NHS
Genomic Medicine Service

Von Hippel-Lindau Disease (VHL)

Information for patients



This booklet was given to you by:

Clinician: _____

Contact details: _____

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Why have I been given this information booklet?

Finding out that you or a family member has von Hippel-Lindau Disease (VHL) can feel worrying. This booklet has been written to help you understand the condition and answer some of the questions you may have.



It explains what VHL is, how it is inherited, the health problems it can cause, and the screening, treatment and support available. We hope this information helps you and your family feel informed and supported.

Many people find that learning more about VHL and understanding their healthcare options helps them feel more in control. Your healthcare team is always happy to answer questions and provide support.

What is von Hippel-Lindau Disease?

Von Hippel-Lindau (VHL) disease is a rare inherited condition caused by a change in a gene called VHL. It is named after the two doctors who first described the condition.

VHL can increase the chance of developing certain growths and tumours in different parts of the body. Some of these can cause health problems if they are not found and treated.

The good news is that regular screening can help find problems early, often before they cause symptoms. When problems are found early, treatment is usually more successful.

There are now many effective treatments available for people with VHL, and research is continuing to improve care and treatment options. Because of this, the outlook for people with VHL is better than ever.

What Health Issues Can VHL Cause?

VHL can affect different parts of the body. The areas most often affected are the eyes, brain (cerebellum), spinal cord, kidneys, adrenal glands and pancreas.

Not everyone with VHL will develop all of these problems. Some people may have only one or two health issues, while others may develop several during their lifetime.

VHL affects each person differently, even within the same family. For example, one family member may develop an eye problem, while another with the same genetic change may develop a kidney problem instead.

The age at which problems develop can also vary. Some people develop symptoms when they are young, while others may not have any problems until later in life.

1) Eyes



Small clusters of abnormal blood vessels can develop on the retina, which is the light-sensitive layer at the back of the eye. These are called **retinal angiomas** or **capillary haemangioblastomas.**

These often do not cause any symptoms and may only be found during an eye examination by an eye specialist.

If a retinal angioma is not found and treated, it can grow larger. This may cause bleeding or damage to the retina, which can affect eyesight and, in some cases, lead to vision loss.

Regular eye checks are important so that retinal angiomas can be found and treated early. Treatment options include laser treatment, injections, radiotherapy or surgery.

2) Brain and Spinal Cord

People with VHL can develop growths called **haemangioblastomas** in the brain or spinal cord. These growths are not cancer and do not spread to other parts of the body.

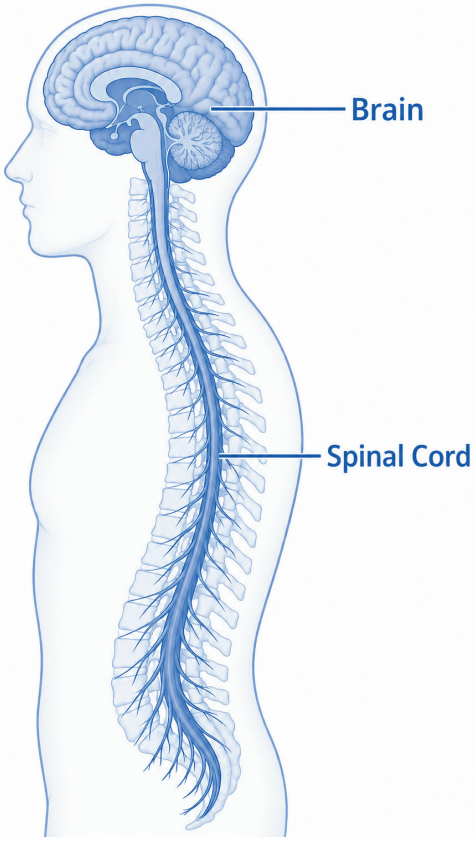
They can be found using MRI scans of the brain and spine. Small haemangioblastomas do not usually cause any symptoms. However, if they grow bigger, they can cause problems.

