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NEWS RELEASE

Canadian, British and American scientists launch major new genome partnership to catalogue all common copy number variations

International team focuses on copy number variants in the human genome

6 July 2007, TORONTO, BOSTON, HINXTON - An international team will use state-of-the-art, high-density microarrays and new computer algorithms to improve the detection of variants in the human genome which are implicated in various diseases. The new systems are the foundation of Phase 2 of the Genome Structural Variation Consortium, which was set up in 2004 and seeks to identify structurally variable regions in the human genome.

In 2006, the Genome Structural Variation Consortium, along with other international collaborators, generated a first-generation map of copy number variants (CNVs) covering the human genome. The CNVs that they identified, which in many cases lead to deletion or duplication of genes along chromosomes, tended to encompass large stretches of DNA ranging from thousands to millions of chemical bases of DNA. These changes are part of the normal variability between apparently healthy people, but some may also predispose individuals to disease. The data also suggested that thousands more CNVs exist, but technological limitations associated with that earlier study precluded the discovery of more CNVs.

The new comprehensive map will be critical for studies attempting to identify genes involved in both rare and common diseases. "Our experiments will generate the highest-resolution CNV catalogue of worldwide populations. The initiative will also complement ours, and other efforts to sequence entire genomes", said Dr. Stephen Scherer, Senior Scientist and Director, The Centre for Applied Genomics (TCAG) at The Hospital for Sick Children (SickKids) in Toronto.

In its second phase, this international research collaboration will develop a comprehensive, higher resolution CNV map for the Human Genome — at a level 100-fold finer than the first map. Working with NimbleGen Systems, Inc. of Madison, Wisconsin, USA, the Consortium has designed a novel set of 2.1-million-feature microarrays that will enable genome-wide detection of CNVs with 42 million probes.

TCAG SickKids wellcome trust sanger institute Harvard Medical School Brigham and Women's Hospital

The set of microarrays have been referred to as "whole-genome oligonucleotide tiling arrays" because they cover almost all DNA along chromosomes, spaced at intervals of only 50 bases, or letters of DNA code.

The international consortium is led by Scherer, Drs. Nigel Carter, Matthew Hurles, and Chris Tyler-Smith of the Wellcome Trust Sanger Institute, and Dr. Charles Lee from Brigham and Women's Hospital and Harvard Medical School.

To better understand CNVs and diseases, researchers and clinicians around the world are now accessing the new CNV maps from the "Database of Genomic Variants" (http://projects.tcag.ca/variation/) hosted by SickKids. TCAG is a genomics platform funded by Genome Canada through OGI.

"Copy Number Variation has emerged over the past few years as a novel and productive focus for understanding variation within the human genome as well as other species' genomes," noted Dr. Christian Burks, President and CEO of the Ontario Genomics Institute (OGI). "We are delighted, in conjunction with funding from Genome Canada and Ontario's Ministry of Research and Innovation, to be supporting this work, and to see Ontario and Canada at the forefront of this area of biomedical research."

"From our Phase 1 map, we suspected there remains much more CNV to be discovered," said Dr. Matthew Hurles, Investigator at the Wellcome Trust Sanger Institute. "Because of the higher resolution of the new systems, we will, for the first time, be able to uncover these smaller variants."

Dr. Nigel Carter, Senior Investigator at the Sanger Institute added, "The availability of flexible high-density oligonucleotide arrays has made a study of such scope possible. The first data from the 42 million probe set has confirmed the ability of this approach to identify copy number variants at least as small as 500 base pairs in size."

The new arrays will be used to scan DNA samples from dozens of individuals of diverse geographic background, in experiments designed to capture all CNVs with a frequency of five percent or greater in the world. As such, the project will generate more than 1.68 billion data points.

"We also anticipate being able to obtain fine-scale information on the anatomy of individual CNVs" said Dr Charles Lee, Director of Cytogenetics (Harvard Cancer Center), Brigham and Women's Hospital and Harvard Medical School. "Ultimately, the data generated from our Phase 2 studies will be important for disease association studies, cancer biomarker studies, and accurate interpretation of many genetic diagnostic tests."

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Notes for editors:

About TCAG

The Centre for Applied Genomics (TCAG) is located in the Research Institute of The Hospital for Sick Children (SickKids), Toronto, and is a Science and Technology Platform of Genome Canada, funded by Genome Canada through the Ontario Genomics Institute. TCAG provides genomics infrastructure to facilitate a wide variety of research, including human genomics and disease, model organisms, and agricultural and food sciences. The Centre's services are available to all clients in the academic, government or private sectors. TCAG supports a number of large-scale Genome Canada projects, other national and international genomics efforts, as well as hundreds of additional researchers in Ontario, Canada, and worldwide. www.tcag.ca

About Wellcome Trust Sanger Institute
The Wellcome Trust Sanger Institute, which receives the majority of its funding from the Wellcome Trust, was founded in 1992 as the focus for UK sequencing efforts. The Institute is responsible for the completion of the sequence of approximately one-third of the human genome as well as genomes of model organisms such as mouse and zebrafish, and more than 90 pathogen genomes. In October 2005, new funding was awarded by the Wellcome Trust to enable the Institute to build on its world-class scientific achievements and exploit the wealth of genome data now available to answer important questions about health and disease. These programmes are built around a Faculty of more than 30 senior researchers. The Wellcome Trust Sanger Institute is based in Hinxton, Cambridge, UK.
www.sanger.ac.uk/humgen/cnv/

About Brigham and Women's Hospital (BWH)
Brigham and Women's Hospital (BWH) is a 747-bed nonprofit teaching affiliate of Harvard Medical School and a founding member of Partners HealthCare System, an integrated health care delivery network. BWH is committed to excellence in patient care with expertise in virtually every specialty of medicine and surgery. The BWH medical preeminence dates back to 1832, and today that rich history in clinical care is coupled with its national leadership in quality improvement and patient safety initiatives and its dedication to educating and training the next generation of health care professionals. Through investigation and discovery conducted at its Biomedical Research Institute (BRI), BWH is an international leader in basic, clinical and translational research on human diseases, involving more than 800 physician-investigators and renowned biomedical scientists and faculty supported by more than $400M in funding. BWH is also home to major landmark epidemiologic population studies, including the Nurses' and Physicians' Health Studies and the Women's Health Initiative.
www.brighamandwomens.org
www.chromosome.bwh.harvard.edu

About Ontario Genomics Institute (OGI)
The Ontario Genomics Institute (OGI) is a private, not-for-profit corporation focused on providing leadership for Ontario in helping build a globally-competitive life sciences sector by creating leverageable genomics resources with top-notch research. Through its relationship with Genome Canada, the Ontario Ministry of Research and Innovation (MRI), and other private and public sector partners, OGI helps Ontario-based scientists secure funding for research in genomics and proteomics as well as for commercialization activities. OGI also focuses on the potential ethical and social issues that can arise with and from such research. www.OntarioGenomics.ca

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