Gareth S Baynam

List of Publications by Year
in descending order

113 papers	6,271 citations	136740 32 h-index	82410 72 g-index
114 all docs	114 docs citations	114 times ranked	11261 citing authors

#	Article	IF	Citations
1	Epidemiology of Rare Craniofacial Anomalies: Retrospective Western Australian Population Data Linkage Study. Journal of Pediatrics, 2022, 241, 162-172.e9.	0.9	11
2	Novel diagnostic DNA methylation episignatures expand and refine the epigenetic landscapes of Mendelian disorders. Human Genetics and Genomics Advances, 2022, 3, 100075.	1.0	42
3	CRISPR single base editing, neuronal disease modelling and functional genomics for genetic variant analysis: pipeline validation using Kleefstra syndrome EHMT1 haploinsufficiency. Stem Cell Research and Therapy, 2022, 13, 69.	2.4	9
4	Association between craniofacial anomalies, intellectual disability and autism spectrum disorder: Western Australian population-based study. Pediatric Research, 2022, 92, 1795-1804.	1.1	8
5	Functional validation of variants of unknown significance using CRISPR gene editing and transcriptomics: A Kleefstra syndrome case study. Gene, 2022, 821, 146287.	1.0	6
6	Common data elements to standardize genomics studies in cerebral palsy. Developmental Medicine and Child Neurology, 2022, 64, 1470-1476.	1.1	4
7	Unlocking sociocultural and community factors for the global adoption of genomic medicine. Orphanet Journal of Rare Diseases, 2022, 17, 191.	1.2	1
8	Further evidence for distinct traits associated with <scp>RBM10</scp> missense variants. Clinical Genetics, 2022, 102, 161-163.	1.0	2
9	The risk of major structural birth defects associated with seasonal influenza vaccination during pregnancy: A populationâ€based cohort study. Birth Defects Research, 2022, 114, 1244-1256.	0.8	5
10	The GA4GH Phenopacket schema defines a computable representation of clinical data. Nature Biotechnology, 2022, 40, 817-820.	9.4	38
11	Fatal Perinatal Mitochondrial Cardiac Failure Caused by Recurrent De Novo Duplications in the ATAD3 Locus. Med, 2021, 2, 49-73.e10.	2.2	33
12	The Human Phenotype Ontology in 2021. Nucleic Acids Research, 2021, 49, D1207-D1217.	6.5	652
13	Culturally competent communication in Indigenous disability assessment: a qualitative study. International Journal for Equity in Health, 2021, 20, 68.	1.5	4
14	Severe congenital cutis laxa: Identification of novel homozygous <scp>LOX</scp> gene variants in two families. Clinical Genetics, 2021, 100, 168-175.	1.0	3
15	â€œThis is my boyâ€™s health! Talk straight to me!â€•perspectives on accessible and culturally safe care among Aboriginal and Torres Strait Islander patients of clinical genetics services. International Journal for Equity in Health, 2021, 20, 103.	1.5	9
16	Genotypeâ€“phenotype correlations and novel molecular insights into the DHX30-associated neurodevelopmental disorders. Genome Medicine, 2021, 13, 90.	3.6	16
17	Birth prevalence of congenital heart defects in Western Australia, 1990â€“2016. Journal of Paediatrics and Child Health, 2021, 57, 1672-1680.	0.4	7
18	Deletion of ERF and CIC causes abnormal skull morphology and global developmental delay. Journal of Physical Education and Sports Management, 2021, 7, a005991.	0.5	3

