Beat Thony

List of Publications by Year in descending order

122 papers	5,764 citations	109321 35 h-index	85541 71 g-index
127 all docs	127 docs citations	127 times ranked	5373 citing authors

#	Article	IF	Citations
1	Tetrahydrobiopterin biosynthesis, regeneration and functions. Biochemical Journal, 2000, 347, 1-16.	3.7	771
2	Tetrahydrobiopterin: biochemistry and pathophysiology. Biochemical Journal, 2011, 438, 397-414.	3.7	390
3	Treatment of a metabolic liver disease by in vivo genome base editing in adult mice. Nature Medicine, 2018, 24, 1519-1525.	30.7	301
4	Tetrahydrobiopterin biosynthesis, regeneration and functions. Biochemical Journal, 2000, 347 Pt 1, 1-16.	3.7	261
5	In vivo adenine base editing of PCSK9 in macaques reduces LDL cholesterol levels. Nature Biotechnology, 2021, 39, 949-957.	17.5	196
6	Tetrahydrobiopterin Deficiencies without Hyperphenylalaninemia: Diagnosis and Genetics of DOPA-Responsive Dystonia and Sepiapterin Reductase Deficiency. Molecular Genetics and Metabolism, 2001, 74, 172-185.	1.1	172
7	Mutations in the BH4-metabolizing genes GTP cyclohydrolase I, 6-pyruvoyl-tetrahydropterin synthase, sepiapterin reductase, carbinolamine-4a-dehydratase, and dihydropteridine reductase. Human Mutation, 2006, 27, 870-878.	2.5	167
8	Molecular genetics of tetrahydrobiopterin-responsive phenylalanine hydroxylase deficiency. Human Mutation, 2008, 29, 167-175.	2.5	158
9	The Genetic Landscape and Epidemiology of Phenylketonuria. American Journal of Human Genetics, 2020, 107, 234-250.	6.2	138
10	Biallelic Mutations in DNAJC12 Cause Hyperphenylalaninemia, Dystonia, and Intellectual Disability. American Journal of Human Genetics, 2017, 100, 257-266.	6.2	127
11	Sepiapterin reductase deficiency: A Treatable Mimic of Cerebral Palsy. Annals of Neurology, 2012, 71, 520-530.	5.3	125
12	Distinct Proteomic, Transcriptomic, and Epigenetic Stress Responses in Dorsal and Ventral Hippocampus. Biological Psychiatry, 2018, 84, 531-541.	1.3	106
13	Mutations in human monoamine-related neurotransmitter pathway genes. Human Mutation, 2008, 29, 891-902.	2.5	92
14	Fusion-dependent formation of lipid nanoparticles containing macromolecular payloads. Nanoscale, 2019, 11, 9023-9031.	5.6	85
15	Consensus guideline for the diagnosis and treatment of tetrahydrobiopterin (BH4) deficiencies. Orphanet Journal of Rare Diseases, 2020, 15, 126.	2.7	85
16	Mutations in the GTP cyclohydrolase I and 6-pyruvoyl-tetrahydropterin synthase genes. Human Mutation, 1997, 10, 11-20.	2.5	75
17	Computationally designed liver-specific transcriptional modules and hyperactive factor IX improve hepatic gene therapy. Blood, 2014, 123, 3195-3199.	1.4	73
18	Administration-route and gender-independent long-term therapeutic correction of phenylketonuria (PKU) in a mouse model by recombinant adeno-associated virus 8 pseudotyped vector-mediated gene transfer. Gene Therapy, 2006, 13, 587-593.	4.5	72

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19	Molecular genetics and impact of residual in vitro phenylalanine hydroxylase activity on tetrahydrobiopterin responsiveness in Turkish PKU population. Molecular Genetics and Metabolism, 2011, 102, 116-121.	1.1	71
20	In vivo prime editing of a metabolic liver disease in mice. Science Translational Medicine, 2022, 14, eabl9238.	12.4	71
21	Aromatic amino acid decarboxylase deficiency: Molecular and metabolic basis and therapeutic outlook. Molecular Genetics and Metabolism, 2019, 127, 12-22.	1.1	66
