Marjan Huizing

List of Publications by Year in descending order

Source: https://exaly.com/author-pdf/6881650/publications.pdf

Version: 2024-02-01

115 papers	6,350 citations	57758 44 h-index	74163 75 g-index
120 all docs	120 docs citations	120 times ranked	6554 citing authors

#	Article	IF	Citations
1	Population Pharmacokinetic Model of N-acetylmannosamine (ManNAc) and N-acetylneuraminic acid (Neu5Ac) in Subjects with GNE Myopathy. Drugs in R and D, 2021, 21, 189-202.	2.2	5
2	Free sialic acid storage disorder: Progress and promise. Neuroscience Letters, 2021, 755, 135896.	2.1	12
3	Safety and efficacy of N-acetylmannosamine (ManNAc) in patients with GNE myopathy: an open-label phase 2 study. Genetics in Medicine, 2021, 23, 2067-2075.	2.4	23
4	Genetic variants associated with Hermansky-Pudlak syndrome. Platelets, 2020, 31, 544-547.	2.3	12
5	Quantitation of cytidineâ€5â€²â€monophosphoâ€Nâ€acetylneuraminic acid in human leukocytes using LCâ€“MS/MS: method development and validation. Biomedical Chromatography, 2020, 34, e4735.	1.7	6
6	Hermanskyâ€“Pudlak syndrome: Mutation update. Human Mutation, 2020, 41, 543-580.	2.5	65
7	Inherited disorders of lysosomal membrane transporters. Biochimica Et Biophysica Acta - Biomembranes, 2020, 1862, 183336.	2.6	18
8	Elevated Plasma Free Sialic Acid Levels in Individuals with Reduced Glomerular Filtration Rates. Kidney360, 2020, 1, 957-961.	2.1	2
9	Rationale and Design for a Phase 1 Study of N-Acetylmannosamine for Primary Glomerular Diseases. Kidney International Reports, 2019, 4, 1454-1462.	0.8	8
10	Hermansky-Pudlak syndrome and oculocutaneous albinism in Chinese children with pigmentation defects and easy bruising. Orphanet Journal of Rare Diseases, 2019, 14, 52.	2.7	13
11	237th ENMC International Workshop: GNE myopathy â€“ current and future research Hoofddorp, The Netherlands, 14â€“16 September 2018. Neuromuscular Disorders, 2019, 29, 401-410.	0.6	5
12	Quantification of lectin fluorescence in GNE myopathy muscle biopsies. Muscle and Nerve, 2018, 58, 286-292.	2.2	13
13	GNE Myopathy: Etiology, Diagnosis, and Therapeutic Challenges. Neurotherapeutics, 2018, 15, 900-914.	4.4	63
14	Hermansky-Pudlak syndrome with a novel genetic variant inHPS1and subsequent accelerated pulmonary fibrosis: significance for phenocopy diseases. Thorax, 2018, 73, 1085-1088.	5.6	7
15	Mutations in KIAA0753 cause Joubert syndrome associated with growth hormone deficiency. Human Genetics, 2017, 136, 399-408.	3.8	30
16	Exome analysis of Smithâ€“Magenis-like syndrome cohort identifies de novo likely pathogenic variants. Human Genetics, 2017, 136, 409-420.	3.8	22
17	Clinical and molecular phenotyping of a child with Hermansky-Pudlak syndrome-7, an uncommon genetic type of HPS. Molecular Genetics and Metabolism, 2017, 120, 378-383.	1.1	25
18	Neurologic involvement in patients with atypical Chediak-Higashi disease. Neurology, 2017, 88, e57-e65.	1.1	20

