

Supplemental Table 1: Epigenetic genes associated with ASDs, continued. For these genes, no direct experimental evidence implicates DNA methylation as impacting their function or regulation. This list includes transcription factors and DNA binding proteins for which no methylated DNA binding data exists, as well as histone modifiers and remodelers. SFARI score refers to the score designated by Simons Foundation Autism Reference Initiative based on clinical data: 1-2, strong evidence; 3-4, suggestive/minimal evidence; 5, hypothesized but untested

Gene	Description	SFARI score	Comorbidities	References
ANKRD11	Interacts with histone deacetylases and p160 coactivators	2	KBG syndrome	(1-12)
ARID1B	Chromatin remodeling, SWI/SNF complexes	1	Coffin-Siris syndrome 1	(2; 6; 8; 9; 12-24)
ASH1L	H3K36 methyltransferase, associates with HOX genes	1	Intellectual disability	(1; 2; 9; 12; 21; 25-27)
ASXL3	Putative polycomb protein	1	Bainbridge-Ropers syndrome	(2; 11; 12; 26; 28-30)
CHD1	Chromatin remodeling	4	Pilarowski-Bjornsson syndrome	(1; 31)
CHD2	Chromatin remodeling	1	Epileptic encephalopathy (childhood-onset)	(1; 2; 9; 11; 12; 21; 23; 26; 31-34)
CHD5	Chromatin remodeling	5		(10; 26; 35)
CHD8	Chromatin remodeling	1		(1; 2; 6; 9; 12; 16; 19; 21; 23; 26; 29; 36-42)
CUX1	DNA binding transcription factor	3		(8; 26)
EHMT1	H3K9 methyltransferase, E2F6 transcriptional repression complex component	3	Kleefstra syndrome	(20; 37; 43-46)
ELP4	HAT component; RNA PolII elongator complex member	3	Aniridia 2, Rolandic epilepsy	(8; 12; 47-49)
EMSY	Interacts with chromatin remodeling complex, interacts with BRCA2	3	Breast and ovarian cancers	(2; 26; 50)
EP400	Exchanges histones for variants, may alter nucleosome/DNA interactions	3		(1; 2; 26; 51; 52)

FOXP1	DNA binding domain, TF, noted as to likely bind CpG sequences	2	Intellectual disability	(2; 5; 8; 9; 12; 21; 23; 42; 53-59)
FOXP2	DNA binding domain, TF	3	Speech-language disorder-1	(60-68)
HDAC3	Histone deacetylase, interacts with DAXX	5		(2)
HIST1H1E	Histone linker protein	N/A	Rahman syndrome	(69)
HMGN1	Binds nucleosome core particles, alters interaction between DNA and histone octamer, associated with active chromatin	3		(70)
HNRNPU	Binds DNA and RNA, involved in several cellular processes such as nuclear chromatin organization, telomere-length regulation, transcription, mRNA alternative splicing and stability, Xist-mediated transcriptional silencing and mitotic cell progression	4	Epileptic encephalopathy (early infantile, 54)	(9; 11; 23)
KAT2B	Histone acetyltransferase for lysine, associates with p300/CBP	2		(1; 2; 9; 33)
KAT6A	H3K9 histone acetyltransferase	3	Intellectual disability	(1; 2)
KDM5B (JARID1B)	H3K4 demethylase	2		(1; 12; 18; 26; 42; 71)
KDM5C (JARID1C)	H3K4 histone demethylase	3	X linked mental retardation	(20; 65; 72)
KDM6B (JMJD3)	H3K27 histone demethylase; may link T-box factors to SWI/SWF remodeling complex	3		(1; 2; 12; 26; 44; 73)
KMT2A	H3K4 methyltransferase, binds to unmethylated promoter CpGs to maintain unmethylated state	1	MLL, Wiedemann-Steiner syndrome	(1; 2; 6; 12; 26)
KMT2C	H3K4 methyltransferase, contains a DNA binding domain	2	Kleefstra syndrome 2	(1; 2; 9; 12; 16; 18; 23; 26; 52; 74)
