Familial Non-VHL Clear Cell Renal Cell Carcinoma [1]

What is familial non-VHL clear cell renal cell carcinoma?

Familial non-VHL clear cell renal cell carcinoma (CCRCC) is a hereditary condition that increases the risk of the clear cell type of renal cell carcinoma (kidney cancer [2]). Currently, no other types of cancer or non-cancerous health problems are known to be associated with familial non-VHL CCRCC. The name separates this condition from von Hippel-Lindau syndrome (VHL), [3] which is the most common cause of hereditary risk for clear cell renal cell carcinoma.

What causes familial non-VHL CCRCC?

Familial non-VHL CCRCC is a genetic condition. This means that the risk of clear cell renal cell carcinoma can be passed from generation to generation in a family. A specific gene causing familial non-VHL CCRCC has not yet been discovered. Some families who appear to have familial non-VHL CCRCC have a translocation (rearrangement) involving chromosome 3. A translocation occurs when pieces of two or more chromosomes break off and reattach on another chromosome. Chromosome translocations can be passed down from generation to generation in a family. Research is ongoing to learn more about familial non-VHL CCRCC.

How is familial non-VHL CCRCC inherited?

Normally, every cell has two copies of each gene: one inherited from the mother and one inherited from the father. Although a specific gene has not been discovered, familial non-VHL CCRCC appears to follow an autosomal dominant inheritance pattern, in which a mutation 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 which 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. For more information, talk with an assisted reproduction specialist at a fertility clinic.

How common is familial non-VHL CCRCC?

Familial non-VHL CCRCC is considered to be very rare. The number of people and families who have familial non-VHL CCRCC is unknown.

How is familial non-VHL CCRCC diagnosed?

Familial non-VHL CCRCC is suspected when multiple family members have clear cell renal cell carcinoma, but no other symptoms of VHL [3].

What are the estimated cancer risks associated with familial non-VHL CCRCC?

The specific risk of clear cell renal cell cancer in families with familial non-VHL CCRCC is unknown.

What are the screening options for familial non-VHL CCRCC?

There are no specific screening guidelines for families suspected of having familial non-VHL CCRCC. Individuals in these families are encouraged to talk with their doctor about screening options for kidney cancer [2], including an ultrasound [4], which uses sound waves to create a picture of the internal organs, computed tomography (CT or CAT) scan [5], which creates a three-dimensional picture of the inside of the body with an x-ray machine. A computer then combines these images into a detailed, cross-sectional view that shows any abnormalities or tumors, and magnetic resonance imaging (MRI) [6], which uses magnetic fields, not x-rays, to produce detailed images of the body.

Screening options may change over time as new technologies are developed and more is learned about non-VHL CCRCC. 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 [7].

Questions to ask the doctor

If you are concerned about your risk for kidney cancer [2], 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 non-VHL CCRCC, consider asking the following questions:

- Does my family history increase my risk of developing kidney cancer?
- Should I meet with a genetic counselor?
- Should I consider genetic testing [8]?

Additional resources

Guide to Kidney Cancer [2]

What to Expect When You Meet With a Genetic Counselor [9]

National Cancer Institute

www.cancer.gov [10]

American Cancer Society
www.cancer.org [11]

Cancer*Care*
www.cancercare.org [12]

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

National Society of Genetic Counselors
www.nsgc.org [13]

National Cancer Institute: Cancer Genetics Services Directory
www.cancer.gov/cancertopics/genetics/directory [14]

Links:
[1] http://www.cancer.net/cancer-types/familial-non-vhl-clear-cell-renal-cell-carcinoma
[2] http://www.cancer.net/node/31256
[3] http://www.cancer.net/node/19322
[4] http://www.cancer.net/node/24714
[5] http://www.cancer.net/node/24486
[6] http://www.cancer.net/node/24578
[7] http://www.cancer.net/node/24959
[8] http://www.cancer.net/node/24895
[9] http://www.cancer.net/node/24907
[10] http://www.cancer.gov/
[11] http://www.cancer.org/
[12] http://www.cancercare.org/
[13] http://www.nsgc.org/
[14] http://www.cancer.gov/cancertopics/genetics/directory