Matthew A Deardorff

List of Publications by Year
in descending order

83	4,490	147801 31	114465 63
papers	citations	h-index	g-index

88	88	88	8686
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	HDAC8 mutations in Cornelia de Lange syndrome affect the cohesin acetylation cycle. Nature, 2012, 489, 313-317.	27.8	488
2	Mutations in Cohesin Complex Members SMC3 and SMC1A Cause a Mild Variant of Cornelia de Lange Syndrome with Predominant Mental Retardation. American Journal of Human Genetics, 2007, 80, 485-494.	6.2	445
3	Analysis of 589,306 genomes identifies individuals resilient to severe Mendelian childhood diseases. Nature Biotechnology, 2016, 34, 531-538.	17.5	273
4	The genomic landscape of balanced cytogenetic abnormalities associated with human congenital anomalies. Nature Genetics, 2017, 49, 36-45.	21.4	251
5	RAD21 Mutations Cause a Human Cohesinopathy. American Journal of Human Genetics, 2012, 90, 1014-1027.	6.2	238
6	Diagnosis and management of Cornelia de Lange syndrome: first international consensus statement. Nature Reviews Genetics, 2018, 19, 649-666.	16.3	223
7	Heterozygous missense mutations in SMARCA2 cause Nicolaides-Baraitser syndrome. Nature Genetics, 2012, 44, 445-449.	21.4	207
8	De Novo Mutations in Protein Kinase Genes CAMK2A and CAMK2B Cause Intellectual Disability. American Journal of Human Genetics, 2017, 101, 768-788.	6.2	136
9	Mutations in SPATA5 Are Associated with Microcephaly, Intellectual Disability, Seizures, and Hearing Loss. American Journal of Human Genetics, 2015, 97, 457-464.	6.2	134
10	CODAS Syndrome Is Associated with Mutations of LONP1, Encoding Mitochondrial AAA+ Lon Protease. American Journal of Human Genetics, 2015, 96, 121-135.	6.2	127
11	Recommendations for the integration of genomics into clinical practice. Genetics in Medicine, 2016, 18, 1075-1084.	2.4	125
12	Germline gain-of-function mutations in AFF4 cause a developmental syndrome functionally linking the super elongation complex and cohesin. Nature Genetics, 2015, 47, 338-344.	21.4	109
13	Recessive DNAH9 Loss-of-Function Mutations Cause Laterality Defects and Subtle Respiratory Ciliary-Beating Defects. American Journal of Human Genetics, 2018, 103, 995-1008.	6.2	92
14	Congenital hyperinsulinism in children with paternal 11p uniparental isodisomy and Beckwithâ€“Wiedemann syndrome. Journal of Medical Genetics, 2016, 53, 53-61.	3.2	76
15	Mutations in the ABCC6 Gene as a Cause of Generalized Arterial Calcification of Infancy: Genotypic Overlap with Pseudoxanthoma Elasticum. Journal of Investigative Dermatology, 2014, 134, 658-665.	0.7	70
16	Phenotypes and genotypes in individuals with SMC1A variants. American Journal of Medical Genetics, Part A, 2017, 173, 2108-2125.	1.2	69
17	Automated Clinical Exome Reanalysis Reveals Novel Diagnoses. Journal of Molecular Diagnostics, 2019, 21, 38-48.	2.8	68
18	Characterization of the Beckwithâ€Wiedemann spectrum: Diagnosis and management. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2019, 181, 693-708.	1.6	62