#	Article	IF	Citations
19	Safety, pharmacokinetics and sialic acid production after oral administration of N -acetylmannosamine (ManNAc) to subjects with GNE myopathy. Molecular Genetics and Metabolism, 2017, 122, 126-134.	1.1	41
20	Delayed diagnosis in a house of correction: Smithâ€“Magenis syndrome due to a de novo nonsense RAI1 variant. American Journal of Medical Genetics, Part A, 2016, 170, 2383-2388.	1.2	4
21	New observation of sialuria prompts detection of liver tumor in previously reported patient. Molecular Genetics and Metabolism, 2016, 118, 92-99.	1.1	4
22	Cystic cerebellar dysplasia and biallelicLAMA1mutations: a lamininopathy associated with tics, obsessive compulsive traits and myopia due to cell adhesion and migration defects. Journal of Medical Genetics, 2016, 53, 318-329.	3.2	25
23	Neurologic involvement in patients with atypical Chediak-Higashi disease. Neurology, 2016, 86, 1320-1328.	1.1	21
24	Expanding the clinical and molecular characteristics of PIGT-CDG, a disorder of glycosylphosphatidylinositol anchors. Molecular Genetics and Metabolism, 2015, 115, 128-140.	1.1	44
25	Quantitative hydrophilic interaction chromatographyâ€“mass spectrometry analysis of N-acetylneuraminic acid and N-acetylmannosamine in human plasma. Journal of Chromatography B: Analytical Technologies in the Biomedical and Life Sciences, 2015, 1000, 105-111.	2.3	21
26	In vitro functional correction of Hermanskyâ€“Pudlak Syndrome type-1 by lentiviral-mediated gene transfer. Molecular Genetics and Metabolism, 2015, 114, 62-65.	1.1	10
27	Sialylation of Thomsenâ€“Friedenreich antigen is a noninvasive blood-based biomarker for GNE myopathy. Biomarkers in Medicine, 2014, 8, 641-652.	1.4	19
28	Atypical presentation of GNE myopathy with asymmetric hand weakness. Neuromuscular Disorders, 2014, 24, 1063-1067.	0.6	21
29	GNE myopathy: New name and new mutation nomenclature. Neuromuscular Disorders, 2014, 24, 387-389.	0.6	61
30	Nonâ€specific accumulation of glycosphingolipids in GNE myopathy. Journal of Inherited Metabolic Disease, 2014, 37, 297-308.	3.6	11
31	Mutation Update forGNEGene Variants Associated with GNE Myopathy. Human Mutation, 2014, 35, 915-926.	2.5	90
32	Two novel compound heterozygous mutations in OPA3 in two siblings with OPA3-related 3-methylglutaconic aciduria. Molecular Genetics and Metabolism Reports, 2014, 1, 114-123.	1.1	6
33	Dysregulation of Galectin-3. Implications for Hermansky-Pudlak Syndrome Pulmonary Fibrosis. American Journal of Respiratory Cell and Molecular Biology, 2014, 50, 605-613.	2.9	42
34	Murine isoforms of UDP-GlcNAc 2-epimerase/ManNAc kinase: Secondary structures, expression profiles, and response to ManNAc therapy. Glycoconjugate Journal, 2013, 30, 609-618.	2.7	3
35	Disorders with similar clinical phenotypes reveal underlying genetic interaction: SATB2 acts as an activator of the UPF3B gene. Human Genetics, 2013, 132, 1383-1393.	3.8	24
36	The <scp>BEACH</scp> Is Hot: A <scp>LYST</scp> of Emerging Roles for <scp>BEACH</scp>â€Domain Containing Proteins in Human Disease. Traffic, 2013, 14, 749-766.	2.7	173

