

## PRESS RELEASE

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# A deep look into population variation in gene activity provides key insight into cell functions and disease susceptibility

**A group of researchers led by the University of Geneva (UNIGE) report the most extensive and comprehensive study regarding impact of genetic variations on gene activity. This publication reveals how the unprecedented resolution for RNA sequencing allows the deep exploration of genetic effects into cellular processes. It uses novel technologies to obtain a detailed picture of how gene activity in blood cells differs among people and to describe the variations in DNA responsible for such differences. Results of this research have important implications for understanding genetic susceptibility to common and rare diseases as well as the basis of natural variation in human characters. The proceeds of this study are published in this week's edition of *Nature*.**

Our DNA contains the information needed to produce different proteins that are the building blocks and key components of cells. Instructions to synthesize such proteins are incorporated into DNA sequences defined as genes. This precious genetic material, however, never leaves the cell's stronghold nucleus. Instead, copies called RNA messengers are made and sent out to the tiny cell's protein factories located outside of the nucleus. Mutations in genes lead to a variation in the abundance or structure of these RNA messengers. This in turn is associated with changes in the protein content of cells, thereby influencing the way certain cellular processes are executed. Such DNA variations thus may contribute to differences in characteristics between individuals and may also cause or predispose to various diseases.

### **A NEW AND DETAILED REALITY EMERGES THROUGH CORRECTED VISION**

In order to elucidate the genetic nature of this variability, a collaboration of researchers from Switzerland, Spain and the UK, under the leadership of the Faculty of medicine of the UNIGE, have used novel technology to study RNA messengers. This cutting edge procedure, called "second generation sequencing", allows an unprecedented level of resolution to determine the abundance and structure of RNA messengers.

Previous studies merely informed us about rough individual differences in the quantity of RNA from each gene in the cell. Moreover, this was only the tip of the iceberg in terms of defining the exact molecular consequences. In this new research project, conducted using blood cells of 60 individuals of European descent, the scientists have obtained a much higher resolution of such processes that allow them to describe in detail the molecular differences in RNA among individuals. "For the first time we are able to "read" the sequence of almost all the RNA molecules in the cell and compare them among individuals" says Dr. Stephen Montgomery from the UNIGE. "As a computational biologist and geneticist, this is a dream come true".

The ability to read the RNA sequence in so many individuals is of unprecedented scale and brings the understanding of genetic variation to a new level. “If we consider Geneva to be a cell and all of the cars to be RNA messengers, then, until today we were only able to tell brands of cars apart. With this new methodology we are able to describe the colour, the size of the car, the size of the engine and other characteristics”, explains prof. Emmanouil Dermitzakis from the Genetics Department of the Faculty of medicine of the UNIGE and the *Frontiers in Genetics* program. “Obviously, such an increase in resolution provides us with a major advance in understanding of cellular processes and the fine detail of differences between humans”.

### **SHARPER INSIGHT WILL PAVE THE WAY TO TAILOR MADE TREATMENTS**

The results of this study, which has received financial support from the *Wellcome Trust*, the *Louis-Jeantet Foundation* and the Spanish Ministry of Science and *Consolider* program, have wider implications for human health. It is well known that DNA variants affecting gene activity may be responsible for disease susceptibility, primarily to common pathologies such as diabetes, cardiovascular diseases and asthma. The understanding of how such hitherto unknown subtle differences modulate gene expression is bound to accelerate the understanding of their mechanisms at the cellular level, enabling faster and more focused development of treatments.

As reported on line in this week’s edition of *Nature*, this study provides a framework towards the full understanding of the impact of genetic variations in cellular interactions, which has very important implications for the understanding of human diseases. ■

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