iPSCORE: A Resource of 222 iPSC Lines Enabling Functional Characterization of Genetic Variation across a Variety of Cell Types.

Journal:	Stem Cell Reports
Publication Year:	2017
Authors:	Athanasia D Panopoulos, Matteo D'Antonio, Paola Benaglio, Roy Williams, Sherin I Hashem, Bernhard M Schuldt, Christopher DeBoever, Angelo D Arias, Melvin Garcia, Bradley C Nelson, Olivier Harismendy, David A Jakubosky, Margaret K R Donovan, William W Greenwald, KathyJean Farnam, Megan Cook, Victor Borja, Carl A Miller, Jonathan D Grinstein, Frauke Drees, Jonathan Okubo, Kenneth E Diffenderfer, Yuriko Hishida, Veronica Modesto, Carl T Dargitz, Rachel Feiring, Chang Zhao, Aitor Aguirre, Thomas J McGarry, Hiroko Matsui, He Li, Joaquin Reyna, Fangwen Rao, Daniel T O'Connor, Gene W Yeo, Sylvia M Evans, Neil C Chi, Kristen Jepsen, Naoki Nariai, Franz-Josef Muller, Lawrence S B Goldstein, Juan Carlos Izpisua Belmonte, Eric Adler, Jeanne F Loring, W Travis Berggren, Agnieszka D'Antonio-Chronowska, Erin N Smith, Kelly A Frazer
PubMed link:	28392216
Funding Grants:	The Stem Cell Matrix: a map of the molecular pathways that define pluripotent cells , World Stem Cell Summit, Collaborative Laboratory for Human Embryonic Stem Cell Research at Sanford-Burnham Medical Research Institute, User-friendly predictive molecular diagnostic assays for quality control of stem cell derivatives for transplantation and drug discovery

Public Summary:

Large-scale collections of induced pluripotent stem cells (iPSCs) could serve as powerful model systems for examining how genetic variation affects biology and disease. Here we describe the iPSCORE resource: a collection of systematically derived and characterized iPSC lines from 222 ethnically diverse individuals that allows for both familial and association-based genetic studies. The iPSCORE collection contains representative individuals for risk and non-risk alleles for 95% of SNPs associated with human phenotypes through genome-wide association studies. Our study demonstrates the utility of iPSCORE for examining how genetic variants influence molecular and physiological traits in iPSCs and derived cell lines.

Scientific Abstract:

Large-scale collections of induced pluripotent stem cells (iPSCs) could serve as powerful model systems for examining how genetic variation affects biology and disease. Here we describe the iPSCORE resource: a collection of systematically derived and characterized iPSC lines from 222 ethnically diverse individuals that allows for both familial and association-based genetic studies. iPSCORE lines are pluripotent with high genomic integrity (no or low numbers of somatic copy-number variants) as determined using high-throughput RNA-sequencing and genotyping arrays, respectively. Using iPSCs from a family of individuals, we show that iPSC-derived cardiomyocytes demonstrate gene expression patterns that cluster by genetic background, and can be used to examine variants associated with physiological and disease phenotypes. The iPSCORE collection contains representative individuals for risk and non-risk alleles for 95% of SNPs associated with human phenotypes through genome-wide association studies. Our study demonstrates the utility of iPSCORE for examining how genetic variants influence molecular and physiological traits in iPSCs and derived cell lines.