BULLETIN

CANADIAN GENETIC DISEASES NETWORK

SCIENTISTS FIND GENE FOR SEVERE FORM OF EPILEPSY

Toronto - September 28, 1998: An international research team led by Canadian Genetic Diseases Network scientist, Dr. Steve Scherer of The Hospital for Sick Children (HSC) and the University of Toronto, has identified a gene responsible for one of the most severe forms of epilepsy: Lafora disease (LD).

Lafora disease occurs during late childhood or early adolescence and is characterized by seizures and progressive neurological degeneration. Death usually occurs within a decade of the first symptoms.

Scientific investigations over the past 50 years have led biochemists to suspect that LD was caused by malfunction of carbohydrate metabolism in the brain; however, the fundamental defect triggering the malfunction was unknown. The LD gene produces a signalling protein which is thought to be involved in the brain's breakdown of carbohydrates. The defective gene interfers with this process, leading to the accumulation of abnormal sugar molecules which likely lead to the destruction of the brain's nerve cells. "Identifying the LD gene gives us the cellular key to determining the cause of the seizures," said Dr. Scherer.

The discovery is reported in the October issue of the scientific journal *Nature Genetics.*

The next step in understanding the disease is to discover the basic mechanisms that lead to the severe epilepsy and ultimately, to develop diagnostic tools and therapies.

Canadian Genetic Diseases Network researchers and facilities have played a major part in the discovery. CGDN members of the research team include Dr. Guy Rouleau of Montreal General Hospital, Dr. Lap-Chee Tsui of HSC, and Sylvia Soder of the Network DNA sequencing facility located at HSC. The facility is one of three sequencing centres and 15 core technology facilities supported by CGDN and located strategically across Canada. The centres are designed to provide rapid and efficient access to advanced technologies which are critical to the success of the scientific discovery process.

Said Dr. Michael Hayden, Scientific Director of the Canadian Genetic Diseases Network from CGDN headquarters in Vancouver, "The discovery of the epilepsy gene is a major breakthrough in the research of this disease. I am extremely pleased that CGDN has played an important role in this success."

The Canadian Genetic Diseases Network is a nation-wide centre of research excellence which creates strategic partnerships to commercialize discoveries for the benefit of health care in Canada and worldwide. Fifty scientists and their teams, based in 18 universities, hospitals, and research centres across Canada, focus on the molecular and cellular causes of inherited disease. In 1997, CGDN was awarded a further seven years' core funding by the federal Networks of Centres of Excellence Program via the Medical Research Council of Canada, to conduct an expanded research program "From Genes to Therapies."

Contact: Carol Smith, Network Manager
Canadian Genetic Diseases Network
Tel. 604) 822-7217/822-7886 Fax 604) 822-7945