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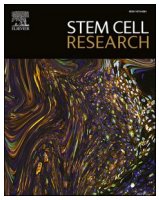

Lab Resource: Genetically-Modified Multiple Cell Lines

Generation of two induced pluripotent stem cell lines and the corresponding isogenic controls from Parkinson's disease patients carrying the heterozygous mutations c.815G > A (p.R272Q) or c.1348C > T (p.R450C) in the RHOT1 gene encoding Miro1

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A B S T R A C T

Fibroblasts from two Parkinson's disease (PD) patients carrying either the heterozygous mutation c.815G > A (Miro1 p.R272Q) or c.1348C > T (Miro1 p.R450C) in the RHOT1 gene, were converted into induced pluripotent stem cells (iPSCs) using RNA-based and episomal reprogramming, respectively. The corresponding isogenic gene-corrected lines have been generated using CRISPR/Cas9 technology. These two isogenic pairs will be used to study Miro1-related molecular mechanisms underlying neurodegeneration in relevant iPSC-derived neuronal models (e.g., midbrain dopaminergic neurons and astrocytes).

1. Resource table

Unique stem cell line identifier	*1. LCSBi009-A* *2. LCSBi009-A-1* *3. LCSBi010-A* *4. LCSBi010-A-1* *5. LCSBi010-A-2*
Alternative name(s) of stem cell line	*1. RHOT1_R272Q_clone1_PD* *2. RHOT1_R272Q_clone18_IsogenicControl* *3. RHOT1_R450C_clone5_PD* *4. RHOT1_R450C_clone6_IsogenicControl* *5. RHOT1_R450C_clone10_IsogenicControl*
Institution	*Luxembourg Centre for Systems Biomedicine (LCSB), University of Luxembourg, Luxembourg*
Contact information of the reported cell line distributor	*Prof. Rejko Krüger; rejko.krueger@uni.lu*
Type of cell line	*iPSCs*
Origin	*Human*
Additional origin info *(applicable for human ESC or iPSC)*	*1. Age at biopsy: 78 years* *Sex: female* *Ethnicity: European White* *2. As in 1.* *3. Age at biopsy: 54 years* *Sex: female* *Ethnicity: European White* *4. As in 3.* *5. As in 3*
Cell Source	*Dermal fibroblasts*

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(*continued*)

Method of reprogramming	*RHOT1_R272Q: RNA transfection* *RHOT1_R450C: nucleofection of episomal reprogramming vectors*
Clonality	*Clonal*
Evidence of the reprogramming transgene loss (including genomic copy if applicable)	*Loss of episomal reprogramming vectors (RHOT1_R450C only)*
The cell culture system used	*iPSCs were maintained under feeder-free conditions, on Matrigel-coated wells, in presence of Essential 8™ (E8) medium*
Type of the Genetic Modification	*Spontaneous point mutation (LCSBi009-A and LCSBi010-A)* *Gene correction (LCSBi009-A-1, LCSBi010-A-1 and LCSBi010-A-2)*
Associated disease	*Parkinson disease (OMIM #168600)*
Gene/locus	*RHOT1 (17q11.2), Gene ID: 55,288*
Method of modification / user-customisable nuclease (UCN) used, the resource used for design optimisation	*CRISPR/Cas9*
User-customisable nuclease (UCN) delivery method	*Nucleofection*
All double-stranded DNA genetic material molecules introduced into the cells	*Yamanaka reprogramming vectors, SpCas9 plasmid*
Analysis of the nuclease-targeted allele status	*Sanger Sequencing of the targeted alleles*
Method of the off-target nuclease activity prediction and surveillance	*The Benchling CRISPR design tool was used to assess on– and off-target potential of selected* guide RNA *sequences*
Descriptive name of the transgene	*N/A*
Eukaryotic selective agent resistance cassettes	*N/A*

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(*continued*)

(including inducible, gene/cell type-specific)	
Inducible/constitutive expression system details	*N/A*
Date archived/stock creation date	*01.12.2022*
Cell line repository/bank	https://hpscreg.eu/cell-line/LCSBi009-A https://hpscreg.eu/cell-line/LCSBi009-A-1 https://hpscreg.eu/cell-line/LCSBi010-A https://hpscreg.eu/cell-line/LCSBi010-A-1 https://hpscreg.eu/cell-line/LCSBi010-A-2
Ethical/GMO work approvals	*The Luxembourgish National Research Ethics Committee (CNER) provided ethical approval for the following project: "Disease modelling of Parkinson's disease using patient-derived fibroblasts and induced pluripotent stem cells" (DiMo-PD, CNER #201411/05).*
Addgene/public access repository recombinant DNA sources' disclaimers (if applicable)	*pCXLE-hUL (Addgene #27080), pCXLE-hSK (Addgene #27078), and pCXLE-hOct3/4 (Addgene #27076)*

