

HLRCC Handbook

Living with HLRCC: a plain-language guide for patients and families.

Educational, not medical advice. Talk with your care team about your own plan.

What is HLRCC?

HLRCC is short for **hereditary leiomyomatosis and renal cell cancer**, and HLRCC is the preferred name. It is sometimes referred to as FH tumor predisposition syndrome. ^[1 , 2]

Older names you may still see include Reed syndrome and multiple cutaneous and uterine leiomyomatosis (MCUL). ^[1 , 2]

- **Hereditary** means it runs in families.
- **Leiomyomatosis** means growths made of smooth muscle. They are not cancer.
- **Renal cell cancer** means kidney cancer.

HLRCC is caused by a change in a gene called **FH**. Genes are like instructions for your body. Everyone has two copies of the *FH* gene. People with HLRCC have one copy that does not work well. ^[1 , 2]

About 1 in 1,000 people may carry a harmful change in the *FH* gene (Shuch 2020). ^[9] In a study of more than 120,000 people who had genetic testing for inherited cancer risk, about 1 in 2,668 of those with no known signs of HLRCC had an HLRCC-related *FH* change. ^[10]

The most important thing to know: having HLRCC does **not** mean you have cancer or that you will get it. It means your risk of kidney cancer is higher than usual. Checking your kidneys every year helps doctors find a problem early. ^[1 , 2]

What can HLRCC cause?

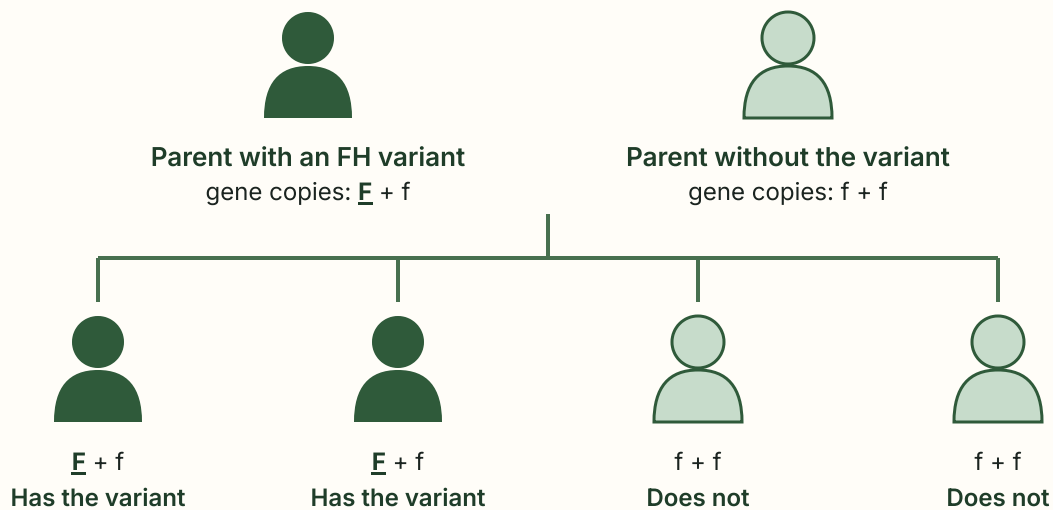
HLRCC is different for everyone, even in the same family. A person may have one, two, all three or none of these: ^[2]

Body part	What can happen	Is it cancer?
Skin	Small firm bumps called leiomyomas. They can hurt, or feel sore in the cold or when touched. ^[2]	No
Uterus (womb)	Fibroids. In HLRCC they are often larger, more numerous and appear at a younger age, around 30 on average. ^[1 , 2]	No
Kidneys	A higher chance of a kind of kidney cancer that can spread even when it is small. ^[1 , 2]	Yes, when it happens

Estimates vary widely. NCI's expert summary says most estimates are 15 to 30 in 100 over a lifetime ^[2]; a 2020 study using large population databases suggests the risk may be much lower ^[9]. Most people with HLRCC do not get kidney cancer.

How HLRCC is passed on

A parent with HLRCC has a **1 in 2 (50%) chance** of passing the *FH* change to each child. Boys and girls have the same chance. ^[1 , 2] Very rarely, a child inherits an *FH* change from both parents. This causes a serious metabolic condition called fumarate hydratase deficiency (also called fumarase deficiency), which starts in newborns and young babies. ^[1 , 2]



Each child has a 1 in 2 chance of getting the FH change. ^[1 , 2]

- If you did **not** get the family's *FH* change, you cannot pass it on to your children. ^[1 , 2]
- Sometimes a parent has the change but never has any signs, so it can look as if it "skipped" a generation. ^[1]
- No test can tell which signs a child with the change will have, or when. ^[1]

How do I know if I have it?