KMT2E	H3K4me1 and H3K4me2 methyltransferase	3		(9; 73; 75)
KMT5B	H4K20 methyltransferase	1	Intellectual disability	(2; 12; 21; 26; 73; 76)
PHF2	Lysine demethylase for H3K9me2, complex with ARID5B	3		(1; 2; 9; 12; 73)
RFX3	TF with winged helix DNA binding domain, roles in immune response, viral/cellular transcriptional regulation, can work in complex with MDBP	4		(18; 26; 29; 77)

SATB2	DNA binding, specifically binds nuclear matrix attachment regions, no evidence that methylation affects this binding	4	Intellectual disability, craniofacial abnormalities, Glass syndrome	(6; 37; 41; 44; 78; 79)
SETD2	H3K36 methyltransferase	3	Luscan-Lumish syndrome	(6; 7; 15; 16; 19; 20; 80; 81)
SETDB1	H3K9 methyltransferase	4		(82; 83)
SETDB2	Histone methyltransferase	4		(84)
SIN3A	MECP2 and histone interacting protein	4	Witteveen-Kolk syndrome	(1; 26; 85)
SMAD4	DNA binding, binds SMAD-binding element, tumor suppressor	3	Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, Myhre syndrome, Pancreatic cancer (somatic), Polyposis (juvenile intestinal)	(1; 79)
SMARCC2	Regulates transcription via SWI/SNF chromatin remodeling	2		(2; 31; 73)
SRCAP	Chromatin remodeling complex, Transcriptional activator, incorporates H2AZ	2	Floating-Harbor syndrome	(1; 9; 12; 21; 26)
TBX1	DNA binding protein (t box gene family), needed for proper development	4	Conotruncal anomaly face syndrome, Digeorge syndrome, Tetralogy of Fallot, Velocardiofacial syndrome	(86)
TSN	Binds DNA at breaks for recombination	5		(87; 88)
USP7	Hydrolase that deubiquitinates DNMT1 and DAXX too	2	neurodevelopmental disorder	(1; 2; 26; 89)
WAC	Regulates H2BK120ub1	2	Desanto-shinawi syndrome	(1; 2; 9; 12; 27)
ZMYND11	Transcriptional corepressor by modulating RNA pol2 and H3K36me3	3	Intellectual disability	(2; 73; 90)

References:

1. Iossifov I, O'Roak BJ, Sanders SJ, Ronemus M, Krumm N, et al. 2014. The contribution of de novo coding mutations to autism spectrum disorder. *Nature* 515:216-21
2. Iossifov I, Levy D, Allen J, Ye K, Ronemus M, et al. 2015. Low load for disruptive mutations in autism genes and their biased transmission. *Proc Natl Acad Sci U S A* 112:E5600-7
3. Marshall CR, Noor A, Vincent JB, Lionel AC, Feuk L, et al. 2008. Structural variation of chromosomes in autism spectrum disorder. *Am J Hum Genet* 82:477-88
4. Willemsen MH, Fernandez BA, Bacino CA, Gerkes E, de Brouwer AP, et al. 2010. Identification of ANKRD11 and ZNF778 as candidate genes for autism and variable cognitive impairment in the novel 16q24.3 microdeletion syndrome. *Eur J Hum Genet* 18:429-35
5. Brett M, McPherson J, Zang ZJ, Lai A, Tan ES, et al. 2014. Massively parallel sequencing of patients with intellectual disability, congenital anomalies and/or autism spectrum disorders with a targeted gene panel. *PLoS One* 9:e93409
6. Deciphering Developmental Disorders S. 2015. Large-scale discovery of novel genetic causes of developmental disorders. *Nature* 519:223-8
7. Butler MG, Rafi SK, Hossain W, Stephan DA, Manzardo AM. 2015. Whole exome sequencing in females with autism implicates novel and candidate genes. *Int J Mol Sci* 16:1312-35
8. Doan RN, Bae BI, Cubelos B, Chang C, Hossain AA, et al. 2016. Mutations in Human Accelerated Regions Disrupt Cognition and Social Behavior. *Cell* 167:341-54 e12
