Adenoid Cystic Carcinoma Research Foundation

# Fusion Gene Discovered in ACC (October 2009)

A landmark finding for ACC patients and researchers was announced today by the Sahlgrenska Academy at the University of Gothenberg (Sweden). Dr. Göran Stenman, a distinguished Professor of Pathology, has identified a protein that is encoded by the fusion of two genes that typically reside on separate chromosomes. All cases of ACC have this gene fusion, according to Dr. Stenman's article in The Proceedings of the National Academy of Sciences, a highly-respected scientific journal (see the press release below).

The diagnostic value of the finding is extraordinary. In the future, pathologists will be able to diagnose ACC patients rapidly and definitively. However, the therapeutic implications may be even more significant. If the fusion gene is found to be essential for the survival and progression of ACC tumors, then researchers will be able to target the specific gene, the protein it encodes, and/or other molecules triggered by the gene's activity. Clearly, this line of research will be a major priority for ACCRF.

ACCRF Co-Founders Marnie and Jeff Kaufman were fortunate to meet Dr. Stenman at the two NIH Workshops on Salivary Gland Cancers that they helped organize in October 2006 and November 2008. As an outgrowth of the first meeting, ACCRF provided funding in late 2006 for the expansion of a genomic study underway in Dr. Stenman's laboratory. Today's announcement demonstrates the importance of supporting the basic research of committed and talented investigators who will drive the important advances in our shared battle against ACC.

These are exciting times in which we look forward to sharing future successes with you...

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Press release from the Sahlgrenska Academy at the University of Gothenburg
13 October 2009

## New cancer gene discovered

A new cancer gene has been discovered by a research group at the Sahlgrenska Academy. The gene causes an insidious form of glandular cancer usually in the head and neck and in women also in the breast. The discovery could lead to quicker and better diagnosis and more effective treatment.

The study is published today in the prestigious scientific journal Proceedings of the National Academy of Sciences (PNAS).

The cancer caused by this new cancer gene is called adenoid cystic carcinoma and is a slow-growing but deadly form of cancer. The research group can now show that the gene is found in 100% of these tumours, which means that a genetic test can easily be used to make a correct diagnosis.

"Now that we know what the cancer is down to, we can also develop new and more effective treatments for this often highly malignant and insidious form of cancer," saysprofessor Göran Stenman, who heads the research group at the Lundberg Laboratory for Cancer Research at the Sahlgrenska Academy. "One possibility might be to develop a drug that quite simply turns off this gene."

The newly discovered cancer gene is what is known as a fusion gene, created when two healthy genes join together as a result of a chromosome change.

"Previously it was thought that fusion genes pretty much only caused leukaemia, but our group can now show that this type of cancer gene is also common in glandular cancer," says Stenman.

One of the two genes that form the fusion gene is known as MYB. Among other things, this gene controls cell growth and makes sure that the body gets rid of cells that are no longer needed. It has long been known to be a highly potent cancer gene in animals, but for a long time there was no evidence of the gene being involved in the development of tumours in humans.

"We suggested back in 1986 that the MYB gene might be involved in this form of cancer, but it's only recently that we've had access to the tools needed to prove it," says Stenman.

The research group has also looked at the mechanism behind the transformation of the

normal MYB gene into a cancer gene. Genes can be compared to blueprints for proteins. Carefully controlled regulating systems then determine when and how much of each protein is formed. One such regulating system, discovered recently, is microRNA, which can turn genes on and off. When this cancer gene forms, this important control system is put out of action, leading to activation of the gene and massive overproduction of an abnormal MYB protein with carcinogenic properties.

"This is an important discovery, because it's a new mechanism which I think will turn out to be quite common in a variety of human cancers," says Stenman.

The study was conducted with support from the Swedish Cancer Society and Sahlgrenska University Hospital, among others.

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The Sahlgrenska Academy is the faculty of health sciences at the University of Gothenburg. Education and research are conducted within the fields of medicine, odontology and health care sciences. About 4 000 undergraduate and 1 000 postgraduate students are enrolled at the academy. There are 1 500 members of staff, of whom 850 are researchers and/or teachers.