Tomas Honzik

List of Publications by Year
in descending order

101 papers	3,501 citations	159358 30 h-index	155451 55 g-index
112 all docs	112 docs citations	112 times ranked	4828 citing authors

#	Article	IF	Citations
1	Integrative Approach to Predict Severity in Nonketotic Hyperglycinemia. Annals of Neurology, 2022, 92, 292-303.	2.8	3
2	A Novel MTTK Gene Variant m.8315A>C as a Cause of MERRF Syndrome. Genes, 2022, 13, 1245.	1.0	1
3	Subjective and polysomnographic evaluation of sleep in mitochondrial optic neuropathies. Journal of Sleep Research, 2021, 30, e13051.	1.7	5
4	GPD1 Deficiency â€“ Underdiagnosed Cause of Liver Disease. Indian Journal of Pediatrics, 2021, 88, 80-81.	0.3	4
5	International consensus guidelines for phosphoglucomutase 1 deficiency (<scp>PGM1â€CDG</scp>): Diagnosis, followâ€up, and management. Journal of Inherited Metabolic Disease, 2021, 44, 148-163.	1.7	27
6	Congenital disorders of glycosylation: Still â€œhotâ€•in 2020. Biochimica Et Biophysica Acta - General Subjects, 2021, 1865, 129751.	1.1	77
7	Brain <scp>MR</scp> patterns in inherited disorders of monoamine neurotransmitters: An analysis of 70 patients. Journal of Inherited Metabolic Disease, 2021, 44, 1070-1082.	1.7	13
8	Guidelines for the diagnosis and management of methylmalonic acidaemia and propionic acidaemia: FirstÂrevision. Journal of Inherited Metabolic Disease, 2021, 44, 566-592.	1.7	118
9	Genetic heterogeneity of neuronal intranuclear inclusion disease: What about the infantile variant?. Annals of Clinical and Translational Neurology, 2021, 8, 994-1001.	1.7	2
10	ALG3-CDG: a patient with novel variants and review of the genetic and ophthalmic findings. BMC Ophthalmology, 2021, 21, 249.	0.6	4
11	Assessment of intellectual impairment, healthâ€related quality of life, and behavioral phenotype in patients with neurotransmitter related disorders: Data from the <scp>iNTD</scp> registry. Journal of Inherited Metabolic Disease, 2021, 44, 1489-1502.	1.7	7
12	Homozygous missense mutation in UQCRC2 associated with severe encephalomyopathy, mitochondrial complex III assembly defect and activation of mitochondrial protein quality control. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2021, 1867, 166147.	1.8	11
13	Impact of Newborn Screening and Early Dietary Management on Clinical Outcome of Patients with Long Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency and Medium Chain Acyl-CoA Dehydrogenase Deficiencyâ€”A Retrospective Nationwide Study. Nutrients, 2021, 13, 2925.	1.7	4
14	Should patients with Phosphomannomutase 2-CDG (PMM2-CDG) be screened for adrenal insufficiency?. Molecular Genetics and Metabolism, 2021, 133, 397-399.	0.5	3
15	Insights into the expanding phenotypic spectrum of inherited disorders of biogenic amines. Nature Communications, 2021, 12, 5529.	5.8	21
16	Should Patients with Kearns-Sayre Syndrome and Corneal Endothelial Failure Be Genotyped for a TCF4 Trinucleotide Repeat, Commonly Associated with Fuchs Endothelial Corneal Dystrophy?. Genes, 2021, 12, 1918.	1.0	0
17	X-linked adrenoleukodystrophy: phenotype-genotype correlation in hemizygous males and heterozygous females with ABCD1 mutations. Neuroendocrinology Letters, 2021, 42, 359-367.	0.2	0
18	Hereditary hyperferritinemia-cataract syndrome in three Czech families: molecular genetic testing and clinical implications. Journal of AAPOS, 2020, 24, 352.e1-352.e5.	0.2	2

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19	An eosinophilic papulopustular rash in a baby. Pediatric Dermatology, 2020, 37, e32-e34.	0.5	0
