

Scientists develop method to visualize a genetic mutation

December 18 2018



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A team of scientists has developed a method that yields, for the first time, visualization of a gene amplifications and deletions known as copy number variants in single cells.

Significantly, the breakthrough, reported in the journal *PLoS Biology*, allows early detection of rare genetic events providing high resolution

analysis of the tempo of evolution. The method may provide a new way of studying mutations in pathogens and [human cancers](#).

"Evolution and disease are driven by mutational events in DNA," explains David Gresham, an associate professor in New York University's Department of Biology and the study's senior author. "However, in populations of [cells](#) these events currently cannot be identified until many cells contain the same mutation. Our method detects these rare events right after they have happened, allowing us to follow their trajectory as the population evolves."

The study, led by Stephanie Lauer, an NYU doctoral candidate, included researchers from NYU's Center for Genomics and Systems Biology and Stanford University.

Copy number variants, or CNVs, are a pervasive source of genetic variation and evolutionary adaptation. However, the dynamics and diversity of CNVs within evolving populations has been unclear.

In the *PLoS Biology* study, the scientists performed evolution experiments in the laboratory by subjecting cells to stressful environments. They studied the [molecular processes](#) underlying CNV formation and the dynamics with which they are generated, selected, and maintained.

The work made use of *Saccharomyces cerevisiae*, a microbe commonly used in biomedical and genomics research, and a fluorescent gene to monitor gene amplifications and deletions in individual cells.

They found that thousands of new CNVs are present in evolving populations in different selective conditions. Most of these mutations never become frequent in the [population](#), but a small number of lucky mutations ultimately survive.

The advance reported in this study shows that CNVs are a main driver of adaptive evolution in evolving microbial populations, the researchers note. As CNVs underlie [drug resistance](#) in some pathogens and tumor formation in some cancers, this advance provides a new way for studying the evolutionary processes that cause disease.

Provided by New York University

Citation: Scientists develop method to visualize a genetic mutation (2018, December 18)
retrieved 29 April 2024 from

<https://phys.org/news/2018-12-scientists-method-visualize-genetic-mutation.html>

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