

Your Genes

and

Your Health

ABGS Module  
Illustrated by Nicki Shaw



# Age-Based Genomic Screening

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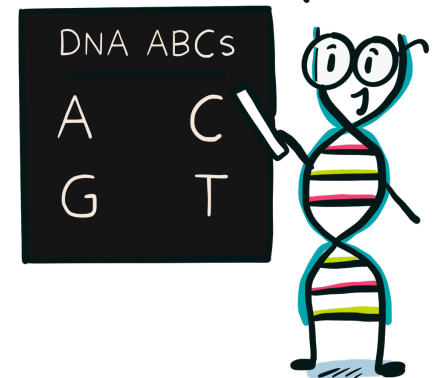
DNA is shaped like a twisted ladder or a double helix.

That's a lot of knowledge!



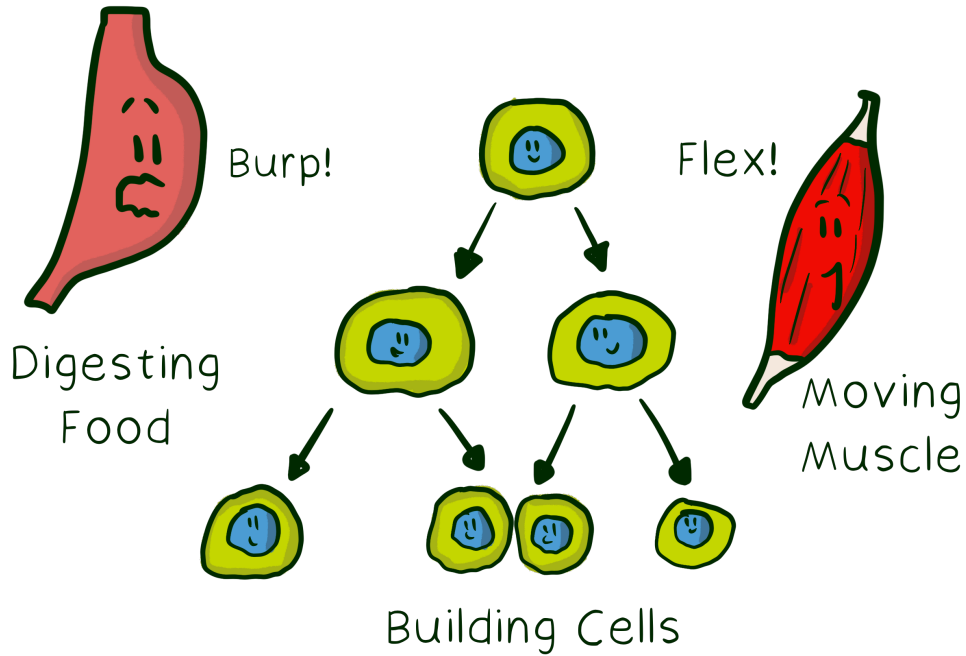
DNA is like an instruction manual for how a living thing is built and works.

♪ A C G T ♪



The DNA "alphabet" uses four letters: A, C, G and T.

Genes provide instructions for traits and body functions.

