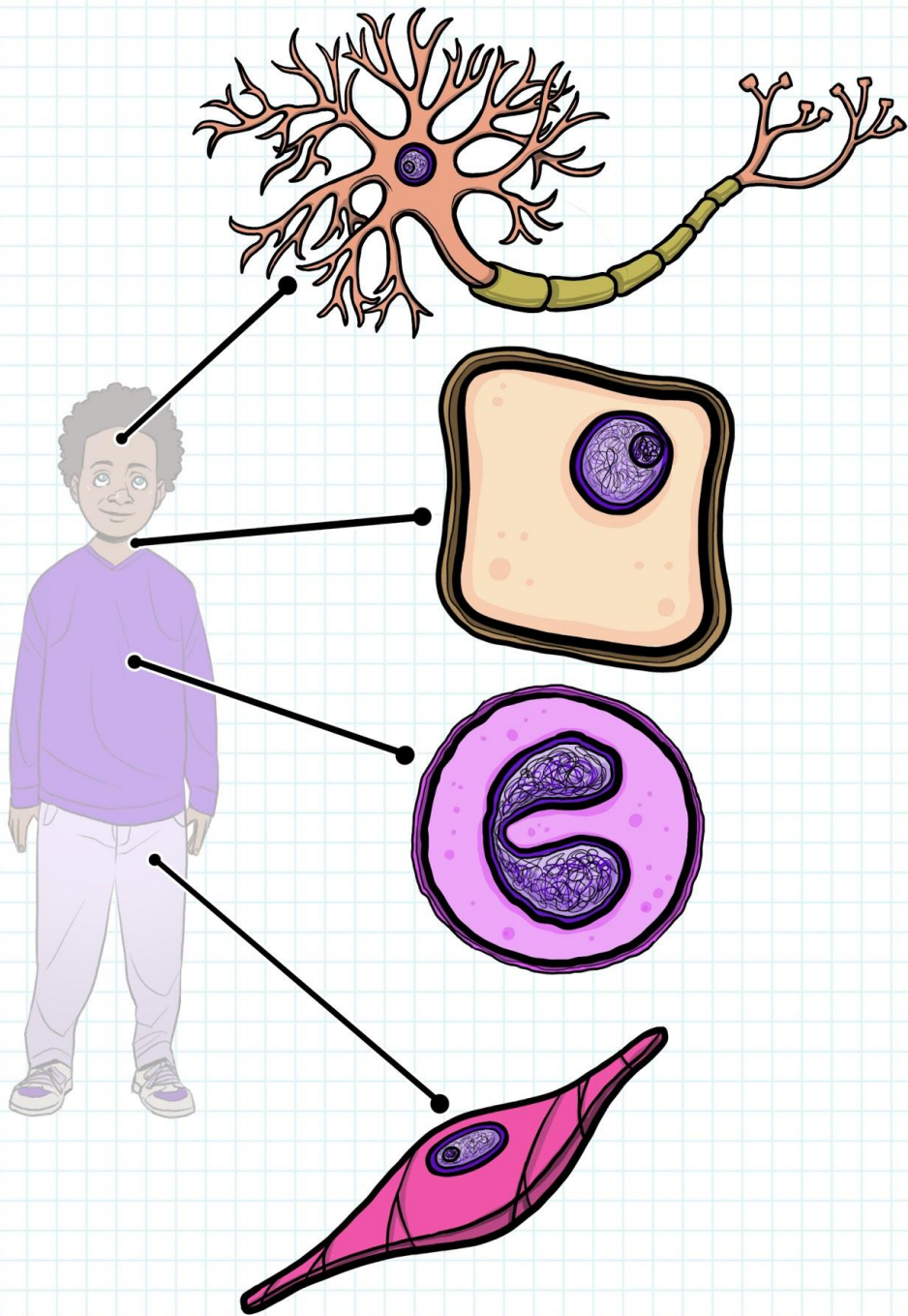


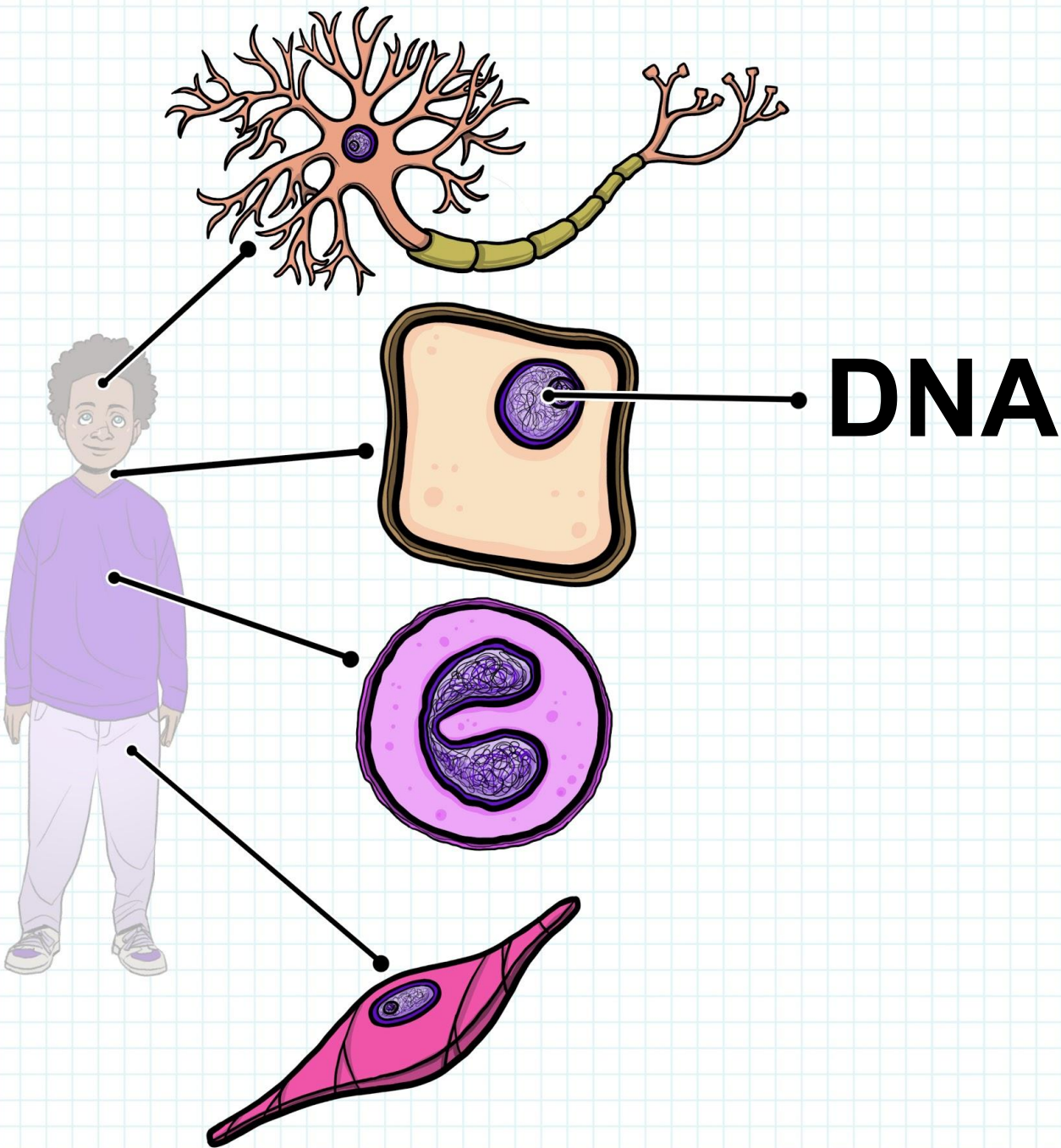








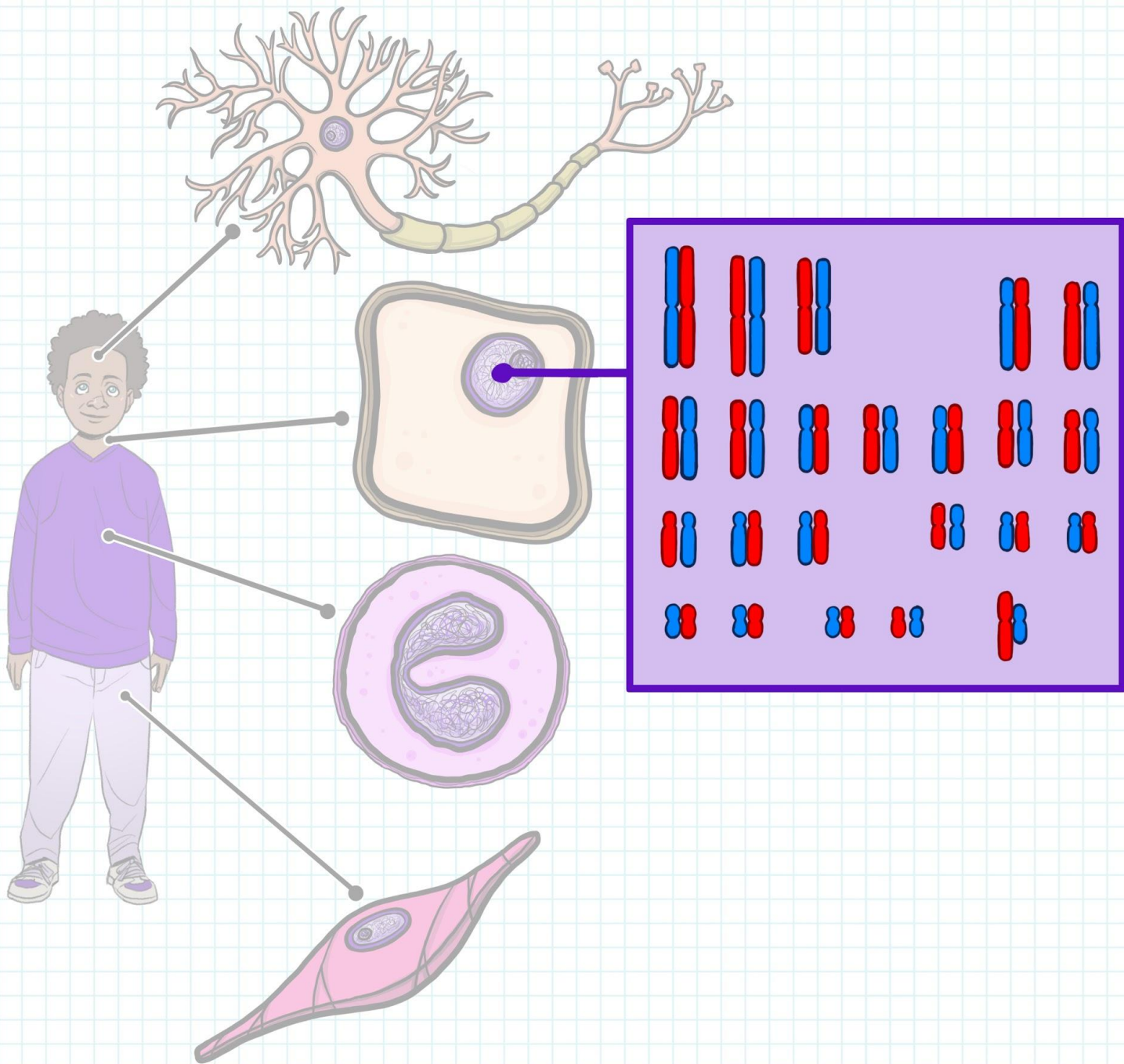


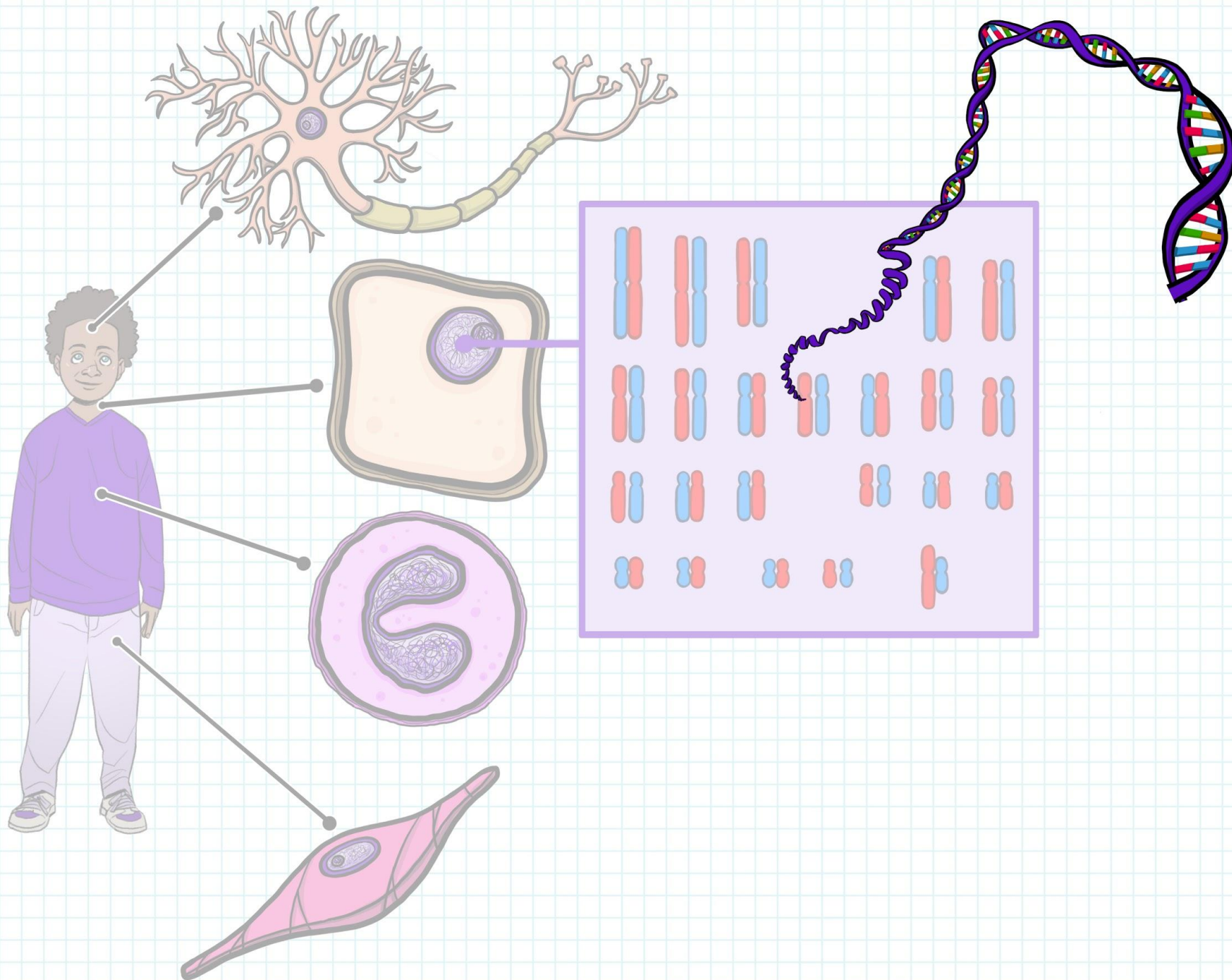


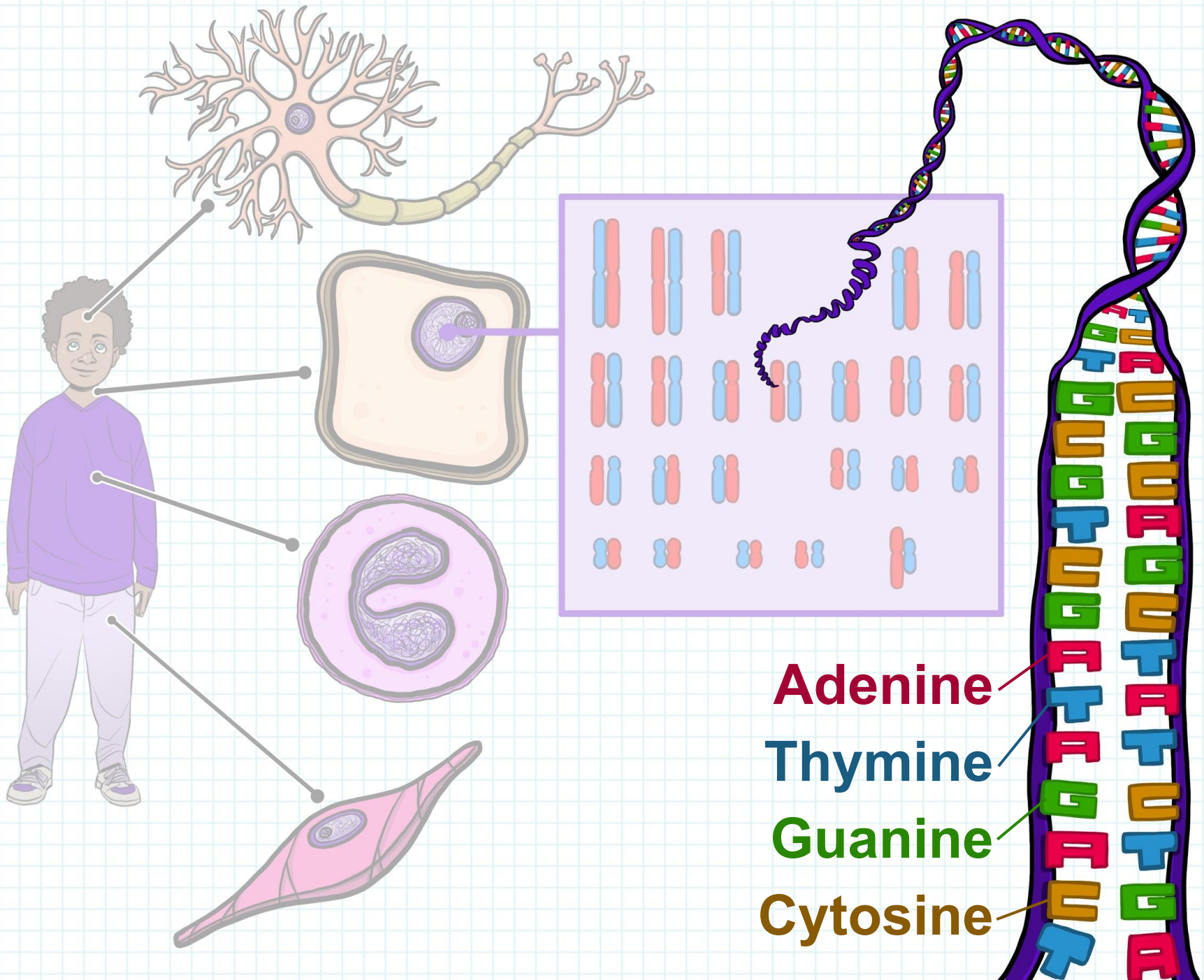
What is genetics?

What is DNA?

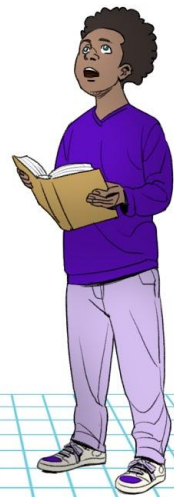








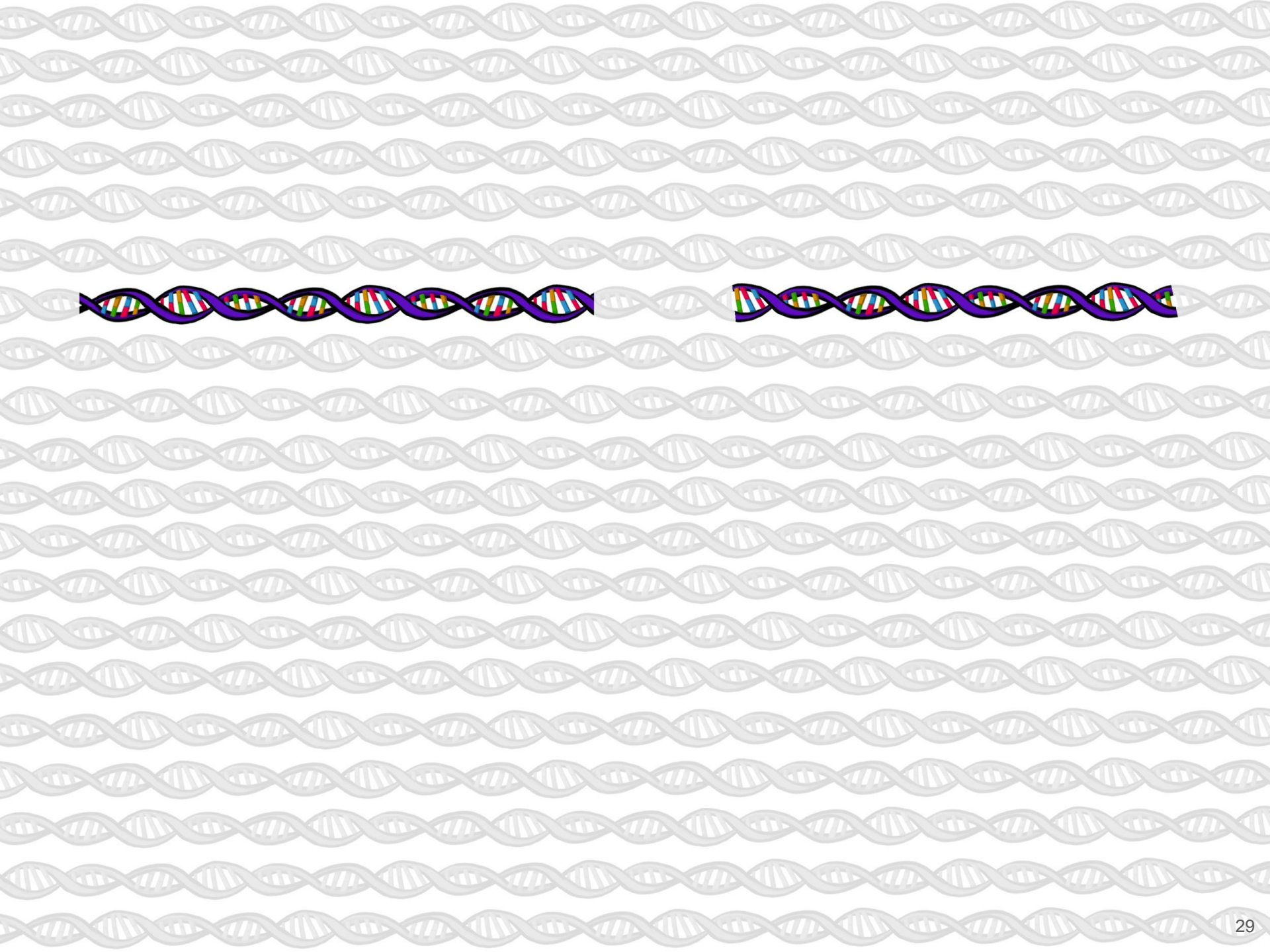
Adenine
Thymine
Guanine
Cytosine



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Quiz!

- What is the function of DNA in our bodies?
- What is a chromosome?
- What is a gene?

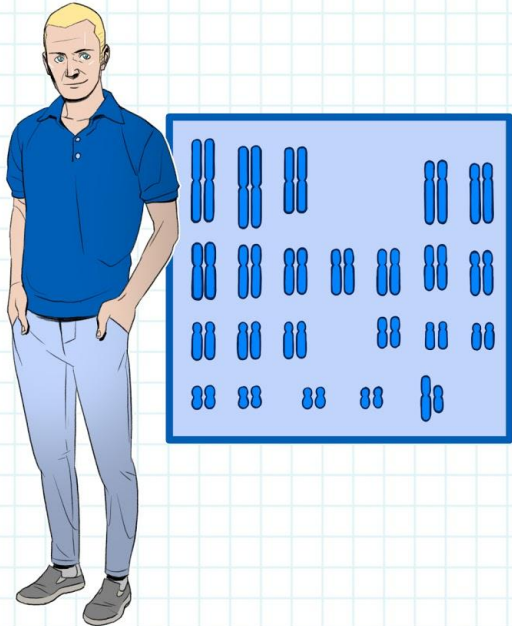
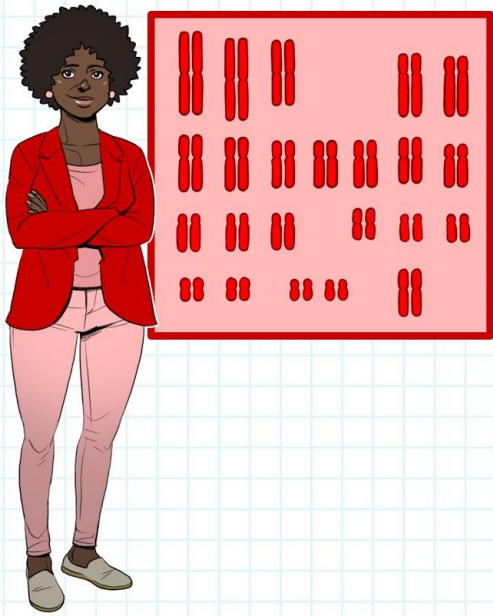
What is genetics?

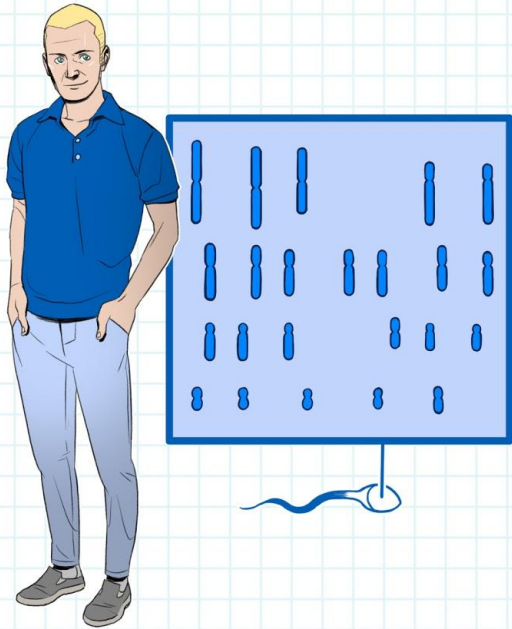
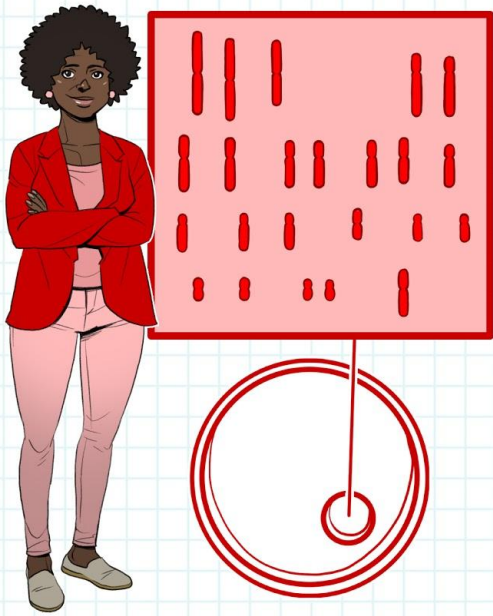
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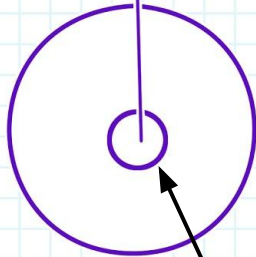
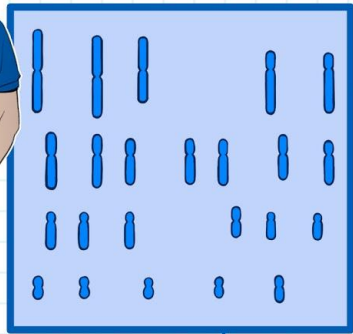
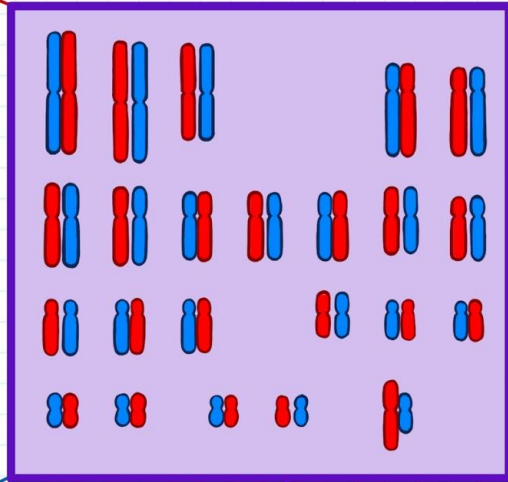
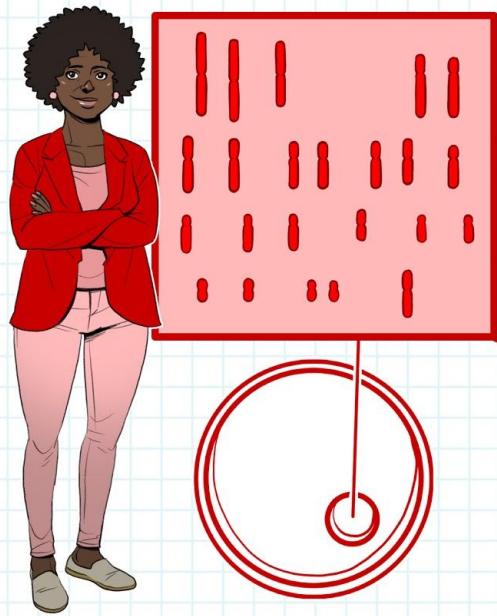
How do we pass on DNA?



