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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	Genetics of Autism Spectrum Disorder: Searching for the Rare to Explain the Common. , 2022, , 299-306.		0
2	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
3	X-linked and mitochondrial disorders. , 2021, , 137-149.		0
4	Insights Into DDX3X Syndrome From a Novel Mouse Model With Construct and Face Validity. Biological Psychiatry, 2021, 90, 732-734.	1.3	0
5	Homozygous deletions implicate non-coding epigenetic marks in Autism spectrum disorder. Scientific Reports, 2020, 10, 14045.	3.3	12
6	KDM5A mutations identified in autism spectrum disorder using forward genetics. ELife, 2020, 9, .	6.0	27
7	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
8	The ubiquitin proteasome pathway in neuropsychiatric disorders. Neurobiology of Learning and Memory, 2019, 165, 106791.	1.9	35
9	<i>PSMD12</i> haploinsufficiency in a neurodevelopmental disorder with autistic features. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2018, 177, 736-745.	1.7	23
10	Variability of Ponto-cerebellar Fibers by Diffusion Tensor Imaging in Diverse Brain Malformations. Journal of Child Neurology, 2017, 32, 271-285.	1.4	5
11	Candidate Genes for Inherited Autism Susceptibility in the Lebanese Population. Scientific Reports, 2017, 7, 45336.	3.3	7
12	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
13	Translating genetic and preclinical findings into autism therapies. Dialogues in Clinical Neuroscience, 2017, 19, 335-343.	3.7	6
14	Arid1b haploinsufficient mice reveal neuropsychiatric phenotypes and reversible causes of growth impairment. ELife, 2017, 6, .	6.0	74
15	MeCP2 and histone deacetylases 1 and 2 in dorsal striatum collectively suppress repetitive behaviors. Nature Neuroscience, 2016, 19, 1506-1512.	14.8	36
16	Evolution of Osteocrin as an activity-regulated factor in the primate brain. Nature, 2016, 539, 242-247.	27.8	120
17	Current Perspectives in Autism Spectrum Disorder: From Genes to Therapy. Journal of Neuroscience, 2016, 36, 11402-11410.	3.6	44
18	The Diverse Genetic Landscape of Neurodevelopmental Disorders. Annual Review of Genomics and Human Genetics, 2014, 15, 195-213.	6.2	146

#	ARTICLE	IF	CITATIONS
19	Synaptic, transcriptional and chromatin genes disrupted in autism. <i>Nature</i> , 2014, 515, 209-215.	27.8	2,254
20	Rett-causing mutations reveal two domains critical for MeCP2 function and for toxicity in MECP2 duplication syndrome mice. <i>ELife</i> , 2014, 3, .	6.0	72
21	Ube3a/E6AP is involved in a subset of MeCP2 functions. <i>Biochemical and Biophysical Research Communications</i> , 2013, 437, 67-73.	2.1	21
22	Using Whole-Exome Sequencing to Identify Inherited Causes of Autism. <i>Neuron</i> , 2013, 77, 259-273.	8.1	383
23	Whole-Exome Sequencing and Homozygosity Analysis Implicate Depolarization-Regulated Neuronal Genes in Autism. <i>PLoS Genetics</i> , 2012, 8, e1002635.	3.5	164
24	Dysfunction in GABA signalling mediates autism-like stereotypies and Rett syndrome phenotypes. <i>Nature</i> , 2010, 468, 263-269.	27.8	1,042
25	Mouse models of MeCP2 disorders share gene expression changes in the cerebellum and hypothalamus. <i>Human Molecular Genetics</i> , 2009, 18, 2431-2442.	2.9	228
26	MeCP2, a Key Contributor to Neurological Disease, Activates and Represses Transcription. <i>Science</i> , 2008, 320, 1224-1229.	12.6	1,582
27	The Story of Rett Syndrome: From Clinic to Neurobiology. <i>Neuron</i> , 2007, 56, 422-437.	8.1	1,097
28	SATB2 Is a Multifunctional Determinant of Craniofacial Patterning and Osteoblast Differentiation. <i>Cell</i> , 2006, 125, 971-986.	28.9	458
29	A novel autosomal recessive non-syndromic hearing impairment locus (DFNB47) maps to chromosome 2p25.1-p24.3. <i>Human Genetics</i> , 2006, 118, 605-610.	3.8	9
30	Novel sequence variants in the TMIE gene in families with autosomal recessive nonsyndromic hearing impairment. <i>Journal of Molecular Medicine</i> , 2006, 84, 226-231.	3.9	19
31	Novel sequence variants in the TMC1 gene in Pakistani families with autosomal recessive hearing impairment. <i>Human Mutation</i> , 2005, 26, 396-396.	2.5	52
32	A novel autosomal recessive nonsyndromic hearing impairment locus (DFNB42) maps to chromosome 3q13.31-q22.3. <i>American Journal of Medical Genetics, Part A</i> , 2005, 133A, 18-22.	1.2	20
33	Mapping of a novel autosomal recessive nonsyndromic deafness locus (DFNB46) to chromosome 18p11.32-p11.31. <i>American Journal of Medical Genetics, Part A</i> , 2005, 133A, 23-26.	1.2	3
34	DFNB44, a Novel Autosomal Recessive Non-Syndromic Hearing Impairment Locus, Maps to Chromosome 7p14.1-q11.22. <i>Human Heredity</i> , 2004, 57, 195-199.	0.8	11