Introducing Human Induced Pluripotent Stem Cells (iPSCs) to the NHGRI Sample Repository for Human Genetic Research

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Background

The NHGRI Sample Repository for Human Genetic Research (NHGRI Repository) established by NHGRI in 2006 at the Coriell Institute for Medical Research facilitates studies of human genetic and genomic variation by establishing, characterizing and distributing a large (over 3,700) and diverse publicly available collection of renewable biospecimens from thousands of people living around the world. The NHGRI Repository includes lymphoblastoid cell lines and DNA samples associated with the International HapMap Project, 1000 Genomes Project and the Human Pangenome Reference Consortium. Participants that have generously donated to the NHGRI Repository consented to their samples and associated data being used for a wide range of general research uses and to public data sharing of largescale genomic data. Through the 1000 Genomes Project, the majority of this collection (N>3200) has been characterized with publicly available whole genome sequencing data and a subset of samples have been characterized with RNA-Seq data (N>460). In addition, web-based search tools of single variant genotypes, multiple variant genotypes, pharmacogenetic and HLA annotations, and lymphoblastoid cell line gene expression profiles can be accessed at https://www.coriell.org/1/Browse/Genomic-Data-Search. In late 2023, the NHGRI Repository incorporated human induced pluripotent stem cells (iPSCs) into the collection for the first time. To date, five iPSCs from the African American People living in St. Louis, Missouri collection have been added to the repository and are available to the research community. The iPSCs were reprogrammed using the Sendai Virus method and subjected to thorough quality control evaluation. Their inclusion in the NHGRI Repository represents a significant addition, providing a valuable resource for future research. Additional information about the NHGRI Repository can be found at https://catalog.coriell.org/NHGRI.

Table 1: NHGRI Repository Population Samples

Population	# Unique Subjects	Population	# Unique Subjects
African American People living in St. Louis, Missouri	9	Indian Telugu People living in the UK	118
African American People living in Southwest, USA	107	Japanese People living in Tokyo, Japan	131
African Caribbean People living in Barbados	120	Kinh People living in Ho Chi Minh City, Vietnam	124
Bengali People living in Bangladesh	144	Luhya People living in Webuye, Kenya	122
British people living in England and Scotland	100	Maasai People living in Kinyawa, Kenya	205
Chinese Dai People living in Xishuangbanna, China	102	Mende People living in Sierra Leone	128
Chinese People living in Metropolitan Denver, CO, USA	129	Mexican American People living in Los Angeles, CA, USA	104
Colombian People living in Medellín, Colombia	136	Peruvian People living in Lima, Peru	122
Esan People living in Nigeria	173	Puerto Rican People living in Puerto Rico	139
Finnish People living in Finland	103	Punjabi People living in Lahore, Pakistan	158
Gambian People living in Western Division, Mandinka	179	Sri Lankan Tamil People living in the UK	128
Gujarati People living in Houston, TX, USA	117	Toscani People living in Italia	117
Han Chinese People living in Beijing, China	162	Yoruba People living in Ibadan, Nigeria	229
Southern Han Chinese People living in China	163	CEPH Collection *	186
Iberian People living in Spain	157		

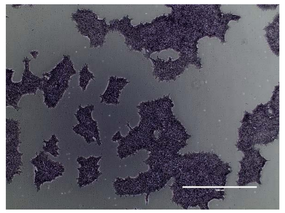
Quality Control of iPSCs

A

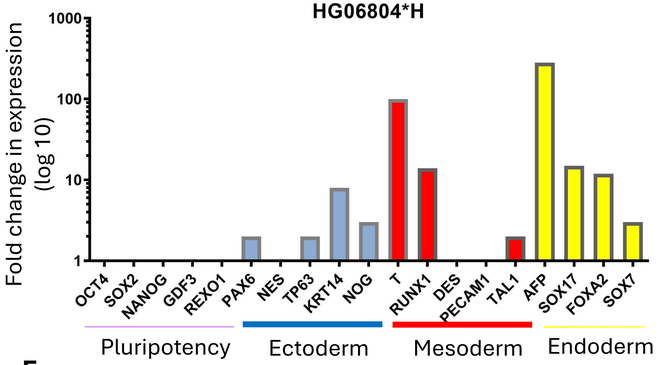
Cell line	Day	Average area (µm²)	Fold increase
HG06800	1	11,566	
	3	102,024	9
HG06801	1	30,675	
	3	289,990	9
HG06802	1	7,043	
	5	197,699	28
HG06803	1	10,471	
	4	253,646	24
HG06804	1	11,122	
	5	132,754	12

B

C



D



E

Gene	Fold change	Gene	Fold change	Gene	Fold change	Gene	Fold change
OCT4	0	PAX6	2	T	100	AFP	281
SOX2	0	NES	0	RUNX1	14	SOX17	15
NANOG	0	TP63	2	DES	1	FOXA2	12
GDF3	1	KRT14	8	PECAM1	0	SOX7	3
REXO1	0	NOG	3	TAL1	2		

Figure 1. A) Post-thaw viability. Colonies must double in diameter within 5 days. B) Cytogenomics analysis of HG06804 confirms normal karyotype. C) HG06804 iPSC colonies stained using the StemTAG Alkaline Phosphatase Staining Kit from CellBiolabs show alkaline phosphatase activity. Scale bar = 1000 µm. D-E) HG06804 Cells are differentiated by embryoid body (EB) formation to assess pluripotency. RNA is extracted and gene expression is measured by quantitative RT-PCR. Ct values are adjusted to the endogenous housekeeping gene GAPDH. Relative gene expression is shown as the fold difference in expression compared to undifferentiated cells. Expression of at least one gene per germ layer should increase by 2 fold or higher.

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