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BULLETIN

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CANADIAN GENETIC DISEASES NETWORK

FOR IMMEDIATE RELEASE
NETWORK SCIENTISTS IN MONTREAL & TORONTO MAKE BREAKTHROUGHS IN *THREE* GENETIC DISEASES

Network scientists Dr. Diane Cox and Dr. John Dick of Toronto's Hospital for Sick Children, and Dr. Steven Narod of McGill University's Montreal General Hospital have recently published news of three research breakthroughs in genetic-related diseases.

The February 1 issues of *Nature Genetics*, *Human Molecular Genetics,* and *Nature Medicine* respectively carried the news.

Just over one year ago, **Dr. Diane Cox'**s team cloned the gene for **Wilson disease**, a devasting syndrom which causes copper poisoning of the liver and brain. In her latest paper, Dr. Cox has announced the discovery of 20 new mutations which, it is believed, will lead to very early diagnosis of the disease in childhood. Another important practical advance is the potential application of these mutations to screening for patients already suspected of having Wilson disease.

Said Dr. Cox "This changes the whole outlook on the diagnosis of liver disease in early childhood and provides a reliable predictive test in families where a diagnosis has been made."

This discovery is of major importance because, unlike many childhood liver diseases, Wilson disease, once identified, is treatable. One in every 30,000 Canadians is believed to carry the defective gene.

Dr. John Dick has made a significance advance in the development of a unique gene therapy model for **thalassemia and sickle cell anemia,** both widespread and severe anemias. Dr. Dick and his team have transplanted into mice, human cells which carry the genetic deficiencies of these diseases. The cells develop into

all blood lineages, including the same defective red blood cells as the anemia sufferer.

Explains Dr. Dick "The importance of this advance is that researchers will now be able to directly examine human cells prior to pre-clinical experiments." He added "This is a good gene therapy model in that we can now assess gene transference into the damaged cells, measure gene expression, and finally follow correction of the red cell defect."

The model is viewed as a valuable stepping stone in anemia research.

Dr. Steven Narod, a member of the research team which recently isolated the breast cancer gene, BRCA1, is now focussing attention on prostate cancer, the most common cancer in men in Canada.

A new study carried out by Dr. Narod and Dr. Fernand Labrie at the University of Laval has found that one in ten men who have brothers with prostate cancer are susceptible also to prostate cancer. Results from a group of over 6,000 randomly selected men between the ages of 50 and 80 showed that cancer was detected in 6% more of the group with brothers with cancer, than in the group with no brothers with cancer. The study also indicates that the cancers in the higher risk group can be detected early with the currently used prostate-specific antigen (PSA) blood screening test.

According to Dr. Narod, men with a family history of prostate cancer should be made aware of their increased risk. No significant excess risk appears to be associated with a father who has prostate cancer.

Dr. Cox, Dr. Dick, and Dr. Narod are members of the Canadian Genetic Diseases Network, a consortium of 38 of Canada's leading geneticists who are linked with twelve universities across Canada, 13 core facilities, and 8 industrial partners to form an "institute without walls". The network carries a mandate under the federal government's Networks of Centres of Excellence Program to perform leading-edge research and to work with industry partners on detection and treatment of genetically-transmitted disease.