Consumer Information Sheet

Genetic variant of unknown significance

This leaflet is for people who had a genetic test, and the genetic test found a variant of unknown significance, also called a variant of uncertain significance or VUS.

Genetic testing is done to answer certain questions. Two common questions are

- Does my condition have a genetic cause?
- Can my relatives inherit my condition?

Unfortunately, genetic testing cannot always answer these questions.

DNA and genes

The human body is made up of millions of cells. Each cell contains DNA. DNA spells out the genetic instructions (genes) the cells need. Some genes tell cells how to grow, divide and work properly. Some genes help keep DNA healthy. DNA is spelt out by the 4 ‘letters’ of the genetic code (A, T, C and G).

DNA variation

Every person’s DNA has many genetic differences or variations.

- Most genetic variations are harmless. They do not cause a genetic condition or health problem. They are called normal variants.
- Some genetic variations change how a gene works and cause a genetic condition or health problem. There are many names for this type of variant. The names include disease-causing variant, pathogenic variant, mutation, genetic error and genetic fault.
- Some variants are not understood yet. They are called variants of unknown significance or VUS. We all have thousands of VUS, but we do not know about them unless we have a genetic test.

What does a VUS mean?

A VUS is neither good nor bad. Its meaning is just not known yet.

What does a VUS mean for a person’s medical care?

A VUS result cannot be used to make medical decisions. Medical care is based on other factors. These include a person’s medical and family history, and the results of other (non-genetic) tests.

Sometimes more testing can help to understand the meaning of a VUS. This is why people with a VUS should see a genetic health specialist.

Science is changing fast. With time the meaning of a VUS may become known. People should regularly ask their doctor if more has been learned about their VUS (usually every 3-5 years).

Where can I get more information or support?

Adult Genetics Unit, Royal Adelaide Hospital	Tel: 08 7074 2697
Centre for Genetic Education	http://www.genetics.edu.au
Genetics Home Reference	https://ghr.nlm.nih.gov/primer

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Talking to family and friends about your test result

This leaflet has been written for people who have had genetic testing for inherited variants in cancer genes. The leaflet does not replace a face-to-face discussion with a healthcare professional.

After getting your genetic test result you may decide to tell family members or friends about your result. People often struggle with the where, when, and how to share important information with family members, especially their children. This can be even harder when talking about potentially upsetting issues.

People may experience a range of emotions. They may want to 'spare' their relatives or children from the pain of difficult information or they may fear how relatives will react to and cope with the information.

Not telling does not change the reality of the situation. In fact, it makes family members powerless to make informed choices about their own current and future healthcare and plans.

Friends and family members may have an unwelcome reaction to your news. This is often because of a lack of knowledge and understanding about inherited risk. They may express different opinions about decisions you make, which can be difficult.

It is important to take care of your own needs and remember that decisions about genetic risk are very personal and may be approached differently by each person.

Talking to young children

Parents may fear telling their children about an inherited risk that may have a detrimental effect on their lives. However, being aware of the situation allows children to cope more effectively. Having knowledge will also help them with future decision making.

Children can be very perceptive. They may already be aware of the family history and be frightened for their parents or themselves. Children are often aware of tension and anxiety within the home. These tensions and anxieties may increase when a relative or parent is unwell or when that person is making difficult decisions around genetic testing.

Sometimes children use their imagination to try and make sense of a change in the adults around them. If their questions are answered in a supportive, loving, open and honest way a lot of their fears and anxieties will be addressed. If they feel listened to and understood they will also feel comfortable to ask more questions as they arise.

Relationships between parents and children can suffer if a parent is trying to hide information. Research has shown many people who are not told about genetic risk before reaching adulthood feel angry and resentful that information was kept from them.

Talking to teenagers

Teenagers are at a difficult developmental age. During this life stage, teenagers start building more relationships outside of the family and press their parents for greater independence. Their emotional and cognitive maturity is also developing. This means that a teenager's response to difficult information may vary from being child-like to being adult-like. This is normal for a teenager but can make communication more complicated.

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You will have already talked to your child about difficult issues as they were growing up (e.g. “the birds and the bees”). Draw on what has worked before and recognise that you already have skills that will help you to talk to your teenager about inherited risk.

Your family situation is unique. Your teenager may already be aware of the family cancer history and be thinking about what this may mean for them in the future or for other relatives. Talking to your teenager may give you a chance to find out what thoughts and feelings they have and to correct any misinformed beliefs.

