Matthew J Edwards

List of Publications by Year
in descending order

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	Standardized practices for RNA diagnostics using clinically accessible specimens reclassifies 75% of putative splicing variants. Genetics in Medicine, 2022, 24, 130-145.	1.1	45
2	Neurogenetic fetal akinesia and arthrogryposis: genetics, expanding genotype-phenotypes and functional genomics. Journal of Medical Genetics, 2021, 58, 609-618.	1.5	46
3	Functional genomics and gene-environment interaction highlight the complexity of congenital heart disease caused by Notch pathway variants. Human Molecular Genetics, 2020, 29, 566-579.	1.4	32
4	Feasibility of Ultra-Rapid Exome Sequencing in Critically Ill Infants and Children With Suspected Monogenic Conditions in the Australian Public Health Care System. JAMA - Journal of the American Medical Association, 2020, 323, 2503.	3.8	160
5	Clinical report: one year of treatment of Proteus syndrome with miransertib (ARQ 092). Journal of Physical Education and Sports Management, 2020, 6, a004549.	0.5	27
6	Atypical nested 22q11.2 duplications between <scp>LCR</scp>22B and <scp>LCR</scp>22D are associated with neurodevelopmental phenotypes including autism spectrum disorder with incomplete penetrance. Molecular Genetics & Genomic Medicine, 2019, 7, e00507.	0.6	26
7	Genetic variation affecting DNA methylation and the human imprinting disorder, Beckwith-Wiedemann syndrome. Clinical Epigenetics, 2018, 10, 114.	1.8	10
8	Knowledge, attitudes and opinions towards measles and the MMR vaccine across two NSW cohorts. Australian and New Zealand Journal of Public Health, 2017, 41, 641-646.	0.8	9
9	ACTB Loss-of-Function Mutations Result in a Pleiotropic Developmental Disorder. American Journal of Human Genetics, 2017, 101, 1021-1033.	2.6	83
10	The Historical Development of Obstetric Anesthesia and Its Contributions to Perinatology. American Journal of Perinatology, 2017, 34, 211-216.	0.6	2
11	The Barker Hypothesis. , 2017, , 1-21.		9
12	Famines, Pregnancy and Effect on the Adults. , 2017, , 357-369.		0
13	Syndrome diagnosis with singleâ€nucleotide polymorphism (<scp>SNP</scp>) microarray. Journal of Paediatrics and Child Health, 2016, 52, 85-89.	0.4	1
14	7p22.1 microdeletions involving ACTB associated with developmental delay, short stature, and microcephaly. European Journal of Medical Genetics, 2016, 59, 502-506.	0.7	10
15	Engagement of undergraduate medical students of paediatrics in special schools for children with disabilities. Journal of Paediatrics and Child Health, 2015, 51, 798-801.	0.4	4
16	A familial 7q36.3 duplication associated with agenesis of the corpus callosum. American Journal of Medical Genetics, Part A, 2015, 167, 2201-2208.	0.7	9
17	Investigation of molybdenum cofactor deficiency due to MOCS2 deficiency in a newborn baby. Meta Gene, 2015, 3, 43-49.	0.3	15
18	Diverse phenotypic consequences of mutations affecting the C-terminus of FLNA. Journal of Molecular Medicine, 2015, 93, 773-782.	1.7	8

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19	<scp>K</scp>awasaki disease in <scp>C</scp>ampbelltown, a suburban hospital. Journal of Paediatrics and Child Health, 2015, 51, 466-467.	0.4	0
20	A warning about tuberculosis. Journal of Paediatrics and Child Health, 2015, 51, 1033-1035.	0.4	0
21	Paediatric hospitalisations for lower respiratory tract infections in Mount Isa. Medical Journal of Australia, 2014, 200, 591-594.	0.8	7
22	Tenâ€year review of <scp>H</scp>enochâ€“<scp>S</scp>chonlein purpura in <scp>C</scp>ampbelltown <scp>H</scp>ospital, southâ€western <scp>S</scp>ydney. Journal of Paediatrics and Child Health, 2014, 50, 840-840.	0.4	0
23	Mutations in KCTD1 Cause Scalp-Ear-Nipple Syndrome. American Journal of Human Genetics, 2013, 92, 621-626.	2.6	65
24	Genetic selection of embryos that later develop the metabolic syndrome. Medical Hypotheses, 2012, 78, 621-625.	0.8	13
25	Evaluation of Satisfaction of Parents With the Use of Videoconferencing for a Pediatric Genetic Consultation. Twin Research and Human Genetics, 2011, 14, 343-346.	0.3	41
26	Premature arthritis is a distinct type II collagen phenotype. Arthritis and Rheumatism, 2010, 62, 1421-1430.	6.7	45
27	The natural history and osteodystrophy of mucolipidosis types II and III. Journal of Paediatrics and Child Health, 2010, 46, 316-322.	0.4	38
28	A novel genetic syndrome characterized by pediatric cataract, dysmorphism, ectodermal features, and developmental delay in an indigenous Australian family. American Journal of Medical Genetics, Part A, 2009, 149A, 633-639.	0.7	2
29	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. Acta Dermato-Venereologica, 2008, 88, 438-448.	0.6	61
30	The Impact of Huntington's Disease on Family Life. Psychosomatics, 2007, 48, 400-404.	2.5	64
31	Paternal uniparental isodisomy for chromosome 14 with mosaicism for a supernumerary marker chromosome 14. American Journal of Medical Genetics, Part A, 2007, 143A, 2165-2171.	0.7	22
32	Molecular consequences of dominant Bethlem myopathy collagen VI mutations. Annals of Neurology, 2007, 62, 390-405.	2.8	66
33	GDF5 Is a Second Locus for Multiple-Synostosis Syndrome. American Journal of Human Genetics, 2006, 78, 708-712.	2.6	83
34	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. Prenatal Diagnosis, 2006, 26, 985-988.	1.1	35
35	Twenty-six novelEFNB1 mutations in familial and sporadic craniofrontonasal syndrome (CFNS). Human Mutation, 2005, 26, 113-118.	1.1	61
36	The adult phenotype in Costello syndrome. American Journal of Medical Genetics, Part A, 2005, 136A, 128-135.	0.7	72

