SUPPLEMENTARY MATERIAL
to the article A.S. Zueva, A.I. Shevchenko, S.P. Medvedev, E.A. Elisafenko, A.A. Sleptsov, M.S. Nazarenko, N.A. Tmoyan, S.M. Zakiyan, I.S. Zakharova "Isogenic induced pluripotent stem cell line ICGi036-A-1 from a patient with familial hypercholesterolaemia, derived by correcting a pathogenic variant of the gene *LDLR c.530C>T*"

Table S1. Oligonucleotides used in the study

Type of test	Locus	Product size, bp	Forward/reverse primer (5'–3')
c.530C>T correction analysis	LDLR_18720F/ LDLR_19091R	372	CTATAGAATGGGCTGGTGTTGG/ CTTAGGCAGTGGAACTCGAAGG
p.1054T>C correction analysis	LDLR_24218F/ LDLR_24476R	259	AGGGACCAACGAATGCTTGGA/ GCAGGTGGAATCTCATGAAACCC
Detection of episomal vectors	*EBNA-1*	196	TCCCGCAGATCTTCCTGCTCCTGTTCCACCG/ CTCAAAGGATCCGGGGTGATAACCATGGACGA
Mycoplasma detection	The ribosomal 16S RNA gene	280	GGGAGCAAACAGGATTAGATACCCT/ TGCACCATCTGTCACTCTGTTAACCTC
Reference gene for quantitative RT-PCR	*ACTB*	308	AGGCACCAGGGCGTGAT/ GATAGCACAGCCTGGATAGCA
	B2M	90	CACCCCCACTGAAAAAGATG/ ATATTAAAAAGCAAGCAAGCAGAA
Pluripotency markers for quantitative RT-PCR (RTqPCR)	*OCT4*	144	GGGAGATTGATAACTGGTGTGTT/ GTGTATATCCCAGGGTGATCCTC
	NANOG	116	TTTGTGGGCCTGAAGAAAACT/ AGGGCTGTCCTGAATAAGCAG
	SOX2	100	GCTTAGCCTCGTCGATGAAC/ AACCCCAAGATGCACAACTC

Table S2. Antibodies used in the study

Marker	Antibody	Dilution	Manufacturer, catalogue number	RRID
Markers of pluripotency	Mouse IgG2b anti-OCT3/4	1:50	Santa Cruz Cat # sc-5279	RRID: AB_628051
	Rabbit IgG anti-NANOG	1:200	ReproCELL Cat # RCAB003P	RRID: AB_2714012
	Rabbit IgG anti-SOX2	1:400	Cell Signaling Cat # 2748	RRID: AB_823640
	Mouse IgG3 anti-SSEA4	1:200	Abcam Cat # ab16287	RRID: AB_778073
Markers of differentiated derivatives	Mouse IgM anti-human PAX6	1:50	Santa Cruz Biotechnology Cat #sc-81649	RRID: AB_1127044
	Rabbit IgG anti-NF200	1:1,000	Sigma Cat # N4142	RRID: AB_477272
	Mouse IgG2a anti-αSMA	1:100	Dako Cat # M0851	RRID: AB_2223500
	Mouse IgG1 anti-human CD90	1:100	eBioscience Cat # 14-0909-82	RRID: AB_763535
	Mouse IgG1 anti-CK18	1:100	Abcam Cat # ab668	RRID: AB_305647
Secondary antibodies	Goat anti-Mouse IgG (H + L) Secondary Antibody, Alexa Fluor 568	1:400	Thermo Fisher Scientific Cat #A11004	RRID: AB_2534072
	Goat anti-Mouse IgM Heavy Chain Cross-Adsorbed Secondary Antibody, Alexa Fluor 568	1:400	Thermo Fisher Scientific Cat #A21043	RRID: AB_2535712
	Goat anti-Mouse IgG3 Cross-Adsorbed Secondary Antibody, Alexa Fluor 488	1:400	Thermo Fisher Scientific Cat #A21151	RRID: AB_2535784
	Goat anti-Mouse IgG1 Cross-Adsorbed Secondary Antibody, Alexa Fluor 568	1:400	Thermo Fisher Scientific Cat #A21124	RRID: AB_2535766
	Goat anti-Rabbit IgG (H + L) Cross-Adsorbed Secondary Antibody, Alexa Fluor 488	1:400	Thermo Fisher Scientific Cat #A11008	RRID: AB_143165

