Support Information Table S1. iPSC disease modeling

Disease	Gene and Mutation	Phenotypes	Control	Reference
Neurological				
Alzheimer's disease	*APP* E603Δ *APP* V717L Sporadic	Intracellular Abeta oligomer accumulation. Increased Abeta 42/40 ratio. ER stress. Oxidative stress. Neuronal death without neurotrophic factors.	healthy iPSC	Cell Stem Cell. 2013 Apr 4;12(4):487-96.
Alzheimer's disease	*APP* duplication Sporadic	Increased Abeta40. Increased phospo-Tau (pT231). Increased active GSK-3beta.	healthy iPSC	Nature. 2012 Jan 25;482(7384):216-20.
Alzheimer's disease	*PS1* A246E *PS2* N141I	Increased Abeta 42/40 ratio.	healthy iPSC	Hum Mol Genet. 2011 Dec 1;20(23):4530-9.
Alzheimer's disease	*presenilin-1* *presenilin-2*	hiN cells from familial Alzheimer disease (FAD) patients with presenilin-1 or -2 mutations exhibit altered. Processing and localization of amyloid precursor protein (APP) and increased production of Aβ.	unaffected hiN cells	Cell. 2011 Aug 5;146(3):359-71.
Frontotemporal dimentia	*PGRN* S116X	Decreased PGRN. Activated MEK-MAPK pathwaty, comparing to PI3-Akt pathway.	healthy iPSCs (An age-matched subject, a clinically normal 64-year-old male with no mutations in GRN, MAPT, or C9ORF7)	Cell Rep. 2012 Oct 25;2(4):789-98.
Frontotemporal dementia	*C9ORF72* intron GGGGCC repeat expansions	GGGGCC repeats instability during neuronal differentiation. Form RNA foci. Gly-Pro dipeptide accumulation. Vulunerability to chloroquine and 3-methyladenine.	healthy iPSC	Acta Neuropathol. 2013 Sep;126(3):385-99.
Amyotrophic lateral sclerosis	*SOD1 L144F*	Sustained SOD1 mutation in iPSCs.	ESCs fibroblast from healthy control individual	Science 2008 Aug 29 321;(5893):1218-21.
Amyotrophic lateral sclerosis	*TARDBP G298S* *TARDBP M337V* *TARDBP Q343R*	Increasd TDP43 in insoble fraction short neurite length.	healthy iPSC	Sci Transl Med. 2012 Aug 1;4(145):145ra104.
Amyotrophic lateral sclerosis	*TARDBP* M337V	Increased TDP43 in soluble fraction neuronal death.	healthy iPSC	PNAS. 2012 Apr 10;109(15):5803-8.
Amyotrophic lateral sclerosis	*TARDBP* M337V	Increased TDP43 in soluble fraction astroglial cell death.	healthy iPSC	PNAS. 2013 Mar 19;110(12):4697-702.
Amyotrophic lateral sclerosis	*VAPB* P56S	Decreaed VAPB. Lack of VAPB positive inclusion. Increased VAPB instability by MG-132.	healthy iPSC ESC	Hum Mol Genet. 2011 Sep 15;20(18):3642-52.
Amyotrophic lateral sclerosis	sporadic	Increased TDP-43 aggregates. Phospho-Ser 409/410 TDP-43 aggregates.	healthy iPSC	Mol Cell Neurosci. 2013 Jul 25;56C:355-364.
Spinal muscular atrophy Parkinson disease Amyotrophic lateral sclerosis	*SMA Exon7 deletion*	n.e.	n.e.	J Reprod Dev. 2012;58(5):515-21. Epub 2012 May 24.
Spinal muscular atrophy type I	*SMA1*	Increase in Fas ligand-mediated apoptosis Increased caspase-8 and -3 activation.	unaffected iPSC	PLoS One. 2012;7(6):e39113. doi: 10.1371/journal.pone.0039113. Epub 2012 Jun 19.
Spinal muscular atrophy type I	*SMA1*	Short axon length, small growth cone, fewer and smaller motor endplates, decreased NMJ.	wild type iPSC heterozygous SMA iPSC genetically corrected patient-specific iPSC	Sci Transl Med. 2012 Dec 19;4(165):165ra162.
Spinal muscular atrophy	*SMN1*	SMA iPSC-derived astrocytes show morphological signs of activation, increased ERK1/2 activation and decreased GDNF production. SMA iPSC-derived astrocyte cultures have disrupted calcium signaling.	healthy iPSC	Glia. 2013 Sep;61(9):1418-28. doi: 10.1002/glia.22522. Epub 2013 Jul 10.
Spinal muscular atrophy	*SMN1* deletion	Reduced differentiation to motoneurons. Decreased number of gem bodies. Abnormal neurite outgrowth.	healthy iPSC	Stem Cells. 2011 Dec;29(12):2090-3.
Spinal muscular atrophy	*SMA* Exon7 deletion	Reduced differentiation to motoneurons. Decreased intranuclear GEM.	healthy iPSC	Nature. 2009 Jan 15;457(7227):277-80.
Spinal muscular atrophy	SMN1 deletion	Lack of response to VPA. Increased expression of the fatty acid translocase CD36 in VPA non-responders.	healthy iPSC ESC	Hum Mol Genet. 2013 Jan 15;22(2):398-407.
Spinobulbar muscular atrophy	*AR* CAG repeat	AR accumulation by Dihydrotestosterone.	healthy iPSC	J Biol Chem. 2013 Mar 22;288(12):8043-52.
Parkinson's disease	*LRRK2* G2019S	Decreased neurite length of dopaminergic neurons. TH positive cell death by H2O2. Increased cleaved-Caspase3 by 6-OHDA.	healthy iPSC	Cell Stem Cell. 2011 Mar 4;8(3):267-80.
Parkinson's disease	*LRRK2* G2019S Sporadic	Accumulation of alpha synuclein. Decreased neurite length of dopaminergic neurons increaed autophagy activity. Increased cleaved-Caspase3 postive neurons.	healthy iPSC	EMBO Mol Med. 2012 May;4(5):380-95.
Parkinson's disease	*LRRK2* G2019S	Decreased neurite length of dopaminergic neurons. Increased cleaved-Caspase3 by 6-OHDA or Rothenone. Increased Tua, phospho-Tau. Increased alpha-Synuclein. Increased phospho-ERK.	healthy iPSC	Cell Stem Cell. 2013 Mar 7;12(3):354-67.