As a parent you know your child or teenager best. You will know how they deal with new and potentially distressing information. You need to feel comfortable about speaking to them. You also need to be prepared for questions when you least expect them. If you do not know the answers to their questions, be honest and say so, but be prepared to look for answers with your child.

Some practical suggestions from other parents

A recent research paper[1] asked parents what advice they would give other parents who need to talk to their children about an inherited gene variant in the family. They emphasised the importance of preparation and careful consideration before deciding when and how to speak to their child.

The parents in the research study suggested:

> Parents consider many things including
  – their child’s maturity, level of anxiety, and awareness of genetics
  – the family history of illness and any current stressful issues in the family
  – their child’s level of concern for their parent who has the gene variant
> Parents be conscious of their own reasons for telling their child, think about their own wishes and hopes about the process and outcome of telling.
> If parents decide to tell, discuss the setting and timing of telling.
> For couples, consider if it is okay for one parent to do the telling on their own if an appropriate time unexpectedly arises.
> Discuss the actual words to use to convey the information (the parents in the research study felt that this was where the most help was needed).
> Think about if it would be useful to have help from friends, relatives or professionals.
> Recognise that you don’t need to tell each child everything at the first discussion; it is important to wait for reactions and answer questions.
> Remember the most important outcome is to keep communication open so fears and misconceptions can be addressed.
> Know that most parents find telling a relief.
> If parents decide not to tell they suggested planning for when and how to convey the information in the future.

Extra support

The staff of the Adult Genetics Unit can help you and your family work through how to speak to children and other relatives.

> Adult Genetics Unit, Royal Adelaide Hospital
  Telephone: 08 7074 2697, Email: adultgenetics@sa.gov.au

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1. Reference: Talking to Children About Maternal BRCA1/2 Genetic Test Results: A Qualitative Study of Parental Perceptions and Advice. Patenaude et al, J Genet Counsel 2013;22:303-314

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Consumer Information Sheet

Coping strategies for people with a cancer gene variant

This leaflet has been written for people who have had a cancer gene variant found on genetic testing. It does not replace the need for a face-to-face discussion with an experienced genetic professional.

Being identified as having a cancer gene may be unexpected and many people will require a period of psychological adjustment. Repeated readjustment may be needed at different times in life as a person's circumstances change.

Coping is the attitudes and behaviours people use to meet goals and challenges in their life, and to promote emotional well-being. Different people use different coping strategies and people may use different strategies at different times in their life. Below are some attitude and behaviour strategies you can consider:

Attitudes

Challenge negative and unhelpful thoughts

- Try to disengage the gene from the cancer diagnosis, remember one does not always equate to the other
- Recall the reasons you undertook genetic testing
- Acknowledge the feelings behind negative thoughts and give yourself permission to have an "off" day

Find positives

- The results might present a new opportunity for cancer treatment
- You may have more cancer screening or prevention opportunities
- Remember genetic information may be valuable for your family members, and could help reduce or prevent cancer

Maintain self-esteem

- Create time to pursue activities or hobbies you enjoy and time to relax
- Reduce the expectations you place on yourself

Reduce stress in your life

- Try to keep positive
- Accept there are things you cannot control
- Be assertive (not aggressive)
- Exercise, eat well, rest and sleep

Take it one day at a time

- Focus on what you can achieve today rather than challenges that may lie ahead
- Don't let your enjoyment of the present be ruined by worrying about the future

Behaviours

Establish a plan

- Set dates for follow up with your specialist
- Discuss the treatment options with your specialists

Find different ways to manage anxiety

- Hyper-vigilance or heightened anxiety will usually reduce over time
- Remember today's feelings won't be forever

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Develop a positive therapeutic relationship with your doctors

- Ensure decision-making is shared
- Build a relationship based on trust and confidence
- Be prepared to seek a second opinion

Adopt a participatory stance

- Asking questions and discussing options may help you feel more in control
- Be aware that avoidance is a tricky long-term strategy

Timing is everything

- Tell people about your test result when you are ready
- Draw boundaries you are comfortable with

Support

Express your emotions

- Find an outlet for your feelings
- Use a professional or “outside” person if you want to protect people close to you or avoid over-burdening them

Reach out for support

- Use friends or family to accompany you to difficult appointments
- Find others in similar circumstances (support groups, peer support or web sites)

Relaxation Exercises

2 minute relaxation. Switch your thoughts to yourself and your breathing. Take a few deep breaths, exhaling slowly. Mentally scan your body. Notice areas that feel tense or cramped. Loosen up these areas, letting go of as much tension as you can. Rotate your head in a smooth, circular motion once or twice. (If any movement causes pain, stop immediately). Roll your shoulders forward and backward several times. Let all your muscles completely relax. Recall a pleasant thought for a few seconds. Take another deep breath and exhale slowly. You should feel relaxed.

