Peter M Van Hasselt

List of Publications by Year
in descending order

110 papers	3,553 citations	117625 34 h-index	168389 53 g-index
111 all docs	111 docs citations	111 times ranked	6062 citing authors

#	Article	IF	Citations
1	Neurofilament light chain and glial fibrillary acidic protein levels in metachromatic leukodystrophy. Brain, 2022, 145, 105-118.	7.6	18
2	The potential and limitations of intrahepatic cholangiocyte organoids to study inborn errors of metabolism. Journal of Inherited Metabolic Disease, 2022, 45, 353-365.	3.6	4
3	Aberrant cyclin C nuclear release induces mitochondrial fragmentation and dysfunction in MED13L syndrome fibroblasts. IScience, 2022, 25, 103823.	4.1	3
4	Modified Delphi procedure-based expert consensus on endpoints for an international disease registry for Metachromatic Leukodystrophy: The European Metachromatic Leukodystrophy initiative (MLDi). Orphanet Journal of Rare Diseases, 2022, 17, 48.	2.7	10
5	Bilateral posterior lamellar corneal transplant surgery in an infant of 17 weeks old: Surgical challenges and the added value of intraoperative optical coherence tomography. Clinical Case Reports (discontinued), 2022, 10, e05637.	0.5	3
6	Towards Understanding Behaviour and Emotions of Children with CLN3 Disease (Batten Disease): Patterns, Problems and Support for Child and Family. International Journal of Environmental Research and Public Health, 2022, 19, 5895.	2.6	2
7	Recognizing differentiating clinical signs of CLN3 disease (Batten disease) at presentation. Acta Ophthalmologica, 2021, 99, 397-404.	1.1	9
8	Deep intronic TIMMDC1 variant delays diagnosis of rapidly progressive complex I deficiency. European Journal of Medical Genetics, 2021, 64, 104120.	1.3	2
9	Missense and truncating variants in CHD5 in a dominant neurodevelopmental disorder with intellectual disability, behavioral disturbances, and epilepsy. Human Genetics, 2021, 140, 1109-1120.	3.8	18
10	Beyond nephronophthisis: Retinal dystrophy in the absence of kidney dysfunction in childhood expands the clinical spectrum of <scp>CEP83</scp> deficiency. American Journal of Medical Genetics, Part A, 2021, 185, 2204-2210.	1.2	7
11	Long-term effect of hematopoietic cell transplantation on systemic inflammation in patients with mucopolysaccharidoses. Blood Advances, 2021, 5, 3092-3101.	5.2	2
12	Therapy-type related long-term outcomes in mucopolysaccaridosis type II (Hunter syndrome) â€“ Case series. Molecular Genetics and Metabolism Reports, 2021, 28, 100779.	1.1	1
13	Quantifying the Effects of Hip Surgery on the Sphericity of the Femoral Head in Patients with Mucopolysaccharidosis Type I. Journal of Bone and Joint Surgery - Series A, 2021, 103, 489-496.	3.0	1
14	Automatic quantification of lymphocyte vacuolization in peripheral blood smears of patients with Batten's disease (CLN3 disease). JIMD Reports, 2021, 58, 100-103.	1.5	0
15	NAA80 bi-allelic missense variants result in high-frequency hearing loss, muscle weakness and developmental delay. Brain Communications, 2021, 3, fcab256.	3.3	14
16	Internalization and Transport of PEGylated Lipid-Based Mixed Micelles across Caco-2 Cells Mediated by Scavenger Receptor B1. Pharmaceutics, 2021, 13, 2022.	4.5	1
17	Glucose transporter type 1 deficiency syndrome and the ketogenic diet. Journal of Inherited Metabolic Disease, 2020, 43, 216-222.	3.6	29
18	Inborn errors of enzymes in glutamate metabolism. Journal of Inherited Metabolic Disease, 2020, 43, 200-215.	3.6	13