Haemangioblastomas in the brain may cause:

- Headaches
- Feeling sick (nausea)
- Dizziness
- Problems with balance or walking

Haemangioblastomas in the spinal cord may cause:

- Pain
- Numbness or tingling
- Muscle stiffness or weakness
- Affect the way you pass urine or open your bowels.

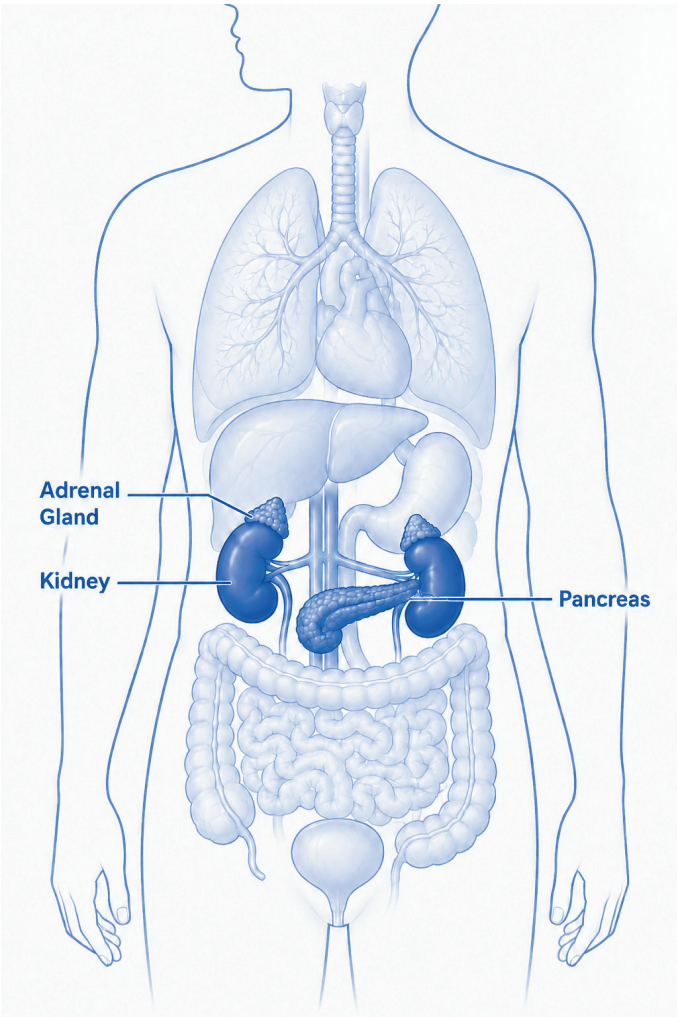


If a haemangioblastoma becomes large and causes symptoms, an operation may be needed to remove it. Some smaller haemangioblastomas can be treated with a type of targeted radiation treatment called **stereotactic radiosurgery**.

Regular MRI scans help to find these growths early so they can be monitored and treated if needed.

3) Kidney and pancreas

People with VHL often develop cysts in their kidneys. Cysts are fluid-filled sacs that are not cancerous and do not usually cause symptoms.



Over time, some kidney cysts can become more abnormal, and a small number may eventually develop into kidney cancer. When kidney cancers are small, they are unlikely to spread to other parts of the body and can usually continue to be monitored.

Regular scans are used to check for kidney cysts and tumours. This helps doctors identify any concerning changes and decide if and when treatment is needed.

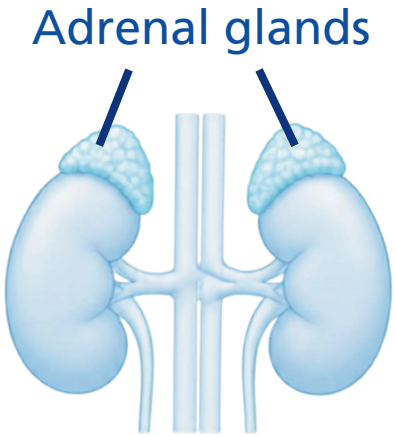
If a kidney cancer grows larger, treatment may be recommended. This may include surgery to remove the tumour. Some people may be able to have a treatment called **cryoablation**, which destroys the tumour by freezing it.