2. Resource utility

We recently identified distinct PD patients carrying different heterozygous mutations in the *RHOT1* gene encoding Miro1 (Berenguer-Escuder et al., 2019, 2020; Grossmann et al., 2019; Grossmann et al., 2020). Here we report the generation of iPSCs – and the corresponding gene-corrected lines – from two carriers of different PD-associated Miro1 mutations, which can be used to investigate the impact of Miro1 deficiency on PD pathogenesis Table 1.

Table 1
Characterization and validation.

Classification (optional *italicized*)	**Test**	**Result**	**Data**
Morphology	Photography	Typical iPSC morphology	Fig. 1 panel A
Pluripotency status evidence for the described cell line	Immunocytochemistry	All iPSC clones display a nuclear localization of the pluripotency markers Nanog and Oct3/4	Fig. 1 panel C
	RT-qPCR	All iPSC clones express the pluripotency markers Nanog and Oct3/4	Fig. 1 panel D
Karyotype	-Illumina iScan S/N: N234 -Karyostat+	No aneuploidies detected	Supplementary Fig. 5
Genotyping for the desired genomic alteration/allelic status of the gene of interest	PCR across the edited site or targeted allele-specific PCR *[mandatory]*	Successful correction of the intended mutations Absence of large deletions in the region targeted by the gRNAs	Fig. 1 panel B Supplementary Fig. 4
	Evaluation of the - (homo-/hetero-/hemi-) zygous status of introduced genomic alteration(s)	*N/A*	*N/A*
	Transgene-specific PCR (when applicable)	*N/A*	*N/A*
Verification of the absence of random plasmid integration events	PCR/Southern *[mandatory]*	Loss of reprogramming vectors confirmed	Supplementary Fig. 1
Parental and modified cell line genetic identity evidence	STR analysis	Identical genotypes between parental and gene-edited lines	Supplementary Fig. 6
Mutagenesis / genetic modification outcome analysis	Sanger Sequencing	Confirmation of the homozygous corrections of the mutated alleles and absence of indel mutations	Fig. 1 panel B
	PCR-based analyses	*N/A*	*N/A*
	Southern Blot or WGS; western blotting (for knock-outs, KOs)	*N/A*	*N/A*
Off-target nuclease activity analysis	Targeted PCR and sequencing for the top 5 predicted off-targets located in genes *[*Optional but highly-recommended if Cas editing is used*]*	*N/A*	*N/A*
Specific pathogen-free status	Mycoplasma *[mandatory]*	Negative	Supplementary Fig. 7
Multilineage differentiation potential	Directed differentiation *[mandatory]*	Ability of all clones to differentiate into the three germ layers	Fig. 1 panel E
Donor screening (OPTIONAL)	HIV 1 + 2 Hepatitis B, Hepatitis C, HTLV 1 + 2	Negative	Supplementary Fig. 7
Genotype - additional histocompatibility info (OPTIONAL)	Blood group genotyping	N/A	N/A
	HLA tissue typing	N/A	N/A

3. Resource details

For the generation of parental iPSC lines (i.e., *LCSBi009-A* and *LCSBi010-A*), we reprogrammed fibroblasts of two PD patients carrying different *RHOT1* heterozygous mutations – the c.815G > A (p.R272Q) and the c.1348C > T (p.R450C) mutation – which are located in the first EF-hand (EF1) and in the C-terminal GTPase (cGTPase) domain of Miro1, respectively (Grossmann et al., 2019). Miro1 p.R272Q fibroblasts were reprogrammed into iPSCs using a virus-free and integration-free RNA-based approach, consisting in the transfection of a synthetic *in vitro* transcribed RNA replicon expressing the reprogramming factors in a single self-replicating polycistronic transcript. Miro1 p.R450C iPSCs

