David P Dimmock

List of Publications by Year
in descending order

128 papers	9,279 citations	61984 43 h-index	43889 91 g-index
137 all docs	137 docs citations	137 times ranked	14482 citing authors

#	Article	IF	Citations
1	Guidelines for investigating causality of sequence variants in human disease. Nature, 2014, 508, 469-476.	27.8	1,130
2	Making a definitive diagnosis: Successful clinical application of whole exome sequencing in a child with intractable inflammatory bowel disease. Genetics in Medicine, 2011, 13, 255-262.	2.4	651
3	Comprehensive Molecular Characterization of Pheochromocytoma and Paraganglioma. Cancer Cell, 2017, 31, 181-193.	16.8	532
4	Assuring the quality of next-generation sequencing in clinical laboratory practice. Nature Biotechnology, 2012, 30, 1033-1036.	17.5	437
5	Meta-analysis of the diagnostic and clinical utility of genome and exome sequencing and chromosomal microarray in children with suspected genetic diseases. Npj Genomic Medicine, 2018, 3, 16.	3.8	420
6	Rapid whole-genome sequencing decreases infant morbidity and cost of hospitalization. Npj Genomic Medicine, 2018, 3, 10.	3.8	314
7	Diversion of aspartate in ASS1-deficient tumours fosters de novo pyrimidine synthesis. Nature, 2015, 527, 379-383.	27.8	271
8	Oral pharmacological chaperone migalastat compared with enzyme replacement therapy in Fabry disease: 18-month results from the randomised phase III ATTRACT study. Journal of Medical Genetics, 2017, 54, 288-296.	3.2	262
9	A Randomized, Controlled Trial of the Analytic and Diagnostic Performance of Singleton and Trio, Rapid Genome and Exome Sequencing in Ill Infants. American Journal of Human Genetics, 2019, 105, 719-733.	6.2	238
10	Diagnosis of genetic diseases in seriously ill children by rapid whole-genome sequencing and automated phenotyping and interpretation. Science Translational Medicine, 2019, 11, .	12.4	203
11	Wastewater sequencing reveals early cryptic SARS-CoV-2 variant transmission. Nature, 2022, 609, 101-108.	27.8	200
12	Successful immune tolerance induction to enzyme replacement therapy in CRIM-negative infantile Pompe disease. Genetics in Medicine, 2012, 14, 135-142.	2.4	183
13	Urea Cycle Dysregulation Generates Clinically Relevant Genomic and Biochemical Signatures. Cell, 2018, 174, 1559-1570.e22.	28.9	183
14	Patient care standards for primary mitochondrial disease: a consensus statement from the Mitochondrial Medicine Society. Genetics in Medicine, 2017, 19, 1380-1397.	2.4	173
15	The NSIGHT1-randomized controlled trial: rapid whole-genome sequencing for accelerated etiologic diagnosis in critically ill infants. Npj Genomic Medicine, 2018, 3, 6.	3.8	156
16	Exploring concordance and discordance for return of incidental findings from clinical sequencing. Genetics in Medicine, 2012, 14, 405-410.	2.4	149
17	Clinical and molecular features of mitochondrial DNA depletion due to mutations in deoxyguanosine kinase. Human Mutation, 2008, 29, 330-331.	2.5	144
18	Project Baby Bear: Rapid precision care incorporating rWGS in 5 California childrenâ€™s hospitals demonstrates improved clinical outcomes and reduced costs of care. American Journal of Human Genetics, 2021, 108, 1231-1238.	6.2	140

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19	Good laboratory practice for clinical next-generation sequencing informatics pipelines. Nature Biotechnology, 2015, 33, 689-693.	17.5	134
20	Allogeneic hematopoietic cell transplantation for XIAP deficiency: an international survey reveals poor outcomes. Blood, 2013, 121, 877-883.	1.4	132
21	Whole exome and whole genome sequencing. Current Opinion in Pediatrics, 2011, 23, 594-600.	2.0	124