Cysts can also develop in the pancreas. These are usually not cancer and often do not need treatment.

Some people with VHL may develop **pancreatic neuroendocrine tumours (PNET)**. These tumours are different to more common types of pancreatic cancer and may grow slowly. Treatment may include surgery, medicines, or both.

4) Adrenal glands

The adrenal glands sit on top of the kidneys. In some people with VHL, a growth called a **phaeochromocytoma** can develop there. These growths can make extra stress hormones, which may cause:



- headaches
- sweating
- a fast or pounding heartbeat
- high blood pressure
- feeling shaky, anxious or panicky

Some people have no symptoms, which is why regular checks are important. If a phaeochromocytoma is found, medicines are usually given to control symptoms before surgery is arranged. Some families with VHL are more likely to develop phaeochromocytomas and may need screening more often.

5) Other possible VHL growths

VHL can also cause less common growths in other parts of the body, including:

- **Inner ear**: rarely a small non-cancerous growth can develop in the inner ear called an **endolymphatic sac tumour**, which can affect hearing or balance. A MRI scan can detect this.
- **Reproductive organs**: Some men with VHL can develop a non-cancerous growth called a cystadenoma in the epididymis, a tube next to the testicle. These growths are usually harmless but may be noticed as a lump.

Not everyone with VHL will develop these growths. Your specialist team will arrange monitoring that is right for you.

What age can VHL problems start?

The age when VHL causes health problems can be very different from person to person. Some people develop signs of VHL in childhood, while others do not have any problems until they are adults.

Eye haemangioblastomas (retinal haemangioblastomas) are often one of the first signs of VHL and can sometimes develop in children.Other VHL-related tumours are more likely to develop in teenagers or young adults, but they can occur at any age. Some people do not develop any VHL-related health problems until their 50s or 60s.

Because eye problems can happen early, regular eye checks usually start from the age of 5 years.

What does screening involve?

The aim of screening is to find health problems early, before they cause symptoms. Finding problems early can make treatment easier and more successful.

The tests and scans you need may vary depending on your age, family history and previous test results. People with VHL usually have regular check-ups throughout their lives.

From 5 years of age

- A yearly appointment with the Clinical Genetics team.
- A yearly eye examination with an eye specialist (ophthalmologist) to check for retinal angiomas at the back of the eye.

From 10 years of age

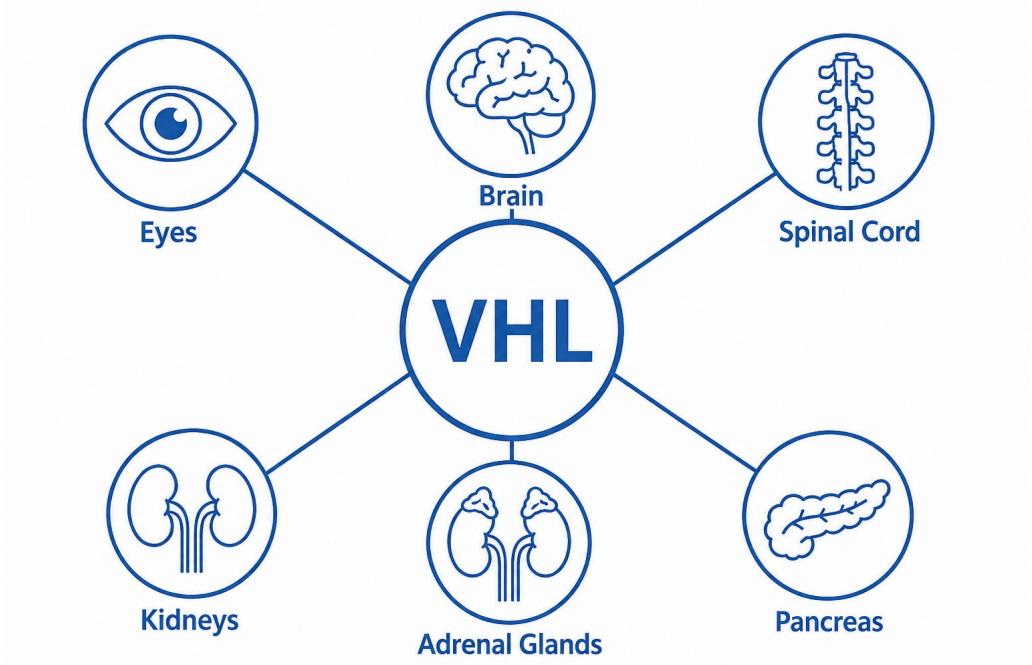
- A urine or blood test every year, or sometimes every 6 months, to look for signs of a phaeochromocytoma. How often this test is needed depends on the type of VHL in the family.

