Supplemental Table 1. A selection of recent iPSC-based drug screens (2016-2018)

Genetic disease	**Screened mutant gene(s)**	**iPSC-derived cell type for screen** (*in vitro* phenotype)	**Screen type** (compounds tested)	**Compound modulator(s)** (disease effect or readout)	***In vivo* disease model** (effect)	**Ref.**
Age-related macular degeneration	***ARMS2 & HTRA1***	**Retinal pigment epithelium** (↑ disease markers)	**Targeted testing** (1 - anti-inflammatory & anti-oxidant)	**Nicotinamide** (↓ disease markers, pro-inflammatory cytokines, & complement factors, and ↑ SIRT1 & NAD biosynthetic pathway)	**Not tested**	(1)
Amyotrophic lateral sclerosis	***C9ORF72***	**Neurons** (↑ intracellular and extracellular poly(GP) protein)	**Targeted testing** (1 - antisense oligonucleotide)	***Antisense oligonucleotide targeting G4C2 repeat C9ORF72 transcript*** (↓ intracellular and extracellular poly(GP) protein)	**Mouse** (↓ CSF and brain poly(GP) protein)	(2)
	C9ORF72	**Motor neurons** (cell death)	**Post-screen validation** (1 – screening hit)	**Vardenafil** (↑ motor neuron survival)	**Mouse** (↑ survival)	(3)
	C9ORF72	**Cortical & motor neurons** (RNA repeat foci & dipeptide repeat proteins)	**Post-screen validation** (3 – screening hits)	**DB1246** **DB1247** **DB1273** (↓ RNA foci & repeat proteins)	***Drosophila*** (↑ survival)	(4)
	FUS	**iPSCs with FUS-eGFP reporter** (↑ stress granule area with sodium arsenite or heat treatment, mislocalization of FUS-eGFP)	**Large collection** 1st: (1000 - chemical genetics) 2nd: (1600 - unbiased)	**Rapamycin** **Torkinib** **Paroxetine** **Promethazine** **Trimipramine** (↓ FUS-eGFP stress granule area with sodium arsenite treatment)	**Not tested**	(5)
	SOD1	**Motor neurons** (cell death)	**Large collection** (1416 - unbiased)	**Bosutinib** (↑ motor neuron survival)	**Mouse** (↑ survival)	(6)

	SOD1	**Spinal motor neurons** (cell death, ↑ markers of ER stress,	**Targeted testing** (6 - inhibitors of p38, ERK, JNK, or CDK kinases, p53, or WNT)	**FR180204** **Pifithrin-α hydrobromide** **SB203580** **SP600125** **XAV 939** (↑ motor neuron survival)	**Not tested**	(7)
Arterial calcification due to deficiency of CD73	***NT5E***	**Mesenchymal stromal cells** (↑ calcification, ↑ TNAP activity, ↓ adenosine production, ↓ PPi	**Targeted testing** (1 – A2bAR agonist & 1 - bisphosphonate)	**BAY 60-6583*** **Etidronate** (↓ calcification, *↓ TNAP activity, & *↓ AKT and p70S6K phosphorylation)	**Mouse with patient iPSC-derived teratomas** (↓ calcification)	(8)
Autism spectrum disorder	***SHANK3***	**Neurons** (↓ Neurite length, neurite branch points, SHANK3 synaptic content, & frequency and intensity of spontaneous calcium oscillations)	**Post-screen validation** (2 – screening hits)	**Lithium** **Valproic acid** (↑ SHANK3 synaptic content & frequency and intensity of spontaneous calcium oscillations)	**Human patients** (↓ autism severity, *only lithium tested)*	(9)
Alzheimer's disease	***APOE***	**Neurons** (↑ amyloid beta [40 & 42], ↑ P-tau, ↓ GABAergic neurons, & ↑ ApoE4 fragments)	**Targeted testing** (1 – structure corrector)	**PH002** (↓ amyloid beta [40 & 42], ↓ P-tau, ↑ GABAergic neurons, & ↓ ApoE4 fragments)	**Not tested**	(10)
	APP	**Neurons** (↑ amyloid beta 1-40 peptides)	**Targeted testing** (2 - retromer stabilizers)	**R33** **R55** (↓ amyloid beta peptides & ↓ tau phosphorylation [+/- APP expression])	**Not tested**	(11)
	PSEN1	**Cortical neurons** (↑ amyloid beta 42/40 ratio)	**Large collection** (1258 - unbiased)	**Bromocriptine** **Cromolyn** **Topiramate** (↓ amyloid beta 42)	**Not tested**	(12)
	PSEN1	**Neurons** (↑ intra- & extracellular amyloid beta 42)	**Targeted testing** (1 - flavonoid)	**Nobiletin** (↑ neprilysin mRNA, ↓ intra- & extracellular amyloid beta 42)	**Not tested**	(13)

