News from SickKids®

FOR IMMEDIATE RELEASE
March 20, 2008

International partnership to produce "gold standard" gene copy reference of the human genome

Wellcome Trust grant expands the Toronto Database

TORONTO - A grant of CDN $870,000 awarded to The Centre for Applied Genomics (TCAG) at The Hospital for Sick Children (SickKids) jointly with the European Molecular Biology Laboratory's European Bioinformatics Institute (EMBL-EBI) will collect, organize and curate genome-wide information, enhancing the translation of genetic information into new diagnostic, prognostic and therapeutic tools for the improved health of children.

The funding will expand operations of TCAG's Database of Genomic Variants (DGV)—known as "the Toronto Database"—the world's premier database housing structural and copy number variation data in the human genome, providing significant support for investigations into structural variation in human biology, considered by *Science* magazine as the breakthrough story of 2007.

"The DGV has served the community well, and with this new grant we can expand our data curation capabilities, as higher resolution micro arrays and DNA sequencing accelerate structural variation discovery," says Dr. Stephen Scherer, DGV founder, senior scientist in Genetics & Genomic Biology at SickKids and professor of Molecular Genetics at the University of Toronto. It was Scherer who with Lars Feuk (Toronto), Charles Lee (Boston) and Matthew Hurles and Nigel Carter (Wellcome Trust Sanger Institute) among other colleagues discovered the existence of CNVs as the most common form of genetic variation in the human genome.

Sequencing of the human genome resulted in discoveries about the differences in our genes and their relationship to the tremendous variety of the human species. Only a few years ago scientists also discovered that certain genes are present in multiple copies in some individuals but not in others, and these copy number variable regions known as CNV have been shown to influence susceptibility to disease and response to treatments. Advances in CNV detection technology have added to the number of known CNV regions in the genome; yet these regions are still considered vastly under-ascertained. Additional technological advances ensure that the rate of data generation will continue to dramatically increase.

“The revolution in medicine arising from our understanding of the human genome is only just beginning,” says Hon. John Wilkinson, Ontario’s Minister of Research and Innovation. “Through the state-of-the-art core genomics resources and services at SickKids that drive this activity, Ontario will lead the exponential growth in information researchers and industry need to develop new diagnostic tests and treatments.”

The Wellcome Trust funding supports the collection, organization and sharing of CNV data that is being generated around the world into a central repository, helping researchers maximize this knowledge for understanding human variation including those genetic differences responsible for disease.

“Analysing data from large-scale studies of human variation allows us to create a detailed map of structural differences in the human genome; this will help scientists to understand the genetic basis of individuality, including differences in susceptibility to many common diseases,” says Dr. Paul Flicek, the lead investigator of the EMBL-EBI team and joint head of the popular Ensembl Genome Browser. The current DGV is accessed daily by thousands of clinicians and scientists worldwide. Among them is Dr. Mansoor Mohammed, chief executive officer of Combimatrix Diagnostics in California whose clinical diagnostics rely on the accurate and updated DGV.

"The biological importance of copy number variability is clear, only three years following its discovery. With the technology now available to discover CNV regions at an unprecedented resolution, CNV research will be greatly facilitated by this database," adds Dr. Alan Schafer, head of Molecular and Physiological Sciences, Wellcome Trust

Key members of the new partnership include SickKids scientists Dr. Lars Feuk of the Genetics & Genome Biology Program at SickKids, along with Jeff MacDonald, Junjun Zhang and Bhooma Thiruvahindrapduram of TCAG. The DGV is currently funded by the Ontario Ministry of Research and Innovation, Genome Canada through the Ontario Genomics Institute, the McLaughlin Centre for Molecular Medicine and the SickKids Foundation.

SickKids, affiliated with the University of Toronto, is Canada’s most research-intensive children’s hospital and the largest centre dedicated to improving children's health in the country. As an innovator in child health, SickKids improves the health of children by integrating care, research and teaching. The mission of SickKids is to provide the best in complex and specialized care by creating scientific and clinical advancements, sharing our knowledge and expertise and championing the development of an accessible, comprehensive and sustainable child health system. For more information, please visit www.sickkids.ca. SickKids is committed to healthier children for a better world.

- 30 -

For more information, please contact:
Janice Nicholson
The Hospital for Sick Children
416-813-6684
janice.nicholson@sickkids.ca