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19	Mutations in chromatin regulators functionally link Cornelia de Lange syndrome and clinically overlapping phenotypes. Human Genetics, 2017, 136, 307-320.	3.8	61
20	De Novo Truncating Mutations in AHDC1 in Individuals with Syndromic Expressive Language Delay, Hypotonia, and Sleep Apnea. American Journal of Human Genetics, 2014, 94, 784-789.	6.2	57
21	IntragenicKANSL1mutations and chromosome 17q21.31 deletions: broadening the clinical spectrum and genotypeâ€“phenotype correlations in a large cohort of patients. Journal of Medical Genetics, 2015, 52, 804-814.	3.2	47
22	TRPV6 Variants Interfere with Maternal-Fetal Calcium Transport through the Placenta and Cause Transient Neonatal Hyperparathyroidism. American Journal of Human Genetics, 2018, 102, 1104-1114.	6.2	47
23	WDR26 Haploinsufficiency Causes a Recognizable Syndrome of Intellectual Disability, Seizures, Abnormal Gait, and Distinctive Facial Features. American Journal of Human Genetics, 2017, 101, 139-148.	6.2	45
24	Cohesin complex-associated holoprosencephaly. Brain, 2019, 142, 2631-2643.	7.6	43
25	Biochemical and Structural Characterization of HDAC8 Mutants Associated with Cornelia de Lange Syndrome Spectrum Disorders. Biochemistry, 2015, 54, 6501-6513.	2.5	41
26	Mutations inSPECC1L, encoding sperm antigen with calponin homology and coiled-coil domains 1-like, are found in some cases of autosomal dominant Opitz G/BBB syndrome. Journal of Medical Genetics, 2015, 52, 104-110.	3.2	40
27	Precision therapy for a new disorder of AMPA receptor recycling due to mutations in ATAD1. Neurology: Genetics, 2017, 3, e130.	1.9	40
28	Cornelia de Lange syndrome in diverse populations. American Journal of Medical Genetics, Part A, 2019, 179, 150-158.	1.2	40
29	Clinical utility gene card for: Cornelia de Lange syndrome. European Journal of Human Genetics, 2015, 23, 1431-1431.	2.8	37
30	P4HA1 mutations cause a unique congenital disorder of connective tissue involving tendon, bone, muscle and the eye. Human Molecular Genetics, 2017, 26, 2207-2217.	2.9	37
31	Nomenclature and definition in asymmetric regional body overgrowth. American Journal of Medical Genetics, Part A, 2017, 173, 1735-1738.	1.2	36
32	A Recurrent De Novo Variant in NACC1 Causes a Syndrome Characterized by Infantile Epilepsy, Cataracts, and Profound Developmental Delay. American Journal of Human Genetics, 2017, 100, 343-351.	6.2	35
33	KCNK9 imprinting syndromeâ€”further delineation of a possible treatable disorder. American Journal of Medical Genetics, Part A, 2016, 170, 2632-2637.	1.2	32
34	Structural aspects of HDAC8 mechanism and dysfunction in Cornelia de Lange syndrome spectrum disorders. Protein Science, 2016, 25, 1965-1976.	7.6	30
35	Tumor screening in Beckwithâ€“Wiedemann syndromeâ€”To screen or not to screen?. American Journal of Medical Genetics, Part A, 2016, 170, 2261-2264.	1.2	28
36	Molecular diagnosis of somatic overgrowth conditions: A singleâ€center experience. Molecular Genetics & Genomic Medicine, 2019, 7, e536.	1.2	28