Mind relaxation. Close your eyes. Breathe normally through your nose. As you exhale, silently say to yourself the word “one”, a short word such as “peaceful”, or a short phrase such as “I feel quiet”. Continue for 10 minutes. If your mind wanders, gently remind yourself to think about your breathing and your chosen word or phrase. Let your breathing become slow and steady.

Deep Breathing Relaxation. Imagine a spot just below your navel. Breathe into that spot and fill your stomach with air. Let the air fill you from the navel up. Let it out, like deflating a balloon. With every long, slow breathe out, you should feel more relaxed.

The staff of the Adult Genetics Unit are available to answer your questions or to help you and your family adjust to learning about a cancer gene variant.

Adult Genetics Unit, Royal Adelaide Hospital	Tel: 08 7074 2697

More information, including peer support, is available online

Inherited Cancers Australia	https://www.inheritedcancers.org.au/

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Emotional responses following genetic testing

This leaflet has been written as an aid for people who have had genetic testing in the cancer setting. The leaflet does not replace a face-to-face discussion with an experienced health professional.

Genetic testing for a familial predisposition to cancer can raise complex emotions. These feelings can occur when the test is done, while waiting for the results, or when you receive the result. Sometimes these feelings can occur later, days, weeks, or month) after you received your test results.

Of course, not everyone has difficult thoughts or feelings. Everyone is different and everyone's experience is different.

This leaflet briefly outlines the range of emotions some people experience so you can be prepared. Expressing these feelings and talking them through with a family member or friend can help. You can also seek professional support.

Waiting for the test result

Many people find waiting for the test result is stressful.

- During the wait it is sensible to be kind to yourself and recognise that you are in an unusual situation. Genetic testing is not commonplace and may be an experience many of your friends, acquaintances or colleagues cannot easily relate to.
- If you find that you are "obsessing" about the result and the "what ifs", try writing down the concerns and questions you have, so you can bring them up with your doctor.
- During the wait for your result try to do activities that you find enjoyable and may take your mind off the waiting.

If you have a cancer gene variant

People who are found to have a cancer gene variant may experience fear, anxiety, sadness, and powerlessness. There may also be a sense of anger.

Some people also experience distress in the form of intrusive thoughts about their health. They may dwell on the result, have recurrent thoughts, or think about it when they don't mean to.

On the other hand, some people are relieved that the uncertainty has been resolved and they may be able to access more or different treatments options. There can also be a heightened sense of being in control.

These thoughts and feelings may fluctuate from time to time, with marked "highs" and "lows" that do not seem to make sense. They may also be emphasised at special times such as anniversaries or if another family member becomes unwell.

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If you do not have a cancer gene variant

People who do not have a cancer gene variant found may have a sense of relief upon receiving their test result. This result may be good news for their family and help reduce worry.

However, some people feel disappointed they do not have a cancer gene variant, as this may reduce or change the treatment options available.

Some people experience both types of emotions. These emotions can be puzzling and distressing but are normal.

Responses in the families

Other members of your family may not feel the same way before and after the test result as you did. Everyone is different and there is no “right” way to respond to having a genetic test.

You may find other family members have significant emotional responses to your test result. For example, a parent may feel anxious or guilty if their child has a gene variant. These are common responses. It is important to be open, supportive, and respectful of other family members responses.

Preparing for your result

People who prepare for how they might feel when they get their test result and who talk openly about this seem to cope better with learning that they have a gene variant.

In addition, having a plan of what steps to take if a gene variant is found helps many people to feel more in control and less anxious.

Extra Support

Remember there people who can provide you with advice and support throughout the process of genetic testing and adjusting to your result.

If you have questions or concerns, or you would like extra support you can discuss your worries with your specialist, family doctor (GP), or contact the Adult Genetics Unit.

> Adult Genetics Unit, Royal Adelaide Hospital	Tel: 08 7074 2697

Knowing there are other people experiencing similar things can also be helpful and online groups may also provide extra support.

> Inherited Cancers Australia	https://www.inheritedcancers.org.au/

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