	Trisomy 21	**Cortical neurons** (Elevated amyloid beta 42/40 & 42/38 ratio)	**Large collection** (1280 - unbiased)	**Selamectin** (↓ amyloid beta 42/38 ratio)	**Not tested**	(14)
Behçet's disease	**Idiopathic**	**Hematopoietic precursor cells** (*AGTR2, CA9, CD44, CXCL1, HTN3, IL-2, PTGER4,* & *TSLP* expression)	**Targeted testing** (34 - immuno-suppressant and anti-inflammatory drugs)	**Multiple corticosteroids** (↓ CXCL1)	**Not tested**	(15)
BH4 metabolism disorders	***PTPS***	**Dopaminergic neurons** (↓ tyrosine hydroxylase, ↓ BH4, ↓ neurons, ↓ tyrosine hydroxylase area, ↓ extraceullar dopamine, & ↑ neopterin)	**Targeted testing** (2 – BH4 supplements)	**BH4** **Sepiapterin** **(**↑ tyrosine hydroxylase area & ↑ extraceullar dopamine [*PTPS* mutants only])	**Not tested**	(16)
Catecholaminergic polymorphic ventricular tachycardia	***CASQ2***	**Cardiomyocytes** (Abnormal Ca^{2+} cycling, isoproterenol-induced arrhythmia)	**Targeted testing** (34 - immuno-suppressant and anti-inflammatory drugs) *Targeted testing: 6 (anti-arrhythmics)*	**JTV-519** **Carvedilol** **Flecainide** **Riluzole** (↓ Ca^{2+} cycling abnormalities)	**Human patients** (↓ ventricular tachy-cardia events, *only flecainide tested*)	(17)
	RYR2	**Cardiomyocytes** (Isoproterenol-induced abnormal diastolic Ca^{2+} increase, & ↑ delayed afterdepolarizations)	**Targeted testing** (1 – stabilizer of the closed state of the ryanodine receptor 2)	**S107** (↓ delayed afterdepolarizations)	**Not tested**	(18)
Diamond-Blackfan anemia	***RPS19*** ***RPL5***	**Hematopoietic stem cells & erythroid progenitors** (↓ Erythroid progenitors in vitro and in vivo [after transplantation])	**Large collection** (1440 - unbiased)	**SMER28** (↑ erythroid progenitors through ATG5)	**Mouse** (↑ Hb) **Zebrafish** (↑ red blood cells)	(19)

Inherited erythromelalgia	***SCN9A***	**Sensory neurons** (↑ excitability)	**Targeted testing** (2 - Nav1.7 inhibitors)	**PF-05089771** (↓ excitability)	**Human patient** (↓ heat-induced pain)	(20)
Familial hypercholesteremia	***LDLR***	**Hepatocyte-like cells** (↑ levels of apoB in culture media, inability to traffic exogenous LDL-cholesterol to endosomes, no ↑ LDL-cholesterol clearance with statins)	**Large collection** (2320 - unbiased)	**Digoxin** **Proscillaridin** (↓ ApoB)	**Humanized mouse** (↓ Serum LDL-cholesterol & apoB) ***Human patient medical records*** (↓ LDL-cholesterol with cardiac glycosides)	(21)
Fibrodysplasia ossificans progressiva	***ACVR1***	**Mesenchymal stromal cells with luciferase reporter** (No wild-type comparisons)	**Large collection** (6809 - unbiased)	**Rapamycin** (↓ activin-A-mediated chondrogenic induction)	**Mouse** (↓ heterotopic ossification)	(22)
	ACVR1	**Osteogenic cells** (No differences to wild-type)	**Targeted testing** (3 - AMPK activators)	**A769962** **AICAR** **Metformin** (↓ osteogenic differentiation)	**Not tested**	(23)
Fragile X syndrome	***FMR1***	**Neural progenitor cells with *FMR1*-nano luciferase reporter** (↓ *FMR1* expression)	**Large collection** 1st: (128 – epigenetic modulators) 2nd: (1134 – unbiased)	**5-aza-dC** **5-aza-C** (↑ *FMR1* expression)	**Not tested**	(24)

Friedreich ataxia	***FXN***	**Neurons** (↓ frataxin, ↓ Fe–S synthesis elements, ↑ oxidative stress, & ↑ neuronal death)	**Targeted testing** (1 – HDAC inhibitor)	**HDAC inhibitor 109** (↑ frataxin, ↑ Fe–S synthesis elements, ↓ oxidative stress, & ↓ neuronal death)	**Not tested**	(25)
Frontotemporal dementia	***C9ORF72***	**Cortical & motor neurons** (RNA repeat foci & dipeptide repeat proteins)	**Post-screen validation** (3 – screening hits)	**DB1246** **DB1247** **DB1273** (↓ RNA foci & repeat proteins)	**Drosophila** (↑ survival)	(4)
	GRN	**Cortical neurons** (↓ *GRN* expression, ↓ PGRN [intracellular & secreted])	**Targeted testing** (1 - HDAC inhibitor)	**SAHA** (↑ *GRN* expression, ↑ PGRN [intracellular & secreted])	**Not tested**	(26)
	GRN	**Neurons** (↓ PGRN)	**Post-screen validation** (1 – screening hit)	**Trehalose** (↑ PGRN)	**Mouse** (↑ brain levels of PGRN)	(27)
	MAPT	**Cortical neurons** (↑ tau, ↑ P-tau, ↑ P-tau in neuronal processes and cell bodies, ↑ mutant tau, ↓ tau solubility, ↑ markers of ER stress and autophagy, ↑ cell death in presence of stressors)	**Targeted testing** (1 - autophagy stimulator)	**Rapamycin** (↑ cell survival in presence of stressors)	**Not tested**	(28)
Gaucher disease	***GBA1***	**Dopaminergic neurons** (↓ gluco-cerebrosidase activity, ↑ gluco-sylceramide, & ↑ gluco-sylsphingosine)	**Targeted testing** (1 - molecular chaperone)	**NCGC607** (↑ gluco-cerebrosidase activity & ↓ α-synuclein [in parkinsonism cells])	**Not tested**	(29)

