

# Donna M Martin

## List of Publications by Year in descending order

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90  
papers

6,514  
citations

109264

35  
h-index

71651

76  
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. <i>Neuron</i> , 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. <i>Neuron</i> , 2011, 70, 863-885.	3.8	1,146
3	Common genetic variants, acting additively, are a major source of risk for autism. <i>Molecular Autism</i> , 2012, 3, 9.	2.6	357
4	Molecular and phenotypic aspects of <i>CHD7</i> mutation in CHARGE syndrome. <i>American Journal of Medical Genetics, Part A</i> , 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. <i>Mammalian Genome</i> , 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. <i>American Journal of Human Genetics</i> , 2016, 99, 831-845.	2.6	146
7	Chd7 cooperates with Sox10 and regulates the onset of CNS myelination and remyelination. <i>Nature Neuroscience</i> , 2016, 19, 678-689.	7.1	142
8	Characterization of progenitor domains in the developing mouse thalamus. <i>Journal of Comparative Neurology</i> , 2007, 505, 73-91.	0.9	141
9	Nestin-Lineage Cells Contribute to the Microvasculature but Not Endocrine Cells of the Islet. <i>Diabetes</i> , 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?. <i>Biological Psychiatry</i> , 2015, 77, 775-784.	0.7	133
11	Atypical phenotypes associated with pathogenic <i>CHD7</i> variants and a proposal for broadening CHARGE syndrome clinical diagnostic criteria. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 344-354.	0.7	122
12	Inappropriate p53 activation during development induces features of CHARGE syndrome. <i>Nature</i> , 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. <i>Development (Cambridge)</i> , 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. <i>Developmental Cell</i> , 2018, 45, 753-768.e8.	3.1	112
15	PITX2 is required for normal development of neurons in the mouse subthalamic nucleus and midbrain. <i>Developmental Biology</i> , 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*. <i>American Journal of Human Genetics</i> , 2002, 71, 618-624.	2.6	93
17	CHD7 functions in the nucleolus as a positive regulator of ribosomal RNA biogenesis. <i>Human Molecular Genetics</i> , 2010, 19, 3491-3501.	1.4	91
18	Oligodendrocyte precursor survival and differentiation requires chromatin remodeling by Chd7 and Chd8. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 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. <i>Biological Psychiatry</i> , 2013, 74, 576-584.	0.7	70
20	Super Enhancers in Cancers, Complex Disease, and Developmental Disorders. <i>Genes</i> , 2015, 6, 1183-1200.	1.0	61
21	Pitx2 Distinguishes Subtypes of Terminally Differentiated Neurons in the Developing Mouse Neuroepithelium. <i>Developmental Biology</i> , 2002, 252, 84-99.	0.9	59
22	Modest Impact on Risk for Autism Spectrum Disorder of Rare Copy Number Variants at 15q11.2, Specifically Breakpoints 1 to 2. <i>Autism Research</i> , 2014, 7, 355-362.	2.1	59
23	Reproductive dysfunction and decreased GnRH neurogenesis in a mouse model of CHARGE syndrome. <i>Human Molecular Genetics</i> , 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. <i>Human Molecular Genetics</i> , 2014, 23, 434-448.	1.4	57
25	Defects in vestibular sensory epithelia and innervation in mice with loss of <i>Chd7</i> function: Implications for human CHARGE syndrome. <i>Journal of Comparative Neurology</i> , 2007, 504, 519-532.	0.9	56
26	<i>De novo</i> dominant <i>ASXL3</i> mutations alter H2A deubiquitination and transcription in Bainbridge-Ropers syndrome. <i>Human Molecular Genetics</i> , 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. <i>NeuroImage</i> , 2012, 59, 2760-2770.	2.1	55
28	Neural crest contributions to the ear: Implications for congenital hearing disorders. <i>Hearing Research</i> , 2019, 376, 22-32.	0.9	54
29	New insights and advances in CHARGE syndrome: Diagnosis, etiologies, treatments, and research discoveries. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , 2017, 175, 397-406.	0.7	46
30	De Novo Heterozygous POLR2A Variants Cause a Neurodevelopmental Syndrome with Profound Infantile-Onset Hypotonia. <i>American Journal of Human Genetics</i> , 2019, 105, 283-301.	2.6	46
31	Genetic analysis of CHARGE syndrome identifies overlapping molecular biology. <i>Genetics in Medicine</i> , 2018, 20, 1022-1029.	1.1	43
32	Branchiootorenal syndrome and oculoauriculovertebral spectrum features associated with duplication of <i>SIX1</i> , <i>SIX6</i> , and <i>OTX2</i> resulting from a complex chromosomal rearrangement. <i>American Journal of Medical Genetics, Part A</i> , 2008, 146A, 2480-2489.	0.7	42
33	Recurrent deletions and duplications of chromosome 2q11.2 and 2q13 are associated with variable outcomes. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 2664-2673.	0.7	42
34	Nestin-Cre mediated deletion of Pitx2 in the mouse. <i>Genesis</i> , 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. <i>Molecular and Cellular Neurosciences</i> , 2008, 37, 696-707.	1.0	38
36	Serotonin transporter genotype impacts amygdala habituation in youth with autism spectrum disorders. <i>Social Cognitive and Affective Neuroscience</i> , 2014, 9, 832-838.	1.5	38