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19	Metabolic fingerprinting reveals extensive consequences of GLS hyperactivity. Biochimica Et Biophysica Acta - General Subjects, 2020, 1864, 129484.	2.4	3
20	Retrospective evaluation of the Dutch preâ€•newborn screening cohort for propionic acidemia and isolated methylmalonic acidemia: What to aim, expect, and evaluate from newborn screening?. Journal of Inherited Metabolic Disease, 2020, 43, 424-437.	3.6	18
21	Hypothesis: determining phenotypic specificity facilitates understanding of pathophysiology in rare genetic disorders. Journal of Inherited Metabolic Disease, 2020, 43, 701-711.	3.6	6
22	Longitudinal Analysis of Ocular Disease in Children with Mucopolysaccharidosis I after Hematopoietic Cell Transplantation. Biology of Blood and Marrow Transplantation, 2020, 26, 928-935.	2.0	6
23	Prime editing for functional repair in patient-derived disease models. Nature Communications, 2020, 11, 5352.	12.8	134
24	Quantifying lymphocyte vacuolization serves as a measure of CLN3 disease severity. JIMD Reports, 2020, 54, 87-97.	1.5	6
25	Hearing loss in patients with mucopolysaccharidoses â€1 and â€6 after hematopoietic cell transplantation: A longitudinal analysis. Journal of Inherited Metabolic Disease, 2020, 43, 1279-1287.	3.6	5
26	Hurdles in treating Hurler disease: potential routes to achieve a â€œrealâ€•cure. Blood Advances, 2020, 4, 2837-2849.	5.2	8
27	The c.1Aâ€‰>â€‰C start codon mutation in CLN3 is associated with a protracted disease course. JIMD Reports, 2020, 52, 23-27.	1.5	6
28	Accurate discrimination of Hartnup disorder from other aminoacidurias using a diagnostic ratio. Molecular Genetics and Metabolism Reports, 2020, 22, 100551.	1.1	3
29	Variants in NGLY1 lead to intellectual disability, myoclonus epilepsy, sensorimotor axonal polyneuropathy and mitochondrial dysfunction. Clinical Genetics, 2020, 97, 556-566.	2.0	19
30	Metachromatic leukodystrophy and transplantation: remyelination, no crossâ€•correction. Annals of Clinical and Translational Neurology, 2020, 7, 169-180.	3.7	45
31	Untargeted Metabolomics for Metabolic Diagnostic Screening with Automated Data Interpretation Using a Knowledge-Based Algorithm. International Journal of Molecular Sciences, 2020, 21, 979.	4.1	16
32	Cross-Omics: Integrating Genomics with Metabolomics in Clinical Diagnostics. Metabolites, 2020, 10, 206.	2.9	20
33	Aminoacyl-tRNA synthetase deficiencies in search of common themes. Genetics in Medicine, 2019, 21, 319-330.	2.4	70
34	Aspartylglycosamine is a biomarker for NGLY1-CDDG, a congenital disorder of deglycosylation. Molecular Genetics and Metabolism, 2019, 127, 368-372.	1.1	31
35	De Novo Heterozygous POLR2A Variants Cause a Neurodevelopmental Syndrome with Profound Infantile-Onset Hypotonia. American Journal of Human Genetics, 2019, 105, 283-301.	6.2	46
36	Early prediction of phenotypic severity in Citrullinemia Type 1. Annals of Clinical and Translational Neurology, 2019, 6, 1858-1871.	3.7	26

