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Scientists develop new human stem cells with half a genome

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Scientists for the first time have generated a type of embryonic stem cell that carries a single copy of the human genome rather than the usual two, a development that could advance research in gene editing, genetic screening and regenerative medicine.

Derived from a female egg, the stem cells are the first human cells known to be capable of cell division with just one copy of the parent cell's genome, according to a study appearing on Wednesday in the journal Nature.

The breakthrough is expected to reduce the complexity of identifying genetic abnormalities, which in turn could advance understanding of many diseases, researchers said.

Human cells are considered diploid because they inherit two sets of chromosomes, 23 from the mother and 23 from the father. Reproductive egg and sperm cells are known as haploid because they contain a single set of chromosomes. They cannot divide to make more eggs and sperm.

"What is fundamentally new is we have cells that can divide and renew with a single genome. That is just unprecedented," said Dieter Egli of Columbia University Medical Center in New York, co-author of the study with Dr. Nissim Benvenisty of The Hebrew University of Jerusalem.

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The researchers, including scientists from The New York Stem Cell Foundation Research Institute, found the haploid stem cells capable of differentiating into many other cell types, such as nerve, heart, and pancreatic cells, while retaining a single set of chromosomes.

Sequencing of the human genome has yielded a wealth of new information about myriad genetic variations and how they interact with each other. But isolating and understanding specific gene abnormalities is challenging with diploid cells because they typically have a copy that is normal and serves as a backup.

"We have two genes of everything and if one is mutated the effect is not so obvious," said Egli in a telephone interview while vacationing in the French Alps. "Because these cells reduce the number of possible combinations and reduce the number of variance, it should be easier to get the answers."

A next logical step in the research, Egli said, is to modify these haploid stem cells either to introduce new disease variances or correct those that are already there.

"That should give us a way to better understand those many, many variances that are being identified in genome sequencing efforts that we think have something to do with disease."

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