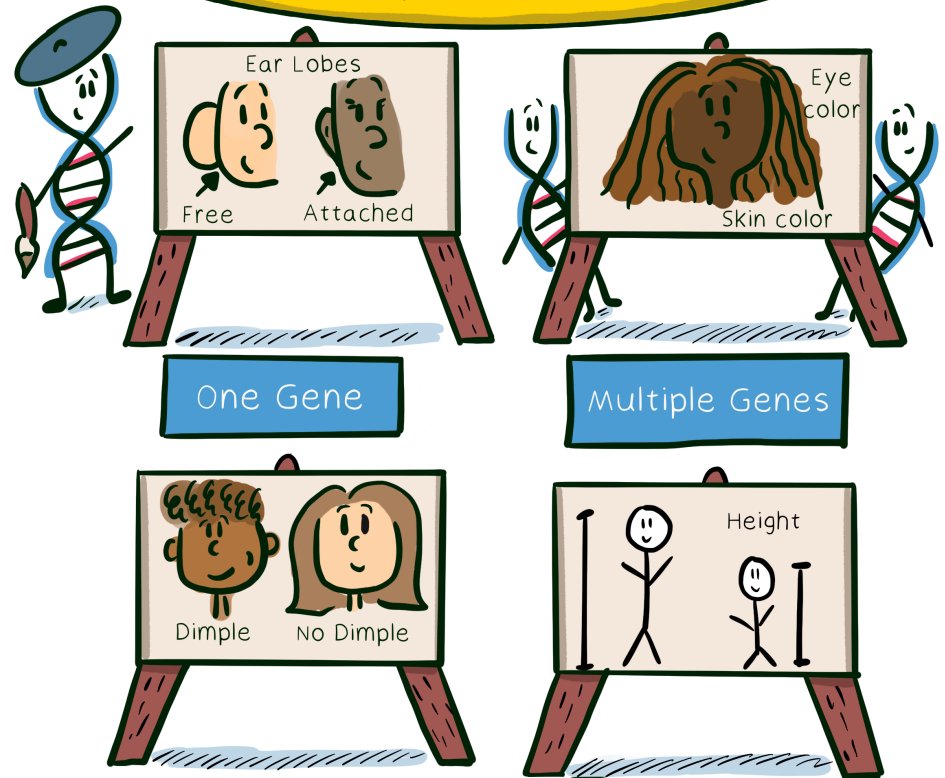


And so much more!



Genes provide the information for your physical traits. A trait is a specific characteristic of an individual.

Art Exhibit:  
Uniquely You!



Some traits are determined entirely by genes, while others are influenced by our environment, or a mixture of both. 🌞 🍴

Some diseases are influenced by a single gene, while others are influenced by multiple genes.

Differences in DNA can change these instructions, just like changing the spelling of a word can affect the meaning of a sentence. These differences are called genetic variants.

The diagram shows three scenarios of genetic influence on diseases:

- Marfan Syndrome:** A whiteboard shows a drawing of a long, thin stick figure and a red, elongated heart. A DNA double helix character stands next to a paint palette.
- Alzheimer's Disease:** A whiteboard shows a drawing of a brain. Two DNA double helix characters are shown, one holding a paint palette.
- Type 2 Diabetes:** A whiteboard shows a drawing of a yellow, speckled pancreas. A DNA double helix character stands next to a paint palette. Below the whiteboard is a blue box containing the text "Genes(s) + Environment".

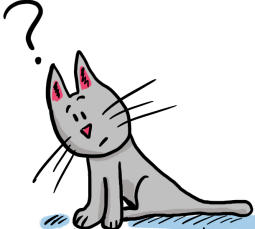


The gray cat ran down the hall.



*Substitution*

The gray cat ran down the ball.



*Deletion*

~~The gray cat ran down the hall.~~



*Repeats*

The gray cat ran **ran ran ran** down the hall.

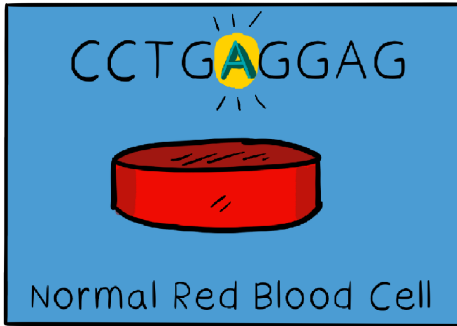
# Sickle Cell Disease (HBB Gene)

In people with sickle cell disease, an A is replaced with a T in both copies of a person's HBB gene. This is an example of a substitution.

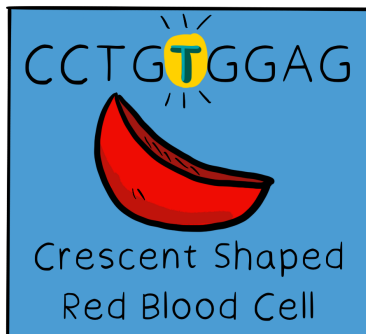


The gray cat ran down the hall.

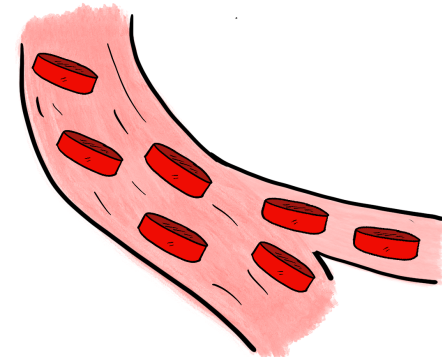
*Substitution*



The gray cat ran down the ball.



