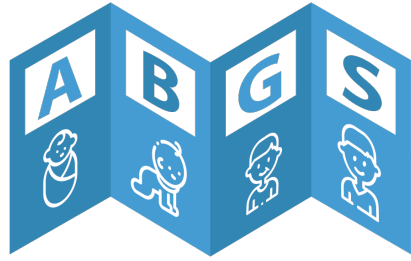


Genetic Testing

and

Screening

ABGS Module
Illustrated by Nicki Shaw



Age-Based Genomic Screening

Illustrated by Nicki Shaw, Olilu Designed
Website: oliludesigned.com

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Website: go.unc.edu/abgs

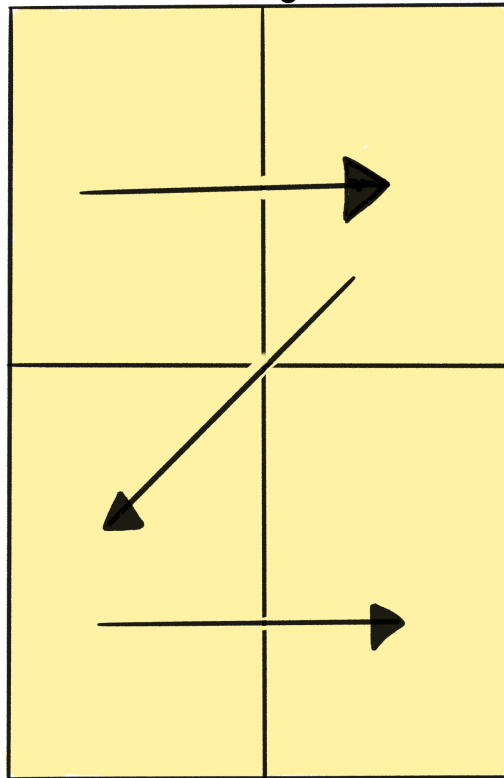
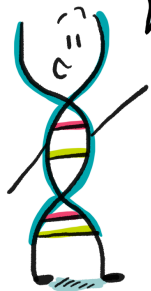
For inquiries and more information,
contact us at ABGS@unc.edu



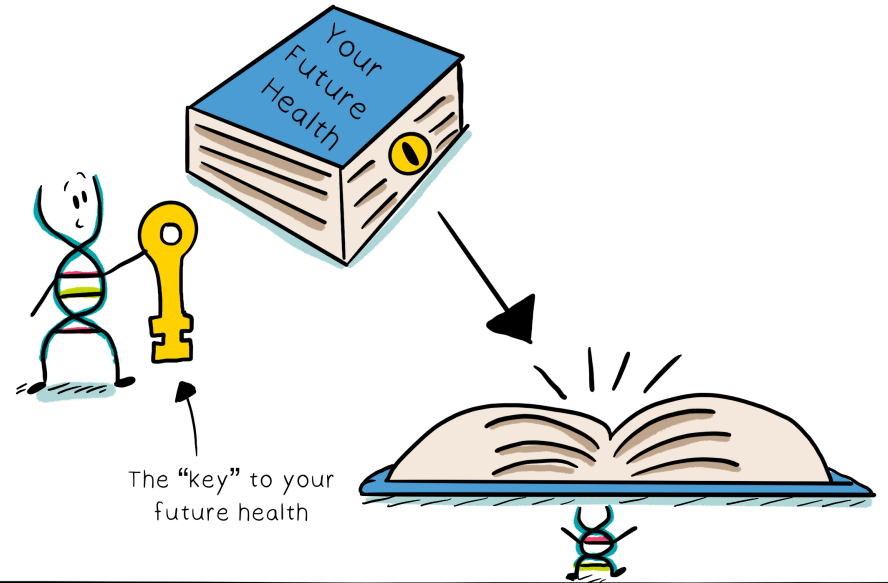
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Sample Comic Page

To read this comic, please start at the top, read left to right, and down the page. Like this!



Your DNA and family history can give us key information about your future health.



Let's take a deeper dive..

