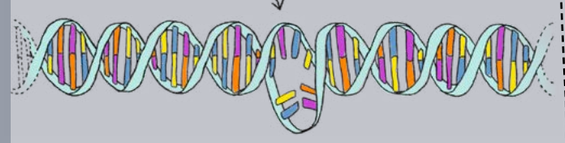
Information about Hereditary Leiomyomatosis and Renal Cell Carcinoma (HLRCC)

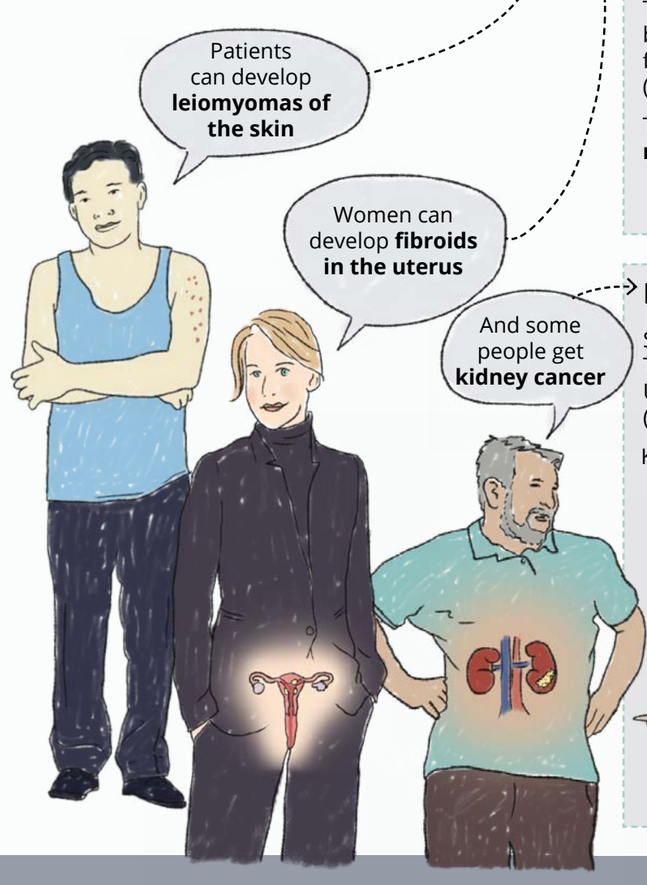
HLRCC is a **rare hereditary syndrome**.
A syndrome is a combination of conditions that arise from one cause. HLRCC is caused by a **DNA abnormality** in the FH gene.



In the Netherlands we know of about **100 families** that have HLRCC.

What complaints and symptoms might you experience?

HLRCC can manifest in different ways.

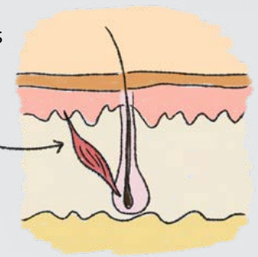


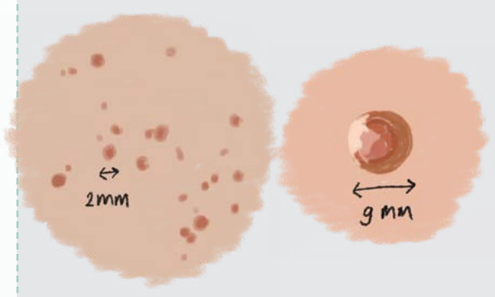
Leiomyomas of the skin

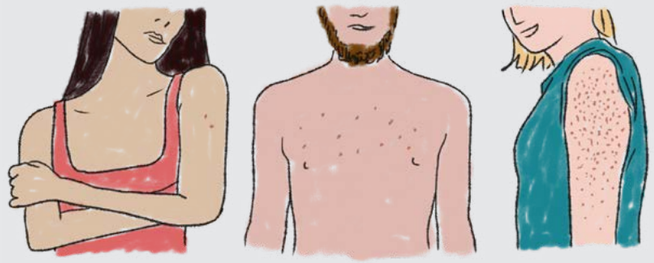
non-cancerous

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**.