Normal Red Blood Cell



Typical Blood Flow

With sickle cell disease



Crescent Shaped Red Blood Cell



## Symptoms

- Pain attacks 
- Swelling in hands and feet 
- Infections 
- Stroke 

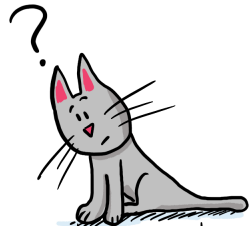
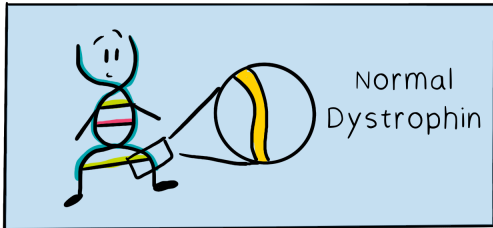
# Duchenne Muscular Dystrophy (DMD Gene)

In people with Duchenne muscular dystrophy, the gene does not work due to the removal of some DNA. This change is an example of deletion.

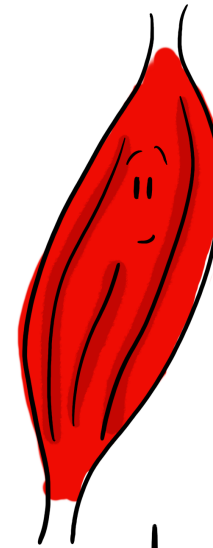
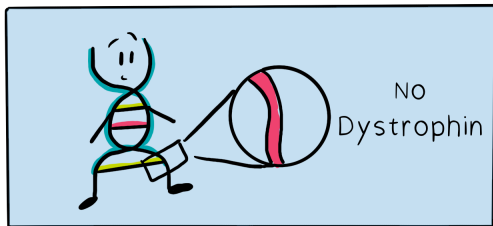


*Deletion*

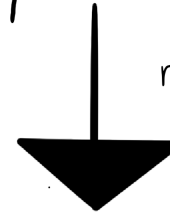
The gray cat ran down the hall.



~~The gray cat ran down the hall.~~



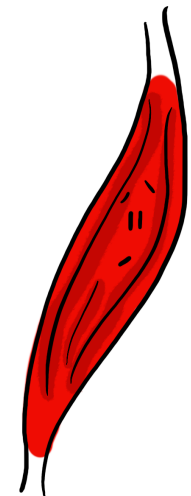
Normal Muscle Growth



with Duchenne muscular dystrophy

## Symptoms

- Loss of Muscle
- Shortened Life Span
- Weak Heart Muscle



# Fragile X Syndrome (FMR1 Gene)

In people with fragile X syndrome, there are repeated segments of DNA within the FMR1 gene which causes disease.

This change is an example of repeats.

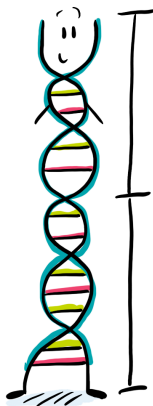


The gray cat ran down the hall.

Repeats



The gray cat ran **ran ran ran** down the hall.



## Possible Symptoms:

•Autism Spectrum Disorder (ASD)