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37	Ectopia lentis phenotypes and theFBN1gene. , 2004, 126A, 284-289.		64
38	Meta-Analysis of 13 Genome Scans Reveals Multiple Cleft Lip/Palate Genes with Novel Loci on 9q21 and 2q32-35. American Journal of Human Genetics, 2004, 75, 161-173.	2.6	200
39	Case-control study of cleft lip or palate after maternal use of topical corticosteroids during pregnancy. American Journal of Medical Genetics Part A, 2003, 120A, 459-463.	2.4	64
40	PEHO and PEHO-like syndromes: Report of five Australian cases. , 2003, 122A, 6-12.		23
41	Esophageal dysmotility in brothers with an FG-like syndrome. American Journal of Medical Genetics Part A, 2000, 91, 185-189.	2.4	5
42	Herrmann multiple synostosis syndrome with neurological complications caused by spinal canal stenosis. American Journal of Medical Genetics Part A, 2000, 95, 118-122.	2.4	14
43	Malignant melanoma in patients with multiple endocrine neoplasia type 1 and involvement of theMEN1 gene in sporadic melanoma. International Journal of Cancer, 2000, 87, 463-467.	2.3	47
44	Nevoid hypertrichosis with multiple patches of hair that underwent almost complete spontaneous resolution. , 1998, 79, 195-196.		12
45	Teratogen update: Gestational effects of maternal hyperthermia due to febrile illnesses and resultant patterns of defects in humans. , 1998, 58, 209-221.		177
46	OA1 Mutations and Deletions in X-Linked Ocular Albinism. American Journal of Human Genetics, 1998, 62, 800-809.	2.6	60
47	Mutation Analysis of the MEN1 Gene in Multiple Endocrine Neoplasia Type 1, Familial Acromegaly and Familial Isolated Hyperparathyroidism. Journal of Clinical Endocrinology and Metabolism, 1998, 83, 2621-2626.	1.8	125
48	Supernumerary ring chromosome 17 identified by fluorescent in situ hybridization. , 1997, 69, 352-355.		3
49	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
50	Agnathia (severe microghathia), aglossia and choanal atresia in an infant. Journal of Paediatrics and Child Health, 1995, 31, 358-361.	0.4	16
51	Scalp-ear-nipple syndrome: Additional manifestations. American Journal of Medical Genetics Part A, 1994, 50, 247-250.	2.4	44
52	Mixoploidy in humans: Two surviving cases of diploid-tetraploid mixoploidy and comparison with diploid-triploid mixoploidy. American Journal of Medical Genetics Part A, 1994, 52, 324-330.	2.4	67
53	Hypertrichosis â€œcubitiâ€•with facial asymmetry. American Journal of Medical Genetics Part A, 1994, 53, 56-58.	2.4	17
54	Clinical and linkage study of a large family with simple ectopia lentis linked to FBN1. American Journal of Medical Genetics Part A, 1994, 53, 65-71.	2.4	26

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55	Deletion in Blood Mitochondrial DNA in Kearns-Sayre Syndrome. Pediatric Research, 1992, 31, 557-560.	1.1	29
56	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. Human Mutation, 1992, 1, 47-54.	1.1	80
57	Familial translocation 5;14 resulting in an unbalanced offspring. American Journal of Medical Genetics Part A, 1991, 39, 362-366.	2.4	6
58	Posterior Nuchal Cystic Hygroma. Clinics in Perinatology, 1990, 17, 611-640.	0.8	37
59	Studies of type I collagen in osteogenesis imperfecta. Journal of Pediatrics, 1990, 117, 67-72.	0.9	10
60	Xâ€linked recessive inheritance of an orofaciodigital syndrome with partial expression in females and survival of affected males. Clinical Genetics, 1988, 34, 325-332.	1.0	33