22	Treatment of phenylketonuria using minicircle-based naked-DNA gene transfer to murine liver. Hepatology, 2014, 60, 1035-1043.	7.3	62
23	In vivo cytidine base editing of hepatocytes without detectable off-target mutations in RNA and DNA. Nature Biomedical Engineering, 2021, 5, 179-189.	22.5	62
24	Sepiapterin reductase deficiency an autosomal recessive DOPA-responsive dystonia. Molecular Genetics and Metabolism, 2006, 89, 116-120.	1.1	61
25	Autism spectrum disorder associated with low serotonin in CSF and mutations in the SLC29A4 plasma membrane monoamine transporter (PMAT) gene. Molecular Autism, 2014, 5, 43.	4.9	59
26	Dwarfism and Low Insulin-like Growth Factor-1 Due to Dopamine Depletion in Pts â€“/â€“ Mice Rescued by Feeding Neurotransmitter Precursors and H4-biopterin. Journal of Biological Chemistry, 2003, 278, 28303-28311.	3.4	57
27	Tetrahydrobiopterin protects phenylalanine hydroxylase activity in vivo: Implications for tetrahydrobiopterin-responsive hyperphenylalaninemia. FEBS Letters, 2004, 577, 507-511.	2.8	55
28	Correction of Murine PKU Following AAV-mediated Intramuscular Expression of a Complete Phenylalanine Hydroxylating System. Molecular Therapy, 2008, 16, 673-681.	8.2	54
29	DNAJC12 deficiency: A new strategy in the diagnosis of hyperphenylalaninemias. Molecular Genetics and Metabolism, 2018, 123, 1-5.	1.1	52
30	Tetrahydrobiopterin shows chaperone activity for tyrosine hydroxylase. Journal of Neurochemistry, 2008, 106, 672-681.	3.9	48
31	Folinic acid treatment for schizophrenia associated with folate receptor autoantibodies. Molecular Genetics and Metabolism, 2014, 113, 307-314.	1.1	47
32	Molecular analysis and long-term follow-up of patients with different forms of 6-pyruvoyl-tetrahydropterin synthase deficiency. European Journal of Pediatrics, 2001, 160, 267-276.	2.7	45
33	Comparison of Adeno-Associated Virus Pseudotype 1, 2, and 8 Vectors Administered by Intramuscular Injection in the Treatment of Murine Phenylketonuria. Human Gene Therapy, 2010, 21, 463-477.	2.7	40
34	Sepiapterin Reductase Deficiency: Clinical Presentation and Evaluation of Long-Term Therapy. Pediatric Neurology, 2006, 35, 308-313.	2.1	39
35	Enhancement of hepatic autophagy increases ureagenesis and protects against hyperammonemia. Proceedings of the National Academy of Sciences of the United States of America, 2018, 115, 391-396.	7.1	39
36	Long-term glial cell line-derived neurotrophic factor overexpression in the intact nigrostriatal system in rats leads to a decrease of dopamine and increase of tetrahydrobiopterin production. Journal of Neurochemistry, 2005, 93, 1482-1486.	3.9	38

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37	Blood phenylalanine reduction corrects CNS dopamine and serotonin deficiencies and partially improves behavioral performance in adult phenylketonuric mice. Molecular Genetics and Metabolism, 2018, 123, 6-20.	1.1	38
38	Structural and Functional Consequences of Mutations in 6-Pyruvoyltetrahydropterin Synthase Causing Hyperphenylalaninemia in Humans. Journal of Biological Chemistry, 1995, 270, 29498-29506.	3.4	37
39	Diagnosis of tetrahydrobiopterin deficiency using filter paper blood spots: further development of the method and 5 years experience. Journal of Inherited Metabolic Disease, 2011, 34, 819-826.	3.6	37
40	Mutations in the pterin-4Î±-carbinolamine dehydratase (PCBD) gene cause a benign form of hyperphenylalaninemia. Human Genetics, 1998, 103, 162-167.	3.8	36