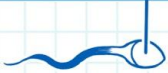
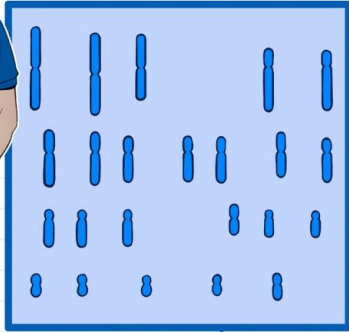
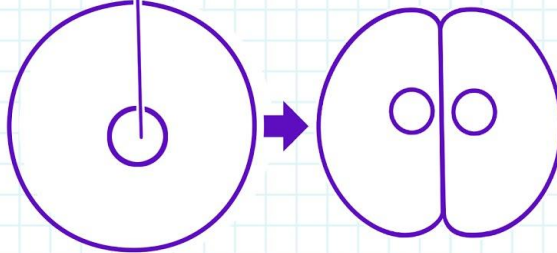
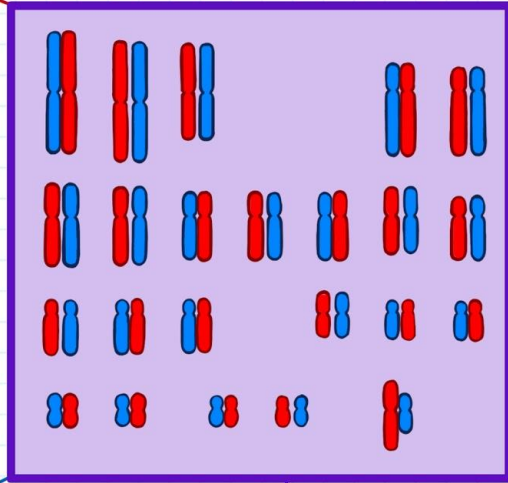
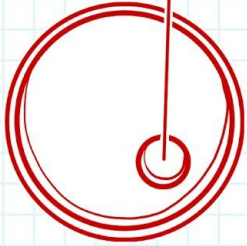
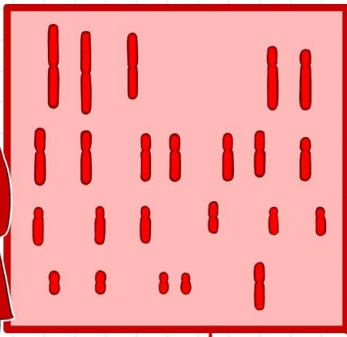


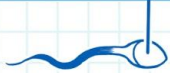
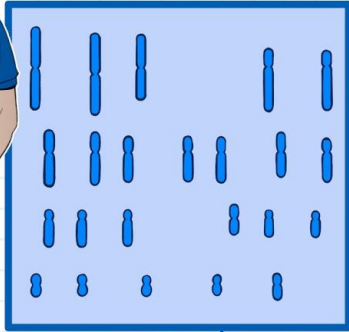
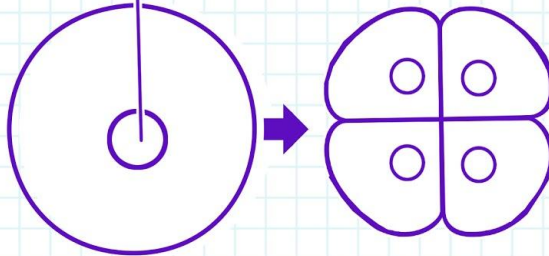
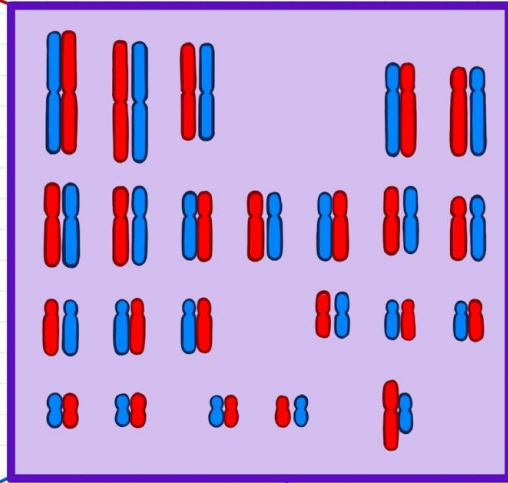
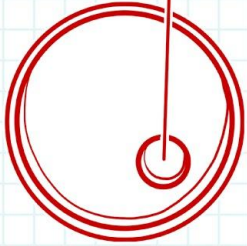
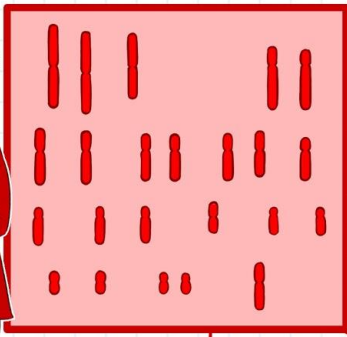


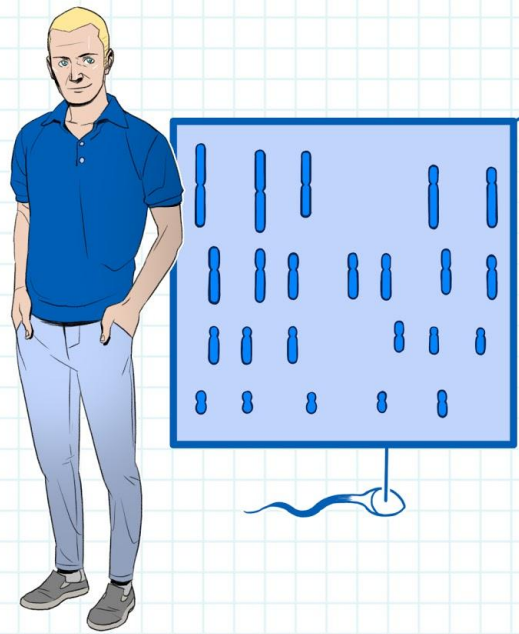
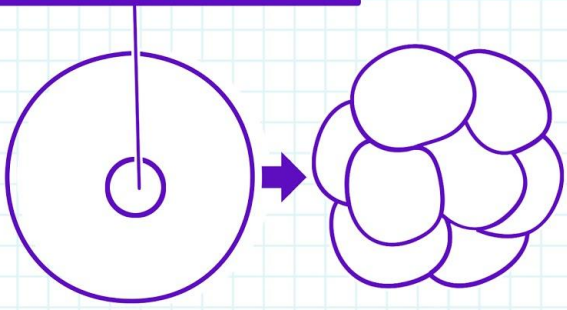
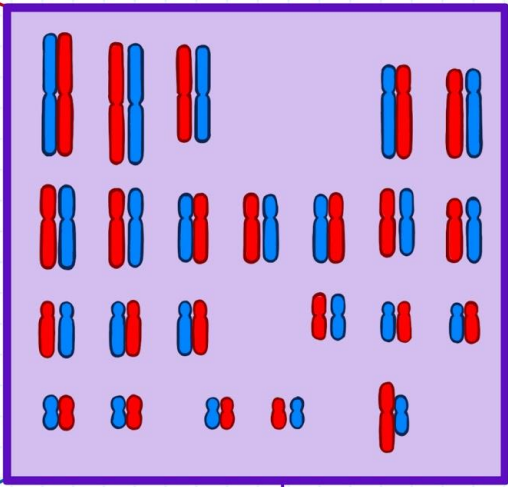
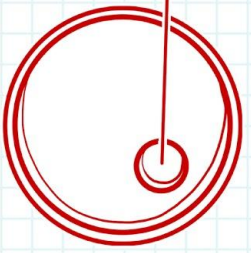
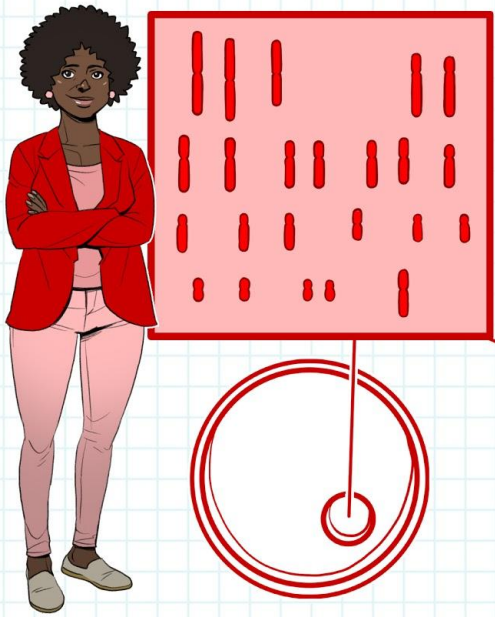


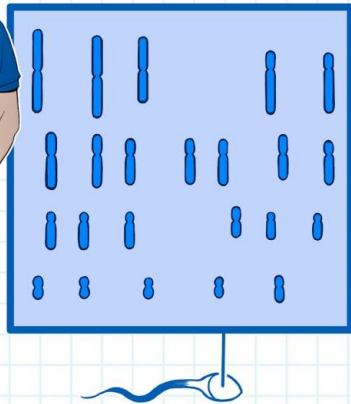
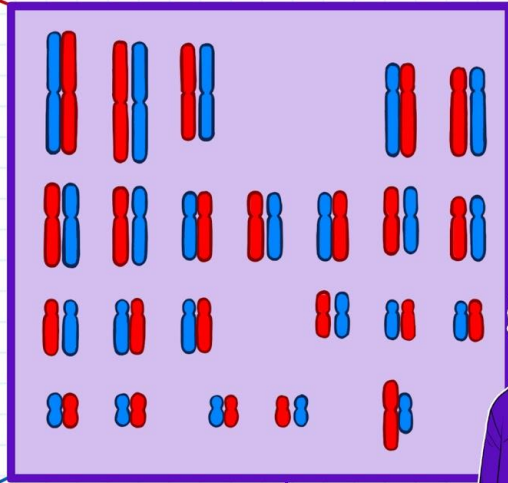
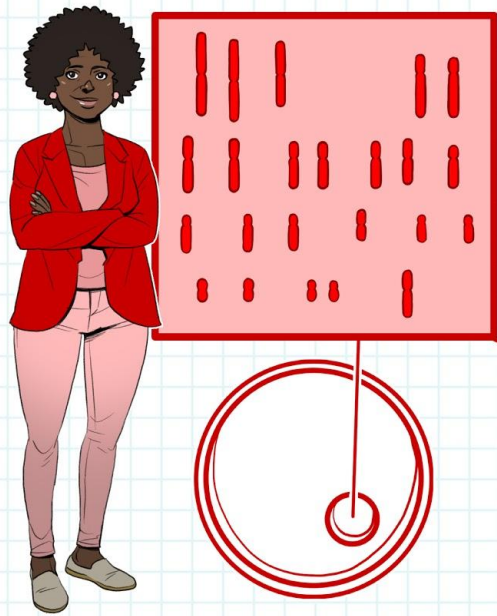


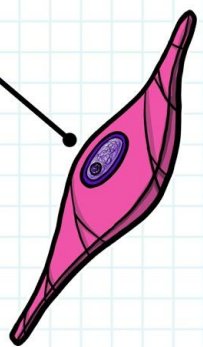
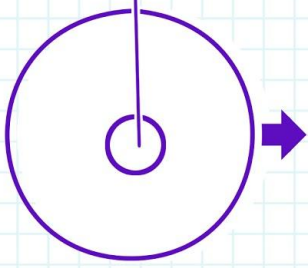
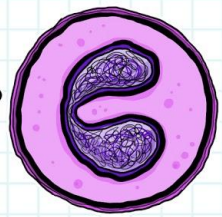
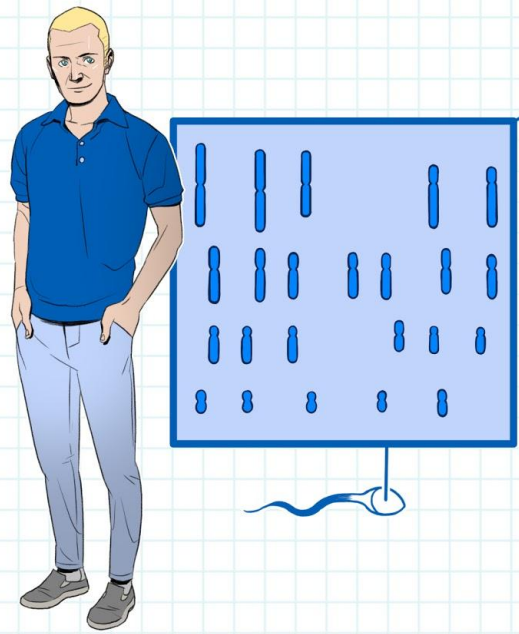
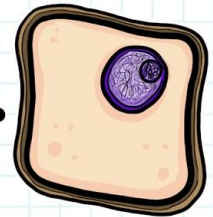
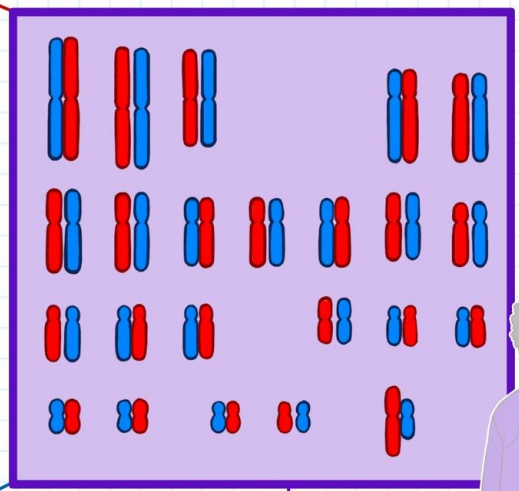
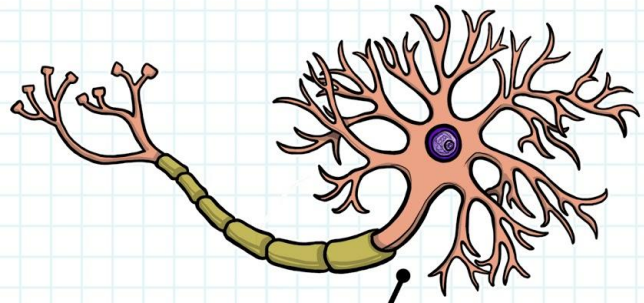
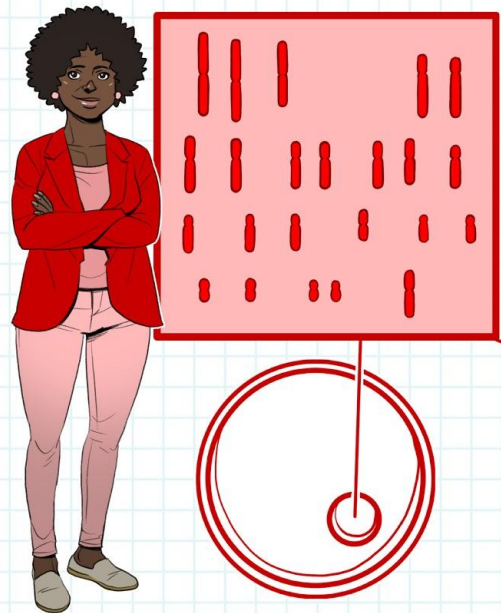
Nucleus

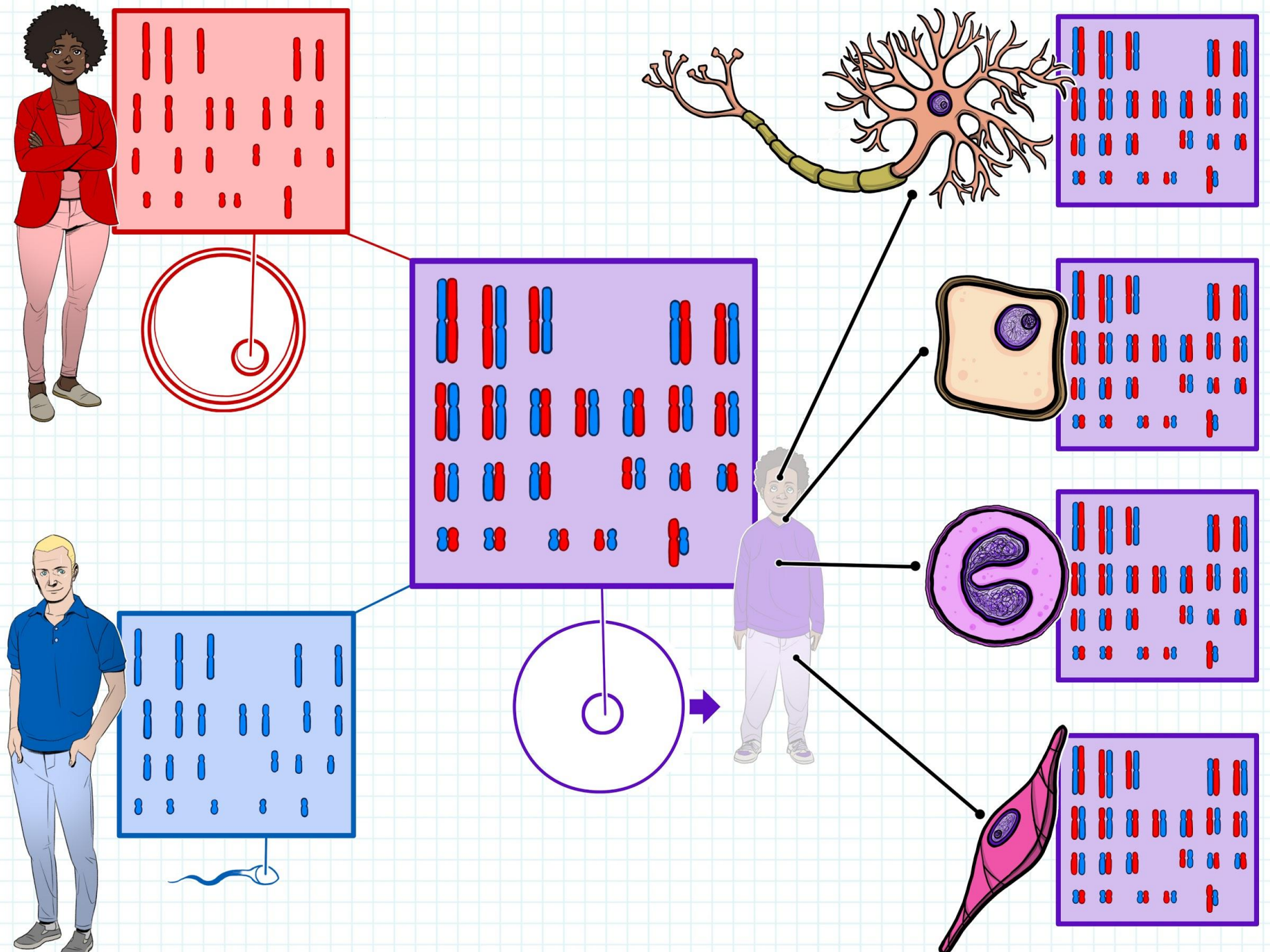










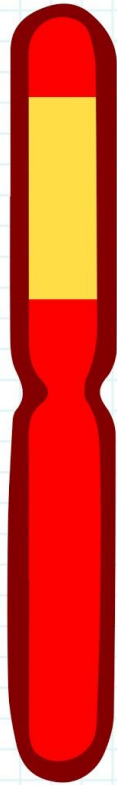


DOMINANT

recessive

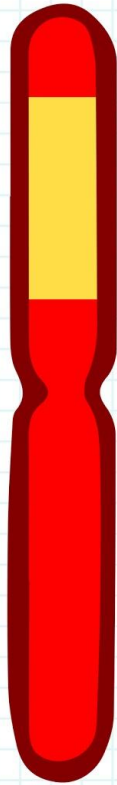
Brown

Blonde



Brown

Blonde



Blonde Blonde



Quiz!

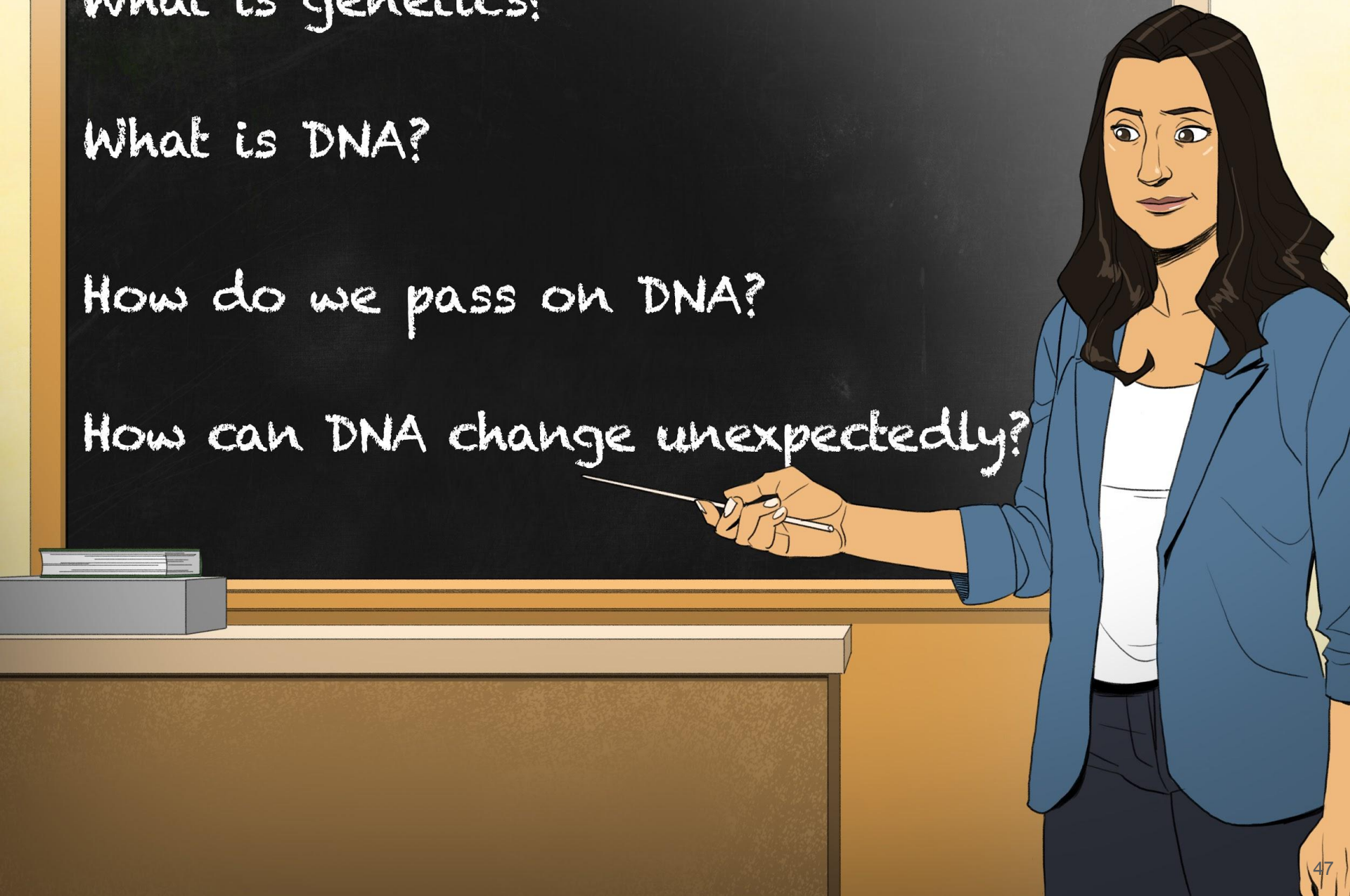
- How many chromosomes does a person typically have?
- How many chromosomes does an egg or sperm have?
- What does “replicate” mean?
- What does it mean if a gene is “dominant” or “recessive?”

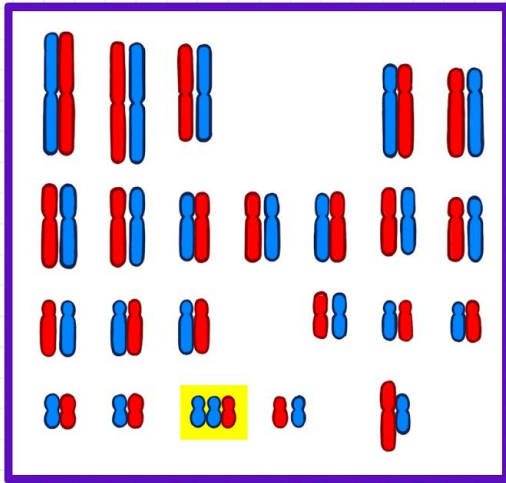
What is genetics?

What is DNA?

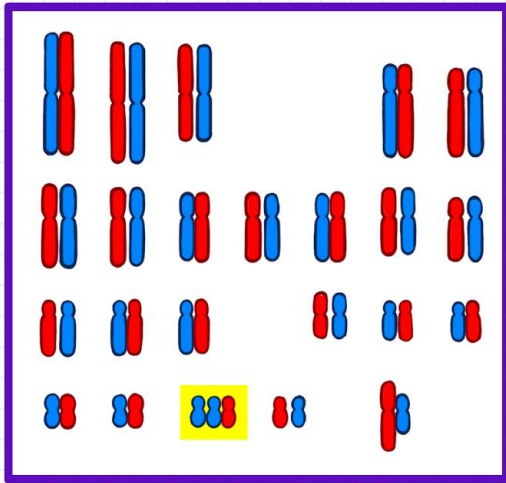
How do we pass on DNA?

How can DNA change unexpectedly?