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19	Ethics and equity in rare disease research and healthcare. Personalized Medicine, 2021, 18, 407-416.	0.8	0
20	Paediatric genomic testing: Navigating genomic reports for the general paediatrician. Journal of Paediatrics and Child Health, 2021, , .	0.4	3
21	Innovation in Informatics to Improve Clinical Care and Drug Accessibility for Rare Diseases in China. Frontiers in Pharmacology, 2021, 12, 719415.	1.6	3
22	Microcephaly in Australian children, 2016â€“2018: national surveillance study. Archives of Disease in Childhood, 2021, 106, 849-854.	1.0	5
23	Childhood rare diseases and the UN convention on the rights of the child. Orphanet Journal of Rare Diseases, 2021, 16, 523.	1.2	7
24	mEDUrare: Supporting Integrated Care for Rare Diseases by Better Connecting Health and Education Through Policy.. Yale Journal of Biology and Medicine, 2021, 94, 693-702.	0.2	1
25	A call for global action for rare diseases in Africa. Nature Genetics, 2020, 52, 21-26.	9.4	31
26	SMART Work Design: Accelerating the Diagnosis of Rare Diseases in the Western Australian Undiagnosed Diseases Program. Frontiers in Pediatrics, 2020, 8, 582.	0.9	3
27	The case for open science: rare diseases. JAMIA Open, 2020, 3, 472-486.	1.0	33
28	Protecting the rare during a rare pandemic. Medical Journal of Australia, 2020, 213, 94.	0.8	2
29	Equitable Expanded Carrier Screening Needs Indigenous Clinical and Population Genomic Data. American Journal of Human Genetics, 2020, 107, 175-182.	2.6	24
30	A flexible computational pipeline for research analyses of unsolved clinical exome cases. Npj Genomic Medicine, 2020, 5, 54.	1.7	4
31	Barriers and Considerations for Diagnosing Rare Diseases in Indigenous Populations. Frontiers in Pediatrics, 2020, 8, 579924.	0.9	25
32	Rights, interests and expectations: Indigenous perspectives on unrestricted access to genomic data. Nature Reviews Genetics, 2020, 21, 377-384.	7.7	141
33	Pathobiologic Mechanisms of Neurodegeneration in Osteopetrosis Derived From Structural and Functional Analysis of 14 ClC-7 Mutants. Journal of Bone and Mineral Research, 2020, 36, 531-545.	3.1	16
34	How many rare diseases are there?. Nature Reviews Drug Discovery, 2020, 19, 77-78.	21.5	204
35	Consideration of a Legislative Framework to Support the Diagnostic Odyssey Commonly Encountered in the Instance of Rare Disease. Journal of Law & Medicine, 2020, 27, 634-644.	0.0	0
36	Editorial: Public Health Genomics. Frontiers in Public Health, 2019, 7, 142.	1.3	1

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37	Hypospadias Prevalence and Trends in International Birth Defect Surveillance Systems, 1980â€“2010. European Urology, 2019, 76, 482-490.	0.9	74
38	Autosomal recessive congenital ichthyosis due to homozygous variants in NIPAL4 with a dramatic response to ustekinumab. Pediatric Dermatology, 2019, 36, 1002-1003.	0.5	19
39	Genomic Testing for Human Health and Disease Across the Life Cycle: Applications and Ethical, Legal, and Social Challenges. Frontiers in Public Health, 2019, 7, 40.	1.3	37
40	Enabling Global Clinical Collaborations on Identifiable Patient Data: The Minerva Initiative. Frontiers in Genetics, 2019, 10, 611.	1.1	14
41	Healthcare System Priorities for Successful Integration of Genomics: An Australian Focus. Frontiers in Public Health, 2019, 7, 41.	1.3	18
42	Genetic or Other Causation Should Not Change the Clinical Diagnosis of Cerebral Palsy. Journal of Child Neurology, 2019, 34, 472-476.	0.7	82
43	Optimizing Precision Medicine for Public Health. Frontiers in Public Health, 2019, 7, 42.	1.3	58
44	A Diagnosis for All Rare Genetic Diseases: The Horizon and the Next Frontiers. Cell, 2019, 177, 32-37.	13.5	113
45	Defining and expanding the phenotype of QARS-associated developmental epileptic encephalopathy. Neurology: Genetics, 2019, 5, e373.	0.9	5
46	Personalised analytics for rare disease diagnostics. Nature Communications, 2019, 10, 5274.	5.8	15
47	Expansion of the Human Phenotype Ontology (HPO) knowledge base and resources. Nucleic Acids Research, 2019, 47, D1018-D1027.	6.5	539
48	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
49	Incidental inequity. European Journal of Human Genetics, 2018, 26, 616-617.	1.4	7
50	Plain-language medical vocabulary for precision diagnosis. Nature Genetics, 2018, 50, 474-476.	9.4	28
51	A Clinical Review of Generalized Overgrowth Syndromes in the Era of Massively Parallel Sequencing. Molecular Syndromology, 2018, 9, 70-82.	0.3	36
52	Maternal variants in NLRP and other maternal effect proteins are associated with multilocus imprinting disturbance in offspring. Journal of Medical Genetics, 2018, 55, 497-504.	1.5	126
53	A review of structural brain abnormalities in Pallisterâ€Killian syndrome. Molecular Genetics & Genomic Medicine, 2018, 6, 92-98.	0.6	14
54	Progress in Rare Diseases Research 2010â€“2016: An IRDiRC Perspective. Clinical and Translational Science, 2018, 11, 11-20.	1.5	104