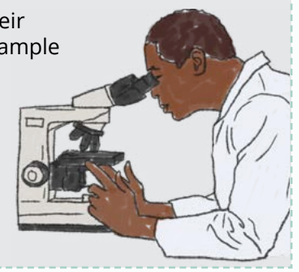




Leiomyomas occur generally between the ages of 20 and 40. The leiomyomas are skin-coloured or red **bumps**, usually on the arms, legs or torso. Sometimes there are a few, sometimes more than 100. They can be **painful** when touched or exposed to cold.

A doctor can diagnose them by their appearance and by examining a sample of tissue (biopsy).

Treatment is **not necessary**, however if the leiomyomas cause a lot of pain we would consider removing them. The risk of a malignant tumour of the skin (leiomyosarcoma) is very small.

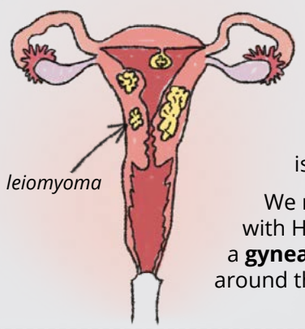


Leiomyomas in the uterus (fibroids)

non-cancerous

Most women with HLRCC develop **fibroids in the uterus**. There can be many and they may be large. The average age at which fibroids are diagnosed is 30 years (range 18-53 years).

The fibroids may lead to serious **menstrual disorders**.



Many women require **surgery** to remove fibroids or the uterus. Half of the women who undergo surgery are younger than 30 years old.

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

We refer women with HLRCC to a **gyneacologist** around the age of 20.

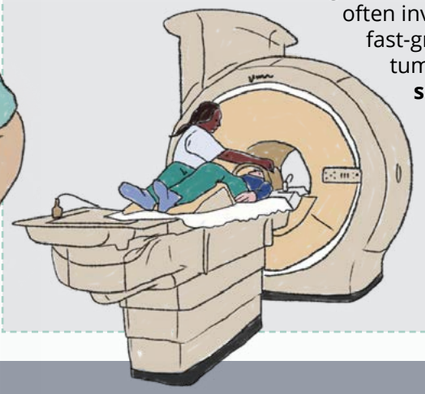
Kidney cancer

cancerous

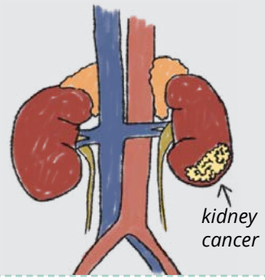
Some people with HLRCC develop **kidney cancer**. The risk of kidney cancer is approximately 15%. Usually it is a specific type of kidney tumour (type II papillary kidney cancer).

Kidney cancer in HLRCC can develop at a young age. Because HLRCC often involves fast-growing tumours, **surgery is usually performed immediately**.

The advice is to have an **MRI scan** of the kidneys every year from the age of 16.

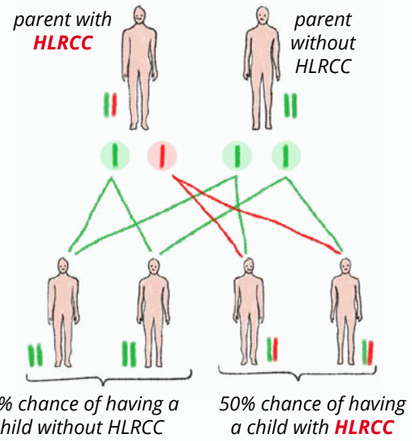


Of 100 people with HLRCC, 15 will develop kidney cancer



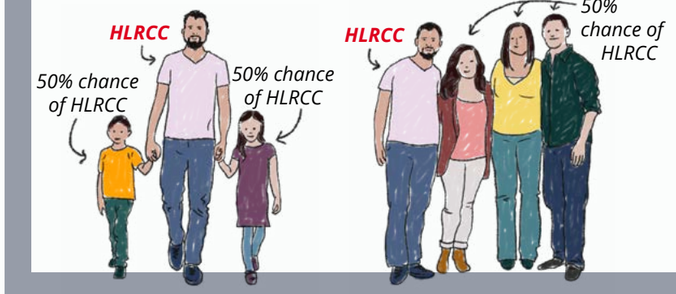
How is HLRCC inherited?

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**.

More information

For more information, please visit:
www.kanker.nl (Dutch)
www.erfelijkheid.nl (Dutch)
www.hlrccinfo.org (English)

Amsterdam UMC

Illustrations & design: Irene Cecile Educational Illustrations, www.irenececile.com.
Clinical geneticist: L.P. van Hest.