22	Pegvaliase for the treatment of phenylketonuria: Results of a long-term phase 3 clinical trial program (PRISM). Molecular Genetics and Metabolism, 2018, 124, 27-38.	1.1	123
23	Quantitative Evaluation of the Mitochondrial DNA Depletion Syndrome. Clinical Chemistry, 2010, 56, 1119-1127.	3.2	119
24	An RCT of Rapid Genomic Sequencing among Seriously Ill Infants Results in High Clinical Utility, Changes in Management, and Low Perceived Harm. American Journal of Human Genetics, 2020, 107, 942-952.	6.2	110
25	Rapid Whole Genome Sequencing Has Clinical Utility in Children in the PICU*. Pediatric Critical Care Medicine, 2019, 20, 1007-1020.	0.5	105
26	Realâ€Time Quantitative PCR Analysis of Mitochondrial DNA Content. Current Protocols in Human Genetics, 2011, 68, Unit 19.7..	3.5	90
27	Genomics in Clinical Practice: Lessons from the Front Lines. Science Translational Medicine, 2013, 5, 194cm5.	12.4	90
28	De Novo Pathogenic Variants in CACNA1E Cause Developmental and Epileptic Encephalopathy with Contractures, Macrocephaly, and Dyskinesias. American Journal of Human Genetics, 2018, 103, 666-678.	6.2	87
29	Citrin Deficiency: A Novel Cause of Failure to Thrive That Responds to a High-Protein, Low-Carbohydrate Diet. Pediatrics, 2007, 119, e773-e777.	2.1	81
30	Citrin deficiency, a perplexing global disorder. Molecular Genetics and Metabolism, 2009, 96, 44-49.	1.1	81
31	Utility of Oligonucleotide Arrayâ€"Based Comparative Genomic Hybridization for Detection of Target Gene Deletions. Clinical Chemistry, 2008, 54, 1141-1148.	3.2	78
32	Mitochondrial Disease Sequence Data Resource (MSeqDR): A global grass-roots consortium to facilitate deposition, curation, annotation, and integrated analysis of genomic data for the mitochondrial disease clinical and research communities. Molecular Genetics and Metabolism, 2015, 114, 388-396.	1.1	76
33	A timely arrival for genomic medicine. Genetics in Medicine, 2011, 13, 195-196.	2.4	75
34	Perspectives of clinical genetics professionals toward genome sequencing and incidental findings: a survey study. Clinical Genetics, 2013, 84, 230-236.	2.0	73
35	SIGIRR Genetic Variants in Premature Infants With Necrotizing Enterocolitis. Pediatrics, 2015, 135, e1530-e1534.	2.1	71
36	Abnormal neurological features predict poor survival and should preclude liver transplantation in patients with deoxyguanosine kinase deficiency. Liver Transplantation, 2008, 14, 1480-1485.	2.4	67

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37	Best practices for the analytical validation of clinical whole-genome sequencing intended for the diagnosis of germline disease. Npj Genomic Medicine, 2020, 5, 47.	3.8	67
38	Whole-exome sequencing supports genetic heterogeneity in childhood apraxia of speech. Journal of Neurodevelopmental Disorders, 2013, 5, 29.	3.1	65
39	A Prospective Study of Parental Perceptions of Rapid Whole-Genome and -Exome Sequencing among Seriously Ill Infants. American Journal of Human Genetics, 2020, 107, 953-962.	6.2	65
40	Evidence- and consensus-based recommendations for the use of pegvaliase in adults with phenylketonuria. Genetics in Medicine, 2019, 21, 1851-1867.	2.4	56
41	Solid organ transplantation in primary mitochondrial disease: Proceed with caution. Molecular Genetics and Metabolism, 2016, 118, 178-184.	1.1	55
42	Successful Application of Whole Genome Sequencing in a Medical Genetics Clinic. Journal of Pediatric Genetics, 2017, 06, 061-076.	0.7	54
43	Antioxidant response genes sequence variants and BPD susceptibility in VLBW infants. Pediatric Research, 2015, 77, 477-483.	2.3	52
