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	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
2	Retrospective identification of prenatal fetal anomalies associated with diagnostic neonatal genomic sequencing results. Prenatal Diagnosis, 2022, 42, 705-716.	2.3	2
3	Healthcare Professionalsâ€™ Attitudes toward Rapid Whole Genome Sequencing in Pediatric Acute Care. Children, 2022, 9, 357.	1.5	4
4	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
5	Better and faster is cheaper. Human Mutation, 2022, 43, 1495-1506.	2.5	2
6	Wastewater sequencing reveals early cryptic SARS-CoV-2 variant transmission. Nature, 2022, 609, 101-108.	27.8	200
7	Metagenomic sequencing and evaluation of the host response in the pediatric aerodigestive population. Pediatric Pulmonology, 2021, 56, 516-524.	2.0	3
8	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
9	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
10	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
11	Rapid whole genome sequencing impacts care and resource utilization in infants with congenital heart disease. Npj Genomic Medicine, 2021, 6, 29.	3.8	27
12	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
13	Novel Variant Findings and Challenges Associated With the Clinical Integration of Genomic Testing. JAMA Pediatrics, 2021, 175, e205906.	6.2	39
14	Ending a diagnostic odyssey: Moving from exome to genome to identify cockayne syndrome. Molecular Genetics & Genomic Medicine, 2021, 9, e1623.	1.2	3
15	Rapid Sequencing-Based Diagnosis of Thiamine Metabolism Dysfunction Syndrome. New England Journal of Medicine, 2021, 384, 2159-2161.	27.0	48
16	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
17	Use of Metagenomic Next-Generation Sequencing to Identify Pathogens in Pediatric Osteoarticular Infections. Open Forum Infectious Diseases, 2021, 8, ofab346.	0.9	17
18	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

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19	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
20	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
21	Expanding the phenotypic spectrum of BCS1Lâ€related mitochondrial disease. Annals of Clinical and Translational Neurology, 2021, 8, 2155-2165.	3.7	11
22	Cost Efficacy of Rapid Whole Genome Sequencing in the Pediatric Intensive Care Unit. Frontiers in Pediatrics, 2021, 9, 809536.	1.9	18
23	The mitochondrial carrier Citrin plays a role in regulating cellular energy during carcinogenesis. Oncogene, 2020, 39, 164-175.	5.9	16
24	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
25	Functional Precision Medicine Identifies New Therapeutic Candidates for Medulloblastoma. Cancer Research, 2020, 80, 5393-5407.	0.9	38
26	Measurement of genetic diseases as a cause of mortality in infants receiving whole genome sequencing. Npj Genomic Medicine, 2020, 5, 49.	3.8	29
27	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
28	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
29	Moving Genomics to Routine Care. Circulation Genomic and Precision Medicine, 2020, 13, 406-416.	3.6	11
30	Clinical utility of genomic sequencing: a measurement toolkit. Npj Genomic Medicine, 2020, 5, 56.	3.8	37
31	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
32	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
33	The Medical Genome Initiative: moving whole-genome sequencing for rare disease diagnosis to the clinic. Genome Medicine, 2020, 12, 48.	8.2	40
34	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
35	Diagnosis of cytomegalovirus infection from clinical whole genome sequencing. Scientific Reports, 2020, 10, 11020.	3.3	6
36	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

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37	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
38	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
39	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
40	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
41	Biallelic mutations in valyl-tRNA synthetase gene VARS are associated with a progressive neurodevelopmental epileptic encephalopathy. Nature Communications, 2019, 10, 707.	12.8	28
42	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
43	373. Critical Care Medicine, 2019, 47, 168.	0.9	0
44	Rapid Whole Genome Sequencing Has Clinical Utility in Children in the PICU*. Pediatric Critical Care Medicine, 2019, 20, 1007-1020.	0.5	105
45	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
46	Evidence- and consensus-based recommendations for the use of pegvaliase in adults with phenylketonuria. Genetics in Medicine, 2019, 21, 1851-1867.	2.4	56
47	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
48	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
49	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
50	Rapid whole-genome sequencing decreases infant morbidity and cost of hospitalization. Npj Genomic Medicine, 2018, 3, 10.	3.8	314
51	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
52	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
53	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
54	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

