Donna M Martin

List of Publications by Year
in descending order

90 papers	6,514 citations	109137 35 h-index	74018 75 g-index
96 all docs	96 docs citations	96 times ranked	10504 citing authors

#	Article	IF	Citations
1	Insights into Autism Spectrum Disorder Genomic Architecture and Biology from 71 Risk Loci. Neuron, 2015, 87, 1215-1233.	3.8	1,219
2	Multiple Recurrent De Novo CNVs, Including Duplications of the 7q11.23 Williams Syndrome Region, Are Strongly Associated with Autism. Neuron, 2011, 70, 863-885.	3.8	1,146
3	Common genetic variants, acting additively, are a major source of risk for autism. Molecular Autism, 2012, 3, 9.	2.6	357
4	Molecular and phenotypic aspects of CHD7 mutation in CHARGE syndrome. American Journal of Medical Genetics, Part A, 2010, 152A, 674-686.	0.7	265
5	Loss of Chd7 function in gene-trapped reporter mice is embryonic lethal and associated with severe defects in multiple developing tissues. Mammalian Genome, 2007, 18, 94-104.	1.0	148
6	Recurrent De Novo and Biallelic Variation of ATAD3A , Encoding a Mitochondrial Membrane Protein, Results in Distinct Neurological Syndromes. American Journal of Human Genetics, 2016, 99, 831-845.	2.6	146
7	Chd7 cooperates with Sox10 and regulates the onset of CNS myelination and remyelination. Nature Neuroscience, 2016, 19, 678-689.	7.1	142
8	Characterization of progenitor domains in the developing mouse thalamus. Journal of Comparative Neurology, 2007, 505, 73-91.	0.9	141
9	Nestin-Lineage Cells Contribute to the Microvasculature but Not Endocrine Cells of the Islet. Diabetes, 2003, 52, 2503-2512.	0.3	137
10	A Genome-wide Association Study of Autism Using the Simons Simplex Collection: Does Reducing Phenotypic Heterogeneity in Autism Increase Genetic Homogeneity?. Biological Psychiatry, 2015, 77, 775-784.	0.7	133
11	Atypical phenotypes associated with pathogenic CHD7 variants and a proposal for broadening CHARGE syndrome clinical diagnostic criteria. American Journal of Medical Genetics, Part A, 2016, 170, 344-354.	0.7	122
12	Inappropriate p53 activation during development induces features of CHARGE syndrome. Nature, 2014, 514, 228-232.	13.7	117
13	The ATP-dependent chromatin remodeling enzyme CHD7 regulates pro-neural gene expression and neurogenesis in the inner ear. Development (Cambridge), 2010, 137, 3139-3150.	1.2	116
14	Dual Requirement of CHD8 for Chromatin Landscape Establishment and Histone Methyltransferase Recruitment to Promote CNS Myelination and Repair. Developmental Cell, 2018, 45, 753-768.e8.	3.1	112
15	PITX2 is required for normal development of neurons in the mouse subthalamic nucleus and midbrain. Developmental Biology, 2004, 267, 93-108.	0.9	94
16	Autosomal Dominant Stapes Ankylosis with Broad Thumbs and Toes, Hyperopia, and Skeletal Anomalies Is Caused by Heterozygous Nonsense and Frameshift Mutations in NOG, the Gene Encoding Noggin*. American Journal of Human Genetics, 2002, 71, 618-624.	2.6	93
17	CHD7 functions in the nucleolus as a positive regulator of ribosomal RNA biogenesis. Human Molecular Genetics, 2010, 19, 3491-3501.	1.4	91
18	Oligodendrocyte precursor survival and differentiation requires chromatin remodeling by Chd7 and Chd8. Proceedings of the National Academy of Sciences of the United States of America, 2018, 115, E8246-E8255.	3.3	81

