

8.12

Genetic testing

Anki Vocab

- **Genetic testing** — The analysis of DNA to identify genes, mutations, or inherited conditions.
 - **DNA** — The molecule that carries genetic information.
 - **Gene** — A section of DNA that codes for a specific trait or protein.
 - **Mutation** — A change in the DNA sequence.
 - **Inherited condition** — A disorder passed from parents to offspring through genes.
 - **Carrier** — A person who has one copy of a recessive allele but does not show symptoms.
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What can we use genetic testing for?

There are four key applications:

1. Identifying inherited conditions
2. Determining whether a person is a carrier
3. Predicting the likelihood of developing certain conditions
4. Confirming biological relationships

It is important to note that within all of these applications, genetic testing does not *change* the DNA, it merely analyses it.

Example 1. Confirmation of biological relationships



(Query on next page)

Query

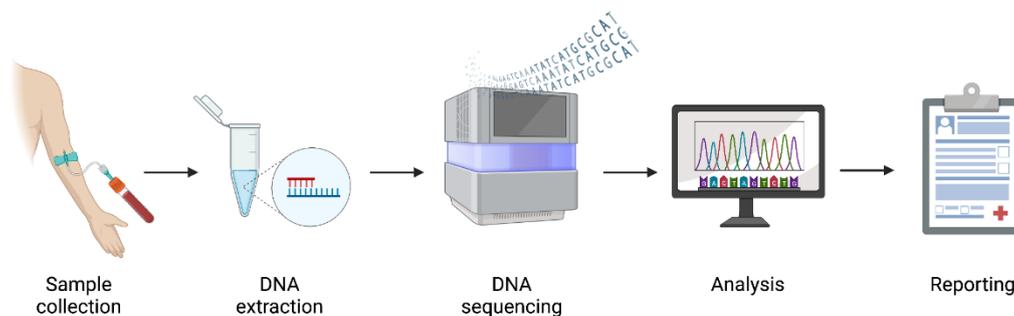
1. What is **Example I** above depicting? And what do you think these structures have to do with DNA and DNA testing?
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How does genetic testing work?

In order to perform a genetic test, we follow three main steps:

1. Collect
 - We need to collect a 'sample' from the individual we're testing that contains their DNA
 - And if we want to look at a specific *gene*, we'll need to 'extract' it from our sample we collected
2. Analyse
 - We can then look at the DNA or gene and even compare it with other samples that we have, to look for any *mutations*.
3. Interpret
 - After we've figured out what the DNA or gene looks like, we can give our results to a specialist, who can then interpret what its implications are for our individual.

Example 2. General genetic testing procedure



Queries

1. During our 'collect' step, what kind of 'sample' could we potentially collect from our individual so that we can access their DNA?
 2. What biological structure(s) contain the DNA in your example of a sample above?
 3. Can you recall the definition of a mutation?
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A genetic testing example: Testing for sickle cell disease

1. Collect

- In this case, our 'sample' is a little bit of blood that is collected from the heel of the individual
- This blood contains white blood cells. A white blood cell, like other somatic cells, has a nucleus. The nucleus of all somatic cells contains a full set of chromosomes. Therefore, our blood sample will contain all of our individual's DNA and genes, including a gene called the HBB gene.

2. Analyse

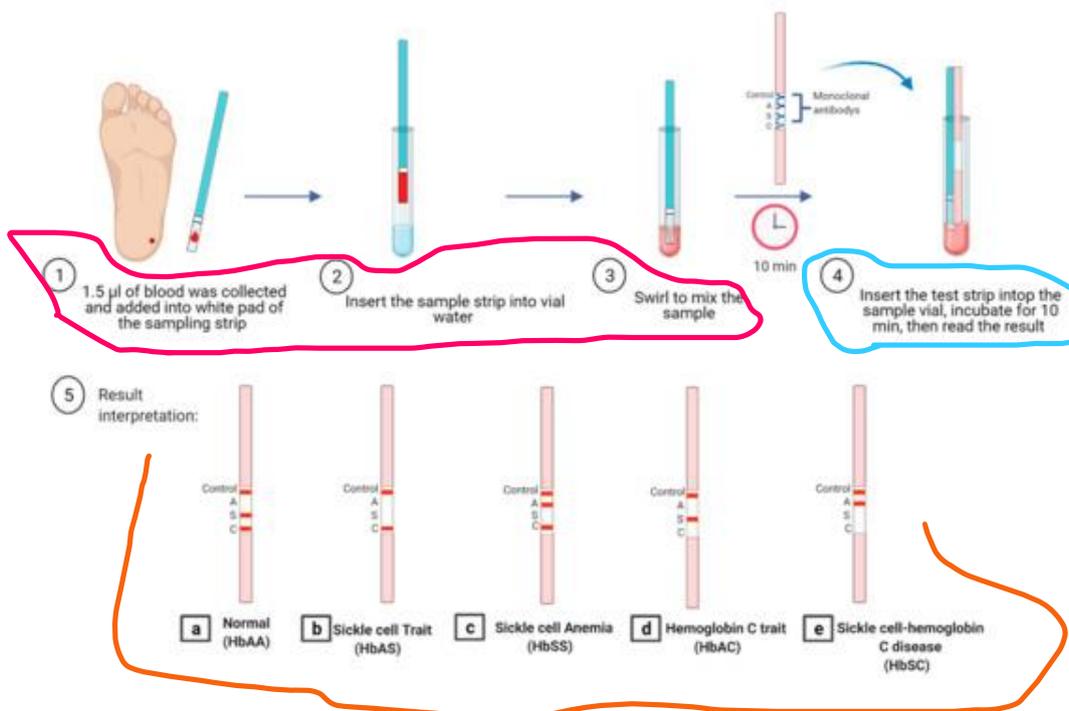
- We can now extract the individual's HBB gene from their DNA blood sample and have a look to see if it has any *mutations*.

