MedicalXpress

# New software traces origins of genetic disorders 20 times more accurately

October 23 2013

In a bioinformatics breakthrough, iMinds – STADIUS – KU Leuven researchers have successfully applied advanced artificial intelligence to enable the automated analysis of huge amounts of genetic data. Their new software suite, eXtasy, automatically generates the most likely cause of a given genetic disorder. The breakthrough directly impacts the treatment of millions of people with a hereditary disease.

At least 5% of the world population suffers from a rare, hereditary disease. Until recently, the origins of these genetic disorders could be correctly identified in only half of all cases. The lack of a conclusive diagnosis prolongs uncertainty for both the patients and their families and marks the beginning of a long search, and expensive, strenuous and even unnecessary treatments.

The introduction of new, cheaper technologies for deciphering the human genome held the promise of a quicker and more accurate diagnosis of hereditary diseases. But this proved challenging –particularly because of the huge amount and complexity of the data to be processed.

The genomes of two healthy individuals show no less than four million differences or mutations. Most of these mutations are harmless, but just one extra, malignant mutation can be enough to cause a genetic anomaly. Existing analytical methods simply do not have the means to reliably and quickly find this needle in the haystack.

The eXtasy software suite developed by iMinds – STADIUS – KU Leuven researchers drastically changes this outlook. The program can trace the origins of genetic disorders twenty times more accurately than existing analytical methods.

"eXtasy uses advanced artificial intelligence to combine whole sets of complex data into a global score that reflects how important a certain mutation is for a certain disease. This data can consist of networks of interacting proteins, but could also include scientific publications or even scores that estimate how harmful a mutation is for the protein in question," explains Prof. Dr. Yves Moreau of iMinds – STADIUS – KU Leuven. "In this way, we can detect disease-causing mutations twenty times more accurately, and provide patients and their families with a much faster and more conclusive diagnosis. We hope this can considerably improve and accelerate the treatment of millions of patients."

"Searching for disease-causing mutations in a patient's genome is really like searching for one specific needle in an enormous pile of needles. eXtasy allows us to formulate more accurate diagnoses, which in turn forms the basis of customized treatments," says Prof. Dr. Joris Vermeesch, who heads the Laboratory for Cytogenetics and Genome Research at KU Leuven.

"Practical applications of genome sequencing technology are possible only if variations can be interpreted accurately. eXtasy is a step in the right direction," adds Prof. Dr. Koen Devriendt, Head of the Department of Human Genetics at the University Hospital of Leuven.

**More information:** Sifrim, A. et al. eXtasy: variant prioritization by genomic data fusion, *Nature Methods* (2013). DOI: 10.1038/nmeth.2656

Provided by KU Leuven