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Canadian researchers play key role in groundbreaking scientific accomplishment

International Consortium completes map of human genetic variation.

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Montréal (QC) - Wednesday October 26, 2005 – The International HapMap Consortium today announced the first comprehensive catalog of human genetic variation, a landmark achievement that is already accelerating the search for genes involved in common diseases, such as asthma, diabetes, cancer and heart disease. Two prominent Canadian researchers, who played a key role in the project, participated in the announcement in Salt Lake City.

Dr. Tom Hudson, from McGill University and Génome Québec Innovation Centre, was the Canadian leader of the HapMap project and one of the original leaders of the HapMap concept paper. Bartha Knoppers, Ph.D., LL.B, from the University of Montréal, was co-chair of the international ethics team and led the ethics discussions in the HapMap Project. Using the high throughput DNA technology platforms of McGill University and Génome Québec Innovation Centre, the Canadian group, analysed chromosomes 2 and 4p of the human genome, work which generated 10% of the data for the HapMap project

HAP MAP - A DESCRIPTION

The Hap Map is a catalogue of common genetic variants that occur in human beings. It describes what these variants are, where they occur in our DNA, and how they are distributed among people within populations and among populations in different parts of the world. In a paper in the October 27, 2005 issue of the journal *Nature,* more than 200 researchers from Canada, China, Japan, Nigeria, the United Kingdom and the United States describe the initial results to chart the patterns of genetic variation that are common in the world's population. The results provide convincing evidence that variation in the human genome is organized into local neighborhoods, called haplotypes.

"The Hap Map is a critical tool that will speed up the discovery of genes responsible for diseases", said Tom Hudson from Salt Lake City. "*To make a comparison, today's announcement is like the first trans-Atlantic flight: once the trip was done, travel was changed forever. With the completion of the first HapMap, human genetics has just crossed a similar barrier".*

THE ORIGINS OF HAPMAP – A CANADIAN CONTRIBUTION

When scientists first announced the draft sequence of the human genome in 2001, the challenge of testing 3 billion DNA letters to find out the location of genes responsible for disease appeared daunting. A group of Canadian (Tom Hudson in Montreal and Kathy Siminovitch in Toronto) and US scientists working on Inflammatory Bowel Disease compared

the sequence differences in a region of chromosome 5 in 200 families from Toronto. To their surprise, the 400 sequence differences that were observed appeared to be organized into simple patterns, which they called haplotype blocks. This structure which was then shown to exist across all human chromosomes, led to the concept of creating a comprehensive HapMap.

"Going from a concept to today's grand scientific accomplishment did not happen by chance. It took many nations, many experts from different fields, communities willing to donate DNA, new technologies, new public databases, and most of all people who believed in the project and were willing to invest in it", mentioned Dr. Martin Godbout, President & CEO of Genome Canada.

"Without the leadership and vision of both Genome Canada and Génome Québec, we could not have gone from a concept to published results in such a short time", added Paul L'Archevêque, President & CEO of Génome Québec .

The Government of Canada, through Genome Canada, and the Government of Québec, through Génome Québec, were the first in the world to approve the HapMap project and announce funding, in April, 2002. Final funding from the other participant countries came six months later, in October, 2002.

"Canada was the first country in this international consortium to approve the Hap Map research project and fund the work of Tom Hudson and his team through Genome Canada in 2002. We are very proud that they, along with their international partners, have made breakthrough progress in bringing to Canadians and to the rest of the world a scientific tool that will accelerate the process of finding disease-causing genes", said David L. Emerson, Minister of Industry. "*Congratulations to Tom Hudson and Bartha Knoppers – they are making Canada very proud and their work confirms Canada's leadership in the field of genomics research"*.

"*In our economic strategy tabled on October 13 - the Quebec Advantage - we identified genomics as one of our key pillar for achieving Quebec's prosperity",* continued Claude Béchard, Minister responsible for Economic Development, Innovation and Export Trade*. "Hap Map has definitely impacted Quebec's economic development and is contributing largely to its international recognition as a leader in the field of research and innovation". Congratulations to Tom and Bartha and thank you to Génome Québec, who played an important role in the development of this project".*

HAPMAP - BENEFITS FOR HUMAN HEALTH

The HapMap provides a tool that can be used in what are called association studies. For these studies, researchers will compare the haplotypes in individuals with a disease to the haplotypes of a comparable group of individuals without a disease (the controls). If a particular haplotype occurs more frequently in affected individuals compared with controls, a gene influencing the disease may be located within or near that haplotype.

"G*etting to know the 10-20 genetic markers that predispose to colon cancer could generate tests with a potential to effectively screen individuals at risk for colon cancer, leading to the removal of the tumours when they are still small and 95% curable*", Dr. Hudson pointed out. "*Getting to know the 10-20 genetic markers for diabetes can help find new treatments that are more specific to individual patients, and so on for all other diseases*".

Significant discoveries may take time to make their way to the clinic as future research is needed. However, the HapMap is a tool that will help researchers around the world significantly speed up the process.

ETHICAL CONSIDERATIONS AT THE HEART OF THE PROJECT

Researchers produced the HapMap using DNA from blood samples collected from 269 volunteers from widely distributed geographic regions: Yoruba in Ibadan, Nigeria; Japanese in Tokyo, Han Chinese in Beijing and Utah residents with ancestry from northern and western Europe. No medical or personal identifying information was obtained from the donors. However, the samples are identified by the population from which they were collected.

"*We have devoted much effort to making sure this project was done as ethically and transparently as possible. Following the precedent set by the Human Genome Project, we have weighed the ethical, legal and social implications of this research from the outset*," said Bartha M. Knoppers, Ph.D., L.L.B., of the University of Montreal. "*For example, we developed a very careful community engagement and sampling strategy to ensure that participants from all the different population groups could give full, informed consent. Still, we know our job is far from over and we stand ready to address whatever ethical, legal and social issues may arise in the future."*

As was the case with the data generated by the Human Genome Project, HapMap data are being made swiftly and freely available to public databases. Researchers can get access to this data through the HapMap Data Coordination Center (www.hapmap.org), the NIH-funded National Center for Biotechnology Information's dbSNP (http://www.ncbi.nlm.nih.gov/SNP/index.html) and the JSNP Database in Japan (http://snp.ims.u-tokyo.ac.jp/).

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Genome Canada is the primary funding and information resource relating to genomics and proteomics in Canada. Dedicated to developing and implementing a national strategy in genomics and proteomics research for the benefit of all Canadians, it has received $600 million from the Government of Canada to date, which, when combined with funding from other partners, totals $1.2 billion invested in 112 innovative research projects and sophisticated science and technology platforms.

Génome Québec is a non-profit organization whose mission is to mobilize the academic and industrial sectors around genomics research. The organization invests and manages funds totalling almost 300 millions dollars from the public and private sectors. It currently manages projects in six major sectors: human health, bioinformatics, ethics, the environment, forestry and agriculture. Since it was launched in 2000, it has generated more than 700 jobs and has led to creation of Montréal's McGill University and Génome Québec Innovation Centre, a world-class research centre than is helping position Québec on the international stage.

Tom Hudson and Bartha Knoppers are available for interviews.

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