Parkinson's disease	*LRRK2 G2019S*	Increased susceptibility to proteasomal stress.	healthy iPSC	Nature. 2012 Nov 22;491(7425):603-7.
Parkinson's disease	*PINK1* V170G	PARKIN translocation to mitochonoria and ubiquitination, depend on PINK1. Dysfunction of Mitophagy.	healthy iPSC	J Biol Chem. 2013 Jan 25;288(4):2223-37.
Parkinson's disease	*PINK1* Q456X *PINK1* V170G	PARKIN translocation to mitochonoria. Decreaed number of mitochondoria. Increased PGC-1alpha.	healthy iPSC	J Neurosci. 2011 Apr 20;31(16):5970-6.
Parkinson's disease	*PINK1* Q456X *LRRK2* R1441C *LRRK2* G2019S	Increaed ROS by Valinomycin. Respilatory dysfunction of mitochondria. Randomaized mitochodorial movement in Axon.	healthy iPSC	Sci Transl Med. 2012 Jul 4;4(141):141ra90.
Parkinson's disease	*PARKIN* ex2-4 deletion *PARKIN* ex6,7 deletion	Increased oxidative stress. Alpha-synuclein accmulation.	healthy iPSC	Mol Brain. 2012 Oct 6;5:35.
Parkinson's disease	*SNCA* triplication	Increased alpha-synuclein of cytosol or culture medium.	healthy iPSC	Nat Commun. 2011 Aug 23;2:440.
Parkinson's disease	*SNCA* triplication	Alpha-synuclein accmulation. Increased cleaved-Caspase3 by H2O2.	healthy iPSC	PLoS One. 2011;6(11):e26159.
Parkinson's disease	*Parkin*	Loss of parkin in human midbrain DA neurons greatly increases the transcription of monoamine oxidases and oxidative stress, significantly reduces DA uptake and increases spontaneous DA release.	healthy iPSC	Nat Commun. 2012 Feb 7;3:668
Parkinson's disease	*idiopathic*	-	non-PD iPSCs ESCs	Cell. 2009 Mar 6;136(5):964-77
Parkinson's disease	-	Mn exposure was associated with significantly higher ROS generation in NPC.	healthy iPSC	Neurotoxicology. 2012 Dec;33(6):1443-9.
Parkinson's disease	*α-Synuclein A53T (G209A)*	-	ESCs	Cell. 2011 Jul 22;146(2):318-31
Gaucher disease	*Gcase* N370S/84GG insertion	Alpha-synuclein accmulation. Increased alpha-synuclein in insoluble fraction.	healthy iPSC	Cell. 2011 Jul 8;146(1):37-52.
Gaucher disease	*GBA1*	Low acid-β-glucosidase activity.	wild-type iPSC isogenic iPSC	Hum Mol Genet. 2013 Feb 15;22(4):633-45
Gaucher disease	*GBA* N370S/N370S *GBA* L444P/RecNcil *GBA* L444P/L444P	Low glucocerebrosidase activity. Accumulated sphingolipids.	healthy iPSC ESC	Proc Natl Acad Sci U S A. 2012 Oct 30;109(44):18054-9.
Huntington disease	*HTT C*AG repeat	Neuronal death without neurotrophic factors.	healthy iPSC	PLoS Curr. 2010 Oct 28;2:RRN1193.
Huntington disease	*HTT C*AG repeat	Increased cleaved-Caspase3. Neuronal death without neurotrophic factors.	healthy iPSC	Cell Stem Cell. 2012 Aug 3;11(2):264-78.
Huntington disease	*HTT C*AG repeat	Increased cleaved-Caspase3. Neuronal death without neurotrophic factors.	healthy iPSC	Cell Stem Cell. 2012 Aug 3;11(2):253-63.
Huntington disease	*HTT CAG repeat*	Increased Susceptibility to HD Aggregate Formation in HD-iPSC	normal iPSC ESC	Stem Cells. 2012 Sep;30(9):2054-62
Huntington disease	*HTT CAG repeat*	Up-regulation of oxidative stress-related proteins in HD-iPSCs. Induction of DNA damage-mediated apoptosis in HD-iPSCs. Down-regulation of cytoskeleton-associated proteins in HD-iPSCs.	normal iPSC ESC	Biochem J. 2012 Sep 15;446(3):359-71
Huntington disease	*HTT CAG repeat*	Astrocytes generated from the HD-iPSCs with increased cytoplasmic vacuolation.	normal iPSC	Mol Brain. 2012 May 21;5:17.
Homozygous and heterozygous Huntington	*HTT CAG repeat*	Enhanced lysosomal activity.	wild type iPSC	Neurobiol Dis. 2012 Apr;46(1):41-51
Fragile X syndrome	*FMR1* CGG repeat expansion	Epigenetic modification of FMR1. Loss of FMR protein expression. Decreased neurite length.	healthy iPSC	PLoS One. 2011;6(10):e26203.
Fragile X syndrome	*FMR1*	Significant DNA methylation differences in the FMR1 promoter and 5' UTR.	iPSC (normal controls)	BMC Med Genet. 2013 Jan 29;14:18.

Fragile X-associated tremor/ataxia syndrome (FXTAS)	*FMR1*	iPS cell-derived neurons harboring the stably-active, expanded allele (EX-Xa) have reduced postsynaptic density protein 95 (PSD95) protein expression, synaptic puncta density and neurite length. EX-Xa eurons are functionally abnormal, with calcium transients of higher amplitude and increased frequency than for neurons harboring the normal-active allele. A sustained calcium elevation was found in the EX-Xa neurons after glutamate application.	iPSC derived from fibroblasts with an active normal allele	Hum Mol Genet. 2012 Sep 1;21(17):3795-805.
Familial dysautonomia	*IKBKAP* Exon20 skip	Increased abnormal splicing variant. Migration dysfunction.	healthy iPSC	Nature. 2009 Sep 17;461(7262):402-6.
Familial dysautonomia	*IKBKAP* Exon20 skip	Decreased IKBAP.	healthy iPSC	Nat Biotechnol. 2012 Dec;30(12):1244-8.
Olivopontocerebellar atrophy(OPCA)	*spinocerebellar ataxia 7 (SCA7)*	-	-	Tohoku J Exp Med. 2012;226(2):151-9.