•Intellectual Disability

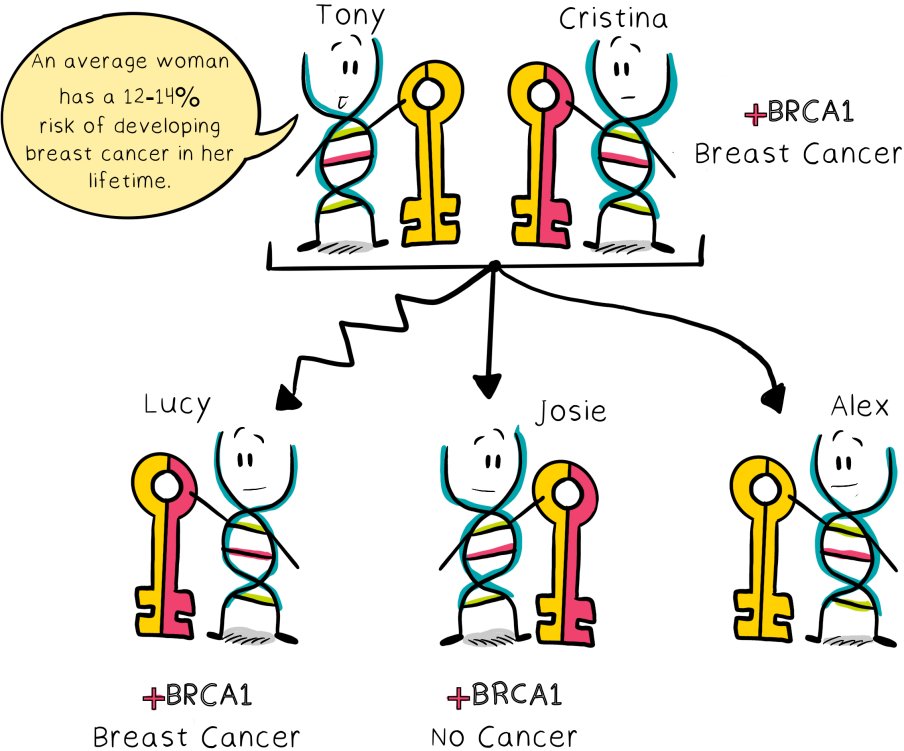


•Murmur/  
Heart Valve Defects



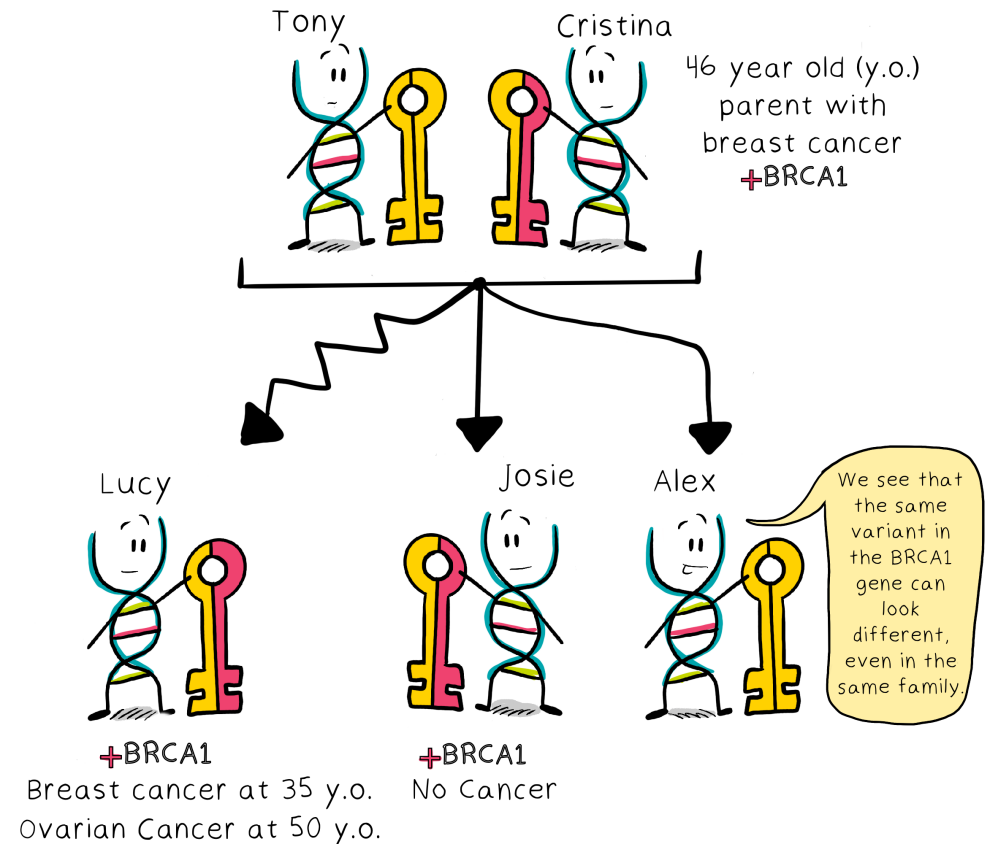
For some genetic conditions, not everyone with a specific disease-related genetic variant will show symptoms of that condition. This difference in disease development is shown in breast cancer. Women who have the BRCA1 disease-related variant have a 50-70% chance of developing breast cancer in their lifetime.