A **genetic test** on a blood or saliva sample looks for a change in the *FH* gene. ^[1 , 3]

- In most cases, if the test finds a **harmful change** in the *FH* gene, it confirms HLRCC. A few *FH* changes carry different risks, so your genetic counselor will explain what your result means. The report may call it "pathogenic" or "likely pathogenic." ^[1 , 2]
- A result called a **variant of uncertain significance (VUS)** is possible. A VUS does not confirm or rule out HLRCC. ^[1 , 39] Doctors follow your own and your family's health history instead. ^[39] Relatives are not offered predictive testing for a VUS, although a laboratory may ask some relatives to be tested to help clarify it. ^[36 , 39] A VUS can be updated later, so ask your genetic counselor to check again from time to time. ^[40] Once one person in a family has a known change, relatives can take a simpler test that looks only for that change. ^[1 , 2]

A **genetic counselor** can help you decide about testing and explain what results mean. ^[43] You can find one through the National Society of Genetic Counselors. ^[19]

What to expect

Your skin

Skin leiomyomas are firm bumps. They can be skin-colored, pink, light brown or reddish-brown. They are most often on the chest, back, arms and legs. They usually first appear between the teen years and the forties, and more can appear over time. ^[1 , 2]

- A doctor needs to take a tiny sample (a **biopsy**) to be sure a bump is a leiomyoma. ^[1]
- Bumps that cause no problems usually need no treatment. ^[2 , 20]
- Painful bumps can be removed or treated with laser or freezing, and some medicines can ease the pain. ^[1 , 2]
- See a skin doctor (dermatologist) every 1 to 2 years for a full skin check. ^[1] NCI notes there is no agreed schedule for skin exams. ^[2]

Fibroids (for women)

Fibroids are growths in the wall of the uterus. They are not cancer. Most women with HLRCC-related fibroids have heavy or irregular periods, and many have pelvic pain. ^[1 , 2 , 32]

- See a gynecologist who knows about HLRCC. NCI's summary suggests a yearly visit with scans from time to time ^[2]; GeneReviews suggests visits as needed for symptoms and family planning ^[1]. Ask your gynecologist how often is right for you.
- Ways to treat fibroids: medicine; a hormone IUD placed in the uterus; surgery to take out only the fibroids (**myomectomy**); surgery to take out the uterus (**hysterectomy**). ^[1 , 2]
- If you hope to have children, talk with your gynecologist early about timing. ^[1] NCCN's patient guideline also suggests considering kidney imaging before a planned pregnancy. ^[3]
- After any fibroid surgery, the tissue should be checked by a lab. ^[1]

Your kidneys

The kidney cancer linked to HLRCC is called **FH-deficient renal cell carcinoma**. It can grow and spread even when it is small. It often causes no symptoms at first. ^[1 , 2 , 11]

- **From age 8, a yearly kidney MRI, for life.** Your urologic oncologist or medical oncologist will decide the exact timing. ^[1 , 2]
- **An ultrasound alone is never enough** to check the kidneys in HLRCC. ^[2]
- If a scan finds a tumor, it should be removed by surgery **soon**, even if it is small. **Do not wait for it to reach 3 cm** (about 1 inch). That rule is used for some other conditions, but not for HLRCC. ^[1 , 2 , 3 , 5]
- Choose a kidney surgeon (urologic oncologist) who has experience with HLRCC. ^[1]
- Tell your doctor if you see blood in your urine, have pain in your lower back, or feel a lump in your belly. ^[1 , 2]

Rare: adrenal and nerve-tissue tumors

A few specific *FH* changes are linked to tumors called **pheochromocytoma** or **paraganglioma**. ^[1 , 20] These can cause high blood pressure, headaches, a racing heart and sweating. ^[44] If your family has one of these changes, your doctor may add yearly blood or urine tests and a scan every two years. ^[1 , 2 , 16 , 20] Most people with HLRCC do not need these extra tests. ^[2]

Your check-up calendar

Use this calendar with your care team. Your own plan may be different.

When	What	Who	Done?
Every year	A kidney MRI with 1 to 3 mm slices through the kidneys. ^[1 , 5] Your radiology team will decide whether contrast dye is needed. ^[20] If a spot needs watching, ask for the same type of scan each time. ^[2] CT if MRI is not possible. ^[3] From age 8, a yearly kidney MRI, for life. Your urologic oncologist or medical oncologist will decide the exact timing. ^[1 , 2]	Radiologist; HLRCC doctor reviews results	☐
Often yearly ^[2] (if you have a uterus); ask your gynecologist ^[1]	Gynecology visit; ultrasound or MRI if advised. ^[1 , 2]	Gynecologist	☐
Every 1–2 years	Full skin check. ^[1] NCI notes there is no agreed schedule for skin exams. ^[2]	Dermatologist	☐
Only for certain FH changes	Check blood pressure at every visit. Have a blood or urine test each year. From age 10, have a body MRI every 2 years. ^[1 , 20]	Your HLRCC doctor	☐
After diagnosis, and before planning a family	Genetic counseling; tell close relatives. ^[1 , 2]	Genetic counselor	☐

My scan record

Year	Scan type and date	Result	Next scan due

Tips for your MRI

- If a spot is being watched, ask for the **same kind of scan** each time, so results can be compared. ^[2]
- If small spaces worry you, tell the team before your visit. Ask if you can go in feet first.
- Bring a copy of your past scan reports, or ask that they be sent ahead.