	GBA1	**Dopaminergic neurons** (↑ ubiquitinated protein species, oxidized α-synuclein, & total α-synuclein)	**Targeted testing** (1 - lysosomal enzyme acid ceramidase inhibitor)	**Carmofur** (↓ ubiquitinated protein species, & oxidized α-synuclein)	**Not tested**	(30)
Huntington's disease	***HTT***	**Medium spiny-like neurons** (↑ cell death)	**Targeted testing** (1 – PPARgamma activator)	**Bexarotene** (↓ cell death)	**Mouse** (↑ motor function, survival, & # of neurons)	(31)
	HTT	**Neurons** (dysregulated transcriptional programs & epigenetic signatures involved in neuronal development)	**Targeted testing** (1 – neuronal differentiation inducer)	**Isoxazole-9** (↓ cell death, ↓ neurite length, & ↑ *NEUROD1* expression)	**Mouse** (cognitive impairment & synaptic pathology)	(32)
	HTT	**Brain microvascular endothelial cells** (↑ migration, abnormal blood-brain barrier function, & perturbed angiogenic & barrier pathways)	**Targeted testing** (1 – WNT inhibitor)	**XAV939** (Rescued angiogenic deficits)	**Not tested**	(33)
Long QT syndrome type 1 & 2	***KCNQ1*** ***KCNH2***	**Cardiomyocytes** (↑ QT interval)	**Targeted testing** (1 - hERG allosteric modulators)	**LUF7346** (↓ QT interval)	**Not tested**	(34)
Long QT syndrome type 3	***SCN5A***	**Cardiomyocytes** (pronounced cardiac late sodium current)	**Targeted testing** (1 - selective inhibitior of cardiac late sodium current)	**GS967** (↓ cardiac late sodium current, altered cardiac late sodium current kinetics, ↓ action potential, & ↓ pro-arrhythmic events)	**Not tested**	(35)

***MECP2* duplication syndrome**	***MECP2***	**Cortical neurons** (↑ synaptogenesis, dendritic arborization, & synchronized burst events)	**Targeted testing** (43 – epigenetic modulators)	**NCH-51** **Scriptaid** (↓ synaptic protein PSD95)	**Not tested**	(36)
Mitochondrial disorders (various)	***MT-ATP6***	**Neural progenitor cells** (↓ ATP, mitochondrial hyperpolarization, & altered calcium homeostasis)	**Large collection** (130 – unbiased)	**Avanafil** (↓ mitochondrial membrane potential)	**Not tested**	(37)
Neuronal ceroid lipofuscinoses	***PPT1 & TPP1***	**Neural stem cells** (↑ Lysosomal lipid accumulation, enlarged lysosomes, & subunit C of mitochondrial ATP synthase puncta)	**Targeted testing** (2 - lysosomal content reducers)	**δ-tocopherol*** **HPBCD** (↓ enlarged lysosomes, *lysosomal lipid accumulation, & *subunit C of mitochondrial ATP synthase puncta)	**Not tested**	(38)
Neutropenia	***ELANE***	**Neutrophils** (↓ Neutrophils)	**Targeted testing** (2 - neutrophil elastase inhibitors)	**MK0339** (↑ Neutrophils)	**Not tested**	(39)
Parkinson's disease	***DJ-1***	**Midbrain dopaminergic neurons** (↑ mitochondrial oxidant stress, oxidized dopamine, neuromelanin, & oxidized, insoluble α-synuclein, L-dopa-mediated ↑ oxidized dopamine, and lysosomal dysfunction)	**Targeted testing** (2 - Cav1 channel antagonists & 2 - antioxidants)	**FK506*** **Isradipine** **mito-TEMPO** **N-acetylcysteine**** (↓ oxidized dopamine, *, **restored lysosomal function, & ↓ **α-synuclein)	**Not tested**	(40)