41	Hyperphenylalaninemia due to defects in tetrahydrobiopterin metabolism: molecular characterization of mutations in 6-pyruvoyl-tetrahydropterin synthase. American Journal of Human Genetics, 1994, 54, 782-92.	6.2	35
42	Disease-causing mutations improving the branch site and polypyrimidine tract: Pseudoexon activation of LINE-2 and antisenseAlulacking the poly(T)-tail. Human Mutation, 2009, 30, 823-831.	2.5	33
43	Quantification of phenylalanine hydroxylase activity by isotope-dilution liquid chromatographyâ€“electrospray ionization tandem mass spectrometry. Molecular Genetics and Metabolism, 2012, 105, 559-565.	1.1	32
44	Variant of dihydropteridine reductase deficiency without hyperphenylalaninaemia: Effect of oral phenylalanine loading. Journal of Inherited Metabolic Disease, 1999, 22, 216-220.	3.6	31
45	Brain catecholamine depletion and motor impairment in aThknock-in mouse with type B tyrosine hydroxylase deficiency. Brain, 2015, 138, 2948-2963.	7.6	31
46	Neurological improvement following intravenous high-dose folinic acid for cerebral folate transporter deficiency caused by FOLR-1 mutation. European Journal of Paediatric Neurology, 2016, 20, 709-713.	1.6	31
47	Tetrahydrobiopterin and inherited hyperphenylalaninemias. Turkish Journal of Pediatrics, 1996, 38, 19-35.	0.6	31
48	Reduced nitric oxide metabolites in CSF of patients with tetrahydrobiopterin deficiency. Journal of Neurochemistry, 2002, 80, 362-364.	3.9	30
49	Chromosomal Localization, Genomic Structure and Characterization of the Human Gene and a Retropseudogene for 6-Pyruvoyltetrahydropterin Synthase. FEBS Journal, 1996, 240, 477-484.	0.2	29
50	Effect of pharmacological chaperones on brain tyrosine hydroxylase and tryptophan hydroxylase 2. Journal of Neurochemistry, 2010, 114, 853-863.	3.9	29
51	Heterogeneous clinical spectrum of DNAJC12-deficient hyperphenylalaninemia: from attention deficit to severe dystonia and intellectual disability. Journal of Medical Genetics, 2018, 55, 249-253.	3.2	29
52	State-of-the-Art 2019 on Gene Therapy for Phenylketonuria. Human Gene Therapy, 2019, 30, 1274-1283.	2.7	29
53	Identification of mutations causing 6-pyruvoyl-tetrahydropterin synthase deficiency in four Italian families. Human Mutation, 1997, 10, 25-35.	2.5	28
54	High dose sapropterin dihydrochloride therapy improves monoamine neurotransmitter turnover in murine phenylketonuria (PKU). Molecular Genetics and Metabolism, 2016, 117, 5-11.	1.1	28

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55	Molecular and metabolic bases of tetrahydrobiopterin (BH4) deficiencies. Molecular Genetics and Metabolism, 2021, 133, 123-136.	1.1	28
56	Nuclear localization of tetrahydrobiopterin biosynthetic enzymes. Biochimica Et Biophysica Acta - General Subjects, 2004, 1670, 56-68.	2.4	26
57	Cysteamine revisited: repair of arginine to cysteine mutations. Journal of Inherited Metabolic Disease, 2017, 40, 555-567.	3.6	26
58	Pseudoexon exclusion by antisense therapy in 6-pyruvoyl-tetrahydropterin synthase deficiency. Human Mutation, 2011, 32, 1019-1027.	2.5	25
59	Low tetrahydrobiopterin biosynthetic capacity of human monocytes is caused by exon skipping in 6-pyruvoyl tetrahydropterin synthase. Biochemical Journal, 2003, 373, 681-688.	3.7	24
60	Stimulation of hepatic phenylalanine hydroxylase activity but not Pah-mRNA expression upon oral loading of tetrahydrobiopterin in normal mice. Molecular Genetics and Metabolism, 2005, 86, 153-155.	1.1	24