#	Article	IF	Citations
37	UDP-GlcNAc 2-Epimerase/ManNAc Kinase (GNE): A Master Regulator of Sialic Acid Synthesis. Topics in Current Chemistry, 2013, 366, 97-137.	4.0	80
38	A BLOCâ€1 mutation screen reveals a novel BLOC1S3 mutation in Hermanskyâ€“Pudlak Syndrome type 8. Pigment Cell and Melanoma Research, 2012, 25, 584-591.	3.3	29
39	Cellular and clinical report of new Griscelli syndrome type III cases. Pigment Cell and Melanoma Research, 2012, 25, 47-56.	3.3	25
40	Oral monosaccharide therapies to reverse renal and muscle hyposialylation in a mouse model of GNE myopathy. Molecular Genetics and Metabolism, 2012, 107, 748-755.	1.1	44
41	The Gne M712T Mouse as a Model for Human Glomerulopathy. American Journal of Pathology, 2012, 180, 1431-1440.	3.8	27
42	Interstitial Lung Disease and Pulmonary Fibrosis in Hermansky-Pudlak Syndrome Type 2, an Adaptor Protein-3 Complex Disease. Molecular Medicine, 2012, 18, 56-64.	4.4	86
43	The <scp>BLOS1</scp>â€Interacting Protein <scp>KXD1</scp> is Involved in the Biogenesis of Lysosomeâ€Related Organelles. Traffic, 2012, 13, 1160-1169.	2.7	31
44	Identification, Tissue Distribution, and Molecular Modeling of Novel Human Isoforms of the Key Enzyme in Sialic Acid Synthesis, UDP-GlcNAc 2-Epimerase/ManNAc Kinase. Biochemistry, 2011, 50, 8914-8925.	2.5	26
45	Hereditary Inclusion Body Myopathy: Single Patient Response to Intravenous Dosing of GNE Gene Lipoplex. Human Gene Therapy, 2011, 22, 1331-1341.	2.7	40
46	NBEAL2 is mutated in gray platelet syndrome and is required for biogenesis of platelet Î±-granules. Nature Genetics, 2011, 43, 732-734.	21.4	223
47	Molecular Analysis of the Retinoic Acid Induced 1 Gene (RAI1) in Patients with Suspected Smith-Magenis Syndrome without the 17p11.2 Deletion. PLoS ONE, 2011, 6, e22861.	2.5	38
48	Retro-orbital injections in mice. Lab Animal, 2011, 40, 155-160.	0.4	398
49	Homozygosity Mapping and Whole-Exome Sequencing to Detect SLC45A2 and G6PC3 Mutations in a Single Patient with Oculocutaneous Albinism and Neutropenia. Journal of Investigative Dermatology, 2011, 131, 2017-2025.	0.7	64
50	Clinical, Molecular, and Cellular Features of Non-Puerto Rican Hermanskyâ€“Pudlak Syndrome Patients of Hispanic Descent. Journal of Investigative Dermatology, 2011, 131, 2394-2400.	0.7	31
51	Hereditary inclusion body myopathy: single patient response to GNE gene Lipoplex therapy. Journal of Gene Medicine, 2010, 12, 403-412.	2.8	26
52	A model of Costeff Syndrome reveals metabolic and protective functions of mitochondrial OPA3. Development (Cambridge), 2010, 137, 2587-2596.	2.5	36
53	Molecular modeling of the bifunctional enzyme UDP-GlcNAc 2-epimerase/ManNAc kinase and predictions of structural effects of mutations associated with HIBM and sialuria. Glycobiology, 2010, 20, 322-337.	2.5	22
54	Two Novel Mutations Identified in an African-American Child with Chediak-Higashi Syndrome. Case Reports in Medicine, 2010, 2010, 1-4.	0.7	10

#	Article	IF	Citations
55	OPA3, mutated in 3-methylglutaconic aciduria type III, encodes two transcripts targeted primarily to mitochondria. Molecular Genetics and Metabolism, 2010, 100, 149-154.	1.1	27
56	Novel 47.5-kb deletion in RAB27A results in severe Griscelli Syndrome Type 2. Molecular Genetics and Metabolism, 2010, 101, 62-65.	1.1	13
57	Gray platelet syndrome: natural history of a large patient cohort and locus assignment to chromosome 3p. Blood, 2010, 116, 4990-5001.	1.4	137
58	Alveolar Macrophage Dysregulation in Hermansky-Pudlak Syndrome Type 1. American Journal of Respiratory and Critical Care Medicine, 2009, 180, 1114-1121.	5.6	71
59	Hermanskyâ€“Pudlak syndrome in two Africanâ€American brothers. American Journal of Medical Genetics, Part A, 2009, 149A, 987-992.	1.2	9
60	Free sialic acid storage disease without sialuria. Annals of Neurology, 2009, 65, 753-757.	5.3	18
61	Identifying Putative Promoter Regions of Hermanskyâ€Pudlak Syndrome Genes by Means of Phylogenetic Footprinting. Annals of Human Genetics, 2009, 73, 422-428.	0.8	2
62	Hereditary Inclusion Body Myopathy: A decade of progress. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2009, 1792, 881-887.	3.8	78
63	Hermanskyâ€“Pudlak syndrome type 1 in patients of Indian descent. Molecular Genetics and Metabolism, 2009, 97, 227-233.	1.1	17
64	Recurrent rhabdomyolysis in a patient with oculocutaneous albinism type 1 and platelet storageâ€pool deficiency. American Journal of Medical Genetics, Part A, 2008, 146A, 3100-3103.	1.2	0
65	A novel mutation in a Turkish patient with Hermanskyâ€“Pudlak syndrome type 5. European Journal of Haematology, 2008, 80, 356-360.	2.2	15
66	Disorders of Lysosome-Related Organelle Biogenesis: Clinical and Molecular Genetics. Annual Review of Genomics and Human Genetics, 2008, 9, 359-386.	6.2	349
67	An immunoblotting assay to facilitate the molecular diagnosis of Hermanskyâ€“Pudlak syndrome. Molecular Genetics and Metabolism, 2008, 93, 134-144.	1.1	17
68	Alleleâ€specific silencing of the dominant disease allele in sialuria by RNA interference. FASEB Journal, 2008, 22, 3846-3852.	0.5	15
69	Platelet alpha granules in BLOC-2 and BLOC-3 subtypes of Hermansky-Pudlak syndrome. Platelets, 2007, 18, 150-157.	2.3	21
70	Analysis of Ocular Hypopigmentation inRab38^{cht/cht}Mice. , 2007, 48, 3905.		31
71	The Slc35d3 gene, encoding an orphan nucleotide sugar transporter, regulates platelet-dense granules. Blood, 2007, 109, 1533-1540.	1.4	42
72	Mutation in the key enzyme of sialic acid biosynthesis causes severe glomerular proteinuria and is rescued by N-acetylmannosamine. Journal of Clinical Investigation, 2007, 117, 1585-1594.	8.2	173