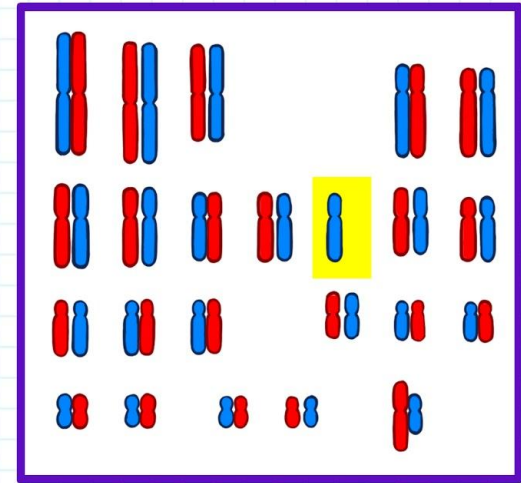




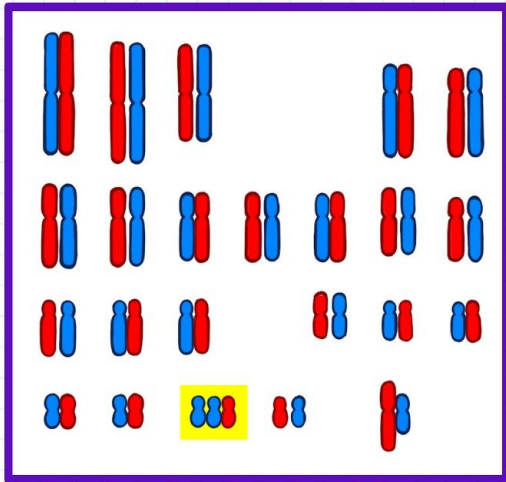
Extra chromosome



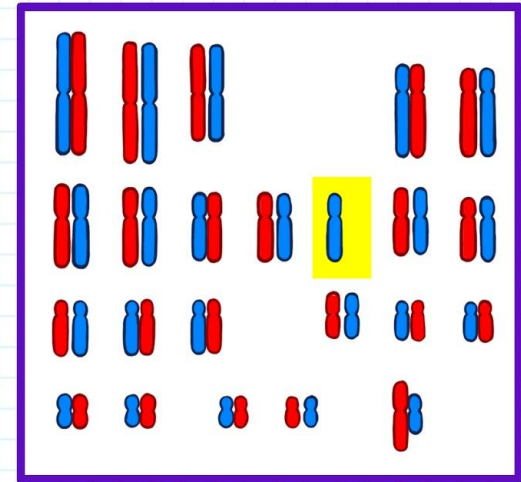
Extra chromosome



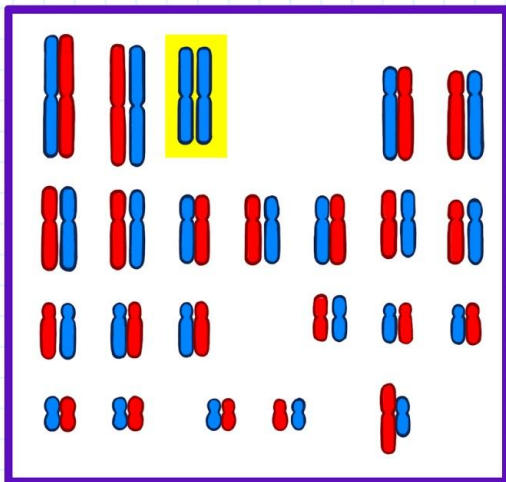
Missing chromosome



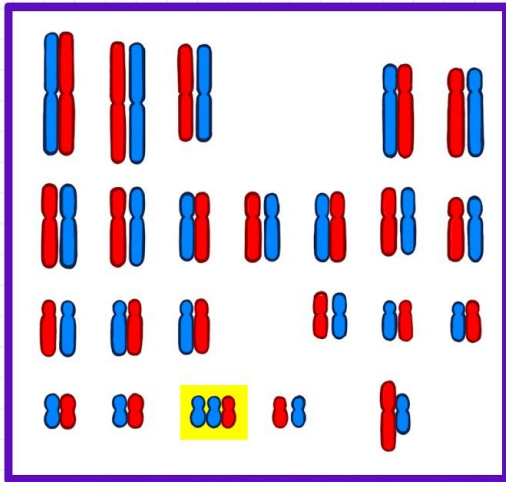
Extra chromosome



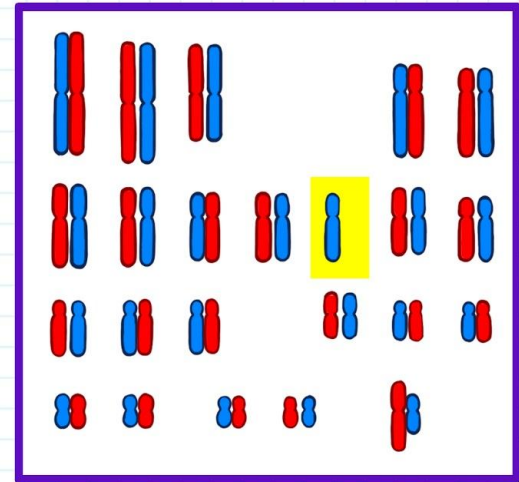
Missing chromosome



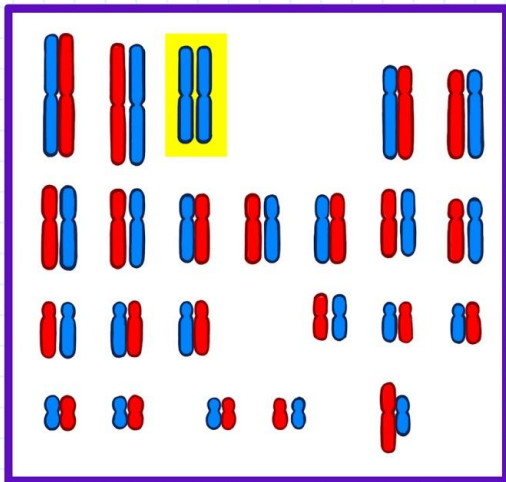
Pair from one parent



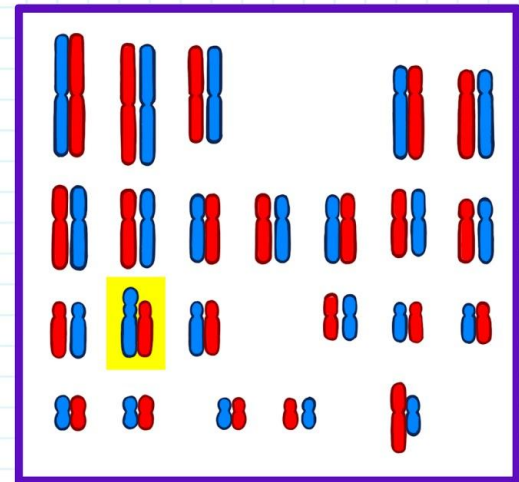
Extra chromosome



Missing chromosome



Pair from one parent



Fragment duplicated



**In genetics, a change at the gene-level
is called a variant.**



Change letters



Change letters

Change letters



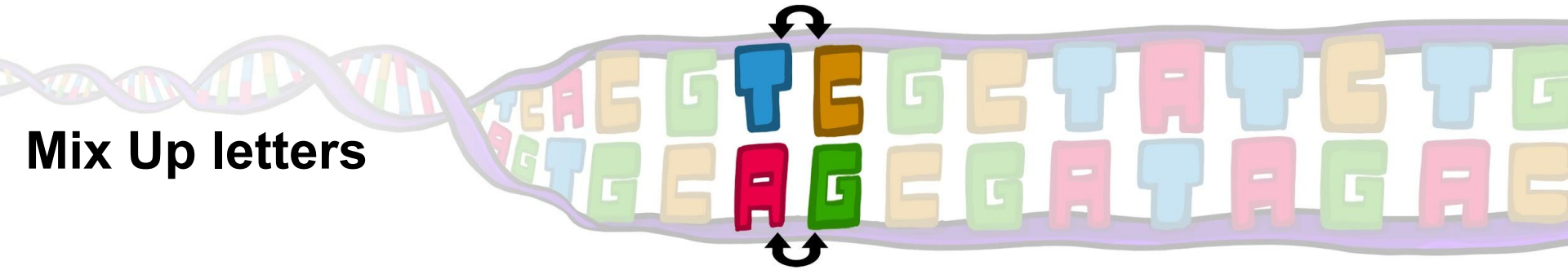
Mix Up letters



Change letters



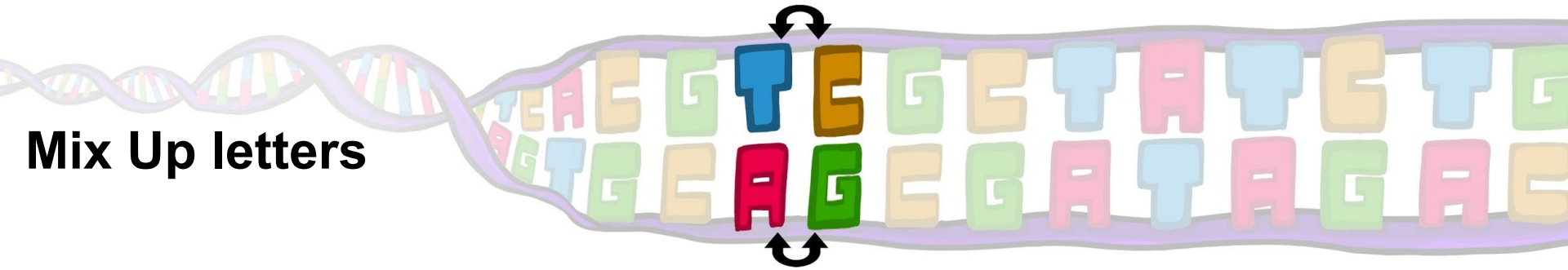
Mix Up letters



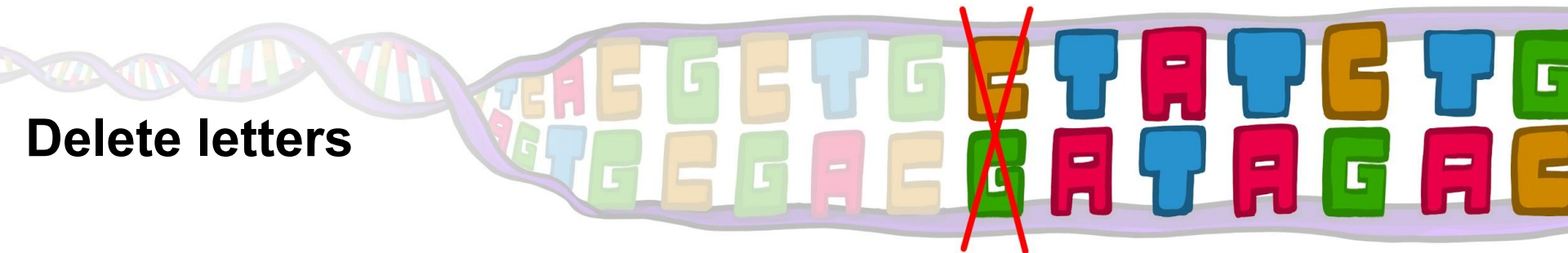
Change letters



Mix Up letters



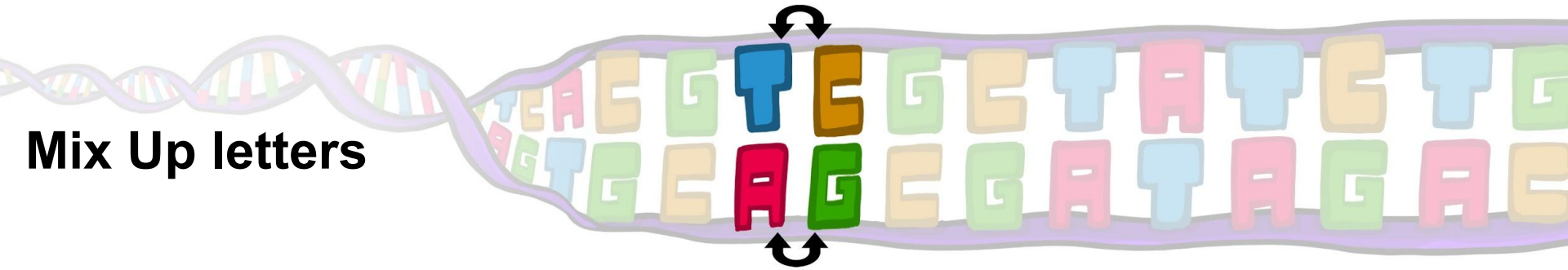
Delete letters



Change letters



Mix Up letters



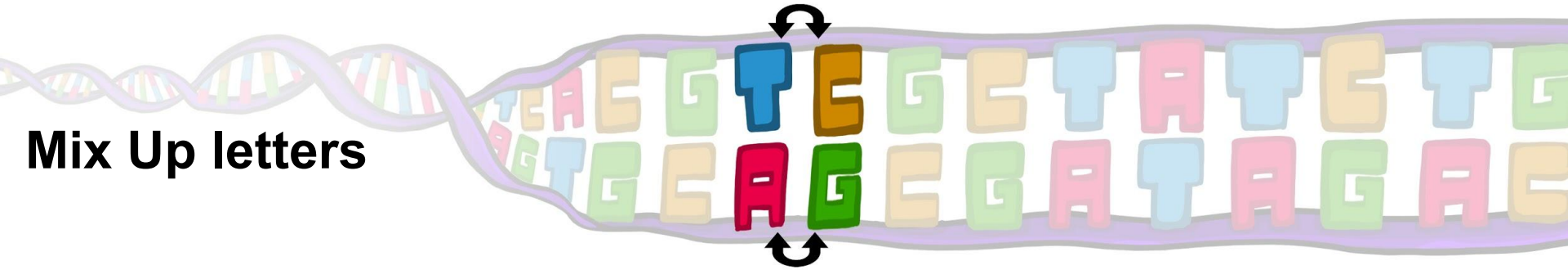
Delete letters



Change letters



Mix Up letters



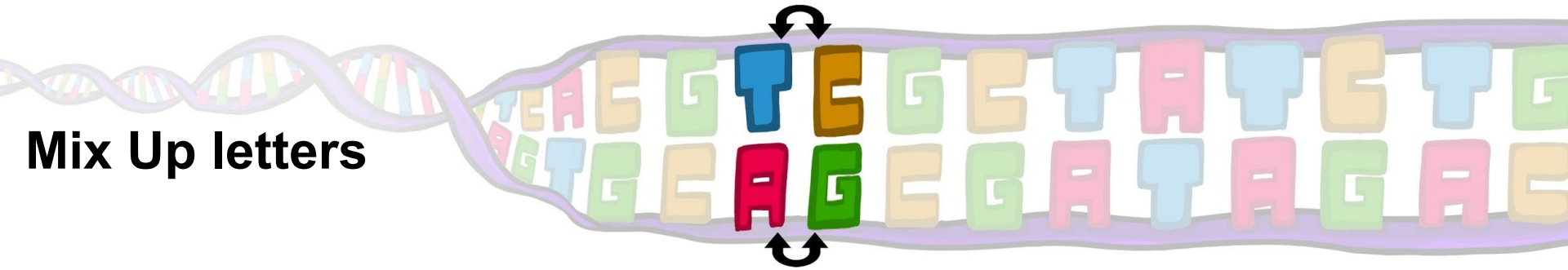
Delete letters



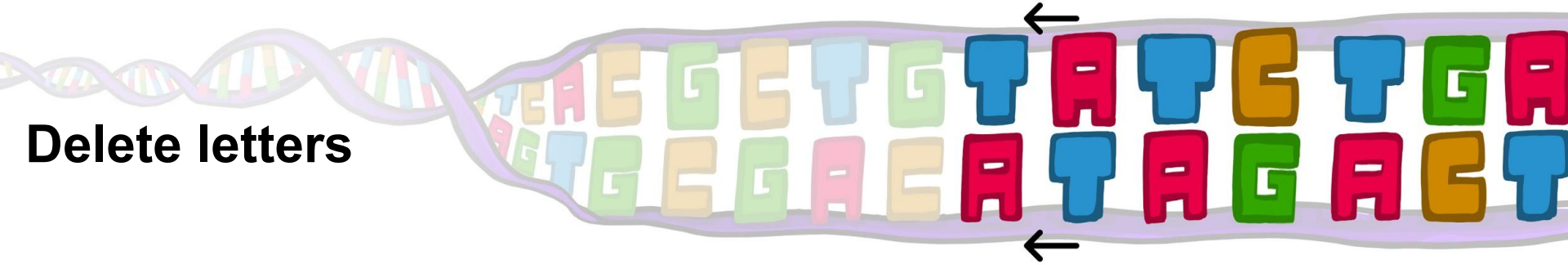
Change letters



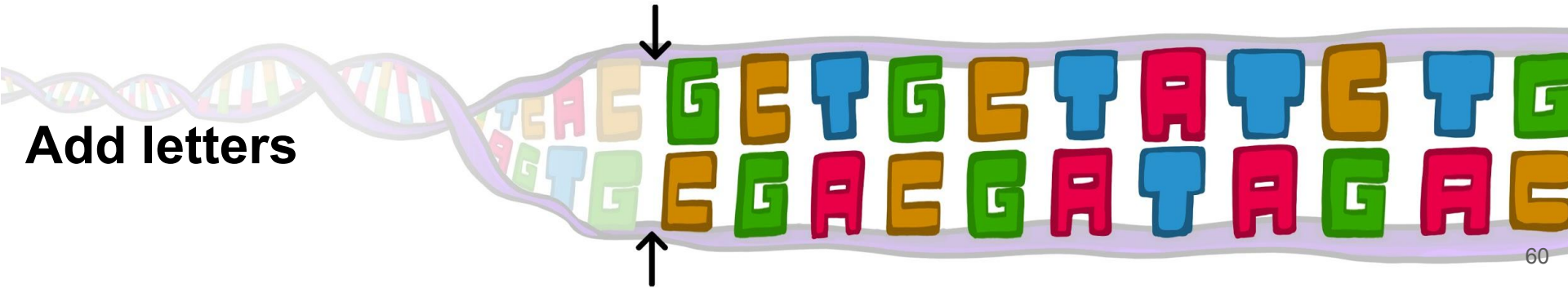
Mix Up letters



Delete letters



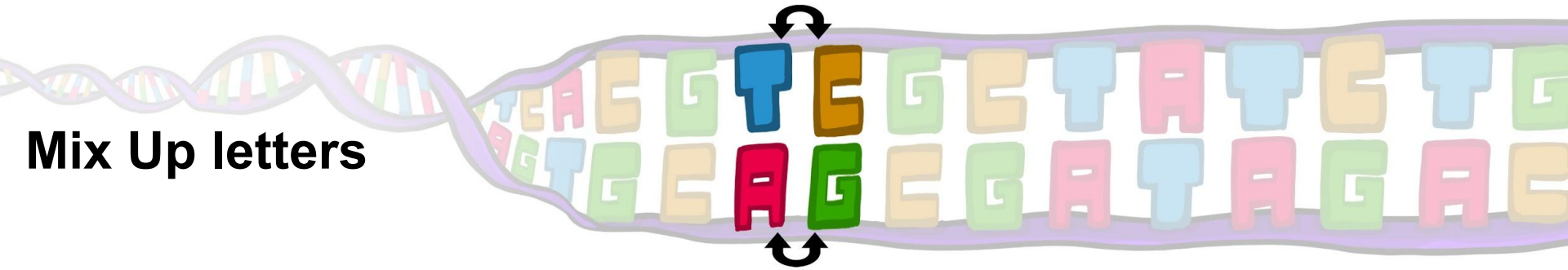
Add letters



Change letters



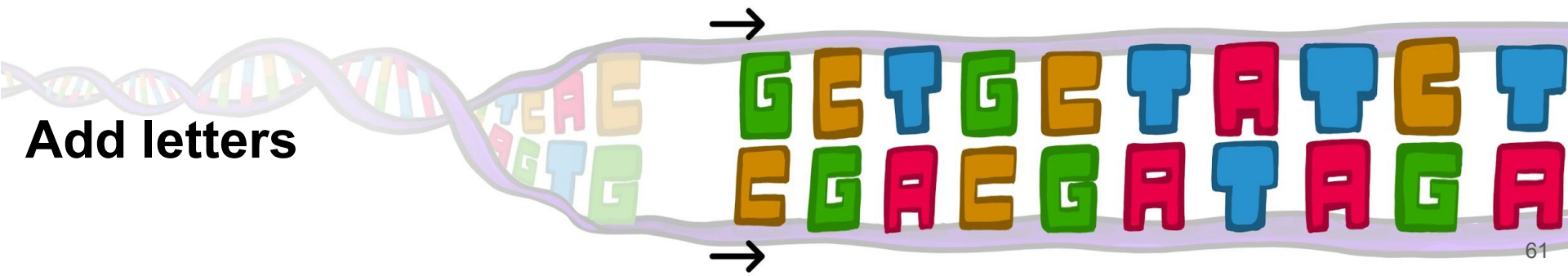
Mix Up letters



Delete letters



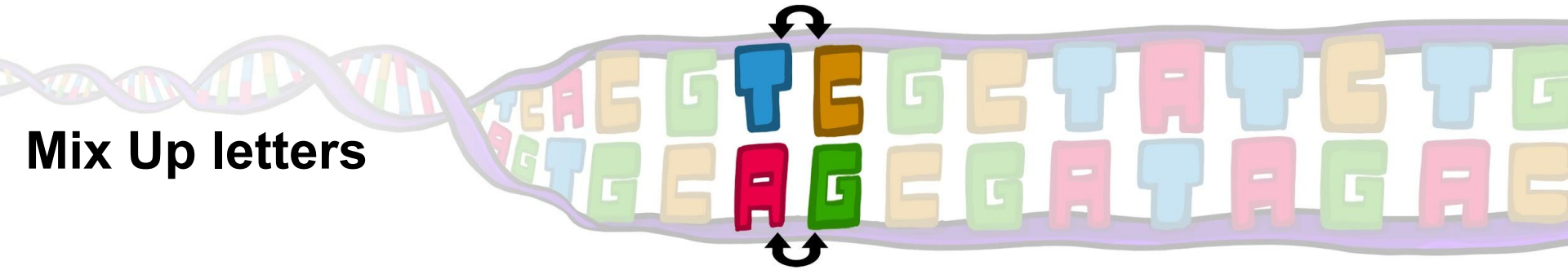
Add letters



Change letters



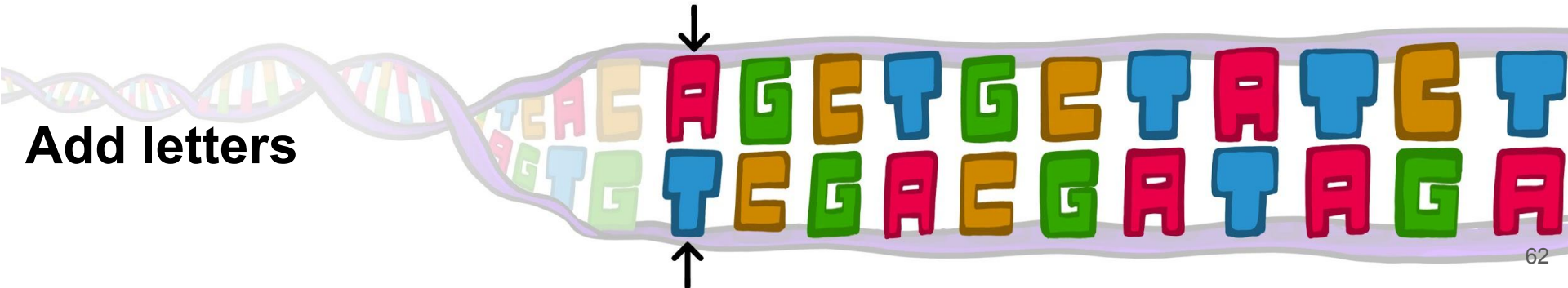
Mix Up letters

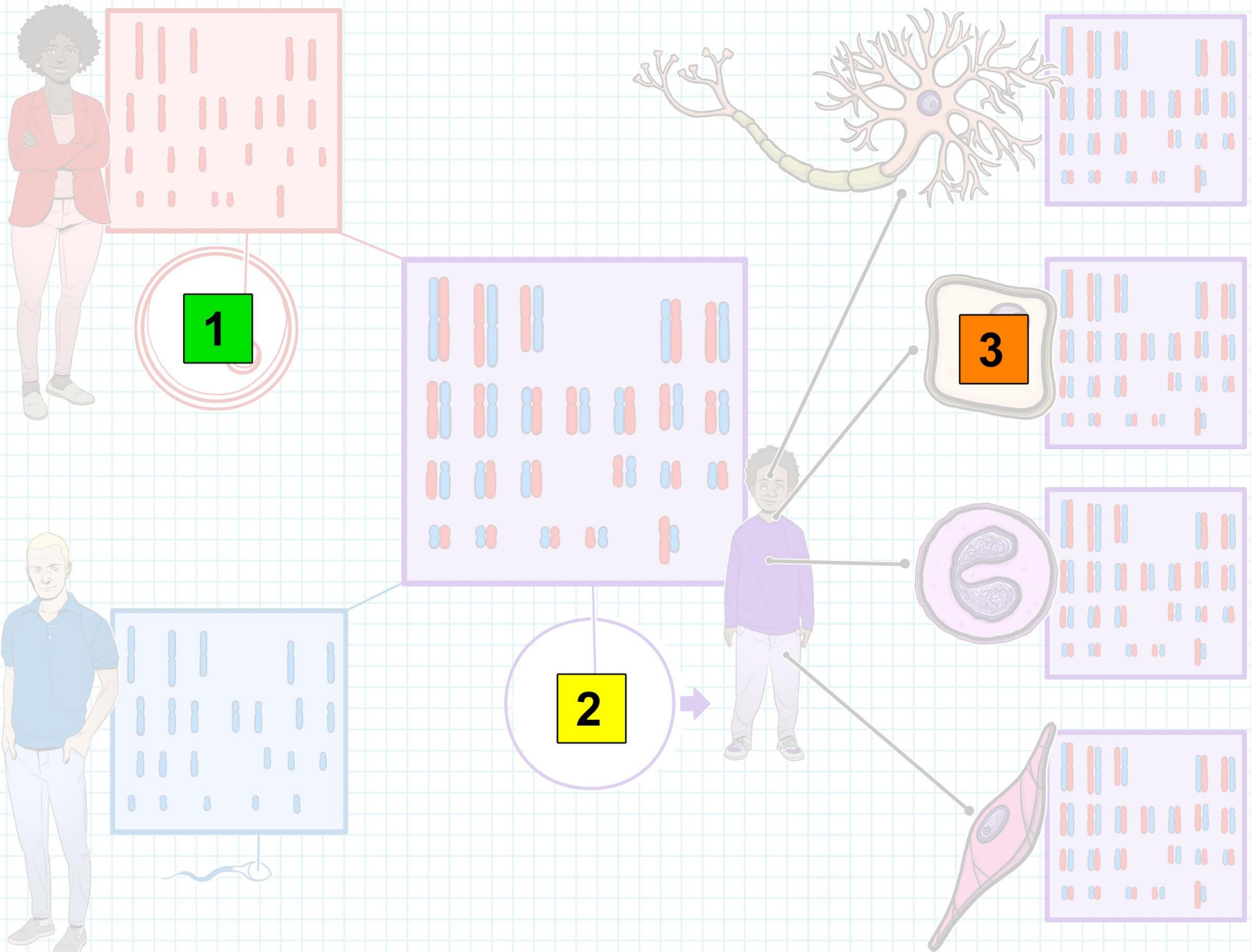


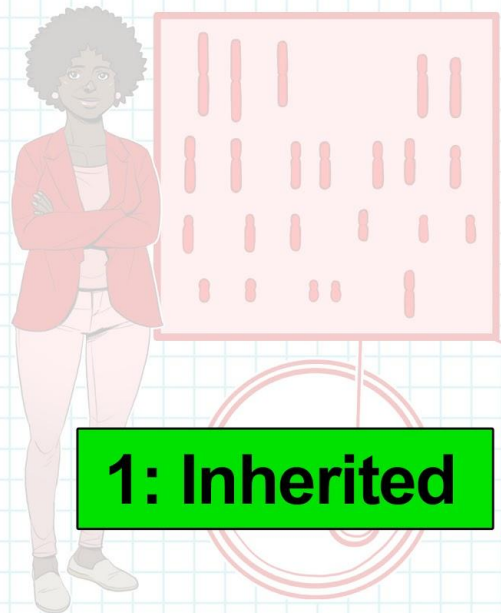
Delete letters



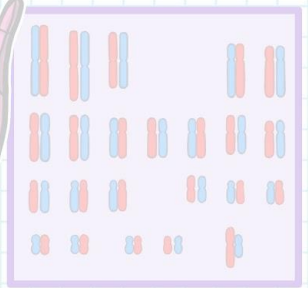
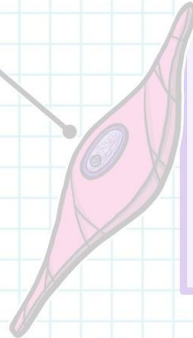
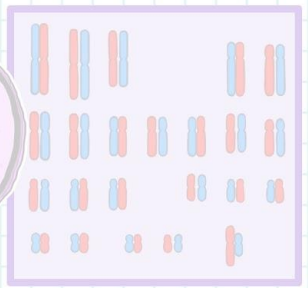
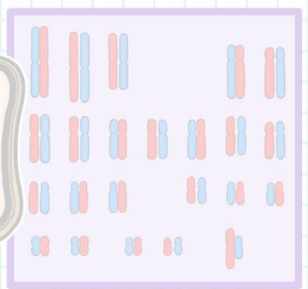
