NHS
East Genomics

Genetic Testing for Inherited Causes of Paraganglioma and Phaeochromocytoma*

Information for patients



*R223

Why am I being offered a genetic test?

You are being offered a test to look for inherited changes (described as variants) in a group of genes that may explain why you have developed a paraganglioma or phaeochromocytoma.

What are paragangliomas and phaeochromocytomas?

Paragangliomas are tumours (growths), that come from special nerve cells that make adrenaline and similar hormones. When these growths form in the adrenal glands, they are called phaeochromocytomas.

Paragangliomas can also develop in other parts of the body, like the abdomen, pelvis, chest, or head and neck. Tumours in the head and neck often don't cause symptoms, except for a small lump you can feel. However, they can cause problems if they slowly grow and press on nearby tissues.

Tumours in the abdomen, pelvis, or chest can sometimes release hormones like adrenaline. This can lead to symptoms similar to a panic attack, such as a fast heartbeat, headaches, anxiety, sweating, and high blood pressure. Most of these tumours are not cancerous, but around 1 in 10 may spread to other parts of the body.

Scan the code or follow the link to find out more:

www.nhs.uk/conditions/phaeochromocytoma

Are paragangliomas and phaeochromocytomas inherited?

Most paragangliomas and phaeochromocytomas happen by chance. But about one in three people with these tumours have an inherited condition that caused them.

Some of these conditions can also raise the risk of other health problems or tumours.

People with an inherited condition are usually offered extra health checks. This helps find any problems early, when they are easier to treat.

What are the benefits from having a genetic test?

Knowing if you have a change in a gene can:

- Help your doctors understand more about why you have a paraganglioma or phaeochromocytoma.
- Affect your future care, like decisions about surgery and whether you need extra health checks later on.
- Give useful information to your family as they may also have a chance of getting similar tumours. If relatives have the same gene change, they may be offered regular health checks to spot problems early, when they are easier to treat.

What is a gene?

Our genes are like a set of instructions inside our body that tell it how to grow, work, and look. Each gene gives instructions for one specific job, like what colour our eyes are or how our body breaks down food.

We have two copies of most of our genes. These come from our parents - one copy from our mother and one copy from our father. That's why we often look like our parents.

Sometimes, a gene can have a change (called a variant) that stops it working properly. If that change causes a health problem, it's called a pathogenic variant. Changes in some genes can increase the chance of developing paragangliomas or phaeochromocytomas.

What genes are tested and what do they mean?

This test looks at genes for conditions that can cause paraganglioma or phaeochromocytoma. The next page lists the different genes and conditions.

Your medical team might ask about your personal and family history to see if there are clues pointing to a genetic condition. This is because some genes can also increase the risk of other health problems or tumours. It is helpful to mention if any relatives have had paraganglioma or phaeochromocytoma, kidney or thyroid cancer, tumours in the parathyroid glands or pituitary (gland at the base of the brain), or other rare tumours.

Your medical team may also check your skin for brown patches or lumps. These can be signs of a different inherited condition that is not included in routine tests.

Gene(s)	Associated condition	What it means
FH	Hereditary Leiomyomatosis and Renal Cell Cancer (HLRCC)	Can cause small red skin lumps, fibroids (muscle growths in the womb) and kidney cancer.
MAX	Hereditary Paraganglioma-Phaeochromocytoma syndrome	Increases the chance of phaeochromocytoma.
MEN1	Multiple endocrine neoplasia type 1	Increases the chance of phaeochromocytoma, pituitary and pancreatic tumours, and hyperparathyroidism*.
RET	Multiple endocrine neoplasia type 2	Can cause thyroid cancer, phaeochromocytoma and hyperparathyroidism*.
SDHA	Hereditary Paraganglioma-Phaeochromocytoma syndrome	Can cause phaeochromocytomas or paragangliomas.