Let's explore this difference in the family below. Cristina has a disease-related variant in one of her BRCA1 genes and her daughters Josie and Lucy have both inherited this variant. However, only Cristina and Lucy have breast cancer (though Josie may develop it too as she ages).



The same disease-related variant can cause different severity of symptoms in a person with the condition. For example, a woman who inherits a disease-related variant in a BRCA1 gene also has a 40-45% chance of developing ovarian cancer in their lifetime.

Let's take another look at the family from before. Here, we see that Cristina and Lucy have the disease-related variant of BRCA1 with development of disease but have different severity of symptoms (types of cancers) and age of onset (46 y.o. vs. 35 y.o. for breast cancer).



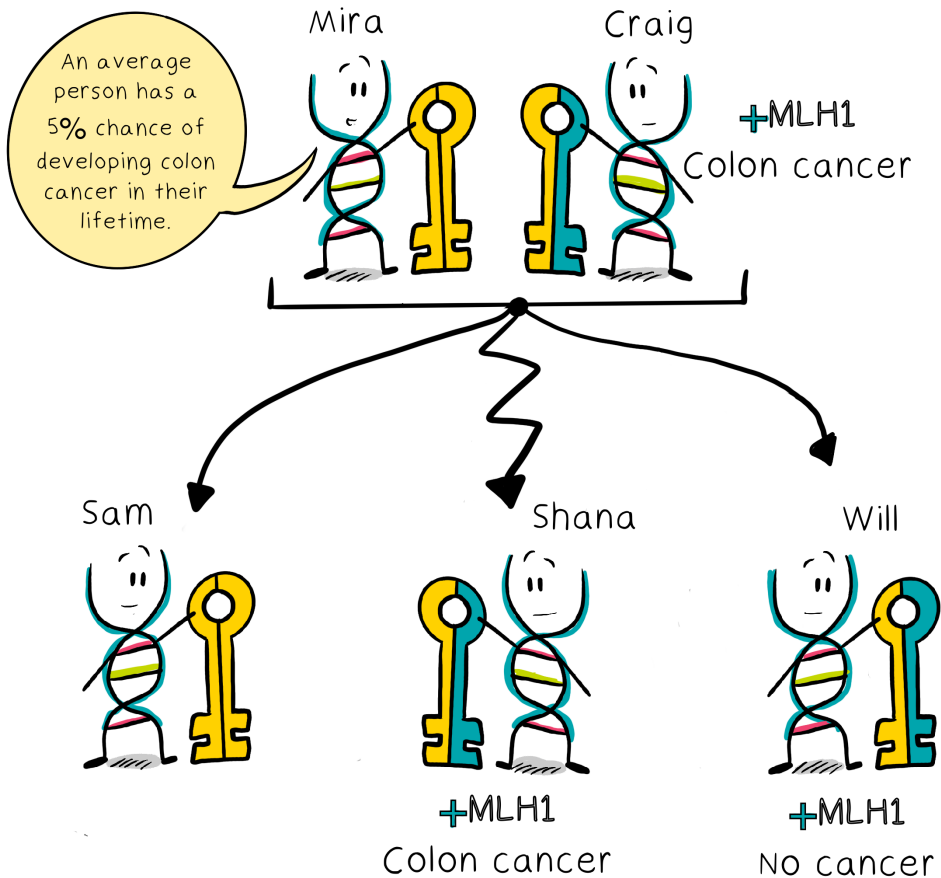


Let's look at another disease example ↘

About 45-60% of individuals with a MLH1 disease-related variant will develop colon cancer in their lifetime.

We can see this difference in disease development in the family below.

Craig has a disease-related variant in one of his MLH1 genes. His daughter Shana and son Will inherited it. However, only Craig and Shana have colon cancer (although Will may develop it too as he ages).

