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CANADIAN GENETIC DISEASES NETWORK

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CANADIAN GENETIC DISEASES NETWORK COLLABORATES IN THE DISCOVERY OF THE GENE RESPONSIBLE FOR SPINAL MUSCULAR ATROPHY (SMA)

The Canadian Genetic Diseases Network has played a key collaborative role in the discovery by University of Ottawa reserchers of the gene responsible for Spinal Muscular Atrophy (SMA), a fatal neurodegenerative disorder. The scientific findings of the research team, led by Dr. Alex MacKenzie of the Children's Hospital of Eastern Ontario (CHEO), are to be published as a cover story in the January 13, 1995 issue of the international scientific journal 'Cell'.

Network Principal Investigator, Dr. Robert Korneluk of CHEO, collaborated in the discovery and conducted the critical large scale genomic DNA sequencing portion of the project. Dr. Korneluk's collaboration was supported financially by the Canadian Genetic Diseases Network with advanced DNA sequencing equipment provided through a Network alliance with the Japan Research and Development Corporation.

The SMA disorder is manifested as a weakness and wasting of the voluntary muscles and, in its most severe form, attacks the fetus in utero or the newborn within the first few months of life. Affected children are unable to sit unsupported and rarely survive their first few years owing to respiratory muscle weakness. The disease is the most common genetic cause of death in Canadian infants, affecting approximately 1,000 Canadian children and adults.

The discovery is the second major breakthrough in neuromuscular disease within three years. In 1992 a team headed by Dr. Korneluk at CHEO located the gene causing Myotonic Dystrophy. Dr. Korneluk's work on neuromuscular diseases led to the SMA collaboration.

Said Dr. Korneluk "As evidenced by Dr. MacKenzie's research, an important aspect in the isolation of the SMA gene is the discovery of a possible link to the prevention of pre-programmed cell death which accounts for other widespread neuromuscular genetic diseases such as Parkinson's Disease, ALS (Lou Gehrig's) and Alzheimer's Disease".

Scientific Director of the Canadian Genetic Diseases Network, Dr. Michael Hayden, commented "This is a remarkable scientific development and an excellent example of the impact of strategic alliances and collaborations available through the federal Networks of Centres of Excellence program".

The Canadian Genetic Diseases Network is a consortium of 38 of Canada's leading geneticists who are linked with nine universities across Canada and 11 technology core facilities 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.

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