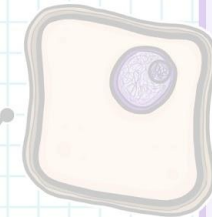
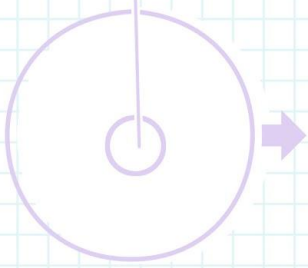
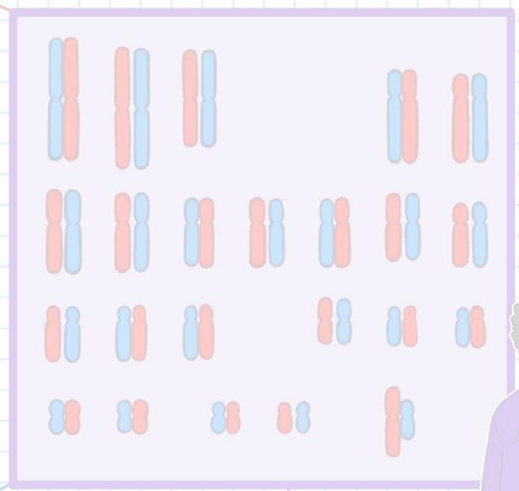
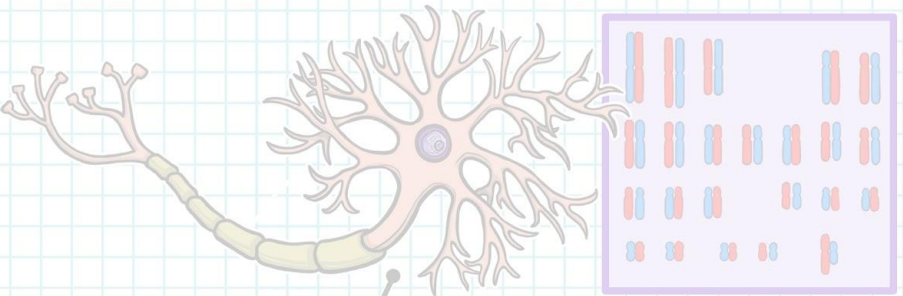
Add letters

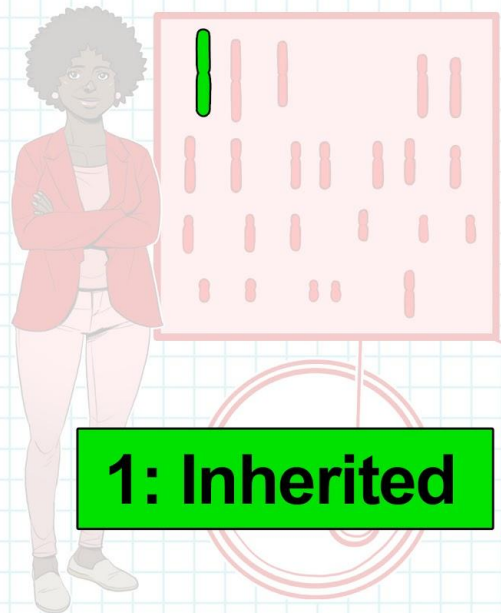




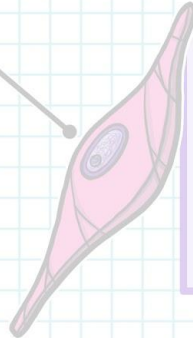
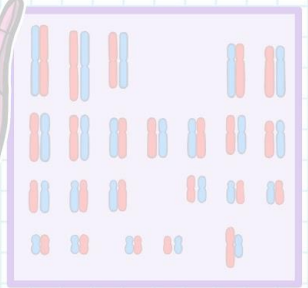
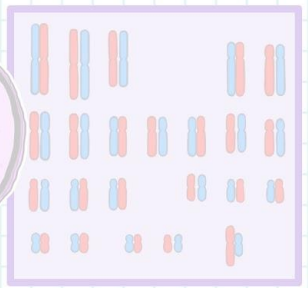
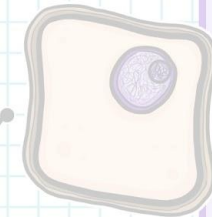
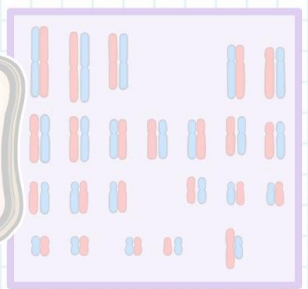
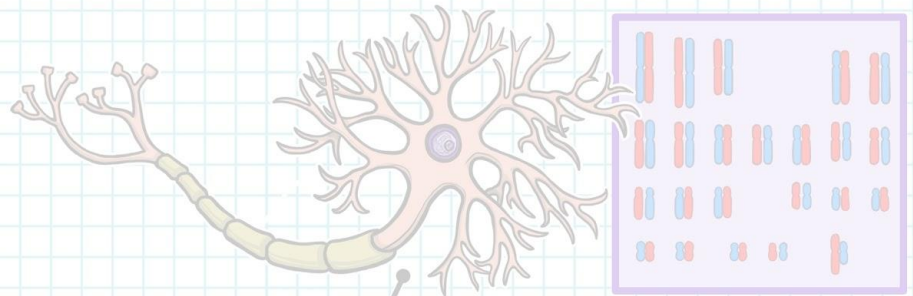
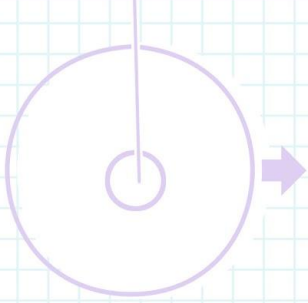
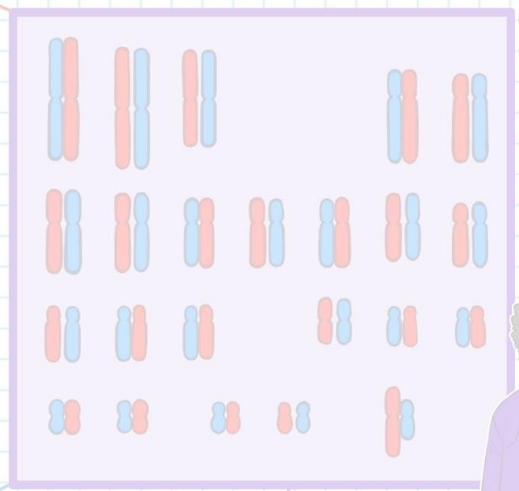


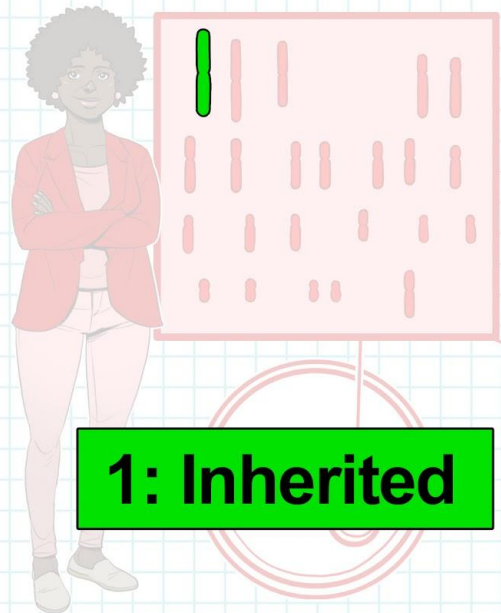
1: Inherited



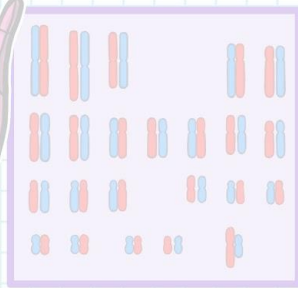
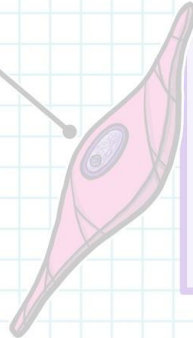
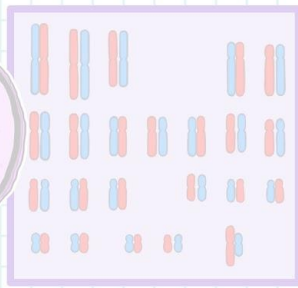
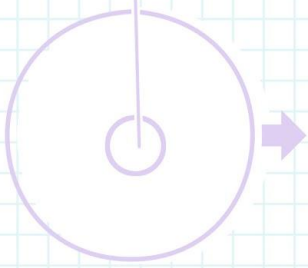
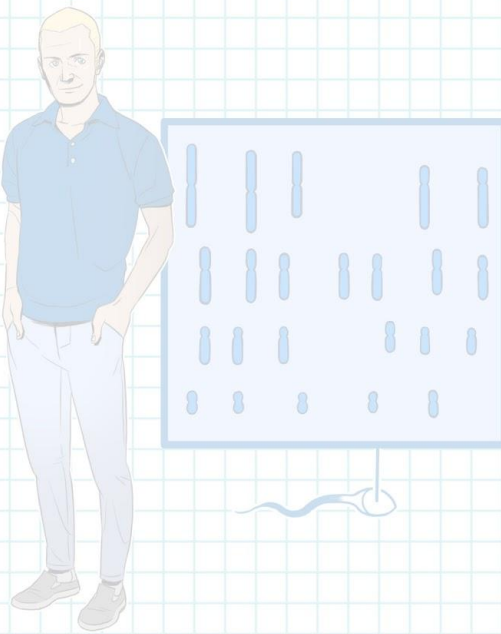
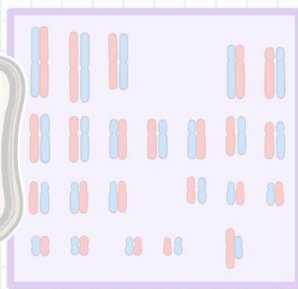
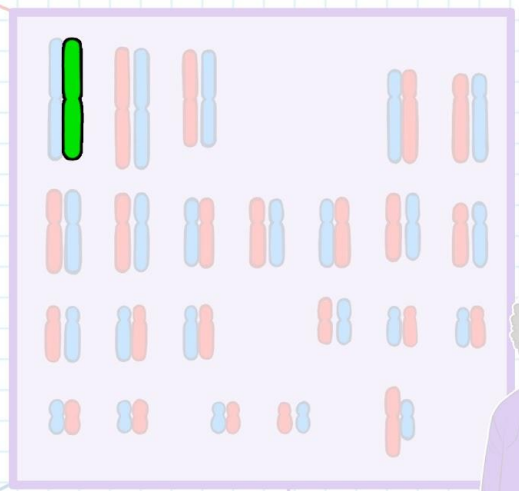
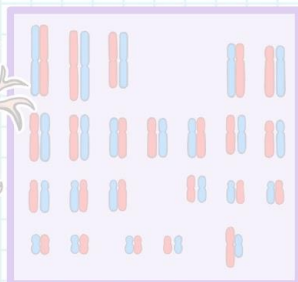
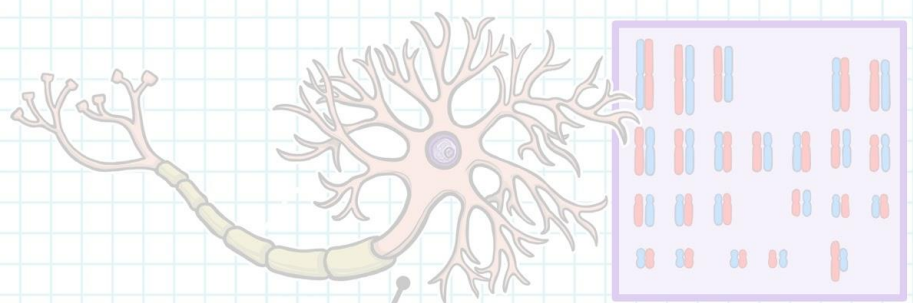


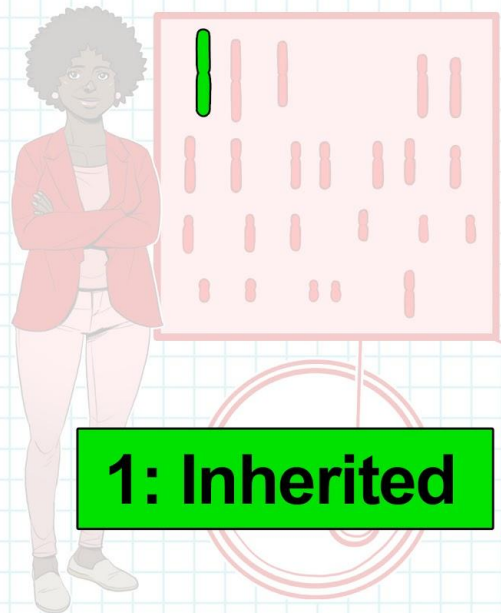
1: Inherited



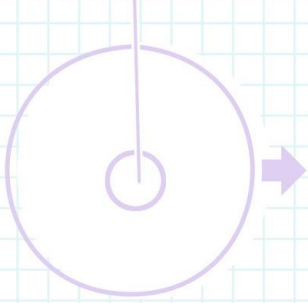
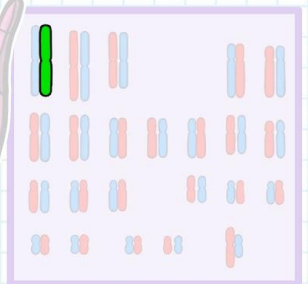
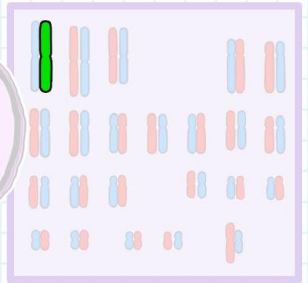
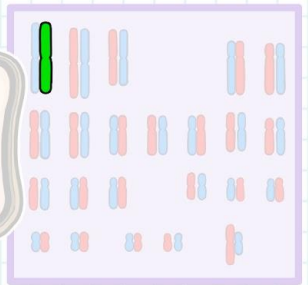
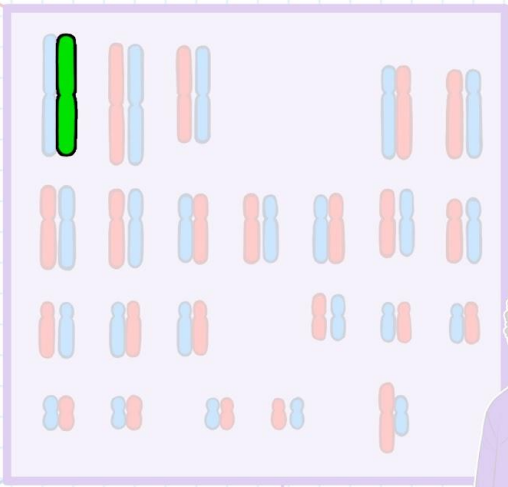
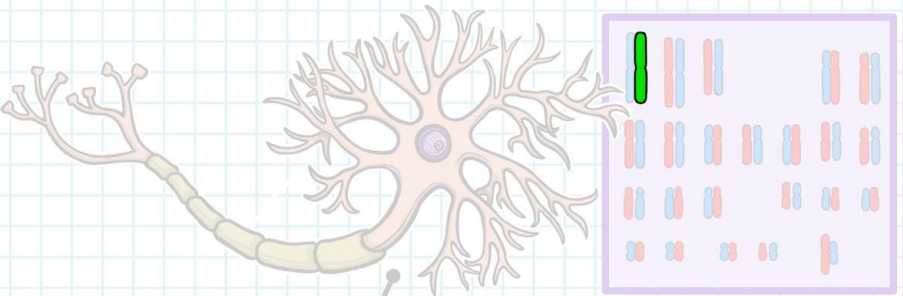


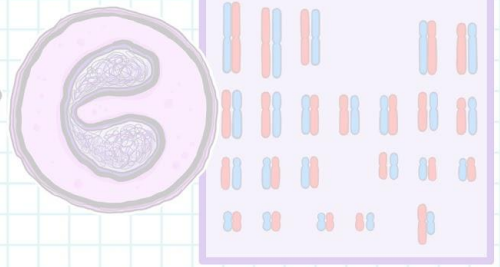
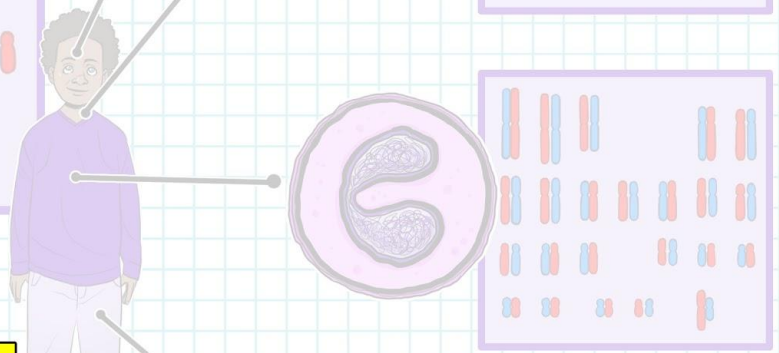
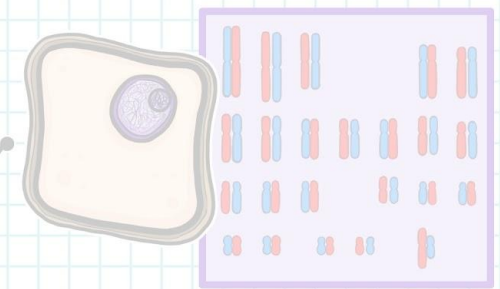
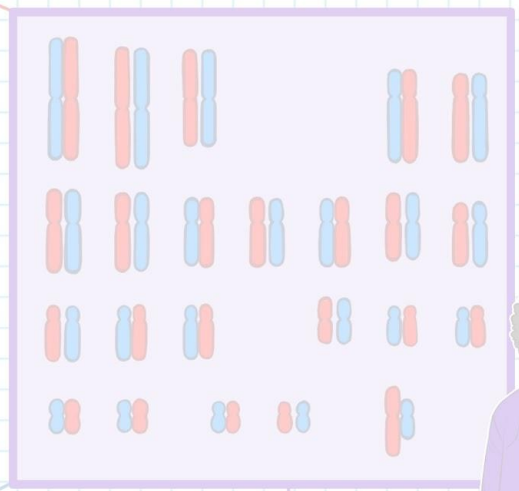
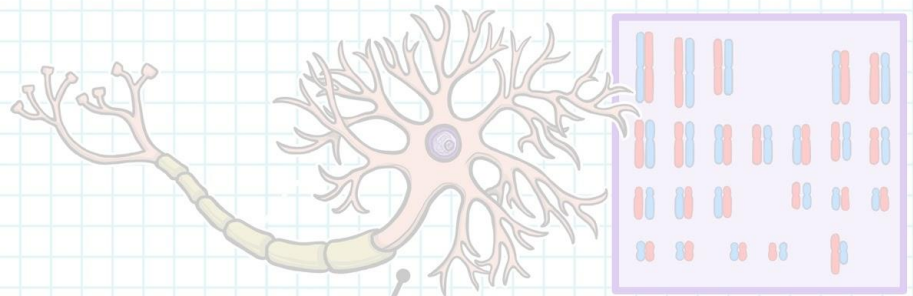
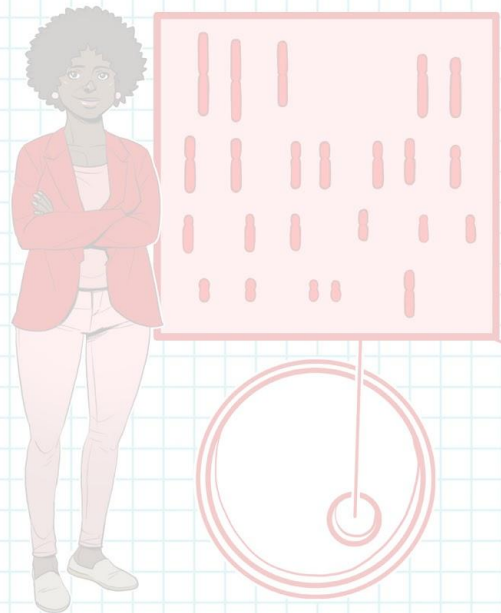
1: Inherited



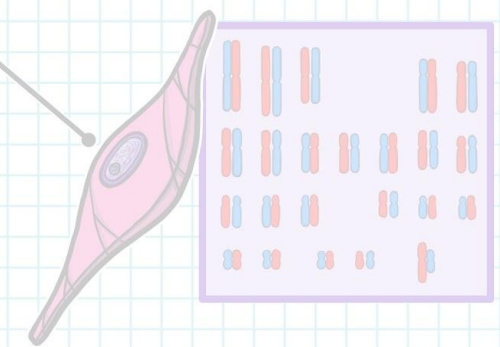


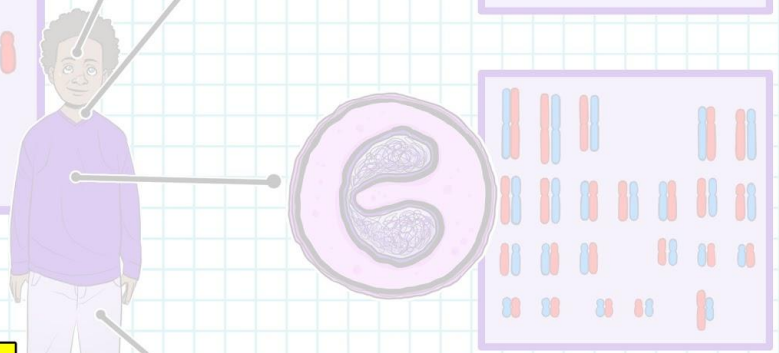
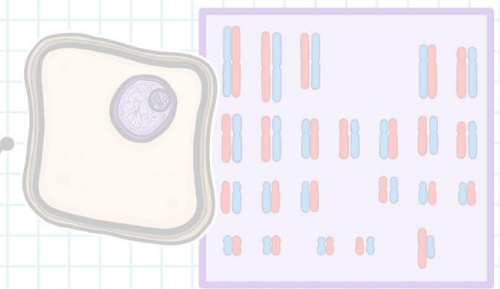
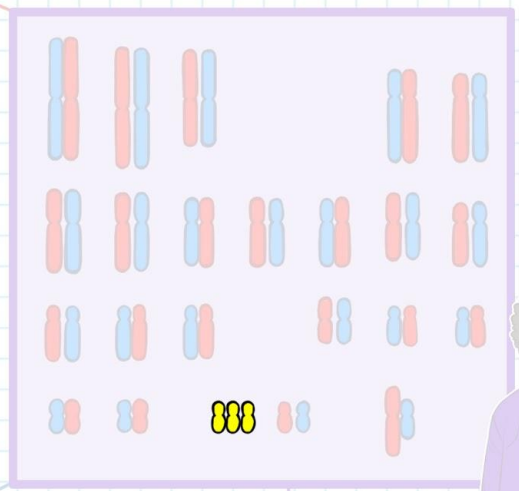
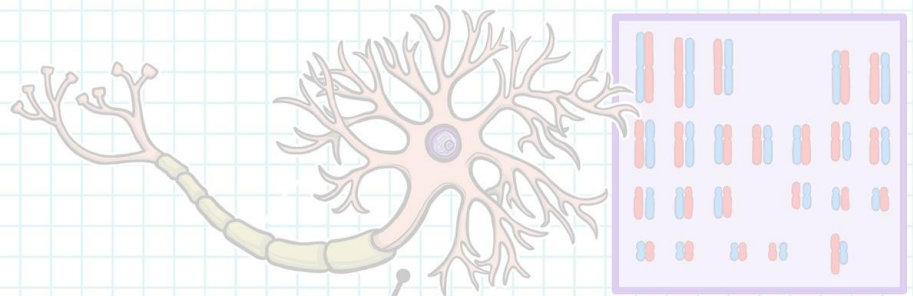
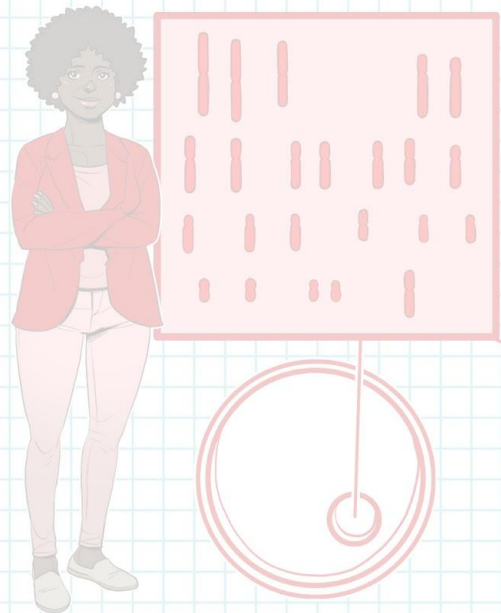
1: Inherited