Spinal cerebral ataxia type2	*ATXN2* hetero	Decreased ataxin-2 expression in neural stem cells or fibroblasts. Short-lived character in Time-lapsed neural growth assay.	healthy iPSC	J Mol Neurosci. 2013 Oct;51(2):237-48. doi: 10.1007/s12031-012-9930-2. Epub 2012 Dec 9
Spinal cerebral ataxia type3 (Machado-Joseph disease)	*ATXN3* CAG repeat	ATXN3 accumulation in insoluble faraction by glutamate stimulation.	healthy iPSC ESC	Nature. 2011 Nov 23;480(7378):543-6.
Friedreich Ataxia (FRDA)	*FXN*	GAA repeat expansion. Reduced FXN mRNA expression.	iPSC (control)	Stem Cell Rev. 2011 Sep;7(3):703-13.
Friedreich Ataxia (FRDA)	*FXN*	Repeat instability of GAA-TTC repeats in FXN. MSH2 highly expressed in pluripotent cells.	healthy iPSC SMA iPSC	Cell Stem Cell. 2010 Nov 5;7(5):631-7.
Friedreich ataxia (FRDA)	*hyperexpansion of the triplet-repeat sequence GAA.TTC within the first intron of the FXN gene*	GAA· TTC triplet-repeat expansion.	Unaffected iPSC	J Biol Chem. 2012 Aug 24;287(35):29861-72.
ataxia-telangiectasia (A-T)	*ATM*	Neural derivatives lack expression of ATM protein. Neural progenitor cells lacked substantial ATM phosphorylation caused by IR(2Gy). Cells from AT patients display decreased mitochondrial membrane potential.	wild type iPSC	Nat Commun. 2013;4:1824.
ataxia-telangiectasia (A-T)	*ATM*	Defective radiation-induced signaling, radiosensitivity, and cell cycle checkpoint defects. Abnormalities in DNA damage signaling pathways.	healthy iPSC SMA iPSC	Stem Cells Transl Med. 2012 Jul;1(7):523-35..
Dravet syndrome	*SCN1A* R1645*	Decreaed GABAergic firing, detected by patch-clamp record.	healthy iPSC	Mol Brain. 2013 May 2;6:19
Dravet syndrome	*SCN1A* F1415I SCN1A Q1923R	Increased paroxysmal deporalization shfit.	healthy iPSC	Hum Mol Genet. 2013 Jun 27
Dravet syndrome	*SCN1A*	DS patient-derived neurons show increased sodium currents in both bipolar-and pyramidal-shaped neurons.	unaffected iPSC	Ann Neurol. 2013 Jul;74(1):128-39.
Rett syndrome	*MECP2* T158M	Decreaed MeCP2. Normal differentiation to Neural precursors, but decreased number of matured TUJ1-positive neurons.	healthy iPSC ESC	Proc Natl Acad Sci U S A. 2011 Aug 23;108(34):14169-74.
Rett syndrome	*MECP2* Q244X *MECP2* R308C	Decreaesd MeCP2. Decreaed VGLUT1 puncta. Decreased cell body size. Decreaed spontanueous firing of neurons. Decreased EPSP firing.	healthy iPSC	Cell. 2010 Nov 12;143(4):527-39.
Rett syndrome	*MECP2*	Inactive X-chromosome in a onrandom pattern. RTT-hiPSC cell-derived neurons demonstrated a reduction in soma size.	isogenic iPS	Hum Mol Genet. 2011 Jun 1;20(11):2103-15.
Rett syndrome	*MECP2*	Smaller nuclear size.	isogenic iPSC	PLoS One. 2011;6(9):e25255. doi: 10.1371/journal.pone.0025255. Epub 2011 Sep 26.
HSV-1 encephalitis	*UNC93B1* c.1034_1037del4 *TLR3* c.1660C>T *TLR3* c.2236G>T	Impaired IFN-beta/lambda1 induction in dsRNA analogue or HSV1 infection.	healthy iPSC	Nature. 2012 Nov 29;491(7426):769-73.
Multiple sclerosis	Sporadic	n.e.	ESCs	Stem Cell Res. 2012 Mar;8(2):259-73.
Pelizaeus-Merzbacher disease	*a rare partial duplication of PLP1*	null PLP1 expression.	healthy iPSC	J Hum Genet. 2012 Sep;57(9):580-6.
mitochondrial encephalomyopathy with lactic acidosis and stroke-like episodes (MELAS)	*ND5 subunit of complex I, G13513A*	Reduction in mitochondrial density and oxygen consumption. Consistent with the cellular mosaicism of the original patient-derived fibroblasts, the MELAS-iPSC clones contained a similar range of mtDNA heteroplasmy of the disease-causing mutation with identical profiles in the remaining mtDNA.	wild type iPSC	Stem Cells. 2013 Jul;31(7):1298-308.
Moyamoya disease (MMD)	*RNF213 R4810K polymorphism*	Angiogenic activities of iPSC-derived vascular endothelial cells (iPSECs) from patients and carriers were lower than from wild-type subjects. Gene expression profiles in iPSECs showed that Securin was down-regulated in carriers and patients.	wild type iPSC	Biochem Biophys Res Commun. 2013 Aug 16;438(1):13-9.
Glioblastoma	-	Widespread resetting of DNA methylation, but malignant cellular behavior in the neural leneage.	iPSC from the neural stem cell line CB660 (normal control) ESCs	Genes Dev. 2013 Mar 15;27(6):654-69.
childhood cerebral ALD (CCALD), adrenomyeloneuropathy (AMN)	*ABCD1*	Abnormal accumulation of VLCFA in oligodendrocytes.	wild-type iPSC	Ann Neurol. 2011 Sep;70(3):402-9.
Muscular				
Duchenne muscular dystrophy	*DMD* del exons 4–43	n.e.	genetically corrected patient-specific iPSC	Mol Ther. 2010 Feb;18(2):386-93.
Facioscapulohumeral Dystrophy (FSHD)	*deletion of a subset of the D4Z4 macrosatellite repeat*	Expression of full-length DUX4 persists in differentiated FSHD iPS cells.	control iPSC	PLoS Genet. 2010 Oct 28;6(10):e1001181.
limb-girdle muscular dystrophy 2D (LGMD2D)	*SGCA*	No SGCA expression in a differentiated myotube.	isogenic iPSC	Sci Transl Med. 2012 Jun 27;4(140):140ra89.
