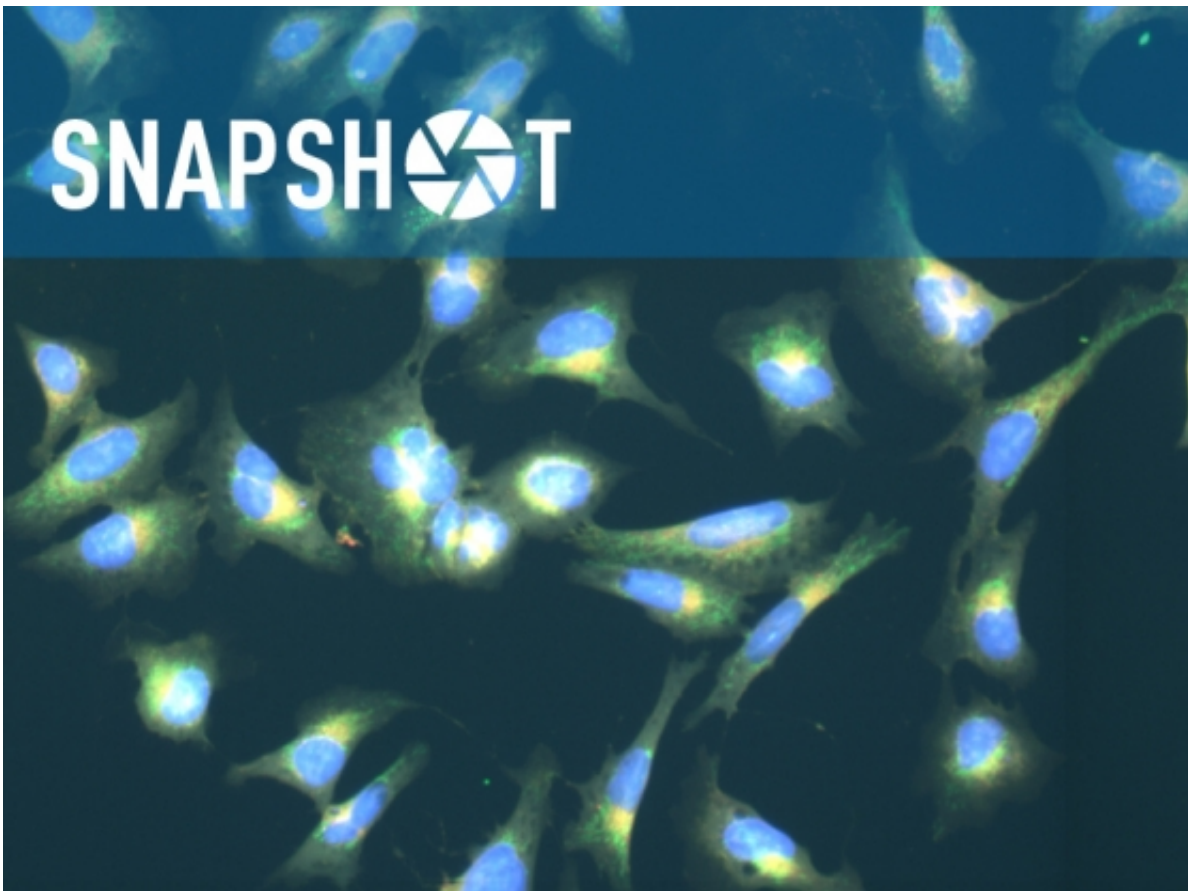


Cellular imaging technique reveal functions of uncharacterized genes or disease-associated gene variants