44	Rapid Sequencing-Based Diagnosis of Thiamine Metabolism Dysfunction Syndrome. New England Journal of Medicine, 2021, 384, 2159-2161.	27.0	48
45	Nextâ€generation Sequencing Facilitates the Diagnosis in a Child With Twinkle Mutations Causing Cholestatic Liver Failure. Journal of Pediatric Gastroenterology and Nutrition, 2012, 54, 291-294.	1.8	42
46	Acute liver failure in neonates with undiagnosed hereditary fructose intolerance due to exposure from widely available infant formulas. Molecular Genetics and Metabolism, 2018, 123, 428-432.	1.1	40
47	The Medical Genome Initiative: moving whole-genome sequencing for rare disease diagnosis to the clinic. Genome Medicine, 2020, 12, 48.	8.2	40
48	Novel Variant Findings and Challenges Associated With the Clinical Integration of Genomic Testing. JAMA Pediatrics, 2021, 175, e205906.	6.2	39
49	Practice patterns of mitochondrial disease physicians in North America. Part 2: treatment, care and management. Mitochondrion, 2013, 13, 681-687.	3.4	38
50	Functional Precision Medicine Identifies New Therapeutic Candidates for Medulloblastoma. Cancer Research, 2020, 80, 5393-5407.	0.9	38
51	Clinical utility of genomic sequencing: a measurement toolkit. Npj Genomic Medicine, 2020, 5, 56.	3.8	37
52	Pathogenic variants in <scp>SQOR</scp> encoding sulfide:quinone oxidoreductase are a potentially treatable cause of Leigh disease. Journal of Inherited Metabolic Disease, 2020, 43, 1024-1036.	3.6	37
53	Practice patterns of mitochondrial disease physicians in North America. Part 1: Diagnostic and clinical challenges. Mitochondrion, 2014, 14, 26-33.	3.4	36
54	Efficient Precision Genome Editing in iPSCs via Genetic Co-targeting withÂSelection. Stem Cell Reports, 2017, 8, 491-499.	4.8	36

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55	A Screen Using iPSC-Derived Hepatocytes Reveals NAD+ as a Potential Treatment for mtDNA Depletion Syndrome. Cell Reports, 2018, 25, 1469-1484.e5.	6.4	36
56	The humanistic burden of Pompe disease: are there still unmet needs? A systematic review. BMC Neurology, 2017, 17, 202.	1.8	31
57	Implementing Rapid Whole-Genome Sequencing in Critical Care: A Qualitative Study of Facilitators and Barriers to New Technology Adoption. Journal of Pediatrics, 2021, 237, 237-243.e2.	1.8	31
58	An online compendium of treatable genetic disorders. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2021, 187, 48-54.	1.6	31
59	Simultaneous detection of mitochondrial DNA depletion and single-exon deletion in the deoxyguanosine gene using array-based comparative genomic hybridisation. Archives of Disease in Childhood, 2009, 94, 55-58.	1.9	29
60	Feasibility of adjunct therapeutic hypothermia treatment for hyperammonemia and encephalopathy due to urea cycle disorders and organic acidemias. Molecular Genetics and Metabolism, 2013, 109, 354-359.	1.1	29
61	The case for early use of rapid whole-genome sequencing in management of critically ill infants: late diagnosis of Coffinâ€“Siris syndrome in an infant with left congenital diaphragmatic hernia, congenital heart disease, and recurrent infections. Journal of Physical Education and Sports Management, 2018, 4, a002469.	1.2	29
62	Measurement of genetic diseases as a cause of mortality in infants receiving whole genome sequencing. Npj Genomic Medicine, 2020, 5, 49.	3.8	29
63	Biallelic mutations in valyl-tRNA synthetase gene VARS are associated with a progressive neurodevelopmental epileptic encephalopathy. Nature Communications, 2019, 10, 707.	12.8	28
64	Rapid whole genome sequencing impacts care and resource utilization in infants with congenital heart disease. Npj Genomic Medicine, 2021, 6, 29.	3.8	27
