

Catalogue of human genetic variation revealed

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A massive effort to study human genetic variation has ended with publication of two papers in the 30 September 2015 issue of the journal *Nature* that catalogue the genomes of more than 2,500 people representing 26 different populations across the globe.



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"This is the definitive resource on human variation. It will help us to ask questions such as why is one person susceptible to disease while another isn't," said Yale's Mark Gerstein, a major contributor to the seven-year old international effort to assess the effects of human genetic variation.

Gerstein's Yale group was instrumental in assessing the functional impact of different kinds of genetic variation. In particular, Gerstein and colleagues found each person possesses on average about 150 variants capable of disabling genes. Most of these variants were mutations of single nucleotides that comprise DNA but some involved large block deletions. While understanding the function of these mutations will help pinpoint causes of disease, researchers were also struck by the ability of humans to accommodate so much variation in their genomes. "I think it is amazing that each of us have such a large number of potentially high impact variations yet so many of us still are healthy and happy," Gerstein said.

For more information, see the following articles:

<http://www.nature.com/nature/journal/v526/n7571/full/nature15393.html>

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Source: [Yale University](#)