Hereditary Leiomyomatosis and Renal Cell Cancer [1]

What is hereditary leiomyomatosis and renal cell cancer?

Hereditary leiomyomatosis and renal cell cancer (HLRCC) is a hereditary condition associated with multiple leiomyomas, which are fibroid skin tumors, uterine fibroids, which are non-cancerous growths in a woman's uterus, and type 2 papillary renal (kidney) cancer [2]. A person with HLRCC can have a few skin tumors or many skin tumors. These skin tumors generally develop in adulthood and occur on the chest, back, arms, and legs; the tumors can be painful, but they are not cancerous. Women with HLRCC can develop uterine fibroids as young as their teens or early 20s.

What causes HLRCC?

HLRCC is a genetic condition. This means that the risk of cancer and other features of HLRCC can be passed from generation to generation in a family. A specific gene called the fumarate hydratase (*FH*) gene is believed to cause most cases of HLRCC. Research is ongoing to learn more about this condition.

How is HLRCC inherited?

Normally, every cell has two copies of each gene: one inherited from the mother and one inherited from the father. HLRCC follows an autosomal dominant inheritance pattern, in which a mutation (alteration) happens in only one copy of the gene. This means that a parent with a gene mutation may pass along a copy of their normal gene or a copy of the gene with the mutation. Therefore, a child who has a parent with a mutation has a 50% chance of inheriting that mutation. A brother, sister, or parent of a person who has a mutation also has a 50% chance of having the same mutation.

Options exist for couples interested in having a child when they know that one of them carries a gene mutation that increases the risk for this hereditary cancer syndrome. Preimplantation genetic diagnosis (PGD) is a medical procedure done in conjunction with in-vitro fertilization (IVF). It allows people who carry a specific known genetic mutation to have children who do not carry the mutation. A woman's eggs are removed and fertilized in a laboratory. When the embryos reach a certain size, one cell is removed and is tested for the hereditary condition in question. The parents can then choose to transfer embryos that do not have the mutation. PGD has been in use for over a decade, and more recently has been used for several hereditary cancer predisposition syndromes. However, this is a complex procedure with financial, physical, and emotional factors for couples to consider before starting. For more information, talk with an assisted reproduction specialist at a fertility clinic.

How common is HLRCC?

HLRCC is considered to be rare. The number of people and families who have this condition is unknown.

How is HLRCC diagnosed?

HLRCC is suspected when a person has a history of multiple leiomyomas. A family history of fibroid tumors and type 2 papillary renal cancer also raises the possibility of HLRCC. Genetic testing [3] to look for mutations in the *FH* gene is available for people suspected of having HLRCC.

What are the estimated cancer risks associated with HLRCC?

HLRCC is associated with an increased risk of type 2 papillary renal cell carcinoma. For people with HLRCC, the estimated risk for this type of kidney cancer is about 15%.

What are the screening options for HLRCC?

There are no specific screening guidelines for HLRCC. The most common screening options for those at a higher than average risk of HLRCC are regular skin examinations and an abdominal/pelvic computed tomography (CT) scan [4], which is a three-dimensional picture of the inside of the body using an x-ray machine, with contrast, meaning a special dye, or a magnetic resonance imaging (MRI) [5] every two years. An MRI uses magnetic fields, not x-rays, to produce detailed images of the body. If a suspicious kidney lesion is found, a CT scan with and without contrast, renal ultrasound examination, and/or an integrated positron emission tomography-computed tomography scan (PET-CT) [6], which is a way to create pictures of organs and tissues inside the body using a small amount of a radioactive substance, may also be done to learn more about the suspicious area. Also, women may be recommended to have regular gynecological examinations and imaging studies, such as an ultrasound [7], to look for uterine fibroids.

Screening options may change over time as new technologies are developed and more is learned about HLRCC. It is important to talk with your doctor about appropriate screening tests.

Learn more about what to expect when having common tests, procedures, and scans [8].

Questions to ask the doctor

If you are concerned about your risk for kidney cancer, talk with your doctor. Consider asking the following questions of your doctor:

- What is my risk of developing kidney cancer?
- What can I do to reduce my risk of cancer?
- What are my options for cancer screening?

If you are concerned about your family history and think your family may have HLRCC, consider asking the following questions:

- Does my family history increase my risk of developing kidney cancer?
- Are the skin changes in me or my family considered leiomyomas?
- Does it suggest the need for a cancer risk assessment?
- Will you refer me to a genetic counselor or other genetics specialist?
- Should I consider genetic testing [3]?

More Information

The Genetics of Cancer [9]

Genetic Testing [3]

What to Expect When You Meet With a Genetic Counselor [10]

Collecting Your Family Cancer History [11]

Sharing Genetic Test Results with Your Family [12]

Additional Resources

Hereditary Leiomyomatosis and Renal Cell Cancer Family Alliance
www.vhl.org/hlrcc [13]

National Cancer Institute
www.cancer.gov [14]

American Cancer Society
www.cancer.org [15]

Cancer*Care*
www.cancercare.org [16]

To find a genetic counselor in your area, ask your doctor or visit the following websites:

National Society of Genetic Counselors
www.nsgc.org [17]

National Cancer Institute: Cancer Genetics Services Directory
www.cancer.gov/cancertopics/genetics/directory [18]

Links
[1] http://www.cancer.net/cancer-types/hereditary-leiomyomatosis-and-renal-cell-cancer
[2] http://www.cancer.net/node/31256
[3] http://www.cancer.net/node/24895
[4] http://www.cancer.net/node/24486
[5] http://www.cancer.net/node/24578
[6] http://www.cancer.net/node/24565
[7] http://www.cancer.net/node/24714
[8] http://www.cancer.net/navigating-cancer-care/diagnosing-cancer/tests-and-procedures
[9] http://www.cancer.net/node/24897
[10] http://www.cancer.net/node/24907
[11] http://www.cancer.net/node/30761
[12] http://www.cancer.net/node/24906
[13] http://www.vhl.org/hlrcc
[14] http://www.cancer.gov/
[15] http://www.cancer.org/
[16] http://www.cancercare.org/
[17] http://www.nsgc.org/
[18] http://www.cancer.gov/cancertopics/genetics/directory