20	Biallelic MADD variants cause a phenotypic spectrum ranging from developmental delay to a multisystem disorder. Brain, 2020, 143, 2437-2453.	3.7	21
21	The Phenotypic Spectrum of 47 Czech Patients with Single, Large-Scale Mitochondrial DNA Deletions. Brain Sciences, 2020, 10, 766.	1.1	8
22	Novel Splicing Variant in the PMM2 Gene in a Patient With PMM2-CDG Syndrome Presenting With Pericardial Effusion: A Case Report. Frontiers in Genetics, 2020, 11, 561054.	1.1	1
23	Consensus guideline for the diagnosis and treatment of tetrahydrobiopterin (BH4) deficiencies. Orphanet Journal of Rare Diseases, 2020, 15, 126.	1.2	85
24	Severe phenotype of <scp>ATP6AP1â€CDG</scp> in two siblings with a novel mutation leading to a differential tissueâ€specific <scp>ATP6AP1</scp> protein pattern, cellular oxidative stress and hepatic copper accumulation. Journal of Inherited Metabolic Disease, 2020, 43, 694-700.	1.7	14
25	Fatal neonatal nephrocutaneous syndrome in 18 Roma children with EGFR deficiency. Journal of Dermatology, 2020, 47, 663-668.	0.6	6
26	Consensus guideline for the diagnosis and management of mannose phosphate isomeraseâ€congenital disorder of glycosylation. Journal of Inherited Metabolic Disease, 2020, 43, 671-693.	1.7	40
27	A new role for dolichol isoform profile in the diagnostics of CDG disorders. Clinica Chimica Acta, 2020, 507, 88-93.	0.5	5
28	Multisystem mitochondrial diseases due to mutations in mtDNA-encoded subunits of complex I. BMC Pediatrics, 2020, 20, 41.	0.7	23
29	Age Dependent Progression of Multiple Epiphyseal Dysplasia and Pseudoachondroplasia Due to Heterozygous Mutations in COMP Gene. Prague Medical Report, 2020, 121, 153-162.	0.4	1
30	The Metabolic Map into the Pathomechanism and Treatment of PGM1-CDG. American Journal of Human Genetics, 2019, 104, 835-846.	2.6	59
31	Evaluation of dietary treatment and amino acid supplementation in organic acidurias and ureaâ€cycle disorders: On the basis of information from a European multicenter registry. Journal of Inherited Metabolic Disease, 2019, 42, 1162-1175.	1.7	30
32	International clinical guidelines for the management of phosphomannomutase 2â€congenital disorders of glycosylation: Diagnosis, treatment and follow up. Journal of Inherited Metabolic Disease, 2019, 42, 5-28.	1.7	91
33	Decreased plasma l-arginine levels in organic acidurias (MMA and PA) and decreased plasma branched-chain amino acid levels in urea cycle disorders as a potential cause of growth retardation: Options for treatment. Molecular Genetics and Metabolism, 2019, 126, 397-405.	0.5	26
34	GP224â€...Multisystem mitochondrial diseases in children with maternally inherited complex I deficiency. , 2019, , .		0
35	Sideroblastic anemia associated with multisystem mitochondrial disorders. Pediatric Blood and Cancer, 2019, 66, e27591.	0.8	13
36	Long-term follow-up in PMM2-CDG: are we ready to start treatment trials?. Genetics in Medicine, 2019, 21, 1181-1188.	1.1	36

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37	Peripapillary microcirculation in Leber hereditary optic neuropathy. Acta Ophthalmologica, 2019, 97, e71-e76.	0.6	23
38	POLR3B-associated leukodystrophy: clinical, neuroimaging and molecular-genetic analyses in four patients: clinical heterogeneity and novel mutations in POLR3B gene. Neurologia I Neurochirurgia Polska, 2019, 53, 369-376.	0.6	0
39	Attenuated Type of Asphyxiating Thoracic Dysplasia due to Mutations in DYNC2H1 Gene. Prague Medical Report, 2019, 120, 124-130.	0.4	2