Myotonic dystrophy type 1 (DM1) Huntington disease	*DMPK1*	Different CTG·CAG repeat lengths among 41 iPSC clones. The correlation between repeat length and expansion rate identified the interval between 57 and 126 repeats as being an important length threshold where expansion rates dramatically increased. Longer repeats showed faster triplet-repeat expansion.	isogenic fibroblasts	Hum Mol Genet. 2013 Aug 28. doi: 10.1093/hmg/ddt386
Miyoshi myopathy	*DYSF* not discribed	Decreased expression of dysferlin. Extensive uptake of FM1-43, indicating defective membrane repair.	healthy iPSC	PLoS One. 2013 Apr 23;8(4):e61540.
Supravalvular aortic stenosis	*ELN* exon9 ins GTAT (premature termination)	Filament bundles of smooth muscle actin down. ELN down.	healthy iPSC	Circulation. 2012 Oct 2;126(14):1695-704.
Ocular				
Gyrate atrophy	*OAT* A226V	n.e.	genetically corrected patiet-specific iPSC	Proc Natl Acad Sci U S A. 2011 Apr 19;108(16):6537-42.
Retinitis pigmentosa (RP)	*RP1* 721Lfs711X *RP1* 721Lfs711X *RP9* H137L *PRPH2* W316G *RHO* G188R	8-OHdG upregulation. in vitro degeneration (Cleaved Casplase-3) up.	healthy iPSC	PLoS One. 2011 Feb 10;6(2):e17084.
Retinitis pigmentosa (RP)	*MAK (male germ cell-associated kinase)*	In the proband with the Alu insertion, the developmental switch to the MAK transcript bearing exons 9 and 12 did not occur.	healthy iPSC non-MAK-associated RP iPSC	Proc Natl Acad Sci U S A. 2011 Aug 23;108(34):E569-76.
Retinitis pigmentosa (RP)	*RHO*	In the differentiated rod cells, diffused distribution of RHO protein in cytoplasm and expressions of endoplasmic reticulum (ER) stress markers strongly indicated the involvement of ER stress. The rod cell numbers decreased significantly after successive culture.	wild type iPSC	Stem Cells Transl Med. 2012 Jun;1(6):503-9.
Autosomal recessive RP	*USH2A*	Analysis of the USH2A transcripts of these cells revealed that one of the patient's mutations causes exonification of intron 40, a translation frameshift and a premature stop codon. Western blotting revealed upregulation of GRP78 and GRP94.	hiPSC (normal)	Elife. 2013 Aug 27;2:e00824. doi: 10.7554/eLife.00824.
Best disease (BD)	*BEST1*	RPE from mutant hiPSCs displayed disrupted fluid flux and increased accrual of autofluorescent material after long-term POS feeding. RHODOPSIN degradation after POS feeding was delayed in BD hiPSC-RPE.	unaffected sibling iPSC	Hum Mol Genet. 2013 Feb 1;22(3):593-607.
Catalact	sporadic	Conexin43 up.	ESCs	PLoS One. 2012;7(3):e32612.
Haemotological				
alpha-thalassemia	*SEA, FIL*	globin chain imbalance	isogenic iPSC ESC	Blood. 2012 Nov 8;120(19):3906-14.
beta-thalassemia	*codon 41/42 4-bp(CTTT) deletion*	-	-	Proc Natl Acad Sci U S A. 2009 Jun 16;106(24):9826-30.

beta-thalassemia	*beta-thalassemia*	-	ESC	J Reprod Dev. 2012;58(4):404-9.
beta-thalassemia	*HBB*	n.e.	n.a.	Cell Res. 2009 Sep;19(9):1120-3.
beta-thalassemia	*HBB*	n.e.	ESCs	Cell Res. 2012 Apr;22(4):637-48.
beta-thalassemia	*HBB*	n.e.	n.a.	Nat Biotechnol. 2011 Jan;29(1):73-8.
Sickle cell disease	*HBB*	n.e.	isogenic iPSC	Blood. 2011 Oct 27;118(17):4599-608
Sickle cell disease	*HBB*	n.e.	isogenic iPSC	Stem Cells. 2011 Nov;29(11):1717-26.
Sickle cell disease	*HBB*	-	isogenic iPSC	Cell Res. 2011 Dec;21(12):1740-4.
Aplastic anemia hypocellular bome marrow	*TERT* *TERC*	Defective telomere elongation and hematopoiesis.	healthy iPSC	J Clin Invest. 2013 May 1;123(5):1952-63.
Fanconi Anemia	*FANCC*	Much higher frequency of chromosomal abnormalities, inability to generate teratoma composed of all three germ layers in vivo.	hiPSC/ESC	Stem Cells. 2013 May;31(5):1022-9.
Fanconi Anemia	*Fanca*	Reduction in reprogramming efficiency. Reprogramming activates the FA pathway and induces DNA damage.	iPSC (normal)	Blood. 2012 Jun 7;119(23):5449-57.
Fanconi anemia	FANCA FANCD2	Failure to relocate to UVC-radiation-induced stalled replication forks.	healthy iPSC ESC	Nature. 2009 Jul 2;460(7251):53-9.
Myeloproliferative disorders	*JAK2-V617F*	Increased erythropoiesis PV-unique gene expression pattern.	normal iPSC	Blood. 2009 Dec 24;114(27):5473-80.
Chronic myeloid leukemia	Philadelphia translocation	Reprogramming of KBM7 cell line results in escape from oncogene addiction, resistant to imanitib.	healthy iPSC	Blood. 2010 May 20;115(20):4039-42.
Chronic myeloid leukemia	Philadelphia translocation (BCR-ABL)	Hematopoietic cells from chronic myeloid leukemia were sensitive to imanitib, iPS cells were not.	healthy iPSC ESC	Blood. 2012 Jun 28;119(26):6234-42.
JMML	*PTPN11 (p.E76K)*	Increased proliferative capacity of myeloid cells. Constitutive activation of GM-CSF. Enhanced STAT5/ERK phosphorylation.	wild-type iPSC	Blood. 2013 Jun 13;121(24):4925-9.
CINCA syndrome	*NLRP3* Y570C	Increased secretion of IL-1beta.	healthy iPSC	Blood. 2012 Aug 9;120(6):1299-308.