From 15 years of age

- Yearly scans of the kidneys, pancreas and adrenal glands. These may include ultrasound scans and MRI scans.
- MRI scans of the brain and spine to check for haemangioblastomas.The timing of future scans will depend on the results.

Some people may need more frequent scans or appointments with other specialist teams.

If any problems are found, your healthcare team will discuss the best plan for monitoring or treatment. Regular screening helps to find problems early and keep you as healthy as possible.



Getting help early

If you have VHL and notice any new symptoms, or if you are worried about your health, please contact your GP or specialist team. Getting advice early can help find and treat any problems as soon as possible.

Are there any other treatment options?

Most people with VHL do not need regular medication for the condition itself.



Treatment usually focuses on screening, monitoring, and treating any tumours or other health problems that develop.

In some situations, doctors may consider a medicine called belzutifan. Belzutifan works by blocking signals linked to the VHL gene that can help tumours grow.

It may help to slow tumour growth or shrink some tumours, but it is not effective for all tumours. Side effects include anaemia (a low red blood cell count) and shortness of breath.

Belzutifan may be considered for people with VHL-related tumours, particularly when other treatment options are not suitable. In the UK it is not currently a routine treatment for everyone with VHL and there are national rules about when it can be used. A team of specialist doctors with experience in treating VHL will carefully consider the possible benefits and side effects, as well as patient preferences, to decide if it is suitable.

If Belzutifan might be helpful for you, your doctor will discuss your case with a specialist team. They will explain the treatment, its possible side effects, and whether it is an appropriate option for you.

If you have questions about Belzutifan, please speak to your doctor, who can advise whether it may be relevant in your individual circumstances.

What is a gene?



Our genes are like a set of instructions inside our body. They tell our body how to grow, work, and look. Each gene has a specific job, such as controlling cell growth or helping prevent cancer.

What is a gene change?

Sometimes, a gene has a change that stops it working properly. If this change causes a health problem, it is called a pathogenic variant.

You may hear different words for this, such as “mutation” or “gene alteration”. In this leaflet, we will simply use the term gene change to mean any change or difference in a gene that causes von Hippel-Lindau (VHL) disease.

What is the VHL gene?

VHL is the name of a gene that helps control how cells grow and divide. It acts as a natural brake to stop cells growing when they should not.

Everyone has two copies of the VHL gene. If one copy contains a gene change, it may not work properly. This can increase the chance of developing certain tumours and cysts in different parts of the body, including the eyes, brain, spinal cord, kidneys, pancreas and adrenal glands.

Is VHL always inherited?

About **8 out of 10 people** with VHL inherit the gene change from a parent who also has the condition.

About **2 out of 10 people** have a new gene change. This means they are the first person in their family to have VHL. This is called a **de novo** gene change.