9. Wang T, Guo H, Xiong B, Stessman HA, Wu H, et al. 2016. De novo genic mutations among a Chinese autism spectrum disorder cohort. *Nat Commun* 7:13316
10. Takata A, Miyake N, Tsurusaki Y, Fukai R, Miyatake S, et al. 2018. Integrative Analyses of De Novo Mutations Provide Deeper Biological Insights into Autism Spectrum Disorder. *Cell Rep* 22:734-47
11. Bowling KM, Thompson ML, Amaral MD, Finnila CR, Hiatt SM, et al. 2017. Genomic diagnosis for children with intellectual disability and/or developmental delay. *Genome Med* 9:43
12. RK CY, Merico D, Bookman M, J LH, Thiruvahindrapuram B, et al. 2017. Whole genome sequencing resource identifies 18 new candidate genes for autism spectrum disorder. *Nat Neurosci* 20:602-11
13. Nord AS, Roeb W, Dickel DE, Walsh T, Kusenda M, et al. 2011. Reduced transcript expression of genes affected by inherited and de novo CNVs in autism. *Eur J Hum Genet* 19:727-31
14. Halgren C, Kjaergaard S, Bak M, Hansen C, El-Schich Z, et al. 2012. Corpus callosum abnormalities, intellectual disability, speech impairment, and autism in patients with haploinsufficiency of ARID1B. *Clin Genet* 82:248-55

15. O'Roak BJ, Vives L, Fu W, Egertson JD, Stanaway IB, et al. 2012. Multiplex targeted sequencing identifies recurrently mutated genes in autism spectrum disorders. *Science* 338:1619-22
16. O'Roak BJ, Vives L, Girirajan S, Karakoc E, Krumm N, et al. 2012. Sporadic autism exomes reveal a highly interconnected protein network of de novo mutations. *Nature* 485:246-50
17. Turner TN, Hormozdiari F, Duyzend MH, McClymont SA, Hook PW, et al. 2016. Genome Sequencing of Autism-Affected Families Reveals Disruption of Putative Noncoding Regulatory DNA. *Am J Hum Genet* 98:58-74
18. Krumm N, Turner TN, Baker C, Vives L, Mohajeri K, et al. 2015. Excess of rare, inherited truncating mutations in autism. *Nat Genet* 47:582-8
19. D'Gama AM, Pochareddy S, Li M, Jamuar SS, Reiff RE, et al. 2015. Targeted DNA Sequencing from Autism Spectrum Disorder Brains Implicates Multiple Genetic Mechanisms. *Neuron* 88:910-7
20. Alvarez-Mora MI, Calvo Escalona R, Puig Navarro O, Madrigal I, Quintela I, et al. 2016. Comprehensive molecular testing in patients with high functioning autism spectrum disorder. *Mutat Res* 784-785:46-52
21. Stessman HA, Xiong B, Coe BP, Wang T, Hoekzema K, et al. 2017. Targeted sequencing identifies 91 neurodevelopmental-disorder risk genes with autism and developmental-disability biases. *Nat Genet* 49:515-26
22. Turner TN, Coe BP, Dickel DE, Hoekzema K, Nelson BJ, et al. 2017. Genomic Patterns of De Novo Mutation in Simplex Autism. *Cell* 171:710-22 e12
23. Lim ET, Uddin M, De Rubeis S, Chan Y, Kamumbu AS, et al. 2017. Rates, distribution and implications of postzygotic mosaic mutations in autism spectrum disorder. *Nat Neurosci* 20:1217-24
24. Zahir FR, Mwenifumbo JC, Chun HE, Lim EL, Van Karnebeek CDM, et al. 2017. Comprehensive whole genome sequence analyses yields novel genetic and structural insights for Intellectual Disability. *BMC Genomics* 18:403
25. Willsey AJ, Sanders SJ, Li M, Dong S, Tebbenkamp AT, et al. 2013. Coexpression networks implicate human midfetal deep cortical projection neurons in the pathogenesis of autism. *Cell* 155:997-1007
