*pharmaceutics*

MDPI

# Supplementary Materials: Patient-derived Induced Pluripotent Stem Cells (iPSCs) and Cerebral Organoids for Drug Screening and Development in Autism Spectrum Disorder: Opportunities and Challenges

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**Table 1.** Main ASD-associated syndromes.

| Chromosomal Disorders | | | | |
|---|---|---|---|---|
| **Genetic Bases** | **Disorder (#OMIM)** | **Gene(s)** | **Location** | **iPSC Models [ref]** |
| Duplication | Chromosome 1q21.1 duplication syndrome (#612475) | Contiguous gene syndromes | 1q21.1 | n.a. |
| Deletion | Chromosome 1q21.1 deletion syndrome (#612474) | | | |
| Deletion | Chromosome 2q37 deletion syndrome (#600430) | Contiguous gene syndrome | 2q37 | n.a. |
| Duplication | Chromosome 7q11.23 duplication syndrome (#609757) | Contiguous gene syndrome | 7q11.23 | n.a. |
| Deletion | Williams-Beuren syndrome (#194050) | | | |
| Inverted duplication with a terminal deletion | Inverted duplication/deletion 8p21–23 (n.a.) | Multiple genes | 8p21–p23 | n.a. |
| Abnormal methylation, paternal uniparental disomy, point mutation of *CDKN1C*, 11p15.5 rearrangement | Beckwith-Wiedemann syndrome (#130650) | *CDKN1C, H19, IGF2, KCNQ1OT1* | 11p15.5 | n.a. |
| Duplication | Maternal 15q11–q13 duplication syndrome (#608636) | *UBE3A* | 15q11–q13 | n.a. |
| Maternal deletion, paternal uniparental disomy, point mutations of *UBE3A*, imprinting defects | Angelman syndrome (#105830) | | | [1–4] |
| Duplication | Paternal 15q11–q13 duplication syndrome (#608636) | *SNRPN, NDN,* possibly others | 15q11–q13 | n.a. |
| Paternal deletion, maternal uniparental disomy, imprinting defects | Prader-Willy syndrome (#176270) | | | [1] |
| Inverted duplication | Inverted duplicated chromosome 15 syndrome (n.a.) | Multiple genes | 15q11.2-q13.1 | n.a. |
| Duplication | Chromosome 16p11.2 duplication syndrome (#614671) | Contiguous gene syndromes | 16p11.2 | n.a. |
| Deletion | Chromosome 16p11.2 deletion syndrome (#611913) | | | |
| Duplication | Potocki–Lupski syndrome (#610883) | *RAI1,* possibly others | 17p11.2 | n.a. |
| Deletion, point mutation of *RAI1* | Smith–Magenis syndrome (#182290) | | | |
| Trisomy 21 | Down syndrome (#190685) | Multiple genes | chr21 | n.a. |

| Chromosomal Disorders | | | | |
|---|---|---|---|---|
| **Genetic Bases** | **Disorder (#OMIM)** | **Gene(s)** | **Location** | **iPSC Models [ref]** |
| Duplication | Chromosome 22q11.21 duplication syndrome (#608363) | Multiple genes | 22q11.21 | n.a. |
| Deletion, point mutation of *TBX1* | DiGeorge syndrome (#188400) Velocardiofacial syndrome (#192430) | *TBX1*, possibly others | | |
| Duplication | Chromosome 22q13.3 duplication syndrome (#615538) | *SHANK3* | 22q13.3 | n.a. |
| Deletion, point mutation of *SHANK3* | Phelan–McDermid syndrome (#606232) | | | [5–9] |
| Monosomy X | Turner syndrome (n.a.) | Multiple genes | chrX | n.a. |

| Monogenic Disorders | | | | |
|---|---|---|---|---|
| **Genetic Bases** | **Disorder (#OMIM)** | **Gene(s)** | **Location** | **iPSC Models [ref]** |
| Point mutation, deletion | Neurexin 1 (*600565) | *NRXN1* | 2p16.3 | n.a. |
| Point mutation, deletion | Contactin 4 (*607280) | *CNTN4* | 3p26.2–p26.3 | n.a. |
| Point mutation | Protocadherin 10 (*608286) | *PCDH10* | 4q28 | n.a. |
| Point mutation | Cornelia de Lange syndrome (#122470) | *NIPBL* | 5p13.2 | n.a. |
| Point mutation, deletion | Sotos syndrome (#117550) | *NSD1* | 5q35.3 | n.a. |
| Point mutation | Joubert syndrome 3 (#608629) | *AHI1* | 6q23.3 | n.a. |
| Point mutation | CHARGE syndrome (#214800) | *CHD7* | 8q12.2 | n.a. |
| Point mutation | Cohen syndrome (#216550) | *COH1* | 8q22.2 | n.a. |
| Point mutation | Tuberous sclerosis (#191100 and #613254) | *TSC1* *TSC2* | 9q34 16p13.3 | [10–15] |
| Point mutation | Macrocephaly/autism syndrome (#605309) | *PTEN* | 10q23.31 | n.a. |
| Point mutation | Smith–Lemli–Opitz syndrome (#270400) | *DHCR7* | 11q13.4 | n.a. |
| Point mutation | Neurodegeneration due to cerebral folate transport deficiency (#613068) | *FOLR1* | 11q13.4 | n.a. |
| Point mutation | Cell adhesion molecule-1 (*605686) | *CADM1* | 11q23.3 | n.a. |
| Point mutation | Timothy syndrome (#601005) | *CACNA1C* | 12p13.33 | [16–20] |
| Point mutation | Phenylketonuria (#261600) | *PAH* | 12q23.2 | n.a. |
| Point mutation | Noonan syndrome (#163950) | *PTPN11* | 12q24.13 | n.a. |
| Point mutation | Moebius syndrome (%157900) | Possibly *REV3L* and *PLXND1* | 13q12.2-q13 | n.a. |
| Point mutation | FOXG1 deletion syndrome (n.a.) | *FOXG1* | 14q12 | [21] |
| Point mutation | Neurofibromatosis type 1 (#162200) | *NF1* | 17q11.2 | n.a. |
| Point mutation | Mucopolysaccharidosis type IIIA (#252900) | *SGSH* | 17q25.3 | n.a. |
| Point mutation | Helsmoortel–Van der Aa syndrome (#615873) | *ADNP* | 20q13.13 | n.a. |
| Point mutation | Adenylosuccinate deficiency (#103050) | *ADSL* | 22q13.1 | n.a. |
| Point mutation | Cyclin-dependent kinase-like 5 disorder (n.a.) | *CDKL5* | Xp22.13 | [22,23] |
| Point mutation | Aarskog-Scott syndrome (#305400) | *FGD1* | Xp11.22 | n.a. |
| Point mutation | Lujan–Fryns syndrome (#309520) | *MED12* | Xq13.1 | n.a. |
| Point mutation, deletion | Neuroligin 3 (*300336) Neuroligin 4 (*300427) | *NLGN3* *NLGN4* | Xq13.1 Xp22.32-p22.31 | n.a. |
| Triplet repeat expansion, point mutation | Fragile X syndrome (#300624) | *FMR1* | Xq27.3 | [24–29] |
| Point mutation | Cerebral creatine deficiency syndrome 1 (#300352) Cerebral creatine deficiency syndrome 2 (#612736) Cerebral creatine deficiency syndrome 3 (#612718) | *SLC6A8* *GAMT* *GATM* | Xq28 19p13.3 15q21.1 | n.a. |
| Point mutation | Rett syndrome (#312750) | *MECP2* | Xq28 | [30–38] |

n.a.: not available.

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