	LRRK2	**Dopaminergic neurons** (↑ p-APP & ↓ tyrosine hydroxylase)	**Targeted testing** (1 - LRRK2 catalytic inhibitor)	**LRRK2-IN-1** (↓ p-APP & ↑ tyrosine hydroxylase)	**Mouse** (HG-10-102-01 treatment: ↓ p-APP, ↑ tyrosine hydroxylase)	(41)
	SNCA	**Neurons** (αSyn aggregation, neurite dysmophoria, & synaptic defects)	**Targeted testing** (3 - modulators of αSyn oligomers)	**NPT100-18A*** **NPT100-14A** **ELN484228** (↓ axonal degeneration, protection from proteasome inhibition, *↑ neurite #)	**Not tested**	(42)
	SNCA	**Neural precursor cells & neurons** (No iPSC-derived wild-type comparisons)	**Post-screen validation** (1 – screening hit)	**Clenbuterol** (↓ rotenone-induced superoxide [in neural precursors] & ↓ SNCA)	**Mouse, wild-type** (↓ SNCA in substantia nigra) ***Patient medical records from Norwegian Prescription Database*** (Parkinson's disease risk ↓ with β2AR agonists, ↑ with β2AR antagonists)	(43)
Pelizaeus-Merzbacher disease	***PLP1***	**Oligodendrocytes** (↓ Process length, ↓ process number, PLP endoplasmic reticulum retention, differentiation defects, & myelination deficits)	**Targeted testing** (2 - endoplasmic reticulum stress modulators)	**Guanabenz** **GSK2656157** (↑ PLP shuttling to processes & ↑ myelination)	**Not tested**	(44)

Pulmonary arterial hypertension	***BMPR2***	**Endothelial cells** (angiogenesis and wound closure assay abnormalities, ↓ BMPR2, & ↓ BMPR2 downstream pathway effectors [after stimulation])	**Targeted testing** (2 - BMPR2 signaling enhancers)	**Elafin** **FK506** (Improved angiogenesis and wound closure assay performance [in responding lines])	**Not tested**	(45)
Retinitis pigmentosa	***MERTK***	**Retinal pigment epithelium** (↓ phagocytosis, no MERTK staining)	**Targeted testing** (2 - translation read-through promoters)	**G418** (Detection of MERTK) **PTC124** (↑ phagocytosis & detection of MERTK)	**Not tested**	(46)
Short QT syndrome - type 1	***KCNH2***	**Cardiomyocytes** (↑ KCNH2, ↓ action potential duration, ↑ rapidly activating delayed rectifier potassium current, ↑ expression of *KCND3*, *KCHIP2*, *CACNA1C*, & *KCNH2*, ↑ intracellular Ca^{2+} level, ↑ arrhythmic events, ↑ carbachol-induced arrhythmic events	**Targeted testing** (3 - anti-arrhythmics)	**Quinidine** (↑ action potential duration & ↓ carbachol-induced arrhythmic events)	**Not tested**	(47)
Spinal muscular atrophy	***SMN1***	**Neurons** (No assessment of phenotype in neurons but ↓ SMN in iPSCs)	**Targeted testing** (14 – tetrapeptide HDAC inhibitors)	**Compound 3** (↑ *SMN2* expression)	**Not tested**	(48)
	SMN1	**Spinal motor neurons** (↓ dendrite length & branching, ↑ apoptosis, ↑ astrocytes)	**Targeted testing** (1 – thyrotropin-releasing hormone analog)	**5-oxo-l-prolyl-l-histidyl-l-prolinamide** (↑ *SMN2* expression, ↑ SMN, ↑ dendrite length & branching, & ↑ p-GSK-3β)	**Human patients** (improved gait metrics)	(49)

Spinocerebellar ataxia type 6	***CACNA1A***	**Purkinje cells** (↑ Cav2.1, ↓ Cav2.1 c-terminal domain [α1ACT], & ↓ cell survival and dendrite deficits [in T3 depletion])	**Targeted testing** (5 – various mechanisms)	**Riluzole** **Thyroid releasing hormone** (↑ cell survival & dendrite deficits [in T3 depletion])	**Not tested**	(50)
Timothy syndrome	***CACNA1C***	**Cardiomyocytes** (↓ spontaneous beating rate & ↑ contraction irregularity)	**Targeted testing** (20 – roscovitine analogs & 4 CDK inhibitors)	**CR8** **DRF053** **Myoseverin-B** **PHA-793887** (↑ spontaneous beating rate & ↓ contraction irregularity, through CDK5 inhibition)	**Not tested**	(51)
VCP-associated disease	***VCP***	**Myogenic lineage cells** (↑ TDP-43, ubiquitin, Light Chain 3-I/II protein, & p62)	**Targeted testing** (7 - autophagy stimulators or inhibitors)	**AT101** **Perifosine** **Rapamycin** (↓ VCP pathological markers)	**Not tested**	(52)
Wolman disease	***LIPA***	**Neural stem cells** (↓ lysosomal acid lipase activity, & ↑ lysosomal content and lipids)	**Targeted testing** (2 - lysosomal content reducers)	**δ-tocopherol** **HPBCD** (↓ lysosomal content and lipids)	**Not tested**	(53)

1 Saini, J.S., Corneo, B., Miller, J.D., Kiehl, T.R., Wang, Q., Boles, N.C., Blenkinsop, T.A., Stern, J.H. and Temple, S. (2017) Nicotinamide Ameliorates Disease Phenotypes in a Human iPSC Model of Age-Related Macular Degeneration. *Cell stem cell*, **20**, 635-647 e637.