26. De Rubeis S, He X, Goldberg AP, Poultney CS, Samocha K, et al. 2014. Synaptic, transcriptional and chromatin genes disrupted in autism. *Nature* 515:209-15
27. Tammimies K, Marshall CR, Walker S, Kaur G, Thiruvahindrapuram B, et al. 2015. Molecular Diagnostic Yield of Chromosomal Microarray Analysis and Whole-Exome Sequencing in Children With Autism Spectrum Disorder. *JAMA* 314:895-903
28. Dinwiddie DL, Soden SE, Saunders CJ, Miller NA, Farrow EG, et al. 2013. De novo frameshift mutation in ASXL3 in a patient with global developmental delay, microcephaly, and craniofacial anomalies. *BMC Med Genomics* 6:32
29. Li J, Wang L, Guo H, Shi L, Zhang K, et al. 2017. Targeted sequencing and functional analysis reveal brain-size-related genes and their networks in autism spectrum disorders. *Mol Psychiatry* 22:1282-90

30. Zhao JJ, Halvardson J, Zander CS, Zaghlool A, Georgii-Hemming P, et al. 2018. Exome sequencing reveals NAA15 and PUF60 as candidate genes associated with intellectual disability. *Am J Med Genet B Neuropsychiatr Genet* 177:10-20
31. Neale BM, Kou Y, Liu L, Ma'ayan A, Samocha KE, et al. 2012. Patterns and rates of exonic de novo mutations in autism spectrum disorders. *Nature* 485:242-5
32. Carvill GL, Heavin SB, Yendle SC, McMahon JM, O'Roak BJ, et al. 2013. Targeted resequencing in epileptic encephalopathies identifies de novo mutations in CHD2 and SYNGAP1. *Nat Genet* 45:825-30
33. Pinto D, Delaby E, Merico D, Barbosa M, Merikangas A, et al. 2014. Convergence of genes and cellular pathways dysregulated in autism spectrum disorders. *Am J Hum Genet* 94:677-94
34. Lebrun N, Parent P, Gendras J, Billuart P, Poirier K, Bienvenu T. 2017. Autism spectrum disorder recurrence, resulting of germline mosaicism for a CHD2 gene missense variant. *Clin Genet* 92:669-70
35. Pisansky MT, Young AE, O'Connor MB, Gottesman, II, Bagchi A, Gewirtz JC. 2017. Mice lacking the chromodomain helicase DNA-binding 5 chromatin remodeler display autism-like characteristics. *Transl Psychiatry* 7:e1152
36. Bernier R, Golzio C, Xiong B, Stessman HA, Coe BP, et al. 2014. Disruptive CHD8 mutations define a subtype of autism early in development. *Cell* 158:263-76
37. Talkowski ME, Rosenfeld JA, Blumenthal I, Pillalamarri V, Chiang C, et al. 2012. Sequencing chromosomal abnormalities reveals neurodevelopmental loci that confer risk across diagnostic boundaries. *Cell* 149:525-37
38. Prontera P, Ottaviani V, Toccaceli D, Rogaia D, Ardisia C, et al. 2014. Recurrent approximately 100 Kb microdeletion in the chromosomal region 14q11.2, involving CHD8 gene, is associated with autism and macrocephaly. *Am J Med Genet A* 164A:3137-41
39. Merner N, Forgeot d'Arc B, Bell SC, Maussion G, Peng H, et al. 2016. A de novo frameshift mutation in chromodomain helicase DNA-binding domain 8 (CHD8): A case report and literature review. *Am J Med Genet A* 170A:1225-35
40. Stolerman ES, Smith B, Chaubey A, Jones JR. 2016. CHD8 intragenic deletion associated with autism spectrum disorder. *Eur J Med Genet* 59:189-94
41. Redin C, Brand H, Collins RL, Kammin T, Mitchell E, et al. 2017. The genomic landscape of balanced cytogenetic abnormalities associated with human congenital anomalies. *Nat Genet* 49:36-45
