Hereditary Papillary Renal Cell Carcinoma [1]

What is hereditary papillary renal cell carcinoma?

Hereditary papillary renal cell carcinoma (HPRCC) is a hereditary condition that increases the risk of the papillary type of renal cell carcinoma (kidney cancer [2]). There are two types of papillary renal cell tumors: type 1 and type 2. The tumors in HPRCC are type 1 tumors. Individuals with HPRCC have an increased risk of multiple kidney tumors and an increased risk of developing tumors on both kidneys. Currently, no other types of cancer or noncancerous health problems are known to be related to HPRCC.

What causes HPRCC?

HPRCC is a genetic condition. This means that the risk for type 1 papillary renal cell carcinoma can be passed from generation to generation in a family. Mutations (alteration) in a gene called *c-met* are linked to HPRCC development. *C-met* is a gene that encodes for the receptor to hepatocyte growth factor. Research is ongoing to learn more about HPRCC.

How is HPRCC inherited?

Normally, every cell has two copies of each gene: one inherited from the mother and one inherited from the father. HPRCC follows 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 HPRCC?

HPRCC is considered to be rare. The number of people and families who have HPRCC is unknown.

How is HPRCC diagnosed?

HPRCC is suspected when multiple family members have type 1 papillary renal cell carcinoma. Genetic testing [3] to look for mutations in the *c-met* gene is available for people suspected of having HPRCC.

What are the estimated cancer risks associated with HPRCC?

The specific risk for type 1 papillary renal cell carcinoma in families with HPRCC is unknown. Studies of families with HPRCC show that not everyone who has inherited the condition will develop kidney cancer [2]. If kidney cancer is diagnosed, talk with your doctor about treatment options. This is an area of active research. One drug, called foretinib, has shown evidence as being effective, and additional testing of this and other medications is ongoing.

What are the screening options for HPRCC?

There are no specific screening guidelines for families suspected of having HPRCC. Individuals in these families are encouraged to talk with their doctor about screening options for kidney cancer [2], including:

- 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). Some doctors suggest that individuals who have HPRCC, or a family history that suggests HPRCC, should have yearly screenings beginning at age 30.

Screening options may change over time as new technologies are developed and more is learned about HPRCC. 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 [2]?
- 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 HPRCC, 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 [3]?

Additional resources
[8]

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/hereditary-papillary-renal-cell-carcinoma
[2] http://www.cancer.net/node/18969
[3] http://www.cancer.net/node/24895
[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/patient/Cancer+Types/Kidney+Cancer
[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/search/geneticsservices