2 Gendron, T.F., Chew, J., Stankowski, J.N., Hayes, L.R., Zhang, Y.J., Prudencio, M., Carlomagno, Y., Daughrity, L.M., Jansen-West, K., Perkerson, E.A. *et al.* (2017) Poly(GP) proteins are a useful pharmacodynamic marker for C9ORF72-associated amyotrophic lateral sclerosis. *Sci Transl Med*, **9**.

3 Osborn, T.M., Beagan, J. and Isacson, O. (2018) Increased motor neuron resilience by small molecule compounds that regulate IGF-II expression. *Neurobiology of disease*, **110**, 218-230.

4 Simone, R., Balendra, R., Moens, T.G., Preza, E., Wilson, K.M., Heslegrave, A., Woodling, N.S., Niccoli, T., Gilbert-Jaramillo, J., Abdelkarim, S. *et al.* (2018) G-quadruplex-binding small molecules ameliorate C9orf72 FTD/ALS pathology in vitro and in vivo. *EMBO Mol Med*, **10**, 22-31.
5 Marrone, L., Poser, I., Casci, I., Japtok, J., Reinhardt, P., Janosch, A., Andree, C., Lee, H.O., Moebius, C., Koerner, E. *et al.* (2018) Isogenic FUS-eGFP iPSC Reporter Lines Enable Quantification of FUS Stress Granule Pathology that Is Rescued by Drugs Inducing Autophagy. *Stem Cell Reports*, **10**, 375-389.
6 Imamura, K., Izumi, Y., Watanabe, A., Tsukita, K., Woltjen, K., Yamamoto, T., Hotta, A., Kondo, T., Kitaoka, S., Ohta, A. *et al.* (2017) The Src/c-Abl pathway is a potential therapeutic target in amyotrophic lateral sclerosis. *Sci Transl Med*, **9**.
7 Bhinge, A., Namboori, S.C., Zhang, X., VanDongen, A.M.J. and Stanton, L.W. (2017) Genetic Correction of SOD1 Mutant iPSCs Reveals ERK and JNK Activated AP1 as a Driver of Neurodegeneration in Amyotrophic Lateral Sclerosis. *Stem Cell Reports*, **8**, 856-869.
8 Jin, H., St Hilaire, C., Huang, Y., Yang, D., Dmitrieva, N.I., Negro, A., Schwartzbeck, R., Liu, Y., Yu, Z., Walts, A. *et al.* (2016) Increased activity of TNAP compensates for reduced adenosine production and promotes ectopic calcification in the genetic disease ACDC. *Sci Signal*, **9**, ra121.
9 Darville, H., Poulet, A., Rodet-Amsellem, F., Chatrousse, L., Pernelle, J., Boissart, C., Heron, D., Nava, C., Perrier, A., Jarrige, M. *et al.* (2016) Human Pluripotent Stem Cell-derived Cortical Neurons for High Throughput Medication Screening in Autism: A Proof of Concept Study in SHANK3 Haploinsufficiency Syndrome. *EBioMedicine*, **9**, 293-305.
10 Wang, C., Najm, R., Xu, Q., Jeong, D.E., Walker, D., Balestra, M.E., Yoon, S.Y., Yuan, H., Li, G., Miller, Z.A. *et al.* (2018) Gain of toxic apolipoprotein E4 effects in human iPSC-derived neurons is ameliorated by a small-molecule structure corrector. *Nature medicine*, in press.
11 Young, J.E., Fong, L.K., Frankowski, H., Petsko, G.A., Small, S.A. and Goldstein, L.S.B. (2018) Stabilizing the Retromer Complex in a Human Stem Cell Model of Alzheimer's Disease Reduces TAU Phosphorylation Independently of Amyloid Precursor Protein. *Stem Cell Reports*, **10**, 1046-1058.
12 Kondo, T., Imamura, K., Funayama, M., Tsukita, K., Miyake, M., Ohta, A., Woltjen, K., Nakagawa, M., Asada, T., Arai, T. *et al.* (2017) iPSC-Based Compound Screening and In Vitro Trials Identify a Synergistic Anti-amyloid beta Combination for Alzheimer's Disease. *Cell Rep*, **21**, 2304-2312.
13 Kimura, J., Shimizu, K., Kajima, K., Yokosuka, A., Mimaki, Y., Oku, N. and Ohizumi, Y. (2018) Nobiletin Reduces Intracellular and Extracellular beta-Amyloid in iPS Cell-Derived Alzheimer's Disease Model Neurons. *Biol Pharm Bull*, **41**, 451-457.
14 Brownjohn, P.W., Smith, J., Portelius, E., Serneels, L., Kvartsberg, H., De Strooper, B., Blennow, K., Zetterberg, H. and Livesey, F.J. (2017) Phenotypic Screening Identifies Modulators of Amyloid Precursor Protein Processing in Human Stem Cell Models of Alzheimer's Disease. *Stem Cell Reports*, **8**, 870-882.

