Yale NewHaven Health
Smilow Cancer Hospital

Yale CANCER CENTER
A Comprehensive Cancer Center Designated by the National Cancer Institute

Hereditary Pancreatic Cancer
Information for patients

In recent years, our knowledge about hereditary pancreatic cancer and genetic testing has increased. While most pancreatic cancer is not hereditary, an estimated 10 percent of pancreatic cancer is due to a hereditary cause.

Hereditary pancreatic cancer is divided into several categories:

- Known hereditary cancer syndromes caused by a gene(s) that increases the risk for pancreatic and other cancers
- Known hereditary pancreatitis syndromes causes by a gene(s) that leads to an increased risk for pancreatic cancer
    - Pancreatitis is a disease that causes the pancreas to become inflamed
- Clustering of two or more relatives with pancreatic cancer in a family, but the specific cause of cancer risk in these families is currently unknown

Finding a hereditary explanation for why some people developed pancreatic cancer can help to:

- Provide a reason why they or their relatives were diagnosed with cancer
- Guide the course of cancer treatment or decision about surgery
- Clarify the risks for other cancers

Genetic testing is one way to understand if a person has a hereditary risk for pancreatic cancer. If a person has genetic testing and finds that he or she has hereditary pancreatic cancer, then it means he or she was born with an increased risk to develop pancreatic and possibly other cancers. Genetic testing for hereditary pancreatic cancer can look for several hereditary cancer syndromes (see next page).

In addition, results of genetic testing is important information to share with relatives because they may have also inherited the same increased risk to develop cancer. When a hereditary explanation is found in a family, relatives can then better understand their risk to develop cancer, which can help guide their decisions about cancer screening, prevention, and management.

Currently, screening for pancreatic cancer is not part of routine medical care. Instead, it is often performed as part of a research study because we do not yet understand the best way to screen for pancreatic cancer. However, there is some early evidence that pancreatic cancer screening can be of value and considered for people who have a high risk to develop pancreatic cancer. Our Smilow experts can discuss the potential benefits, risks, and limitations of pancreatic cancer screening, as well as research studies for people at increased risk for pancreatic cancer.

Hereditary Cancer Syndromes	Associated Gene(s)	Lifetime Risk of Pancreatic Cancer	Other Cancers at Increased Risks
Hereditary Breast and Ovarian Cancer Syndrome (HBOC)	*BRCA1* *BRCA2*	4-8%	Breast cancer Ovarian cancer Prostate cancer Melanoma
Familial Atypical Multiple Mole Melanoma Syndrome (FAMMM)	*CDKN2A*	10-19%	Melanoma
Familial Adenomatous Polyposis Syndrome (FAP)	*APC*	2-4%	Multiple precancerous colon polyps (totaling 20 or more) Colorectal cancer Gastrointestinal cancers
Lynch Syndrome (Hereditary Non-Polyposis Colorectal Cancer or HNPCC)	*EPCAM* *MLH1* *MSH2* *MSH6* *PMS2*	3-4%	Colorectal cancer Uterine and ovarian cancer Gastrointestinal cancers Urinary tract cancer Sebaceous cancer
Peutz-Jeghers Syndrome (PJS)	*STK11*	11-36%	Gastrointestinal tract polyps Gastrointestinal cancers Colorectal cancer Breast cancer Gynecological cancers
Other syndromes	*ATM* *PALB2*	Increased, but not well defined	Female breast cancer

Hereditary Pancreatitis and Pancreatic Cancer Syndromes	Associated Gene(s)	Lifetime Risk of Pancreatic Cancer	Other Increased Risks
Hereditary Pancreatitis (HP) Genes:	*PRSS1* *SPINK1*	25-40%	Multiple events of severe pancreatitis
Cystic Fibrosis (CF)	*CFTR*	Not as well defined	Chronic lung and pancreatic disease