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37	Mutation update for the <i>SATB2</i> gene. <i>Human Mutation</i> , 2019, 40, 1013-1029.	1.1	38
38	Duplication 16p11.2 in a child with infantile seizure disorder. <i>American Journal of Medical Genetics, Part A</i> , 2010, 152A, 1567-1574.	0.7	37
39	Mature middle and inner ears express <i>Chd7</i> and exhibit distinctive pathologies in a mouse model of CHARGE syndrome. <i>Hearing Research</i> , 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. <i>Developmental Dynamics</i> , 2014, 243, 1055-1066.	0.8	34
41	Genotype-phenotype correlations in individuals with pathogenic <i>RERE</i> variants. <i>Human Mutation</i> , 2018, 39, 666-675.	1.1	34
42	Delayed fusion and altered gene expression contribute to semicircular canal defects in <i>Chd7</i> deficient mice. <i>Mechanisms of Development</i> , 2012, 129, 308-323.	1.7	33
43	GABAergic and glutamatergic identities of developing midbrain <i>Pitx2</i> neurons. <i>Developmental Dynamics</i> , 2011, 240, 333-346.	0.8	32
44	A novel chromosome 19p13.12 deletion in a child with multiple congenital anomalies. <i>American Journal of Medical Genetics, Part A</i> , 2009, 149A, 396-402.	0.7	31
45	Chromatin Remodeling in Development and Disease: Focus on CHD7. <i>PLoS Genetics</i> , 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). <i>American Journal of Medical Genetics Part A</i> , 2001, 99, 115-119.	2.4	30
47	Dysregulation of cotranscriptional alternative splicing underlies CHARGE syndrome. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2018, 115, E620-E629.	3.3	28
48	Genotype-phenotype analysis of 523 patients by genetics evaluation and clinical exome sequencing. <i>Pediatric Research</i> , 2020, 87, 735-739.	1.1	28
49	CHD7 represses the retinoic acid synthesis enzyme <i>ALDH1A3</i> during inner ear development. <i>JCI Insight</i> , 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. <i>Otology and Neurotology</i> , 2014, 35, 1466-1470.	0.7	25
52	Harnessing molecular motors for nanoscale pulldown in live cells. <i>Molecular Biology of the Cell</i> , 2017, 28, 463-475.	0.9	25
53	Incorporation of exome-based CNV analysis makes trio-WES a more powerful tool for clinical diagnosis in neurodevelopmental disorders: A retrospective study. <i>Human Mutation</i> , 2021, 42, 990-1004.	1.1	25
54	Characterization of a Stapes Ankylosis Family with a <i>NOG</i> Mutation. <i>Otology and Neurotology</i> , 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. <i>Scientific Reports</i> , 2020, 10, 17445.	1.6	23
56	Epigenetic Developmental Disorders: CHARGE Syndrome, a Case Study. <i>Current Genetic Medicine Reports</i> , 2015, 3, 1-7.	1.9	19
57	Association of Salary Differences Between Medical Specialties With Sex Distribution. <i>JAMA Pediatrics</i> , 2021, 175, 524.	3.3	19
58	Systemic lupus erythematosus in a man with Noonan syndrome. <i>American Journal of Medical Genetics Part A</i> , 2001, 102, 59-62.	2.4	18
59	Age-related effect of serotonin transporter genotype on amygdala and prefrontal cortex function in adolescence. <i>Human Brain Mapping</i> , 2014, 35, 646-658.	1.9	18
60	Disruption of RAB40AL function leads to Martin-Probst syndrome, a rare X-linked multisystem neurodevelopmental human disorder. <i>Journal of Medical Genetics</i> , 2012, 49, 332-340.	1.5	17
61	A novel <i>Taulacz</i> allele reveals a requirement for <i>Pitx2</i> in formation of the mammillothalamic tract. <i>Genesis</i> , 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. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , 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. <i>Developmental Cognitive Neuroscience</i> , 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. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , 2017, 175, 439-449.	0.7	16
65	Nervous system development and disease: A focus on trithorax related proteins and chromatin remodelers. <i>Molecular and Cellular Neurosciences</i> , 2018, 87, 46-54.	1.0	16
66	Single Cell Transcriptomics Reveal Abnormalities in Neurosensory Patterning of the Chd7 Mutant Mouse Ear. <i>Frontiers in Genetics</i> , 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. <i>NeuroImage: Clinical</i> , 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. <i>Molecular Therapy - Methods and Clinical Development</i> , 2021, 23, 319-333.	1.8	15
69	Atopic disorders in CHARGE syndrome: A retrospective study and literature review. <i>European Journal of Medical Genetics</i> , 2018, 61, 225-229.	0.7	12
70	Pleiotropic and isoform-specific functions for Pitx2 in superior colliculus and hypothalamic neuronal development. <i>Molecular and Cellular Neurosciences</i> , 2013, 52, 128-139.	1.0	11
71	Duplication 2p25 in a child with clinical features of CHARGE syndrome. <i>American Journal of Medical Genetics, Part A</i> , 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. <i>Molecular and Cellular Neurosciences</i> , 2012, 49, 32-43.	1.0	10

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