

Matthew J Edwards

List of Publications by Year in descending order

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Version: 2024-02-01

60
papers

2,331
citations

201385

27
h-index

243296

44
g-index

61
all docs

61
docs citations

61
times ranked

3512
citing authors

#	ARTICLE	IF	CITATIONS
1	Meta-Analysis of 13 Genome Scans Reveals Multiple Cleft Lip/Palate Genes with Novel Loci on 9q21 and 2q32-35. <i>American Journal of Human Genetics</i> , 2004, 75, 161-173.	2.6	200
2	Teratogen update: Gestational effects of maternal hyperthermia due to febrile illnesses and resultant patterns of defects in humans. , 1998, 58, 209-221.		177
3	Feasibility of Ultra-Rapid Exome Sequencing in Critically Ill Infants and Children With Suspected Monogenic Conditions in the Australian Public Health Care System. <i>JAMA - Journal of the American Medical Association</i> , 2020, 323, 2503.	3.8	160
4	Mutation Analysis of the MEN1 Gene in Multiple Endocrine Neoplasia Type 1, Familial Acromegaly and Familial Isolated Hyperparathyroidism. <i>Journal of Clinical Endocrinology and Metabolism</i> , 1998, 83, 2621-2626.	1.8	125
5	GDF5 Is a Second Locus for Multiple-Synostosis Syndrome. <i>American Journal of Human Genetics</i> , 2006, 78, 708-712.	2.6	83
6	ACTB Loss-of-Function Mutations Result in a Pleiotropic Developmental Disorder. <i>American Journal of Human Genetics</i> , 2017, 101, 1021-1033.	2.6	83
7	Recurrence of lethal osteogenesis imperfecta due to parental mosaicism for a mutation in the COL1A2 gene of type I collagen. The mosiac parent exhibits phenotypic features of a mild form of the disease. <i>Human Mutation</i> , 1992, 1, 47-54.	1.1	80
8	The adult phenotype in Costello syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2005, 136A, 128-135.	0.7	72
9	Mixoploidy in humans: Two surviving cases of diploid-tetraploid mixoploidy and comparison with diploid-triploid mixoploidy. <i>American Journal of Medical Genetics Part A</i> , 1994, 52, 324-330.	2.4	67
10	Molecular consequences of dominant Bethlem myopathy collagen VI mutations. <i>Annals of Neurology</i> , 2007, 62, 390-405.	2.8	66
11	Mutations in KCTD1 Cause Scalp-Ear-Nipple Syndrome. <i>American Journal of Human Genetics</i> , 2013, 92, 621-626.	2.6	65
12	Case-control study of cleft lip or palate after maternal use of topical corticosteroids during pregnancy. <i>American Journal of Medical Genetics Part A</i> , 2003, 120A, 459-463.	2.4	64
13	Ectopia lentis phenotypes and theFBN1gene. , 2004, 126A, 284-289.		64
14	The Impact of Huntington's Disease on Family Life. <i>Psychosomatics</i> , 2007, 48, 400-404.	2.5	64
15	Twenty-six novelEFNB1 mutations in familial and sporadic craniofrontonasal syndrome (CFNS). <i>Human Mutation</i> , 2005, 26, 113-118.	1.1	61
16	Differential Expression of Pyloric Atresia in Junctional Epidermolysis Bullosa with ITGB4 Mutations Suggests that Pyloric Atresia is due to Factors Other than the Mutations and Not Predictive of a Poor Outcome: Three Novel Mutations and a Review of the Li. <i>Acta Dermato-Venereologica</i> , 2008, 88, 438-448.	0.6	61
17	OA1 Mutations and Deletions in X-Linked Ocular Albinism. <i>American Journal of Human Genetics</i> , 1998, 62, 800-809.	2.6	60
18	Malignant melanoma in patients with multiple endocrine neoplasia type 1 and involvement of theMEN1 gene in sporadic melanoma. <i>International Journal of Cancer</i> , 2000, 87, 463-467.	2.3	47