Table S3. Results of short tandem repeat analysis at 26 genomic loci

Locus	IPSC line name		
	ICGi036-A-1	ICGi036-A	Mononuclear cells
AMEL	X,X	X,X	X,X
SRY	–	–	–
D3S1358	16,16	16,16	16,16
TH01	9,9.3	9,9.3	9,9.3
D12S391	20,24	20,24	20,24
D5S818	11,12	11,12	11,12
TPOX	11,11	11,11	11,11
Yindel	–	–	–
D2S441	11,11	11,11	11,11
D7S820	8,10	8,10	8,10
D13S317	8,9	8,9	8,9
FGA	21,21.2	21,21.2	21,21.2
D22S1045	11,15	11,15	11,15
D18S51	16,16	16,16	16,16
D16S539	11,12	11,12	11,12
D8S1179	13,15	13,15	13,15
CSF1PO	10,12	10,12	10,12
D6S1043	11,16	11,16	11,16
vWA	16,18	16,18	16,18
D21S11	29,31.2	29,31.2	29,31.2
SE33	19,28.2	19,28.2	19,28.2
D10S1248	14,17	14,17	14,17
D1S1656	15.3,17.3	15.3,17.3	15.3,17.3
D19S433	13,14	13,14	13,14
D2S1338	20,24	20,24	20,24
DYS391	–	–	–

Table S4. Cell line ICGi036-A-1 passport

Unique stem cell line identifier	ICGi036-A-1
Alternative name for the cell line	FH 1.3.1S_130S5
Institution	Institute of Cytology and Genetics, Siberian Branch of the Russian Academy of Sciences, Novosibirsk, Russia
Contact information for inquiring about the cell line	Irina Zakharova, zakharova@bionet.nsc.ru
Cell type	iPSC
Species of organism	*Homo sapiens*
Additional information on the origin of the cell line	Age: 32 Gender: Female Ethnicity: unknown
Source of the original cells	Peripheral blood mononuclear cells
Reprogramming method	Non-integrating episomal plasmid vectors
Genetic modification type	Correction of single nucleotide substitution
Disease	OMIM 143890, familial hypercholesterolaemia type IIa
The gene/locus in which the genetic modification has occurred	*LDLR*: c.530C>T (p.Ser177Leu), ClinVar ID 3686; rs121908026; OMIM:606945.0004
Modification method/site-specific nuclease used	Base editing systems: xCas9(3.7)-ABE(7.10), addgene #108382; xCas9(3.7)-BE4, addgene #108381. Vector containing guide RNA spacer sequences designed as part of this work: pC9-sgRNA-mCherry. Software used to select guide RNAs is PnB Designer (https://fgcz-shiny.uzh.ch/PnBDesigner/)
Delivery method of programmable nucleases	Lipofection by Lipofectamine® 3000
Genetic material introduced into cells	xCas9(3.7)-ABE(7.10) is a plasmid containing adenine deaminase and nicase xCas9n; xCas9(3.7)-BE4 is a plasmid containing cytosine deaminase and xCas9n nicase; pC9-1054_2-mCherry, pC9-530-mCherry are plasmids with an integrated guide RNA spacer sequence
Method for analysing a modification made	PCR followed by Sanger sequencing of exon 4 in the *LDLR* gene
Method for assessing off-target activity	Site prediction software PnB Designer (https://fgcz-shiny.uzh.ch/PnBDesigner/)
Morphology	Rounded colonies, growing in a monolayer, with clear edges, in the shape corresponding to human pluripotent stem cells
Pluripotency	Validated in the test for the generation of derivatives of three germ layers resulting from spontaneous differentiation
Karyotype	46,XX
Contamination detection	No bacteria, fungi, mycoplasma detected
Application field	The cell line will be used to study the effect of a pathogenic genetic variant in the *LDLR* gene on the function of relevant differentiated derivatives; to study the contribution of the remaining uncorrected likely pathogenic substitution; to establish a system of isogenic cell lines for the development of test systems for potential drugs to treat FH
Cultivation system	Based on mitotically inactivated mouse embryonic fibroblasts
Cultivation medium	Based on DMEM/F12 medium and KnockOut SR serum replacement
Temperature, ℃	37
CO_2 concentration, %	5
O_2 concentration, %	Atmospheric (about 21 %)
Seeding method	Recombinant enzyme TrypLE™ Express (ThermoFisher Scientific)
Seeding rate	1:10
Cryopreservation	90 % FBS, 10 % DMSO
Storage conditions	Liquid nitrogen
Registry entry	https://hpscreg.eu/cell-line/ICGi036-A-1
Date of passporting/depositing	03.04.2024