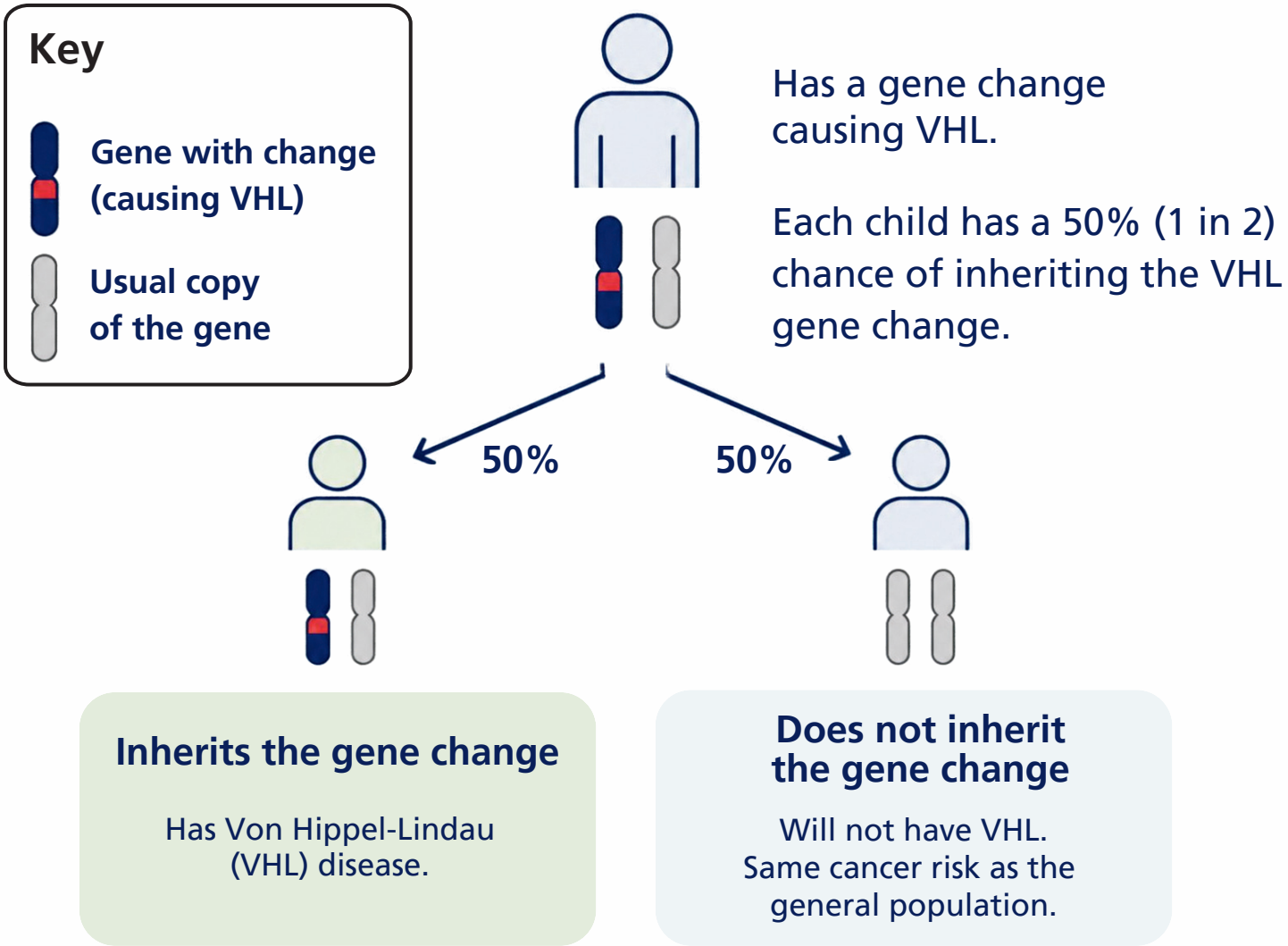
How is a VHL gene change inherited?

Everyone has two copies of the VHL gene, one inherited from each parent. If a person has a VHL gene change, each of their children has:

- a 50% (1 in 2) chance of inheriting the VHL gene change
- a 50% (1 in 2) chance of inheriting the usual working copy of the gene

This chance is the same for both men and women. If a person inherits the VHL gene change, they have von Hippel-Lindau (VHL) disease. This pattern of inheritance is called autosomal dominant inheritance.

