Jeffrey W Innis

List of Publications by Year
in descending order

47	2,118	394421	243625
papers	citations	19 h-index	44 g-index

47	47	47	5459
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Novel multilocus imprinting disturbances in a child with expressive language delay and intellectual disability. American Journal of Medical Genetics, Part A, 2022, , .	1.2	2
2	Cardiac phenotype in familial partial lipodystrophy. Clinical Endocrinology, 2021, 94, 1043-1053.	2.4	7
3	Transcriptome analysis of MBD5-associated neurodevelopmental disorder (MAND) neural progenitor cells reveals dysregulation of autism-associated genes. Scientific Reports, 2021, 11, 11295.	3.3	4
4	Metreleptin therapy for nonalcoholic steatohepatitis: Open-label therapy interventions in two different clinical settings. Med, 2021, 2, 814-835.e6.	4.4	12
5	AHDC1 missense mutations in Xia-Gibbs syndrome. Human Genetics and Genomics Advances, 2021, 2, 100049.	1.7	5
6	Histone H3.3 beyond cancer: Germline mutations in Histone 3 Family 3A and 3B cause a previously unidentified neurodegenerative disorder in 46 patients. Science Advances, 2020, 6, .	10.3	43
7	Phenotypic spectrum and transcriptomic profile associated with germline variants in TRAF7. Genetics in Medicine, 2020, 22, 1215-1226.	2.4	22
8	Natural history of ROHHAD syndrome: development of severe insulin resistance and fatty liver disease over time. Clinical Diabetes and Endocrinology, 2019, 5, 9.	2.7	17
9	Expanding the phenotypic spectrum of MBOAT7â€related intellectual disability. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2019, 180, 483-487.	1.7	19
10	Compound heterozygosity for loss-of-function FARSB variants in a patient with classic features of recessive aminoacyl-tRNA synthetase-related disease. Human Mutation, 2018, 39, 834-840.	2.5	30
11	Potential association of LMNA-associated generalized lipodystrophy with juvenile dermatomyositis. Clinical Diabetes and Endocrinology, 2018, 4, 6.	2.7	8
12	A Novel Generalized Lipodystrophy-Associated Progeroid Syndrome Due to Recurrent Heterozygous LMNA p.T10I Mutation. Journal of Clinical Endocrinology and Metabolism, 2018, 103, 1005-1014.	3.6	47
13	Interstitial microdeletion of the 1p34.3p34.2 region. Molecular Genetics & Genomic Medicine, 2018, 6, 673-677.	1.2	7
14	Spectrum of disease associated with partial lipodystrophy: lessons from a trial cohort. Clinical Endocrinology, 2017, 86, 698-707.	2.4	72
15	Integrative clinical genomics of metastatic cancer. Nature, 2017, 548, 297-303.	27.8	685
16	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.	1.2	42
17	Chromosomal Imbalances in Patients with Congenital Cardiac Defects: A Meta-analysis Reveals Novel Potential Critical Regions Involved in Heart Development. Congenital Heart Disease, 2015, 10, 193-208.	0.2	24
18	Nine de novo duplications affecting both maternal and paternal chromosomes and an inherited 15q11.2 deletion, in a patient with developmental delay. Clinical Case Reports (discontinued), 2015, 3, 396-401.	0.5	1