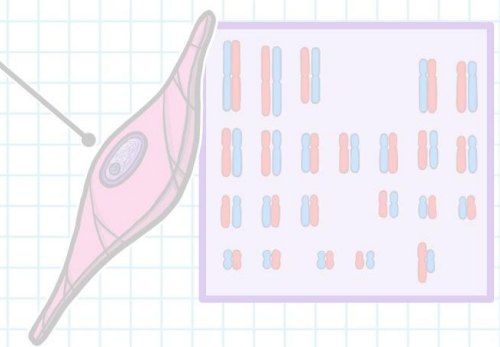


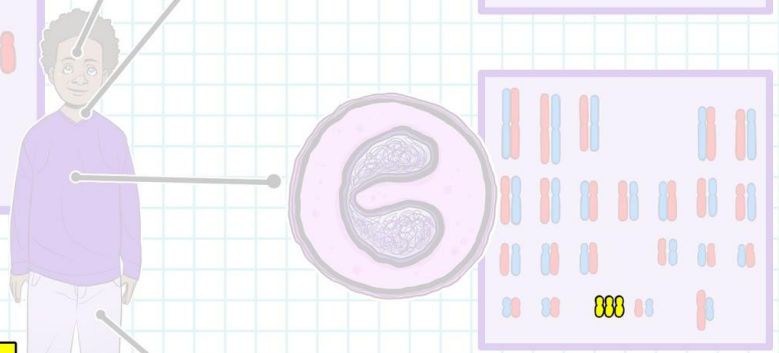
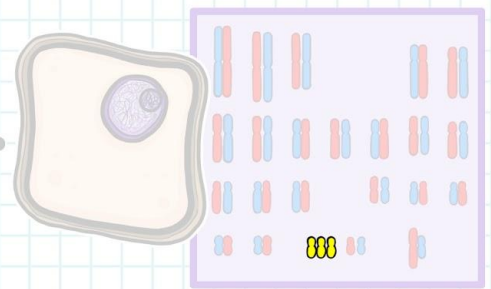
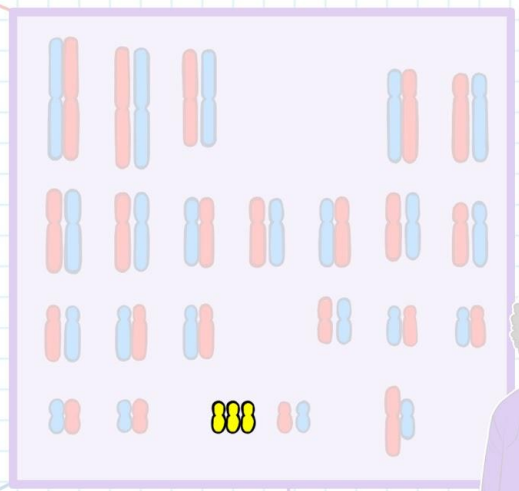
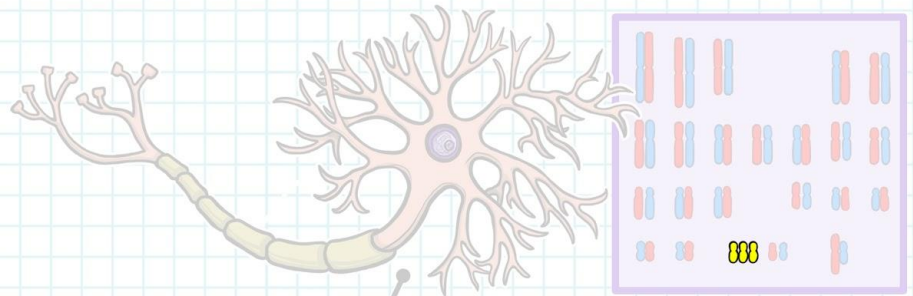
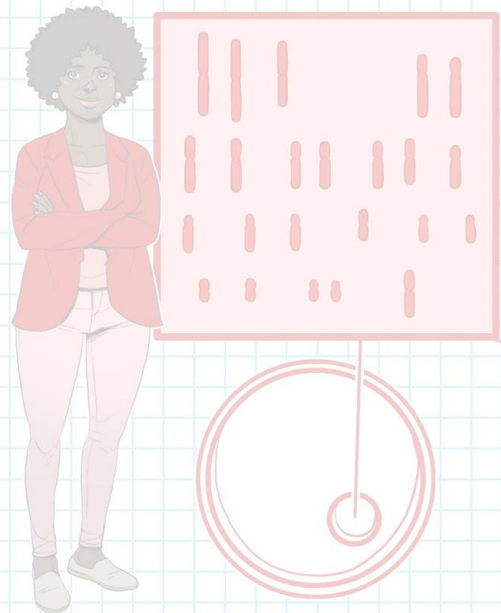
2: De Novo



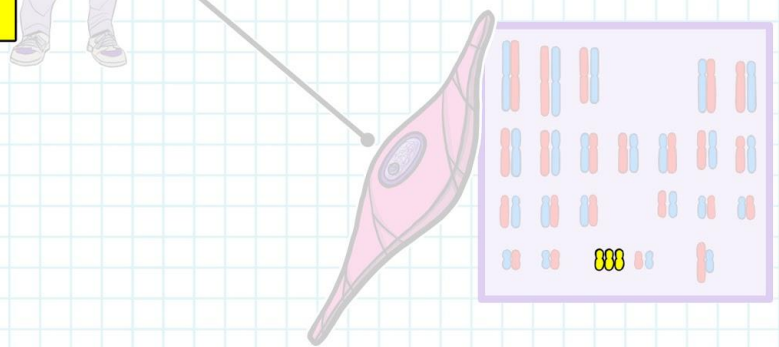


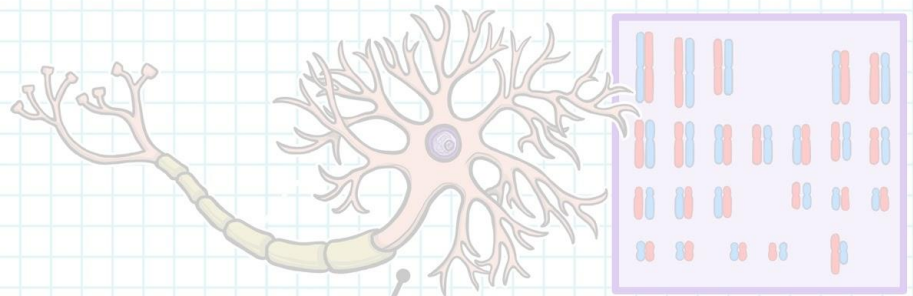
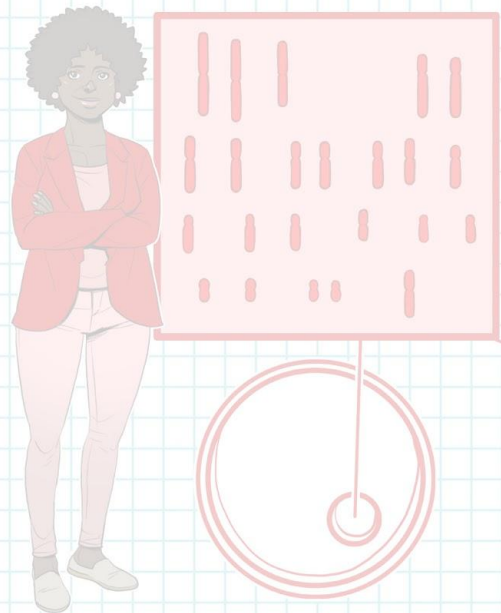
2: De Novo



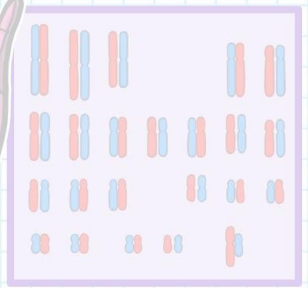
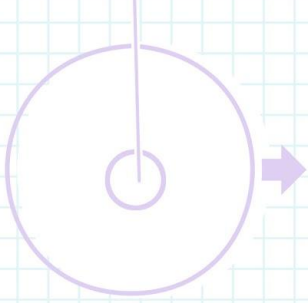
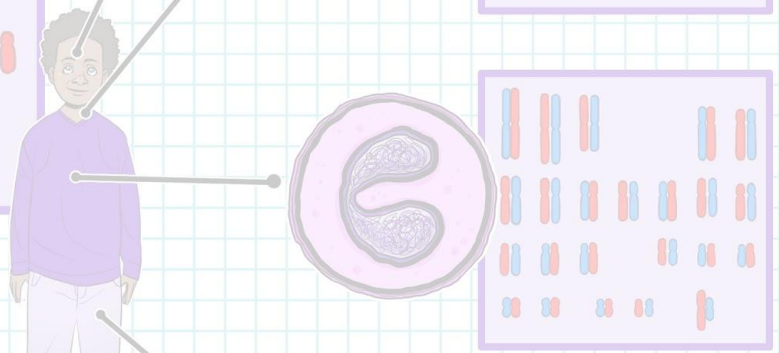
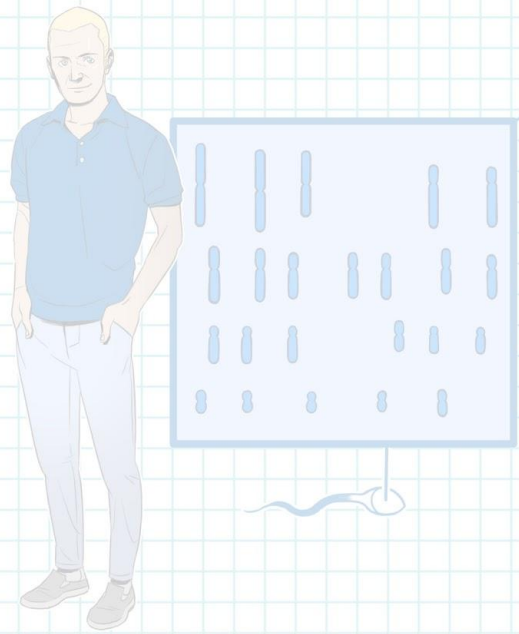
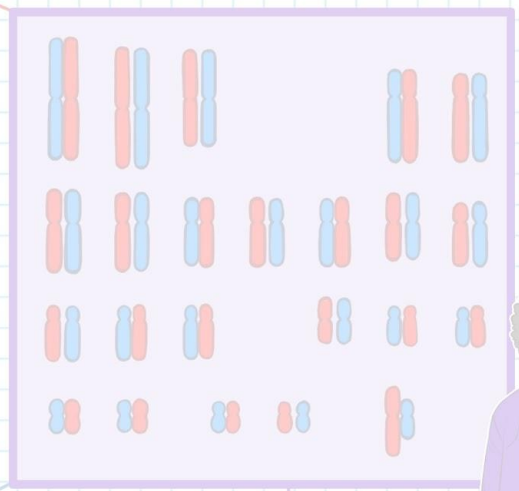


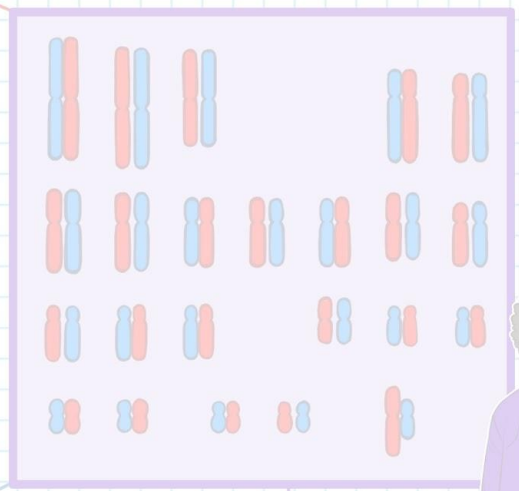
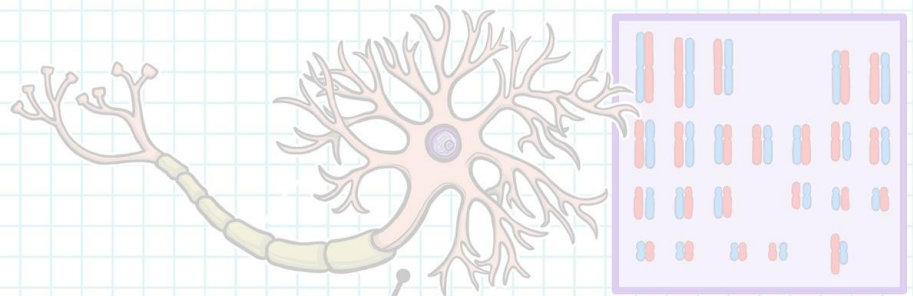
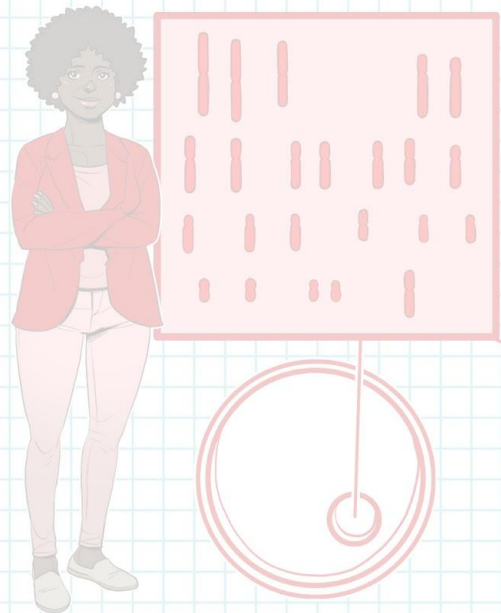
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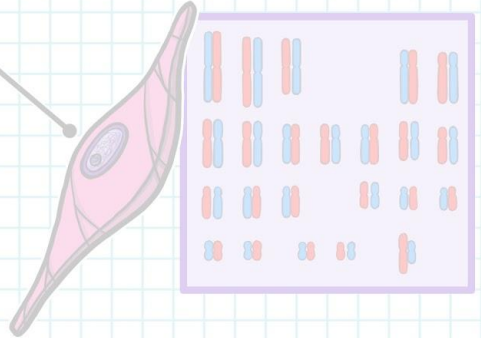
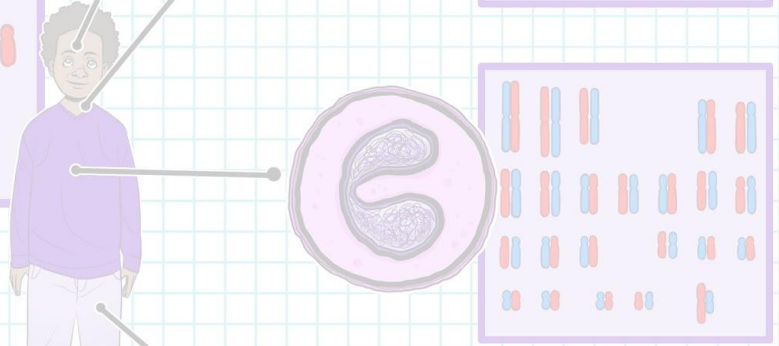
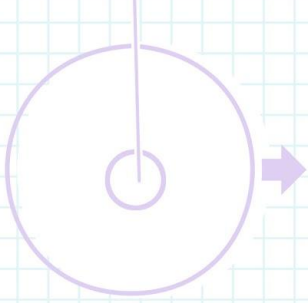
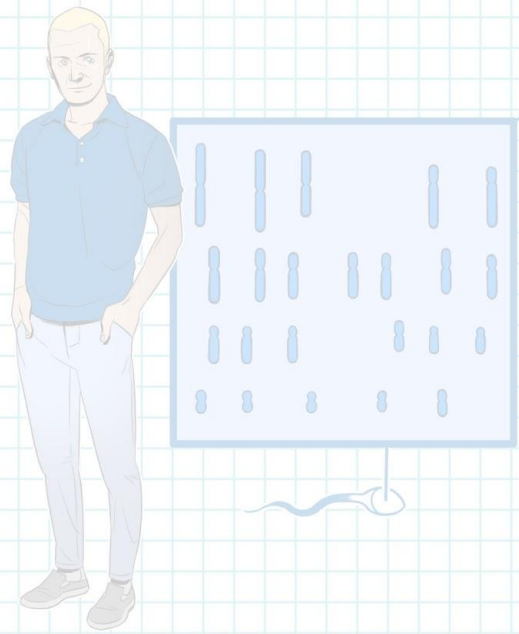


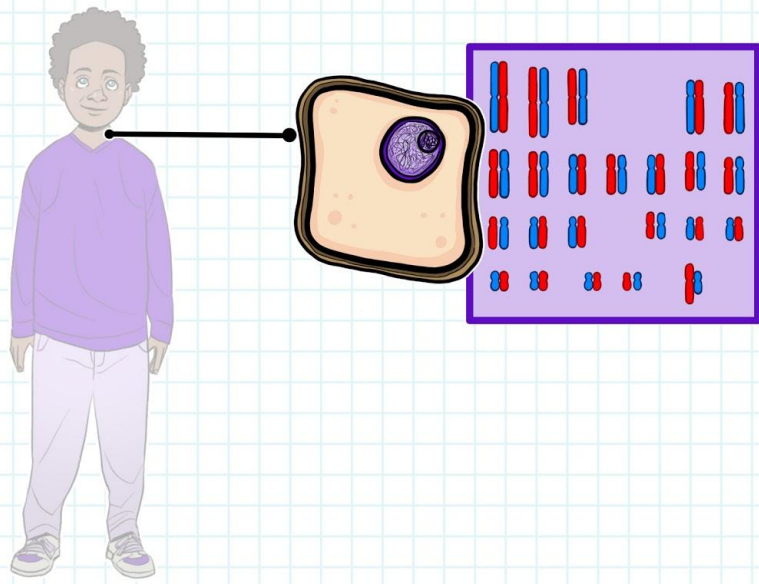
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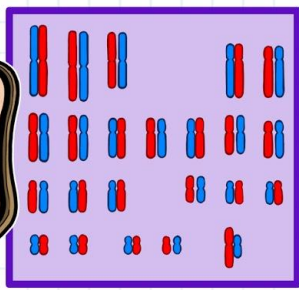
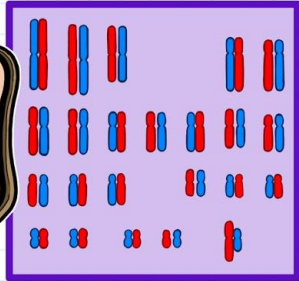
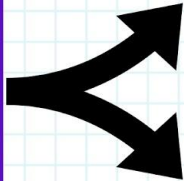
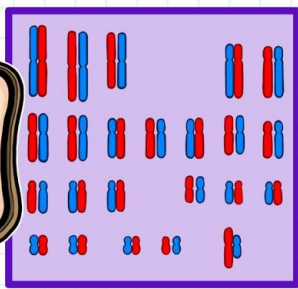


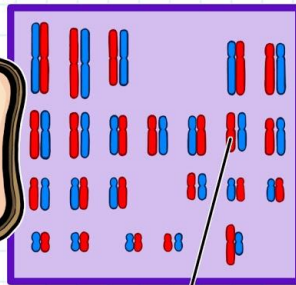
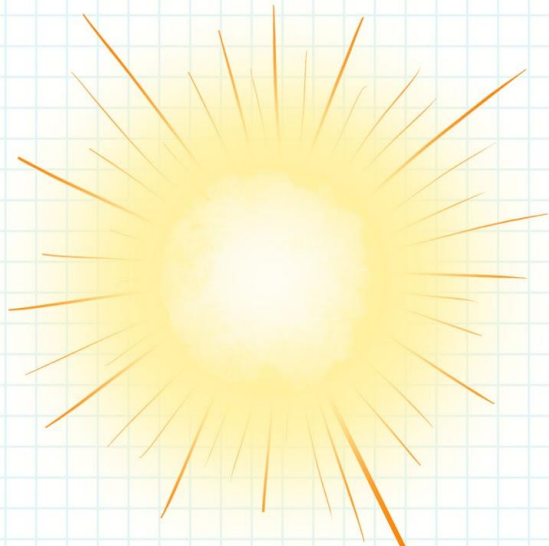


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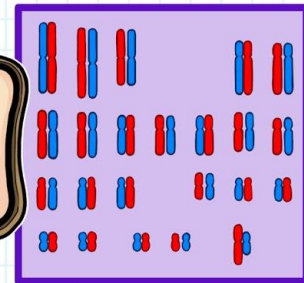
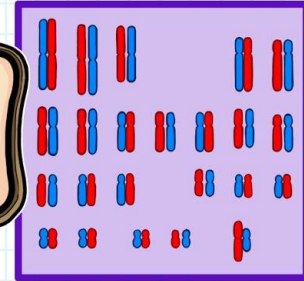
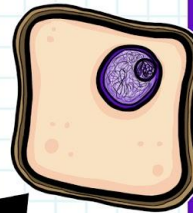
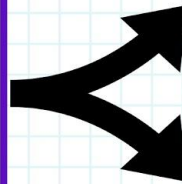


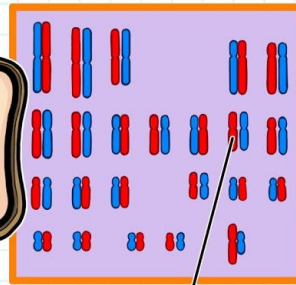
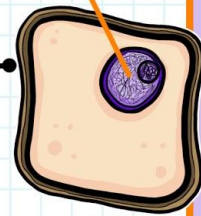
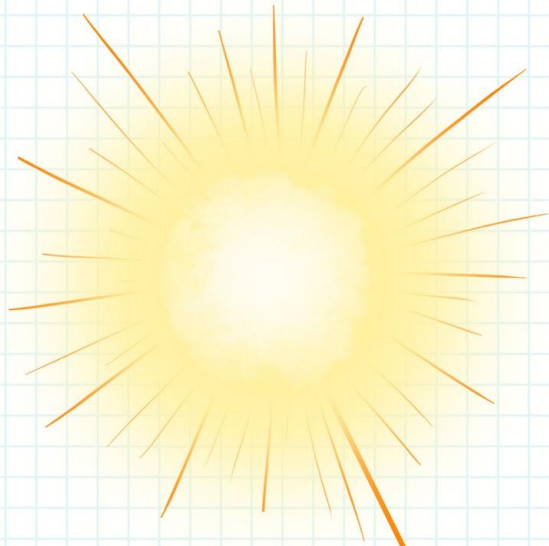




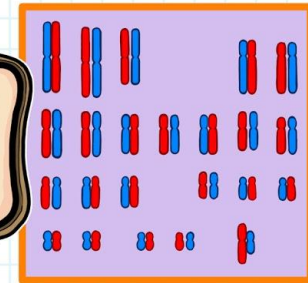
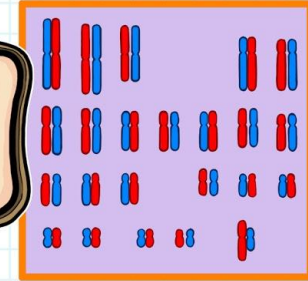
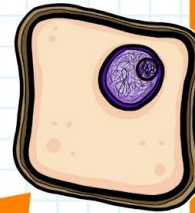
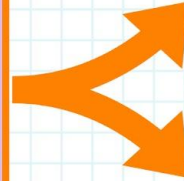


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AAGCTTATGCC
TCTGTGGTCTG
GCAGAGCAACC
TCTGATCGTC
TAGTTGCC
GGGT





TTGAC
GATCCAC
ATTTCGACACA
AAGCTTATGCC
TCTGTGGTCTG
GCAGAGCAACC
CTGATCGTC
TAGTTGCC
GGGT



“Benign”

Variant but no harm



“Benign”

Variant but no harm



“Deleterious” or “Pathogenic”

Variant may cause harm or increase risk



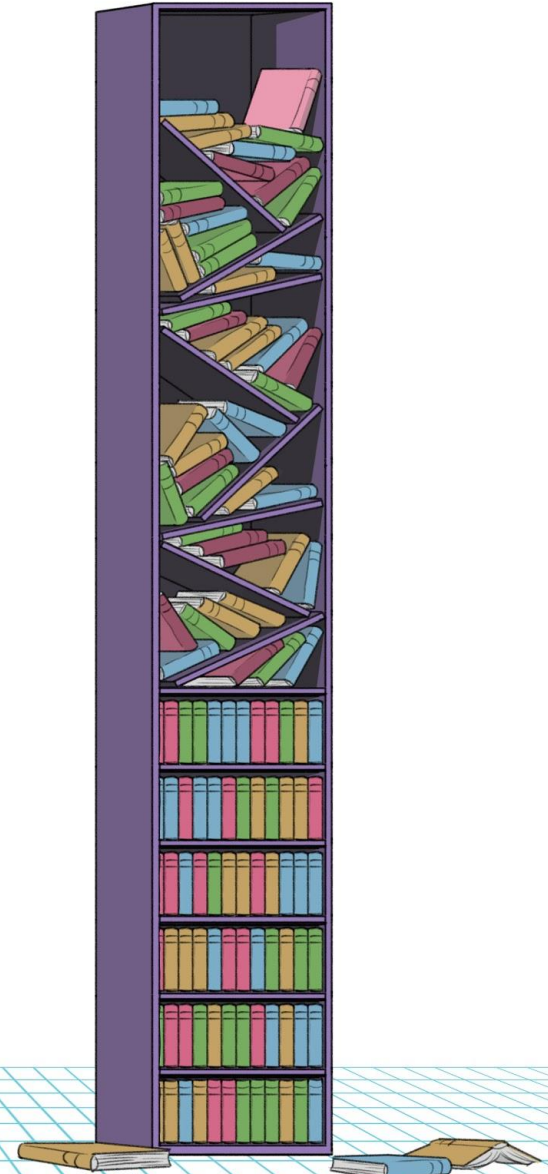
“Benign”

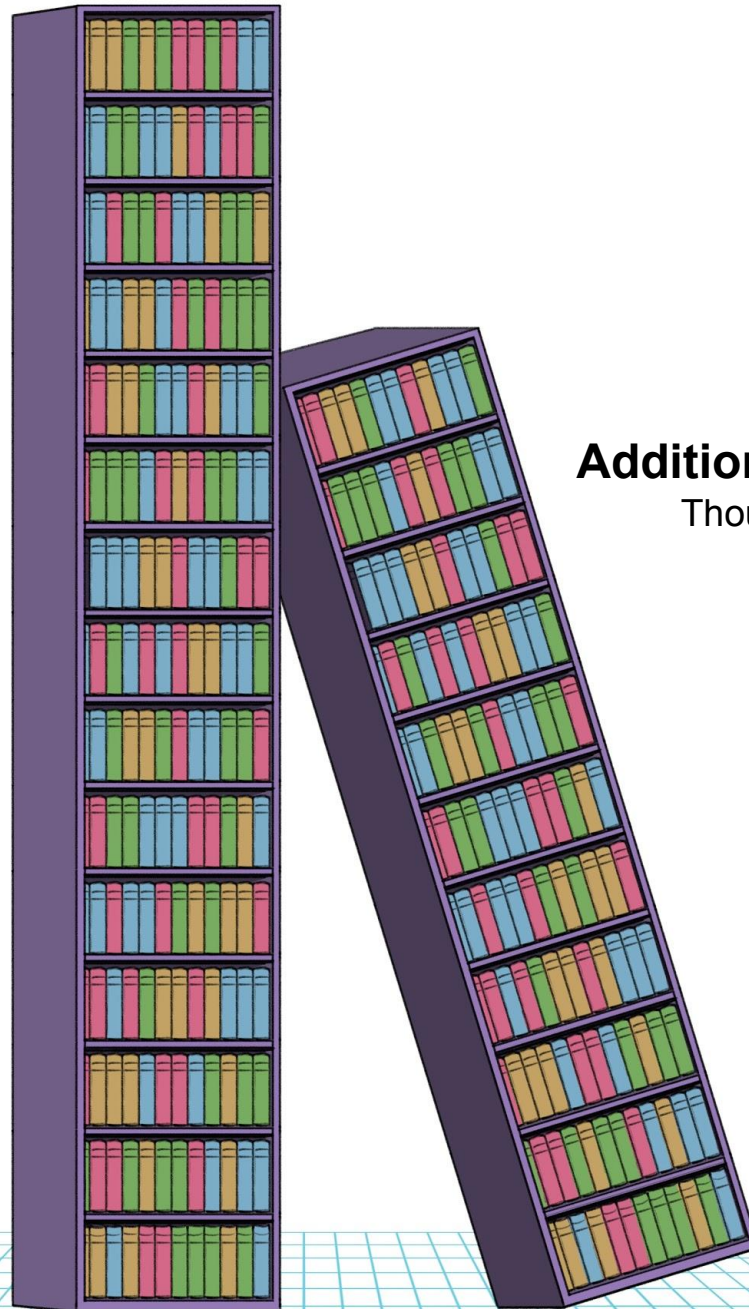
Variant but no harm



“Deleterious” or “Pathogenic”

Variant may cause harm or increase risk





Additional Chromosome
Thousands of genes!