61	Two Greek siblings with sepiapterin reductase deficiency. Molecular Genetics and Metabolism, 2008, 94, 403-409.	1.1	23
62	Characterization of the Human PCBD Gene Encoding the Bifunctional Protein Pterin-4Î±-carbinolamine Dehydratase/Dimerization Cofactor for the Transcription Factor Hnf-1Î±. Biochemical and Biophysical Research Communications, 1995, 210, 966-973.	2.1	22
63	Autism associated with low 5-hydroxyindolacetic acid in CSF and the heterozygous SLC6A4 gene Gly56Ala plus 5-HTTLPR L/L promoter variants. Molecular Genetics and Metabolism, 2011, 102, 368-373.	1.1	22
64	Phenylalanine hydroxylase variants interact with the coâ€chaperone DNAJC12. Human Mutation, 2019, 40, 483-494.	2.5	22
65	Expression and characterization of recombinant human and rat liver 6-pyruvoyl tetrahydropterin synthase. Modified cysteine residues inhibit the enzyme activity. FEBS Journal, 1994, 219, 497-502.	0.2	20
66	Functional Characterization of the spf/ash Splicing Variation in OTC Deficiency of Mice and Man. PLoS ONE, 2015, 10, e0122966.	2.5	20
67	Tetrahydrobiopterin Loading Test in Xanthine Dehydrogenase and Molybdenum Cofactor Deficiencies. Biochemical and Molecular Medicine, 1996, 58, 199-203.	1.4	18
68	Low-Dose Gene Therapy for Murine PKU Using Episomal Naked DNA Vectors Expressing PAH from Its Endogenous Liver Promoter. Molecular Therapy - Nucleic Acids, 2017, 7, 339-349.	5.1	18
69	A simple dried blood spot-method for in vivo measurement of ureagenesis by gas chromatographyâ€“mass spectrometry using stable isotopes. Clinica Chimica Acta, 2017, 464, 236-243.	1.1	17
70	Oxidative Stress, Folate Receptor Autoimmunity, and CSF Findings in Severe Infantile Autism. Autism Research & Treatment, 2020, 2020, 1-14.	0.5	17
71	Pterins and Related Enzymes. , 2008, , 665-701.		17
72	Stereospecificity and Catalytic Function of Histidine Residues in 4a-Hydroxy-tetrahydropterin Dehydratase/DCoHâ€. Biochemistry, 1998, 37, 11246-11254.	2.5	16

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73	Immunohistochemical localisation of pterin-4Î±-carbinolamine dehydratase in rat peripheral organs. Histochemistry and Cell Biology, 1999, 111, 381-390.	1.7	16
74	Four Years of Diagnostic Challenges with Tetrahydrobiopterin Deficiencies in Iranian Patients. JIMD Reports, 2016, 32, 7-14.	1.5	16
75	Sensory stimulus-sensitive drop attacks and basal ganglia calcification: new findings in a patient with FOLR1 deficiency. Epileptic Disorders, 2014, 16, 88-92.	1.3	15
76	Reconstitution of a metabolic pathway with triple-cistronic IRES-containing retroviral vectors for correction of tetrahydrobiopterin deficiency. Journal of Gene Medicine, 2000, 2, 22-31.	2.8	14
77	Sepiapterin reductase deficiency in a 2â€yearâ€old girl with incomplete response to treatment during shortâ€term followâ€up. Journal of Inherited Metabolic Disease, 2009, 32, 5-10.	3.6	14
78	Hepatocyte Transfection in Small Pigs After Weaning by Hydrodynamic Intraportal Injection of Naked DNA/Minicircle Vectors. Human Gene Therapy Methods, 2015, 26, 181-192.	2.1	14
79	Treatment of Cystathionine Î²-Synthase Deficiency in Mice Using a Minicircle-Based Naked DNA Vector. Human Gene Therapy, 2019, 30, 1093-1100.	2.7	14
80	Dominant negative allele (N47D) in a compound heterozygote for a variant of 6-pyruvoyltetrahydropterin synthase deficiency causing transient hyperphenylalaninemia. Human Mutation, 1999, 13, 286-289.	2.5	13
