Hilary J Vernon

List of Publications by Year
in descending order

56 papers	1,868 citations	279798 23 h-index	289244 40 g-index
60 all docs	60 docs citations	60 times ranked	3944 citing authors

#	Article	IF	Citations
1	Clinical whole exome sequencing in child neurology practice. Annals of Neurology, 2014, 76, 473-483.	5.3	228
2	Reanalysis of Clinical Exome Sequencing Data. New England Journal of Medicine, 2019, 380, 2478-2480.	27.0	205
3	Cardiolipin, Mitochondria, and Neurological Disease. Trends in Endocrinology and Metabolism, 2021, 32, 224-237.	7.1	113
4	A ketogenic diet rescues hippocampal memory defects in a mouse model of Kabuki syndrome. Proceedings of the National Academy of Sciences of the United States of America, 2017, 114, 125-130.	7.1	102
5	Inborn Errors of Metabolism. JAMA Pediatrics, 2015, 169, 778.	6.2	99
6	Unlocking the Secrets of Mitochondria in the Cardiovascular System. Circulation, 2019, 140, 1205-1216.	1.6	91
7	A phase 2/3 randomized clinical trial followed by an open-label extension to evaluate the effectiveness of elamipretide in Barth syndrome, a genetic disorder of mitochondrial cardiolipin metabolism. Genetics in Medicine, 2021, 23, 471-478.	2.4	59
8	De novo POGZ mutations are associated with neurodevelopmental disorders and microcephaly. Journal of Physical Education and Sports Management, 2015, 1, a000455.	1.2	51
9	FGF21 underlies a hormetic response to metabolic stress in methylmalonic acidemia. JCI Insight, 2018, 3, .	5.0	50
10	Missense variants in the chromatin remodeler CHD1 are associated with neurodevelopmental disability. Journal of Medical Genetics, 2018, 55, 561-566.	3.2	49
11	Hypoxia tolerance in the Norrin-deficient retina and the chronically hypoxic brain studied at single-cell resolution. Proceedings of the National Academy of Sciences of the United States of America, 2019, 116, 9103-9114.	7.1	44
12	WACloss-of-function mutations cause a recognisable syndrome characterised by dysmorphic features, developmental delay and hypotonia and recapitulate 10p11.23 microdeletion syndrome. Journal of Medical Genetics, 2015, 52, 754-761.	3.2	41
13	A detailed analysis of methylmalonic acid kinetics during hemodialysis and after combined liver/kidney transplantation in a patient with mut⁰ methylmalonic acidemia. Journal of Inherited Metabolic Disease, 2014, 37, 899-907.	3.6	40
14	Neuroimaging Findings of Organic Acidemias and Aminoacidopathies. Radiographics, 2018, 38, 912-931.	3.3	40
15	Noninvasive monitoring of chronic kidney disease using pH and perfusion imaging. Science Advances, 2019, 5, eaaw8357.	10.3	38
16	De Novo Variants in CNOT1, a Central Component of the CCR4-NOT Complex Involved in Gene Expression and RNA and Protein Stability, Cause Neurodevelopmental Delay. American Journal of Human Genetics, 2020, 107, 164-172.	6.2	37
17	Introduction of sapropterin dihydrochloride as standard of care in patients with phenylketonuria. Molecular Genetics and Metabolism, 2010, 100, 229-233.	1.1	35
18	Phenotypic expansion of POGZâ€related intellectual disability syndrome (Whiteâ€Sutton) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50 6	1.2	35

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19	Clinical laboratory studies in Barth Syndrome. Molecular Genetics and Metabolism, 2014, 112, 143-147.	1.1	34
20	Mutations in FARS2 and nonâ€fatal mitochondrial dysfunction in two siblings. American Journal of Medical Genetics, Part A, 2015, 167, 1147-1151.	1.2	33
21	New targets for monitoring and therapy in Barth syndrome. Genetics in Medicine, 2016, 18, 1001-1010.	2.4	32
22	Deoxysphingolipid precursors indicate abnormal sphingolipid metabolism in individuals with primary and secondary disturbances of serine availability. Molecular Genetics and Metabolism, 2018, 124, 204-209.	1.1	31
23	Metabolomics Reveals New Mechanisms for Pathogenesis in Barth Syndrome and Introduces Novel Roles for Cardiolipin in Cellular Function. PLoS ONE, 2016, 11, e0151802.	2.5	31
24	Functional exercise capacity, strength, balance and motion reaction time in Barth syndrome. Orphanet Journal of Rare Diseases, 2019, 14, 37.	2.7	24
25	Nutritional Interventions for Mitochondrial OXPHOS Deficiencies: Mechanisms and Model Systems. Annual Review of Pathology: Mechanisms of Disease, 2018, 13, 163-191.	22.4	22
26	GATAD2B-associatedneurodevelopmental disorder (GAND): clinical and molecular insights into a NuRD-relateddisorder. Genetics in Medicine, 2020, 22, 878-888.	2.4	22
27	Aprt/Opn double knockout mice: Osteopontin is a modifier of kidney stone disease severity. Kidney International, 2005, 68, 938-947.	5.2	21
28	6p25 microdeletion: White matter abnormalities in an adult patient. American Journal of Medical Genetics, Part A, 2013, 161, 1686-1689.	1.2	21
29	Clinical presentation and natural history of Barth Syndrome: An overview. Journal of Inherited Metabolic Disease, 2022, 45, 7-16.	3.6	21
30	In Vitro Models to Study the Blood Brain Barrier. Methods in Molecular Biology, 2011, 758, 153-168.	0.9	18
31	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
32	Multi-omics studies in cellular models of methylmalonic acidemia and propionic acidemia reveal dysregulation of serine metabolism. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2019, 1865, 165538.	3.8	17
33	Identification and Functional Studies of Regulatory Variants Responsible for the Association of <i>NRG3</i> with a Delusion Phenotype in Schizophrenia. Molecular Neuropsychiatry, 2015, 1, 36-46.	2.9	14
34	Current and future treatment approaches for Barth syndrome. Journal of Inherited Metabolic Disease, 2022, 45, 17-28.	3.6	14
35	Designing clinical trials for rare diseases: unique challenges and opportunities. Nature Reviews Methods Primers, 2022, 2, .	21.2	14
36	Kinetic and structural changes in <scp>H</scp>smt<scp>P</scp>he<scp>RS</scp>, induced by pathogenic mutations in human <scp>FARS</scp>2. Protein Science, 2017, 26, 1505-1516.	7.6	13