3. Interpret

- We can then present our findings from the previous step to a professional, who can determine whether the individual's HBB gene has the mutation which causes sickle cell disease.

What's fascinating about this is that thanks to advances in biotechnology, we only need to worry about the collection step, as we now have 'sampling strips' that can do the analysis and interpretation for us (see **Example 3**).

Example 3. Genetic testing for sickle cell disease using a sampling strip



Collecting (done by us)

Analysing

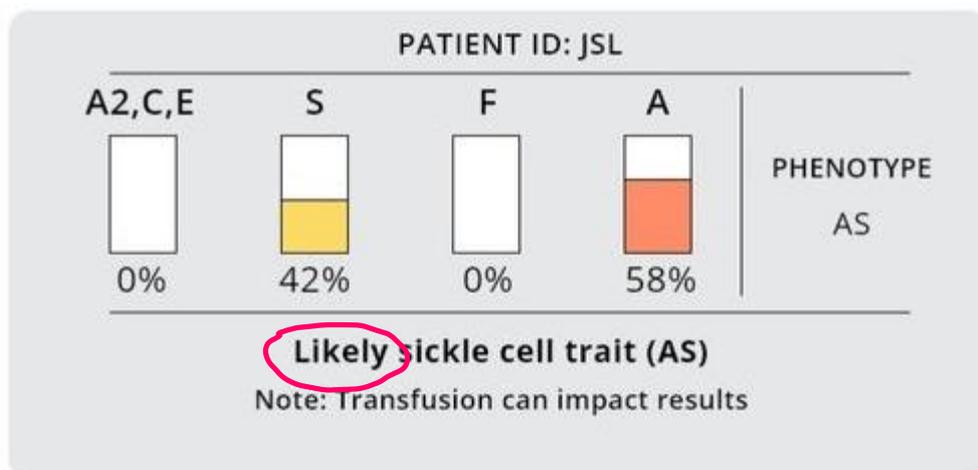
Interpreting

Genetic testing is useful, but limited

Genetic testing has various practical limitations:

- Testing 'positive' for a certain DNA test does not guarantee that a certain disease will occur. (See **Example 4**)
- Some results can be uncertain or inconclusive.
- Not all conditions have available tests.
- Environmental factors could still impact an individual's health.

Example 4. A sickle cell screening from 'Grace Laboratories'



'likely' ≠ guaranteed

Genetic testing also has various *ethical* considerations (**Queries**):

- **Privacy** — Who do you believe should be able access to your genetic test? And what protections, if any, do you think should be enacted before they can do so?
- **Discrimination** — What might an employer or insurance company do with your genetic test if they had access?
- **Psychological impact** — How would knowing that you are 'likely' to develop a condition affect your day-to-day life ? (even though it isn't guaranteed)
- **Consent** — Do you believe that testing should be voluntary and informed?

(cont. next page)

Benefits

Despite its limitations and ethical considerations, genetic testing presents various potential benefits:

- Early detection of diseases (see **Example 5**).
- Earlier and more informed treatment planning.
- Informed lifestyle and health choices.
- Informed family planning.

Example 5. Early detection of diseases via genetic testing is socially beneficial

Health Report

More Americans surviving cancer

Knowing genetic risk can increase early detection

By TIMOTHY POOR

ST. LOUIS – Finding out about her genetic risk for cancer may have saved Jackie Walker's life.

Four years ago, Walker decided to get a genetic test after a close friend was diagnosed with Parkinson's, an inherited disease. She met with a genetic counselor and had her cheek swabbed for DNA, then it was sent to a local lab and analyzed. The test showed that she had a genetic risk for colon cancer.

When she told her doctor what she learned, he said she should get a colonoscopy to check for colon cancer. She did, and the results showed early signs of cancer. Walker was 42.

Screening for colon cancer usually begins at age 50, but because Walker knew she had a genetic risk, she was tested sooner. Her cancer was found early and treated and Walker survived. Now 46 and in full remission, she runs three days a week and enjoys weekly outings in Forest Park with her husband and two daughters.

CANCER'S IMPACT
Like Walker, more Americans are



Jackie Walker says genetic testing helped find her cancer

can also determine if you have a greater risk of developing certain diseases. If you have the genetic marker for colon cancer, you are more likely to get it than someone without the marker.

The National Cancer Institute encourages Americans to understand their genetic make up. That means getting a genetic test, and going over the results with your doctor. That way, the doctor can determine your possible genetic risk for certain health problems.

Hereditary colon cancer typically occurs as a result of defects in a single gene received from one or both parents. People with this defective gene have a higher chance of getting the disease. Hereditary colon cancer is thought to cause up to 10 percent of all colon cancers.

WALKER'S MESSAGE
Walker says she wouldn't have known her risk for colon cancer if she hadn't done the genetic testing and told her doctor what she learned. Walker thinks that if others knew about genetic risks and genetic testing, more people would be screened earlier and

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Queries

1. Certain individuals (understandably) have reservations about genetic testing and could hence miss out on the benefits of early disease detection. What government based initiatives could you suggest that could help to alleviate these concerns and increase the percentage of the population that seeks out genetic testing early?