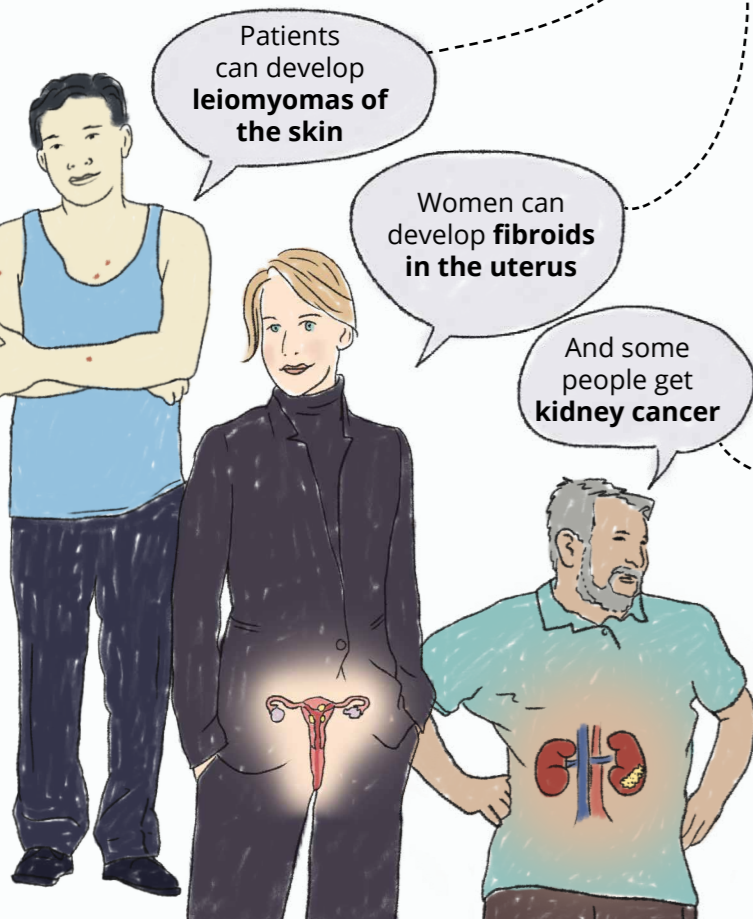


Information about Hereditary Leiomyomatosis and Renal Cell Carcinoma (HLRCC)

HLRCC is a **rare hereditary syndrome**. A syndrome is a combination of conditions that arise from one cause. HLRCC is caused by a **DNA abnormality** in the FH gene. In the Netherlands we know of about **160 families** that have HLRCC.

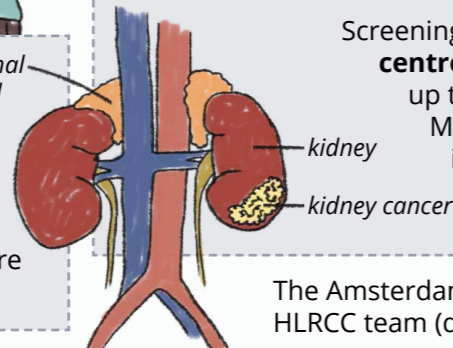
What complaints and symptoms might you experience?

HLRCC can manifest in different ways.



Paragangliomas

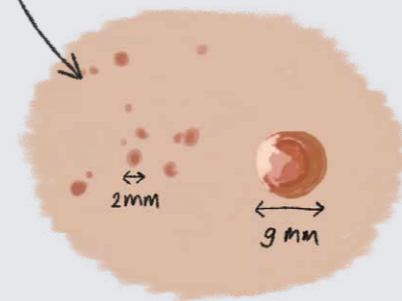
About 1% of people with HLRCC are diagnosed with **paragangliomas**. These are rare, usually benign tumours that are located close to nerves and blood vessels. They can occur in the adrenal glands (where they are called pheochromocytomas), among other places.



→ Leiomyomas of the skin

Our skin contains small muscle cells that raise the hairs when someone gets goosebumps. Leiomyomas are **non-cancerous tumours** in these muscle cells.

The leiomyomas can **differ in size**.



Leiomyomas occur generally between the ages of 20 and 40. They are skin-coloured or red or brown **bumps**, usually on the arms, legs or torso. Sometimes there are a few, sometimes more than 100. They can be **painful** when touched or exposed to cold.

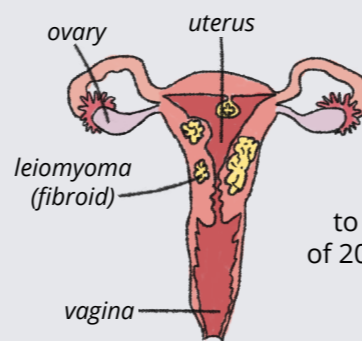
Treatment is **not necessary**. Only if the leiomyomas cause problems, treatment is considered. The risk of a malignant tumour of the skin (leiomyosarcoma) is very small.



→ Leiomyomas in the uterus (fibroids)

Most women with HLRCC develop **fibroids in the uterus**. There can be many and they may be large. Women often experience (severe) symptoms, such as heavy and painful periods and reduced fertility.

Many women need **surgery** to remove the myomas or the uterus (average age 30-35 years). Sometimes there are other treatment options, such as medication.



The risk of a malignant tumour of the uterus (leiomyosarcoma) is very small.