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55	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
56	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
57	Rapid Diagnosis of KCNQ2-Associated Early Infantile Epileptic Encephalopathy Improved Outcome. Pediatric Neurology, 2018, 86, 69-70.	2.1	15
58	Urea Cycle Dysregulation Generates Clinically Relevant Genomic and Biochemical Signatures. Cell, 2018, 174, 1559-1570.e22.	28.9	183
59	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
60	Response to Metcalfe et al.. Genetics in Medicine, 2018, 20, 1093-1093.	2.4	0
61	Comprehensive Molecular Characterization of Pheochromocytoma and Paraganglioma. Cancer Cell, 2017, 31, 181-193.	16.8	532
62	Efficient Precision Genome Editing in iPSCs via Genetic Co-targeting withÂSelection. Stem Cell Reports, 2017, 8, 491-499.	4.8	36
63	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
64	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
65	Successful Application of Whole Genome Sequencing in a Medical Genetics Clinic. Journal of Pediatric Genetics, 2017, 06, 061-076.	0.7	54
66	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
67	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
68	Should we implement population screening for fragile X?. Genetics in Medicine, 2017, 19, 1295-1299.	2.4	7
69	Choices of incidental findings of individuals undergoing genome wide sequencing, a single center's experience. Clinical Genetics, 2017, 91, 137-140.	2.0	26
70	Presentation and Diagnostic Evaluation of Mitochondrial Disease. Pediatric Clinics of North America, 2017, 64, 161-171.	1.8	17
71	The humanistic burden of Pompe disease: are there still unmet needs? A systematic review. BMC Neurology, 2017, 17, 202.	1.8	31
72	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

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73	Should states adopt newborn screening for early infantile Krabbe disease?. Genetics in Medicine, 2016, 18, 217-220.	2.4	13
74	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
75	Solid organ transplantation in primary mitochondrial disease: Proceed with caution. Molecular Genetics and Metabolism, 2016, 118, 178-184.	1.1	55
76	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
77	Citrin Deficiency. , 2016, , .		0
78	Protocol for the â€œImplementation, adoption, and utility of family history in diverse care settingsâ€• study. Implementation Science, 2015, 10, 163.	6.9	19
79	SIGIRR Genetic Variants in Premature Infants With Necrotizing Enterocolitis. Pediatrics, 2015, 135, e1530-e1534.	2.1	71
80	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
81	Good laboratory practice for clinical next-generation sequencing informatics pipelines. Nature Biotechnology, 2015, 33, 689-693.	17.5	134
82	A novel FOXF1 mutation associated with alveolar capillary dysplasia and coexisting colobomas and hemihyperplasia. Journal of Perinatology, 2015, 35, 155-157.	2.0	5
83	Antioxidant response genes sequence variants and BPD susceptibility in VLBW infants. Pediatric Research, 2015, 77, 477-483.	2.3	52
84	Diversion of aspartate in ASS1-deficient tumours fosters de novo pyrimidine synthesis. Nature, 2015, 527, 379-383.	27.8	271
85	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
86	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
87	Ethical Issues in DNA Sequencing in the Neonate. Clinics in Perinatology, 2014, 41, 993-1000.	2.1	5
88	Screening an Asymptomatic Person for Genetic Risk. New England Journal of Medicine, 2014, 370, 2442-2445.	27.0	9
89	Practice patterns of mitochondrial disease physicians in North America. Part 1: Diagnostic and clinical challenges. Mitochondrion, 2014, 14, 26-33.	3.4	36
90	Guidelines for investigating causality of sequence variants in human disease. Nature, 2014, 508, 469-476.	27.8	1,130