#	Article	IF	Citations
73	Improper Trafficking of Melanocyte-Specific Proteins in Hermanskyâ€“Pudlak Syndrome Type-5. Journal of Investigative Dermatology, 2007, 127, 1471-1478.	0.7	41
74	Cellular Defects in Chediakâ€“Higashi Syndrome Correlate with the Molecular Genotype and Clinical Phenotype. Journal of Investigative Dermatology, 2007, 127, 2674-2677.	0.7	64
75	Intravenous immune globulin in hereditary inclusion body myopathy: a pilot study. BMC Neurology, 2007, 7, 3.	1.8	49
76	Intestinal Disease in Hermansky-Pudlak Syndrome: Occurrence of Colitis and Relation to Genotype. Clinical Gastroenterology and Hepatology, 2006, 4, 73-80.	4.4	51
77	Normal sialylation of serum N-linked and O-GalNAc-linked glycans in hereditary inclusion-body myopathy. Molecular Genetics and Metabolism, 2006, 88, 389-390.	1.1	15
78	Ileal Crohn's disease in a woman with Hermansky-Pudlak syndrome. Gastroenterologie Clinique Et Biologique, 2006, 30, 621-624.	0.9	10
79	A new genetic isolate with a unique phenotype of syndromic oculocutaneous albinism: clinical, molecular, and cellular characteristics. Human Mutation, 2006, 27, 1158-1158.	2.5	23
80	Melanocytes Derived from Patients with Hermanskyâ€“Pudlak Syndrome Types 1, 2, and 3 Have Distinct Defects in Cargo Trafficking. Journal of Investigative Dermatology, 2005, 124, 420-427.	0.7	53
81	Association of the Hermansky-Pudlak syndrome type-3 protein with clathrin. BMC Cell Biology, 2005, 6, 33.	3.0	20
82	Use of a cell-free system to determine UDP-N-acetylglucosamine 2-epimerase and N-acetylmannosamine kinase activities in human hereditary inclusion body myopathy. Glycobiology, 2005, 15, 1102-1110.	2.5	43
83	Slc7a11 gene controls production of pheomelanin pigment and proliferation of cultured cells. Proceedings of the National Academy of Sciences of the United States of America, 2005, 102, 10964-10969.	7.1	186
84	Melanocyte-Specific Proteins Are Aberrantly Trafficked in Melanocytes of Hermansky-Pudlak Syndrome-Type 3. American Journal of Pathology, 2005, 166, 231-240.	3.8	46
85	Single nucleotide polymorphisms in the dystroglycan gene do not correlate with disease severity in hereditary inclusion body myopathy. Molecular Genetics and Metabolism, 2005, 86, 244-249.	1.1	6
86	Optic atrophies in metabolic disorders. Molecular Genetics and Metabolism, 2005, 86, 51-60.	1.1	19
87	Disease mechanisms associated with mutations of the GNE gene. Drug Discovery Today Disease Mechanisms, 2005, 2, 519-527.	0.8	13
88	Molecular Defects that Affect Platelet Dense Granules. Seminars in Thrombosis and Hemostasis, 2004, 30, 537-547.	2.7	113
89	Rab27b is Up-Regulated in Human Griscelli Syndrome Type II Melanocytes and Linked to the Actin Cytoskeleton via Exon F-Myosin Va Transcripts. Pigment Cell & Melanoma Research, 2004, 17, 498-505.	3.6	39
90	Cellular, Molecular and Clinical Characterization of Patients with Hermanskyâ€“Pudlak Syndrome Type 5. Traffic, 2004, 5, 711-722.	2.7	64

