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	Integrative clinical genomics of metastatic cancer. Nature, 2017, 548, 297-303.	27.8	685
2	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
3	Human HOX gene disorders. Molecular Genetics and Metabolism, 2014, 111, 4-15.	1.1	138
4	Familial Large Vestibular Aqueduct Syndrome. Laryngoscope, 1996, 106, 960-965.	2.0	94
5	Spectrum of disease associated with partial lipodystrophy: lessons from a trial cohort. Clinical Endocrinology, 2017, 86, 698-707.	2.4	72
6	Whole-exome resequencing distinguishes cystic kidney diseases from phenocopies in renal ciliopathies. Kidney International, 2014, 85, 880-887.	5.2	67
7	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
8	Range of HOX/TALE superclass associations and protein domain requirements for HOXA13:MEIS interaction. Developmental Biology, 2005, 277, 457-471.	2.0	60
9	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
10	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
11	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
12	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
13	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
14	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
15	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
16	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
17	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
18	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

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19	Developmental Biology: Frontiers for Clinical Genetics: Limb development: molecular dysmorphology is at hand!. Clinical Genetics, 1998, 53, 337-348.	2.0	23
20	Phenotypic spectrum and transcriptomic profile associated with germline variants in TRAF7. Genetics in Medicine, 2020, 22, 1215-1226.	2.4	22
21	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
22	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
23	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
24	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
25	Metreleptin therapy for nonalcoholic steatohepatitis: Open-label therapy interventions in two different clinical settings. Med, 2021, 2, 814-835.e6.	4.4	12
26	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
27	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
28	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
29	Two highly polymorphic CA repeats in the Menkes gene (ATP7A). Human Genetics, 1995, 96, 355-6.	3.8	8
30	Possible third case of Lin-Gettig syndrome. American Journal of Medical Genetics Part A, 2002, 110, 380-383.	2.4	8
31	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
32	Potential association of LMNA-associated generalized lipodystrophy with juvenile dermatomyositis. Clinical Diabetes and Endocrinology, 2018, 4, 6.	2.7	8
33	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
34	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
35	Interstitial microdeletion of the 1p34.3p34.2 region. Molecular Genetics & Genomic Medicine, 2018, 6, 673-677.	1.2	7
36	Cardiac phenotype in familial partial lipodystrophy. Clinical Endocrinology, 2021, 94, 1043-1053.	2.4	7

#	Article	IF	Citations
37	Apparently new syndrome of sensorineural hearing loss, retinal pigment epithelium lesions, and discolored teeth. , 1998, 75, 13-17.		6
38	A Novel Intergenic ETnII-Î² Insertion Mutation Causes Multiple Malformations in Polypodia Mice. PLoS Genetics, 2013, 9, e1003967.	3.5	6
39	Priming the search forHOX mutations. Teratology, 2002, 65, 47-49.	1.6	5
40	AHDC1 missense mutations in Xia-Gibbs syndrome. Human Genetics and Genomics Advances, 2021, 2, 100049.	1.7	5
41	Integrative biology and the developing limb bud1. Evolution & Development, 2002, 4, 378-389.	2.0	4
42	Expanded HOXA13 polyalanine tracts in a monotreme. Evolution & Development, 2008, 10, 433-438.	2.0	4
43	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
44	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
45	Exclusion of BMP6 as a candidate gene for cleidocranial dysplasia. , 1997, 71, 292-297.		1
46	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
47	A mouse transgene drives embryonic dorsal posterior commissure expression. Transgenic Research, 2007, 16, 823-828.	2.4	0