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19	Neurogenetic fetal akinesia and arthrogryposis: genetics, expanding genotype-phenotypes and functional genomics. <i>Journal of Medical Genetics</i> , 2021, 58, 609-618.	1.5	46
20	Premature arthritis is a distinct type II collagen phenotype. <i>Arthritis and Rheumatism</i> , 2010, 62, 1421-1430.	6.7	45
21	Standardized practices for RNA diagnostics using clinically accessible specimens reclassifies 75% of putative splicing variants. <i>Genetics in Medicine</i> , 2022, 24, 130-145.	1.1	45
22	Scalp-ear-nipple syndrome: Additional manifestations. <i>American Journal of Medical Genetics Part A</i> , 1994, 50, 247-250.	2.4	44
23	Evaluation of Satisfaction of Parents With the Use of Videoconferencing for a Pediatric Genetic Consultation. <i>Twin Research and Human Genetics</i> , 2011, 14, 343-346.	0.3	41
24	The natural history and osteodystrophy of mucopolidosis types II and III. <i>Journal of Paediatrics and Child Health</i> , 2010, 46, 316-322.	0.4	38
25	Posterior Nuchal Cystic Hygroma. <i>Clinics in Perinatology</i> , 1990, 17, 611-640.	0.8	37
26	Prenatal Diagnosis of congenital disorder of glycosylation type Ia (CDG-Ia) by cordocentesis and transferrin isoelectric focussing of serum of a 27-week fetus with non-immune hydrops. <i>Prenatal Diagnosis</i> , 2006, 26, 985-988.	1.1	35
27	X-linked recessive inheritance of an orofaciogigital syndrome with partial expression in females and survival of affected males. <i>Clinical Genetics</i> , 1988, 34, 325-332.	1.0	33
28	Functional genomics and gene-environment interaction highlight the complexity of congenital heart disease caused by Notch pathway variants. <i>Human Molecular Genetics</i> , 2020, 29, 566-579.	1.4	32
29	Deletion in Blood Mitochondrial DNA in Kearns-Sayre Syndrome. <i>Pediatric Research</i> , 1992, 31, 557-560.	1.1	29
30	Clinical report: one year of treatment of Proteus syndrome with miransertib (ARQ 092). <i>Journal of Physical Education and Sports Management</i> , 2020, 6, a004549.	0.5	27
31	Clinical and linkage study of a large family with simple ectopia lentis linked to FBN1. <i>American Journal of Medical Genetics Part A</i> , 1994, 53, 65-71.	2.4	26
32	Atypical nested 22q11.2 duplications between $\langle scp \rangle LCR \langle /scp \rangle 22B$ and $\langle scp \rangle LCR \langle /scp \rangle 22D$ are associated with neurodevelopmental phenotypes including autism spectrum disorder with incomplete penetrance. <i>Molecular Genetics & Genomic Medicine</i> , 2019, 7, e00507.	0.6	26
33	PEHO and PEHO-like syndromes: Report of five Australian cases. , 2003, 122A, 6-12.		23
34	Paternal uniparental isodisomy for chromosome 14 with mosaicism for a supernumerary marker chromosome 14. <i>American Journal of Medical Genetics, Part A</i> , 2007, 143A, 2165-2171.	0.7	22
35	Hypertrichosis \hat{a} cubiti \hat{a} with facial asymmetry. <i>American Journal of Medical Genetics Part A</i> , 1994, 53, 56-58.	2.4	17
36	Agnathia (severe microgathia), aglossia and choanal atresia in an infant. <i>Journal of Paediatrics and Child Health</i> , 1995, 31, 358-361.	0.4	16