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55	Future of Rare Diseases Research 2017â€“2027: An IRDiRC Perspective. Clinical and Translational Science, 2018, 11, 21-27.	1.5	154
56	Silver Russel syndrome in an aboriginal patient from Australia. American Journal of Medical Genetics, Part A, 2018, 176, 2561-2563.	0.7	13
57	Editorial: Precision Public Health. Frontiers in Public Health, 2018, 6, 121.	1.3	50
58	Changes to the Employers' Use of Genetic Information and Non-discrimination for Health Insurance in the USA: Implications for Australians. Frontiers in Public Health, 2018, 6, 183.	1.3	1
59	Elucidating the genetic architecture of Adams-Oliver syndrome in a large European cohort. Human Mutation, 2018, 39, 1246-1261.	1.1	31
60	The Human Phenotype Ontology in 2017. Nucleic Acids Research, 2017, 45, D865-D876.	6.5	699
61	â€˜IRDiRC Recognized Resourcesâ€™: a new mechanism to support scientists to conduct efficient, high-quality research for rare diseases. European Journal of Human Genetics, 2017, 25, 162-165.	1.4	30
62	Genotype and phenotype spectrum of NRAS germline variants. European Journal of Human Genetics, 2017, 25, 823-831.	1.4	36
63	International Cooperation to Enable the Diagnosis of All Rare Genetic Diseases. American Journal of Human Genetics, 2017, 100, 695-705.	2.6	305
64	The International Rare Diseases Research Consortium: Policies and Guidelines to maximize impact. European Journal of Human Genetics, 2017, 25, 1293-1302.	1.4	62
65	Improved Diagnosis and Care for Rare Diseases through Implementation of Precision Public Health Framework. Advances in Experimental Medicine and Biology, 2017, 1031, 55-94.	0.8	20
66	The collective impact of rare diseases in Western Australia: an estimate using a population-based cohort. Genetics in Medicine, 2017, 19, 546-552.	1.1	77
67	Indigenous Genetics and Rare Diseases: Harmony, Diversity and Equity. Advances in Experimental Medicine and Biology, 2017, 1031, 511-520.	0.8	15
68	Outcomes of an International Workshop on Preconception Expanded Carrier Screening: Some Considerations for Governments. Frontiers in Public Health, 2017, 5, 25.	1.3	28
69	3-Dimensional Facial Analysisâ€”Facing Precision Public Health. Frontiers in Public Health, 2017, 5, 31.	1.3	13
70	Initiating an undiagnosed diseases program in the Western Australian public health system. Orphanet Journal of Rare Diseases, 2017, 12, 83.	1.2	24
71	Prevalence of microcephaly in an Australian populationâ€based birth defects register, 1980â€“2015. Medical Journal of Australia, 2017, 206, 351-356.	0.8	14
72	Translating Aboriginal genomics â€” four letters Closing the Gap. Medical Journal of Australia, 2016, 205, 379-379.	0.8	3