65	Choices of incidental findings of individuals undergoing genome wide sequencing, a single center's experience. Clinical Genetics, 2017, 91, 137-140.	2.0	26
66	The role of molecular testing and enzyme analysis in the management of hypomorphic citrullinemia. American Journal of Medical Genetics, Part A, 2008, 146A, 2885-2890.	1.2	24
67	Implementation, adoption, and utility of family health history risk assessment in diverse care settings: evaluating implementation processes and impact with an implementation framework. Genetics in Medicine, 2019, 21, 331-338.	2.4	24
68	Recessive deoxyguanosine kinase deficiency causes juvenile onset mitochondrial myopathy. Molecular Genetics and Metabolism, 2012, 107, 92-94.	1.1	22
69	Protocol for the â€œImplementation, adoption, and utility of family history in diverse care settingsâ€• study. Implementation Science, 2015, 10, 163.	6.9	19
70	Reduced Mitochondrial DNA Content and Heterozygous Nuclear Gene Mutations in Patients With Acute Liver Failure. Journal of Pediatric Gastroenterology and Nutrition, 2013, 57, 438-443.	1.8	18
71	Long-term developmental progression in infants and young children taking sapropterin for phenylketonuria: a two-year analysis of safety and efficacy. Genetics in Medicine, 2015, 17, 365-373.	2.4	18
72	221 newborn-screened neonates with medium-chain acyl-coenzyme A dehydrogenase deficiency: Findings from the Inborn Errors of Metabolism Collaborative. Molecular Genetics and Metabolism, 2016, 119, 75-82.	1.1	18

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73	Metagenomic Next-Generation Sequencing for Pathogen Detection and Transcriptomic Analysis in Pediatric Central Nervous System Infections. Open Forum Infectious Diseases, 2021, 8, ofab104.	0.9	18
74	Cost Efficacy of Rapid Whole Genome Sequencing in the Pediatric Intensive Care Unit. Frontiers in Pediatrics, 2021, 9, 809536.	1.9	18
75	Progressive myofiber loss with extensive fibro-fatty replacement in a child with mitochondrial DNA depletion syndrome and novel thymidine kinase 2 gene mutations. Neuromuscular Disorders, 2009, 19, 784-787.	0.6	17
76	Presentation and Diagnostic Evaluation of Mitochondrial Disease. Pediatric Clinics of North America, 2017, 64, 161-171.	1.8	17
77	Use of Metagenomic Next-Generation Sequencing to Identify Pathogens in Pediatric Osteoarticular Infections. Open Forum Infectious Diseases, 2021, 8, ofab346.	0.9	17
78	Correction of Hyperbilirubinemia in Gunn Rats Using Clinically Relevant Low Doses of Helper-Dependent Adenoviral Vectors. Human Gene Therapy, 2011, 22, 483-488.	2.7	16
79	Cystic fibrosis mutations for p.F508del compound heterozygotes predict sweat chloride levels and pancreatic sufficiency. Clinical Genetics, 2012, 82, 546-551.	2.0	16
80	Potentially diagnostic electron paramagnetic resonance spectra elucidate the underlying mechanism of mitochondrial dysfunction in the deoxyguanosine kinase deficient rat model of a genetic mitochondrial DNA depletion syndrome. Free Radical Biology and Medicine, 2016, 92, 141-151.	2.9	16
81	The mitochondrial carrier Citrin plays a role in regulating cellular energy during carcinogenesis. Oncogene, 2020, 39, 164-175.	5.9	16
82	Mortality in a neonate with molybdenum cofactor deficiency illustrates the need for a comprehensive rapid precision medicine system. Journal of Physical Education and Sports Management, 2020, 6, a004705.	1.2	16
83	Rapid whole-genome sequencing identifies a novel homozygous NPC1 variant associated with Niemannâ€“Pick type C1 disease in a 7-week-old male with cholestasis. Journal of Physical Education and Sports Management, 2017, 3, a001966.	1.2	15
