

David Watkins

List of Publications by Year in descending order

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Version: 2024-02-01

32
papers

1,175
citations

471477

17
h-index

395678

33
g-index

38
all docs

38
docs citations

38
times ranked

1381
citing authors

#	ARTICLE	IF	CITATIONS
1	Mutations in ABCD4 cause a new inborn error of vitamin B12 metabolism. <i>Nature Genetics</i> , 2012, 44, 1152-1155.	21.4	191
2	Inborn errors of cobalamin absorption and metabolism. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , 2011, 157, 33-44.	1.6	167
3	An X-Linked Cobalamin Disorder Caused by Mutations in Transcriptional Coregulator HCFC1. <i>American Journal of Human Genetics</i> , 2013, 93, 506-514.	6.2	110
4	Update and new concepts in vitamin responsive disorders of folate transport and metabolism. <i>Journal of Inherited Metabolic Disease</i> , 2012, 35, 665-670.	3.6	67
5	Novel inborn error of folate metabolism: identification by exome capture and sequencing of mutations in the MTHFD1 gene in a single proband. <i>Journal of Medical Genetics</i> , 2011, 48, 590-592.	3.2	66
6	A PRDX1 mutant allele causes a MMACHC secondary epimutation in cblC patients. <i>Nature Communications</i> , 2018, 9, 67.	12.8	64
7	Human mutations in methylenetetrahydrofolate dehydrogenase 1 impair nuclear de novo thymidylate biosynthesis. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2015, 112, 400-405.	7.1	56
8	Mutations in THAP11 cause an inborn error of cobalamin metabolism and developmental abnormalities. <i>Human Molecular Genetics</i> , 2017, 26, 2838-2849.	2.9	47
9	Defective lysosomal release of vitamin B12 (cblF): A hereditary cobalamin metabolic disorder associated with sudden death. <i>American Journal of Medical Genetics Part A</i> , 1989, 33, 555-563.	2.4	41
10	Severe Methylenetetrahydrofolate Reductase Deficiency. <i>JAMA Neurology</i> , 2014, 71, 901.	9.0	38
11	Shifting landscapes of human MTHFR missense-variant effects. <i>American Journal of Human Genetics</i> , 2021, 108, 1283-1300.	6.2	33
12	Lessons in biology from patients with inborn errors of vitamin B12 metabolism. <i>Biochimie</i> , 2013, 95, 1019-1022.	2.6	31
13	Inborn Error of Cobalamin Metabolism Associated with the Intracellular Accumulation of Transcobalamin-Bound Cobalamin and Mutations in <i>ZNF143</i> , Which Codes for a Transcriptional Activator. <i>Human Mutation</i> , 2016, 37, 976-982.	2.5	30
14	Sacrificial Cobalt-Carbon Bond Homolysis in Coenzyme B ₁₂ as a Cofactor Conservation Strategy. <i>Journal of the American Chemical Society</i> , 2018, 140, 13205-13208.	13.7	24
15	Added value of next generation gene panel analysis for patients with elevated methylmalonic acid and no clinical diagnosis following functional studies of vitamin B12 metabolism. <i>Molecular Genetics and Metabolism</i> , 2016, 117, 363-368.	1.1	23
16	Severe 5,10-Methylenetetrahydrofolate Reductase Deficiency and Two MTHFR Variants in an Adolescent With Progressive Myoclonic Epilepsy. <i>Pediatric Neurology</i> , 2014, 51, 266-270.	2.1	21
17	Lessons in biology from patients with inherited disorders of vitamin B12 and folate metabolism. <i>Biochimie</i> , 2016, 126, 3-5.	2.6	17
18	Allosteric Regulation of Oligomerization by a B12 Trafficking G-Protein Is Corrupted in Methylmalonic Aciduria. <i>Cell Chemical Biology</i> , 2019, 26, 960-969.e4.	5.2	16

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19	Immunodeficiency and inborn disorders of vitamin B12 and folate metabolism. <i>Current Opinion in Clinical Nutrition and Metabolic Care</i> , 2020, 23, 241-246.	2.5	15
20	Methylmalonyl-coA epimerase deficiency: A new case, with an acute metabolic presentation and an intronic splicing mutation in the MCEE gene. <i>Molecular Genetics and Metabolism Reports</i> , 2016, 9, 19-24.	1.1	14
21	Inherited defects of cobalamin metabolism. <i>Vitamins and Hormones</i> , 2022, 119, 355-376.	1.7	12
22	Methionine synthase deficiency: Variable clinical presentation and benefit of early diagnosis and treatment. <i>Journal of Inherited Metabolic Disease</i> , 2022, 45, 157-168.	3.6	10
23	Epimutations in both the TESK2 and MMACHC promoters in the Epi-cblC inherited disorder of intracellular metabolism of vitamin B12. <i>Clinical Epigenetics</i> , 2022, 14, 52.	4.1	10
24	New insights into the metabolic and nutritional determinants of severe combined immunodeficiency. <i>Rare Diseases (Austin, Tex)</i> , 2015, 3, e1112479.	1.8	9
25	Next generation sequencing of patients with mut methylmalonic aciduria: Validation of somatic cell studies and identification of 16 novel mutations. <i>Molecular Genetics and Metabolism</i> , 2016, 118, 264-271.	1.1	9
26	The emerging role of the mitochondrial fatty-acid synthase (mtFASII) in the regulation of energy metabolism. <i>Biochimica Et Biophysica Acta - Molecular and Cell Biology of Lipids</i> , 2019, 1864, 1629-1643.	2.4	9
27	Biochemical analysis of patients with mutations in MTHFD1 and a diagnosis of methylenetetrahydrofolate dehydrogenase 1 deficiency. <i>Molecular Genetics and Metabolism</i> , 2020, 130, 179-182.	1.1	9
28	Methionine dependence in tumor cells: The potential role of cobalamin and MMACHC. <i>Molecular Genetics and Metabolism</i> , 2021, 132, 155-161.	1.1	9
29	Epimutation in inherited metabolic disorders: the influence of aberrant transcription in adjacent genes. <i>Human Genetics</i> , 2022, , 1.	3.8	7
30	Genome and RNA sequencing in patients with methylmalonic aciduria of unknown cause. <i>Genetics in Medicine</i> , 2020, 22, 432-436.	2.4	4
31	Probing the functional consequence and clinical relevance of <i>CD320</i> p.E88del, a variant in the transcobalamin receptor gene. <i>American Journal of Medical Genetics, Part A</i> , 2022, 188, 1124-1141.	1.2	2
32	Confounding factors in identification of disease-resilient individuals. <i>Nature Biotechnology</i> , 2016, 34, 1103-1104.	17.5	0