15 Son, M.Y., Kim, Y.D., Seol, B., Lee, M.O., Na, H.J., Yoo, B., Chang, J.S. and Cho, Y.S. (2017) Biomarker Discovery by Modeling Behcet's Disease with Patient-Specific Human Induced Pluripotent Stem Cells. *Stem Cells Dev*, **26**, 133-145.
16 Ishikawa, T., Imamura, K., Kondo, T., Koshiba, Y., Hara, S., Ichinose, H., Furujo, M., Kinoshita, M., Oeda, T., Takahashi, J. *et al.* (2016) Genetic and pharmacological correction of aberrant dopamine synthesis using patient iPSCs with BH4 metabolism disorders. *Hum Mol Genet*, **25**, 5188-5197.
17 Maizels, L., Huber, I., Arbel, G., Tijsen, A.J., Gepstein, A., Khoury, A. and Gepstein, L. (2017) Patient-Specific Drug Screening Using a Human Induced Pluripotent Stem Cell Model of Catecholaminergic Polymorphic Ventricular Tachycardia Type 2. *Circ Arrhythm Electrophysiol*, **10**.
18 Sasaki, K., Makiyama, T., Yoshida, Y., Wuriyanghai, Y., Kamakura, T., Nishiuchi, S., Hayano, M., Harita, T., Yamamoto, Y., Kohjitani, H. *et al.* (2016) Patient-Specific Human Induced Pluripotent Stem Cell Model Assessed with Electrical Pacing Validates S107 as a Potential Therapeutic Agent for Catecholaminergic Polymorphic Ventricular Tachycardia. *PLoS One*, **11**, e0164795.
19 Doulatov, S., Vo, L.T., Macari, E.R., Wahlster, L., Kinney, M.A., Taylor, A.M., Barragan, J., Gupta, M., McGrath, K., Lee, H.Y. *et al.* (2017) Drug discovery for Diamond-Blackfan anemia using reprogrammed hematopoietic progenitors. *Sci Transl Med*, **9**.
20 Cao, L., McDonnell, A., Nitzsche, A., Alexandrou, A., Saintot, P.P., Loucif, A.J., Brown, A.R., Young, G., Mis, M., Randall, A. *et al.* (2016) Pharmacological reversal of a pain phenotype in iPSC-derived sensory neurons and patients with inherited erythromelalgia. *Sci Transl Med*, **8**, 335ra356.
21 Cayo, M.A., Mallanna, S.K., Di Furio, F., Jing, R., Tolliver, L.B., Bures, M., Urick, A., Noto, F.K., Pashos, E.E., Greseth, M.D. *et al.* (2017) A Drug Screen using Human iPSC-Derived Hepatocyte-like Cells Reveals Cardiac Glycosides as a Potential Treatment for Hypercholesterolemia. *Cell stem cell*, **20**, 478-489 e475.
22 Hino, K., Horigome, K., Nishio, M., Komura, S., Nagata, S., Zhao, C., Jin, Y., Kawakami, K., Yamada, Y., Ohta, A. *et al.* (2017) Activin-A enhances mTOR signaling to promote aberrant chondrogenesis in fibrodysplasia ossificans progressiva. *J Clin Invest*, **127**, 3339-3352.
23 Lin, H., Ying, Y., Wang, Y.Y., Wang, G., Jiang, S.S., Huang, D., Luo, L., Chen, Y.G., Gerstenfeld, L.C. and Luo, Z. (2017) AMPK downregulates ALK2 via increasing the interaction between Smurf1 and Smad6, leading to inhibition of osteogenic differentiation. *Biochim Biophys Acta*, **1864**, 2369-2377.
24 Li, M., Zhao, H., Ananiev, G.E., Musser, M.T., Ness, K.H., Maglaque, D.L., Saha, K., Bhattacharyya, A. and Zhao, X. (2017) Establishment of Reporter Lines for Detecting Fragile X Mental Retardation (FMR1) Gene Reactivation in Human Neural Cells. *Stem cells*, **35**, 158-169.
25 Codazzi, F., Hu, A., Rai, M., Donatello, S., Salerno Scarzella, F., Mangiameli, E., Pelizzoni, I., Grohovaz, F. and Pandolfo, M. (2016) Friedreich ataxia-induced pluripotent stem cell-derived neurons show a cellular phenotype that is corrected by a benzamide HDAC inhibitor. *Hum Mol Genet*, **25**, 4847-4855.