X-linked chronic granulomatous disease	*CYBB* L153R	Lack ROS production.	healthy iPSC	Blood. 2011 May 26;117(21):5561-72.
chronic granulomatous disease	p47 phox gp91phox	Lack ROS production. Similar cytokine profile to those of the control iPSC.	healthy iPSC ESC	Stem Cells. 2012 Apr;30(4):599-611.
Congenital amegakaryocytic thrombocytopenia	*MPL* Q186X *MPL* 1499delT	Decreased megakaryopoiesis or erythropoiesis, improved by complimentary transduction of MPL. Increased FLI1 response to TPO signaling.	healthy iPSC ESC	J Clin Invest. 2013 Sep 3;123(9):3802-14.
Severe congenital neutropenia (SCN)	*ELANE*	Neutrophil maturation arrest. Little sensitivity to G-CSF.	healthy iPSC	Proc Natl Acad Sci U S A. 2013 Feb 19;110(8):3023-8.
Shwachman-Diamond syndrome (SDS)	*SDBS*	Deficits in exocrine pancreatic and hematopoietic differentiation. Enhanced aptosis. Elevated protease levels in culture supernatants.	SBDS-deficient ESCs	Cell Stem Cell. 2013 Jun 6;12(6):727-36.
Cardiac				
Long QT syndrome type 1	*KCNQ1* R190Q	Prolonged action potentials and arryhthmogenic early afterdepolarizations due to reduced IKs current. Dominant-negative effect of R190Q-KCNQ1 ion channel subunits on the trafficking of wild type KCNQ1 channels to the plasma membrane.	healthy iPSC	N Engl J Med. 2010 Oct 7;363(15):1397-409.
Long QT syndrome type 2	*KCNH2* G1681A	Prolonged action potentials.	healthy iPSC	Eur Heart J. 2011 Apr;32(8):952-62.
Long QT syndrome type 2	*KCNH2* R107W	Asymptomatic carrier with LQT2 family history used to diagnose LQT2 as hiPSCcardiomyocytes showed prolonged FPD/APD.	healthy iPSC	Dis Model Mech. 2012 Mar;5(2):220-30.
Long QT syndrome type 2	*KCNH2* A614V	Prolonged action potentials due to reduced IKr current. Early afterdepolarizations and triggered arrhythmias.	healthy iPSC	Nature. 2011 Mar 10;471(7337):225-9.
Long QT syndrome type 3	-	-	-	Circ Res. 2011 Sep 30;109(8):841-7.
Long QT syndrome type 3	*SCN5A mutation (c.5287G>A; p.V1763M)*	Dominant mutant SCN5A allele gene expression significantly prolonged action potential duration or APD an increased tetrodotoxin (TTX)-sensitive late or persistent Na+ current. A positive shift of steady state inactivation and a faster recovery from inactivation.	healthy iPSC	Int J Cardiol. 2013 Oct 15;168(6):5277-86. doi: 10.1016/j.ijcard.2013.08.015. Epub 2013 Aug 15
Long QT syndrome	*The proband was found to have a de novo SCN5A LQT-3 mutation (F1473C) and a polymorphism (K897T) in KCNH2, the gene for LQT-2.*	Analysis of the biophysics and molecular pharmacology of ion channels expressed in cardiomyocytes (CMs) differentiated from these iPSCs (iPSC-CMs) demonstrates a primary LQT-3 (Na(+) channel) defect responsible for the arrhythmias not influenced by the KCNH2 polymorphism.	wild type iPSC	J Gen Physiol. 2013 Jan;141(1):61-72.
Long QT syndrome	*KCNQ1*	A markedly prolonged field potential duration (FPD) frequent severe arrhythmia only in LQTS-iPSC-derived Ebs by the IKr blocker E4031. The IKs blocker chromanol 293B did not prolong FPD in the LQTS-iPSC-derived Ebs.	healthy iPSC	Cardiovasc Res. 2012 Sep 1;95(4):419-29.
Long QT syndrome type 1 Hypertrophic cardiomyopathy Dilated cardiomyopathy	*KCNQ1* G269S *MYH7* R663H *TNNT2* R173W	Increased susceptibility to known cardiotoxic drugs as measured by action potential duration and quantification of drug-induced arrhythmias such as early afterdepolarizations and delayed afterdepolarizations.	healthy iPSC ESC	Circulation. 2013 Apr 23;127(16):1677-91.
Catecholaminergic polymorphic ventricular tachycardia type 1	*RYR2* F2483I	Elevated diastolic Ca concentrations, reduced SR Ca content, increased susceptibility to DADs and arrhythmias after catecholaminergic stimulation.	healthy iPSC ESC	Cell Physiol Biochem. 2011;28(4):579-92.
Catecholaminergic polymorphic ventricular tachycardia type 1	*RYR2* S406L	Increased susceptibility to DADs and arrhythmia. Elevated diastolic Ca2+ concentrations, a reduced SR Ca2+.	healthy iPSC	EMBO Mol Med. 2012 Mar;4(3):180-91.
Catecholaminergic polymorphic ventricular tachycardia type 1	*RYR2* M4109R	Increased susceptibility to delayed afterdepolarizations. Whole-cell [Ca2+] transient irregularities at Ca imaging.	healthy iPSC	J Am Coll Cardiol. 2012 Sep 11;60(11):990-1000.
Catecholaminergic polymorphic ventricular tachycardia type 2	*CASQ2* D307H	Increased susceptibility to delayed afterdepolarizations. Oscillatory arrhythmic prepotentials, after-contractions and diastolic [Ca2+](i) rise. More immature phenotype with less organized myofibrils and enlarged sarcoplasmic reticulum cisternae at electron microscopy analysis.	healthy iPSC	J Cell Mol Med. 2012 Mar;16(3):468-82.
Catecholaminergic polymorphic ventricular tachycardia (CPVT)	*P2328S mutation in RyR2*	Defects in Ca(2+) cycling and electrophysiology in CPVT CMs. Catecholaminergic stress led to abnormal Ca(2+) signaling and induced arrhythmias in CPVT CMs. CPVT CMs displayed reduced sarcoplasmic reticulum Ca(2+) content. Patch-clamp recordings of CPVT CMs revealed both delayed afterdepolarizations during spontaneous beating and in response to adrenaline and also early afterdepolarizations during spontaneous beating.	healthy iPSC	PLoS One. 2012;7(9):e44660. doi: 10.1371/journal.pone.0044660. Epub 2012 Sep 4.
