**Supplementary Table I. Gene association to disease.**

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| **Gene** | **Disease associations** |
| *ABCC8* | Three patients reported with intermediate/ permanent DEND syndrome, transient neonatal diabetes and/or minor dystonia.1-4 |
| *CNTN4* | Autism spectrum disorder.5,6  Candidate gene for SCA16 in a Japanese family (most likely only ascertained to this family, not confirmed in the Japanese population).7,8  One patient with 3p deletion syndrome without neurological symptoms.9  One patient with bilateral optical nerve aplasia.10  Three patients with speech impairment, motor, and cognitive delay; two of these patients had seizures.11 |
| *CTNNA3* | Late-onset Alzheimer’s disease in women.12  Susceptibility to ADHD.13  Three patients from one family with essential tremor (rare in the general population).14,15 |
| *FHIT* | Tourette syndrome and autism.16  Major depressive disorder.17  Susceptibility for multiple sclerosis.18 |
| *GAP43* | Candidate gene for 3q13.2–q13.31 deletion syndrome.19 |
| *KCNJ11* | Moderate intellectual disability (IQ 40-55), (intermediate) DEND, impaired visuomotor integration, autism, and ADHD in V59M mutations.3,21-23 Milder features in non-V59M mutations.21 The majority of the patients described with a mutation in KCNJ11 and a DCD diagnosis had non-V59M mutations.4 |
| *KLF7* | Candidate gene for 2q33.3q34 deletion.24  Candidate gene for mild mental impairment in children (IQ 55-60, 70-75).25  One patient with Rett-like features (bruxism and repetitive movements of the hand).26 |
| *LSAMP* | Schizophrenia and bipolar disorder.27  Candidate gene for Meniere's disease in one family.27,28 |
| *PTPRN2* | Candidate gene for ADHD.29 |
| *RBFOX1* | Autism and ADHD.16  Increased risk for generalized epilepsy.16,30 |
| *SHANK3* | Autism.31  Schizophrenia, bipolar disorder, catatonia.32,33  Epilepsy.33  One patient with two *de novo* mutations (frameshift and missense) with ataxia, autism, delayed motor- and language development.34  Haploinsufficiency causes Phelan-McDermid syndrome.35 |
| *VIPR2* | Candidate gene for holoprosencephaly.36  Schizophrenia.37  One patient with autism, intellectual disability, and no speech.38 |

**Footnote**. List of 12 DCD-associated genes and their disease associations. References, indicated with superscript numbers, can be found below. DEND: developmental delay, epilepsy and neonatal diabetes; SCA16: spinocerebellar ataxia type 16; ADHD: attention deficit hyperactivity disorder; IQ: intelligence quotient.

