Gene mapping project offers new clues about humans

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* 95 percent of human genetic differences cataloged

* Findings offer new tools for studying disease

* More discoveries yet to come, team says

By Julie Steenhuysen

CHICAGO, Oct 27 (Reuters) - Early data from the 1,000 Genomes Project, an international effort to build a detailed map of human genetic variation, is already offering new clues about human disease, including why some people are more severely affected by disease than others.

Dr. Evan Eichler of the University of Washington in Seattle and colleagues used findings from the pilot phase of the project to identify subtle differences among people in areas of the genome where DNA sequences are often repeated many times.

"I believe this is where we will make huge inroads in understanding the genetic basis of human disease," Eichler said.

His findings were released online on Wednesday in the journal Science to coincide with publication in Nature of the first data from the 1000 Genomes Project -- a public-private effort that aims to map to sequence and compare the genomes of 2,500 people from several different regions of the globe.

The data is expected to offer researchers a new set of tools to help understand the genetic causes of disease.

Eichler said differences in DNA sequences, known as copy number variation, have traditionally been difficult to compare, but they may explain why some people have certain diseases but not others.

And they may also explain why some people are more severely affected by disease than others.

Duplications of segments of the genome appear to have led to many of the qualities that distinguish human beings from other primate species, Eichler told a press briefing.

And they also may be linked with diseases like schizophrenia and autism, he said.

RAPID GAINS

The findings are possible because of advances in machines that sequence genetic information made by companies such as Illumina (ILMN.O: Quote, Profile, Research) and Roche (ROG.VX: Quote, Profile, Research).

These are allowing researchers to make rapid gains in their understanding of all of the different variations in human genes, researchers from the 1000 Genomes Project team said on Wednesday.

"Already, just in the pilot phase, we've identified over 15 million genetic differences by looking at 179 people. Over half of those differences haven't been seen before," said Dr. Richard Durbin, group leader of the Wellcome Trust Sanger Institute and co-chair of the 1000 Genomes Project.

"This is the largest catalogue of its kind, and having it in the public domain will help maximize the efficiency of human genetics research," Durbin told a news briefing.

The pilot includes data from more than 800 people, highlighting around 16 million variations in the genetic code that were previously unknown.

Durbin said researchers now know 95 percent of the genetic variants in any individual, which will help as teams try to assess the genetic causes of both rare and common genetic diseases.

The team found that each individual carries a significant number of potentially dangerous genetic mutations, with maybe 250 to 300 genes that have defective copies.

And they said the new map will allow researchers to look at the effects of recent evolution on the human genome. (Edited by David Storey)