Arrhythmogenic right ventricular dysplasia/cardiomyopathy	*PKP2* c.2484C.T	Abnormal plakoglobin nuclear translocation and decreased beta-catenin activity. Abnormal PPAR-gamma activation only in adult-like metabolic energetics. Exaggerated lipogenesis and apoptosis Ca-handling deficits.	ESCs	Nature. 2013 Feb 7;494(7435):105-10.
Arrhythmogenic right ventricular cardiomyopathy	*PKP2*	Gene expression levels of PKP2 and plakoglobin in cardiomyocytes from ARVC-iPSCs(ARVC iPSC-CM) were significantly lower. ARVC iPSC-CMs exhibited markedly reduced immunofluorescence signals when stained for PKP2 and plakoglobin. ARVC iPSC-CMs were larger and contained darker lipid droplets.	healthy iPSC	Eur Heart J. 2013 Apr;34(15):1122-33.
Timothy Syndrome	*CACNA1C* G406R	Irregular cardiac myocyte contraction and electrical activity, excess Ca influx and prolonged APD.	healthy iPSC	Nature. 2011 Mar 10;471(7337):230-4.
Timothy syndrome	*CACNA1C* G406	Increased TH expression. Increased Norepinephrine and dopamine secretion.	healthy iPSC	Nat Med. 2011 Nov 27;17(12):1657-62
Timothy syndrome	*A point mutation in the gene encoding Ca(V)1.2*	Activity-dependent dendrite retraction.	iPSC (unaffected control)	Nat Neurosci. 2013 Feb;16(2):201-9.

Dilated cardiomyopathy	*LMNA* R225X *LMNA* exon4 ins GCCA (Frame-shit change)	abnormal nuclear morphology increased senescence and apoptosis by field electric stimulation	healthy iPSC	Aging (Albany NY). 2012 Nov;4(11):803-822.
Dilated cardiomyopathy	*TNNT2* R173W	Decreased contractility and abnormal sarcomeric alpha-actinin distribution.	healthy iPSC	Sci Transl Med. 2012 Apr 18;4(130):130ra47.
Dilated cardiomyopathy	*DES*	Diffuse abnormal DES aggregations in DCM-induced-pluripotent stem cell (iPSC)-derived cardiomyocytes the diminished maximum rate of calcium ion re-uptake, slower spontaneous beating rate failure to have sustained respond to the inotropic stress induced by administration of isoproterenol.	healthy iPSC	Hum Mol Genet. 2013 Apr 1;22(7):1395-403.
Familial hypertrophic cardiomyopathy	*MYH7* R663H	Cellular enlargement and contractile arrhythmia dysregulation of Ca2+ cycling and elevation in intracellular Ca2+.	healthy iPSC ESC	Cell Stem Cell. 2013 Jan 3;12(1):101-13.
LEOPARD Syndrome	*PTPN11*	Large cardiomyocyte having a higher degree of sarcomeric organization and preferential localization of NFATC4 in the nucleus.	wild-type iPSC	Nature. 2010 Jun 10;465(7299):808-12.
Marfan syndrome	*FBN1*	Enhanced activation of TGFβ signaling observed in MFS cells decreased their endogenous BMP signaling exogenous BMP antagonized the enhanced TGFβ signaling in both MFS stem cells and MFSiPS cells.	wild type iPSC ESC	Stem Cells. 2012 Dec;30(12):2709-19.
Marfan syndrome	*FBN1*	Osteogenic differentiation of ESCs with a FBN1 mutation is inhibited. Chondrogenesis is not perturbated and occurs in a TGF-β cell-autonomous fashion. Skeletal phenotypes observed in hESCs carrying the monogenic FBN1 mutation (MFS cells) are faithfully phenocopied by cells differentiated from iPSCs derived independently from MFS patient fibroblasts.	wild type iPSC	Proc Natl Acad Sci U S A. 2012 Jan 3;109(1):215-20.

Metabolic

Wilson's disease	*ATP7B* M769V	n.e.	healthy iPSC	Protein Cell. 2012 Nov;3(11):855-63.
Wilson's disease	*ATP7B*	Abnormal cytoplasmic localization of mutated ATP7B and defective copper transport in hepatocyte-like cells from Wilson's disease.	IMR90 iPSC ESC	Hum Mol Genet. 2011 Aug 15;20(16):3176-87.
1) Alpha1-antitrypsin deficiency 2) Familial hypercholesterolemia 3) Glycogen storage disease type 1a	1) *A1AT* E342L 2) *LDL-R* n.a. 3) *G6PC*	1) increased polymeric alpha1-antitrypsin 2) decreased LDL-uptake 3) glycogen accumulation	healthy iPSC	J Clin Invest. 2010 Sep;120(9):3127-36.
Mucopolysaccharidosis type IIIB	*NAGLU* R482W	Accumulated heparan sulfate and proteoglycans. Golgi complex alterations.	healthy iPSC	Hum Mol Genet. 2011 Sep 15;20(18):3653-66.
Pompe disease	*GAA*	Low GAA activity and high glycogen content. Multiple ultrastructural aberrances.	healthy iPSC ESC	Hum Mol Genet. 2011 Dec 15;20(24):4851-64.
Lesch-Nyhan syndrome	*HPRT1* not described	Erosion of X chromosome inactivation after long time passage.	healthy iPSC	Cell Stem Cell. 2012 May 4;10(5):595-609.
Type 1 and type 2 diabetes	-	-	-	Stem Cells Transl Med. 2012 Jun;1(6):451-61.
Type 1 diabetes	-	-	-	Proc Natl Acad Sci U S A. 2009 Sep 15;106(37):15768-73.
Type 1 diabetes (T1D)	-	Notable intrapatient variation was evident upon further guided differentiation into HNF4α- and HNF1β-expressing primitive gut tube, and INS- and glucagon (GCG)-expressing islet-like cells.	nondiabetic iPSC	Mol Ther. 2013 Jan;21(1):228-39.
Type 2 diabetes (T2D)	-	Derived iPS clones acquired a rejuvenated state, characterized by elongated telomeres and suppressed senescence-related p15INK4b/p16INK4a gene expression and oxidative stress signaling.	iPSCs from non-diabetic patients	Aging (Albany NY). 2012 Jan;4(1):60-73.
Maturity onset diabetes of the young (MODY)	*HNF4A, GCK, HNF1A, HNF1B, CEL*	-	healthy iPSC	J Biol Chem. 2013 Feb 22;288(8):5353-6.