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19	Adjusting Head Circumference for Covariates in Autism: Clinical Correlates of a Highly Heritable Continuous Trait. Biological Psychiatry, 2013, 74, 576-584.	0.7	70
20	Super Enhancers in Cancers, Complex Disease, and Developmental Disorders. Genes, 2015, 6, 1183-1200.	1.0	61
21	Pitx2 Distinguishes Subtypes of Terminally Differentiated Neurons in the Developing Mouse Neuroepithelium. Developmental Biology, 2002, 252, 84-99.	0.9	59
22	Modest Impact on Risk for Autism Spectrum Disorder of Rare Copy Number Variants at 15<scp>q</scp>11.2, Specifically Breakpoints 1 to 2. Autism Research, 2014, 7, 355-362.	2.1	59
23	Reproductive dysfunction and decreased GnRH neurogenesis in a mouse model of CHARGE syndrome. Human Molecular Genetics, 2011, 20, 3138-3150.	1.4	57
24	CHD7 and retinoic acid signaling cooperate to regulate neural stem cell and inner ear development in mouse models of CHARGE syndrome. Human Molecular Genetics, 2014, 23, 434-448.	1.4	57
25	Defects in vestibular sensory epithelia and innervation in mice with loss of Chd7 function: Implications for human CHARGE syndrome. Journal of Comparative Neurology, 2007, 504, 519-532.	0.9	56
26	De novodominantASXL3mutations alter H2A deubiquitination and transcription in Bainbridgeâ€“Ropers syndrome. Human Molecular Genetics, 2016, 25, 597-608.	1.4	56
27	The impact of serotonin transporter (5-HTTLPR) genotype on the development of resting-state functional connectivity in children and adolescents: A preliminary report. NeuroImage, 2012, 59, 2760-2770.	2.1	55
28	Neural crest contributions to the ear: Implications for congenital hearing disorders. Hearing Research, 2019, 376, 22-32.	0.9	54
29	New insights and advances in CHARGE syndrome: Diagnosis, etiologies, treatments, and research discoveries. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2017, 175, 397-406.	0.7	46
30	De Novo Heterozygous POLR2A Variants Cause a Neurodevelopmental Syndrome with Profound Infantile-Onset Hypotonia. American Journal of Human Genetics, 2019, 105, 283-301.	2.6	46
31	Genetic analysis of CHARGE syndrome identifies overlapping molecular biology. Genetics in Medicine, 2018, 20, 1022-1029.	1.1	43
32	Branchiootorenal syndrome and oculoauriculovertebral spectrum features associated with duplication ofSIX1,SIX6, andOTX2resulting from a complex chromosomal rearrangement. American Journal of Medical Genetics, Part A, 2008, 146A, 2480-2489.	0.7	42
33	Recurrent deletions and duplications of chromosome 2q11.2 and 2q13 are associated with variable outcomes. American Journal of Medical Genetics, Part A, 2015, 167, 2664-2673.	0.7	42
34	Nestin-Cre mediated deletion ofPitx2 in the mouse. Genesis, 2006, 44, 336-344.	0.8	41
35	Cre fate mapping reveals lineage specific defects in neuronal migration with loss of Pitx2 function in the developing mouse hypothalamus and subthalamic nucleus. Molecular and Cellular Neurosciences, 2008, 37, 696-707.	1.0	38
36	Serotonin transporter genotype impacts amygdala habituation in youth with autism spectrum disorders. Social Cognitive and Affective Neuroscience, 2014, 9, 832-838.	1.5	38