40	Review of SRD5A3 Disease-Causing Sequence Variants and Ocular Findings in Steroid 5Î±-Reductase Type 3 Congenital Disorder of Glycosylation, and a Detailed New Case. Folia Biologica, 2019, 65, 134-141.	0.8	5
41	Variable X-chromosome inactivation and enlargement of pericentral glutamine synthetase zones in the liver of heterozygous females with OTC deficiency. Virchows Archiv Fur Pathologische Anatomie Und Physiologie Und Fur Klinische Medizin, 2018, 472, 1029-1039.	1.4	12
42	Mutations in Vps15 perturb neuronal migration in mice and are associated with neurodevelopmental disease in humans. Nature Neuroscience, 2018, 21, 207-217.	7.1	30
43	Aberrant apolipoprotein C-III glycosylation in glycogen storage disease type III and IX. Metabolism: Clinical and Experimental, 2018, 82, 135-141.	1.5	9
44	Revisiting mitochondrial diagnostic criteria in the new era of genomics. Genetics in Medicine, 2018, 20, 444-451.	1.1	62
45	Integrating glycomics and genomics uncovers SLC10A7 as essential factor for bone mineralization by regulating post-Golgi protein transport and glycosylation. Human Molecular Genetics, 2018, 27, 3029-3045.	1.4	37
46	Changes in Transcription Pattern Lead to a Marked Decrease in COX, CS and SQR Activity After the Developmental Point of the 22nd Gestational Week. Physiological Research, 2018, 67, 79-91.	0.4	3
47	Advances in treatment of inherited metabolic disorders with neurological symptomatology. Neurologie Pro Praxi, 2018, 19, 100-103.	0.0	0
48	Activity of the liver enzyme ornithine carbamoyltransferase (OTC) in blood: LC-MS/MS assay for non-invasive diagnosis of ornithine carbamoyltransferase deficiency. Clinical Chemistry and Laboratory Medicine, 2017, 55, 1168-1177.	1.4	10
49	Oral D-galactose supplementation in PGM1-CDG. Genetics in Medicine, 2017, 19, 1226-1235.	1.1	55
50	<scp>OPA</scp>1 analysis in an international series of probands with bilateral optic atrophy. Acta Ophthalmologica, 2017, 95, 363-369.	0.6	7
51	Thymidine kinase 2 and alanyl-tRNA synthetase 2 deficiencies cause lethal mitochondrial cardiomyopathy: case reports and review of the literature. Cardiology in the Young, 2017, 27, 936-944.	0.4	28
52	Muscular dystrophies and myopathies: the spectrum of mutated genes in the Czech Republic. Clinical Genetics, 2017, 91, 463-469.	1.0	32
53	Hereditary Multiple Exostoses: Clinical, Molecular and Radiologic Survey in 9 Families. Prague Medical Report, 2017, 118, 87-94.	0.4	2
54	The phenotypic spectrum of fifty Czech m.3243A>G carriers. Molecular Genetics and Metabolism, 2016, 118, 288-295.	0.5	29

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55	The International Working Group on Neurotransmitter related Disorders (iNTD): A worldwide research project focused on primary and secondary neurotransmitter disorders. Molecular Genetics and Metabolism Reports, 2016, 9, 61-66.	0.4	48
56	Defining the Phenotype and Assessing Severity in Phosphoglucomutase-1ÂDeficiency. Journal of Pediatrics, 2016, 175, 130-136.e8.	0.9	43
57	Unique presentation of LHON/MELAS overlap syndrome caused by m.13046T>C in MTND5. Ophthalmic Genetics, 2016, 37, 419-423.	0.5	10
58	Recessive <scp>ITPA</scp> mutations cause an early infantile encephalopathy. Annals of Neurology, 2015, 78, 649-658.	2.8	45
59	Hyperammonemic crisis in a child with ATP synthase deficiency caused by mtDNA mutation m.8851T>C. Molecular Genetics and Metabolism Reports, 2015, 2, 46.	0.4	0
60	DCDC2 Mutations Cause a Renal-Hepatic Ciliopathy by Disrupting Wnt Signaling. American Journal of Human Genetics, 2015, 96, 81-92.	2.6	98