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73	The rare and undiagnosed diseases diagnostic service â€“ application of massively parallel sequencing in a state-wide clinical service. Orphanet Journal of Rare Diseases, 2016, 11, 77.	1.2	48
74	Mutation Update for Kabuki Syndrome GenesKMT2DandKDM6Aand Further Delineation of X-Linked Kabuki Syndrome Subtype 2. Human Mutation, 2016, 37, 847-864.	1.1	134
75	A high-throughput sequencing test for diagnosing inherited bleeding, thrombotic, and platelet disorders. Blood, 2016, 127, 2791-2803.	0.6	157
76	Automatic concept recognition using the Human Phenotype Ontology reference and test suite corpora. Database: the Journal of Biological Databases and Curation, 2015, 2015, bav005-bav005.	1.4	55
77	A germline MTOR mutation in Aboriginal Australian siblings with intellectual disability, dysmorphism, macrocephaly, and small thoraces. American Journal of Medical Genetics, Part A, 2015, 167, 1659-1667.	0.7	35
78	Undiagnosed Diseases Network International (UDNI): White paper for global actions to meet patient needs. Molecular Genetics and Metabolism, 2015, 116, 223-225.	0.5	69
79	Report and review of described associations of <scp>M</scp>ayerâ€<scp>R</scp>okitanskyâ€<scp>K</scp>Ã¼sterâ€<scp>H</scp>auser syndrome and <scp>S</scp>ilverâ€“<scp>R</scp>ussell syndrome. Journal of Paediatrics and Child Health, 2015, 51, 555-560.	0.4	10
80	The Human Phenotype Ontology: Semantic Unification of Common and Rare Disease. American Journal of Human Genetics, 2015, 97, 111-124.	2.6	203
81	Mutations in a TGF-Î² Ligand, TGFB3, CauseÂSyndromic Aortic Aneurysms andÂDissections. Journal of the American College of Cardiology, 2015, 65, 1324-1336.	1.2	238
82	Phenotyping: Targeting genotype's rich cousin for diagnosis. Journal of Paediatrics and Child Health, 2015, 51, 381-386.	0.4	29
83	Monitoring of Therapy for Mucopolysaccharidosis Type I Using Dysmorphometric Facial Phenotypic Signatures. JIMD Reports, 2015, 22, 99-106.	0.7	7
84	Modeling 3D Facial Shape from DNA. PLoS Genetics, 2014, 10, e1004224.	1.5	190
85	Multinodular Goiter in Children: An Important Pointer to a Germline DICER1 Mutation. Journal of Clinical Endocrinology and Metabolism, 2014, 99, 1947-1948.	1.8	30
86	Rare Disease Research Roadmap: Navigating the bioinformatics and translational challenges for improved patient health outcomes. Health Policy and Technology, 2014, 3, 325-335.	1.3	9
87	The Facial Evolution: Looking Backward and Moving Forward. Human Mutation, 2013, 34, 14-22.	1.1	36
88	Overexpression of Aromatase Associated With Loss of Heterozygosity of theSTK11Gene Accounts for Prepubertal Gynecomastia in Boys with Peutz-Jeghers Syndrome. Journal of Clinical Endocrinology and Metabolism, 2013, 98, E1979-E1987.	1.8	29
89	Objective Monitoring of mTOR Inhibitor Therapy by Three-Dimensional Facial Analysis. Twin Research and Human Genetics, 2013, 16, 840-844.	0.3	14
90	A Dysmorphometric Analysis to Investigate Facial Phenotypic Signatures as a Foundation for Non-invasive Monitoring of Lysosomal Storage Disorders. JIMD Reports, 2012, 8, 31-39.	0.7	9