84	Common data elements for clinical research in mitochondrial disease: a National Institute for Neurological Disorders and Stroke project. Journal of Inherited Metabolic Disease, 2017, 40, 403-414.	3.6	15
85	Rapid Diagnosis of KCNQ2-Associated Early Infantile Epileptic Encephalopathy Improved Outcome. Pediatric Neurology, 2018, 86, 69-70.	2.1	15
86	A personal perspective on returning secondary results of clinical genome sequencing. Genome Medicine, 2012, 4, 54.	8.2	14
87	A novel c.592-4_c.592-3delTT mutation in DGUOK gene causes exon skipping. Mitochondrion, 2010, 10, 188-191.	3.4	13
88	Should states adopt newborn screening for early infantile Krabbe disease?. Genetics in Medicine, 2016, 18, 217-220.	2.4	13
89	At the intersection of precision medicine and population health: an implementation-effectiveness study of family health history based systematic risk assessment in primary care. BMC Health Services Research, 2020, 20, 1015.	2.2	13
90	De Novo Mutations in POLG Presenting with Acute Liver Failure or Encephalopathy. Journal of Pediatric Gastroenterology and Nutrition, 2009, 49, 126-129.	1.8	11

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91	Rapid whole-genome sequencing identifies a novel AIRE variant associated with autoimmune polyendocrine syndrome type 1. Journal of Physical Education and Sports Management, 2018, 4, a002485.	1.2	11
92	Moving Genomics to Routine Care. Circulation Genomic and Precision Medicine, 2020, 13, 406-416.	3.6	11
93	Expanding the phenotypic spectrum of BCS1Lâ€related mitochondrial disease. Annals of Clinical and Translational Neurology, 2021, 8, 2155-2165.	3.7	11
94	Novel Factor XIII variant identified through whole-genome sequencing in a child with intracranial hemorrhage. Journal of Physical Education and Sports Management, 2018, 4, a003525.	1.2	10
95	Sdha+/- Rats Display Minimal Muscle Pathology Without Significant Behavioral or Biochemical Abnormalities. Journal of Neuropathology and Experimental Neurology, 2018, 77, 665-672.	1.7	10
96	Quantitative analysis of the natural history of prolidase deficiency: description of 17 families and systematic review of published cases. Genetics in Medicine, 2021, 23, 1604-1615.	2.4	10
97	Postmortem diagnosis of PPA2-associated sudden cardiac death from dried blood spot in a neonate presenting with vocal cord paralysis. Journal of Physical Education and Sports Management, 2020, 6, a005611.	1.2	10
98	Screening an Asymptomatic Person for Genetic Risk. New England Journal of Medicine, 2014, 370, 2442-2445.	27.0	9
99	Results and Lessons of a Pilot Study of Cascade Screening for Familial Hypercholesterolemia in US Primary Care Practices. Journal of General Internal Medicine, 2020, 35, 351-353.	2.6	9
100	Effect of Sociodemographic Factors on Uptake of a Patient-Facing Information Technology Family Health History Risk Assessment Platform. Applied Clinical Informatics, 2019, 10, 180-188.	1.7	8
101	Whole Genome Sequencing: A Considered Approach to Clinical Implementation. Current Protocols in Human Genetics, 2013, 77, Unit9.22.	3.5	7
102	Should we implement population screening for fragile X?. Genetics in Medicine, 2017, 19, 1295-1299.	2.4	7
103	Diagnosis of cytomegalovirus infection from clinical whole genome sequencing. Scientific Reports, 2020, 10, 11020.	3.3	6
104	Abnormal SCID Newborn Screening and Spontaneous Recovery Associated with a Novel Haploinsufficiency IKZF1 Mutation. Journal of Clinical Immunology, 2021, 41, 1241-1249.	3.8	6