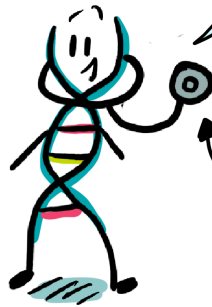


Chapter 1: Genetic Testing

Doctor!
What are
you doing
here?

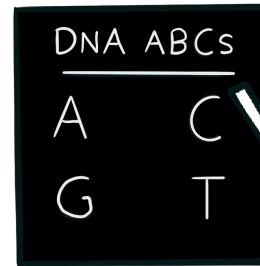


Hey Gene!
I'm here to help you find out
how you can learn more
about your future health!



stethoscope

The instructions that lie within your genes are written out with a special alphabet of A, G, C, and T. Sometimes changes in these genes cause the gene to work differently. These spelling differences within genes are called disease-related genetic variants.

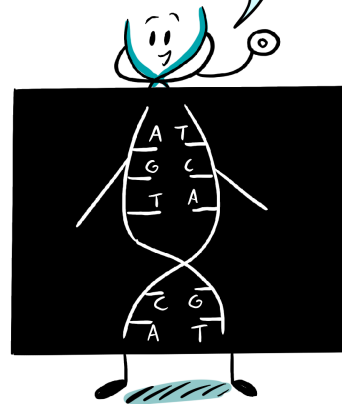


One way is through genetic testing! The goals of genetic testing are:

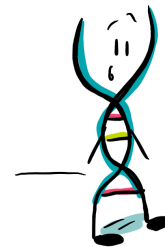
- To find a reason for symptoms or disease a person already has or health condition that runs in their family.
- Better health with options for treatment.



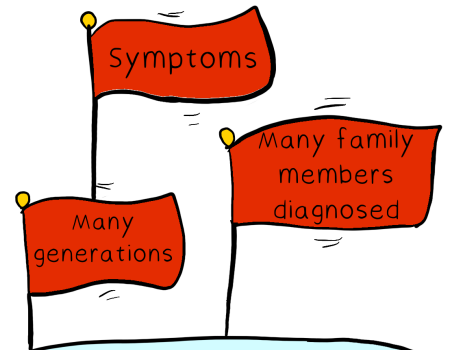
One of the tools we use for testing is DNA sequencing! It reads the instructions in our genes.



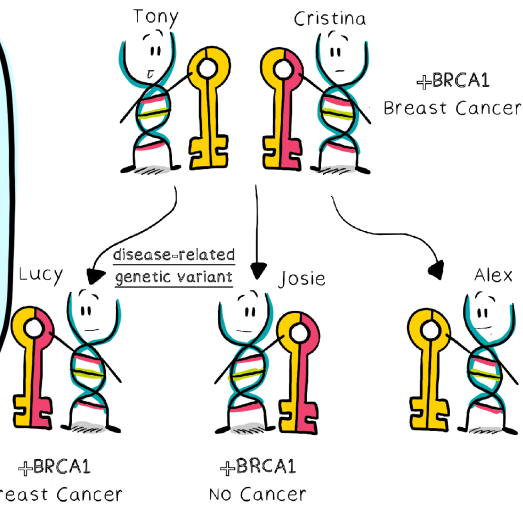
A person's medical history or family history can show red flags that they might have an inherited disease.



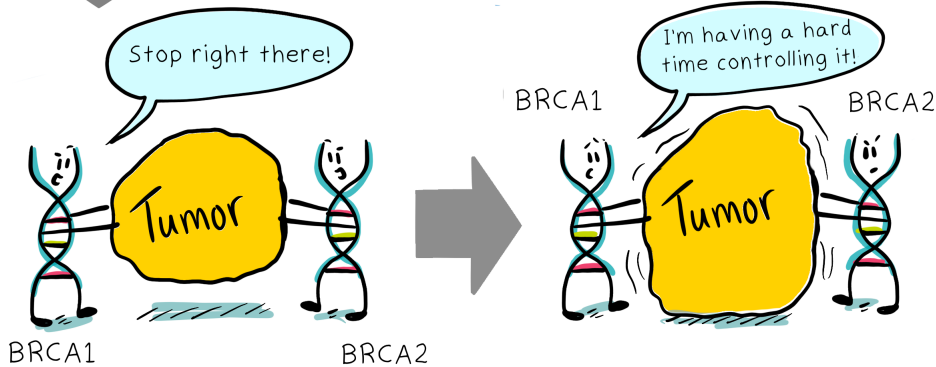
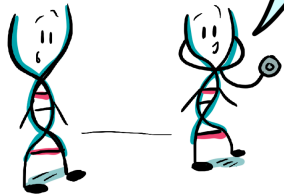
DNA sequencing to look for disease-related variants in people with symptoms or a family history is called genetic testing.