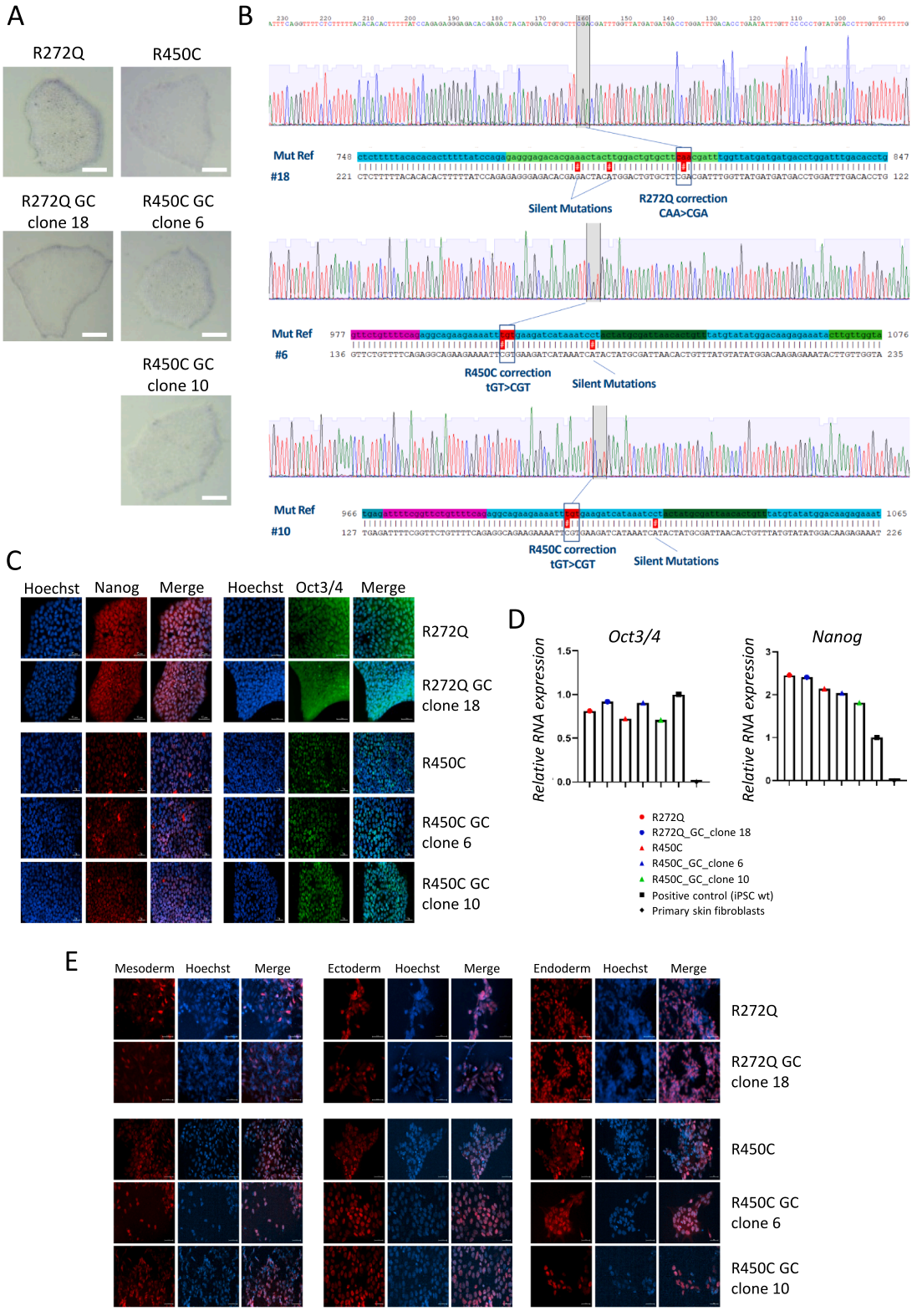

Fig. 1.