We refer women with HLRCC to a **gynaecologist** from the age of 20 onwards.



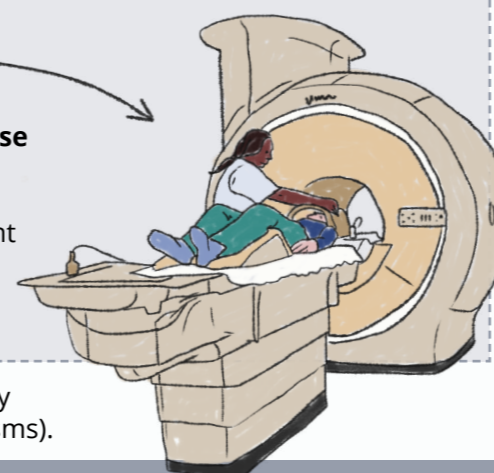
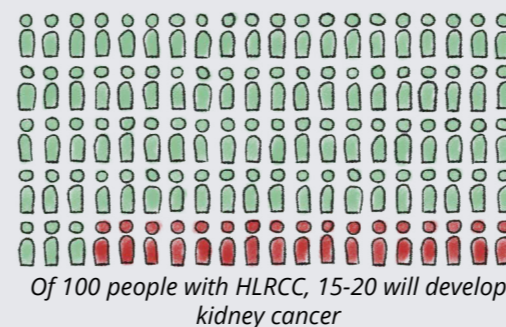
→ Kidney cancer

Some people with HLRCC develop **kidney cancer**. The risk of kidney cancer is approximately 15-20%. Usually it is a specific type of kidney cancer (FH-deficient renal cancer). Because HLRCC often involves fast-growing tumours, **surgery is usually performed immediately**.

Kidney cancer in HLRCC can develop at a young age. The advice is to have an **MRI scan** of the kidneys every year from the age of 8-10.

Screening in a (kidney cancer) **expertise centre** is necessary. For children up to the age of 18, the Princess Máxima Centre (PMC) in Utrecht is the expertise centre.

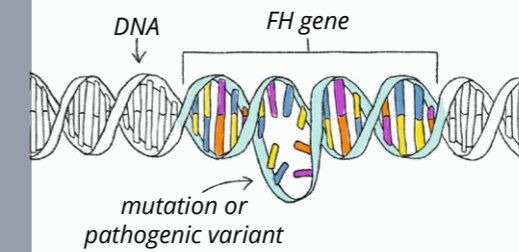
The Amsterdam UMC has a multidisciplinary HLRCC team (doctors from various specialisms).



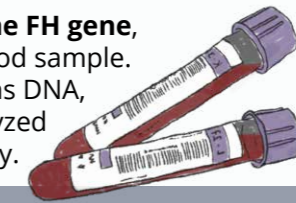
DNA, genes and mutations

A **gene** is a small piece of **DNA**. An abnormality in a gene is called a **mutation** or **pathogenic variant**.

People with a mutation in the **FH gene** have HLRCC.

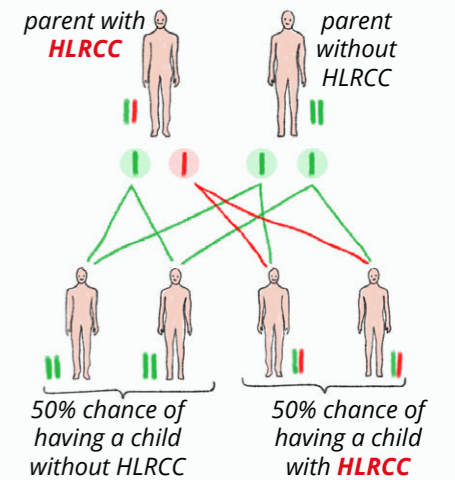


To **test for the FH gene**, we take a blood sample. Blood contains DNA, which is analyzed in a laboratory.



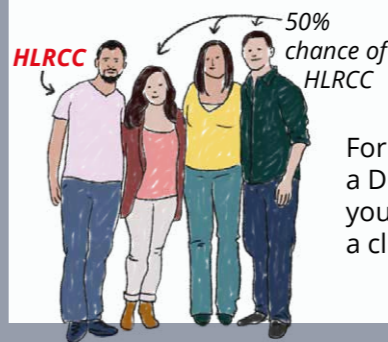
How is HLRCC inherited?

HLRCC is inherited in an **autosomal dominant** manner. Children of a parent with HLRCC each have a **50% (1 in 2) chance** of also having HLRCC. This applies to both sons and daughters.



What does this mean for family members?

Children and siblings of people with HLRCC have a **50% chance** of also having this predisposition.



For genetic advice and a DNA test, you can ask your GP to refer you to a clinical geneticist.

What are the options if you want to have children?

More information about hereditary conditions when **planning a family** can be found at www.erfelijkheid.nl/kinderwens/wat-als-je-een-erfelijke-ziekte-kunt-doorgeven.



For tailor-made information, you can make an appointment with a **clinical geneticist**.

Rare situation

Very occasionally, a child inherits an FH mutation from both parents. The child then has **two FH mutations**. This causes a very serious metabolic disorder in childhood (Fumarate Hydratase Deficiency, FHD).

More information

www.erfelijkheid.nl (Dutch)
www.hlrccinfo.org (English)