For some people with cancer, we can find a **disease-related variant** in a certain gene which increases a person's risk of cancer. These include variants in the BRCA1 and BRCA2 genes which are linked to increased risk of getting breast cancer.



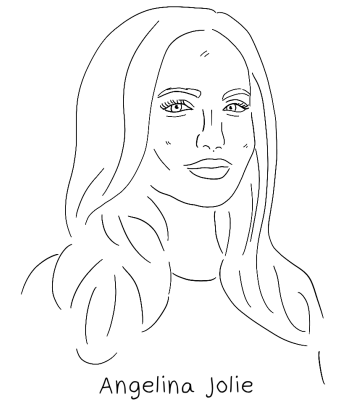
Josie and Lucy get the **disease-related variant** from their mom. However, as we see here, just having the variant does not guarantee a person will develop cancer but they have a higher risk of getting it.



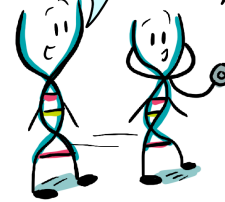
The job of the BRCA1 and BRCA2 genes is to help our body stop tumors from growing.

People who inherit this type of variant in a copy of their BRCA1 or BRCA2 gene are more likely to develop cancer (especially breast cancer).

For example, Angelina Jolie's mother had breast cancer and ovarian cancer and died at age 56. Her aunt and grandmother had it too. Genetic testing found that Angelina inherited the **disease-related variant** in BRCA1 from her mother.

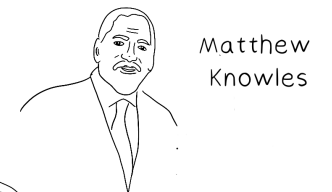


I love her movies!

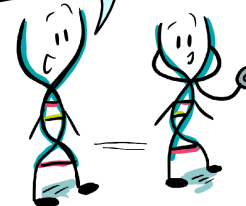


Because she learned of her high risk of breast and ovarian cancer, she decided to have surgery to remove her breasts and ovaries.

Matthew Knowles was diagnosed with breast cancer. Genetic testing found that he had a **disease-related variant** in one copy of his BRCA2 genes. Beyonce and Solange had genetic testing that found they did not inherit this variant from their father.



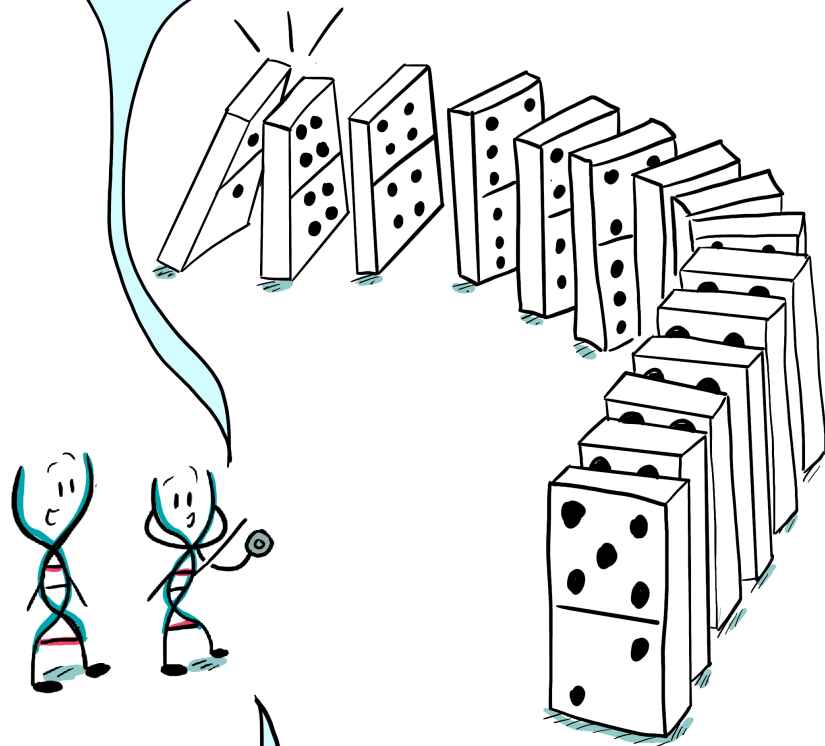
I love their music!