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19	Integrative Clinical Sequencing in the Management of Refractory or Relapsed Cancer in Youth. JAMA - Journal of the American Medical Association, 2015, 314, 913.	7.4	333
20	Whole-exome resequencing distinguishes cystic kidney diseases from phenocopies in renal ciliopathies. Kidney International, 2014, 85, 880-887.	5.2	67
21	Human HOX gene disorders. Molecular Genetics and Metabolism, 2014, 111, 4-15.	1.1	138
22	Functional analysis of candidate genes in 2q13 deletion syndrome implicates FBLN7 and TMEM87B deficiency in congenital heart defects and FBLN7 in craniofacial malformations. Human Molecular Genetics, 2014, 23, 4272-4284.	2.9	41
23	A Novel Intergenic ETnII-Î² Insertion Mutation Causes Multiple Malformations in Polypodia Mice. PLoS Genetics, 2013, 9, e1003967.	3.5	6
24	Analysis of De Novo <scp>HOXA</scp>13 Polyalanine Expansions Supports Replication Slippage Without Repair in Their Generation. American Journal of Medical Genetics, Part A, 2013, 161, 1019-1027.	1.2	8
25	Maternal intrachromosomal insertional translocation leads to recurrent 1q21.3q23.3 deletion in two siblings. American Journal of Medical Genetics, Part A, 2012, 158A, 2591-2601.	1.2	7
26	Multiple congenital anomalies and developmental delay in a boy associated with a de novo 16p13.3 deletion. American Journal of Medical Genetics, Part A, 2011, 155, 612-617.	1.2	7
27	BAC transgenic analysis reveals enhancers sufficient for Hoxa13 and neighborhood gene expression in mouse embryonic distal limbs and genital bud. Evolution & Development, 2008, 10, 421-432.	2.0	25
28	Expanded HOXA13 polyalanine tracts in a monotreme. Evolution & Development, 2008, 10, 433-438.	2.0	4
29	Molecular characterization of HOXA13 polyalanine expansion proteins in handâ€“footâ€“genital syndrome. American Journal of Medical Genetics, Part A, 2007, 143A, 3161-3168.	1.2	25
30	A mouse transgene drives embryonic dorsal posterior commissure expression. Transgenic Research, 2007, 16, 823-828.	2.4	0
31	Description and genetic mapping of Polypodia: an X-linked dominant mouse mutant with ectopic caudal limbs and other malformations. Mammalian Genome, 2006, 17, 903-913.	2.2	14
32	Range of HOX/TALE superclass associations and protein domain requirements for HOXA13:MEIS interaction. Developmental Biology, 2005, 277, 457-471.	2.0	60
33	Polyalanine expansion in HOXA13: three new affected families and the molecular consequences in a mouse model. Human Molecular Genetics, 2004, 13, 2841-2851.	2.9	47
34	Conserved expression domains for genes upstream and within the HoxA and HoxD clusters suggests a long-range enhancer existed before cluster duplication. Evolution & Development, 2004, 6, 423-430.	2.0	35
35	Microcephaly, jejunal atresia, aberrant right bronchus, ocular anomalies, and XY sex reversal. American Journal of Medical Genetics Part A, 2004, 125A, 293-298.	2.4	11
36	Two patients with monomelic ulnar duplication with mirror hand polydactyly: Segmental Laurin-Sandrow syndrome. American Journal of Medical Genetics Part A, 2004, 131A, 77-81.	2.4	11

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37	Juberg-Hayward syndrome: Report of a new patient with severe phenotype and novel clinical features. American Journal of Medical Genetics Part A, 2003, 122A, 257-260.	2.4	9
38	Possible third case of Lin-Gettig syndrome. American Journal of Medical Genetics Part A, 2002, 110, 380-383.	2.4	8
39	AHOXA13 allele with a missense mutation in the homeobox and a dinucleotide deletion in the promoter underlies Guttmacher syndrome. Human Mutation, 2002, 19, 573-574.	2.5	62
40	Integrative biology and the developing limb bud1. Evolution & Development, 2002, 4, 378-389.	2.0	4
41	Priming the search forHOX mutations. Teratology, 2002, 65, 47-49.	1.6	5
42	Apparently new syndrome of sensorineural hearing loss, retinal pigment epithelium lesions, and discolored teeth. , 1998, 75, 13-17.		6
43	Developmental Biology: Frontiers for Clinical Genetics: Limb development: molecular dysmorphology is at hand!. Clinical Genetics, 1998, 53, 337-348.	2.0	23
44	Autosomal dominant microcephaly with normal intelligence, short palpebral fissures, and digital anomalies. American Journal of Medical Genetics Part A, 1997, 71, 150-155.	2.4	12
45	Exclusion of BMP6 as a candidate gene for cleidocranial dysplasia. , 1997, 71, 292-297.		1
46	Familial Large Vestibular Aqueduct Syndrome. Laryngoscope, 1996, 106, 960-965.	2.0	94
47	Two highly polymorphic CA repeats in the Menkes gene (ATP7A). Human Genetics, 1995, 96, 355-6.	3.8	8