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	The Human Phenotype Ontology in 2017. Nucleic Acids Research, 2017, 45, D865-D876.	6.5	699
2	The Human Phenotype Ontology in 2021. Nucleic Acids Research, 2021, 49, D1207-D1217.	6.5	652
3	Expansion of the Human Phenotype Ontology (HPO) knowledge base and resources. Nucleic Acids Research, 2019, 47, D1018-D1027.	6.5	539
4	International Cooperation to Enable the Diagnosis of All Rare Genetic Diseases. American Journal of Human Genetics, 2017, 100, 695-705.	2.6	305
5	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
6	How many rare diseases are there?. Nature Reviews Drug Discovery, 2020, 19, 77-78.	21.5	204
7	The Human Phenotype Ontology: Semantic Unification of Common and Rare Disease. American Journal of Human Genetics, 2015, 97, 111-124.	2.6	203
8	Modeling 3D Facial Shape from DNA. PLoS Genetics, 2014, 10, e1004224.	1.5	190
9	Extending the phenotypes associated withDICER1mutations. Human Mutation, 2011, 32, 1381-1384.	1.1	173
10	A high-throughput sequencing test for diagnosing inherited bleeding, thrombotic, and platelet disorders. Blood, 2016, 127, 2791-2803.	0.6	157
11	Future of Rare Diseases Research 2017â€“2027: An IRDiRC Perspective. Clinical and Translational Science, 2018, 11, 21-27.	1.5	154
12	Rights, interests and expectations: Indigenous perspectives on unrestricted access to genomic data. Nature Reviews Genetics, 2020, 21, 377-384.	7.7	141
13	Mutation Update for Kabuki Syndrome GenesKMT2DandKDM6Aand Further Delineation of X-Linked Kabuki Syndrome Subtype 2. Human Mutation, 2016, 37, 847-864.	1.1	134
14	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
15	A Diagnosis for All Rare Genetic Diseases: The Horizon and the Next Frontiers. Cell, 2019, 177, 32-37.	13.5	113
16	Progress in Rare Diseases Research 2010â€“2016: An IRDiRC Perspective. Clinical and Translational Science, 2018, 11, 11-20.	1.5	104
17	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
18	Genetic or Other Causation Should Not Change the Clinical Diagnosis of Cerebral Palsy. Journal of Child Neurology, 2019, 34, 472-476.	0.7	82

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19	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
20	Hypospadias Prevalence and Trends in International Birth Defect Surveillance Systems, 1980â€“2010. European Urology, 2019, 76, 482-490.	0.9	74
21	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
22	Fetal akinesia: review of the genetics of the neuromuscular causes. Journal of Medical Genetics, 2011, 48, 793-801.	1.5	65
23	The International Rare Diseases Research Consortium: Policies and Guidelines to maximize impact. European Journal of Human Genetics, 2017, 25, 1293-1302.	1.4	62
24	Optimizing Precision Medicine for Public Health. Frontiers in Public Health, 2019, 7, 42.	1.3	58
25	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
26	Editorial: Precision Public Health. Frontiers in Public Health, 2018, 6, 121.	1.3	50
27	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
28	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
29	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
30	CANT1 mutation is also responsible for Desbuquois dysplasia, type 2 and Kim variant. Journal of Medical Genetics, 2011, 48, 32-37.	1.5	39
31	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
32	The GA4GH Phenopacket schema defines a computable representation of clinical data. Nature Biotechnology, 2022, 40, 817-820.	9.4	38
33	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
34	The Facial Evolution: Looking Backward and Moving Forward. Human Mutation, 2013, 34, 14-22.	1.1	36
35	Genotype and phenotype spectrum of NRAS germline variants. European Journal of Human Genetics, 2017, 25, 823-831.	1.4	36
36	A Clinical Review of Generalized Overgrowth Syndromes in the Era of Massively Parallel Sequencing. Molecular Syndromology, 2018, 9, 70-82.	0.3	36

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37	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
38	Carpenter syndrome: extended RAB23 mutation spectrum and analysis of nonsenseâ€mediated mRNA decay. Human Mutation, 2011, 32, E2069-78.	1.1	34
39	The case for open science: rare diseases. JAMIA Open, 2020, 3, 472-486.	1.0	33
40	Fatal Perinatal Mitochondrial Cardiac Failure Caused by Recurrent De Novo Duplications in the ATAD3 Locus. Med, 2021, 2, 49-73.e10.	2.2	33
41	A call for global action for rare diseases in Africa. Nature Genetics, 2020, 52, 21-26.	9.4	31
42	Elucidating the genetic architecture of Adams-Oliver syndrome in a large European cohort. Human Mutation, 2018, 39, 1246-1261.	1.1	31
43	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
44	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
45	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
46	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
47	Phenotyping: Targeting genotype's rich cousin for diagnosis. Journal of Paediatrics and Child Health, 2015, 51, 381-386.	0.4	29
48	Outcomes of an International Workshop on Preconception Expanded Carrier Screening: Some Considerations for Governments. Frontiers in Public Health, 2017, 5, 25.	1.3	28
49	Plain-language medical vocabulary for precision diagnosis. Nature Genetics, 2018, 50, 474-476.	9.4	28
50	Parental smoking impairs vaccine responses inÂchildren with atopic genotypes. Journal of Allergy and Clinical Immunology, 2007, 119, 366-374.	1.5	27
51	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
52	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
53	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
54	Gender-specific effects of cytokine gene polymorphisms on childhood vaccine responses. Vaccine, 2008, 26, 3574-3579.	1.7	25