Gene(s)	Associated condition	What it means
SDHAF2	Hereditary Paraganglioma-Phaeochromocytoma syndrome	Risk mostly affects people who inherit a gene change from their father. Head and neck paraganglioma are more common.
SDHB	Hereditary Paraganglioma-Phaeochromocytoma syndrome	Increases the chance of phaeochromocytoma and also kidney cancer.
SDHC	Hereditary Paraganglioma-Phaeochromocytoma syndrome	Head and neck paraganglioma are common.
SDHD	Hereditary Paraganglioma-Phaeochromocytoma syndrome	Risk mostly affects people who inherit a gene change from their father. Head and neck paraganglioma are common.

Gene(s)	Associated condition	What it means
TMEM127	Hereditary Paraganglioma-Phaeochromocytoma syndrome	Increases the chance of phaeochromocytoma.
VHL	Von Hippel-Lindau disease	Can cause kidney cancer, benign tumours in the eye, brain or spinal cord, pancreatic tumours and phaeochromocytoma.

***Hyperparathyroidism** = overactivity of the parathyroid gland in the neck leading to high levels of calcium*

Do I have to have the test?

No. It's your choice.

The test might help guide your care and give useful information for relatives. If you decide not to have it, your doctors will still give you the best care they can.

You can choose to have the test later if you're not ready now.

If you decide to go ahead in the future, your hospital team can help arrange it, or your GP can refer you to the clinical genetics team.

What if I'm not sure about the test?

If you haven't already seen the Genetics team, you can ask to have an appointment with them.

They can help you think through the pros and cons of testing.

Will my information be kept private?

Yes. Your information will be kept safe and private. Only your care team and a few trained staff will see your details.

Everyone involved must follow strict rules to protect your privacy.

What will happen next if I say yes?

You'll be asked if you agree to the test. You'll also be asked about your choices for research.

A blood sample will be taken for the test.

What results might I get?

1 No gene change found

This is the most common result. It means we didn’t find a genetic reason for your paraganglioma or phaeochromocytoma. This can be reassuring, but sometimes there may still be a genetic cause that we don’t yet know about. If any relatives have also had related tumours you may be offered a clinic appointment with the Genetics team, if you haven't already seen them.

2 A gene change is found

This means there is a genetic reason for your paraganglioma or phaeochromocytoma. Your care team will use this information to help plan your treatment. Your family may also be offered testing to see if they have the same gene change. You will be offered an appointment with the Genetics team to talk more about what this means.

3 A gene change is found, but we’re not sure what it means

This is rare. Sometimes we find a change in a gene, but we don’t know yet if it causes paraganglioma or phaeochromocytoma. We call this a variant of unknown significance or VUS. You’ll be offered an appointment with the Genetics team to look into this more.

How will I get my results?

The team who arranged your test will give you the results, usually in about 3-4 months.

They will set up a time to talk with you about what the results mean.

What happens if a gene change is found?

If a gene change is found, you will be offered an appointment with the Clinical Genetics team.

They will explain more about the gene change and what it means for you.

This is a chance to talk about how the result may affect your health, if you need extra health care, and if your relatives can have genetic tests.

You can also ask any other questions at this appointment.

We understand that finding a gene change can feel worrying, especially if it's linked to other health problems or a higher chance of cancer.

But these results can help us plan the best care for you in the future.

Screening for people with an inherited cause for phaeochromocytoma and paraganglioma

People who have a gene change causing an increased chance of developing phaeochromocytoma or paraganglioma have an appointment each year with an endocrinologist (doctor who specialises in conditions that affect hormones).

Blood and urine tests can monitor hormone levels. MRI scans may also be suggested to look for tumours, and sometimes surgery is required to remove a growth.

The care arranged for you will depend on the gene change found and your own individual circumstances.

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

Produced by: NHS East Genomics, September 2025 (V1)
This information was correct when published
Lead authors: Dr Anna Wilsdon and Dr Rachel Harrison
Review: September 2029
www.eastgenomics.nhs.uk

NHS East Genomics