42. Chen R, Davis LK, Guter S, Wei Q, Jacob S, et al. 2017. Leveraging blood serotonin as an endophenotype to identify de novo and rare variants involved in autism. *Mol Autism* 8:14
43. Kleefstra T, Brunner HG, Amiel J, Oudakker AR, Nillesen WM, et al. 2006. Loss-of-function mutations in euchromatin histone methyl transferase 1 (EHMT1) cause the 9q34 subtelomeric deletion syndrome. *Am J Hum Genet* 79:370-7
44. Jiang YH, Yuen RK, Jin X, Wang M, Chen N, et al. 2013. Detection of clinically relevant genetic variants in autism spectrum disorder by whole-genome sequencing. *Am J Hum Genet* 93:249-63

45. Balan S, Iwayama Y, Maekawa M, Toyota T, Ohnishi T, et al. 2014. Exon resequencing of H3K9 methyltransferase complex genes, EHMT1, EHTM2 and WIZ, in Japanese autism subjects. *Mol Autism* 5:49
46. Anazi S, Maddirevula S, Salpietro V, Asi YT, Alsahli S, et al. 2017. Expanding the genetic heterogeneity of intellectual disability. *Hum Genet* 136:1419-29
47. Strug LJ, Clarke T, Chiang T, Chien M, Baskurt Z, et al. 2009. Centrotemporal sharp wave EEG trait in rolandic epilepsy maps to Elongator Protein Complex 4 (ELP4). *Eur J Hum Genet* 17:1171-81
48. Addis L, Ahn JW, Dobson R, Dixit A, Ogilvie CM, et al. 2015. Microdeletions of ELP4 Are Associated with Language Impairment, Autism Spectrum Disorder, and Mental Retardation. *Hum Mutat* 36:842-50
49. Davis LK, Meyer KJ, Rudd DS, Librant AL, Epping EA, et al. 2008. Pax6 3' deletion results in aniridia, autism and mental retardation. *Hum Genet* 123:371-8
50. Gupta S, Ellis SE, Ashar FN, Moes A, Bader JS, et al. 2014. Transcriptome analysis reveals dysregulation of innate immune response genes and neuronal activity-dependent genes in autism. *Nat Commun* 5:5748
51. Chahrour MH, Yu TW, Lim ET, Ataman B, Coulter ME, et al. 2012. Whole-exome sequencing and homozygosity analysis implicate depolarization-regulated neuronal genes in autism. *PLoS Genet* 8:e1002635
52. Krupp DR, Barnard RA, Duffourd Y, Evans SA, Mulqueen RM, et al. 2017. Exonic Mosaic Mutations Contribute Risk for Autism Spectrum Disorder. *Am J Hum Genet* 101:369-90
53. Autism Spectrum Disorders Working Group of The Psychiatric Genomics C. 2017. Meta-analysis of GWAS of over 16,000 individuals with autism spectrum disorder highlights a novel locus at 10q24.32 and a significant overlap with schizophrenia. *Mol Autism* 8:21
54. O'Roak BJ, Deriziotis P, Lee C, Vives L, Schwartz JJ, et al. 2011. Exome sequencing in sporadic autism spectrum disorders identifies severe de novo mutations. *Nat Genet* 43:585-9
55. Lozano R, Vino A, Lozano C, Fisher SE, Deriziotis P. 2015. A de novo FOXP1 variant in a patient with autism, intellectual disability and severe speech and language impairment. *Eur J Hum Genet* 23:1702-7
56. Hamdan FF, Daoud H, Rochefort D, Piton A, Gauthier J, et al. 2010. De novo mutations in FOXP1 in cases with intellectual disability, autism, and language impairment. *Am J Hum Genet* 87:671-8
57. Palumbo O, D'Agruma L, Minenna AF, Palumbo P, Stallone R, et al. 2013. 3p14.1 de novo microdeletion involving the FOXP1 gene in an adult patient with autism, severe speech delay and deficit of motor coordination. *Gene* 516:107-13