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91	Hypertrophic Cardiomyopathy: A New Mutation Illustrates the Need for Family-Centered Care. Pediatric Cardiology, 2014, 35, 1474-1477.	1.3	2
92	Perspectives of clinical genetics professionals toward genome sequencing and incidental findings: a survey study. Clinical Genetics, 2013, 84, 230-236.	2.0	73
93	Whole-exome sequencing supports genetic heterogeneity in childhood apraxia of speech. Journal of Neurodevelopmental Disorders, 2013, 5, 29.	3.1	65
94	Practice patterns of mitochondrial disease physicians in North America. Part 2: treatment, care and management. Mitochondrion, 2013, 13, 681-687.	3.4	38
95	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
96	Whole Genome Sequencing: A Considered Approach to Clinical Implementation. Current Protocols in Human Genetics, 2013, 77, Unit9.22.	3.5	7
97	Genomics in Clinical Practice: Lessons from the Front Lines. Science Translational Medicine, 2013, 5, 194cm5.	12.4	90
98	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
99	Allogeneic hematopoietic cell transplantation for XIAP deficiency: an international survey reveals poor outcomes. Blood, 2013, 121, 877-883.	1.4	132
100	Successful immune tolerance induction to enzyme replacement therapy in CRIM-negative infantile Pompe disease. Genetics in Medicine, 2012, 14, 135-142.	2.4	183
101	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
102	Recessive deoxyguanosine kinase deficiency causes juvenile onset mitochondrial myopathy. Molecular Genetics and Metabolism, 2012, 107, 92-94.	1.1	22
103	A personal perspective on returning secondary results of clinical genome sequencing. Genome Medicine, 2012, 4, 54.	8.2	14
104	Exploring concordance and discordance for return of incidental findings from clinical sequencing. Genetics in Medicine, 2012, 14, 405-410.	2.4	149
105	Cystic fibrosis mutations for p.F508del compound heterozygotes predict sweat chloride levels and pancreatic sufficiency. Clinical Genetics, 2012, 82, 546-551.	2.0	16
106	Assuring the quality of next-generation sequencing in clinical laboratory practice. Nature Biotechnology, 2012, 30, 1033-1036.	17.5	437
107	Clinical diagnostic whole genome sequencing in a paediatric population: experience from our WGS genetics clinic. BMC Proceedings, 2012, 6, .	1.6	3
108	Realâ€Time Quantitative PCR Analysis of Mitochondrial DNA Content. Current Protocols in Human Genetics, 2011, 68, Unit 19.7..	3.5	90

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109	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
110	A timely arrival for genomic medicine. Genetics in Medicine, 2011, 13, 195-196.	2.4	75
111	Whole exome and whole genome sequencing. Current Opinion in Pediatrics, 2011, 23, 594-600.	2.0	124
112	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
113	A Novel XIAP Mutation Detected by Genome Wide Sequencing Causes Early Onset Inflammatory Bowel Disease. Clinical Immunology, 2010, 135, S105.	3.2	0
114	A novel c.592-4_c.592-3delTT mutation in DGUOK gene causes exon skipping. Mitochondrion, 2010, 10, 188-191.	3.4	13
115	Quantitative Evaluation of the Mitochondrial DNA Depletion Syndrome. Clinical Chemistry, 2010, 56, 1119-1127.	3.2	119
116	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
117	Citrin deficiency, a perplexing global disorder. Molecular Genetics and Metabolism, 2009, 96, 44-49.	1.1	81
118	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
119	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
120	Novel human pathological mutations. Gene symbol: ASS1. Disease: Citrullinaemia. Human Genetics, 2009, 126, 341.	3.8	1
121	Novel human pathological mutations. Gene symbol: ASS1. Disease: Citrullinaemia. Human Genetics, 2009, 126, 342.	3.8	0
122	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
123	Clinical and molecular features of mitochondrial DNA depletion due to mutations in deoxyguanosine kinase. Human Mutation, 2008, 29, 330-331.	2.5	144
124	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
125	Utility of Oligonucleotide Arrayâ€"Based Comparative Genomic Hybridization for Detection of Target Gene Deletions. Clinical Chemistry, 2008, 54, 1141-1148.	3.2	78
126	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

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127	213. Improving the Therapeutic Index of Helper-Dependent Adenoviral Vector for Crigler-Najjar Gene Therapy. Molecular Therapy, 2006, 13, S82.	8.2	0
128	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