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37	Milestones in treatments for inborn errors of metabolism: Reflections on <scp>Where chemistry and medicine meet</scp>. American Journal of Medical Genetics, Part A, 2021, 185, 3350-3358.	1.2	13
38	Mitochondrial ataxias. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2018, 155, 129-141.	1.8	11
39	Variants in the transcriptional corepressor BCORL1 are associated with an Xâ€linked disorder of intellectual disability, dysmorphic features, and behavioral abnormalities. American Journal of Medical Genetics, Part A, 2019, 179, 870-874.	1.2	11
40	An improved functional assay in blood spot to diagnose Barth syndrome using the monolysocardiolipin/cardiolipin ratio. Journal of Inherited Metabolic Disease, 2022, 45, 29-37.	3.6	11
41	Prospective diagnosis of MT-ATP6-related mitochondrial disease by newborn screening. Molecular Genetics and Metabolism, 2021, 134, 37-42.	1.1	10
42	A New Mouse Model of Mild Ornithine Transcarbamylase Deficiency (spf-j) Displays Cerebral Amino Acid Perturbations at Baseline and upon Systemic Immune Activation. PLoS ONE, 2015, 10, e0116594.	2.5	8
43	Mitochondrial disease disrupts hepatic allostasis and lowers the threshold for immune-mediated liver toxicity. Molecular Metabolism, 2020, 37, 100981.	6.5	8
44	The management of pregnancy and delivery in 3-hydroxy-3-methylglutaryl-CoA lyase deficiency. , 2016, 170, 1600-1602.		7
45	Diverse mitochondrial abnormalities in a new cellular model of TAFFAZZIN deficiency are remediated by cardiolipin-interacting small molecules. Journal of Biological Chemistry, 2021, 297, 101005.	3.4	7
46	Accurate assignment of disease liability to genetic variants using only population data. Genetics in Medicine, 2022, 24, 87-99.	2.4	4
47	Cover Image, Volume 179A, Number 5, May 2019. , 2019, 179, i-i.		3
48	PARS2-associated mitochondrial disease: A case report of a patient with prolonged survival and literature review. Molecular Genetics and Metabolism Reports, 2020, 24, 100613.	1.1	3
49	Cardiolipinâ€™s Remodeling Rules Revealed: The Role of the Cellular Lipidome. Cell Reports, 2020, 30, 3949-3950.	6.4	3
50	Arginine kinetics are altered in a pilot sample of adolescents and young adults with Barth syndrome. Molecular Genetics and Metabolism Reports, 2020, 25, 100675.	1.1	2
51	High-resolution mass spectrometric analysis of cardiolipin profiles in Barth syndrome. Mitochondrion, 2021, 60, 27-32.	3.4	2
52	Barth syndrome and the many fascinating aspects of cardiolipin. Journal of Inherited Metabolic Disease, 2022, 45, 1-2.	3.6	1
53	Finding Treatments for Genetic Metabolic Disease. Current Pediatrics Reports, 2016, 4, 173-177.	4.0	0
54	Case Report: SATB2-Associated Syndrome Overlapping With Clinical Mitochondrial Disease Presentation: Report of Two Cases. Frontiers in Genetics, 2021, 12, 692087.	2.3	0

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55	Investigating Mitochondrial Dysfunction in Barth Syndrome. FASEB Journal, 2022, 36, .	0.5	0
56	Quality of life in Barth syndrome. Therapeutic Advances in Rare Disease, 2022, 3, 263300402210937.	0.7	0