Because neither of the Knowles sisters inherited their father's variant, they do not have an increased risk for breast or ovarian cancer.

It is important to understand your family health history and learn if any relatives have undergone genetic testing for an inherited disease.

If there is a disease-related variant in the family, other relatives can have genetic testing for the same variant, called cascade testing.



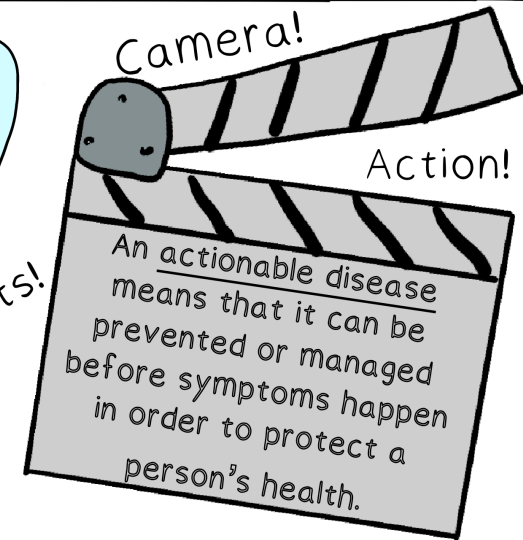
This is the type of testing that Beyonce, Solange, and Angelina had to look to see if they inherited their parents' disease-related variants. Genetic counselors can help with cascade testing.

Chapter 2: Population Screening

Next up, we're going to review routine population screening! This looks for diseases or health conditions that are actionable.

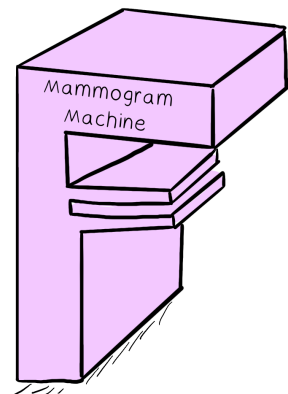
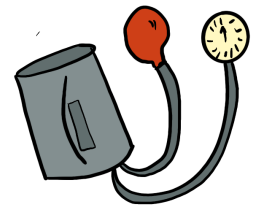
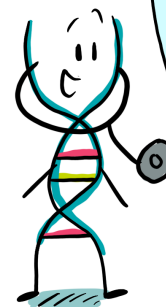


Lights!



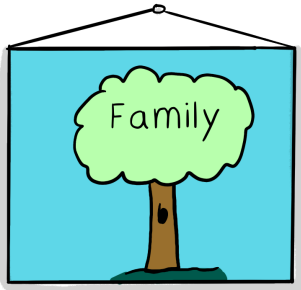
Examples of these screenings are:

- Blood pressure screenings can help people with increased risk of heart attack or stroke.
- Cancer screenings like mammograms are used to find tumors when they are newer and smaller.



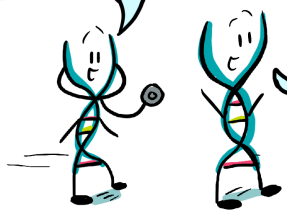
Chapter 3: Personalized Screening

Next up, personalized screening! This looks for **actionable** health conditions and diseases in people with **personal risk factors** and family history of disease.

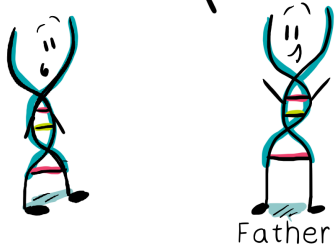


Oh, my parents are here!