26 Almeida, S., Gao, F., Coppola, G. and Gao, F.B. (2016) Suberoylanilide hydroxamic acid increases progranulin production in iPSC-derived cortical neurons of frontotemporal dementia patients. *Neurobiol Aging*, **42**, 35-40.
27 Holler, C.J., Taylor, G., McEachin, Z.T., Deng, Q., Watkins, W.J., Hudson, K., Easley, C.A., Hu, W.T., Hales, C.M., Rossoll, W. *et al.* (2016) Trehalose upregulates progranulin expression in human and mouse models of GRN haploinsufficiency: a novel therapeutic lead to treat frontotemporal dementia. *Mol Neurodegener*, **11**, 46.
28 Silva, M.C., Cheng, C., Mair, W., Almeida, S., Fong, H., Biswas, M.H.U., Zhang, Z., Huang, Y., Temple, S., Coppola, G. *et al.* (2016) Human iPSC-Derived Neuronal Model of Tau-A152T Frontotemporal Dementia Reveals Tau-Mediated Mechanisms of Neuronal Vulnerability. *Stem Cell Reports*, **7**, 325-340.
29 Aflaki, E., Borger, D.K., Moaven, N., Stubblefield, B.K., Rogers, S.A., Patnaik, S., Schoenen, F.J., Westbroek, W., Zheng, W., Sullivan, P. *et al.* (2016) A New Glucocerebrosidase Chaperone Reduces alpha-Synuclein and Glycolipid Levels in iPSC-Derived Dopaminergic Neurons from Patients with Gaucher Disease and Parkinsonism. *The Journal of neuroscience : the official journal of the Society for Neuroscience*, **36**, 7441-7452.
30 Kim, M.J., Jeon, S., Burbulla, L.F. and Krainc, D. (2018) Acid ceramidase inhibition ameliorates alpha-synuclein accumulation upon loss of GBA1 function. *Hum Mol Genet*, in press.
31 Dickey, A.S., Sanchez, D.N., Arreola, M., Sampat, K.R., Fan, W., Arbez, N., Akimov, S., Van Kanegan, M.J., Ohnishi, K., Gilmore-Hall, S.K. *et al.* (2017) PPARdelta activation by bexarotene promotes neuroprotection by restoring bioenergetic and quality control homeostasis. *Sci Transl Med*, **9**.
32 Consortium, H.D.i. (2017) Developmental alterations in Huntington's disease neural cells and pharmacological rescue in cells and mice. *Nat Neurosci*, **20**, 648-660.
33 Lim, R.G., Quan, C., Reyes-Ortiz, A.M., Lutz, S.E., Kedaigle, A.J., Gipson, T.A., Wu, J., Vatine, G.D., Stocksdale, J., Casale, M.S. *et al.* (2017) Huntington's Disease iPSC-Derived Brain Microvascular Endothelial Cells Reveal WNT-Mediated Angiogenic and Blood-Brain Barrier Deficits. *Cell Rep*, **19**, 1365-1377.
34 Sala, L., Yu, Z., Ward-van Oostwaard, D., van Veldhoven, J.P., Moretti, A., Laugwitz, K.L., Mummery, C.L., AP, I.J. and Bellin, M. (2016) A new hERG allosteric modulator rescues genetic and drug-induced long-QT syndrome phenotypes in cardiomyocytes from isogenic pairs of patient induced pluripotent stem cells. *EMBO Mol Med*, **8**, 1065-1081.
35 Portero, V., Casini, S., Hoekstra, M., Verkerk, A.O., Mengarelli, I., Belardinelli, L., Rajamani, S., Wilde, A.A.M., Bezzina, C.R., Veldkamp, M.W. *et al.* (2017) Anti-arrhythmic potential of the late sodium current inhibitor GS-458967 in murine Scn5a-1798insD+/- and human SCN5A-1795insD+/- iPSC-derived cardiomyocytes. *Cardiovasc Res*, **113**, 829-838.
36 Nageshappa, S., Carromeu, C., Trujillo, C.A., Mesci, P., Espuny-Camacho, I., Pasciuto, E., Vanderhaeghen, P., Verfaillie, C.M., Raitano, S., Kumar, A. *et al.* (2016) Altered neuronal network and rescue in a human MECP2 duplication model. *Mol Psychiatry*, **21**, 178-188.

