

Hereditary Pancreatitis [1]

What is hereditary pancreatitis?

Hereditary pancreatitis (HP) is a condition associated with recurrent pancreatitis, which is inflammation of the pancreas, and an increased risk of [pancreatic cancer](#) [2]. Cancer begins when normal cells begin to change and grow uncontrollably, forming a mass called a tumor. A tumor can be benign, meaning noncancerous, or malignant, meaning cancerous. When a tumor is malignant, it can spread to other parts of the body.

In people with HP, the first episode of pancreatitis usually occurs in childhood. However, the age when symptoms start and the severity can vary widely among people with HP, even within the same family.

What causes HP?

HP is a genetic condition. This means that the risk of pancreatitis and pancreatic cancer can be passed from generation to generation in a family. The gene associated with HP is called *PRSS1*. A mutation (alteration) in the *PRSS1* gene gives a person an increased risk of pancreatitis and pancreatic cancer. Mutations in two other genes, called *SPINK1* and *CFTR*, have also been linked to HP; however, it is unknown if mutations in these genes cause an increased risk of pancreatic cancer. Researchers believe that other genes may be associated with HP, and studies are ongoing to learn more about this condition.

How is HP inherited?

Normally, every cell has two copies of each gene: one inherited from the mother and one inherited from the father. HP follows an autosomal dominant inheritance pattern, in which a mutation needs to happen in only one copy of the gene for the person to have an increased risk of getting that disease. 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. 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 HP?

The specific incidence of HP is unknown. HP is estimated to account for about 3% to 6% of all pancreatitis cases.

How is HP diagnosed?

The diagnosis of HP is considered when two or more close family members, meaning parents, siblings, or children, in at least two generations have recurrent pancreatitis. Genetic testing is available for mutations in the *PRSS1*, *SPINK1*, and *CFTR* genes for people who may have HP.

What are the estimated cancer risks associated with HP?

The risk of pancreatic cancer [2] in people with HP has been estimated to be up to 40%.

What are the screening options for HP?

Screening for pancreatic cancer is suggested for people known to have HP. However, the effectiveness of current screening techniques for the early diagnosis of pancreatic cancer is not proven. Available screening options include:

- Magnetic resonance imaging (MRI) [3] or computed tomography (CT or CAT) [4] scan of the pancreas
- Endoscopic ultrasound [5], which is a thin, lighted tube guided into the esophagus through the mouth, and a transducer sends out sound waves that can identify if a tumor is present.
- Endoscopic retrograde cholangiopancreatography (ERCP). An endoscope is passed into the small intestine through the mouth and stomach. A catheter (smaller tube) is passed through the endoscope and into the bile ducts and pancreatic ducts to look for cancer.
- Blood test for CA19-9 level. This test aims to detect a protein found to be elevated in the blood of some people with pancreatic cancer.

Screening options may change over time as new technologies are developed and more is learned about HP. 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 [6].

Questions to ask the doctor

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

- What is my risk of developing pancreatic 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 you or other family members may have HP, consider asking the following questions:

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

More Information

[The Genetics of Cancer](#) [7]

[Genetic Testing](#) [8]

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

[Collecting Your Family Cancer History](#) [10]

[Sharing Genetic Test Results with Your Family](#) [11]

Additional Resources

Pancreatic Cancer Action Network

www.pancan.org [12]

National Cancer Institute

www.cancer.gov [13]

American Cancer Society

www.cancer.org [14]

CancerCare

www.cancer.org [15]

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

National Society of Genetic Counselors

www.nsgc.org [16]

National Cancer Institute: Cancer Genetics Services Directory

www.cancer.gov/cancertopics/genetics/directory [17]

Links:

- [1] <http://www.cancer.net/cancer-types/hereditary-pancreatitis>
- [2] <http://www.cancer.net/node/31388>
- [3] <http://www.cancer.net/node/24578>
- [4] <http://www.cancer.net/node/24486>
- [5] <http://www.cancer.net/node/24714>
- [6] <http://www.cancer.net/node/24959>
- [7] <http://www.cancer.net/node/24897>
- [8] <http://www.cancer.net/node/24895>
- [9] <http://www.cancer.net/node/24907>
- [10] <http://www.cancer.net/node/30761>
- [11] <http://www.cancer.net/node/24906>
- [12] <http://www.pancan.org/>
- [13] <http://www.cancer.gov/>
- [14] <http://www.cancer.org/>
- [15] <http://www.cancercare.org/>
- [16] <http://www.nsgc.org/>
- [17] <http://www.cancer.gov/cancertopics/genetics/directory>