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37	A narrative review of factors associated with the development and progression if nonâ€alcoholic fatty liver disease. GastroHep, 2019, 1, 180.	0.6	4
38	Biallelic Variants in ASNA1 , Encoding a Cytosolic Targeting Factor of Tail-Anchored Proteins, Cause Rapidly Progressive Pediatric Cardiomyopathy. Circulation Genomic and Precision Medicine, 2019, 12, 397-406.	3.6	16
39	Glutaminase Deficiency Caused by Short Tandem Repeat Expansion inGLS. New England Journal of Medicine, 2019, 381, 1185-1185.	27.0	2
40	Disruptive variants of CSDE1 associate with autism and interfere with neuronal development and synaptic transmission. Science Advances, 2019, 5, eaax2166.	10.3	35
41	Treatment of thoracolumbar kyphosis in patients with mucopolysaccharidosis type I: results of an international consensus procedure. Orphanet Journal of Rare Diseases, 2019, 14, 17.	2.7	11
42	Motor function impairment is an early sign of CLN3 disease. Neurology, 2019, 93, e293-e297.	1.1	13
43	Pathophysiology of propionic and methylmalonic acidemias. Part 2: Treatment strategies. Journal of Inherited Metabolic Disease, 2019, 42, 745-761.	3.6	27
44	Pathophysiology of propionic and methylmalonic acidemias. Part 1: Complications. Journal of Inherited Metabolic Disease, 2019, 42, 730-744.	3.6	68
45	Increasing the dose of oral vitamin K prophylaxis and its effect on bleeding risk. European Journal of Pediatrics, 2019, 178, 1033-1042.	2.7	13
46	Direct-infusion based metabolomics unveils biochemical profiles of inborn errors of metabolism in cerebrospinal fluid. Molecular Genetics and Metabolism, 2019, 127, 51-57.	1.1	13
47	Impact of newborn screening for veryâ€longâ€chain acylâ€CoA dehydrogenase deficiency on genetic, enzymatic, and clinical outcomes. Journal of Inherited Metabolic Disease, 2019, 42, 414-423.	3.6	36
48	Synapse alterations precede neuronal damage and storage pathology in a human cerebral organoid model of CLN3-juvenile neuronal ceroid lipofuscinosis. Acta Neuropathologica Communications, 2019, 7, 222.	5.2	49
49	Emotional and behavioral problems, quality of life and metabolic control in NTBC-treated Tyrosinemia type 1 patients. Orphanet Journal of Rare Diseases, 2019, 14, 285.	2.7	19
50	GLS hyperactivity causes glutamate excess, infantile cataract and profound developmental delay. Human Molecular Genetics, 2019, 28, 96-104.	2.9	23
51	IQSEC2-related encephalopathy in males and females: a comparative study including 37 novel patients. Genetics in Medicine, 2019, 21, 837-849.	2.4	47
52	Identification of a Loss-of-Function Mutation in the Context of Glutaminase Deficiency and Neonatal Epileptic Encephalopathy. JAMA Neurology, 2019, 76, 342.	9.0	33
53	Expanding the Spectrum of BAF-Related Disorders: De Novo Variants in SMARCC2 Cause a Syndrome with Intellectual Disability and Developmental Delay. American Journal of Human Genetics, 2019, 104, 164-178.	6.2	59
54	Direct Infusion Based Metabolomics Identifies Metabolic Disease in Patientsâ€™ Dried Blood Spots and Plasma. Metabolites, 2019, 9, 12.	2.9	48

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55	Clinical, neuroradiological, and biochemical features of SLC35A2â€CDG patients. Journal of Inherited Metabolic Disease, 2019, 42, 553-564.	3.6	32
56	Timing of cognitive decline in CLN3 disease. Journal of Inherited Metabolic Disease, 2018, 41, 257-261.	3.6	24
57	Beneficial Effect of BH4 Treatment in a 15-Year-Old Boy with Biallelic Mutations in DNAJC12. JIMD Reports, 2018, 42, 99-103.	1.5	15
58	KIAA1109 Variants Are Associated with a Severe Disorder of Brain Development and Arthrogryposis. American Journal of Human Genetics, 2018, 102, 116-132.	6.2	46
59	Hypothesis: lobe A (COG1â€“4)-CDG causes a more severe phenotype than lobe B (COG5â€“8)-CDG. Journal of Medical Genetics, 2018, 55, 137-142.	3.2	14
60	Autism spectrum disorder: an early and frequent feature in cerebrotendinous xanthomatosis. Journal of Inherited Metabolic Disease, 2018, 41, 641-646.	3.6	19
61	Early and late outcomes after cord blood transplantation for pediatric patients with inherited leukodystrophies. Blood Advances, 2018, 2, 49-60.	5.2	45
62	Mixed micellar system stabilized with saponins for oral delivery of vitamin K. Colloids and Surfaces B: Biointerfaces, 2018, 170, 521-528.	5.0	16
63	Influence of PEGylation of Vitamin-K-Loaded Mixed Micelles on the Uptake by and Transport through Caco-2 Cells. Molecular Pharmaceutics, 2018, 15, 3786-3795.	4.6	6
64	Bi-allelic mutations in TRAPPC2L result in a neurodevelopmental disorder and have an impact on RAB11 in fibroblasts. Journal of Medical Genetics, 2018, 55, 753-764.	3.2	39
65	Salivary Î±-Iduronidase Activity as a Potential New Biomarker for the Diagnosis and Monitoring the Effect of Therapy in Mucopolysaccharidosis Type I. Biology of Blood and Marrow Transplantation, 2018, 24, 1808-1813.	2.0	3
66	Incomplete biomarker response in mucopolysaccharidosis type I after successful hematopoietic cell transplantation. Molecular Genetics and Metabolism, 2017, 122, 86-91.	1.1	4
67	Ankyrin repeat and zinc-finger domain-containing 1 mutations are associated with infantile-onset inflammatory bowel disease. Journal of Biological Chemistry, 2017, 292, 7904-7920.	3.4	29
68	Mutations in MDH2, Encoding a Krebs Cycle Enzyme, Cause Early-Onset Severe Encephalopathy. American Journal of Human Genetics, 2017, 100, 151-159.	6.2	63
69	A Case of Unexpected Adult-Onset Neurologic Decline in CLN3-Associated Retinal Degeneration. JAMA Ophthalmology, 2017, 135, 1451.	2.5	5
70	Progressive deafnessâ€“dystonia due to SERAC1 mutations: A study of 67 cases. Annals of Neurology, 2017, 82, 1004-1015.	5.3	63
71	Quality of life of Hurler syndrome patients after successful hematopoietic stem cell transplantation. Blood Advances, 2017, 1, 2236-2242.	5.2	19
72	Synaptic UNC13A protein variant causes increased neurotransmission and dyskinetic movement disorder. Journal of Clinical Investigation, 2017, 127, 1005-1018.	8.2	84

