NEWS RELEASE – FOR IMMEDIATE RELEASE

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Canadian Scientists Advancing Identification of Rare Disease Genes

Vancouver, BC— There are over 7,000 known genetic diseases. Although each one is rare, as a group they have an enormous impact on the well-being of Canadians, affecting the lives of approximately 500,000 children and 2.5 million adults and their families. These disorders cause a variety of medical problems such as birth defects, intellectual disability, difficulty with physical growth and organ failure. Surprisingly, the majority of genes causing these conditions are still unknown, but a group of Canadian scientists wants to change this.

Finding of Rare Disease Genes in Canada (FORGE Canada), funded in part by Genome British Columbia, is a project that aims to change the way that rare diseases are identified and treated. The advent of next-generation sequencing technologies allows for rapid identification of disease-causing genes in small families or a few unrelated individuals with the same rare condition. This technology maximizes a tremendous opportunity as there is presently no efficient mechanism in our healthcare system to diagnose rare diseases.

The FORGE Canada team will begin by studying 70 different rare diseases and they are optimistic that families participating in this research project will begin to benefit in a matter of weeks. Upon diagnosis they can better understand the impact and treatment options: unlike people who suffer from more common disorders such as cancer or heart disease, families affected by rare diseases have nowhere to turn. Research is often the only way to get answers to their questions.

Research into rare diseases also has enormous implications for more common diseases. Dr. Jan Friedman, a scientist at the Child & Family Research Institute and professor of Medical Genetics at the University of British Columbia, is a co-leader on the FORGE Canada project. Dr. Friedman cites the example of research into a group of patients with a rare genetic disorder that impaired their ability to clear cholesterol from their bodies. Children born with this rare disease would be at risk for heart attacks in their early teens and would therefore have to undergo bypass surgery. A result from this specific research is a drug called Lipitor, which is a cholesterol-reducing drug used by millions of people.

“If we can understand the condition then we can gain insight into possible treatment,” says Dr. Friedman. “The treatment could be drug therapy, but might be something as simple as vitamin or dietary intervention. It could be a novel discovery or just a new way of using existing information never applied to a certain condition before.”

Better diagnoses will allow Canadian healthcare teams to reduce or prevent patient complications and to develop tailored treatments. In the long term, identification of disease genes is an essential step toward the development of drugs that will one day improve the lives of affected children.

“Helping Canadian families is why we support these exceptional projects,” says Dr. Alan Winter, President and CEO of Genome BC. “Without this research thousands of Canadian families would remain isolated, frustrated and without support for the rare diseases which afflict them.” Primary funding for this project comes from Genome BC, Genome Canada, Genome Quebec and the Canadian Institutes of Health Research (CIHR).

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About Genome British Columbia:
Genome British Columbia is a catalyst for the life sciences cluster on Canada’s West Coast, and manages a cumulative portfolio of over $450M in technology platforms and research projects. Working with governments, academia and industry across sectors such as forestry, fisheries, agriculture, environment, bioenergy, mining and human health, the goal of the organization is to generate social and economic benefits for British Columbia and Canada.
www.genomebc.ca

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