

HLRCC QUICK FACTS

- HLRCC stands for Hereditary Leiomyomatosis and Renal Cell Cancer (sometimes Carcinoma). It is also known as Reed's Syndrome.
- HLRCC is caused by an inherited genetic alteration in the Fumarate Hydratase (FH) gene. There is a 50% risk of passing this on and the severity of the disease can vary a lot from person to person. It can be diagnosed by the detection of this genetic alteration (mutation).
- Many women with HLRCC develop large uterine fibroids in their twenties. Although benign, the fibroids may result in early treatment.
- Both men and women tend to develop benign skin leiomyomas (or "skin bumps") in their twenties. These two symptoms together, fibroids and skin leiomyomas, offer an important clue to the need for genetic testing of the FH gene. Because of the absence of uterine fibroids, HLRCC is more likely to go undetected in men, and early diagnosis is less likely.
- Screening requires an annual MRI scan of the abdomen using pre- and post-contrast 3D acquisition with 3mm slice reconstruction for the detection of kidney cancer. Be sure to read the Handbook section "Suggested Screening Guidelines" for more information.
- Even small HLRCC kidney tumors can metastasize, or spread, very quickly to the bones, lungs and brain. Unlike some other cancers, there is no curative treatment for kidney cancer once it metastasizes, although life may be extended with the latest class of drugs. Annual screening helps our goal to prevent metastasis and to stay healthy.
- If you live in the United States, you may want to consider being part of a clinical trial at the National Institutes of Health (US-NIH) in Bethesda, MD. This is the only trial open at the time of writing. We expect that more will be opened in the future, possibly at other locations around the world.



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- It is recommended that children who have a parent with HLRCC have genetic testing by age 8 as screening is recommended from that age for children at risk. Please read more about this in the Handbook.
- It is a newly identified condition (2002) and is currently being studied at several locations around the world.
- There are approximately 200-300 families currently diagnosed with HLRCC with perhaps 1500 patients. It is an under-diagnosed condition because of its rarity.
- Being diagnosed with HLRCC can be a very scary thing, mostly because it has the word cancer in its heading. If you have HLRCC it does not mean that you have kidney cancer or will necessarily get it. However it does mean that you have an increased risk factor for kidney cancer, and you need to screen yourself so that doctors can detect even the smallest HLRCC cancer in your kidneys.
- Our motto is *Knowledge Saves*. Although it is sometimes difficult to come to terms with a new diagnosis, we provide you and your family with information to protect yourselves and future generations. Knowledge truly is a gift.
- We encourage you to look through our website and the Handbook. We also encourage you to reach out to us and ask for support – or offer it to another member. We are a small group, but we are very active, even on Facebook! We hope you will join us!
- <http://www.hlrccinfo.org> This has all the links under “Join Us”
- <http://vhl.inspire.com>
- <https://www.rareconnect.org>
- The HLRCC Family Alliance 1-800-767-4845 extension 709
- HLRCC Family Alliance c/o VHLA, 2001 Beacon Street, Suite 208, Boston, MA 02135 USA.

Enjoy this beautiful day!



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