David Watkins

List of Publications by Year
in descending order

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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	Methionine synthase deficiency: Variable clinical presentation and benefit of early diagnosis and treatment. Journal of Inherited Metabolic Disease, 2022, 45, 157-168.	3.6	10
2	Probing the functional consequence and clinical relevance of <scp>CD320</scp> p.E88del, a variant in the transcobalamin receptor gene. American Journal of Medical Genetics, Part A, 2022, 188, 1124-1141.	1.2	2
3	Inherited defects of cobalamin metabolism. Vitamins and Hormones, 2022, 119, 355-376.	1.7	12
4	Epimutation in inherited metabolic disorders: the influence of aberrant transcription in adjacent genes. Human Genetics, 2022, , 1.	3.8	7
5	Epimutations in both the TESK2 and MMACHC promoters in the Epi-cblC inherited disorder of intracellular metabolism of vitamin B12. Clinical Epigenetics, 2022, 14, 52.	4.1	10
6	Methionine dependence in tumor cells: The potential role of cobalamin and MMACHC. Molecular Genetics and Metabolism, 2021, 132, 155-161.	1.1	9
7	Shifting landscapes of human MTHFR missense-variant effects. American Journal of Human Genetics, 2021, 108, 1283-1300.	6.2	33
8	Genome and RNA sequencing in patients with methylmalonic aciduria of unknown cause. Genetics in Medicine, 2020, 22, 432-436.	2.4	4
9	Immunodeficiency and inborn disorders of vitamin B12 and folate metabolism. Current Opinion in Clinical Nutrition and Metabolic Care, 2020, 23, 241-246.	2.5	15
10	Biochemical analysis of patients with mutations in MTHFD1 and a diagnosis of methylenetetrahydrofolate dehydrogenase 1 deficiency. Molecular Genetics and Metabolism, 2020, 130, 179-182.	1.1	9
11	The emerging role of the mitochondrial fatty-acid synthase (mtFASII) in the regulation of energy metabolism. Biochimica Et Biophysica Acta - Molecular and Cell Biology of Lipids, 2019, 1864, 1629-1643.	2.4	9
12	Allosteric Regulation of Oligomerization by a B12 Trafficking G-Protein Is Corrupted in Methylmalonic Aciduria. Cell Chemical Biology, 2019, 26, 960-969.e4.	5.2	16
13	A PRDX1 mutant allele causes a MMACHC secondary epimutation in cblC patients. Nature Communications, 2018, 9, 67.	12.8	64
14	Sacrificial Cobaltâ€“Carbon Bond Homolysis in Coenzyme B₁₂ as a Cofactor Conservation Strategy. Journal of the American Chemical Society, 2018, 140, 13205-13208.	13.7	24
15	Mutations in THAP11 cause an inborn error of cobalamin metabolism and developmental abnormalities. Human Molecular Genetics, 2017, 26, 2838-2849.	2.9	47
16	Lessons in biology from patients with inherited disorders of vitamin B12 and folate metabolism. Biochimie, 2016, 126, 3-5.	2.6	17
17	Inborn Error of Cobalamin Metabolism Associated with the Intracellular Accumulation of Transcobalamin-Bound Cobalamin and Mutations inZNF143, Which Codes for a Transcriptional Activator. Human Mutation, 2016, 37, 976-982.	2.5	30
18	Methylmalonyl-coA epimerase deficiency: A new case, with an acute metabolic presentation and an intronic splicing mutation in the MCEE gene. Molecular Genetics and Metabolism Reports, 2016, 9, 19-24.	1.1	14

#	Article	IF	Citations
19	Confounding factors in identification of disease-resilient individuals. Nature Biotechnology, 2016, 34, 1103-1104.	17.5	0
20	Next generation sequencing of patients with mut methylmalonic aciduria: Validation of somatic cell studies and identification of 16 novel mutations. Molecular Genetics and Metabolism, 2016, 118, 264-271.	1.1	9
21	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. Molecular Genetics and Metabolism, 2016, 117, 363-368.	1.1	23
22	New insights into the metabolic and nutritional determinants of severe combined immunodeficiency. Rare Diseases (Austin, Tex), 2015, 3, e1112479.	1.8	9
23	Human mutations in methylenetetrahydrofolate dehydrogenase 1 impair nuclear de novo thymidylate biosynthesis. Proceedings of the National Academy of Sciences of the United States of America, 2015, 112, 400-405.	7.1	56
24	Severe Methylenetetrahydrofolate Reductase Deficiency. JAMA Neurology, 2014, 71, 901.	9.0	38
25	Severe 5,10-Methylenetetrahydrofolate Reductase Deficiency and Two MTHFR Variants in an Adolescent With Progressive Myoclonic Epilepsy. Pediatric Neurology, 2014, 51, 266-270.	2.1	21
26	An X-Linked Cobalamin Disorder Caused by Mutations in Transcriptional Coregulator HCFC1. American Journal of Human Genetics, 2013, 93, 506-514.	6.2	110
27	Lessons in biology from patients with inborn errors of vitamin B12 metabolism. Biochimie, 2013, 95, 1019-1022.	2.6	31
28	Mutations in ABCD4 cause a new inborn error of vitamin B12 metabolism. Nature Genetics, 2012, 44, 1152-1155.	21.4	191
29	Update and new concepts in vitamin responsive disorders of folate transport and metabolism. Journal of Inherited Metabolic Disease, 2012, 35, 665-670.	3.6	67
30	Inborn errors of cobalamin absorption and metabolism. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2011, 157, 33-44.	1.6	167
31	Novel inborn error of folate metabolism: identification by exome capture and sequencing of mutations in the MTHFD1 gene in a single proband. Journal of Medical Genetics, 2011, 48, 590-592.	3.2	66
32	Defective lysosomal release of vitamin B12 (cblF): A hereditary cobalamin metabolic disorder associated with sudden death. American Journal of Medical Genetics Part A, 1989, 33, 555-563.	2.4	41