Maturity-onset diabetes of the young type 2 (MODY2)	*GCK*	β cells required higher glucose levels to stimulate insulin secretion.	healthy +A152:D154iPSC	J Clin Invest. 2013 Jul 1;123(7):3146-53.
Mitochondrial diabetes	*mtDNA A3243G mutation*	About half of the clones had undetectable levels of the mutation.	-	Diabetologia. 2012 Jun;55(6):1689-98.
familial hypercholesterolemia (FH)	*LDLR*	Inability of uptake labeled LDL particles.	genetically corrected patient-specific iPSC	Mol Biotechnol. 2013 Jul;54(3):863-73.
familial hypercholesterolemia (FH)	*LDLR*	iPSC-derived hepatocytes show deficiencies in uptake of LDL-C and in their response to lovastatin and a marked elevation in secretion of lipidated apolipoprotein B-100.	iPSC ESC	Hepatology. 2012 Dec;56(6):2163-71.
Fabry disease	*mutations in the retinal pigment epithelium (RPE) gene BESTROPHIN1*	massive membranous cytoplasmic bodies	-	Mol Genet Metab. 2013 Aug;109(4):386-9.
Mucopolysaccharidosis type I (Hurler Syndrome)	*IDUA(Y167X, W402X)*	accumulation of GAG	genetically corrected patient-specific iPSC	Blood. 2011 Jan 20;117(3):839-47.
1)Tyrosinemia type I 2)Glycogen straoge type Ib 3)Progressive familial hereditary cholestasis 4)Crigler-Najjar Syndrome	1)*FAH* Q64H 2)*SLC37A* c.1124-2A>G 3)Multifactorial 4)*UGT1A1* L413P	n.e.	n.e.	Stem Cell Rev. 2010 Dec;6(4):622-32.

Imprinting

Angelman syndrome Pradder-Willi syndrome	*UBE3A* (15q11-q13) deletion	No evidence of DNA methylation imprint erasure at the cis-acting PSW imprinting center. UBE3A paternalimprinting re-established during hiPSC neuronal differentiation.	healthy iPSC	Proc Natl Acad Sci U S A. 2010 Oct 12;107(41):17668-73.
Pradder-Willi syndrome	*UBE3A* (15q11-q13) deletion	iPSCs retain a high level of DNA methylation in the imprinting center of the maternal allele and show concomitant reduced expression of the disease-associated small nucleolar RNA HBII-85/SNORD116.	healthy iPSC	J Biol Chem. 2010 Dec 17;285(51):40303-11.

Skin

Recessive dystrophic epidermolysis bullosa (RDEB)	*COL7A1* E2059G *COL7A1* G1640fsX70/R2751fsX20 *COL7A1* R261X	No Col7 was detected in the skin-like structures derived from RDEB iPS cells.	healthy iPSC	J Invest Dermatol. 2011 Apr;131(4):848-56.
Recessive dystrophic epidermolysis bullosa (RDEB)	*COL7A1*	No expression of Type VII collagen.	healthy iPSC	Proc Natl Acad Sci U S A. 2011 May 24;108(21):8797-802.
Dyskeratosis congenita	*TERT* P704S *TERT* R979W DKC1 L54V TCAB1 H*Y/G*R	Disrupts telomere elongation during reprogramming telomerase mislocalizes from Cajal bodies to nucleoli and decreased telomerase activity.	healthy iPSC	Nature. 2011 May 22;474(7351):399-402.
Dyskeratosis Congenita	*DKC1*	Telomere elongation, upregulation of TERC and DKC1.	wild-type iPSC	Nature. 2010 Mar 11;464(7286):292-6.
Hermansky-Pudlak syndrome Chediak-Higashi syndrome	*HP1* exon15 16bp dupl. *HP2* G2028T *LYST* Codon40 GCA to GGCA	Decreased melanin content melanosome abnormality.	healthy iPSC	Cell Rep. 2013 Apr 25;3(4):1140-52.

other

Down syndrome (trisomy 21)	Chr.21 trisomy	Increased Abeta 40. Fibrillar Abeta accmulation. Increased phospo-Tau (pT231, pS396).	healthy iPSC	Sci Transl Med. 2012 Mar 7;4(124):124ra29.
Down syndrome (trisomy 21)	Chr.21 trisomy	The derived, disomic cells proliferated faster and produced more endothelia in vivo than their otherwise isogenic trisomic counterparts, but in vitro hematopoietic differentiation was not consistently altered.	isogenic disomic iPSC	Cell Stem Cell. 2012 Nov 2;11(5):615-9.
Down syndrome (trisomy 21)	Chr.21 trisomy	Yielded Ts21 iPSCs and an isogenic control that is disomic for human chromosome 21 (HSA21). Ts21 neurons displayed reduced synaptic activity.	isogenic disomic iPSC	Proc Natl Acad Sci U S A. 2013 Jun 11;110(24):9962-7.
Down syndrome (trisomy 21)	Chr.21 trisomy	The level of amyloid precursor protein was significantly increased in NPCs derived from T21 AF-iPS cells. The expression levels of miR-155 and miR-802 in T21 AF-iPS-NPCs were highly elevated in the presence of low expression of MeCP2. T21 iPS-NPCs generated fewer neurons and exhibit developmental defects during neurogenesis.	normal iPSC	Exp Cell Res. 2013 Feb 15;319(4):498-505.
Down syndrome (trisomy 21)	Chr.21 trisomy	Blood progenitor populations generated from T21 iPSCs were present at normal frequency and proliferated normally. However, their developmental potential was altered with enhanced erythropoiesis and reduced myelopoiesis, but normal megakaryocyte production	euploid iPSC	Proc Natl Acad Sci U S A. 2012 Oct 23;109(43):17573-8.
Down syndrome (trisomy 21)	Chr.21 trisomy	-	-	Stem Cell Res Ther. 2012 Apr 18;3(2):14.

Down syndrome (trisomy 21)	Chr.21 trisomy	Trisomic cells of hES, iPS, or isogenic origins exhibited a two- to fivefold increase in a population of CD43(+)(Leukosialin)/CD235(+)(Glycophorin A) hematopoietic cells.	isogenic iPSC	Proc Natl Acad Sci U S A. 2012 Oct 23;109(43):17567-72.