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37	Investigation of molybdenum cofactor deficiency due to MOCS2 deficiency in a newborn baby. <i>Meta Gene</i> , 2015, 3, 43-49.	0.3	15
38	Herrmann multiple synostosis syndrome with neurological complications caused by spinal canal stenosis. <i>American Journal of Medical Genetics Part A</i> , 2000, 95, 118-122.	2.4	14
39	Genetic selection of embryos that later develop the metabolic syndrome. <i>Medical Hypotheses</i> , 2012, 78, 621-625.	0.8	13
40	Nevoid hypertrichosis with multiple patches of hair that underwent almost complete spontaneous resolution. , 1998, 79, 195-196.		12
41	Studies of type I collagen in osteogenesis imperfecta. <i>Journal of Pediatrics</i> , 1990, 117, 67-72.	0.9	10
42	7p22.1 microdeletions involving ACTB associated with developmental delay, short stature, and microcephaly. <i>European Journal of Medical Genetics</i> , 2016, 59, 502-506.	0.7	10
43	Genetic variation affecting DNA methylation and the human imprinting disorder, Beckwith-Wiedemann syndrome. <i>Clinical Epigenetics</i> , 2018, 10, 114.	1.8	10
44	A familial 7q36.3 duplication associated with agenesis of the corpus callosum. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 2201-2208.	0.7	9
45	Knowledge, attitudes and opinions towards measles and the MMR vaccine across two NSW cohorts. <i>Australian and New Zealand Journal of Public Health</i> , 2017, 41, 641-646.	0.8	9
46	The Barker Hypothesis. , 2017, , 1-21.		9
47	Diverse phenotypic consequences of mutations affecting the C-terminus of FLNA. <i>Journal of Molecular Medicine</i> , 2015, 93, 773-782.	1.7	8
48	Paediatric hospitalisations for lower respiratory tract infections in Mount Isa. <i>Medical Journal of Australia</i> , 2014, 200, 591-594.	0.8	7
49	Familial translocation 5;14 resulting in an unbalanced offspring. <i>American Journal of Medical Genetics Part A</i> , 1991, 39, 362-366.	2.4	6
50	Esophageal dysmotility in brothers with an FG-like syndrome. <i>American Journal of Medical Genetics Part A</i> , 2000, 91, 185-189.	2.4	5
51	Engagement of undergraduate medical students of paediatrics in special schools for children with disabilities. <i>Journal of Paediatrics and Child Health</i> , 2015, 51, 798-801.	0.4	4
52	Supernumerary ring chromosome 17 identified by fluorescent in situ hybridization. , 1997, 69, 352-355.		3
53	A novel genetic syndrome characterized by pediatric cataract, dysmorphism, ectodermal features, and developmental delay in an indigenous Australian family. <i>American Journal of Medical Genetics, Part A</i> , 2009, 149A, 633-639.	0.7	2
54	The Historical Development of Obstetric Anesthesia and Its Contributions to Perinatology. <i>American Journal of Perinatology</i> , 2017, 34, 211-216.	0.6	2

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55	Predictive diagnosis of multiple endocrine neoplasia (MEN 1) in four Australian kindreds. Australian and New Zealand Journal of Medicine, 1996, 26, 27-32.	0.5	1
56	Syndrome diagnosis with single nucleotide polymorphism (<sc>SNP</sc>) microarray. Journal of Paediatrics and Child Health, 2016, 52, 85-89.	0.4	1
57	Ten-year review of <sc>H</sc>enoch-<sc>S</sc>chonlein purpura in <sc>C</sc>ampbelltown <sc>H</sc>ospital, south-western <sc>S</sc>ydney. Journal of Paediatrics and Child Health, 2014, 50, 840-840.	0.4	0
58	<sc>K</sc>awasaki disease in <sc>C</sc>ampbelltown, a suburban hospital. Journal of Paediatrics and Child Health, 2015, 51, 466-467.	0.4	0
59	A warning about tuberculosis. Journal of Paediatrics and Child Health, 2015, 51, 1033-1035.	0.4	0
60	Famines, Pregnancy and Effect on the Adults. , 2017, , 357-369.		0