58. Girirajan S, Dennis MY, Baker C, Malig M, Coe BP, et al. 2013. Refinement and discovery of new hotspots of copy-number variation associated with autism spectrum disorder. *Am J Hum Genet* 92:221-37
59. Chien WH, Gau SS, Chen CH, Tsai WC, Wu YY, et al. 2013. Increased gene expression of FOXP1 in patients with autism spectrum disorders. *Mol Autism* 4:23

60. Newbury DF, Bonora E, Lamb JA, Fisher SE, Lai CS, et al. 2002. FOXP2 is not a major susceptibility gene for autism or specific language impairment. *Am J Hum Genet* 70:1318-27
61. Wassink TH, Piven J, Vieland VJ, Pietila J, Goedken RJ, et al. 2002. Evaluation of FOXP2 as an autism susceptibility gene. *Am J Med Genet* 114:566-9
62. Gauthier J, Joobor R, Mottron L, Laurent S, Fuchs M, et al. 2003. Mutation screening of FOXP2 in individuals diagnosed with autistic disorder. *Am J Med Genet A* 118A:172-5
63. Marui T, Koishi S, Funatogawa I, Yamamoto K, Matsumoto H, et al. 2005. No association of FOXP2 and PTPRZ1 on 7q31 with autism from the Japanese population. *Neurosci Res* 53:91-4
64. Chien YL, Wu YY, Chiu YN, Liu SK, Tsai WC, et al. 2011. Association study of the CNS patterning genes and autism in Han Chinese in Taiwan. *Prog Neuropsychopharmacol Biol Psychiatry* 35:1512-7
65. Toma C, Hervas A, Torrico B, Balmana N, Salgado M, et al. 2013. Analysis of two language-related genes in autism: a case-control association study of FOXP2 and CNTNAP2. *Psychiatr Genet* 23:82-5
66. Li H, Yamagata T, Mori M, Momoi MY. 2005. Absence of causative mutations and presence of autism-related allele in FOXP2 in Japanese autistic patients. *Brain Dev* 27:207-10
67. Chien YL, Wu YY, Chen HI, Tsai WC, Chiu YN, et al. 2017. The central nervous system patterning gene variants associated with clinical symptom severity of autism spectrum disorders. *J Formos Med Assoc* 116:755-64
68. Gong X, Jia M, Ruan Y, Shuang M, Liu J, et al. 2004. Association between the FOXP2 gene and autistic disorder in Chinese population. *Am J Med Genet B Neuropsychiatr Genet* 127B:113-6
69. Duffney LJ, Valdez P, Tremblay MW, Cao X, Montgomery S, et al. 2018. Epigenetics and autism spectrum disorder: A report of an autism case with mutation in H1 linker histone HIST1H1E and literature review. *Am J Med Genet B Neuropsychiatr Genet* 177:426-33
70. Abuhatzira L, Shamir A, Schones DE, Schaffer AA, Bustin M. 2011. The chromatin-binding protein HMGN1 regulates the expression of methyl CpG-binding protein 2 (MECP2) and affects the behavior of mice. *J Biol Chem* 286:42051-62
71. Al-Mubarak B, Abouelhoda M, Omar A, Aldhalaan H, Aldosari M, et al. 2017. Whole exome sequencing reveals inherited and de novo variants in autism spectrum disorder: a trio study from Saudi families. *Sci Rep* 7:5679
72. Adegbola A, Gao H, Sommer S, Browning M. 2008. A novel mutation in JARID1C/SMCX in a patient with autism spectrum disorder (ASD). *Am J Med Genet A* 146A:505-11
73. Iossifov I, Ronemus M, Levy D, Wang Z, Hakker I, et al. 2012. De novo gene disruptions in children on the autistic spectrum. *Neuron* 74:285-99
74. Freed D, Pevsner J. 2016. The Contribution of Mosaic Variants to Autism Spectrum Disorder. *PLoS Genet* 12:e1006245