April 12 2017



Credit : Lauren Solomon, Broad Communications, and Broad Target Accelerator Team

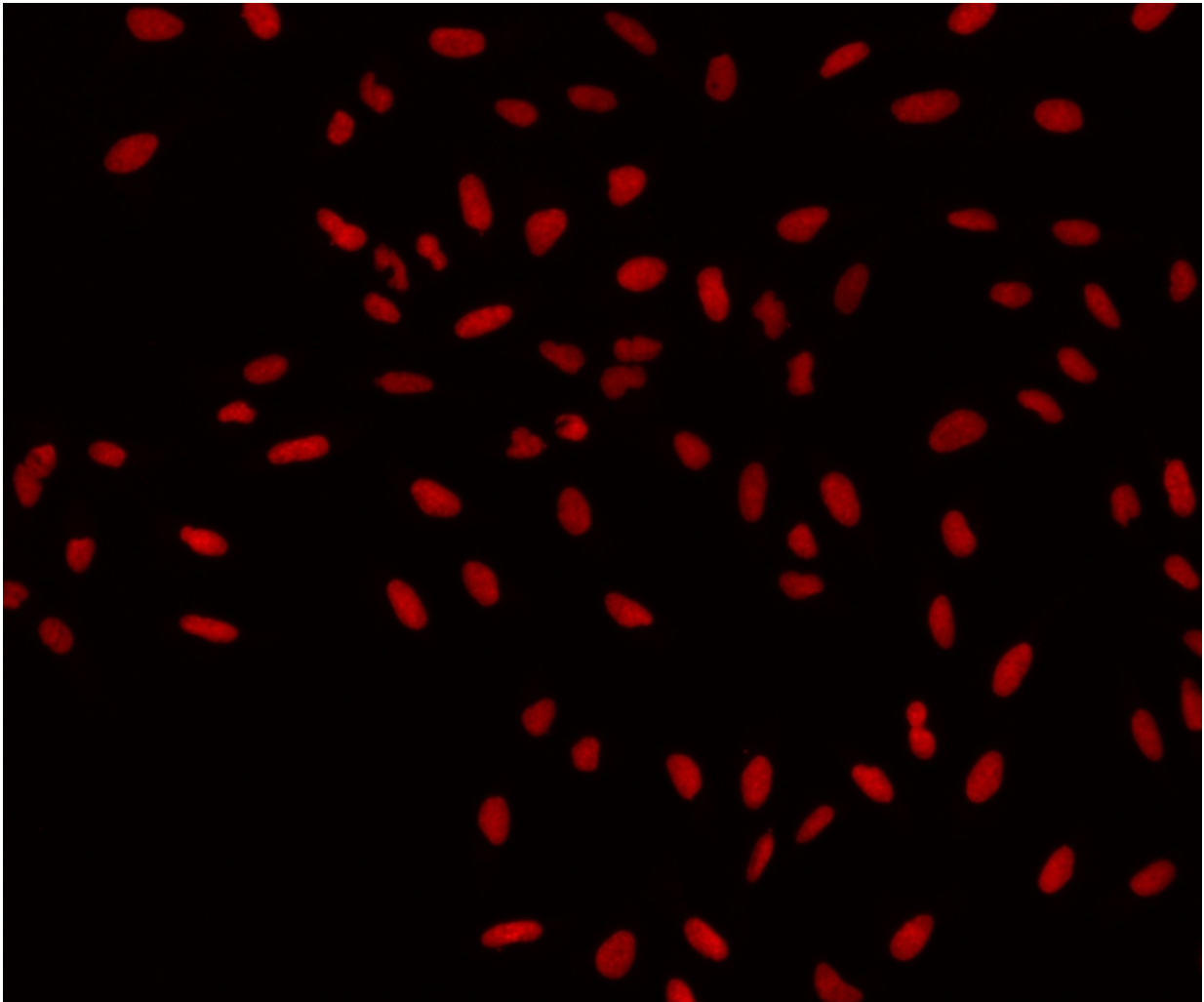
Scientists have used cells' visual appearance, or morphology, as a way to

help understand their state and identity essentially since the invention of the microscope. Now a research team led by Broad Institute Imaging Platform director Anne Carpenter and postdoctoral fellow Mohammad Rohban has shown that a high-throughput, computerized imaging technique for studying morphology, called Cell Painting, can provide insight into the cellular roles of genes or disease-linked gene alleles whose function or impact is unknown.

With Cell Painting, a technique developed at the Broad, researchers tag eight cellular components and organelles (actin, cytoplasmic RNA, [endoplasmic reticulum](#), Golgi apparatus, mitochondria, nucleus, nucleolus, and the plasma membrane) with fluorescent dyes. They then image the [cells](#) microscopically and computationally generate morphological profiles or signatures—measurable and reproducible changes in more than 1,500 features of the cells' appearance—that arise in response to a given perturbation.

