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CANADIAN GENETIC DISEASES NETWORK

FOR IMMEDIATE RELEASE: April 1, 1996

SCIENTIFIC ADVANCES IN EYE DISEASE & FANCONI ANEMIA

Research teams led by two members of the Canadian Genetic Diseases Network based at Toronto's Hospital for Sick Children have today announced significant scientific advances in two important healthcare areas.

Dr. Roderick McInnes of the Department of Genetics and Paediatrics at HSC, together with colleagues from the U.S., has identified a major regulatory gene, called Chx10, that is a critical control of retina and eye development in mammals. While Chx10 was discovered by the McInnes lab several years ago, it is these latest findings which demonstrate that Chx10 is essential for the development of a normal eye.

In studies of mice, the team found that a malfunctioning Chx10 gene was responsible for profound retinal abnormalities such as eye size, reduced number of cells in the retina, and the absence or incomplete formation of one of the major retinal cell types, bipolar cells. Recent research has indicated that the network of genes required to form an eye may be very similar across a wide range of species, and indicates that the Chx10 gene may be essential for normal eye development in humans.

The discovery of genes which control retinal development may have long term implications for the treatment of conditions that cause destruction of the human retina and may eventually lead to therapies for these disorders if a healthy retina could be regenerated from a damaged one.

The results of the findings are published in the April issue of *Nature Genetics*.

A second team of scientists, led by Dr. Manuel Buchwald at the Hospital for Sick Children has successfully reproduced characteristics of Fanconi Anemia in mice. The development of a mouse model of this rare inherited blood disorder (one in 350,000 people) provides researchers with a means of testing novel treatments for the disease, including gene therapy. The team's findings are published in the April issue of *Nature Genetics.*

Since 1992 when Dr. Buchwald's research group cloned the gene responsible for one form of Fanconi Anemia, the team has worked towards obtaining a better understanding of the parameters and biochemical properties of the disease. The mouse model, which shares several important characteristics of the human disease, significantly assists the ongoing research. The most severe effect of Fanconi Anemia is the failure of the bone marrow to make red and white blood cells, and platelets. The devastating results leave sufferers anemic and weak and prone to severe bleeding due to insufficient blood clotting. Patients are also at increased risk for leukemia and cannot fight infection. They rarely survive to adulthood.

Commented Dr. Michael Hayden, Scientific Director of the Canadian Genetic Diseases Network "I congratulate Dr. McInnes and Dr. Buchwald on these outstanding scientific contributions. These results are an excellent example of the benefits of NCE collaborations and facilities across Canada which are crucial to ongoing research of this calibre."

The Canadian Genetic Diseases Network is a consortium of 38 of Canada's leading geneticists who are linked with 12 universities, 9 hospitals, and 11 core facilities across the country. The Network performs leading-edge research on common genetically transmitted diseases and works with industry partners on methods of detection and treatment of the diseases. Core funding for Network programs is provided by the federal Networks of Centres of Excellence Program through the Medical Research Council of Canada.

For further information contact:
Carol Smith, Network Manager
Canadian Genetic Diseases Network
Tel. 604-822-7217/7886