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37	Mutation update for the SATB2 gene. Human Mutation, 2019, 40, 1013-1029.	1.1	38
38	Duplication 16p11.2 in a child with infantile seizure disorder. American Journal of Medical Genetics, Part A, 2010, 152A, 1567-1574.	0.7	37
39	Mature middle and inner ears express Chd7 and exhibit distinctive pathologies in a mouse model of CHARGE syndrome. Hearing Research, 2011, 282, 184-195.	0.9	36
40	The chromatin remodeling protein CHD7, mutated in CHARGE syndrome, is necessary for proper craniofacial and tracheal development. Developmental Dynamics, 2014, 243, 1055-1066.	0.8	34
41	Genotype-phenotype correlations in individuals with pathogenicREREvariants. Human Mutation, 2018, 39, 666-675.	1.1	34
42	Delayed fusion and altered gene expression contribute to semicircular canal defects in Chd7 deficient mice. Mechanisms of Development, 2012, 129, 308-323.	1.7	33
43	GABAergic and glutamatergic identities of developing midbrain Pitx2 neurons. Developmental Dynamics, 2011, 240, 333-346.	0.8	32
44	A novel chromosome 19p13.12 deletion in a child with multiple congenital anomalies. American Journal of Medical Genetics, Part A, 2009, 149A, 396-402.	0.7	31
45	Chromatin Remodeling in Development and Disease: Focus on CHD7. PLoS Genetics, 2010, 6, e1001010.	1.5	31
46	CHARGE association With choanal atresia and inner ear hypoplasia in a child with a de novo chromosome translocation t(2;7)(p14;q21.11). American Journal of Medical Genetics Part A, 2001, 99, 115-119.	2.4	30
47	Dysregulation of cotranscriptional alternative splicing underlies CHARGE syndrome. Proceedings of the National Academy of Sciences of the United States of America, 2018, 115, E620-E629.	3.3	28
48	Genotypeâ€“phenotype analysis of 523 patients by genetics evaluation and clinical exome sequencing. Pediatric Research, 2020, 87, 735-739.	1.1	28
49	CHD7 represses the retinoic acid synthesis enzyme ALDH1A3 during inner ear development. JCI Insight, 2018, 3, .	2.3	27
50	Mouse Models for the Dissection of CHD7 Functions in Eye Development and the Molecular Basis for Ocular Defects in CHARGE Syndrome. , 2015, 56, 7923.		26
51	CHD7 Mutations and CHARGE Syndrome in Semicircular Canal Dysplasia. Otology and Neurotology, 2014, 35, 1466-1470.	0.7	25
52	Harnessing molecular motors for nanoscale pulldown in live cells. Molecular Biology of the Cell, 2017, 28, 463-475.	0.9	25
53	Incorporation of exomeâ€based CNV analysis makes trioâ€WES a more powerful tool for clinical diagnosis in neurodevelopmental disorders: A retrospective study. Human Mutation, 2021, 42, 990-1004.	1.1	25
54	Characterization of a Stapes Ankylosis Family with a NOG Mutation. Otology and Neurotology, 2003, 24, 210-215.	0.7	24

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55	CHD7 promotes neural progenitor differentiation in embryonic stem cells via altered chromatin accessibility and nascent gene expression. Scientific Reports, 2020, 10, 17445.	1.6	23
56	Epigenetic Developmental Disorders: CHARGE Syndrome, a Case Study. Current Genetic Medicine Reports, 2015, 3, 1-7.	1.9	19
57	Association of Salary Differences Between Medical Specialties With Sex Distribution. JAMA Pediatrics, 2021, 175, 524.	3.3	19
58	Systemic lupus erythematosus in a man with Noonan syndrome. American Journal of Medical Genetics Part A, 2001, 102, 59-62.	2.4	18
59	Ageâ€related effect of serotonin transporter genotype on amygdala and prefrontal cortex function in adolescence. Human Brain Mapping, 2014, 35, 646-658.	1.9	18
60	Disruption of RAB40AL function leads to Martinâ€“Probst syndrome, a rare X-linked multisystem neurodevelopmental human disorder. Journal of Medical Genetics, 2012, 49, 332-340.	1.5	17
61	A novel TaulacZ allele reveals a requirement for Pitx2 in formation of the mammillothalamic tract. Genesis, 2012, 50, 67-73.	0.8	17
62	Congenital heart defects in CHARGE: The molecular role of CHD7 and effects on cardiac phenotype and clinical outcomes. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2020, 184, 81-89.	0.7	17
63	The influence of 5-HTTLPR transporter genotype on amygdala-subgenual anterior cingulate cortex connectivity in autism spectrum disorder. Developmental Cognitive Neuroscience, 2017, 24, 12-20.	1.9	16
64	Inner ear manifestations in CHARGE: Abnormalities, treatments, animal models, and progress toward treatments in auditory and vestibular structures. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2017, 175, 439-449.	0.7	16
65	Nervous system development and disease: A focus on trithorax related proteins and chromatin remodelers. Molecular and Cellular Neurosciences, 2018, 87, 46-54.	1.0	16
66	Single Cell Transcriptomics Reveal Abnormalities in Neurosensory Patterning of the Chd7 Mutant Mouse Ear. Frontiers in Genetics, 2018, 9, 473.	1.1	16
67	The impact of serotonin transporter genotype on default network connectivity in children and adolescents with autism spectrum disorders. NeuroImage: Clinical, 2013, 2, 17-24.	1.4	15
68	GJB2 gene therapy and conditional deletion reveal developmental stage-dependent effects on inner ear structure and function. Molecular Therapy - Methods and Clinical Development, 2021, 23, 319-333.	1.8	15
69	Atopic disorders in CHARGE syndrome: A retrospective study and literature review. European Journal of Medical Genetics, 2018, 61, 225-229.	0.7	12
70	Pleiotropic and isoform-specific functions for Pitx2 in superior colliculus and hypothalamic neuronal development. Molecular and Cellular Neurosciences, 2013, 52, 128-139.	1.0	11
71	Duplication 2p25 in a child with clinical features of CHARGE syndrome. American Journal of Medical Genetics, Part A, 2016, 170, 1148-1154.	0.7	11
72	Distinct populations of GABAergic neurons in mouse rhombomere 1 express but do not require the homeodomain transcription factor PITX2. Molecular and Cellular Neurosciences, 2012, 49, 32-43.	1.0	10