were established following electroporation of patient fibroblasts with episomal vectors expressing the four Yamanaka reprogramming factors. Clonal iPSC populations were identified by morphology, isolated and expanded (Fig. 1A, upper panels). The absence of plasmid integration in their genome was confirmed by PCR analysis (Supplementary Fig. 1). Isogenic, gene-corrected lines (i.e., *LCSBi009-A-1*, *LCSBi010-A-1* and *LCSBi010-A-2*) were generated by Axol Bioscience using a CRISPR/Cas9-mediated gene editing approach (Supplementary Figs. 2 and 3). For both Miro1 R272Q and Miro1 R450C gene correction, two distinct gRNAs targeting the intended mutation site were selected based on prior *in silico* analysis considering their distance from the target, predicted activity and off-target profile (Table 2). After testing their cutting efficiency in the corresponding parental iPSC lines, gRNAs of higher activity were chosen (C1917A-R272Q-g2 and C1917B-R450C-g1, respectively). Cas9-mediated re-cutting was prevented by introducing silent mutations in the gRNA binding sites present on the editing donor, consisting of a single-stranded oligodeoxynucleotide (ssODN) with homology arms flanking the target site (Table 2). Gene editing factors were introduced in the parental iPSC lines by nucleofection. Several iPSC colonies were screened by PCR and Sanger sequencing, confirming the successful corrections and the absence of indel mutations in one Miro1 p.R272Q (clone 18) and in two Miro1 p.R450C (clone 6 and 10) gene-edited clones (Table 2, Fig. 1B). qPCR analysis excluded the presence of large deletions in the region targeted by the respective gRNAs (Supplementary Fig. 4). The three gene-corrected lines, all displaying a typical iPSC morphology (Fig. 1A, lower panels), were then expanded and subjected to further characterization. Immunocytochemistry and RT-qPCR analyses confirmed that both patient-derived and gene-edited clones

Table 2
Reagent details.

Antibodies and stains used for immunocytochemistry/flow-cytometry			
	Antibody	**Dilution**	**Company Cat # and RRID**
Pluripotency Markers	*Rabbit anti Nanog*	*1:500*	*Abcam, Cat #: ab21624; RRID: AB_446437*
Pluripotency Markers	*Mouse anti Oct3/4*	*1:1000*	*Santa Cruz, Cat #: sc-5279; RRID: AB_628051*
Secondary antibody	*Alexa Fluor 488 Goat anti-Mouse IgG (H + L)*	*1.1000*	*Invitrogen, Cat #: A11029; RRID: AB_138404*
Secondary antibody	*Alexa Fluor 568 Goat anti-Rabbit IgG (H + L)*	*1:1000*	*Invitrogen, Cat #: A11036; RRID: AB_143011*
Mesoderm Marker	*Goat anti Brachyury*	*1:500*	*Human Pluripotent Stem Cell Functional Identification Kit (R&D Systems, Cat No.*
Endoderm Marker	*Goat anti SOX-17*	*1:500*	*SC027B)*
Ectoderm Marker	*Goat anti OTX2*	*1:500*	
Secondary antibody	*Alexa Fluor 647 Donkey anti Goat IgG (H + L)*	*1:1000*	*Invitrogen, Cat No. A21447; RRID: AB_141844*
Site-specific nuclease			
Nuclease information	*Nuclease type/version*	*SpCas9*	
Delivery method	*Nucleofection*	*Amaxa 4D*	
Selection/enrichment strategy	*Selection cassette(s), FACS*	*N/A*	
Primers and Oligonucleotides used in this study			
	Target	**Forward/Reverse primer (5′-3′)**	
Pluripotency Marker (RT-qPCR)	*OCT4-FAM*	*Hs00999632_g1 (Thermo Fisher Scientific)*	
Pluripotency Marker (RT-qPCR)	*NANOG-FAM*	*Hs02387400_g1 (Thermo Fisher Scientific)*	
Housekeeping gene (RT-qPCR)	*ACTB-VIC*	*Hs03023880_g1 (Thermo Fisher Scientific)*	
Potential random integration-detecting PCRs	*OriP fw*	*TTCCACGAGGGTAGTGAACC*	
Potential random integration-detecting PCRs	*OriP rv*	*TCGGGGGTGTTAGAGACAAC*	
gRNA Miro1 c.815G > A mutation	*RHOT1 exon 11*	*GAGGGAGACACGAAACTACT*	
gRNA Miro1 c.1348C > T mutation	*RHOT1 exon 16*	*ATTTTCGGTTCTGTTTTCAG*	
Miro1 c.815G > A ssODN repair template	*RHOT1 exon 11*	*GGAACAAATATTCAGGTGTCAAATCCAGGTCATCATCATAACC AAATCGTCGAAGCACAGTCCATGTAGTCTCGTGTCTCCCTCTCT GGATAAAAAGTGTGTGTAAAAAGAGAAAACCTGA*	
Miro1 c.1348C > T ssODN repair template	*RHOT1 exon 16*	*TCCATATACATAAACAGTGTTAATCGCATAGTAGGATTTATGA TCTTCACGAATTTTCTTCTGTCTCTGAAAACAGAACCGAAAAT CTCAATATCTGCAGTAACTAGTTTTCAA*	
Screening PCR Miro1 c.815G > A gene-corrected clones	*RHOT1 exon 11*	*GAGGCACAGTGAAGTTAATGAGATTG / CTTTAATCACAGCAATTTGGGAGGC*	
Screening PCR Miro1 c.1348C > T gene-corrected clones	*RHOT1 exon 16*	*ATGGTATAATGAAGATTGCCAGCAG / ACCACGCCTAGCCAAAAGAC*	
qPCR allelic status Miro1 c.815G > A gene-corrected clones	*RHOT1 intron 10*	*GGCCATTTCCTCTAGTGCTC / ATACGGATGCTAGCTTTGCATTG*	
qPCR allelic status Miro1 c.1348C > T gene-corrected clones	*RHOT1 intron 17*	*ACTTGTTGGTAAGAAATTCTGTGGC / CAGAAGTTCTACAGAGAGTCATTCA*	
Housekeeping gene (qPCR)	*GAPDH*	*TCTCCTGGAAGGGCTTCGTA / TAAGGCATGGCTGCAACTGA*	
Sanger sequencing Miro1 c.815G > A gene-corrected clones	*RHOT1 exon 11*	*GAGGCACAGTGAAGTTAATGAGATTG*	
Sanger sequencing Miro1 c.1348C > T gene-corrected clones	*RHOT1 exon 16*	*GAACAAATACATCTTTTATTTTAG*	