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37	MESP1 Mutations in Patients with Congenital Heart Defects. Human Mutation, 2016, 37, 308-314.	2.5	26
38	Utility of genetic evaluation in infants with congenital heart defects admitted to the cardiac intensive care unit. American Journal of Medical Genetics, Part A, 2016, 170, 3090-3097.	1.2	26
39	ECHS1 Deficiency as a Cause of Severe Neonatal Lactic Acidosis. JIMD Reports, 2016, 30, 33-37.	1.5	26
40	Complex management of a patient with a contiguous Xp11.4 gene deletion involving ornithine transcarbamylase: A role for detailed molecular analysis in complex presentations of classical diseases. Molecular Genetics and Metabolism, 2008, 94, 498-502.	1.1	25
41	New insights into DNA methylation signatures: SMARCA2 variants in Nicolaides-Baraitser syndrome. BMC Medical Genomics, 2019, 12, 105.	1.5	25
42	De novo missense variants in MEIS2 recapitulate the microdeletion phenotype of cardiac and palate abnormalities, developmental delay, intellectual disability and dysmorphic features. American Journal of Medical Genetics, Part A, 2018, 176, 1845-1851.	1.2	21
43	ANKRD11 variants: <scp>KBG</scp> syndrome and beyond. Clinical Genetics, 2021, 100, 187-200.	2.0	21
44	A human case of SLC35A3â€related skeletal dysplasia. American Journal of Medical Genetics, Part A, 2017, 173, 2758-2762.	1.2	20
45	Ganglioglioma in a Sotos syndrome patient with anNSD1 deletion. American Journal of Medical Genetics Part A, 2004, 130A, 393-394.	2.4	18
46	Beckwithâ€“Wiedemann syndrome in diverse populations. American Journal of Medical Genetics, Part A, 2019, 179, 525-533.	1.2	18
47	Bohringâ€Opitz syndrome caused by an ASXL1 mutation inherited from a germline mosaic mother. American Journal of Medical Genetics, Part A, 2018, 176, 1249-1252.	1.2	17
48	Diagnosis and management of the phenotypic spectrum of twins with Beckwithâ€Wiedemann syndrome. American Journal of Medical Genetics, Part A, 2019, 179, 1139-1147.	1.2	17
49	Rapid and accurate interpretation of clinical exomes using Phenoxome: a computational phenotype-driven approach. European Journal of Human Genetics, 2019, 27, 612-620.	2.8	17
50	Improved molecular detection of mosaicism in Beckwith-Wiedemann Syndrome. Journal of Medical Genetics, 2021, 58, 178-184.	3.2	17
51	Pathogenic variants in SMARCA5 , a chromatin remodeler, cause a range of syndromic neurodevelopmental features. Science Advances, 2021, 7, .	10.3	17
52	Hyperinsulinemic hypoglycemia in seven patients with de novo NSD1 mutations. American Journal of Medical Genetics, Part A, 2019, 179, 542-551.	1.2	16
53	EP300 â€related Rubinsteinâ€“Taybi syndrome: Highlighted rare phenotypic findings and a genotypeâ€“phenotype metaâ€analysis of 74 patients. American Journal of Medical Genetics, Part A, 2020, 182, 2926-2938.	1.2	16
54	Delineating the molecular and phenotypic spectrum of the SETD1B-related syndrome. Genetics in Medicine, 2021, 23, 2122-2137.	2.4	16