Questions to ask your doctor

About my diagnosis

- ☐ What is my exact *FH* change? Can I have a copy of my genetic test report?
- ☐ Do I need to see a genetic counselor?
- ☐ Is my *FH* change one of the few linked to pheochromocytoma or paraganglioma?

About my kidney scans

- ☐ When is my next kidney MRI? Who will read it?
- ☐ Does the radiologist know about HLRCC?
- ☐ Will you use the same kind of scan each year?
- ☐ What happens if the scan finds a cyst or a spot?

If a kidney tumor is found

- ☐ Is the surgeon experienced with HLRCC?
- ☐ Will the tumor be removed soon, with a wide margin?
- ☐ Is [a clinical trial](#) right for me?

About fibroids and skin

- ☐ How often should I see a gynecologist and a dermatologist?
- ☐ What are my treatment choices if I want to have children?
- ☐ What can I do about painful skin bumps?

About my family

- ☐ Who in my family should be tested?
- ☐ At what age should my children be tested and start scans?

Genetic testing, insurance and feelings

Insurance (United States)

A U.S. law called **GINA** helps keep health insurers and many employers from treating you unfairly because of a gene test. ^[18]

- Health insurers cannot use your genetic test results to deny coverage or set your price. ^[18]
- Employers with 15 or more workers cannot use them to hire, fire or promote. ^[18]
- GINA does **not** cover life, disability or long-term care insurance. ^[18] Ask a genetic counselor about this.

Feelings

Learning you have HLRCC can be scary. Many people feel worried, angry, guilty about passing it on, or relieved to finally have an answer. All of these feelings are normal.

- Talk with people you trust.
- Ask your care team about a counselor or social worker.
- Connect with other families through the HLRCC Foundation.
- Keep a simple record of your scans and results. It helps you feel in control.

Support and resources

- **HLRCC Foundation:** hlrcc.org. [Questions and answers](#), genetic testing information, videos and [a map of HLRCC specialists](#).
- **Find a genetic counselor:** findageneticcounselor.nsgc.org ^[19]
- **Research studies:** the U.S. National Cancer Institute runs [a long-term HLRCC study](#) (NCT00050752). Other studies were open or planned too: NCT04981509 (open at some places only) and NCT07716735. ^[17, 23] Studies open and close often. This list was checked on October 30, 2026, so **check ClinicalTrials.gov** for "hereditary leiomyomatosis" before you act.
- **Easy-to-read pages:** the National Cancer Institute's page on kidney cancer that runs in families ^[24], and the NIH GARD page on HLRCC. ^[25]
- **For your doctor:** GeneReviews ^[1] and the NCI summary on HLRCC ^[2].
- **Support the Foundation:** hlrcc.org/donate-now/

Words to know

Biopsy

Taking a tiny piece of tissue to look at under a microscope.

Gene change (variant)

A difference in a gene. A harmful change can raise the risk of disease.

Genetic counselor

An expert who helps you understand genetic tests and family risk.

MRI

A scan that uses magnets, not X-rays, to take detailed pictures inside the body.

Surveillance

Regular check-ups and scans to catch problems early.

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Share with your family

Tear out or copy this page to give to a relative.

Someone in your family has HLRCC. HLRCC is a condition that runs in families. It is caused by a change in a gene called *FH*. It can cause skin bumps, fibroids in the uterus, and a higher chance of kidney cancer. ^[1 , 2]

Why this matters to you: you may have the same gene change. A simple test can tell you. If you have it, a kidney MRI every year can help find problems early. ^[1 , 2 , 3]

Who may be at risk?

- **Children** of a person with HLRCC each have a **1 in 2 (50%)** chance of having the same change. **Brothers, sisters and parents** may also have it. ^[1 , 2]
- **Aunts, uncles, nieces, nephews and grandparents** may also be at risk. Their chance depends on how they are related. ^[1]
- If your parent tested **negative** for the family change, you did not get it from that parent. ^[1 , 2]

What should I do?

1. **Ask your relative for a copy of their genetic test report.** It names the exact *FH* change. This makes your test simpler. ^[1 , 2]
2. **See a genetic counselor** or your doctor. Find one at findageneticcounselor.nsgc.org. ^[19]
3. **Decide about testing.** You can test for the one family change. Testing is your choice. ^[1 , 2]
4. **Children:** From age 8, a yearly kidney MRI, for life. Your urologic oncologist or medical oncologist will decide the exact timing. ^[1 , 2] Talk with a genetic counselor about testing. ^[5]

If you test positive

You will be offered a kidney MRI every year, skin checks, and gynecology visits if you have a uterus. ^[1 , 2]

If you test negative for the family change

You do not need HLRCC check-ups, and you cannot pass this change to your children. ^[1 , 2]

Learn more: hlrcc.org · Educational, not medical advice ·

Support the Foundation

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