**Supplementary References**

1. Proks P, Arnold AL, Bruining J, et al. A heterozygous activating mutation in the sulphonylurea receptor SUR1 (ABCC8) causes neonatal diabetes. *Hum Mol Genet*. 2006;15(11):1793–1800
2. Babenko AP, Polak M, Cavé H, et al. Activating mutations in the ABCC8 gene in neonatal diabetes mellitus. *N Engl J Med.* 2006;355(5):456-466
3. Patch AM, Flanagan SE, Boustred C, Hattersley AT, Ellard S. Mutations in the ABCC8 gene encoding the SUR1 subunit of the KATP channel cause transient neonatal diabetes, permanent neonatal diabetes or permanent diabetes diagnosed outside the neonatal period. *Diabetes Obes Metab*.2007;9:28-39
4. Busiah K, Drunat S, Vaivre-Douret L, et al. Neuropsychological dysfunction and developmental defects associated with genetic changes in infants with neonatal diabetes mellitus: a prospective cohort study. *Lancet Diabetes Endocrinol*. 2013;1(3):199-207.
5. Roohi J, Montagna C, Tegay DH, et al. Disruption of contactin 4 in three subjects with autism spectrum disorder. *J Med Genet*. 2009;46: 176-182
6. Zuko A, Kleijer KT, Oguro-Ando A, et al. Contactins in the neurobiology of autism. *Eur J Pharmacol*. 2013;719(1-3):63-74
7. Miura S, Shibata H, Furuya H, et al. The contactin 4 gene locus at 3p26 is a candidate gene of SCA16. *Neurology*. 2006;67(7):1236-1241
8. Tanaka E, Maruyama H, Morino H, Nakajima E, Kawakami H. The CNTN4 c.4256C> T mutation is rare in Japanese with inherited spinocerebellar ataxia. *J Neurol Sci.* 2008;266(1-2):180-181
9. Fernandez T, Morgan T, Davis N, et al. Disruption of contactin 4 (CNTN4) results in developmental delay and other features of 3p deletion syndrome. *Am J Hum Genet*. 2004;74(6):1286-1293
10. Prasov L, Masud T, Khaliq S, et al. ATOH7 mutations cause autosomal recessive persistent hyperplasia of the primary vitreous. *Hum Mol Genet*. 2012;21(16):3681–3694
11. Zhang SQ, Fleischer J, Al-Kateb H, Mito Y, Amarillo I, Shinawi M. Intragenic CNTN4 copy number variants associated with a spectrum of neurobehavioral phenotypes. *Eur J Med Genet.* 2020;63(3):103736
12. Miyashita A, Arai H, Asada T,et al. Genetic association of CTNNA3 with late-onset Alzheimer's disease in females. *Hum Mol Genet*. 2007;16(23):2854-69
13. Bacchelli E, Ceroni F, Pinto D, et al. A CTNNA3 compound heterozygous deletion implicates a role for αT-catenin in susceptibility to autism spectrum disorder. *J Neurodev Disord*. 2014;6(1):17
14. Houle G, Ambalavanan A, Schmouth JF, et al. No rare deleterious variants from STK32B, PPARGC1A, and CTNNA3 are associated with essential tremor. *Neurol Genet*. 2017;3(5):e195
15. Müller SH, Girard SL, Hopfner F, et al. Genome-wide association study in essential tremor identifies three new loci. *Brain*. 2016;139(12):3163-3169
16. Mosca SJ, Langevin LM, Dewey D, et al. Copy-number variations are enriched for neurodevelopmental genes in children with developmental coordination disorder. *J Med Genet.* 2016;53(12):812-819
17. Direk N, Williams S, Smith JA, et al. An Analysis of Two Genome-wide Association Meta-analyses Identifies a New Locus for Broad Depression Phenotype. *Biol Psychiatry*. 2017;82(5):322–329
18. Mahurkar S, Moldovan M, Suppiah V, et al. Response to interferon-beta treatment in multiple sclerosis patients: a genome-wide association study. *Pharmacogenomics J.* 2017;17(4):312-318
19. Shuvarikov A, Campbell IM, Dittwald P, et al. Recurrent HERV‐H‐Mediated 3q13. 2–q13. 31 Deletions Cause a Syndrome of Hypotonia and Motor, Language, and Cognitive Delays. *Hum Mutat*. 2013;34(10):1415-1423
20. Svalastoga P, Sulen Å, Fehn JR, et al. Intellectual Disability in KATP Channel Neonatal Diabetes. *Diabetes Care.* 2020*;*43(3):526–533
21. Slingerland AS, Nuboer R, Hadders-Algra M, Hattersley AT, Bruining GJ. Improved motor development and good long-term glycaemic control with sulfonylurea treatment in a patient with the syndrome of intermediate developmental delay, early-onset generalised epilepsy and neonatal diabetes associated with the V59M mutation in the KCNJ11 gene. *Diabetologia*. 2006;49:2559–2563
22. Landmeier KA, Lanning M, Carmody D, Greeley SAW, Msall ME. ADHD, learning difficulties and sleep disturbances associated with KCNJ11‐related neonatal diabetes. *Pediatr Diabete*s. 2017;18(7):518-523
23. Bowman P, Broadbridge E, Knight BA, et al. Psychiatric morbidity in children with KCNJ 11 neonatal diabetes. *Diabet Med.* 2016;33(10):1387-1391
24. Powis Z, Petrik I, Cohen JS, et al. De novo variants in KLF7 are a potential novel cause of developmental delay/intellectual disability, neuromuscular and psychiatric symptoms. *Clin Genet.* 2018;93(5):1030-1038
25. Butcher LM, Maburn E, Knight J, et al. SNPs, microarrays and pooled DNA: identification of four loci associated with mild mental impairment in a sample of 6000 children. *Hum Mol Genet.* 2005;14(10):1315–1325
26. Jang DH, Chae H, Kim M. Autistic and Rett‐like features associated with 2q33.3–q34 interstitial deletion. *Am J Hum Genet Part A.* 2015;167(9):2213-2218
27. Escalera-Balsera A, Roman-Naranjo P, Lopez-Escamez JA. Systematic Review of Sequencing Studies and Gene Expression Profiling in Familial Meniere Disease. *Genes*. 2020;11(12):1414
28. Mehrjoo Z, Kahrizi K, Mohseni M, et al. Limbic System Associated Membrane Protein Mutation in an Iranian Family Diagnosed with Ménière’s Disease. *Arch Iran Med*. 2020;23(5):319-325
29. Lionel AC, Crosbie J, Barbosa N, et al. Rare copy number variation discovery and cross-disorder comparisons identify risk genes for ADHD. *Sci Transl Med.* 2011;3(95):95ra75-95ra75
30. Lal D, Trucks H, Moller RS, et al. Rare exonic deletions of the RBFOX1 gene increase risk of idiopathic generalized epilepsy. *Epilepsia*.2013;54(2):265-271
31. Bruno LP, Doddato G, Valentino F, et al. New Candidates for Autism/Intellectual Disability Identified by Whole-Exome Sequencing. *Int J Mol Sci.* 2021;22(24):13439
32. Accogli A, Yang R, Blain-Juste ME, Braverman N, Shah J, Trakadis Y. SHANK3 Mutation and Mosaic Turner Syndrome in a Female Patient With Intellectual Disability and Psychiatric Features. *J Neuropsychiatry Clin Neurosci.* 2019;31(3):272-275
33. Kolevzon A, Delaby E, Berry-Kravis E, Buxbaum JD, Betancur C. Neuropsychiatric decompensation in adolescents and adults with Phelan-McDermid syndrome: a systematic review of the literature. *Mol Autism.* 2019;10(1):1-22
34. Zhu W, Li J, Chen S, et al. Two de novo novel mutations in one SHANK3 allele in a patient with autism and moderate intellectual disability. *Am J Hum Genet Part A.* 2018;176(4):973-979
35. Gong X, Jiang Y, Zhang X, et al. High proportion of 22q13 deletions and SHANK3 mutations in Chinese patients with intellectual disability. *PloS One*. 2012;7(4):e34739
36. Fang X, Zhang J, Lu T, et al. Identification of novel candidate pathogenic genes in pituitary stalk interruption syndrome by whole‐exome sequencing. *J Cell Mol Med* 24.20 (2020): 11703-11717.
37. Vacic V, McCarthy S, Malhotra D, et al. Duplications of the neuropeptide receptor gene VIPR2 confer significant risk for schizophrenia. *Nature.* 2011;471(7339):499-503
38. Firouzabadi SG, Kariminejad R, Vameghi R, et al. Copy Number Variants in Patients with Autism and Additional Clinical Features: Report of VIPR2 Duplication and a Novel Microduplication Syndrome. *Mol Neurobiol.* 2017;54:7019–7027