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73	Is FGF13 a major contributor to genetic epilepsy with febrile seizures plus?. Epilepsy Research, 2016, 128, 48-51.	1.6	7
74	A New Approach for Fast Metabolic Diagnostics in CMAMMA. JIMD Reports, 2016, 30, 15-22.	1.5	7
75	Efficacy of hematopoietic cell transplantation in metachromatic leukodystrophy: the Dutch experience. Blood, 2016, 127, 3098-3101.	1.4	56
76	Neurodevelopmental Outcome after Hematopoietic Cell Transplantation in Inborn Errors of Metabolism: Current Considerations and Future Perspectives. Neuropediatrics, 2016, 47, 285-292.	0.6	29
77	A Mixed Micelle Formulation for Oral Delivery of Vitamin K. Pharmaceutical Research, 2016, 33, 2168-2179.	3.5	37
78	Gallbladder and the risk of polyps and carcinoma in metachromatic leukodystrophy. Neurology, 2016, 87, 103-111.	1.1	40
79	Effectiveness of whole-exome sequencing and costs of the traditional diagnostic trajectory in children with intellectual disability. Genetics in Medicine, 2016, 18, 949-956.	2.4	148
80	Destabilized SMC5/6 complex leads to chromosome breakage syndrome with severe lung disease. Journal of Clinical Investigation, 2016, 126, 2881-2892.	8.2	65
81	Vitamin B6 in Plasma and Cerebrospinal Fluid of Children. PLoS ONE, 2015, 10, e0120972.	2.5	23
82	Hematopoietic Cell Transplantation for MPS Patients Is Safe and Effective: Results after Implementation of International Guidelines. Biology of Blood and Marrow Transplantation, 2015, 21, S93.	2.0	4
83	Pain: a prevalent feature in patients with mucopolysaccharidosis. Results of a crossâ€sectional national survey. Journal of Inherited Metabolic Disease, 2015, 38, 323-331.	3.6	34
84	Monocarboxylate Transporter 1 Deficiency and Ketone Utilization. New England Journal of Medicine, 2015, 372, 578-579.	27.0	2
85	Hematopoietic Cell Transplantation for Mucopolysaccharidosis Patients Is Safe and Effective: Results after Implementation of International Guidelines. Biology of Blood and Marrow Transplantation, 2015, 21, 1106-1109.	2.0	138
86	Key features and clinical variability of COG6-CDG. Molecular Genetics and Metabolism, 2015, 116, 163-170.	1.1	49
87	Eyes on MEGDEL: Distinctive Basal Ganglia Involvement in Dystonia Deafness Syndrome. Neuropediatrics, 2015, 46, 098-103.	0.6	34
88	Pitfalls in Diagnosing Neuraminidase Deficiency: Psychosomatics and Normal Sialic Acid Excretion. JIMD Reports, 2015, 25, 9-13.	1.5	15
89	Suitability of methylmalonic acid and total homocysteine analysis in dried bloodspots. Analytica Chimica Acta, 2015, 853, 435-441.	5.4	13
90	Survival and Psychomotor Development With Early Betaine Treatment in Patients With Severe Methylenetetrahydrofolate Reductase Deficiency. JAMA Neurology, 2014, 71, 188.	9.0	60

