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List of Publications by Year
in descending order

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34 papers	8,116 citations	394421 19 h-index	434195 31 g-index
40 all docs	40 docs citations	40 times ranked	13450 citing authors

#	Article	IF	Citations
1	Synaptic, transcriptional and chromatin genes disrupted in autism. Nature, 2014, 515, 209-215.	27.8	2,254
2	MeCP2, a Key Contributor to Neurological Disease, Activates and Represses Transcription. Science, 2008, 320, 1224-1229.	12.6	1,582
3	The Story of Rett Syndrome: From Clinic to Neurobiology. Neuron, 2007, 56, 422-437.	8.1	1,097
4	Dysfunction in GABA signalling mediates autism-like stereotypies and Rett syndrome phenotypes. Nature, 2010, 468, 263-269.	27.8	1,042
5	SATB2 Is a Multifunctional Determinant of Craniofacial Patterning and Osteoblast Differentiation. Cell, 2006, 125, 971-986.	28.9	458
6	Using Whole-Exome Sequencing to Identify Inherited Causes of Autism. Neuron, 2013, 77, 259-273.	8.1	383
7	Mouse models of MeCP2 disorders share gene expression changes in the cerebellum and hypothalamus. Human Molecular Genetics, 2009, 18, 2431-2442.	2.9	228
8	Whole-Exome Sequencing and Homozygosity Analysis Implicate Depolarization-Regulated Neuronal Genes in Autism. PLoS Genetics, 2012, 8, e1002635.	3.5	164
9	The Diverse Genetic Landscape of Neurodevelopmental Disorders. Annual Review of Genomics and Human Genetics, 2014, 15, 195-213.	6.2	146
10	Evolution of Osteocrin as an activity-regulated factor in the primate brain. Nature, 2016, 539, 242-247.	27.8	120
11	Disruption of the ATXN1â€“CIC complex causes a spectrum of neurobehavioral phenotypes in mice and humans. Nature Genetics, 2017, 49, 527-536.	21.4	113
12	Arid1b haploinsufficient mice reveal neuropsychiatric phenotypes and reversible causes of growth impairment. ELife, 2017, 6, .	6.0	74
13	Rett-causing mutations reveal two domains critical for MeCP2 function and for toxicity in MECP2 duplication syndrome mice. ELife, 2014, 3, .	6.0	72
14	Novel sequence variants in theTMC1 gene in Pakistani families with autosomal recessive hearing impairment. Human Mutation, 2005, 26, 396-396.	2.5	52
15	Current Perspectives in Autism Spectrum Disorder: From Genes to Therapy. Journal of Neuroscience, 2016, 36, 11402-11410.	3.6	44
16	MeCP2 and histone deacetylases 1 and 2 in dorsal striatum collectively suppress repetitive behaviors. Nature Neuroscience, 2016, 19, 1506-1512.	14.8	36
17	The ubiquitin proteasome pathway in neuropsychiatric disorders. Neurobiology of Learning and Memory, 2019, 165, 106791.	1.9	35
18	The ubiquitin ligase UBE3B, disrupted in intellectual disability and absent speech, regulates metabolic pathways by targeting BCKDK. Proceedings of the National Academy of Sciences of the United States of America, 2019, 116, 3662-3667.	7.1	27

#	Article	IF	Citations
19	KDM5A mutations identified in autism spectrum disorder using forward genetics. ELife, 2020, 9, .	6.0	27
20	PSMD12 haploinsufficiency in a neurodevelopmental disorder with autistic features. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2018, 177, 736-745.	1.7	23
21	Ube3a/E6AP is involved in a subset of MeCP2 functions. Biochemical and Biophysical Research Communications, 2013, 437, 67-73.	2.1	21
22	A novel autosomal recessive nonsyndromic hearing impairment locus (DFNB42) maps to chromosome 3q13.31-q22.3. American Journal of Medical Genetics, Part A, 2005, 133A, 18-22.	1.2	20
23	Novel sequence variants in the TMIE gene in families with autosomal recessive nonsyndromic hearing impairment. Journal of Molecular Medicine, 2006, 84, 226-231.	3.9	19
24	Analysis of recent shared ancestry in a familial cohort identifies coding and noncoding autism spectrum disorder variants. Npj Genomic Medicine, 2022, 7, 13.	3.8	18
25	Homozygous deletions implicate non-coding epigenetic marks in Autism spectrum disorder. Scientific Reports, 2020, 10, 14045.	3.3	12
26	DFNB44, a Novel Autosomal Recessive Non-Syndromic Hearing Impairment Locus, Maps to Chromosome 7p14.1-q11.22. Human Heredity, 2004, 57, 195-199.	0.8	11
27	A novel autosomal recessive non-syndromic hearing impairment locus (DFNB47) maps to chromosome 2p25.1-p24.3. Human Genetics, 2006, 118, 605-610.	3.8	9
28	Candidate Genes for Inherited Autism Susceptibility in the Lebanese Population. Scientific Reports, 2017, 7, 45336.	3.3	7
29	Translating genetic and preclinical findings into autism therapies. Dialogues in Clinical Neuroscience, 2017, 19, 335-343.	3.7	6
30	Variability of Ponto-cerebellar Fibers by Diffusion Tensor Imaging in Diverse Brain Malformations. Journal of Child Neurology, 2017, 32, 271-285.	1.4	5
31	Mapping of a novel autosomal recessive nonsyndromic deafness locus (DFNB46) to chromosome 18p11.32-p11.31. American Journal of Medical Genetics, Part A, 2005, 133A, 23-26.	1.2	3
32	X-linked and mitochondrial disorders. , 2021, , 137-149.		0
33	Genetics of Autism Spectrum Disorder: Searching for the Rare to Explain the Common. , 2022, , 299-306.		0
34	Insights Into DDX3X Syndrome From a Novel Mouse Model With Construct and Face Validity. Biological Psychiatry, 2021, 90, 732-734.	1.3	0