61	TMEM70 deficiency: longâ€term outcome of 48 patients. Journal of Inherited Metabolic Disease, 2015, 38, 417-426.	1.7	51
62	The phenotypic spectrum of organic acidurias and urea cycle disorders. Part 2: the evolving clinical phenotype. Journal of Inherited Metabolic Disease, 2015, 38, 1059-1074.	1.7	175
63	Analysis of Mitochondrial Network Morphology in Cultured Myoblasts from Patients with Mitochondrial Disorders. Ultrastructural Pathology, 2015, 39, 340-350.	0.4	5
64	Triple trouble â€“ DMD, autism, epilepsy. Neuromuscular Disorders, 2015, 25, S243.	0.3	0
65	Isoelectric Focusing of Serum Apolipoprotein C-III as a Sensitive Screening Method for the Detection of O-glycosylation Disturbances. Prague Medical Report, 2015, 116, 73-86.	0.4	7
66	Proposed guidelines for the diagnosis and management of methylmalonic and propionic acidemia. Orphanet Journal of Rare Diseases, 2014, 9, 130.	1.2	482
67	Large copy number variations in combination with point mutations in the TYMP and SCO2 genes found in two patients with mitochondrial disorders. European Journal of Human Genetics, 2014, 22, 431-434.	1.4	11
68	Sebelipase alfa over 52weeks reduces serum transaminases, liver volume and improves serum lipids in patients with lysosomal acid lipase deficiency. Journal of Hepatology, 2014, 61, 1135-1142.	1.8	81
69	Novel FBN1 gene mutation and maternal germinal mosaicism as the cause of neonatal form of Marfan syndrome. American Journal of Medical Genetics, Part A, 2014, 164, 1559-1564.	0.7	18
70	Mosaic tissue distribution of the tandem duplication of LAMP2 exons 4 and 5 demonstrates the limits of Danon disease cellular and molecular diagnostics. Journal of Inherited Metabolic Disease, 2014, 37, 117-124.	1.7	17
71	Mutation of Nogo-B Receptor, a Subunit of cis-Prenyltransferase, Causes a Congenital Disorder of Glycosylation. Cell Metabolism, 2014, 20, 448-457.	7.2	104
72	Glycogen storage disease-like phenotype with central nervous system involvement in a PGM1-CDG patient. Neuroendocrinology Letters, 2014, 35, 137-41.	0.2	27

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73	Non-invasive screening of cytochrome c oxidase deficiency in children using a dipstick immunocapture assay. Folia Biologica, 2014, 60, 268-74.	0.8	1
74	Lipoprotein lipase deficiency: clinical, biochemical and molecular characteristics in three patients with novel mutations in the LPL gene. Folia Biologica, 2014, 60, 235-43.	0.8	6
75	Different laboratory and muscle biopsy findings in a family with an m.8851T>C mutation in the mitochondrial MTATP6 gene. Molecular Genetics and Metabolism, 2013, 108, 102-105.	0.5	9
76	Clinical effect and safety profile of recombinant human lysosomal acid lipase in patients With cholesteryl ester storage disease. Hepatology, 2013, 58, 950-957.	3.6	99
77	Novel Mutations in the TAZ Gene in Patients with Barth Syndrome. Prague Medical Report, 2013, 114, 139-153.	0.4	16
78	Neonatal onset of mitochondrial disorders in 129 patients: clinical and laboratory characteristics and a new approach to diagnosis. Journal of Inherited Metabolic Disease, 2012, 35, 749-759.	1.7	65
79	Clinical picture of S-adenosylhomocysteine hydrolase deficiency resembles phosphomannomutase 2 deficiency. Molecular Genetics and Metabolism, 2012, 107, 611-613.	0.5	30
80	RFT1-CDG in adult siblings with novel mutations. Molecular Genetics and Metabolism, 2012, 107, 760-762.	0.5	13
81	Hypertrophic Cardiomyopathy Due to the Mitochondrial DNA Mutation m.3303C>T Diagnosed in an Adult Male. International Heart Journal, 2012, 53, 383-387.	0.5	16