75. Dong S, Walker MF, Carriero NJ, DiCola M, Willsey AJ, et al. 2014. De novo insertions and deletions of predominantly paternal origin are associated with autism spectrum disorder. *Cell Rep* 9:16-23
76. Sanders SJ, Murtha MT, Gupta AR, Murdoch JD, Raubeson MJ, et al. 2012. De novo mutations revealed by whole-exome sequencing are strongly associated with autism. *Nature* 485:237-41
77. Tabet AC, Verloes A, Pilorge M, Delaby E, Delorme R, et al. 2015. Complex nature of apparently balanced chromosomal rearrangements in patients with autism spectrum disorder. *Mol Autism* 6:19
78. Coe BP, Witherspoon K, Rosenfeld JA, van Bon BW, Vulto-van Silfhout AT, et al. 2014. Refining analyses of copy number variation identifies specific genes associated with developmental delay. *Nat Genet* 46:1063-71
79. Geisheker MR, Heymann G, Wang T, Coe BP, Turner TN, et al. 2017. Hotspots of missense mutation identify neurodevelopmental disorder genes and functional domains. *Nat Neurosci* 20:1043-51
80. Fromer M, Pocklington AJ, Kavanagh DH, Williams HJ, Dwyer S, et al. 2014. De novo mutations in schizophrenia implicate synaptic networks. *Nature* 506:179-84
81. Lumish HS, Wynn J, Devinsky O, Chung WK. 2015. Brief Report: SETD2 Mutation in a Child with Autism, Intellectual Disabilities and Epilepsy. *J Autism Dev Disord* 45:3764-70
82. Cukier HN, Rabionet R, Konidari I, Rayner-Evans MY, Baltos ML, et al. 2010. Novel variants identified in methyl-CpG-binding domain genes in autistic individuals. *Neurogenetics* 11:291-303
83. Jiang Y, Loh YE, Rajarajan P, Hirayama T, Liao W, et al. 2017. The methyltransferase SETDB1 regulates a large neuron-specific topological chromatin domain. *Nat Genet* 49:1239-50
84. Cukier HN, Lee JM, Ma D, Young JI, Mayo V, et al. 2012. The expanding role of MBD genes in autism: identification of a MECP2 duplication and novel alterations in MBD5, MBD6, and SETDB1. *Autism Res* 5:385-97
85. Farwell Hagman KD, Shinde DN, Mroske C, Smith E, Radtke K, et al. 2017. Candidate-gene criteria for clinical reporting: diagnostic exome sequencing identifies altered candidate genes among 8% of patients with undiagnosed diseases. *Genet Med* 19:224-35
86. Paylor R, Glaser B, Mupo A, Ataliotis P, Spencer C, et al. 2006. Tbx1 haploinsufficiency is linked to behavioral disorders in mice and humans: implications for 22q11 deletion syndrome. *Proc Natl Acad Sci U S A* 103:7729-34
87. Stein JM, Bergman W, Fang Y, Davison L, Brensinger C, et al. 2006. Behavioral and neurochemical alterations in mice lacking the RNA-binding protein translin. *J Neurosci* 26:2184-96
88. Aoki K, Suzuki K, Sugano T, Tasaka T, Nakahara K, et al. 1995. A novel gene, Translin, encodes a recombination hotspot binding protein associated with chromosomal translocations. *Nat Genet* 10:167-74
89. Hao YH, Fountain MD, Jr., Fon Tacer K, Xia F, Bi W, et al. 2015. USP7 Acts as a Molecular Rheostat to Promote WASH-Dependent Endosomal Protein Recycling and Is Mutated in a Human Neurodevelopmental Disorder. *Mol Cell* 59:956-69

90. Halvardson J, Zhao JJ, Zaghlool A, Wentzel C, Georgii-Hemming P, et al. 2016. Mutations in HECW2 are associated with intellectual disability and epilepsy. *J Med Genet* 53:697-704