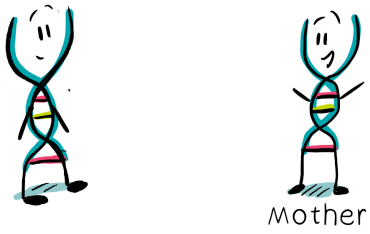
Hello!



Since we have a family member who had colon cancer at a young age, early colonoscopies are recommended for close relatives, like you!



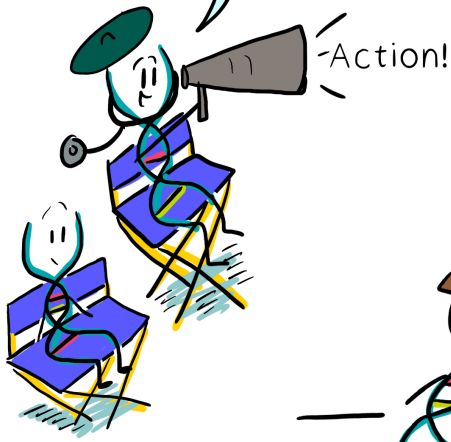
Since we have a family member who had a heart attack, blood pressure and cholesterol levels are checked more often for their close relatives, like you!



Chapter 4: Genetic Screening

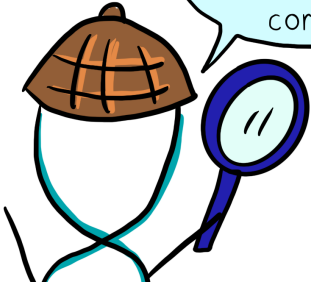
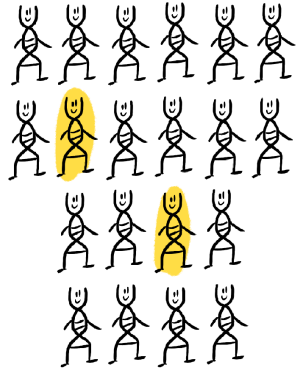
Lastly, we have genetic screening! Genetic screening is a type of routine population screening, like blood pressure screening.

It involves reading the DNA sequence of certain genes to look for **disease-related variants** that increase the risk of **actionable** disease for healthy people.



The Case of the Disease-Related Variant!

Ah hah! I found people with **disease-related variants** that cause their genes not to work correctly.

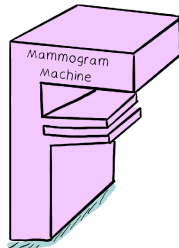


Genetic screening identifies about 1 person with a genetic condition out of every 20 people

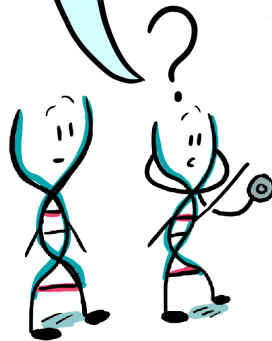
If a disease-related variant is found, doctors can take action to prevent or treat the disease early.

Medications to lower cholesterol can be taken.