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91	Perioperative complications in patients diagnosed with mucopolysaccharidosis and the impact of enzyme replacement therapy followed by hematopoietic stem cell transplantation at early age. Paediatric Anaesthesia, 2014, 24, 521-527.	1.1	23
92	Expanding the clinical phenotype of COG6 deficiency. Journal of Medical Genetics, 2014, 51, 425.1-425.	3.2	2
93	Monocarboxylate Transporter 1 Deficiency and Ketone Utilization. New England Journal of Medicine, 2014, 371, 1900-1907.	27.0	86
94	Impaired Cognitive Functioning in Patients with Tyrosinemia Type I Receiving Nitisinone. Journal of Pediatrics, 2014, 164, 398-401.	1.8	71
95	Loss of Syntaxin 3 Causes Variant Microvillus Inclusion Disease. Gastroenterology, 2014, 147, 65-68.e10.	1.3	151
96	Treatment of hip dysplasia in patients with mucopolysaccharidosis type I after hematopoietic stem cell transplantation: results of an international consensus procedure. Orphanet Journal of Rare Diseases, 2013, 8, 155.	2.7	39
97	Supplementation with a powdered blend of PUFAs normalizes DHA and AA levels in patients with PKU. Molecular Genetics and Metabolism, 2013, 109, 121-124.	1.1	12
98	Improvement of White Matter Changes on Neuroimaging Modalities After Stem Cell Transplant in Metachromatic Leukodystrophy. JAMA Neurology, 2013, 70, 779.	9.0	44
99	Mutations in the phospholipid remodeling gene SERAC1 impair mitochondrial function and intracellular cholesterol trafficking and cause dystonia and deafness. Nature Genetics, 2012, 44, 797-802.	21.4	175
100	Application of exome sequencing in the search for genetic causes of rare disorders of copper metabolism. Metallomics, 2012, 4, 606.	2.4	6
101	Mutation in subdomain G' of mitochondrial elongation factor G1 is associated with combined OXPHOS deficiency in fibroblasts but not in muscle. European Journal of Human Genetics, 2011, 19, 275-279.	2.8	42
102	Intracranial bleeding due to vitaminÂK deficiency: advantages of using a pediatric intensive care registry. Intensive Care Medicine, 2011, 37, 1014-1020.	8.2	18
103	Metabolic Profiles in Children During Fasting. Pediatrics, 2011, 127, e1021-e1027.	2.1	74
104	Hydrolysed Formula Is a Risk Factor for Vitamin K Deficiency in Infants With Unrecognised Cholestasis. Journal of Pediatric Gastroenterology and Nutrition, 2010, 51, 773-776.	1.8	6
105	Fatal outcome due to deficiency of subunit 6 of the conserved oligomeric Golgi complex leading to a new type of congenital disorders of glycosylation. Human Molecular Genetics, 2010, 19, 3623-3633.	2.9	97
106	The influence of bile acids on the oral bioavailability of vitamin K encapsulated in polymeric micelles. Journal of Controlled Release, 2009, 133, 161-168.	9.9	55
107	Prevention of Vitamin K Deficiency Bleeding in Breastfed Infants: Lessons From the Dutch and Danish Biliary Atresia Registries. Pediatrics, 2008, 121, e857-e863.	2.1	74
108	Unmasking of a hemizygous WFS1 gene mutation by a chromosome 4p deletion of 8.3â€‰Mb in a patient with Wolfâ€“Hirschhorn syndrome. European Journal of Human Genetics, 2007, 15, 1132-1138.	2.8	37

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109	Multiple oxidative phosphorylation deficiencies in severe childhood multi-system disorders due to polymerase gamma (POLG1) mutations. European Journal of Pediatrics, 2007, 166, 229-234.	2.7	66
110	Altered Immune Function in Human Newborns after Prenatal Administration of Betamethasone: Enhanced Natural Killer Cell Activity and Decreased T Cell Proliferation in Cord Blood. Pediatric Research, 1999, 45, 306-312.	2.3	66