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Genetic Link Between Pancreatitis and Alcohol Consumption, Says Pitt Team

PITTSBURGH, Nov. 12, 2012 - A new study published online today in *Nature Genetics* reveals a genetic link between chronic pancreatitis and alcohol consumption. Researchers from the University of Pittsburgh School of Medicine and more than 25 other health centers across the United States found a genetic variant on chromosome X near the claudin-2 gene (*CLDN2*) that predicts which men who are heavy drinkers are at high risk of developing chronic pancreatitis.

This finding enables doctors to identify people with early signs of pancreatitis or an attack of acute pancreatitis who are at very high risk for progressing to chronic pancreatitis, allowing them to take preventative action to slow the development of the disease, and give the pancreas a chance to heal. Once an individual develops pancreatitis it takes several years for the pancreas to deteriorate.

“The discovery that chronic pancreatitis has a genetic basis solves a major mystery about why some people develop chronic pancreatitis and others do not,” said David C. Whitcomb, M.D., professor of medicine, cell biology and physiology, and human genetics at the University of Pittsburgh School of Medicine and lead author of the report. “We also knew there was an unexpected higher risk of men developing pancreatitis with alcohol consumption, but until now we weren’t sure why. Our discovery of this new genetic variant on chromosome X helps explain this mystery as well.”

Over 100,000 Americans suffer from chronic pancreatitis, a progressive inflammatory disease characterized by abdominal pain and permanent damage to the pancreas. Most studies report excessive alcohol consumption as the major risk factor for adult-onset chronic pancreatitis. However, according to Dr. Whitcomb, only 3 percent of individuals who are alcoholics develop chronic pancreatitis, suggesting a pancreas-specific risk factor.

The study was conducted over 10 years and involved more than 2,000 patients, all of whom underwent DNA testing in a study funded by the National Institutes of Health. Researchers discovered that there was a common DNA variant on the X chromosome that is present in 26 percent of men without pancreatitis, but jumps to nearly 50 percent of men diagnosed with alcoholic pancreatitis. Women have two X chromosomes, so most women with the high-risk DNA variant on one X chromosome appear to be protected from alcoholic chronic pancreatitis by the other X chromosome, if it is normal. Men have one X chromosome and one Y chromosome, so if they inherit a high-risk X chromosome, there is no protection.

The factor on chromosome X does not appear to cause pancreatitis but if pancreatic injury occurs for any reason such as gallstone pancreatitis or abdominal trauma, it is more likely that the person will develop chronic pancreatitis - especially if they also drink alcohol.

“This information is important because the high-risk chromosome can be identified in patients who drink and have early signs of pancreatic injury,” said Dhiraj Yadav, M.D., M.P.H., a co-investigator on the study. “If pancreatic injury and acute pancreatitis occur, patients must stop drinking immediately.”

Nationally, 16 percent of men drink alcohol at levels defined by the National Institute on Alcohol Abuse and Alcoholism (NIAAA) as high risk. Twenty-six percent of these men who drink heavily are at high risk of chronic pancreatitis following pancreas injury. Only 10 percent of women drink alcohol at dangerous levels, and of these only 6 percent have the X chromosome variant on both X chromosomes.

“Previous discoveries show that chronic pancreatitis without alcohol involvement has a strong genetic link. This helps to eliminate the previous stigma that patients with chronic pancreatitis must also be heavy drinkers,” added Dr. Whitcomb. “This study proves that there is a genetic element to the disease.”

Referrals of at-risk patients are welcome at UPMC and other large academic centers. The Pancreas Clinic within the UPMC Digestive Disorder Center is designed to evaluate patients using genetic and other data to provide treatment that is individualized to each patient. In addition to clinical care, the physician-scientists who staff this clinic are actively involved in teaching physicians and trainees the art and science of personalized medicine for chronic pancreatitis.

Collaborators on this study include Bernie Devlin, Ph.D., Adam Slivka, M.D., Ph.D., Dhiraj Yadav, M.D., M.P.H., Randall E. Brand, M.D., Vijay Singh, M.D., Alyssa M Krasinskas, M.D., all of the [University of Pittsburgh](#); Jill P. Smith, M.D., of [Pennsylvania State - Hershey](#); John P. Neoptolemos, M.D., of the [University of Liverpool](#); Markus M. Lerch, M.D., of the [University of Greifswald](#); and others.

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About the University of Pittsburgh School of Medicine

As one of the nation’s leading academic centers for biomedical research, the University of Pittsburgh School of Medicine integrates advanced technology with basic science across a broad range of disciplines in a continuous quest to harness the power of new knowledge and improve the human condition. Driven mainly by the School of Medicine and its affiliates, Pitt has ranked among the top 10 recipients of funding from the National Institutes of Health since 1997. In rankings recently released by the National Science Foundation, Pitt ranked fifth among all American universities in total federal science and engineering research and development support.

Likewise, the School of Medicine is equally committed to advancing the quality and strength of its medical and graduate education programs, for which it is recognized as an innovative leader, and to training highly skilled, compassionate clinicians and creative scientists well-equipped to engage in world-class research. The School of Medicine is the academic partner of [UPMC](#), which has collaborated with the University to raise the standard of medical excellence in Pittsburgh and to position health care as a driving force behind the region’s economy. For more information about the School of Medicine, see www.medschool.pitt.edu.

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