37 Lorenz, C., Lesimple, P., Bukowiecki, R., Zink, A., Inak, G., Mlody, B., Singh, M., Semtner, M., Mah, N., Aure, K. *et al.* (2017) Human iPSC-Derived Neural Progenitors Are an Effective Drug Discovery Model for Neurological mtDNA Disorders. *Cell stem cell*, **20**, 659-674 e659.
38 Sima, N., Li, R., Huang, W., Xu, M., Beers, J., Zou, J., Titus, S., Ottinger, E.A., Marugan, J.J., Xie, X. *et al.* (2018) Neural stem cells for disease modeling and evaluation of therapeutics for infantile (CLN1/PPT1) and late infantile (CLN2/TPP1) neuronal ceroid lipofuscinoses. *Orphanet J Rare Dis*, **13**, 54.
39 Makaryan, V., Kelley, M.L., Fletcher, B., Bolyard, A.A., Aprikyan, A.A. and Dale, D.C. (2017) Elastase inhibitors as potential therapies for ELANE-associated neutropenia. *J Leukoc Biol*, **102**, 1143-1151.
40 Burbulla, L.F., Song, P., Mazzulli, J.R., Zampese, E., Wong, Y.C., Jeon, S., Santos, D.P., Blanz, J., Obermaier, C.D., Strojny, C. *et al.* (2017) Dopamine oxidation mediates mitochondrial and lysosomal dysfunction in Parkinson's disease. *Science*, **357**, 1255-1261.
41 Chen, Z.C., Zhang, W., Chua, L.L., Chai, C., Li, R., Lin, L., Cao, Z., Angeles, D.C., Stanton, L.W., Peng, J.H. *et al.* (2017) Phosphorylation of amyloid precursor protein by mutant LRRK2 promotes AICD activity and neurotoxicity in Parkinson's disease. *Sci Signal*, **10**.
42 Kouroupi, G., Taoufik, E., Vlachos, I.S., Tsioras, K., Antoniou, N., Papastefanaki, F., Chroni-Tzartou, D., Wrasidlo, W., Bohl, D., Stellas, D. *et al.* (2017) Defective synaptic connectivity and axonal neuropathology in a human iPSC-based model of familial Parkinson's disease. *Proceedings of the National Academy of Sciences of the United States of America*, **114**, E3679-E3688.
43 Mittal, S., Bjornevik, K., Im, D.S., Flierl, A., Dong, X., Locascio, J.J., Abo, K.M., Long, E., Jin, M., Xu, B. *et al.* (2017) beta2-Adrenoreceptor is a regulator of the alpha-synuclein gene driving risk of Parkinson's disease. *Science*, **357**, 891-898.
44 Nevin, Z.S., Factor, D.C., Karl, R.T., Douvaras, P., Laukka, J., Windrem, M.S., Goldman, S.A., Fossati, V., Hobson, G.M. and Tesar, P.J. (2017) Modeling the Mutational and Phenotypic Landscapes of Pelizaeus-Merzbacher Disease with Human iPSC-Derived Oligodendrocytes. *Am J Hum Genet*, **100**, 617-634.
45 Sa, S., Gu, M., Chappell, J., Shao, N.Y., Ameen, M., Elliott, K.A., Li, D., Grubert, F., Li, C.G., Taylor, S. *et al.* (2017) Induced Pluripotent Stem Cell Model of Pulmonary Arterial Hypertension Reveals Novel Gene Expression and Patient Specificity. *Am J Respir Crit Care Med*, **195**, 930-941.
46 Ramsden, C.M., Nommiste, B., A, R.L., Carr, A.F., Powner, M.B., M, J.K.S., Chen, L.L., Muthiah, M.N., Webster, A.R., Moore, A.T. *et al.* (2017) Rescue of the MERTK phagocytic defect in a human iPSC disease model using translational read-through inducing drugs. *Sci Rep*, **7**, 51.
47 El-Battrawy, I., Lan, H., Cyganek, L., Zhao, Z., Li, X., Buljubasic, F., Lang, S., Yucel, G., Sattler, K., Zimmermann, W.H. *et al.* (2018) Modeling Short QT Syndrome Using Human-Induced Pluripotent Stem Cell-Derived Cardiomyocytes. *J Am Heart Assoc*, **7**.

48 Lai, J.I., Leman, L.J., Ku, S., Vickers, C.J., Olsen, C.A., Montero, A., Ghadiri, M.R. and Gottesfeld, J.M. (2017) Cyclic tetrapeptide HDAC inhibitors as potential therapeutics for spinal muscular atrophy: Screening with iPSC-derived neuronal cells. *Bioorg Med Chem Lett*, **27**, 3289-3293.
49 Ohuchi, K., Funato, M., Kato, Z., Seki, J., Kawase, C., Tamai, Y., Ono, Y., Nagahara, Y., Noda, Y., Kameyama, T. *et al.* (2016) Established Stem Cell Model of Spinal Muscular Atrophy Is Applicable in the Evaluation of the Efficacy of Thyrotropin-Releasing Hormone Analog. *Stem Cells Transl Med*, **5**, 152-163.
50 Ishida, Y., Kawakami, H., Kitajima, H., Nishiyama, A., Sasai, Y., Inoue, H. and Muguruma, K. (2016) Vulnerability of Purkinje Cells Generated from Spinocerebellar Ataxia Type 6 Patient-Derived iPSCs. *Cell Rep*, **17**, 1482-1490.
51 Song, L., Park, S.E., Isseroff, Y., Morikawa, K. and Yazawa, M. (2017) Inhibition of CDK5 Alleviates the Cardiac Phenotypes in Timothy Syndrome. *Stem Cell Reports*, **9**, 50-57.
52 Llewellyn, K.J., Nalbandian, A., Weiss, L.N., Chang, I., Yu, H., Khatib, B., Tan, B., Scarfone, V. and Kimonis, V.E. (2017) Myogenic differentiation of VCP disease-induced pluripotent stem cells: A novel platform for drug discovery. *PLoS One*, **12**, e0176919.
53 Aguisanda, F., Yeh, C.D., Chen, C.Z., Li, R., Beers, J., Zou, J., Thorne, N. and Zheng, W. (2017) Neural stem cells for disease modeling of Wolman disease and evaluation of therapeutics. *Orphanet J Rare Dis*, **12**, 120.