Inheritance of VHL
(Von Hippel-Lindau Disease)



Genetic testing for VHL

It is now possible to find the VHL gene change in most families with VHL. Once a gene change has been found, other family members can have a genetic test to see whether they have inherited the same gene change. This is called a predictive genetic test.

Testing someone who has VHL
The first test is usually carried out on someone who has a health problem linked to VHL. This is called a diagnostic genetic test. The test looks for a change in a gene that may explain the person's health problems. Results can take a few months, depending on the laboratory used.

There are three possible results:

1. **Negative result - No VHL gene change is found.**
This means it is unlikely that the person's health problems are caused by a change in the VHL gene. However, current testing does not identify every VHL gene change. A small number of people (about 5%) with VHL may still receive a negative test result despite having the condition.

   If this result happens, the genetics team will review the person's medical and family history to decide whether any further screening or genetic testing is needed.

   If no VHL gene change is found, predictive genetic testing cannot be offered to other family members.

2. **Positive result - A VHL gene change is found.**
This confirms that the person has VHL and may be at risk of developing health problems linked to the condition.

   Other family members can then be offered a genetic test to find out whether they have inherited the same gene change. This is called a predictive genetic test.

3 Uncertain result

Sometimes a change is found in the VHL gene, but it is not clear if it causes VHL. This is called a variant of uncertain significance (VUS).

Everyone has small differences in their genes. Most of these differences are harmless, but some can cause VHL. Sometimes there is not enough information to know whether a gene change is harmless or causes VHL. More information about the gene change may become available in the future.

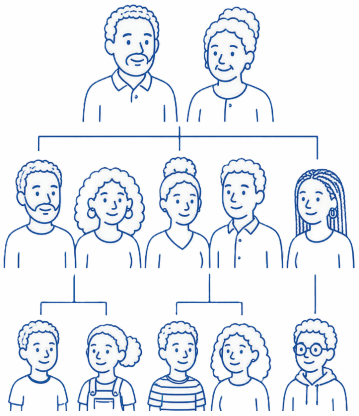
The genetics team will review the family history and advise whether any extra screening is needed for other family members.

Because it is not clear whether the gene change causes VHL, predictive genetic testing is not offered to other family members.



Can my relatives be tested for VHL?

Yes, if a gene change in the VHL gene is found in the family genetic testing can be offered to other family members. This is called a predictive genetic test.



Predictive genetic testing involves a blood test. Before the test, their doctor or genetic counsellor will explain what the results could mean for you and your family and answer any questions you may have.

Who is usually tested first?

Testing is usually offered in steps. This means that close relatives, such as parents, brothers, sisters, and children from the age of 4 years, are normally tested first.

How do I decide whether testing is right for me?

Having a predictive genetic test is a personal choice, and not everyone wants to know this information. It is important to think about the timing of the test, your age, the screening options available to you, and how you might feel about the result. If you decide not to have testing now, you can contact the Genetics Service again in the future to discuss it.

Will a predictive genetic test affect my insurance?

In the UK, there is a Code on Genetic Testing and Insurance. This means insurance companies cannot ask for the results of a predictive genetic test, except in very limited situations.

More information is available from the Association of British Insurers - www.abi.org.uk - or Genetic Alliance - www.geneticalliance.org.uk/insurance-and-genetic-conditions-faqs

If you decide to take out life, critical illness, or income protection insurance, it is a good idea to use an accredited insurance broker and read the policy details carefully.

Is there an alternative to predictive testing?

We strongly encourage people who may have inherited VHL to consider genetic testing. The test can show whether you have inherited the VHL gene change and whether you need regular screening.

People with VHL need regular health checks and scans. These can help find problems early, when treatment is often easier and more successful. These checks are not needed for people who have not inherited VHL.

I am worried about testing my child

It is normal to feel worried about genetic testing for your child. The Clinical Genetics team is here to support you and your family throughout the process.

Genetic testing can show whether a child has inherited the VHL gene change and whether they need regular screening. Screening can help find problems early, often before symptoms develop.

