BULLETIN

CANADIAN GENETIC DISEASES NETWORK

# SCRIVER RESEARCH GROUP ANTICIPATES RELIEF FOR PKU SUFFERERS

**Montreal - March 3, 1999:** A group of researchers at McGill University, led by Canadian Genetic Diseases Network principal investigator, Dr. Charles Scriver, has developed an enzyme substitution approach to treating the disease phenylketonuria (PKU). Sufferers of the disease carry extremely high levels of phenylalanine, an amino acid nutrient in the blood which, when left untreated, commonly leads to mental retardation.

The disease is due to the absence of an enzyme which is normally present in the liver for the purpose of degrading the phenylalanine ingested through foods such as milk, cheese, meat, fish, breads and nuts. In an article published March 2 in the *Proceedings of the NationalAcademy of Sciences USA*, under the first authorship of CGDN graduate student, Christineh Sarkissian, the Montreal group describes a method of producing the missing enzyme in large quantities in bacterial cells. Results of the study indicate a 30% to 50% reduction in phenylalanine concentration in the blood of laboratory mice.

Although successful treatment of PKU requires a 70% to 80% reduction in blood phenylalanine levels, researchers are hopeful that these new findings may eventually lead to a therapy in humans and a significant increase in the tolerance of PKU sufferers for many common foods. Currently, the disease imposes a stringent, lifelong controlled diet and a dependence on nutritional supplements to reduce blood levels of phenylalanine and prevent mental retardation.

Dr. Scriver's research of PKU has been supported by the Canadian Genetic Diseases Network for nine years. The latest findings have been developed in collaboration with a CGDN affiliate, IBEX Technologies of Montreal.

The Canadian Genetic Diseases Network is a not-for-profit nation-wide centre of research excellence under the federal Networks of Centres of Excellence Program. CGDN 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 Networks of Centres of Excellence Program via the Medical Research Council of Canada, to conduct an expanded research program "From Genes to Therapies."

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