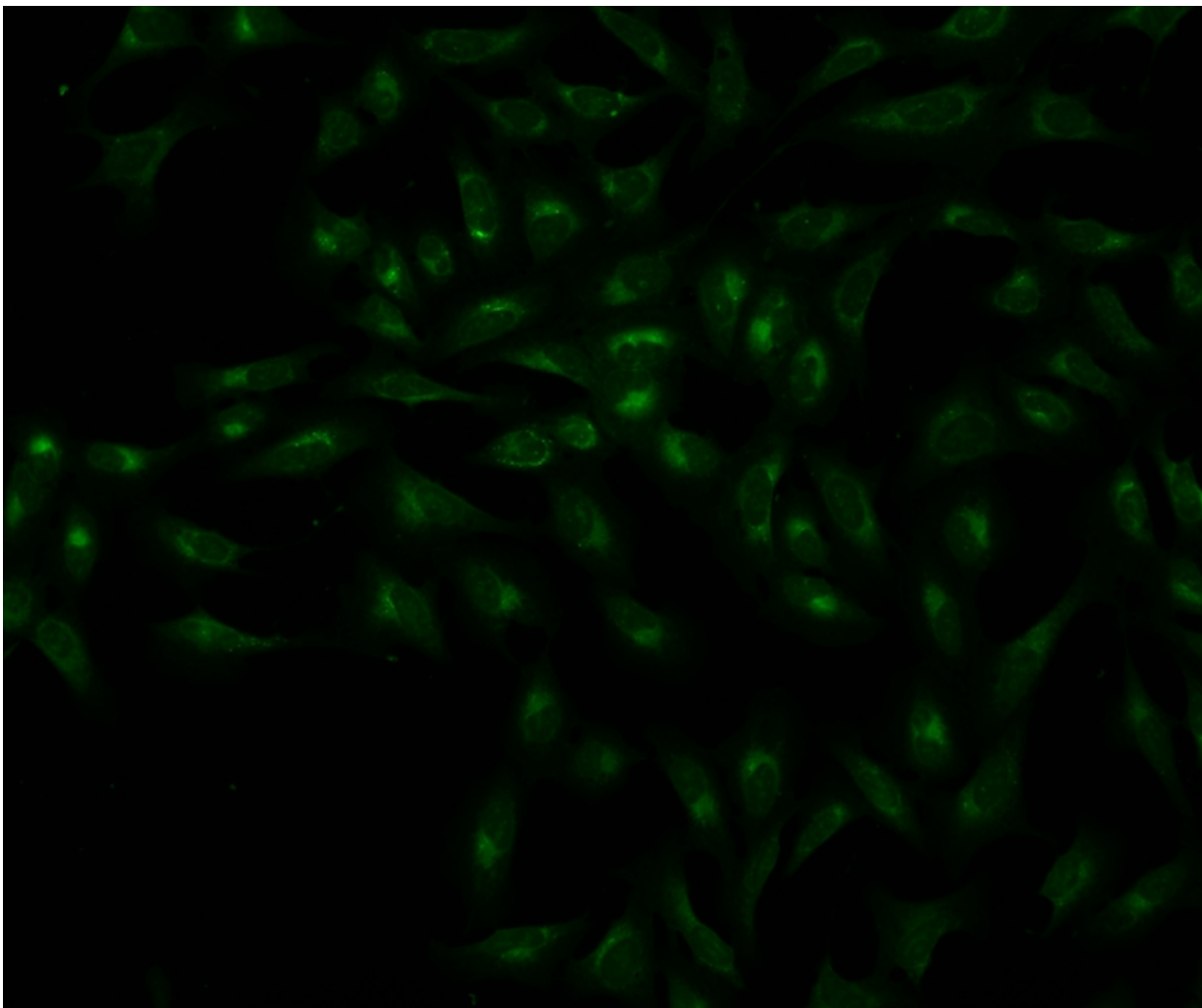
In the current study, published online in *eLife*, Rohban, Carpenter, and their colleagues overexpressed 220 [genes](#) one-by-one in [cultured cells](#), dyed the cells, and searched for signatures associated with each gene.

The team found that they could identify such signatures for half of the genes studied. Moreover, they found that they could group genes by function based on those signatures. For instance, overexpressing genes that reshape the cytoskeleton (the cell's structural scaffolding) resulted in similar signatures, as did overexpressing genes within or related to particular cell signaling pathways.