Quiz!

- What is a genetic change at the chromosomal level called?
- What is a genetic change at the gene level called?
- What does it mean if a change is “benign”?
- What does it mean if a change is “deleterious?”

Got all that?





Medical
Center

Registration







Category	Item	Value	Unit
Patient Data	Name	John Doe	
	Age	45	Years
	Gender	Male	
	Weight	180	Lbs
Vital Signs	BP	120/80	mmHg
	HR	75	Beats/min
	Temp	98.6	F
Lab Results	Hemoglobin	14	g/dL
	Glucose	100	mg/dL



My job as a genetic counselor is to help people understand their risk for conditions that may run in families, like cancer sometimes does.



My job as a genetic counselor is to help people understand their risk for conditions that may run in families, like cancer sometimes does.

I'll take a health history for you, and for your family. Then I'll tell you about genetic testing, if it seems like that might be helpful.

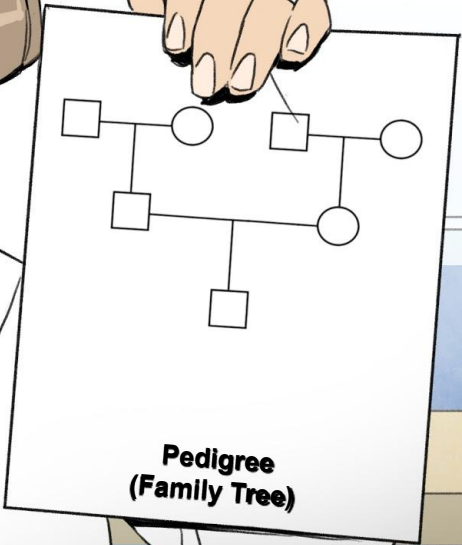


My job as a genetic counselor is to help people understand their risk for conditions that may run in families, like cancer sometimes does.

I'll take a health history for you, and for your family. Then I'll tell you about genetic testing, if it seems like that might be helpful.

And if you DO decide to get tested, I'll be here afterward to explain the results and to help you decide what to do next.





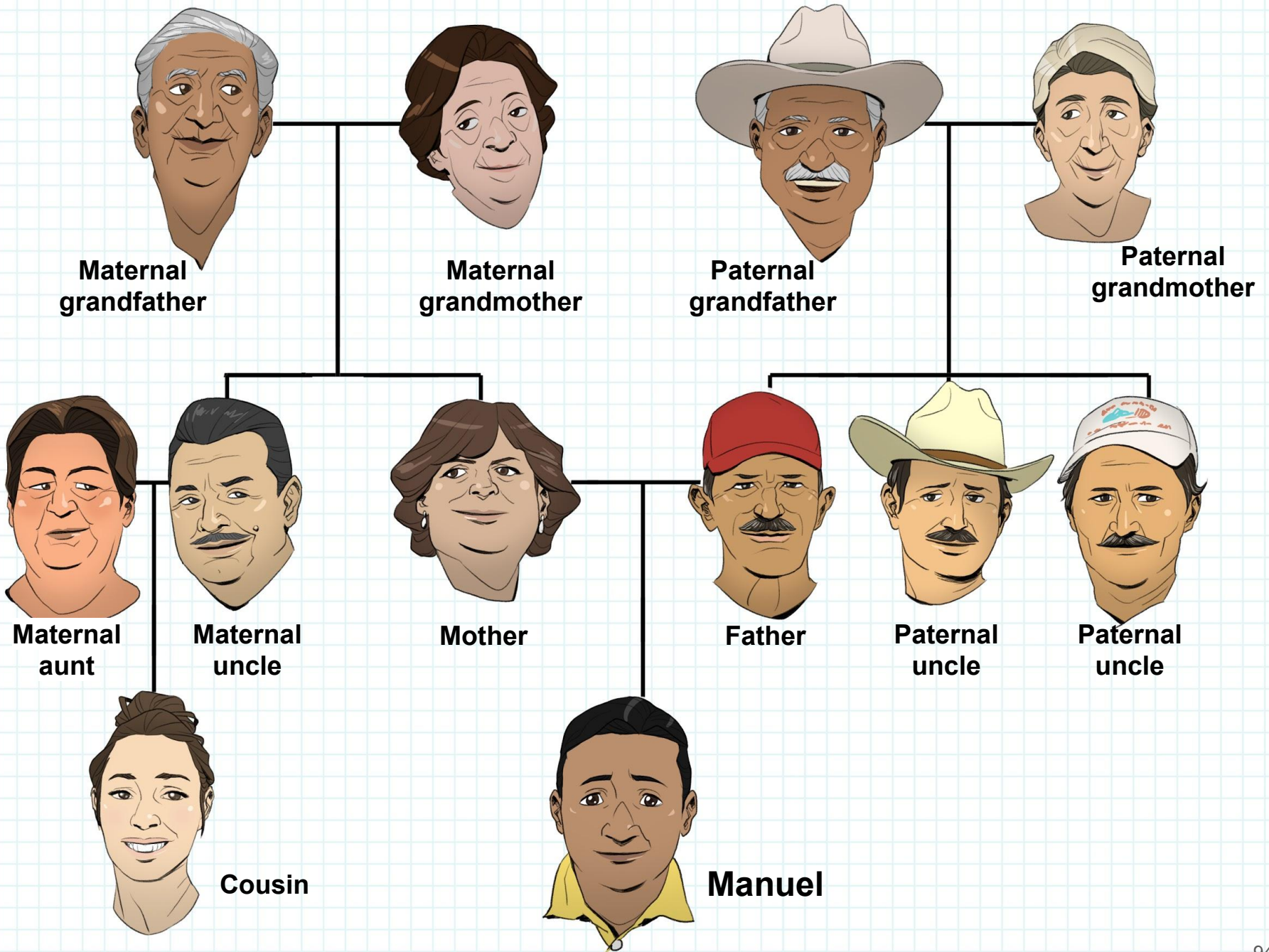
I don't know what my dad's parents died of. I know my grandmother on my mom's side had some kind of cancer, but I don't know what kind.

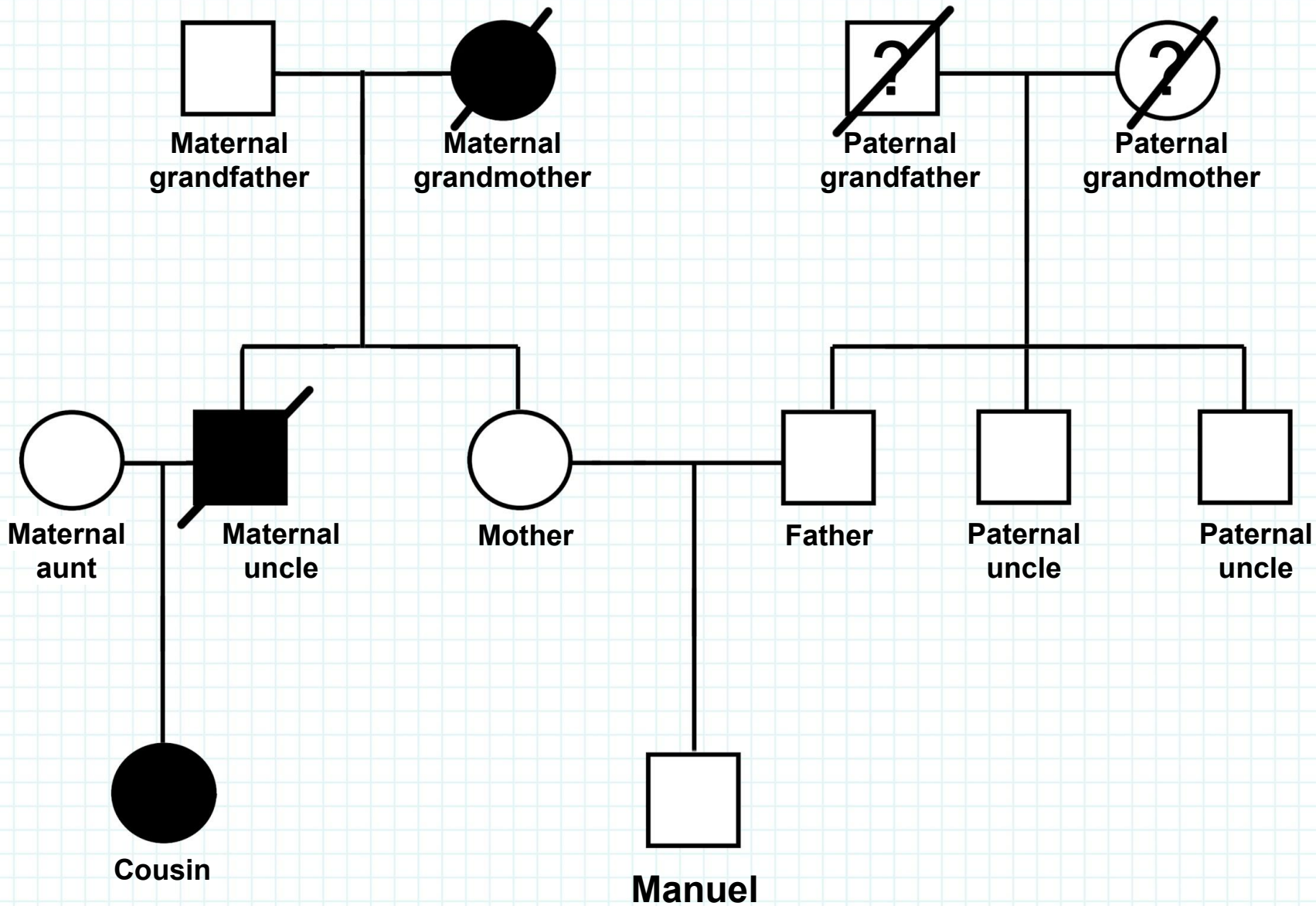



I don't know what my dad's parents died of. I know my grandmother on my mom's side had some kind of cancer, but I don't know what kind.

I guess I could ask. But half the family is back in Mexico, and we don't like to talk about things like this.







A man with dark hair, wearing a white button-down shirt, is shown from the chest up. He has a thoughtful or questioning expression, with his hands clasped together in front of him. The background is a simple blue wall with a window on the right and a green plant on the left.

But what does this have to do with genetics?

It's just bad luck, right?

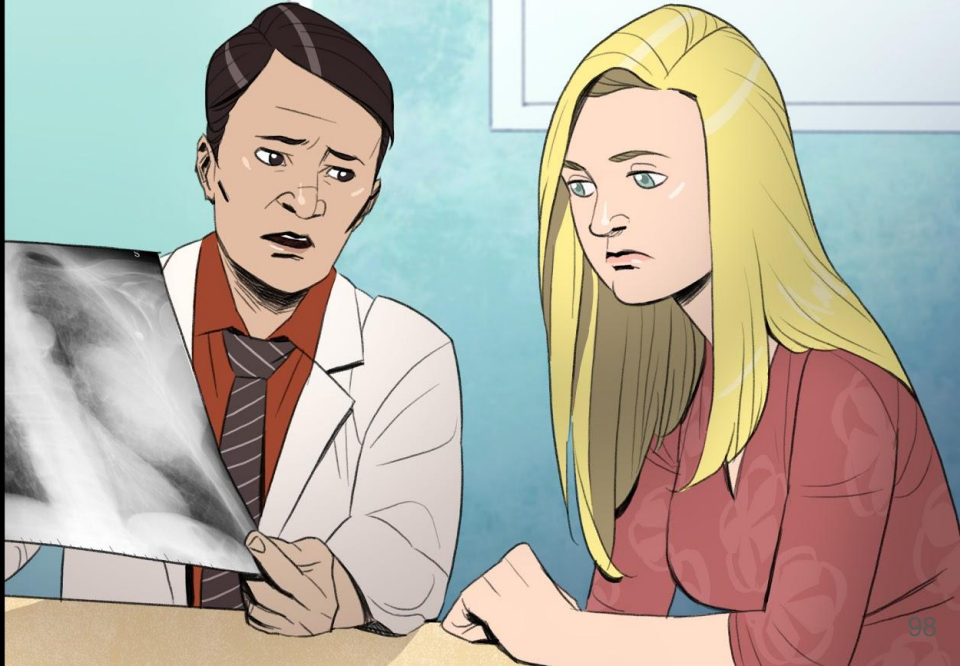
Environmental causes or random mutations



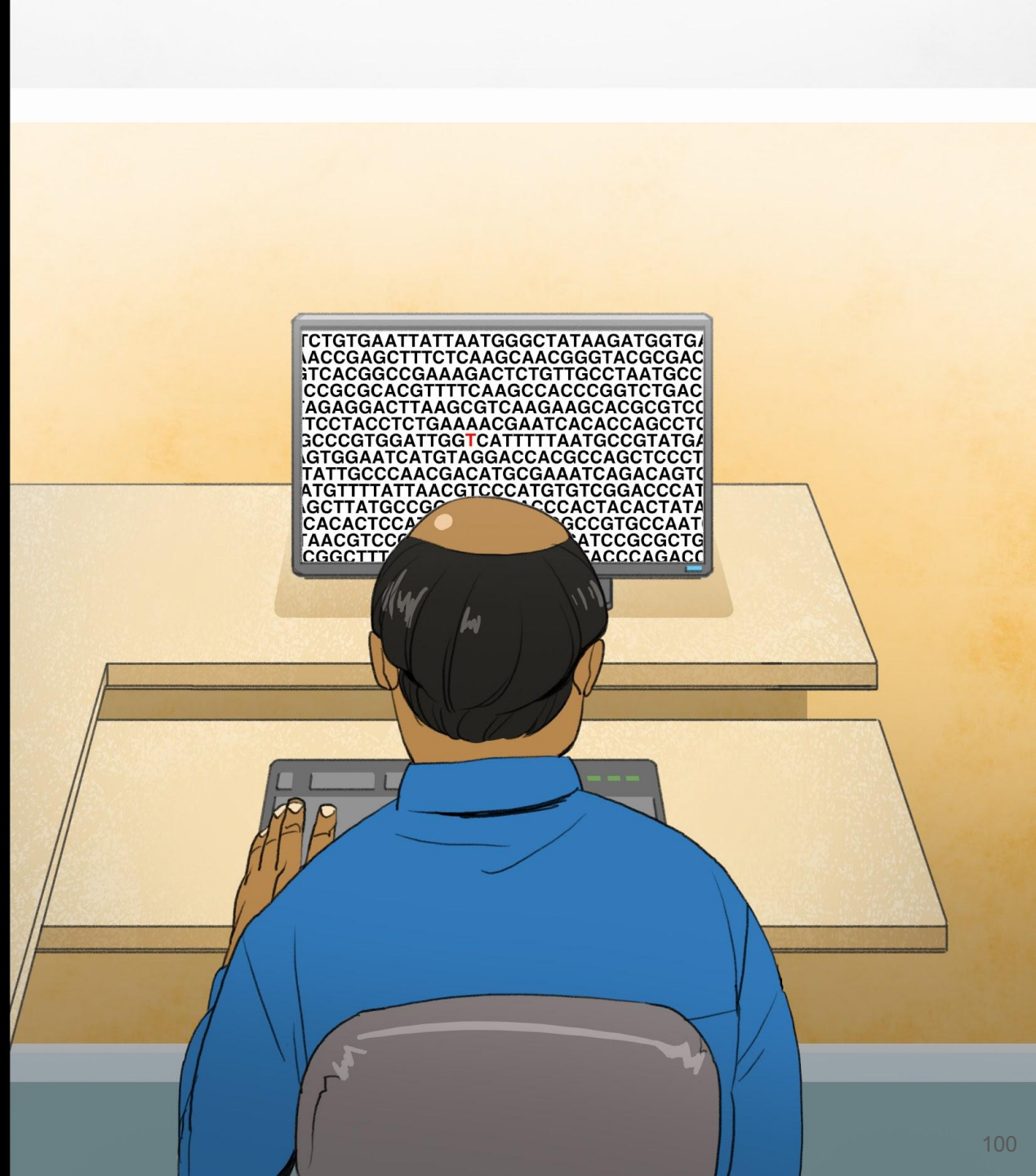
Environmental causes or random mutations



Gene mutation or variant







Single site analysis

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CGTAAGAGTCGCGCTGAA
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ACACTATACCCAGACCAC
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AGCAGAGCA **A** CCGAGCTTTCTCAAG
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GGCTGATCGTCACGGCCGAAAGACTCT
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Individual gene testing

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Gene panel

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Exome sequencing




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Genome sequencing




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A man with dark hair, wearing a white button-down shirt, is shown from the chest up. He has a worried expression, with furrowed brows and a slightly open mouth. His hands are clasped together on a surface in front of him. To his left is a large green plant in a brown pot. The background is a light blue wall with a window on the right. A yellow speech bubble is positioned above the plant.

So, this test would tell us if I have cancer? Or if I'm going to get cancer?


An illustration of a woman with brown hair, wearing a white lab coat over a red top, sitting at a table with her hands clasped. She is looking towards a man whose back is to the viewer. A speech bubble above her contains text. The background shows a window with a white frame and a blue wall.

Genetic testing can tell us if a person has certain genetic conditions, but it doesn't tell you if you have cancer.



Genetic testing can tell us if a person has certain genetic conditions, but it doesn't tell you if you have cancer.

It tells you if you have a gene that makes it more likely for you to get certain cancers in the future.



Genetic testing can tell us if a person has certain genetic conditions, but it doesn't tell you if you have cancer.

It tells you if you have a gene that makes it more likely for you to get certain cancers in the future.

But even then, we can't know if you would develop the disease or condition for sure.

Exome sequencing also provides a lot of other genetic information. From this test, we can learn about whether you have other genetic conditions that are unrelated to cancer.



Exome sequencing also provides a lot of other genetic information. From this test, we can learn about whether you have other genetic conditions that are unrelated to cancer.

We can also learn if you are a carrier of any gene variants that are related to particular illnesses. In this case, you wouldn't get the illness yourself because the gene is recessive.



But you could pass the affected gene on to your children. If your partner also passes on an affected gene, your children could get the illness.



But you could pass the affected gene on to your children. If your partner also passes on an affected gene, your children could get the illness.

You can decide if you want this other information or only the information about cancer genes.



What can exome sequencing tell Manuel?

Information he may want to know:

- Cancer risk

Information he may **NOT** want to know:

- Risk of other non-cancer illness
- Carrier status of other genes



Only looks at 1% – 2% of a person's entire genome.



```
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May find an unknown variant in a known gene.



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 GTAGGACCACCGCAGCTCCCTGCGAGGCTCCAATTTTTTTGTTCACTCTATTGCCAACGACAT
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Some types of changes are not visible.




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Quiz!

- What is genetic testing?
- Why would a genetic counselor recommend genetic testing?
- What is looked at in single site analysis?
- What is looked at in individual gene testing?
- What is looked at in gene panel testing?
- What is looked at in exome testing?
- What is looked at in genome testing?
- What are some limitations to gene sequencing?


It sounds like this test can tell me if I have certain genetic conditions, but I don't have any symptoms of those.






It sounds like this test can tell me if I have certain genetic conditions, but I don't have any symptoms of those.

And I guess it could tell me if I carry some gene variant that I might pass to my children. I'm most worried about cancer.


An illustration of a man with dark hair and a white button-down shirt, looking slightly worried or questioning. He is sitting at a table with his hands clasped. A woman with blonde hair is seen from the back, facing him. A green plant is on the table between them. A speech bubble points to the man's mouth.

This test just tells me if I have a higher risk than everyone else of getting cancer, right?

A man with dark hair, wearing a white button-down shirt, is sitting at a table. He has a worried expression on his face, with furrowed brows and a slightly open mouth. He is gesturing with his hands as if in the middle of a conversation. In the foreground, the back of a woman's head with blonde hair is visible, looking towards the man. The background features a blue wall, a window with white trim, and a green potted plant.

This test just tells me if I have a higher risk than everyone else of getting cancer, right?

But so what? If I'm going to get cancer, there's nothing I can do about it, right?



This test just tells me if I have a higher risk than everyone else of getting cancer, right?

But so what? If I'm going to get cancer, there's nothing I can do about it, right?

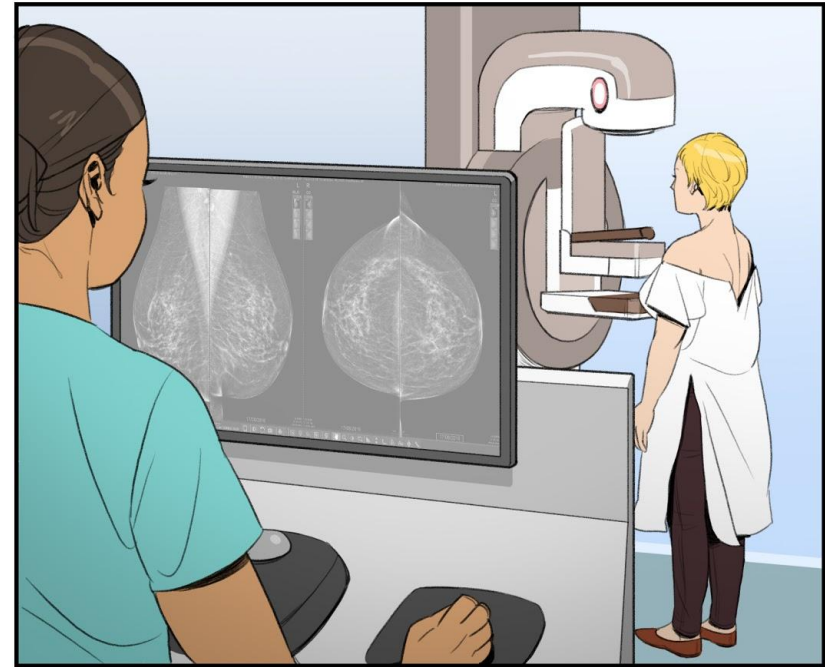
So why bother getting tested? What does it matter?

“It actually does matter...”

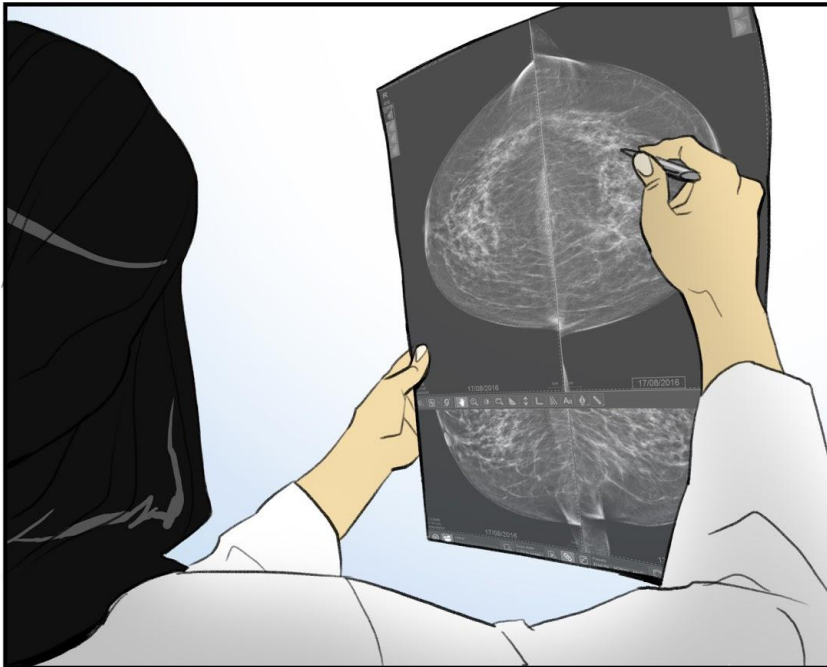
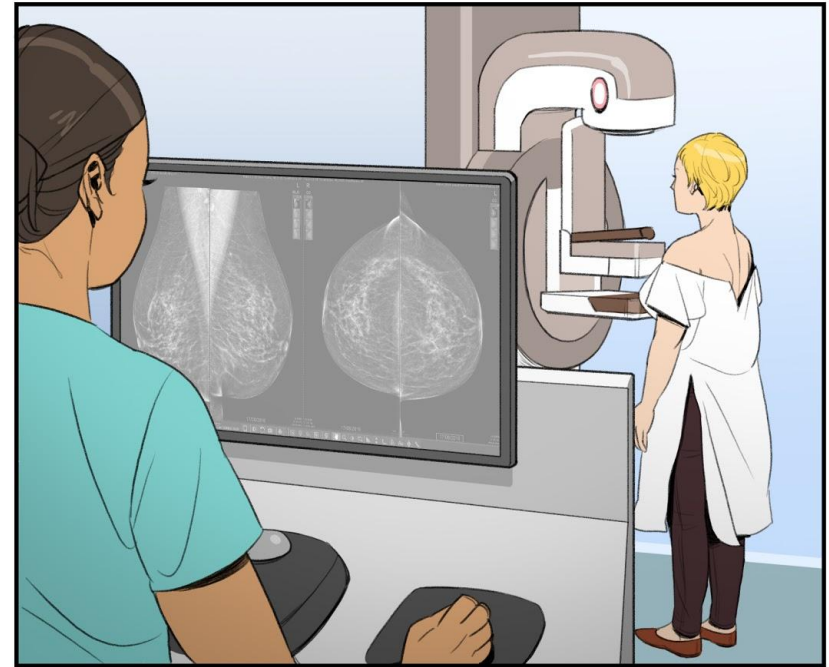
"It actually does matter..."



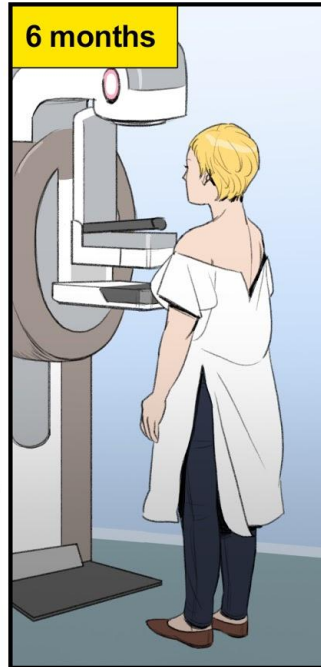
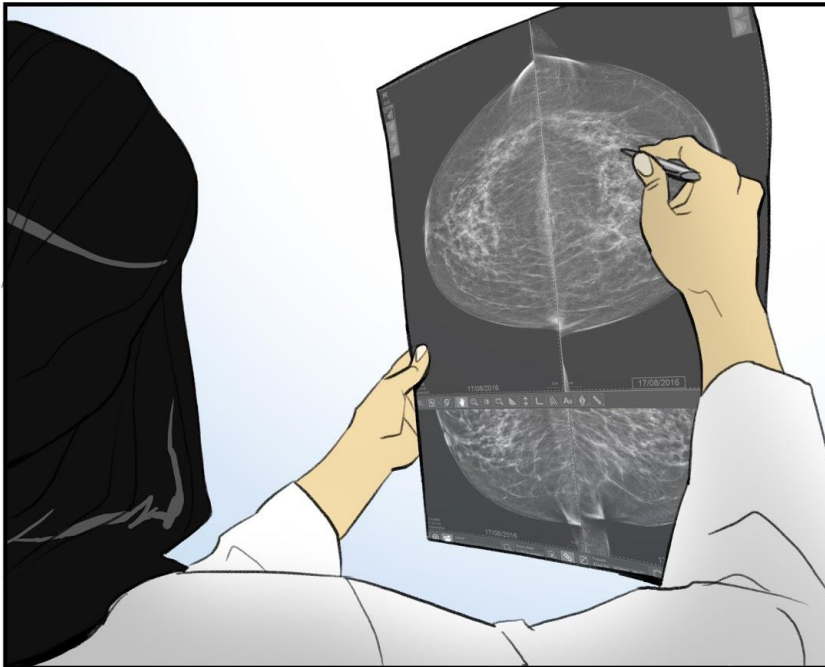
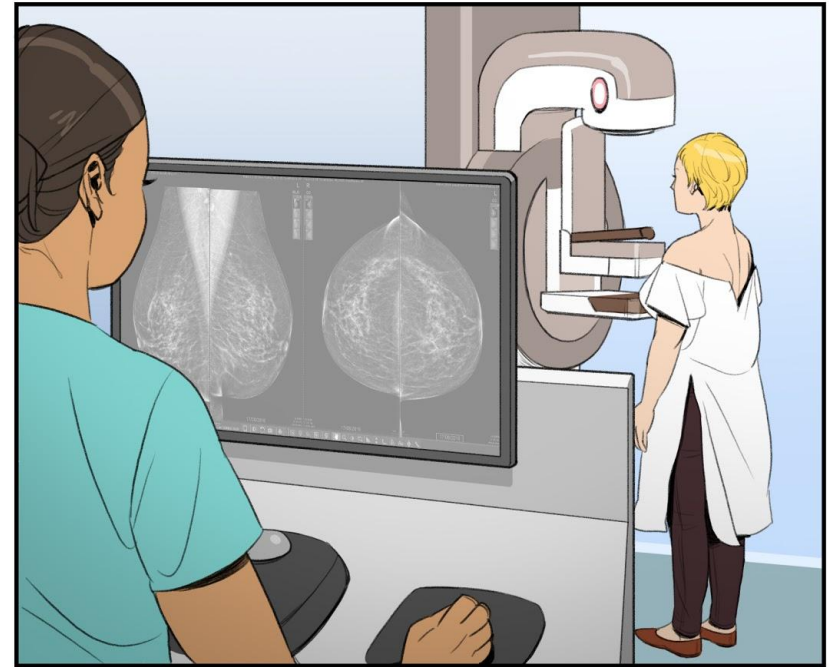
"It actually does matter..."



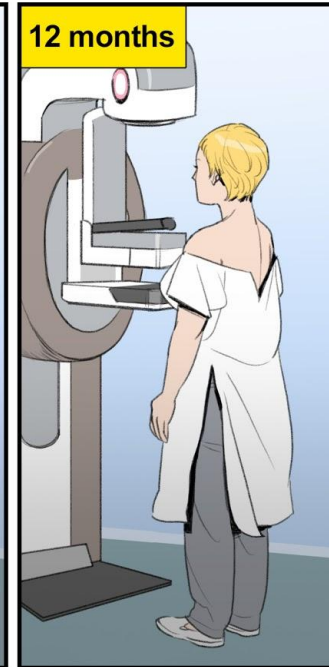
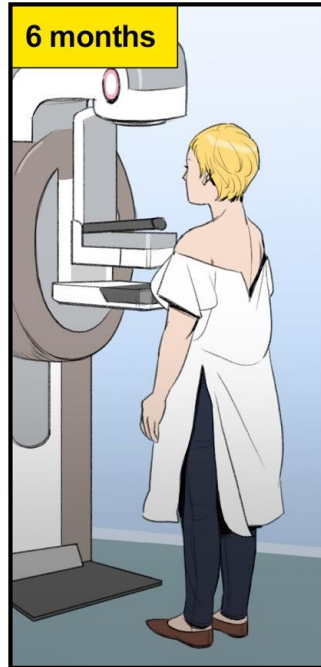
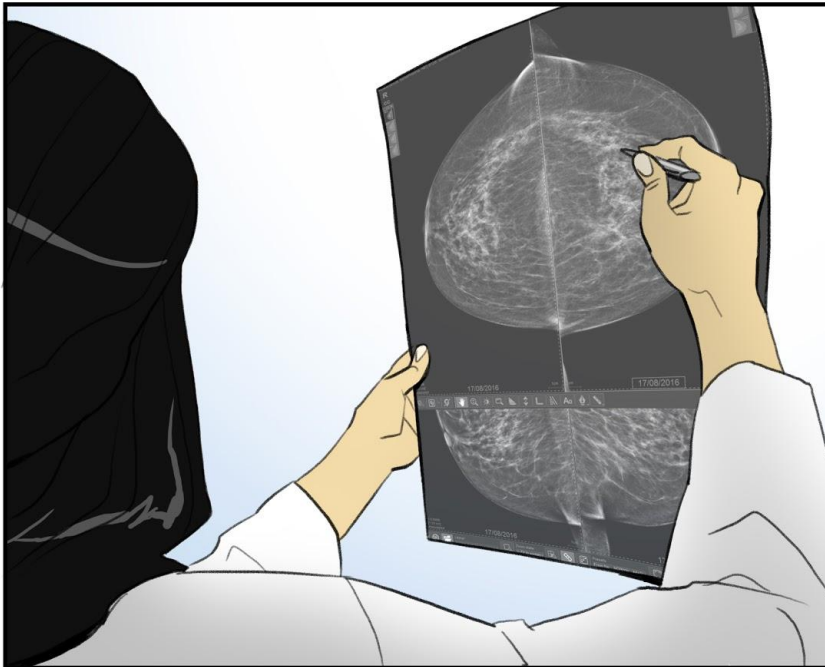
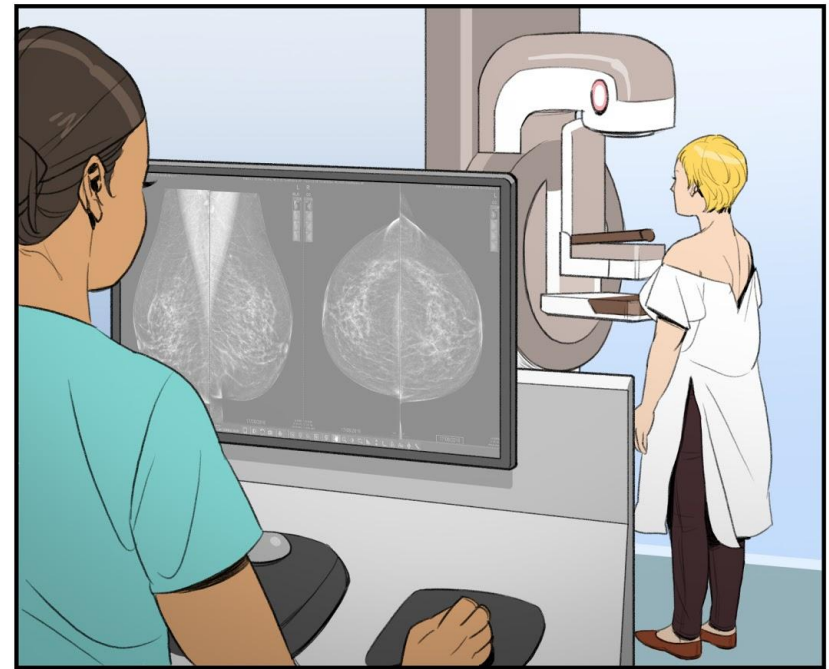
"It actually does matter..."



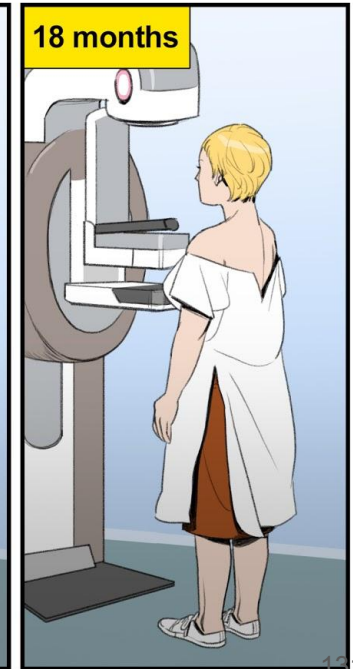
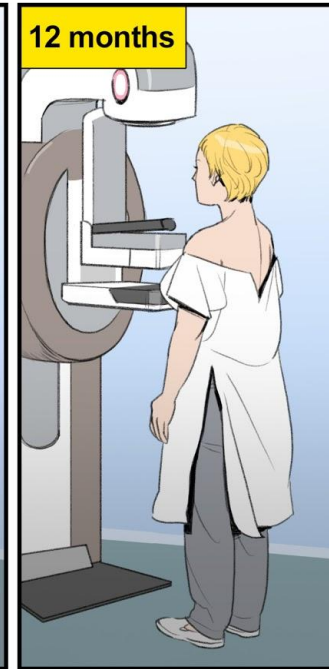
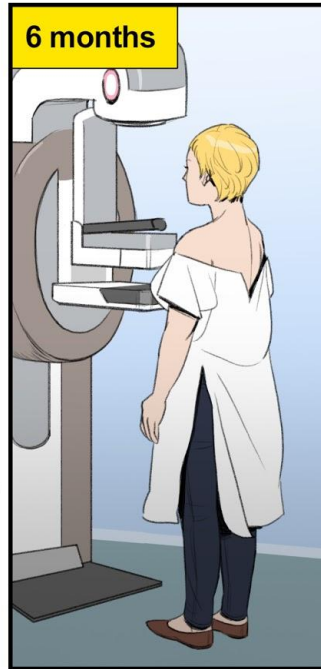
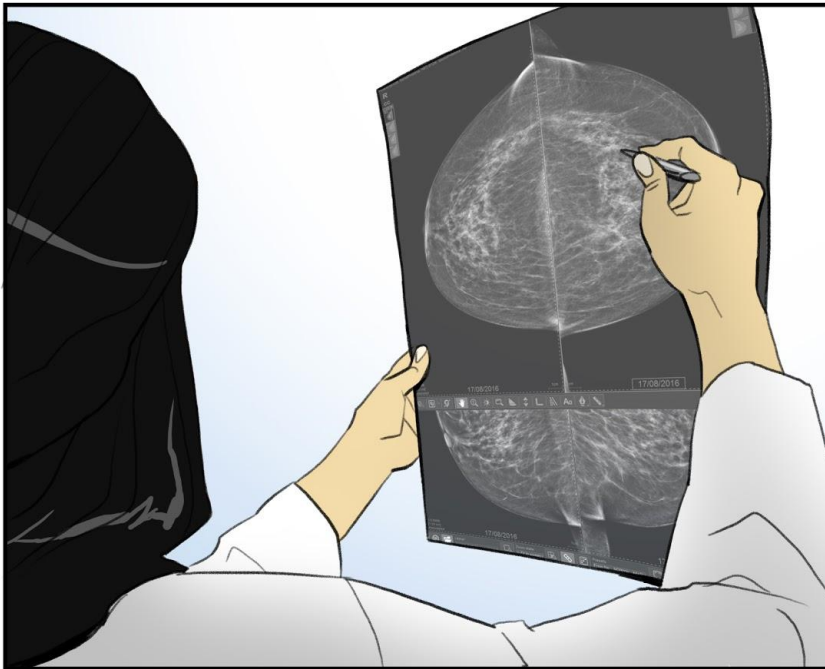
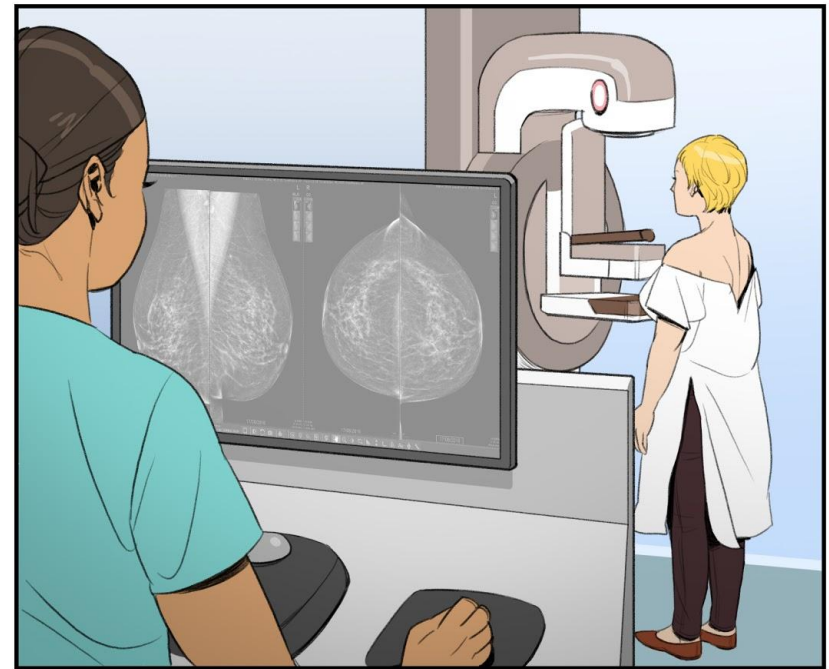
"It actually does matter..."



"It actually does matter..."




"It actually does matter..."



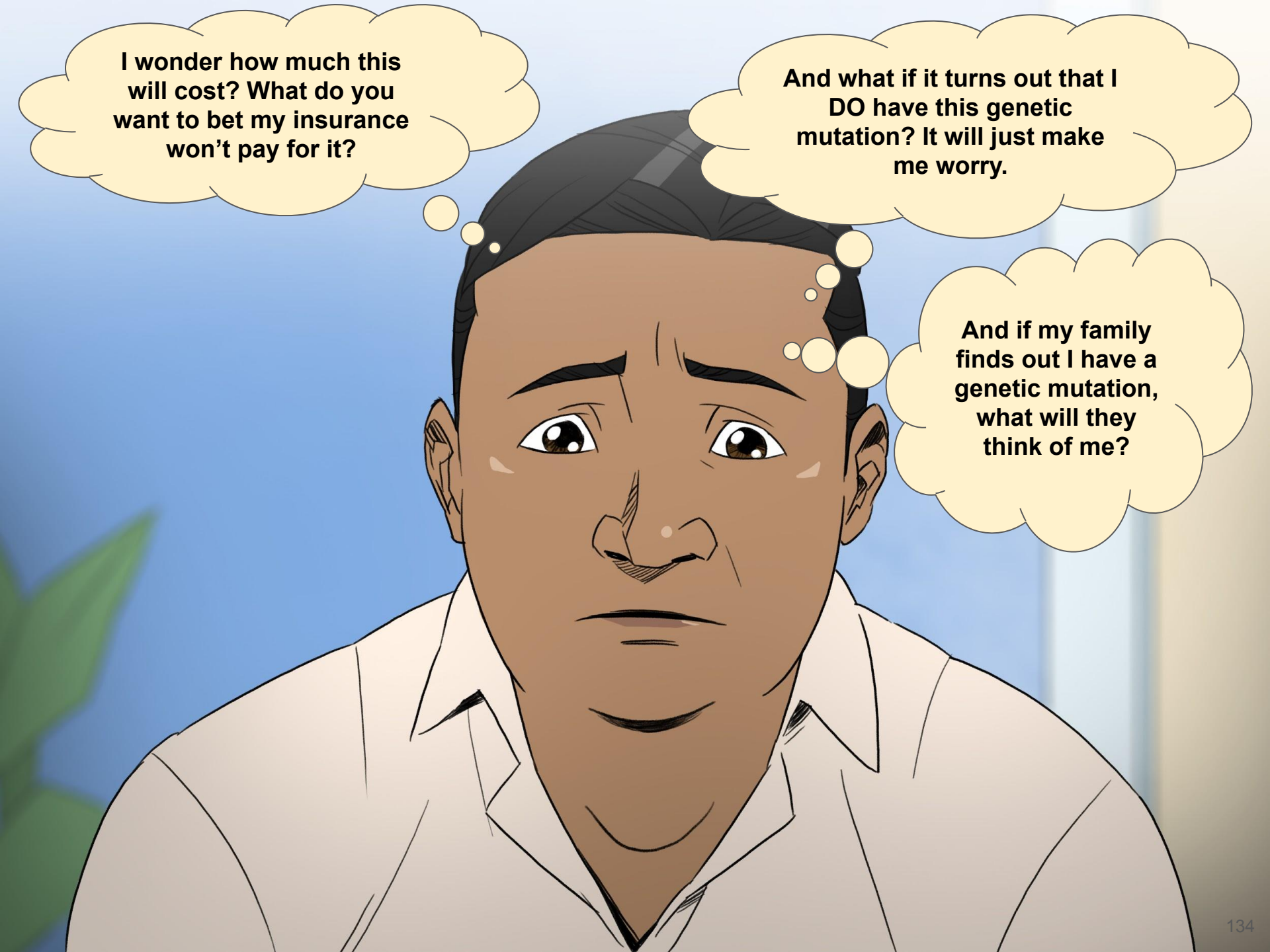
**I wonder how much this
will cost? What do you
want to bet my insurance
won't pay for it?**





I wonder how much this will cost? What do you want to bet my insurance won't pay for it?

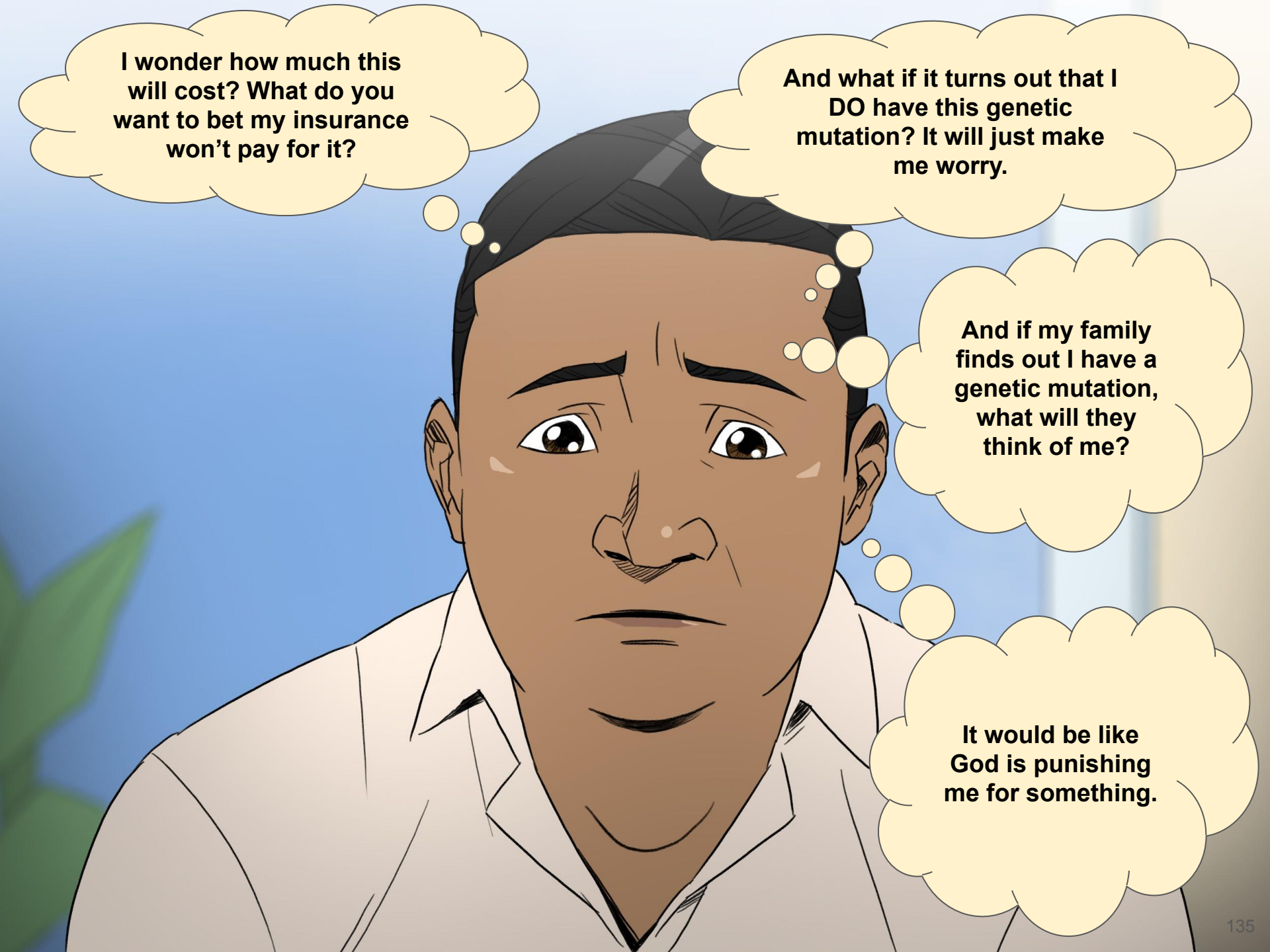
And what if it turns out that I DO have this genetic mutation? It will just make me worry.

A man with dark skin and short black hair is shown from the chest up, wearing a light-colored collared shirt. He has a worried expression with furrowed brows and wide eyes. Three yellow thought bubbles are connected to his head by small circles. The background is a soft-focus indoor setting with a blue wall and a green plant on the left.

I wonder how much this will cost? What do you want to bet my insurance won't pay for it?

And what if it turns out that I DO have this genetic mutation? It will just make me worry.

And if my family finds out I have a genetic mutation, what will they think of me?


A man with a worried expression, surrounded by thought bubbles. The man has dark skin, short dark hair, and is wearing a light-colored collared shirt. He has a furrowed brow and wide, anxious eyes. Four yellow thought bubbles are connected to his head by thin lines, each containing a different worry. The background is a simple blue and green gradient.

I wonder how much this will cost? What do you want to bet my insurance won't pay for it?

And what if it turns out that I DO have this genetic mutation? It will just make me worry.

And if my family finds out I have a genetic mutation, what will they think of me?

It would be like God is punishing me for something.

A man with a worried expression, surrounded by thought bubbles. He has dark hair, a high forehead, and is wearing a light-colored collared shirt. The background is a soft-focus outdoor scene with blue sky and green foliage. Five yellow thought bubbles of varying sizes are connected to his head by thin lines, containing his internal monologue.


I wonder how much this will cost? What do you want to bet my insurance won't pay for it?

And what if it turns out that I DO have this genetic mutation? It will just make me worry.

Or what if they terminate my health insurance because they know I'm going to get cancer?

And if my family finds out I have a genetic mutation, what will they think of me?

It would be like God is punishing me for something.

A man with a worried expression is shown from the chest up. He has dark skin, short dark hair, and is wearing a light-colored collared shirt. He is surrounded by several yellow thought bubbles of various sizes, some connected to his head by thin lines. The background is a soft-focus outdoor scene with blue sky and green foliage.

I wonder how much this will cost? What do you want to bet my insurance won't pay for it?

And what if it turns out that I DO have this genetic mutation? It will just make me worry.