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73	Chromatin remodeler CHD7 is critical for cochlear morphogenesis and neurosensory patterning. Developmental Biology, 2021, 477, 11-21.	0.9	10
74	Changing the editorial process at JCI and JCI Insight in response to the COVID-19 pandemic. Journal of Clinical Investigation, 2020, 130, 2147-2147.	3.9	10
75	Interrupted aortic arch in a child with trisomy 5q31.1q35.1 due to a maternal (20;5) balanced insertion. American Journal of Medical Genetics Part A, 2003, 116A, 268-271.	2.4	9
76	Meis2 Is Required for Inner Ear Formation and Proper Morphogenesis of the Cochlea. Frontiers in Cell and Developmental Biology, 2021, 9, 679325.	1.8	8
77	C-terminals in the mouse branchiomotor nuclei originate from the magnocellular reticular formation. Neuroscience Letters, 2013, 548, 137-142.	1.0	7
78	Response to correspondence to Hale et al. atypical phenotypes associated with pathogenic CHD7 variants and a proposal for broadening CHARGE syndrome clinical diagnostic criteria. American Journal of Medical Genetics, Part A, 2016, 170, 3367-3368.	0.7	7
79	Itâ€™s All in the Delivery: Improving AAV Transfection Efficiency with Exosomes. Molecular Therapy, 2017, 25, 309-311.	3.7	6
80	Genetic specification of leftâ€“right asymmetry in the diaphragm muscles and their motor innervation. ELife, 2017, 6, .	2.8	6
81	Gene-based diagnostic and treatment methods for tinnitus. International Tinnitus Journal, 2003, 9, 3-10.	0.1	6
82	Skewed X-inactivation in carriers establishes linkage in an X-linked deafness-mental retardation syndrome. American Journal of Medical Genetics Part A, 2004, 131A, 209-212.	2.4	5
83	Genetics of subthalamic nucleus in development and disease. Experimental Neurology, 2005, 192, 320-330.	2.0	5
84	Microarray oligonucleotide probe designer: a Web service. Open Access Bioinformatics, 2010, 2, 145.	0.9	5
85	Axial levelâ€specific regulation of neuronal development: Lessons from PITX2. Journal of Neuroscience Research, 2015, 93, 195-198.	1.3	5
86	Balancing dual demands on the physician-scientist workforce. Journal of Clinical Investigation, 2018, 128, 3204-3205.	3.9	5
87	Epigenetic mechanisms of inner ear development. Hearing Research, 2022, 426, 108440.	0.9	5
88	Have You Heard? Viral-Mediated Gene Therapy Restores Hearing. Neuron, 2012, 75, 188-190.	3.8	2
89	Development and implementation of an electronic medical record module to track genetic testing results. Genetics in Medicine, 2021, 23, 972-975.	1.1	2
90	Support for the Diagnosis of CHARGE Syndrome. JAMA Otolaryngology - Head and Neck Surgery, 2017, 143, 634.	1.2	0