105	In the Absence of Evidentiary Harm, Existing Societal Norms Regarding Parental Authority Should Prevail. American Journal of Bioethics, 2014, 14, 24-26.	0.9	5
106	Ethical Issues in DNA Sequencing in the Neonate. Clinics in Perinatology, 2014, 41, 993-1000.	2.1	5
107	A novel FOXF1 mutation associated with alveolar capillary dysplasia and coexisting colobomas and hemihyperplasia. Journal of Perinatology, 2015, 35, 155-157.	2.0	5
108	The nucleotide prodrug CERC â€913 improves mtDNA content in primary hepatocytes from DGUOKâ€deficient rats. Journal of Inherited Metabolic Disease, 2021, 44, 492-501.	3.6	5

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109	Rapid whole-genome sequencing in critically Ill children: shifting from unease to evidence, education, and equitable implementation. Journal of Pediatrics, 2021, 238, 343.	1.8	5
110	Necrotizing Enterocolitis Is Not Associated With Sequence Variants in Antioxidant Response Genes in Premature Infants. Journal of Pediatric Gastroenterology and Nutrition, 2016, 62, 420-423.	1.8	4
111	Diagnosis and treatment of a boy with IPEX syndrome presenting with diabetes in early infancy. Clinical Case Reports (discontinued), 2019, 7, 2123-2127.	0.5	4
112	Healthcare Professionalsâ€™ Attitudes toward Rapid Whole Genome Sequencing in Pediatric Acute Care. Children, 2022, 9, 357.	1.5	4
113	Clinical diagnostic whole genome sequencing in a paediatric population: experience from our WGS genetics clinic. BMC Proceedings, 2012, 6, .	1.6	3
114	Metagenomic sequencing and evaluation of the host response in the pediatric aerodigestive population. Pediatric Pulmonology, 2021, 56, 516-524.	2.0	3
115	Ending a diagnostic odyssey: Moving from exome to genome to identify cockayne syndrome. Molecular Genetics & Genomic Medicine, 2021, 9, e1623.	1.2	3
116	683. Bronchoscope-Guided, Targeted Lobar Aersolization of HDAd into the Lungs of Nonhuman Primate Results in Exceedingly High Pulmonary Transduction Uniformally throughout the Entire Lung with Negligible Toxicity. Molecular Therapy, 2006, 13, S264.	8.2	2
117	Hypertrophic Cardiomyopathy: A New Mutation Illustrates the Need for Family-Centered Care. Pediatric Cardiology, 2014, 35, 1474-1477.	1.3	2
118	Retrospective identification of prenatal fetal anomalies associated with diagnostic neonatal genomic sequencing results. Prenatal Diagnosis, 2022, 42, 705-716.	2.3	2
119	Better and faster is cheaper. Human Mutation, 2022, 43, 1495-1506.	2.5	2
120	Expanding the phenotypic and molecular spectrum of NFS1 â€related disorders that cause functional deficiencies in mitochondrial and cytosolic ironâ€“sulfur cluster containing enzymes. Human Mutation, 2022, 43, 305-315.	2.5	1
121	Novel human pathological mutations. Gene symbol: ASS1. Disease: Citrullinaemia. Human Genetics, 2009, 126, 341.	3.8	1
122	213. Improving the Therapeutic Index of Helper-Dependent Adenoviral Vector for Crigler-Najjar Gene Therapy. Molecular Therapy, 2006, 13, S82.	8.2	0
123	A Novel XIAP Mutation Detected by Genome Wide Sequencing Causes Early Onset Inflammatory Bowel Disease. Clinical Immunology, 2010, 135, S105.	3.2	0
124	373. Critical Care Medicine, 2019, 47, 168.	0.9	0
125	Citrin Deficiency. , 2016, , .		0
126	Response to Metcalfe et al.. Genetics in Medicine, 2018, 20, 1093-1093.	2.4	0

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127	Novel human pathological mutations. Gene symbol: ASS1. Disease: Citrullinaemia. Human Genetics, 2009, 126, 342.	3.8	0
128	eP286: Genome-to-treatment: A system to guide the acute management of genetic disorders in children. Genetics in Medicine, 2022, 24, S181.	2.4	0