Or what if they terminate my health insurance because they know I'm going to get cancer?

And if my family finds out I have a genetic mutation, what will they think of me?

If they find out at work, could I get fired?

It would be like God is punishing me for something.

Manuel, you look worried. What's going through your mind?



GINA: the Genetic Information Nondiscrimination Act of 2008

PUBLIC LAW 110-233—MAY 21, 2008

122 STAT. 881

Public Law 110-233
110th Congress

An Act

To prohibit discrimination on the basis of genetic information with respect to health insurance and employment.

May 21, 2008
[H.R. 493]

Be it enacted by the Senate and House of Representatives of the United States of America in Congress assembled,

SECTION 1. SHORT TITLE; TABLE OF CONTENTS.

(a) SHORT TITLE.—This Act may be cited as the “Genetic Information Nondiscrimination Act of 2008”.

(b) TABLE OF CONTENTS.—The table of contents of this Act is as follows:

Sec. 1. Short title; table of contents.
Sec. 2. Findings.

TITLE I—GENETIC NONDISCRIMINATION IN HEALTH INSURANCE

Sec. 101. Amendments to Employee Retirement Income Security Act of 1974.
Sec. 102. Amendments to the Public Health Service Act.
Sec. 103. Amendments to the Internal Revenue Code of 1986.
Sec. 104. Amendments to title XVIII of the Social Security Act relating to medigap.
Sec. 105. Privacy and confidentiality.
Sec. 106. Assuring coordination.

TITLE II—PROHIBITING EMPLOYMENT DISCRIMINATION ON THE BASIS OF GENETIC INFORMATION

Sec. 201. Definitions.
Sec. 202. Employer practices.
Sec. 203. Employment agency practices.
Sec. 204. Labor organization practices.
Sec. 205. Training programs.
Sec. 206. Confidentiality of genetic information.
Sec. 207. Remedies and enforcement.
Sec. 208. Disparate impact.
Sec. 209. Construction.
Sec. 210. Medical information that is not genetic information.
Sec. 211. Regulations.
Sec. 212. Authorization of appropriations.
Sec. 213. Effective date.

TITLE III—MISCELLANEOUS PROVISIONS

Sec. 301. Severability.
Sec. 302. Child labor protections.

SEC. 2. FINDINGS.

Congress makes the following findings:

(1) Deciphering the sequence of the human genome and other advances in genetics open major new opportunities for medical progress. New knowledge about the genetic basis of illness will allow for earlier detection of illnesses, often before symptoms have begun. Genetic testing can allow individuals to take steps to reduce the likelihood that they will contract

Genetic
Information
Non-
discrimination
Act of 2008
42 USC 2000ff
note.

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122 STAT. 881

TITLE I—GENETIC NONDISCRIMINATION IN HEALTH INSURANCE

“(3) NO GROUP-BASED DISCRIMINATION ON BASIS OF GENETIC INFORMATION.—

“(A) IN GENERAL.—For purposes of this section, a group health plan, and a health insurance issuer offering group health insurance coverage in connection with a group health plan, may not adjust premium or contribution amounts for the group covered under such plan on the basis of genetic information.”

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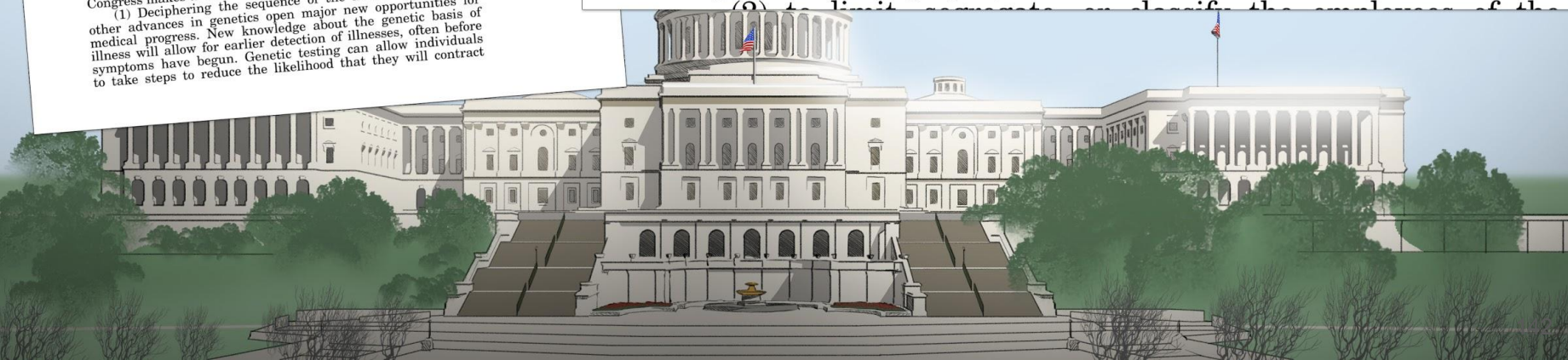
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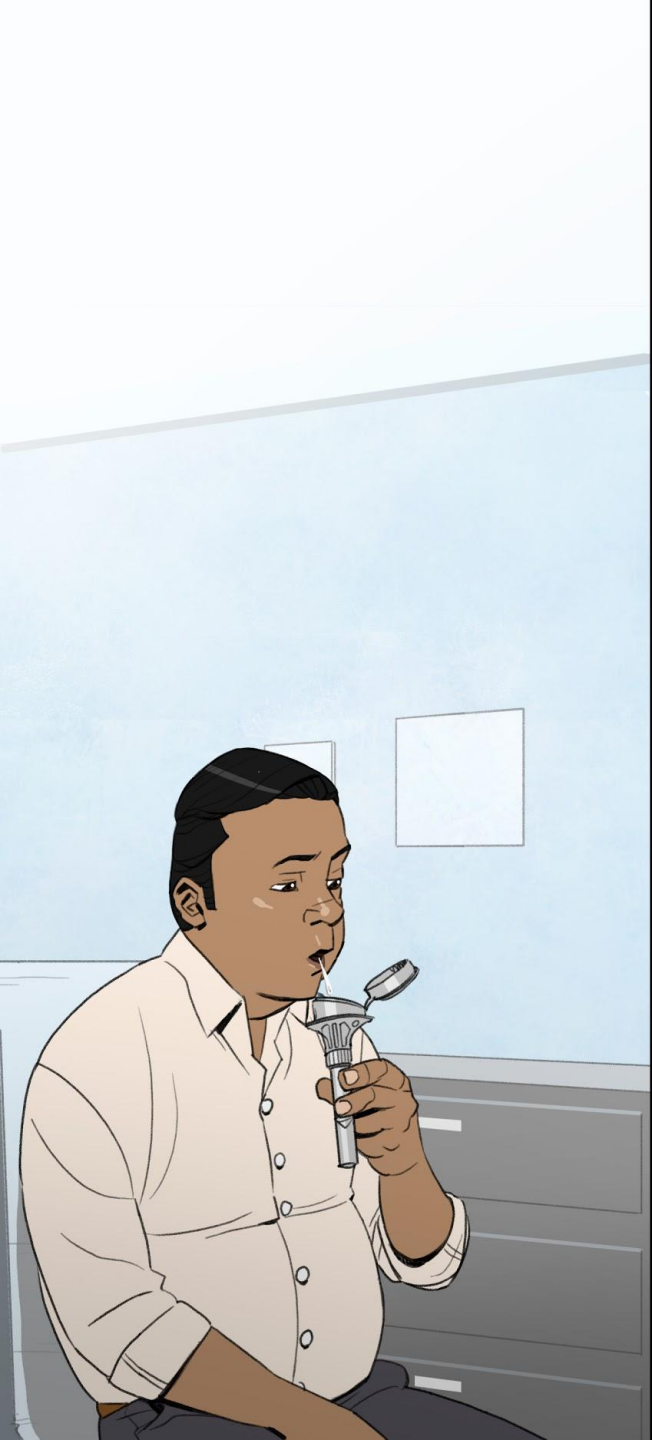
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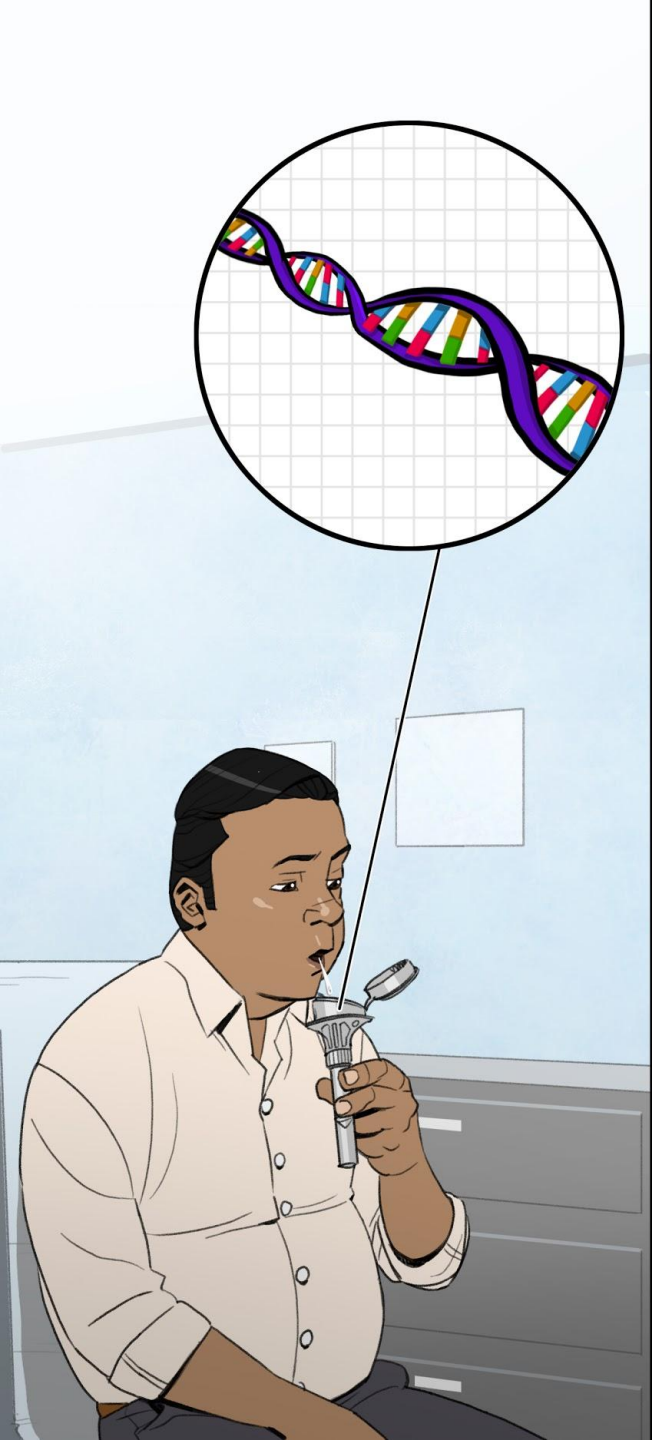
(a) DISCRIMINATION BASED ON GENETIC INFORMATION.—It shall be an unlawful employment practice for an employer—

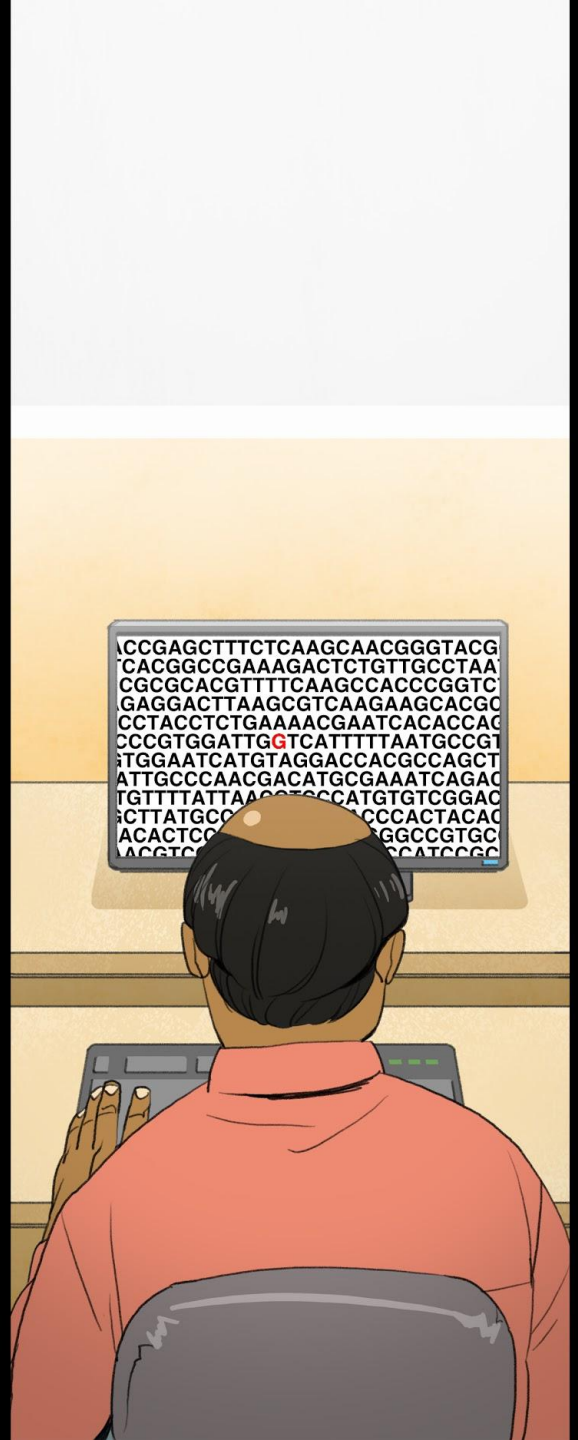
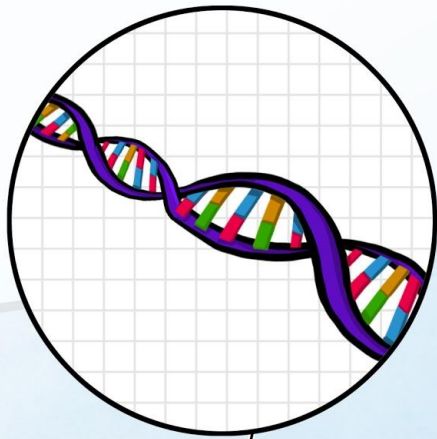
(1) to fail or refuse to hire, or to discharge, any employee, or otherwise to discriminate against any employee with respect to the compensation, terms, conditions, or privileges of employment of the employee, because of genetic information with respect to the employee; or

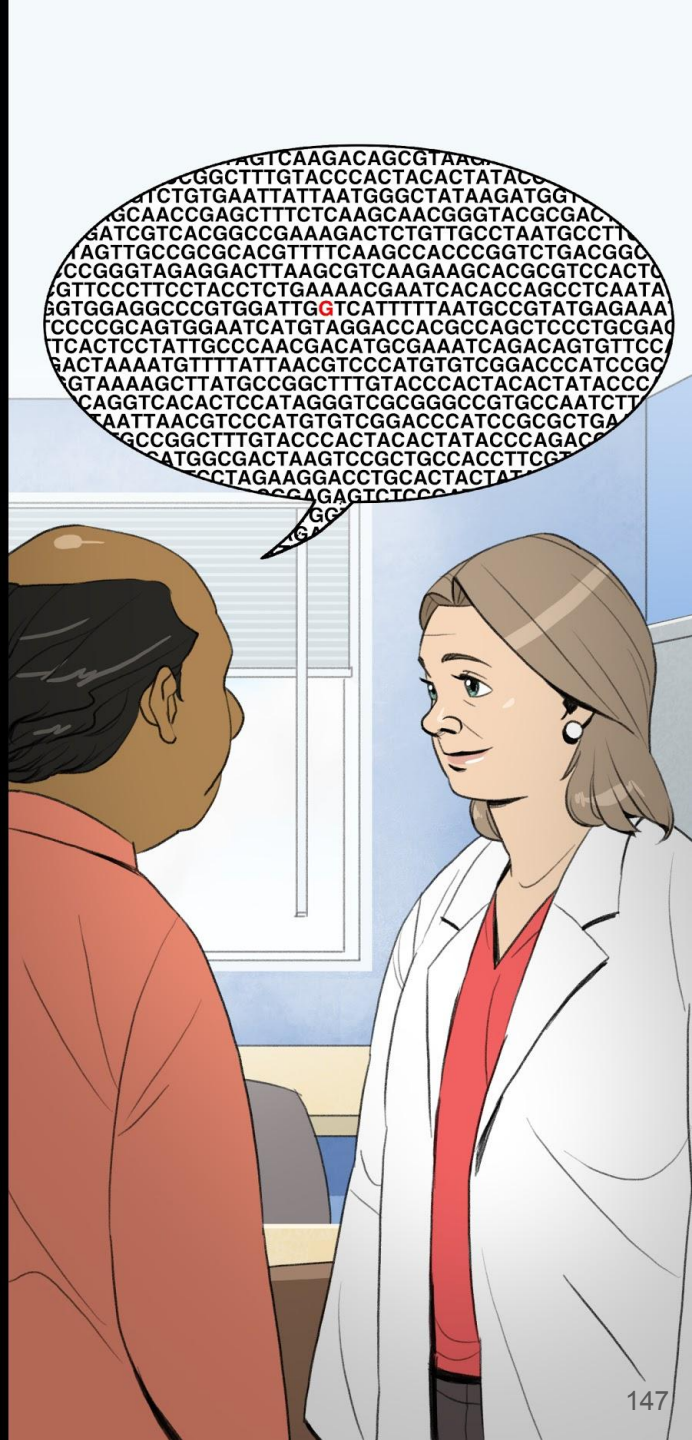
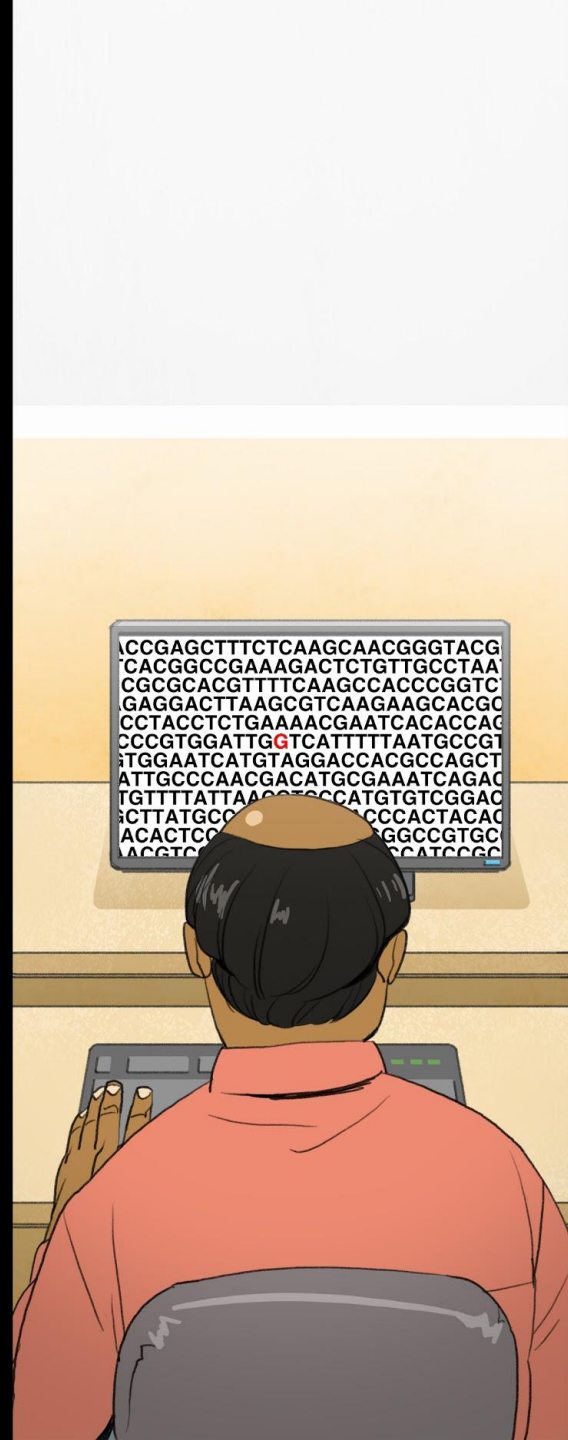
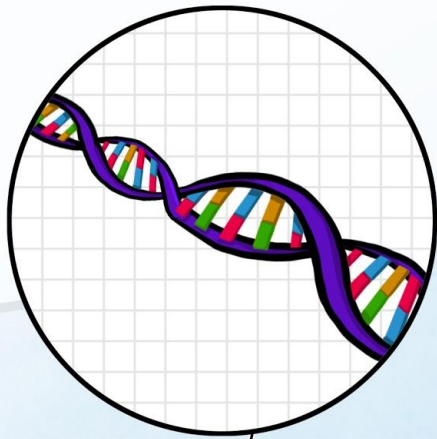
42 USC 2000f note.











Quiz!

- What's the purpose of genetic testing?
- What could be some of the benefits to genetic testing?
- Why might some people be wary of testing?
- What is GINA?
- Does a patient have to hear all the results from exome testing?
- Why might a patient NOT want to hear all the results?



Medical
Center



**"Negative"
(Normal)**



"Negative"
(Normal)



"Positive"
(Deleterious mutation)



"Negative"
(Normal)



"VUS"
(Variant of uncertain
significance)




"Positive"
(Deleterious mutation)



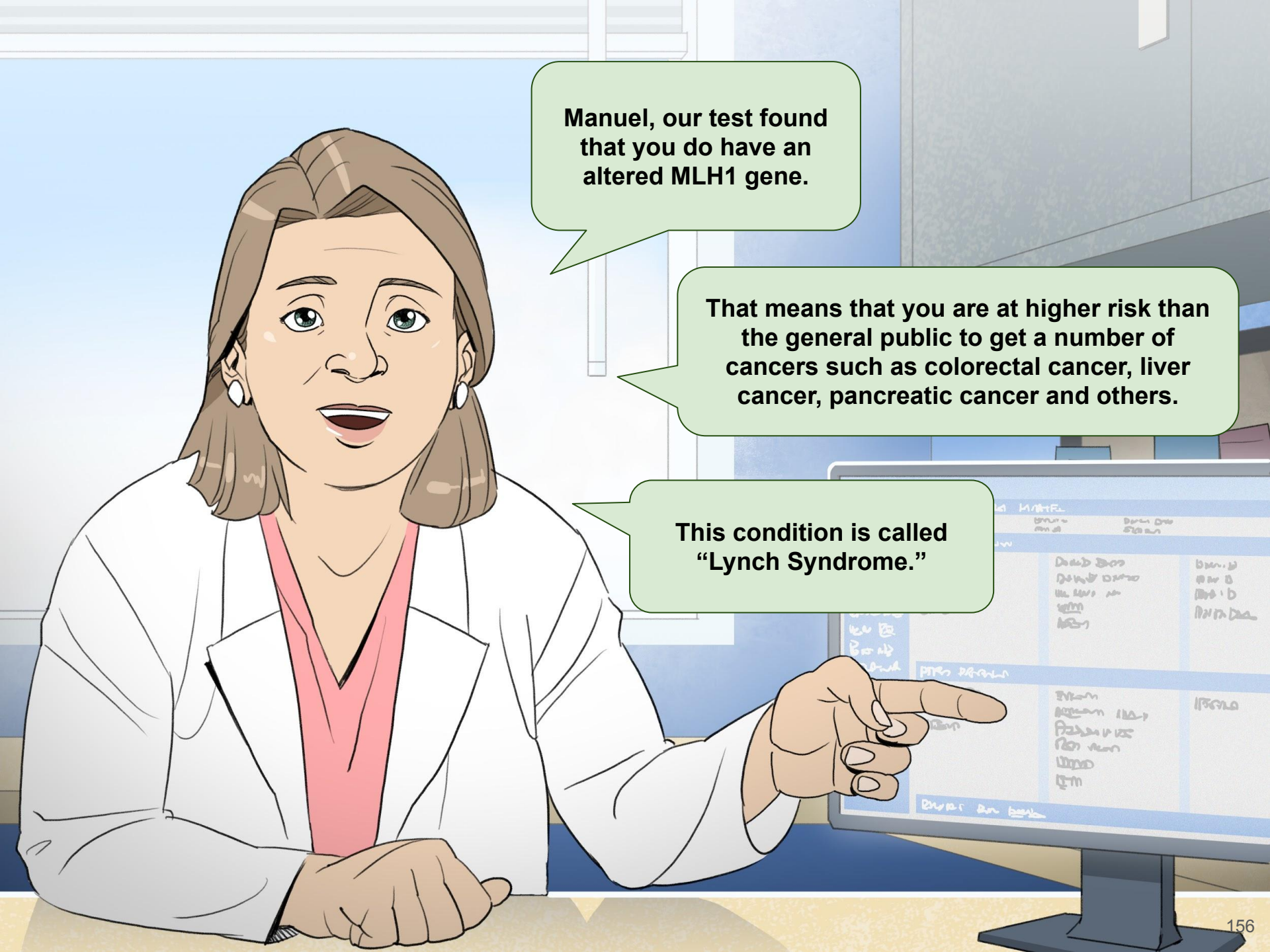
Manuel, our test found that you do have an altered MLH1 gene.





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That means that you are at higher risk than the general public to get a number of cancers such as colorectal cancer, liver cancer, pancreatic cancer and others.



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This condition is called “Lynch Syndrome.”

So, let's set up an appointment for a colonoscopy for you. It would probably be a good idea to do them more frequently than usual, now that we know you have a high risk of colon cancer.



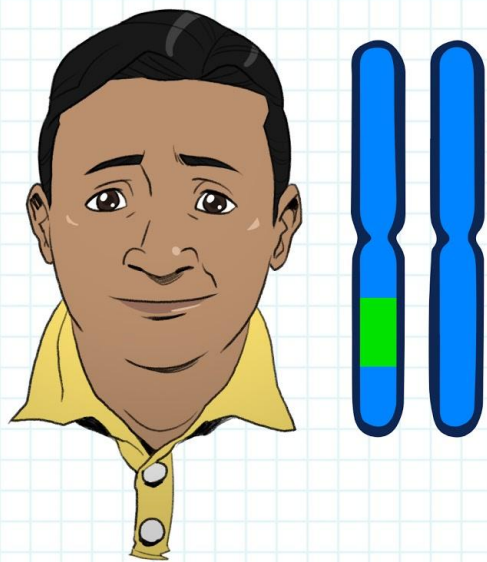
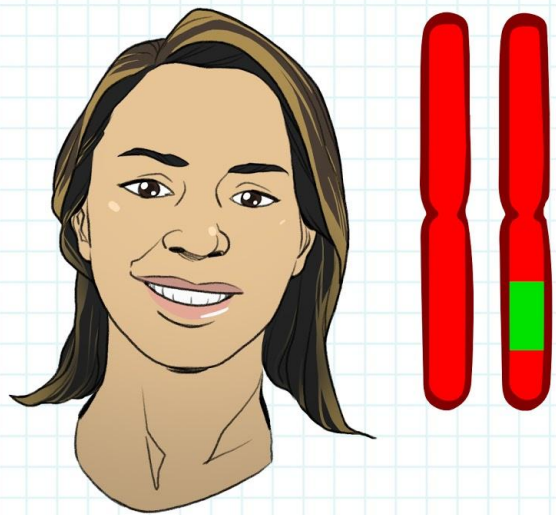
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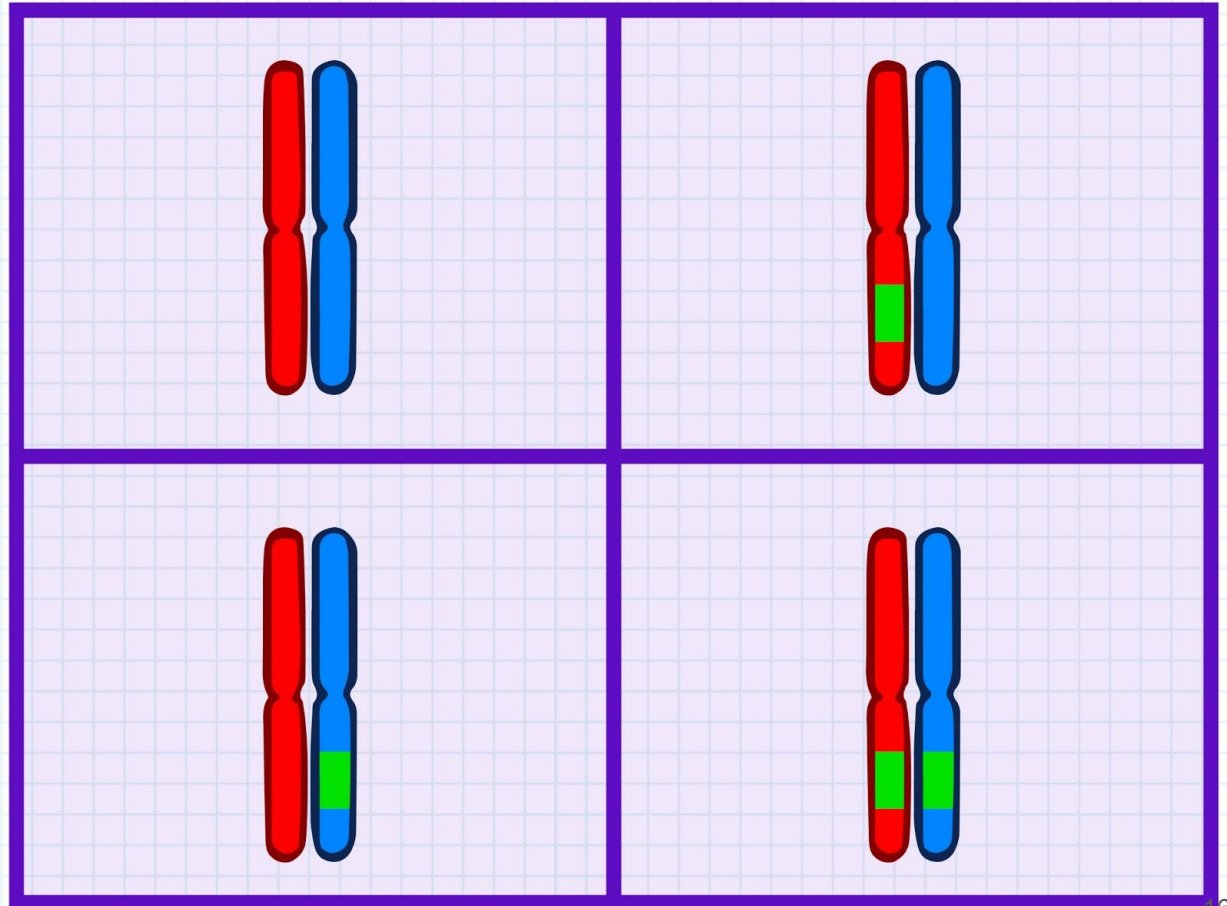
We know that removing polyps can help prevent cancer, AND catch it early if it starts.



That makes sense. Let's make that appointment.









Wow, this is good to know. My wife and I have been talking about having children.



A man with dark hair, wearing a yellow polo shirt, is shown from the chest up. He has a thoughtful expression, with his hands clasped in front of him. The background is a simple blue wall with a window on the right and a green plant on the left.

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I need to talk to her about this. Can she get tested to find out if she is a carrier?

Absolutely. Have her call in for an appointment and she can get “carrier testing” too.





Nurses Station

Quiz!

- What sort of sample is taken to do genetic testing?
- What does it mean to have a positive result?
- What does it mean to have a negative result?
- What does it mean to variant of uncertain significance?
- What does it mean to be a carrier of a genetic condition?

QUESTIONS?