Because VHL screening starts from around 5 years of age, we usually offer predictive genetic testing by 4 years of age. Children who have inherited VHL are introduced to screening gradually as they get older.

If a child has not inherited the VHL gene change, they will not need VHL screening or follow-up appointments.

Before testing, you will have the opportunity to talk with a genetics healthcare professional and ask any questions you may have.

If I have not inherited VHL can I still get cancer?

Yes, there is still a chance of developing tumours or cancer, because cancer is common in the general population and cancer risk is affected by many different factors. Your chance of developing cancer would be similar to anyone else in the general population.

Do lifestyle and diet affect cancer risk?

There is currently no clear evidence that any specific diet or lifestyle change can prevent VHL-related tumours or cancers.

However, research in the general population shows that some healthy lifestyle choices can help reduce the overall risk of cancer. These include:



- Not smoking
- Limiting alcohol
- Being physically active
- Maintaining a healthy weight

Eating a healthy, balanced diet can also help you stay well and maintain a healthy weight. This includes:



- Plenty of fruit and vegetables
- Wholegrain foods such as wholemeal bread, brown rice and oats
- Beans, peas and lentils (pulses)

These healthy habits are recommended for everyone, whether or not they have VHL.

Are there any research studies ?

You may be able to take part in research studies if you would like to.Research studies may not help you directly, but they can help doctors learn more about VHL and improve care for people in the future.

Taking part in research is completely your choice. If you decide not to take part, this will not affect your NHS care in any way. **If you would like more information about research studies, please speak to your healthcare team.**

Where can I get further information/support ?

Your Clinical Genetics Team can provide additional information and support about VHL. Other sources of support include:

- **VHL UK / Ireland Charity -** support and information for people affected by VHL: www.vhl-uk-ireland.org
- **VHL Alliance (VHLA)** - information and support for individuals and families affected by VHL. Their website includes the Active Surveillance Guidelines, which explain recommended screening and monitoring for VHL-related conditions, as well as the VHL Handbook, information on treatment and care, and resources for children and young people: www.vhl.org
- International information, education and support for people and families affected by VHL: www.vhl.org
- **Genetic Alliance UK** - information and support for people and families affected by genetic conditions: www.geneticalliance.org.uk
- **Macmillan Cancer Support** - emotional support, practical advice, and reliable information about cancer, treatment, and living well: www.macmillan.org.uk
- **Cancer Research UK** – reliable information about cancer, treatment and screening: www.cancerresearchuk.org
- **Maggie's Centres** – emotional and practical support for people affected by cancer: www.maggies.org

Glossary

Plain-language meanings for words used in this leaflet.

Adrenal glands
Small glands above the kidneys that make hormones, including adrenaline and cortisol.

Cyst
A fluid-filled sac that can form in an organ or tissue.

Gene
A section of DNA that carries instructions for how the body grows and works.

Genetic change
A difference in a gene. Some genetic changes can affect health.

Haemangioblastoma
A usually slow-growing, non-cancerous tumour of blood vessels, often in the brain or spinal cord.

Inherited
Passed from a parent to a child through genes.

MRI scan
A scan that uses a strong magnetic field and radio waves to make detailed pictures of the body.

Pancreas
An organ behind the stomach that helps digest food and controls blood sugar.

Retina
The light-sensitive layer at the back of the eye that helps you see.

Screening
Regular checks to look for a health problem early, before symptoms appear.

Tumour
A growth of cells. Tumours can be non-cancerous or cancerous.

VHL
Short for von Hippel-Lindau disease, an inherited condition that can cause certain tumours and cysts.

About this leaflet

This leaflet provides general information only. Your care will be based on your own needs and the services available in your area. This leaflet does not replace guidance from your healthcare professional.

Need more help?

If you want more information or need help understanding this leaflet, please contact your healthcare team.

Your feedback

We would value your feedback on this leaflet, whether you are a patient, carer, family member or NHS clinician. Please scan the QR code or follow the link below to provide your feedback.

https://bit.ly/leaflet-feedback

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