NCE·GENETICS

BULLETIN

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CANADIAN GENETIC DISEASES NETWORK

SCIENTISTS FIND MAJOR FANCONI ANEMIA GENE

Toronto, November 1, 1996: An international team of scientists, including a group of researchers led by Dr. Manuel Buchwald, a Network principal investigator and Director of the Research Institute at Hospital for Sick Children (HSC) in Toronto, has discovered the gene which is responsible for more than 60% of Fanconi anemia cases. The rare disease is most severely characterized by the failure of bone marrow which produces all the body's blood cells and affects one in 350,000 people.

In 1992 Dr. Buchwald led a team at Hospital for Sick Children which cloned the first gene known to be responsible for 15% of cases of Fanconi anemia (FAC). Subsequently, in April 1966, Dr. Buchwald's team successfully reproduced characteristics of Fanconi anemia in mice. The latest discovery of the major Fanconi anemia gene (FAA), thought to be one of five different genes which contribute to the disease, now means that scientists have a better chance of understanding the basic defect leading to Fanconia anemia. The results were published in the November issue of the international journal *Nature Genetics*.

The affects of the disease are devastating, leaving patients weak and prone to severe bleeding due to insufficient blood clotting. Said Dr. Buchwald, "Patients can be treated with transfusions, but the only cure is a bone marrow transplant which is particularly difficult for them because of the sensitivity to chemotherapy which Fanconi anemia produces." He added that patients are also at an increased risk for developing leukemia and many do not survive to adulthood.

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The cloning of the FAA gene increases the likelihood that eventual gene therapy treatment for the disease will benefit a broader range of Fanconia anemia patients.

Funding and support for Dr. Buchwald's project is provided in part by the Canadian Genetic Diseases Network through the federal government's Networks of Centres of Excellence program and the Medical Research Council of Canada. Said Dr. Buchwald, "The network made important contributions to the research especially through its infrastructure." Dr. Lap-Chee Tsui, Geneticist-in-Chief at the Hospital for Sick Children added "Since the inception of the NCE program in 1990, significant advances have been made in genetics research projects at HSC which are supported by the Canadian Genetic Diseases Network through R&D funding, core technology facilities, and technology management."

The Network is a consortium of 38 of Canada's leading geneticists and their research teams who are linked with 11 universities, 8 hospitals, and 9 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.

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