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55	The utility of alphaâ€fetoprotein screening in Beckwithâ€“Wiedemann syndrome. American Journal of Medical Genetics, Part A, 2017, 173, 581-584.	1.2	14
56	Phenotypic predictors and final diagnoses in patients referred for RASopathy testing by targeted next-generation sequencing. Genetics in Medicine, 2017, 19, 715-718.	2.4	14
57	The variability of <scp>SMARCA4</scp>â€related <scp>Coffinâ€“Siris</scp> syndrome: Do nonsense candidate variants add to milder phenotypes?. American Journal of Medical Genetics, Part A, 2020, 182, 2058-2067.	1.2	14
58	Exome sequencing reveals a nonsense mutation in MMP13 as a new cause of autosomal recessive metaphyseal anadysplasia. European Journal of Human Genetics, 2015, 23, 264-266.	2.8	13
59	A novel pathogenic missense ADAMTS17 variant that impairs secretion causes Weill-Marchesani Syndrome with variably dysmorphic hand features. Scientific Reports, 2020, 10, 10827.	3.3	13
60	<scp>EGFR</scp> mutations cause a lethal syndrome of epithelial dysfunction with progeroid features. Molecular Genetics & Genomic Medicine, 2015, 3, 452-458.	1.2	12
61	Fetal akinesia deformation sequence due to a congenital disorder of glycosylation. American Journal of Medical Genetics, Part A, 2015, 167, 2411-2417.	1.2	12
62	Growth hormone deficiency in megalencephalyâ€capillary malformation syndrome: An association with activating mutations in PIK3CA. American Journal of Medical Genetics, Part A, 2020, 182, 162-168.	1.2	11
63	Correspondence to Gripp et al. nephroblastomatosis or Wilms tumor in a fourth patient with a somatic PIK3CA mutation. American Journal of Medical Genetics, Part A, 2017, 173, 2293-2295.	1.2	10
64	Variants in <scp>NAA15</scp> cause pediatric hypertrophic cardiomyopathy. American Journal of Medical Genetics, Part A, 2021, 185, 228-233.	1.2	10
65	Copy number variation in CEP57L1 predisposes to congenital absence of bilateral ACL and PCL ligaments. Human Genomics, 2015, 9, 31.	2.9	9
66	Hematopoietic Stem Cell Transplant for the Treatment of X-MAID. Frontiers in Pediatrics, 2019, 7, 170.	1.9	9
67	The chromatin remodeler ISWI acts during Drosophila development to regulate adult sleep. Science Advances, 2021, 7, .	10.3	9
68	Evolution of histomorphologic, cytogenetic, and genetic abnormalities in an untreated patient with MIRAGE syndrome. Cancer Genetics, 2020, 245, 42-48.	0.4	7
69	Molecular Diagnostic Outcomes from 700 Cases. Journal of Molecular Diagnostics, 2022, 24, 274-286.	2.8	7
70	Utility and limitations of exome sequencing as a genetic diagnostic tool for conditions associated with pediatric sudden cardiac arrest/sudden cardiac death. Human Genomics, 2015, 9, 15.	2.9	5
71	Argininosuccinic Acid Lyase Deficiency Missed by Newborn Screen. JIMD Reports, 2016, 34, 43-47.	1.5	4
72	Cover Image, Volume 179A, Number 4, April 2019. , 2019, 179, i-i.		4

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73	Segmental congenital hemangiomas: Three cases of a rare entity. Pediatric Dermatology, 2020, 37, 548-553.	0.9	4
74	NKX2â€6 related congenital heart disease: Biallelic homeodomainâ€disrupting variants and truncus arteriosus. American Journal of Medical Genetics, Part A, 2020, 182, 1454-1459.	1.2	3
75	Nonlethal presentations of CYP26B1 â€related skeletal anomalies and multiple synostoses syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 2766-2775.	1.2	3
76	Whose Data, Whose Risk? Omics Privacy Concerns Should be Defined by Individuals, not Researchers. American Journal of Bioethics, 2021, 21, 67-70.	0.9	2
77	Another tool in the genome-wide association study arsenal: population-based detection of somatic gene conversion. BMC Medicine, 2011, 9, 13.	5.5	1
78	MG-102â€...Coffin-siris syndrome caused by a missense mutation in arid1a. Journal of Medical Genetics, 2015, 52, A1.3-A2.	3.2	1
79	Response to: Toriello et al., â€œUpdate on the Torielloâ€“Carey Syndrome.â€•Further delineation of a young woman with deletion 1q42.12â€q42.2. American Journal of Medical Genetics, Part A, 2017, 173, 1988-1991.	1.2	1
80	A PIK3CA mutation in an acquired capillary malformation. Pediatric Dermatology, 2020, 37, 246-247.	0.9	1
81	Cornelia de Lange syndrome and the Cohesin complex: Abstracts from the 9th Biennial Scientific and Educational Virtual Symposium 2020. American Journal of Medical Genetics, Part A, 2022, 188, 1005-1014.	1.2	1
82	Cornelia de Lange Syndrome: A Variable Disorder of Cohesin Pathology. Current Genetic Medicine Reports, 2015, 3, 74-81.	1.9	0
83	Cover Image, Volume 173A, Number 7, July 2017. American Journal of Medical Genetics, Part A, 2017, 173, i.	1.2	0