82	Elevated CSF-lactate is a reliable marker of mitochondrial disorders in children even after brief seizures. European Journal of Paediatric Neurology, 2011, 15, 101-108.	0.7	27
83	Response to the â€˜letter to the Editorâ€™. European Journal of Paediatric Neurology, 2011, 15, 378.	0.7	0
84	Vascular presentation of cystathionine betaâ€synthase deficiency in adulthood. Journal of Inherited Metabolic Disease, 2011, 34, 33-37.	1.7	47
85	Clinical presentation and metabolic consequences in 40 breastfed infants with nutritional vitamin B12 deficiency â€“ What have we learned?. European Journal of Paediatric Neurology, 2010, 14, 488-495.	0.7	116
86	Developmental changes of mitochondrial DNA content and expression of genes involved in mtDNA transcription and maintenance in human fetal liver and muscle tissues. Biochimica Et Biophysica Acta - Bioenergetics, 2010, 1797, 108-109.	0.5	2
87	Mitochondrial encephalocardio-myopathy with early neonatal onset due to TMEM70 mutation. Archives of Disease in Childhood, 2010, 95, 296-301.	1.0	72
88	237 Csf-Lactate as a Marker of Mitochondrial Disorders Even in Children After Brief Seizures. Pediatric Research, 2010, 68, 123-123.	1.1	21
89	Mitochondrial DNA content and expression of genes involved in mtDNA transcription, regulation and maintenance during human fetal development. Mitochondrion, 2010, 10, 321-329.	1.6	57
90	A new case of ALG8 deficiency (CDG Ih). Journal of Inherited Metabolic Disease, 2009, 32, 259-264.	1.7	140

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91	Activities of respiratory chain complexes and pyruvate dehydrogenase in isolated muscle mitochondria in premature neonates. Early Human Development, 2008, 84, 269-276.	0.8	30
92	TMEM70 mutations cause isolated ATP synthase deficiency and neonatal mitochondrial encephalocardiomyopathy. Nature Genetics, 2008, 40, 1288-1290.	9.4	183
93	Development of a human mitochondrial oligonucleotide microarray (h-MitoArray) and gene expression analysis of fibroblast cell lines from 13 patients with isolated F1Fo ATP synthase deficiency. BMC Genomics, 2008, 9, 38.	1.2	22
94	The impact of mitochondrial tRNA mutations on the amount of ATP synthase differs in the brain compared to other tissues. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2008, 1782, 317-325.	1.8	37
95	Mitochondrial DNA Haplogroups in the Czech Population Compared to Other European Countries. Human Biology, 2008, 80, 669-674.	0.4	6
96	The developmental changes in mitochondrial DNA content per cell in human cord blood leukocytes during gestation. Physiological Research, 2008, 57, 947-955.	0.4	24
97	Prolonged impairment of polymorphonuclear cells functions in one infant with transient zinc deficiency: a case report. Prague Medical Report, 2008, 109, 184-93.	0.4	1
98	Lethal Fetal and Early Neonatal Presentation of Adenylosuccinate Lyase Deficiency: Observation of 6 Patients in 4 Families. Journal of Pediatrics, 2007, 150, 57-61.e2.	0.9	58
99	Specific Properties of Heavy Fraction of Mitochondria from Human-term Placenta â€“ Glycerophosphate-dependent Hydrogen Peroxide Production. Placenta, 2006, 27, 348-356.	0.7	16
100	Carnitine concentrations in term and preterm newborns at birth and during the first days of life. Prague Medical Report, 2005, 106, 297-306.	0.4	12
101	Polarographic evaluation of mitochondrial enzymes activity in isolated mitochondria and in permeabilized human muscle cells with inherited mitochondrial defects. Physiological Research, 2003, 52, 781-8.	0.4	16