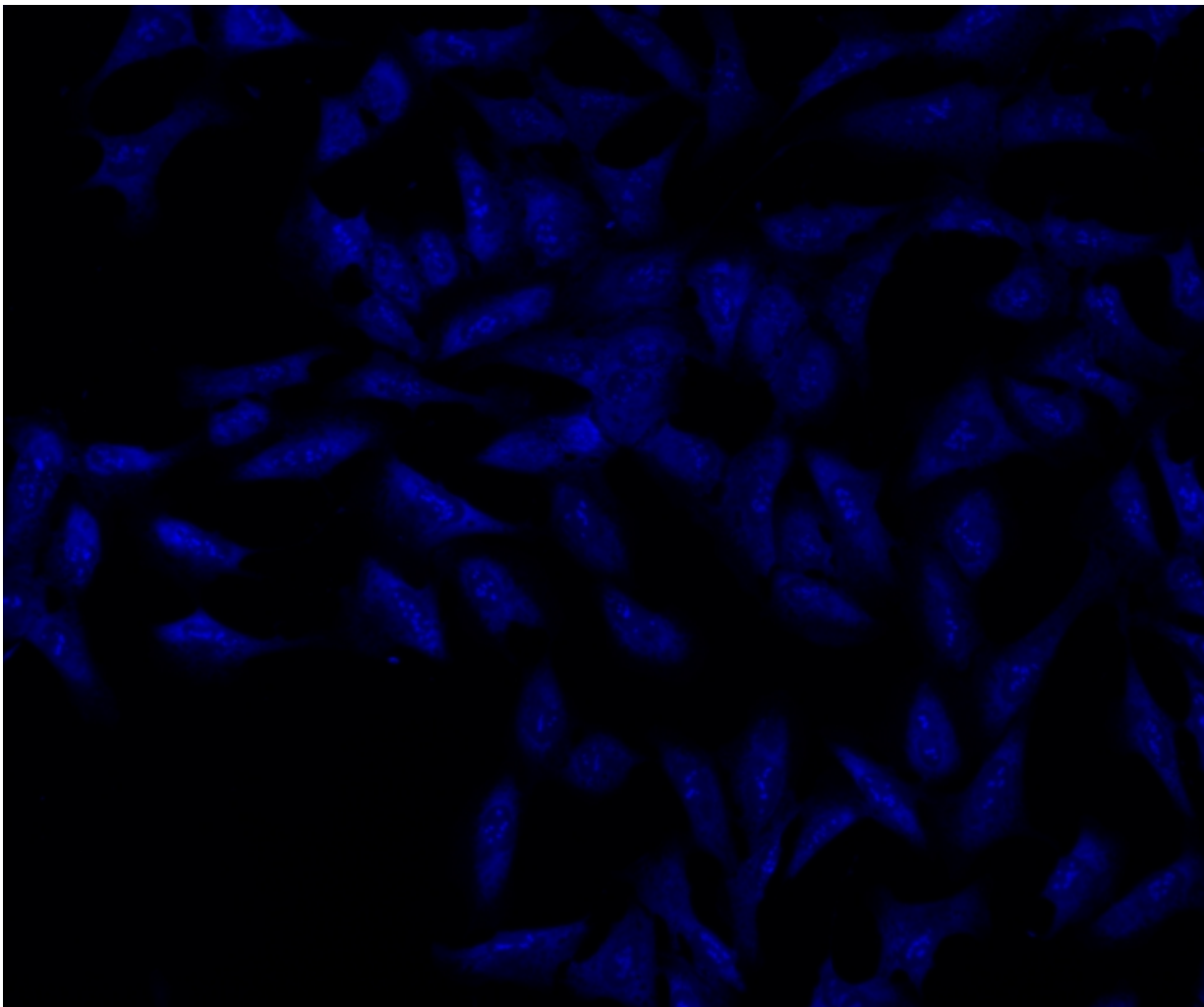


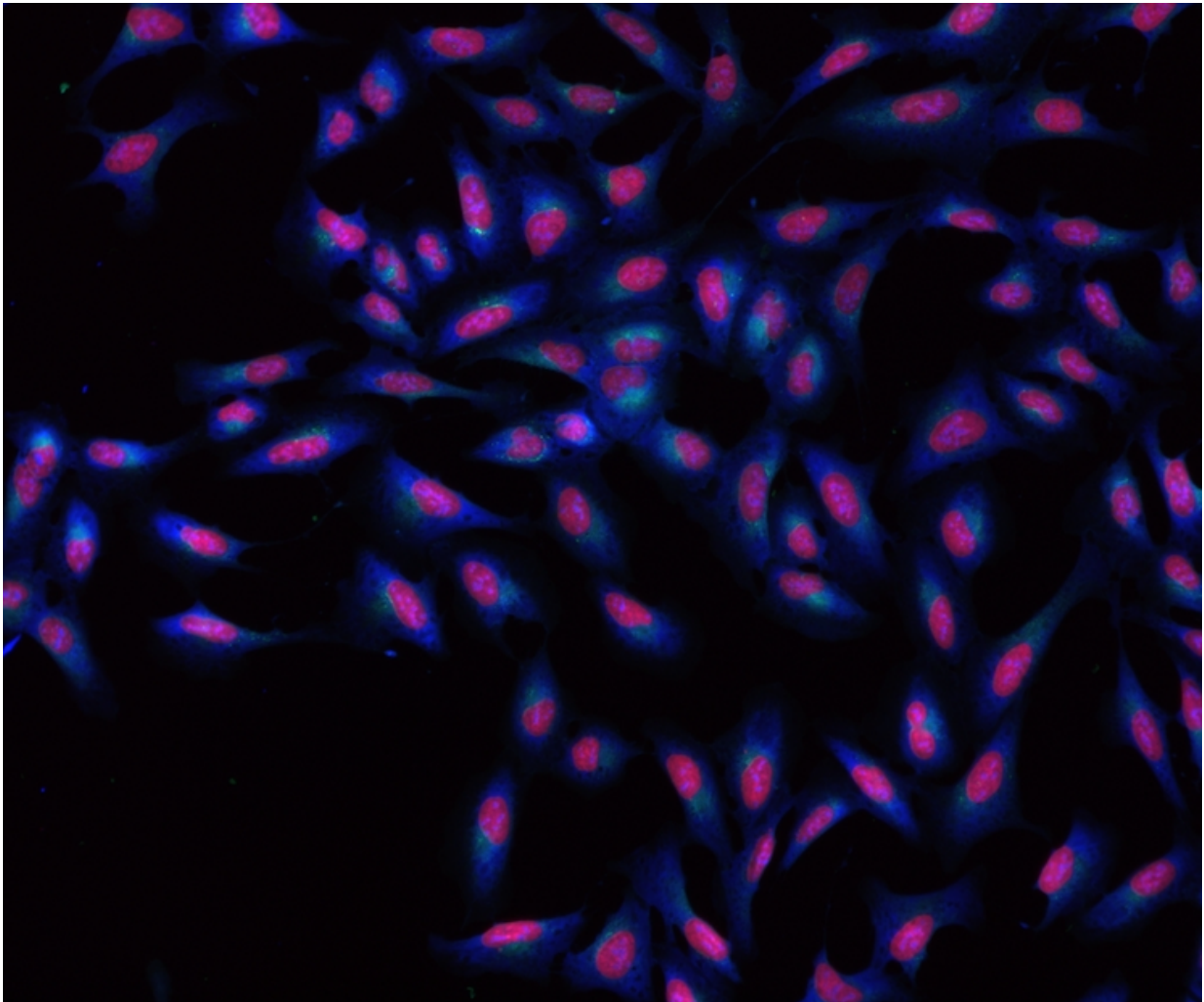
Credit: Broad Institute of MIT and Harvard

The findings suggest that Cell Painting may provide researchers with an inexpensive, high-throughput means to understanding the functions of uncharacterized genes—a category that currently includes more than 30 percent of the human genome. In addition, Cell Painting could help identify the biological mechanisms behind genetic variations flagged by genome-wide association and other large-scale genetic studies of traits and disease.



Credit: Broad Institute of MIT and Harvard





More information: Mohammad Hossein Rohban et al. Systematic morphological profiling of human gene and allele function via Cell Painting, *eLife* (2017). [DOI: 10.7554/eLife.24060](https://doi.org/10.7554/eLife.24060)

Provided by Broad Institute of MIT and Harvard

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