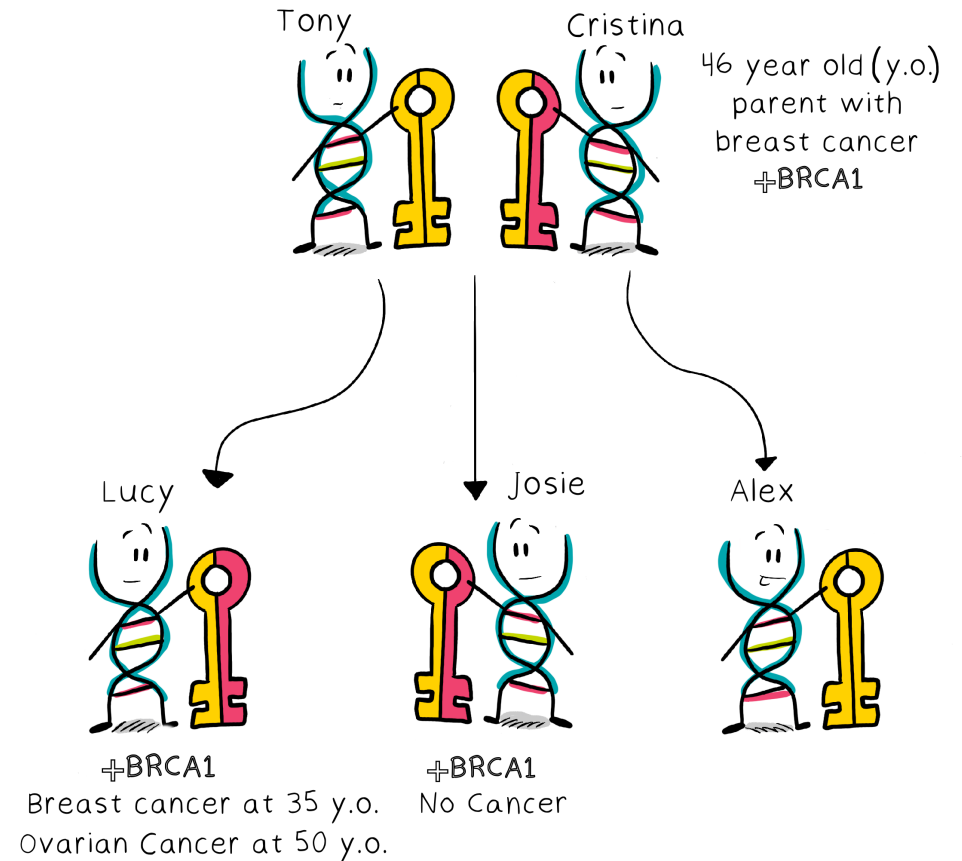
Mammograms can be used to look for breast cancer.



Sometimes people with the same genetic condition can experience it in different ways starting earlier or later in life and this can be challenging for doctors and patients.



Not everyone with the disease-related variant will have symptoms in their lifetime.

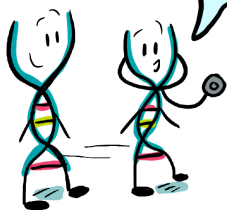


People with the genetic condition may have symptoms starting earlier or later in life and their symptoms may be more or less severe. This means that the actions that doctors recommend may help some people more than others and it is difficult to predict who will benefit the most.

Chapter 5: Summary

	Genetic Testing	Genetic Screening
Who?	For people with personal or family history of disease.	For the general population.
What?	Genes related to the disease in the family.	Genes related to certain actionable diseases.
Why?	Understand if a disease is due to an inherited risk in the family.	To prevent or treat disease early.
When?	Symptoms have already shown up in a person or their family.	Before symptoms appear.
Where?	Specialty Genetics Provider	Public health setting or primary care provider

Here's a handy chart to summarize everything we've talked about! Remember: Genetic screening is not a substitute for genetic testing if you have family history that could be explained by inherited disease.



Also, this information does not replace a good conversation with your doctor. Please talk to me about any specifics of your situation.

-Testing and screening are both used to help find health conditions, but are used in different situations and do not replace each other.

-**Genetic testing** is for people with certain personal or family histories to see whether they have an inherited condition. It can provide a diagnosis for symptoms.

-**Genetic screening** finds about 1 out of every 20 people with an actionable inherited condition they didn't know about. It can give information about risk and need for additional follow-up.

-**Actionable** conditions can be prevented or managed before symptoms occur.

-People with the same inherited condition can develop symptoms differently or not at all and this complicates follow up health care.

There are many more chapters in your health journey to come. Don't ever hesitate to call me with any questions!



Thanks, doctor!



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.

○ Population Screening: screening of the general public to find people who need health care to prevent or treat a disease they don't know about before symptoms happen.

○ Genetic Screening: a specific type of population screening that involves reading a person's DNA to see if they have any potentially disease-causing or "pathogenic" variants.

○ Genetic Testing: reading the DNA of certain genes in a person with family history or personal history of disease; can help diagnose a disease if a person is already showing symptoms.

○ Actionable: a condition that can be prevented or managed before symptoms happen in order to protect health.

○ Cascade Testing: a specific type of genetic testing to find out if a person inherited a disease-causing genetic variant that is known to run in a family.