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55	Barriers and Considerations for Diagnosing Rare Diseases in Indigenous Populations. Frontiers in Pediatrics, 2020, 8, 579924.	0.9	25
56	Initiating an undiagnosed diseases program in the Western Australian public health system. Orphanet Journal of Rare Diseases, 2017, 12, 83.	1.2	24
57	Equitable Expanded Carrier Screening Needs Indigenous Clinical and Population Genomic Data. American Journal of Human Genetics, 2020, 107, 175-182.	2.6	24
58	Cornelia de Lange Syndrome. Advances in Experimental Medicine and Biology, 2010, , 113-123.	0.8	20
59	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
60	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
61	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
62	Healthcare System Priorities for Successful Integration of Genomics: An Australian Focus. Frontiers in Public Health, 2019, 7, 41.	1.3	18
63	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
64	Genotypeâ€“phenotype correlations and novel molecular insights into the DHX30-associated neurodevelopmental disorders. Genome Medicine, 2021, 13, 90.	3.6	16
65	Indigenous Genetics and Rare Diseases: Harmony, Diversity and Equity. Advances in Experimental Medicine and Biology, 2017, 1031, 511-520.	0.8	15
66	Personalised analytics for rare disease diagnostics. Nature Communications, 2019, 10, 5274.	5.8	15
67	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
68	Objective Monitoring of mTOR Inhibitor Therapy by Three-Dimensional Facial Analysis. Twin Research and Human Genetics, 2013, 16, 840-844.	0.3	14
69	Prevalence of microcephaly in an Australian populationâ€based birth defects register, 1980â€“2015. Medical Journal of Australia, 2017, 206, 351-356.	0.8	14
70	A review of structural brain abnormalities in Pallisterâ€Killian syndrome. Molecular Genetics & Genomic Medicine, 2018, 6, 92-98.	0.6	14
71	Enabling Global Clinical Collaborations on Identifiable Patient Data: The Minerva Initiative. Frontiers in Genetics, 2019, 10, 611.	1.1	14
72	3-Dimensional Facial Analysisâ€”Facing Precision Public Health. Frontiers in Public Health, 2017, 5, 31.	1.3	13

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73	Silver Russel syndrome in an aboriginal patient from Australia. American Journal of Medical Genetics, Part A, 2018, 176, 2561-2563.	0.7	13
74	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
75	The need for genetic studies of Indigenous Australians. Medical Journal of Australia, 2012, 196, 313-313.	0.8	12
76	Associations of a novel IL4RA polymorphism, Ala57Thr, in Greenlander Inuit. Journal of Allergy and Clinical Immunology, 2006, 118, 627-634.	1.5	11
77	Epidemiology of Rare Craniofacial Anomalies: Retrospective Western Australian Population Data Linkage Study. Journal of Pediatrics, 2022, 241, 162-172.e9.	0.9	11
78	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
79	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
80	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
81	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
82	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
83	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
84	Association between craniofacial anomalies, intellectual disability and autism spectrum disorder: Western Australian population-based study. Pediatric Research, 2022, 92, 1795-1804.	1.1	8
85	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
86	Monitoring of Therapy for Mucopolysaccharidosis Type I Using Dysmorphometric Facial Phenotypic Signatures. JIMD Reports, 2015, 22, 99-106.	0.7	7
87	Incidental inequity. European Journal of Human Genetics, 2018, 26, 616-617.	1.4	7
88	Birth prevalence of congenital heart defects in Western Australia, 1990â€“2016. Journal of Paediatrics and Child Health, 2021, 57, 1672-1680.	0.4	7
89	Childhood rare diseases and the UN convention on the rights of the child. Orphanet Journal of Rare Diseases, 2021, 16, 523.	1.2	7
90	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

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91	Defining and expanding the phenotype of QARS-associated developmental epileptic encephalopathy. Neurology: Genetics, 2019, 5, e373.	0.9	5
92	Microcephaly in Australian children, 2016â€“2018: national surveillance study. Archives of Disease in Childhood, 2021, 106, 849-854.	1.0	5
93	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
94	A flexible computational pipeline for research analyses of unsolved clinical exome cases. Npj Genomic Medicine, 2020, 5, 54.	1.7	4
95	Culturally competent communication in Indigenous disability assessment: a qualitative study. International Journal for Equity in Health, 2021, 20, 68.	1.5	4
96	Common data elements to standardize genomics studies in cerebral palsy. Developmental Medicine and Child Neurology, 2022, 64, 1470-1476.	1.1	4
97	Translating Aboriginal genomics â€" four letters Closing the Gap. Medical Journal of Australia, 2016, 205, 379-379.	0.8	3
98	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
99	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
100	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
101	Paediatric genomic testing: Navigating genomic reports for the general paediatrician. Journal of Paediatrics and Child Health, 2021, , .	0.4	3
102	Innovation in Informatics to Improve Clinical Care and Drug Accessibility for Rare Diseases in China. Frontiers in Pharmacology, 2021, 12, 719415.	1.6	3
103	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
104	Protecting the rare during a rare pandemic. Medical Journal of Australia, 2020, 213, 94.	0.8	2
105	Further evidence for distinct traits associated with <scp>RBM10</scp> missense variants. Clinical Genetics, 2022, 102, 161-163.	1.0	2
106	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
107	Editorial: Public Health Genomics. Frontiers in Public Health, 2019, 7, 142.	1.3	1
108	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

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109	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
110	Unlocking sociocultural and community factors for the global adoption of genomic medicine. Orphanet Journal of Rare Diseases, 2022, 17, 191.	1.2	1
111	Ethics and equity in rare disease research and healthcare. Personalized Medicine, 2021, 18, 407-416.	0.8	0
112	Digit-all: Rare Diseases. European Medical Journal Innovations, 0, , 11-16.	2.0	0
113	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