

My Genes My Choice

Genetic Testing Toolkit for Patients and Their Families



Name: _____

Hi! This is a toolkit designed especially for you to guide your discussions with your doctor/health team about having a genetic test. Before testing, discuss with your doctor/health team the possible benefits and limitations for you so that you can decide whether genetic testing is right for you at this time.

How to use this toolkit?



1. Read

You will find 9 boxes with key aspects that it might be helpful for you to consider when thinking about doing a genetic test. Read carefully each box and work through each numbered section in your own time.

2. Rate

After reading the information shared in boxes 1-8, use the 1-5/face scale to record your feelings and your understanding of the information.

At the end, count your final score by adding up the numbers you have put in each box. Box 9 is a space for you to write down any questions you have for your doctor/health team.



3. Discuss



We have suggested some questions for you to ask your doctor/health team but feel free to ask them whatever you want!

It may help you to get the best information you need if you share your thoughts and questions with your doctor/health team and share your final score with them.

How to use the 1-5/face scale?

After reading the information presented in each box, ask yourself:

- Is this information clear to me?
- Do I have questions/doubts I would like to share with my doctor/health team or somebody else?
- How do I feel, is there anything in particular I am concerned about?



1-5/face scale

Is this information clear to you?

Circle the face or number that fits best.



1. Don't Understand I have a lot of questions / doubts/
I feel very worried / confused



2. Not Sure I have questions / doubts/
I feel worried / confused



3. Understand Some I have some questions / doubts/
I am a bit concerned



4. Understand Most I have some questions / doubts/
I feel mostly calm and clear



5. Understand I do not have any questions / doubts/
I feel calm and certain

1 What are genes?

Our bodies are made up of billions of cells. Cells form the different parts of our bodies, for example our muscles, brain, heart. Inside each cell is our DNA, which contains thousands of genes. Genes act like instructions that tell our bodies how to make proteins, helping us grow, develop and stay healthy.

We all have natural differences in our DNA, called genetic variants. Some variants can affect how a protein is made and may increase the chance of disease. Others do not affect the protein and are not linked to disease.

We inherit two copies of most genes, one from each parent.

Is this information clear to you?

Circle the face or number that fits best.



1

Don't Understand



2

Not Sure



3

Understand Some



4

Understand Most



5

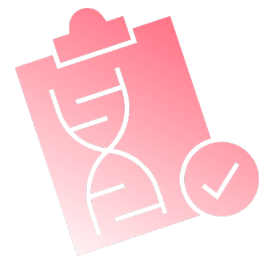
Understand

2. What is a genetic test?

A genetic test will look to see if your genes are giving your cells the correct instructions.

The test is arranged by your doctor/health team.

They will arrange to send a sample (either blood, saliva, cheek or skin cells) to a laboratory.



Is this information clear to you?

Circle the face or number that fits best.



1

Don't Understand



2

Not Sure



3

Understand Some



4

Understand Most



5

Understand

3. Why have a genetic test?

A genetic test can sometimes help explain the cause of a health condition. Knowing the genetic cause may:

- Help guide treatment, care and monitoring.
- Provide information about ways to reduce the chance of illness or make lifestyle changes.
- Help some people better understand why they have a particular condition.
- Show whether other family members may have a higher chance of having the same condition.
- Help people understand the chance of passing a genetic condition on to their children.

Genetic testing does not always provide clear answers, but the information can sometimes be useful for both you and your family.



Ask your doctor/health team:

- About possible improvements in your care
- About possible lifestyle changes that might help
- How your family might get access to a genetic test if you have a genetic condition

How do I feel about this information?

Circle the face or number that fits best.



