

Understanding the effects of genes on human traits

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Recent technological developments in genomics have revealed a large number of genetic influences on common complex diseases, such as diabetes, asthma, cancer or schizophrenia. However, discovering a genetic variant predisposing to a disease is only a first step. To apply this knowledge towards prevention or cure, including tailoring treatment to the patient's genetic profile –also known as personalized medicine – we need to know how this genetic variant affects health.

In a study published today in *Nature Communications*, Dr. Constantin Polychronakos from the Research Institute of the McGill University Health Centre (RI-MUHC), and collaborators from McGill University and The University of Texas, propose a novel approach for scanning the entire genome that will help us understand the effect of genes on human traits.

"This completely new methodology really opens up different ways of understanding how the genome affects the biology of the human body", says Dr. Polychronakos, corresponding author of the study and Director of the Endocrine Genetics Laboratory at the Montreal Children's Hospital and Professor in the Departments of Pediatrics and Human Genetics at McGill University.

DNA is the blueprint according to which our body is constructed and functions. Cells "read" this blueprint by transcribing the information into RNA, which is then used as a template to construct proteins – the body's building blocks. Genes are scanned based on the association of their



RNA with ribosomes – particles in which <u>protein synthesis</u> takes place.

"Until now, researchers have been focusing on the effects of disease-associated genomic variants on DNA-to-RNA transcription, instead of the challenging question of effects on RNA-to-<u>protein translation</u>," says Dr. Polychronakos. "Thanks to this methodology, we can now better understand the effect of genetic variants on translation of RNA to protein – a powerful way of developing biomarkers for personalized medicine and new therapies."

More information: www.nature.com/ncomms/2013/130 ... full/ncomms3260.html

Provided by McGill University Health Centre

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