81	A liverâ€humanized mouse model of carbamoyl phosphate synthetase 1â€deficiency. Journal of Inherited Metabolic Disease, 2019, 42, 1054-1063.	3.6	13
82	Long-term correction of murine phenylketonuria by viral gene transfer: liver versus muscle. Journal of Inherited Metabolic Disease, 2010, 33, 677-680.	3.6	12
83	The mechanism of BH4-responsive hyperphenylalaninemia-As it occurs in the ENU1/2 genetic mouse model. Human Mutation, 2012, 33, 1464-1473.	2.5	12
84	A female case of aromatic l-amino acid decarboxylase deficiency responsive to MAO-B inhibition. Brain and Development, 2016, 38, 959-963.	1.1	12
85	A novel common large genomic deletion and two new missense mutations identified in the Romanian phenylketonuria population. Gene, 2016, 576, 182-188.	2.2	12
86	Natural history, with clinical, biochemical, and molecular characterization of classical homocystinuria in the Qatari population. Journal of Inherited Metabolic Disease, 2019, 42, 818-830.	3.6	12
87	Correction of a urea cycle defect after exÂvivo gene editing of human hepatocytes. Molecular Therapy, 2021, 29, 1903-1917.	8.2	12
88	Isolated central form of tetrahydrobiopterin deficiency associated with hemizygosity on chromosome 11q and a mutant allele of PTPS. Human Mutation, 2000, 16, 54-60.	2.5	11
89	The spectrum of phenylketonuria genotypes in the Armenian population: Identification of three novel mutant PAH alleles. Molecular Genetics and Metabolism, 2011, 104, S93-S96.	1.1	11
90	The Pah-R261Q mouse reveals oxidative stress associated with amyloid-like hepatic aggregation of mutant phenylalanine hydroxylase. Nature Communications, 2021, 12, 2073.	12.8	11

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91	Mildly compromised tetrahydrobiopterin cofactor biosynthesis due to Pts variants leads to unusual body fat distribution and abdominal obesity in mice. Journal of Inherited Metabolic Disease, 2016, 39, 309-319.	3.6	10
92	Tetrahydrobiopterin treatment reduces brain Lâ€Phe but only partially improves serotonin in hyperphenylalaninemic ENU1/2 mice. Journal of Inherited Metabolic Disease, 2018, 41, 709-718.	3.6	10
93	In silico and in vivo models for Qatari-specific classical homocystinuria as basis for development of novel therapies. Human Mutation, 2019, 40, 230-240.	2.5	10
94	Preclinical evaluation of liposome-supported peritoneal dialysis for the treatment of hyperammonemic crises. Journal of Controlled Release, 2020, 328, 503-513.	9.9	10
95	A novel Pah-exon1 deleted murine model of phenylalanine hydroxylase (PAH) deficiency. Molecular Genetics and Metabolism, 2020, 131, 306-315.	1.1	10
96	Peripheral biopterin and neopterin in schizophrenia and depression. Psychiatry Research, 2021, 297, 113745.	3.3	10
97	Retrovirus-Mediated Gene Transfer of 6-Pyruvoyl-Tetrahydropterin Synthase Corrects Tetrahydrobiopterin Deficiency in Fibroblasts from Hyperphenylalaninemic Patients. Human Gene Therapy, 1996, 7, 1587-1593.	2.7	9
98	Retrovirus-Mediated Double Transduction of the GTPCH and PTPS Genes Allows 6-Pyruvoyltetrahydropterin Synthase-Deficient Human Fibroblasts to Synthesize and Release Tetrahydrobiopterin. Journal of Neurochemistry, 2002, 71, 33-40.	3.9	9
99	Comprehensive characterization of ureagenesis in the spf^{ash} mouse, a model of human ornithine transcarbamylase deficiency, reveals ageâ€dependency of ammonia detoxification. Journal of Inherited Metabolic Disease, 2019, 42, 1064-1076.	3.6	9
100	Short Communication. Biological Chemistry, 1998, 379, 1441-1456.	2.5	8
