

Study shows that hundreds of genetic variations determine the height of individuals

By Dr. Guillaume Lettre

Hundreds of common genetic variations in the human genome influence people's height, according to a major study published recently in the journal *Nature*. For this new study, close to 300 researchers from 200 institutions within the GIANT Consortium (Genetic Investigation of ANthropometric Traits) analyzed the DNA of 183,000 individuals with the objective of identifying genetic variations called SNPs (single nucleotide polymorphisms, pronounced "snips"), which are associated with height.

Height is a complex genetic trait in that it is influenced by an individual's genetic baggage and by his or her environment. In the specific case of height, an estimated 80 per cent of the variation observed in a population is attributable to variations

in DNA, with the remaining 20 per cent attributed to environmental factors such as nutrition. It is well known that height is determined mainly by heredity. What we had yet to demonstrate prior to this publication was the sheer number of genes involved in growth and height.

To identify genes that influence height, my colleagues and I from the United States, the United Kingdom and the Netherlands used a pangenomic association approach whereby close to 2.4 million SNPs throughout the genome were tested in 183,000 participants to assess the existence of a link with height. "Our team of researchers therefore identified 180 DNA regions, each of which explains a fraction of the normal variation in height in the population. This represents the most important step toward improving our knowledge of the genetic factors that explain why we are

all of different height," said Dr. Joel N. Hirschhorn, professor at Harvard University and the Broad Institute, and co-director of the study.

Because this was one of the largest studies on pangenomic association ever conducted owing to the sheer number of participants assessed, it was possible to test several new hypotheses. We demonstrated that the genes associated with complex human traits such as height come together in common signalling pathways. In other words, even if genetic variations that influence height are extremely numerous, we've observed that there is nothing random about their dispersion in the genome and that these variations are manifest in genes with similar biological functions. In doing so, our study responds to a major criticism against studies on pangenomic association, namely that the identification of

great numbers of SNPs linked to a given trait holds little scientific value, since they would usually be randomly dispersed in the genome. The study of biological signalling pathways identified by the GIANT Consortium thus opens up new possibilities, for example in endocrinology research.

In spite of the scope of the study, the 180 SNPs identified by researchers only explain some 10 per cent of hereditary variation in height – far from the 80% estimated. "Even if studies on pangenomic association are powerful tools, we're still far from fully understanding how differences between genomes influence traits such as height," cautions Professor Tim Frayling of the Peninsula Medical School and the University of Exeter, also a collaborator on the study. "We found that a trait such as height is far more complex than we anticipated and that

other strategies are necessary to understand how our genetic differences have an impact on individual characteristics such as height or predisposition to illness."

Indeed, contrary to so-called simple genetic diseases such as cystic fibrosis and sickle-cell anemia, where a single mutation causes the disease, height is influenced by the sum of hundreds of genetic variations that we all carry in our DNA. Knowing this, if we can understand the genetics of height, we can then apply this knowledge to other complex hereditary human diseases, notably cardiovascular diseases.

Dr. Guillaume Lettre is co-director of the study and researcher at the Montreal Heart Institute, as well as associate professor at the Université de Montréal.

Headwaters Health Care Centre Embraces Technology to Improve Patient Care

By Ann Cain

The message at the September Hospital board meeting at Headwater Health Care Centre was that electronic health records will improve patient care. "A province-wide electronic medical record or EMR will provide authorized access to patient information to help health-care providers make

better decisions," says Dr. Ken Derksen, Chief of Staff at Headwaters.

Two local physician groups in Orangeville are participating in a provincial pilot project for electronic medical records that will allow authorized physicians to access patient records online. "I was able to review the details of a patient's surgery that took place at 9 am within hours from my office," says

Derksen. In the past, the physician's office would have to wait for a paper report from the Hospital.

"By being early adopters of technology, the Hospital and area physicians are able to help improve patient care," says Cholly Boland, President and CEO, Headwaters Health Care Centre.

The Hospital currently participates in the Reach portal,

an online network that provides clinicians access to their patient's medical record from five area hospitals – including Headwaters Health Care Centre, William Osler Health System, The Credit Valley Hospital, Trillium Health Centre, and Halton Healthcare Services. Headwaters also participates in the province-wide electronic Child Health Network.

Headwaters will par-

ticipate in the World Health Organization (WHO) SAVE LIVES: Clean Your Hands global hand hygiene day on October 15th. The WHO's global call to action invites health-care workers, facilities and organizations throughout the world to campaign for improved hand hygiene to reduce health care associated infections. To date over 10,000 health-care facilities around the world have participated.

To view Headwaters' publicly reported infection rates visit www.headwatershealth.ca.

The Board approved the Hospital's new strategic directions. "Based on input and guidance from community partners, members of the Board of Directors, the management team, medical staff and Hospital employees we've developed a set of priorities that will help us change and grow to meet the health-care needs of the community we serve," says Boland. The Hospital has set a clear course that will provide safe and high-quality health care services; build partnerships to improve access to health care services; make Headwaters a workplace of choice for staff, physicians and volunteers; and demonstrate accountability in all we do.

Ann Cain is Manager of Public Relations and Communications at Headwaters Health Care Centre.

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