

As population exploded, more rare genes entered human genome

May 11 2012, By Krishna Ramanujan

(Medical Xpress) -- As the Earth's human population has skyrocketed since the rise of agriculture some 10,000 years ago -- to 7 billion people from a few million -- so, too, has the number of rare genetic variants.

Since about 2,000 years ago (fewer than 100 generations), the human population has experienced an explosive growth after 8,000 years of moderate exponential growth.

This recent accelerated growth has created more [genetic mutations](#) and rare gene variants, which may play a role in boosting the risks of complex diseases in which genes play a role, say Cornell researchers in the May 11 issue of the [journal Science](#).

The study found that when a large sample of 10,000 individuals was used in a model of human population growth, rare genetic variants were detected far more frequently than in previous studies. The new model also showed that the vast majority of these rare variants were due to mutations that arose in the past 2,500 years, coinciding with the explosive growth.

Previous models had used small samples with 62 or fewer individuals, or they did not account for the recent increased rate of population growth, and, therefore, they predicted lower numbers of rare genetic variants.

"It is expected that the number of rare alleles [variants of genes] would increase with population growth," said Alon Keinan, the Robert N.

Noyce Assistant Professor in Life Science and Technology in the Department of Biological Statistics and [Computational Biology](#), who co-authored the study with Andrew Clark, the Jacob Gould Schurman Professor of [Population Genetics](#) and Nancy and Peter Meinig Family Investigator.

"Our conclusions show just how huge of an effect recent explosive growth in humans could have in this regard," Keinan said, adding that the study also shows the importance of considering a very large sample in order to observe the rare variants in the population overall.

The research also has implications for models of genetic disease. Because many different genetic variants can contribute to complex diseases, more rare and new disease-related alleles can increase the risk of disease in different individuals, he said.

The study was funded by the National Institutes of Health and the Alfred P. Sloan Foundation.

More information: "Recent Explosive Human Population Growth Has Resulted in an Excess of Rare Genetic Variants," by A. Keinan; A.G. Clark at Cornell University in Ithaca, NY. *Science* (2012).

Provided by Cornell University

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