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91	Congenital diaphragmatic hernia interval on chromosome 8p23.1 characterized by genetics and protein interaction networks. American Journal of Medical Genetics, Part A, 2012, 158A, 3148-3158.	0.7	42
92	Sexual dimorphism in multiple aspects of 3D facial symmetry and asymmetry defined by spatially dense geometric morphometrics. Journal of Anatomy, 2012, 221, 97-114.	0.9	84
93	The need for genetic studies of Indigenous Australians. Medical Journal of Australia, 2012, 196, 313-313.	0.8	12
94	Fetal akinesia: review of the genetics of the neuromuscular causes. Journal of Medical Genetics, 2011, 48, 793-801.	1.5	65
95	Carpenter syndrome: extended RAB23 mutation spectrum and analysis of nonsenseâ€mediated mRNA decay. Human Mutation, 2011, 32, E2069-78.	1.1	34
96	Extending the phenotypes associated withDICER1mutations. Human Mutation, 2011, 32, 1381-1384.	1.1	173
97	Intersections of Epigenetics, Twinning and Developmental Asymmetries: Insights Into Monogenic and Complex Diseases and a Role for 3D Facial Analysis. Twin Research and Human Genetics, 2011, 14, 305-315.	0.3	18
98	CANT1 mutation is also responsible for Desbuquois dysplasia, type 2 and Kim variant. Journal of Medical Genetics, 2011, 48, 32-37.	1.5	39
99	A child with an STK11 mutation and Sotos syndrome-like features: can STK11 mutations produce a Sotos syndrome phenocopy?. BMJ Case Reports, 2011, 2011, bcr0720114445-bcr0720114445.	0.2	1
100	A recurrence of a hydrop lethal skeletal dysplasia showing similarity to Desbuquois dysplasia and a proposed new sign: The Upsilon sign. American Journal of Medical Genetics, Part A, 2010, 152A, 966-969.	0.7	9
101	A Child With an FGFR3 Mutation, a Laterality Disorder and an Hepatoblastoma: Novel Associations and Possible Geneâ€“Environment Interactions. Twin Research and Human Genetics, 2010, 13, 297-300.	0.3	14
102	Cornelia de Lange Syndrome. Advances in Experimental Medicine and Biology, 2010, , 113-123.	0.8	20
103	Craniometaphyseal dysplasia and chondrocalcinosis cosegregating in a family with an ANKH mutation. American Journal of Medical Genetics, Part A, 2009, 149A, 1331-1333.	0.7	12
104	45,X/46,XY mosaicism and Oculo-Auriculo-Vertebral Spectrum following an IVF pregnancy: a report and a discussion of their interrelationships. Journal of Maxillofacial and Oral Surgery, 2009, 8, 279-282.	0.6	2
105	Deletion of 8p23.1 with features of Cornelia de Lange syndrome and congenital diaphragmatic hernia and a review of deletions of 8p23.1 to 8pter ? A further locus for Cornelia de Lange syndrome. American Journal of Medical Genetics, Part A, 2008, 146A, 1565-1570.	0.7	26
106	A c.1019Aâ€‰>â€‰G mutation in FGFR2, which predicts p.Tyr340Cys, in a lethally malformed fetus with Pfeiffer syndrome and multiple pterygia. American Journal of Medical Genetics, Part A, 2008, 146A, 2301-2303.	0.7	7
107	Mutation screening of fumarate hydratase by multiplex ligation-dependent probe amplification: detection of exonic deletion in a patient with leiomyomatosis and renal cell cancer. Cancer Genetics and Cytogenetics, 2008, 183, 83-88.	1.0	30
108	Gender-specific effects of cytokine gene polymorphisms on childhood vaccine responses. Vaccine, 2008, 26, 3574-3579.	1.7	25

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109	Impact of genetic variants in IL-4, IL-4 RA and IL-13 on the anti-pneumococcal antibody response. Vaccine, 2007, 25, 306-313.	1.7	38
110	Parental smoking impairs vaccine responses inÂchildren with atopic genotypes. Journal of Allergy and Clinical Immunology, 2007, 119, 366-374.	1.5	27
111	Associations of a novel IL4RA polymorphism, Ala57Thr, in Greenlander Inuit. Journal of Allergy and Clinical Immunology, 2006, 118, 627-634.	1.5	11
112	A case of 3q29 microdeletion with novel features and a review of cytogenetically visible terminal 3q deletions. Clinical Dysmorphology, 2006, 15, 145-148.	0.1	26
113	Digit-all: Rare Diseases. European Medical Journal Innovations, 0, , 11-16.	2.0	0