101	Improvement of DNA Vector Delivery of DOTAP Lipoplexes by Short-Chain Aminolipids. ACS Omega, 2020, 5, 24724-24732.	3.5	8
102	Development of Covalent Chitosan-Polyethylenimine Derivatives as Gene Delivery Vehicle: Synthesis, Characterization, and Evaluation. International Journal of Molecular Sciences, 2021, 22, 3828.	4.1	8
103	A Sensitive Assay System To Test Antisense Oligonucleotides for Splice Suppression Therapy in the Mouse Liver. Molecular Therapy - Nucleic Acids, 2014, 3, e193.	5.1	7
104	Analysis of the Qatari R336C cystathionine Î²â€synthase protein in mice. Journal of Inherited Metabolic Disease, 2019, 42, 831-838.	3.6	7
105	Role of protein structure in variant annotation: structural insight of mutations causing 6-pyruvoyl-tetrahydropterin synthase deficiency. Pathology, 2019, 51, 274-280.	0.6	7
106	Novel Protein Biomarkers of Monoamine Metabolism Defects Correlate with Disease Severity. Movement Disorders, 2021, 36, 690-703.	3.9	7
107	Tetrahydrobiopterin deficiencies: Lesson from clinical experience. JIMD Reports, 2021, 59, 42-51.	1.5	7
108	Mutation Analysis In Patients with 6-Pyruvoyl-Tetrahydropterin Synthase Deficiency. Pteridines, 1995, 6, 141-143.	0.5	7

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109	Antisense Oligonucleotide Rescue of Deep-Intronic Variants Activating Pseudoexons in the 6-Pyruvoyl-Tetrahydropterin Synthase Gene. Nucleic Acid Therapeutics, 2022, 32, 378-390.	3.6	7
110	Delivery of non-viral naked DNA vectors to liver in small weaned pigs by hydrodynamic retrograde intrabiliary injection. Molecular Therapy - Methods and Clinical Development, 2022, 24, 268-279.	4.1	6
111	Mutational spectrum of PTS gene and in silico pathological assessment of a novel variant in Mexico. Brain and Development, 2018, 40, 530-536.	1.1	5
112	Development of a porcine model of phenylketonuria with a humanized R408W mutation for gene editing. PLoS ONE, 2021, 16, e0245831.	2.5	5
113	Characterization of Human Pterin-4a-carbinolamine Dehydratase/Dimerization Cofactor of Hepatocyte Nuclear Factor-1Î±, and of the Cys81-mutant Involved in Hyperphenylalanincemia. Pteridines, 1995, 6, 123-125.	0.5	3
114	Phenotypic Reversion of Fair Hair upon Gene Therapy of the Phenylketonuria Mice. Human Gene Therapy, 2014, 25, 573-574.	2.7	3
115	Tyrosine hydroxylase deficiencyâ€"Clinical insights and a novel deletion in TH gene in an Indian patient. JIMD Reports, 2020, 53, 12-15.	1.5	3
116	Modeling the cognitive effects of diet discontinuation in adults with phenylketonuria (PKU) using pegvaliase therapy in PAH-deficient mice. Molecular Genetics and Metabolism, 2022, 136, 46-64.	1.1	3
117	Murine models of inherited monoaminergic and GABAergic neurotransmitter disorders. Future Neurology, 2006, 1, 665-676.	0.5	2
118	Editorial. Journal of Inherited Metabolic Disease, 2019, 42, 1041-1043.	3.6	2
119	Sapropterin dihydrochloride therapy in dihydropteridine reductase deficiency: Insight from the first case with molecular diagnosis in Brazil. JIMD Reports, 2021, 61, 19-24.	1.5	1
120	Isolation of 6-Pyruvoyl-tetrahydropterin Synthase cDNAs from Human Brain. Pteridines, 1996, 7, 91-93.	0.5	1
121	Human Pterinâ€4Î±â€Carbinolamine Dehydratase/Dimerization Cofactor of Hepatocyte Nuclear Factorâ€1Î±. FEBS Journal, 1995, 231, 414-423.	0.2	0
122	Challenges of experimental gene therapy for urea cycle disorders. Journal of Pediatric Biochemistry, 2015, 04, 065-073.	0.2	0