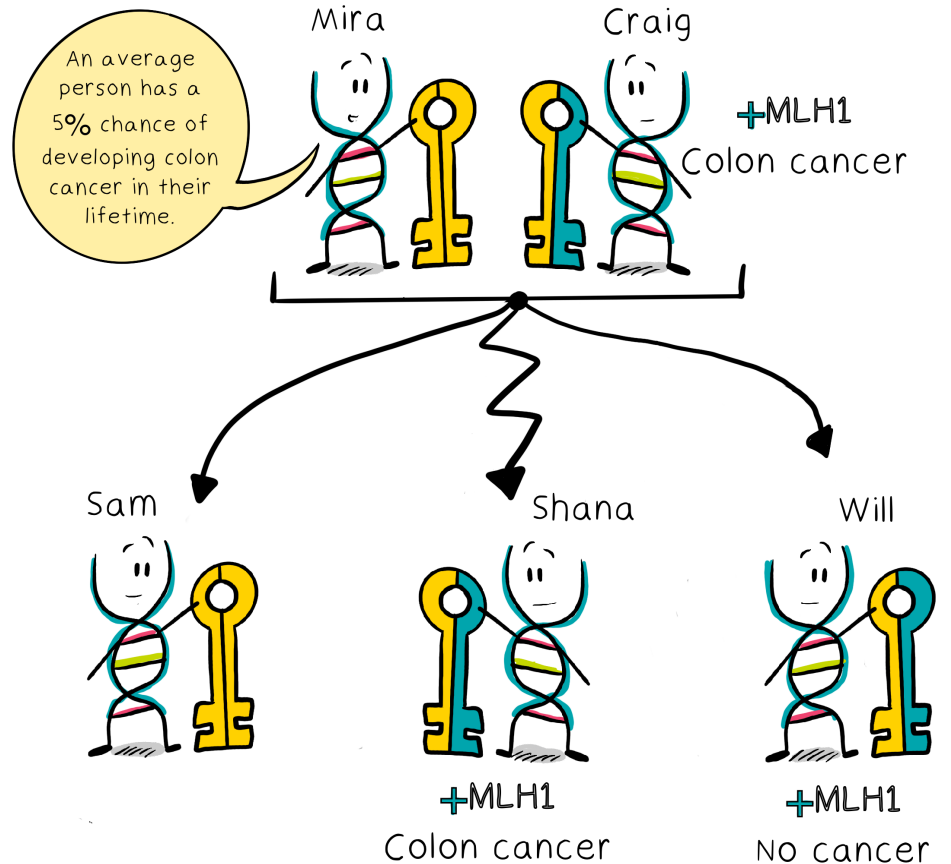


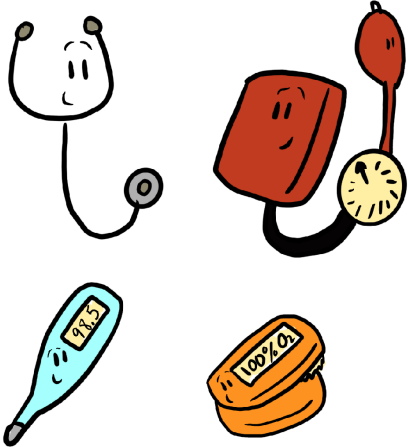
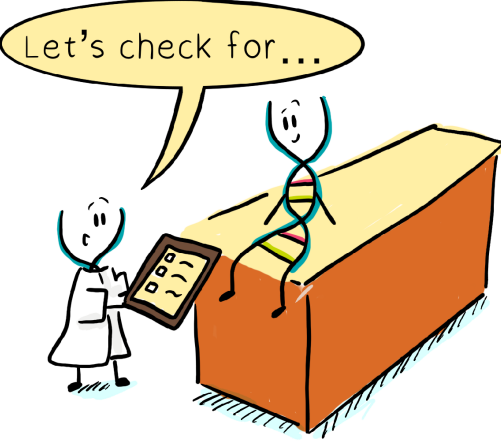