Turner syndrome(monosomy X) trisomy 8 (Warkany syndrome 2) trisomy9 trisomy 13 (Patau syndrome) partial trisomy 11;22 (Emanuel syndrome)	-	TS iPSCs displayed lower levels of the pseudoautosomal genes ASMTL and PPP2R3B, and displayed insufficient up-regulation of the pseudoautosomal placental gene CSF2RA during embryoid body formation.	IMR90 iPSC ESC	Hum Mol Genet. 2012 Jan 1;21(1):32-45.
Klinefelter's Syndrome	*47, XXY*	Aberrantly expressed genes associated with the clinical features of KS.	normal iPSC	J Biol Chem. 2012 Nov 9;287(46):38970-9.
Atypical Werner Syndrome (AWS) Hutchinson Gilford Progeria Syndrome (HGPS) Dilated Cardiomyopathy (DCM)	*lamin A/C*	low expression of lamin A/C in iPSC nuclear abnormality, increased senescence and susceptibility to apoptosis in iPSC-derived fibroblasts	normal hiPSC	Aging (Albany NY). 2011 Apr;3(4):380-90.
Hutchinson Gilford Progeria Syndrome (HGPS)	*LMNA* c.1824C>T (splice alterlation)	Increased DNA damage and cell death at hypoxic condition. Transplantation effect for ischemic Murine Hind Limb were decreased.	healthy iPSC ESC	Proc Natl Acad Sci U S A. 2010 Oct 12;107(41):17668-73.G181
Hutchinson Gilford Progeria Syndrome (HGPS)	*LMNA*	Absence of progerin. Lack of the nuclear envelope. Epigenetic alterations normally associated with premature ageing.	wid type iPSC	Nature. 2011 Apr 14;472(7342):221-5.
Hutchinson Gilford Progeria Syndrome (HGPS)	*LMNA*	Increased gene expression of progerin.	healthy iPSC genetically corrected patient-specific iPSC	Cell Stem Cell. 2011 Jun 3;8(6):688-94.
Cockayne syndrome (CS)	*ERCC6*	Elevated cell death rate and higher reactive oxygen species (ROS) production. Up-regulation of TXNIP and TP53 transcriptional expression.	wild type iPSC	Hum Mol Genet. 2012 Sep 1;21(17):3825-34.
Schizophrenia	-	Perturbations in neural differentiation and mitochondrial function.	healthy iPSC	Mol Psychiatry. 2013 Oct;18(10):1067-76.
Schizophrenia	Sporadic	Decreaed synaptic junction. Decreaed neurite length. Decreaed number of synapse. Spontanueous action potential or EPSP firing ratio were unchanged.	healthy iPSC	Nature. 2011 May 12;473(7346):221-5.
Schizophrenia, PD	-	Disease-specific alterations in gene expression, protein epression and cell function.	healthy ONS(olfactory neurosphere-derived) cells	Dis Model Mech. 2010 Nov-Dec;3(11-12):785-98.
autism spectrum disorder	-	-	-	Neurosci Lett. 2012 May 10;516(1):9-14.
Cystic fibrosis α-1 antitrypsin deficiency-related emphysema scleroderma skckle-cell disease	-	-	ESCs	Stem Cells. 2010 Oct;28(10):1728-40.
α1 antitrypsin deficiency	*A1AT, also known as SERPINA1*	Polymeric A1AT protein in hepatocyte-like cells decreased A1AT enzymatic inhibitory activity.	genetically corrected patient-specific iPSC	Nature. 2011 Oct 12;478(7369):391-4.
α1 antitrypsin deficiency	*A1AT*	-		Blood. 2011 Aug 18;118(7):1801-5.
ectrodactyly, ectodermal dysplasia, and cleft lip/palate (EEC) syndrome	*p63*	Early ectodermal commitment into K18(+) cells but failed to further differentiate into K14(+) cells (epidermis/limbus) or K3/K12(+) cells (corneal epithelium).	healthy iPSC	Proc Natl Acad Sci U S A. 2013 Feb 5;110(6):2152-6.
Fibrodysplasia ossificans progressiva (FOP)	*activin receptor-like kinase 2 (ALK2)*	Generation of induced pluripotent stem cells (iPSCs) from FOP-derived skin fibroblasts is repressed.	healthy iPSC	Stem Cells. 2012 Nov;30(11):2437-49.
primary immunodeficiencies	*RAG1* *STAT1* *TLR3* *RMRP*	-	healthy iPSC	J Allergy Clin Immunol. 2011 Jun;127(6):1400-7.e4.
XLF/Cernunnos syndrome	*XLF(homozygous R57X)*	Alterd in vitro differentiation capacity and inability to generate teratomas comprised of all three layers in vitro.	unaffected iPSC ESC	Stem Cells. 2013 Jul 2. doi: 10.1002/stem.1456.
familial Alzheimer's disease familial Parkinson disease	*PARK4*	increased Aβ42/40, α-synuclein	centenarian donors	PLoS One. 2012;7(7):e41572. doi: 10.1371/journal.pone.0041572. Epub 2012 Jul 25.
Alzheimer's disease Parkinson's disease juvenile-onset, type I diabetes mellitus Duchenne type muscular dystrophy	-	n.e.	n.e.	Exp Mol Med. 2012 Mar 31;44(3):202-13.
1)ADA-SCID 2)Gaucher disease type III 3)Duchenne muscular dystrophy 4)Becker muscular dystrophy 5)Down Syndrome 6)Parkinson disease 7)Juvenile diabetes mellitus 8)Shwachman-Bodian-Diamond syndrome 9)Huntington disease 10)Lesch-Nyhan Syndrome (carrier)	1)*ADA* exon7 GGG>AGG/exon10 Del(GAAGA) 2)*GBA* exon9 AAC>AGC/84G insertion 3)*dystrophin* exon45–52 deletion 4)*dystrophin* unidentified 5)Chr.21 trisomy 6)sporadic 7)sporadic 8)*SBDS* IV2+2T>C/IVS3-1G>A 9)*HTT* CAG repeat 10)*HPRT1* Heterozygosity	n.e.	n.e.	Cell. 2008 Sep 5;134(5):877-86.
ADPKD SLE Wilms tumor	*W3842X mutation in exon 41 of PKD1 gene*	-	-	Stem Cell Res Ther. 2011 Dec 6;2(6):48.
HIV	-	HIV-1 resistant and functional macrophage	healthy iPSC	Mol Ther. 2011 Mar;19(3):584-93.