Pancreatic Cancer Genetic Epidemiology (PACGENE)

By Heidi Rothenmund

In 2002, the National Cancer Institute in the United States provided funding for pancreatic cancer collaboration called PACGENE. This collaboration includes 7 centres involved with pancreatic cancer research, including our Pancreas Cancer Registry at Mount Sinai Hospital in Toronto. The goal is to understand what causes pancreatic cancer, with particular interest in identifying the gene(s) that cause hereditary pancreatic cancer. It is estimated that approximately 5-10% of patients with pancreatic cancer will have a family history of this disease. Families with 2 or more relatives diagnosed with pancreatic adenocarcinoma (this is the most common type of pancreatic cancer) are eligible for this study.

A recent publication about this collaboration ("Pancreatic Cancer Genetic Epidemiology Consortium", Peterson, et al., Cancer Epidemiology, Biomarkers & Prevention, 2006) summarizes our progress to date. Eligible families are identified by screening newly diagnosed patients (e.g., The Ontario Pancreas Cancer Study), physician referrals, and self-referrals (often via the internet). Collectively, over 13,000 patients were screened for a family history of pancreatic cancer. So far, approximately 475 families with familial pancreatic cancer have been enrolled, including the participation of over 1900 relatives. These families have provided information on diet, lifestyle, family history, medical history, and many have provided blood and/or tissue samples for genetic studies.

The mean age of diagnosis of pancreatic cancer in these families was approximately 64 years (with ages ranging from 30-97). This is approximately 5 years younger than the mean age of diagnosis in the general population. However, they did not see decreasing ages of diagnosis in families where there were a higher number of relatives affected with pancreatic cancer. This was unexpected since other types of hereditary cancers (such as hereditary breast and colorectal cancer) tend to have diagnoses decades younger than the general population. Since smoking is known to be a significant risk factor for pancreatic cancer, there is also interest on the smoking status of these patients. They found that 38% of patients were "never-smokers" and 47% were either smokers or ex-smokers.

Our recruitment of new families and data collection continues and the significant task of finding the pancreatic cancer gene(s) is on-going. We have recently provided the second batch of samples for genetic analysis. A very large number of families need to be screened to identify the few families that likely have a gene causing hereditary pancreatic cancer. The barriers to this type of research include the prognosis of this disease, the accuracy of family history details, the interest of family members to participate, the availability of medical records to confirm cancer diagnoses in the family, and the availability of blood and/or tissue samples from key family members. Almost all families have limitations as to what information and samples are available, and it may be difficult for any one research site to have enough families and data to determine the causes of this disease. The PACGENE Consortium is a valuable resource that will hopefully improve our understanding of pancreatic cancer. Please contact us if you or your family is interested in participating.