expressed the pluripotency markers OCT3/4 and Nanog (Fig. 1C and D). Moreover, they were all able to differentiate into the three germ layers (Fig. 1E). Finally, all iPSC lines displayed a normal karyotype (Supplementary Fig. 5), were genetically identical to the corresponding fibroblasts (Supplementary Fig. 6) and free of contamination by human pathogens (Supplementary Fig. 7).

4. Materials and methods

4.1. Cell culture and reprogramming

PD patient-derived fibroblasts were grown in DMEM medium containing 4.5 g/L D-glucose, 10% FBS and 1% Pen/Strep (Thermo Fisher Scientific). For the generation of Miro1 p.R272Q iPSCs, fibroblasts were reprogrammed using the Simplicon™ RNA Reprogramming Kit (Merck/EMD Millipore, Cat No. SCR550), according to the manufacturer's instructions. Briefly, cells were pre-treated with the B18R Protein (Millipore, Cat. No. GF156) to suppress the cellular interferon response, and then transfected with the VEE-OKS-iG RNA replicon expressing the reprogramming factors Oct4, Klf4, Sox2 and Glis1. To support iPSC induction, the TeSR™-E7™ reprogramming medium (STEMCELL Technologies, Cat No. 05914) was used until first iPSC colonies became visible; TeSR™-E7™ was then replaced by Essential 8 (E8) medium (Thermo Fisher Scientific). For the generation of Miro1 p.R450C iPSCs, episomal vectors expressing Yamanaka reprogramming factors, namely pCXLE-hUL (Addgene #27080), pCXLE-hSK (Addgene #27078) and pCXLE-hOct3/4 (Addgene #27076), were introduced into patient fibroblasts by electroporation, using the Amaxa Nucleofector II system (Lonza Bioscience) program B16. Transfected cells were plated on Matrigel-coated wells and fed every day with E8 medium. Undifferentiated iPSC colonies were identified by morphology, isolated and expanded; passages were performed every 3–4 days with 0.5 mM EDTA in PBS. Bright field images taken after expansion confirmed the typical iPSC colony morphology for all lines (Fig. 1A).

4.2. Reprogramming vector analysis

Loss of reprogramming vectors was confirmed by PCR using primers specific to the OriP region (Table 2, Supplementary Fig. 1), which is present in all plasmids encoding Yamanaka factors.