1
Don't Understand



2
Not Sure



3
Understand Some



4
Understand Most



5
Understand

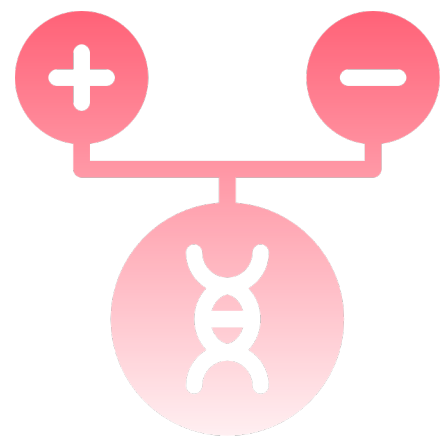
4. Are there any downsides to having a test?

Genetic testing can be helpful, but there can also be some limitations.

- The results may not always explain the cause of your symptoms or condition.
- Sometimes a test may find a genetic change that we do not fully understand, which can leave some uncertainty.
- Occasionally, testing may identify a chance of developing another health condition that was not the main reason for testing.
- A negative result does not always mean that there is no genetic cause or no future increased chance of developing a disease.
- Some people find that uncertain or unexpected results can cause worry or anxiety.
- In some countries genetic test results may influence health and life insurances, careers and plans to live abroad.

Ask your doctor/health team:

- What might happen if this is the case
- About what can be done for you and your family if you have a negative result, tell them your worries about doing the test.
- If they are aware of anyone in your country with experience of this.



How do I feel about this information?

Circle the face or number that fits best.



1

Don't Understand



2

Not Sure



3

Understand Some



4

Understand Most



5

Understand

5. What about the emotional side?

Genetic testing can sometimes bring up difficult emotions. Some people experience feelings of anxiety, guilt, blame or worry about themselves or their family. These are normal reactions and can take time to process.

Genetic test results may also affect family relationships. For example, family members may receive different results or have different views about whether they want genetic information.

Because genetic information is shared within families, your test result could reveal information about relatives that they may not wish to know. This can sometimes create tension or lead to difficult conversations.



Ask your doctor/health team:

- If support is available to discuss any concerns you may have.

How do I feel about this information?

Circle the face or number that fits best.



1

Don't Understand



2

Not Sure



3

Understand Some



4

Understand Most



5

Understand

6. What the results might say

Your result will say one of 3 things.

1. It will confirm that you have a genetic variant causing your condition.
2. It will say that no genetic variant was found.
3. You may have a genetic variant of uncertain significance (VUS)

A VUS is a genetic change where we do not yet know whether it affects how the protein works. Because of this, we cannot currently say whether it has any impact on health.

Ask your doctor/health team about what can be done for you and your family if you have a VUS.

Is this information clear to you?

Circle the face or number that fits best.



1

Don't Understand



2

Not Sure



3

Understand Some



4

Understand Most



5

Understand

7. How long will it take to get a result?

Genetic testing is often complex. It can take months, and in some cases, years to get a result.

Ask your doctor/health team how long it might take and how your result will be given.

Is this information clear to you?

Circle the face or number that fits best.



1

Don't Understand



2

Not Sure



3

Understand Some



4

Understand Most



5

Understand

Calculate your final score by adding up the numbers you put in each of the boxes above.

Record your score in the box to the right.

Genetic Testing Toolkit

8. How do I feel about genetic testing?

Below you will find some points and questions that may help you to think about what genetic testing means to you. You may want to ask yourself these questions and/or you may want to talk to your family about them. You can write down your thoughts here if it helps.

1. How important is it for me to have certainty about my diagnosis?

2. How would I feel if I didn't get a result?

3. How would I feel if I got an unexpected result?

4. What does my doctor/health team say about potential improvements in my care or treatment?

5. Does anyone in my family have similar symptoms to me?

6. If I have a confirmed genetic condition who else in my family has a chance of having the same condition?

7. Do they know that this is a possibility?

8. Would they want to be tested too?

9. Am I or is anyone in the family planning to have children?

10. Would I/they want or be able to have prenatal genetic testing?

How do I feel about my decision?

Circle the face or number that fits best.



1

Don't Understand



2

Not Sure



3

Understand Some



4

Understand Most



5

Understand



9