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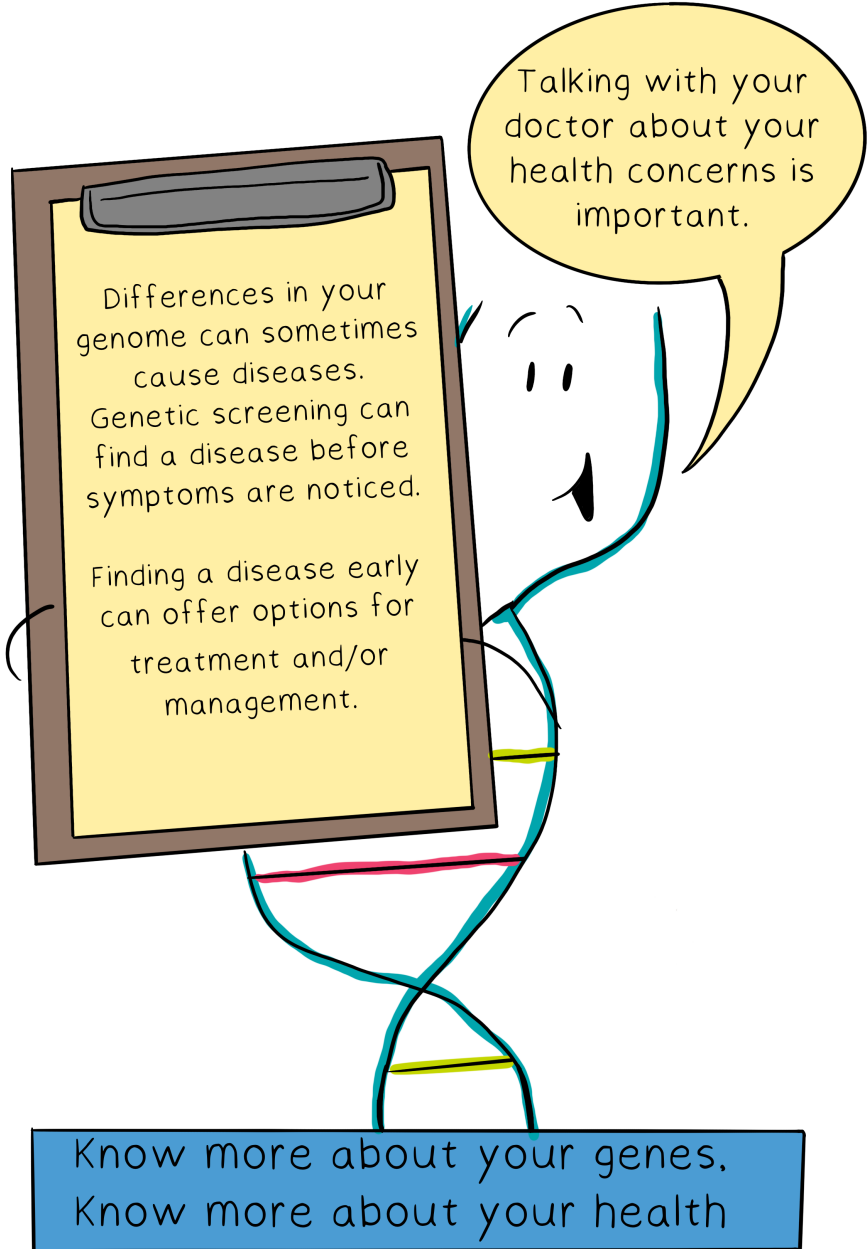
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<p>Genetic screening can help to find symptoms or evidence of disease early.</p>	<p>Finding the diagnosis can help doctors treat the symptoms.</p>
	
<p>If someone has symptoms, we do genetic testing.</p>	<p>Finding a disease early can improve options for prevention and better health outcomes.</p>
	



Talking with your doctor about your health concerns is important.

Differences in your genome can sometimes cause diseases. Genetic screening can find a disease before symptoms are noticed.

Finding a disease early can offer options for treatment and/or management.

Know more about your genes. Know more about your health

## Plain Language Glossary

### ○ DNA:

The blueprint for a person which is written in the letters "A, G, C, T" and inherited across each generation.

### ○ Gene:

A small piece of DNA that provides the code for specific functions or traits.

### ○ Trait:

Any characteristic of a person; can be based on one or multiple genes as well as the setting that a person lives.

### ○ Chromosomes:

Tight coils of DNA, containing multiple genes, held within each cell.

### ○ Genome:

All of a person's DNA organized in chromosomes.

### ○ Genetic variant:

A difference in the spelling of a gene's "ATGC" alphabet. Synonyms: mutation, variation, change.

### ■ Disease-related genetic variant:

A spelling difference that causes the gene to not work correctly and can cause disease.

### ○ Inherited:

A trait, disease, or condition that is passed down through a family by each generation.

### ○ Recessive disease:

A disease that happens when both of a person's two copies of a gene are not working.

### ○ Dominant disease:

A disease that happens when either one of a person's two copies of a gene is not working.

### ○ Affected:

A person who has symptoms of a disease or condition.

### ○ Unaffected:

A person who does not have symptoms of a disease or condition.