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Frontiers in
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A NEW LIGHT SHED ON GENETIC REGULATION'S ROLE IN THE PREDISPOSITION TO COMMON DISEASES

An international team co-
led by Professor Emmanouil
Dermitzakis of the University
of Geneva has discovered
several thousands new
genetic variants impacting
gene expression some of
which are responsible for
predisposition to common
diseases, bringing closer to
the biological interpretation
of personal genomes.

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Why are some people more likely than others to suffer from diseases such as type 2 diabetes or heart dysfunction? It is partly due to genetics. Several genetic variants are indeed known to increase the risk of developing these common and multifactorial diseases. Among these differences, the majority concerns modifications in the genome that affect the level of expression of certain genes. With colleagues from King's College, Oxford University and the Wellcome Trust Sanger Institute, Emmanouil Dermitzakis, Louis-Jeantet Professor at the Faculty of Medicine of the University of Geneva (UNIGE), and his team discovered several thousands variants affecting the expression levels of genes, 358 of which seem to play a key role in the predisposition to certain diseases. This study is being published in the journal *Nature Genetics*.

Genetic disease risk differences between one individual and another are based on complex aetiology. Indeed, they may reflect differences in the genes themselves, or else differences at the heart of the regions involved in the regulation of these same genes.

By gene regulation we mean the decision that the cell makes as to when, where and at what level to activate or suppress the expression of a gene. In theory, two people could thus share a gene that is perfectly identical and yet show differences in their predisposition to a disease due to genetic differences concerning the regulation (overexpression or underexpression) of this same gene.

Numerous teams are currently trying to draw up a map of regions involved in gene regulation. Not an easy task, but invaluable since it allows us to understand all the genetic causes that can explain the predisposition to certain diseases.

Working with twins

Emmanouil Dermitzakis, Louis-Jeantet Professor at the Faculty of Medicine and member of the NCCR Frontiers in Genetics and the Institute of Genetics and Genomics of Geneva (IGE₃), is a specialist in what is called the genetics of complex traits. With an international team co-led by Professor Tim Spector (Kings College), Professor Mark McCarthy (Oxford University) and Dr. Panos Deloukas (Wellcome Trust Sanger Institute), he publishes a study highlighting thousands of these genetic variants that seem to explain individual differences in gene expression.

For this work, the researchers used samples of three different tissue types (adipose tissue, skin and blood cells) collected from more than 800 homozygotic (identical) and dizygotic twins.

«Identifying variants which control the activity of many genes is a greater challenge than we anticipated but we are developing appropriate tools to uncover them and understand their contribution to disease,» comments Panos Deloukas. «Modern human genetics combined with samples donated by the participants in studies such as TwinsUK is making great strides towards finding the genetic culprits behind human disease.»

The method researchers followed allowed them to uncover nearly 358 variants apparently involved in the predisposition to certain diseases including quantifying the contribution of rare regulatory variants that was previously not possible to identify by conventional analysis methods.

«Our work adds to those who have previously demonstrated the contribution of common variants in the predisposition to these disorders», explains Emmanouil Dermitzakis. «Thanks to this new level of knowledge, and if we manage to adapt this methodology to search for these variants in each individual, this will be a powerful tool to help prognose the predisposition to certain diseases and more importantly understand the biological aetiology in order to develop and employ individualized treatments.»

contact

Emmanouil Dermitzakis

022 379 54 83

emmanouil.dermitzakis@unige.ch

UNIVERSITY OF GENEVA

Press office

24 rue du Général-Dufour

CH-1211 Genève 4

Tél. 022 379 77 17

media@unige.ch

www.unige.ch