4.3. Gene editing

Gene-corrected iPSC lines were generated by Axol Bioscience using the CRISPR/Cas9 technology (Supplementary Fig. 2 and 3). Briefly, iPSCs were co-transfected with the SpCas9 plasmid, the specific gRNA and a single-stranded oligodeoxynucleotide (ssODN) repair template incorporating the corrected allele and also containing synonymous base changes that facilitate PCR screening and prevent Cas9-mediated re-cutting (Table 2, Supplementary Fig. 2 and 3). Cells were then plated on Matrigel-coated wells and fed with mTESR™1 (STEMCELL Technologies).

4.4. PCR and Sanger sequencing

Gene-edited iPSC clones were screened by PCR using the indicated primers (Table 2, Supplementary Fig. 4). PCR amplicons were then sequenced to confirm the successful gene editing and the absence of indel mutations (Table 2, Fig. 1B). Positive clones (clone 18 for Miro1 p.R272Q_GC, clones 6 and 10 for Miro1 p.R450C_GC) were finally expanded and gene correction further confirmed by PCR and sequencing. Location of the corrected alleles and silent mutations introduced are indicated by the hashtags highlighted in red (Fig. 1B).

4.5. Pluripotency assay

Expression of stemness markers was evaluated by both immunocytochemistry (ICC) and RT-qPCR analysis. For the ICC, iPSCs were plated on Matrigel-coated coverslips and fixed in PBS + 4% PFA for 15 min. Cells were then permeabilized and blocked for 1 h in PBS supplemented with 10% goat serum, 2% BSA and 0.4% Triton-X 100, followed by overnight incubation at 4 °C with Nanog and Oct3/4 primary antibodies (Table 2), diluted in 1% goat serum, 0.2% BSA and 0.1% Triton-X 100. The following day, cells were washed in PBS and incubated with the corresponding secondary antibodies (Table 2). Finally, nuclei were stained with Hoechst and images acquired using a Zeiss AxioObserverZ1 microscope (Carl Zeiss Microimaging GmBH). Scale bar = 50 μm (Fig. 1C).

For the RT-qPCR analysis, total RNA was extracted using the RNeasy Mini Kit (QIAGEN) and retrotranscribed using the Transcriptor High Fidelity cDNA Synthesis Kit (Roche). The resulting cDNAs were subjected to multiplex qPCR using the LightCycler® 480 Probes Master Kit (Roche) and the hydrolysis probes NANOG-FAM, OCT3/4-FAM and ACTB-VIC (Table 2). Relative RNA levels were expressed as fold change compared to a wild-type (wt) iPSC line that was used as positive control. cDNA obtained from primary skin fibroblasts was used as negative control (Fig. 1D).

4.6. Three germ layer differentiation

iPSCs were plated on Matrigel-coated coverslips two days before the *in vitro* differentiation procedure started. The Human Pluripotent Stem Cell Functional Identification Kit (R&D Systems, Cat No. SC027B) was used to verify iPSCs capacity to differentiate into the three germ layers. ICC of the ectodermal marker OTX2, the mesodermal marker Brachyury and the endodermal marker SOX17 was performed. Nuclei were stained with Hoechst and images were acquired using a Zeiss AxioObserverZ1 microscope. Scale bar = 50 μm (Fig. 1E).

4.7. Karyotyping and genetic identity

Genomic DNA was isolated from iPSCs and karyotype analysis was performed to exclude the presence of chromosomal aberrations. Karyotype of patient-derived iPSCs (Miro1 p.R272Q and p.R450C mutant lines) was assessed at Life&Brain Genomics (Bonn, Germany) using an Illumina iScan S/N: N234 array (Supplementary Fig. 5, upper panels). Karyotyping of gene-edited lines was performed by Life Technologies (Thermo Fisher Scientific, Madison, WI, USA) using a CytoScan HT-CMA 96F array for Karyostat + analysis (Supplementary Fig. 5, lower panels). Genetic identity between gene-edited and parental lines (both fibroblasts and mutant iPSCs) was confirmed by STR analysis (Cell Line Service, GmbH, Germany, Supplementary Fig. 6).

4.8. Human pathogens analysis

iPSCs were subjected to PCR analysis to exclude the presence of contamination by human pathogens, including Mycoplasma, Hepatitis, HIV and HTLV. All clones resulted negative (Supplementary Fig. 7).

Declaration of Competing Interest

The authors declare that they have no known competing financial interests or personal relationships that could have appeared to influence the work reported in this paper.

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Appendix A. Supplementary data

Supplementary data to this article can be found online at https://doi.org/10.1016/j.scr.2023.103145.

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