#	Article	IF	Citations
91	Hermanskyâ€“Pudlak syndrome type 4 in a patient from Sri Lanka with pulmonary fibrosis. American Journal of Medical Genetics, Part A, 2004, 127A, 201-207.	1.2	36
92	Hypoglycosylation of Î±-dystroglycan in patients with hereditary IBM due to GNE mutations. Molecular Genetics and Metabolism, 2004, 81, 196-202.	1.1	107
93	Milder ocular findings in Hermanskyâ€“Pudlak syndrome type 3 compared with Hermanskyâ€“Pudlak syndrome type 1. Ophthalmology, 2004, 111, 1599-1603.	5.2	23
94	Reduced pigmentation (rp), a mouse model of Hermansky-Pudlak syndrome, encodes a novel component of the BLOC-1 complex. Blood, 2004, 104, 3181-3189.	1.4	48
95	Hermansky-Pudlak syndrome typeÂ4 (HPS-4): clinical and molecular characteristics. Human Genetics, 2003, 113, 10-17.	3.8	138
96	Sialic acid storage disease of the Salla phenotype in American monozygous twin female sibs. American Journal of Medical Genetics Part A, 2003, 120A, 23-27.	2.4	12
97	Biochemical and molecular analyses of infantile free sialic acid storage disease in North American children. American Journal of Medical Genetics Part A, 2003, 120A, 28-33.	2.4	31
98	Cappuccino, a mouse model of Hermansky-Pudlak syndrome, encodes a novel protein that is part of the pallidin-muted complex (BLOC-1). Blood, 2003, 101, 4402-4407.	1.4	79
99	Nonsense Mutations in ADTB3A Cause Complete Deficiency of the Î²3A Subunit of Adaptor Complex-3 and Severe Hermansky-Pudlak Syndrome Type 2. Pediatric Research, 2002, 51, 150-158.	2.3	150
100	Disorders of Vesicles of Lysosomal Lineage: The Hermansky- Pudlak Syndromes. Current Molecular Medicine, 2002, 2, 451-467.	1.3	109
101	Evidence that Griscelli Syndrome with Neurological Involvement Is Caused by Mutations in RAB27A, Not MYO5A. American Journal of Human Genetics, 2002, 71, 407-414.	6.2	67
102	Hermansky-Pudlak syndrome type 1: gene organization, novel mutations, and clinical-molecular review of non-Puerto Rican cases. Human Mutation, 2002, 20, 482-482.	2.5	57
103	Hermanskyâ€“Pudlak Syndrome: Vesicle Formation from Yeast to Man. Pigment Cell & Melanoma Research, 2002, 15, 405-419.	3.6	102
104	Characterization of the Murine Gene Corresponding to Human Hermansky-Pudlak Syndrome Type 3: Exclusion of the Subtle Gray (sut) Locus. Molecular Genetics and Metabolism, 2001, 74, 217-225.	1.1	5
105	Dominant Inheritance of Sialuria, an Inborn Error of Feedback Inhibition. American Journal of Human Genetics, 2001, 68, 1419-1427.	6.2	48
106	Hermansky-Pudlak Syndrome Type 3 in Ashkenazi Jews and Other Nonâ€“Puerto Rican Patients with Hypopigmentation and Platelet Storage-Pool Deficiency. American Journal of Human Genetics, 2001, 69, 1022-1032.	6.2	90
107	Molecular cloning and characterization of human VPS18, VPS 11, VPS16, and VPS33. Gene, 2001, 264, 241-247.	2.2	83
108	Hermansky-Pudlak Syndrome and Chediak-Higashi Syndrome: Disorders of Vesicle Formation and Trafficking. Thrombosis and Haemostasis, 2001, 86, 233-245.	3.4	115

#	Article	IF	Citations
109	Mutation of a new gene causes a unique form of Hermanskyâ€“Pudlak syndrome in a genetic isolate of central Puerto Rico. Nature Genetics, 2001, 28, 376-380.	21.4	205
110	AP-3 Mediates Tyrosinase but Not TRP-1 Trafficking in Human Melanocytes. Molecular Biology of the Cell, 2001, 12, 2075-2085.	2.1	138
111	Pulmonary Function and High-Resolution CT Findings in Patients With an Inherited Form of Pulmonary Fibrosis, Hermansky-Pudlak Syndrome, Due to Mutations in HPS-1. Chest, 2000, 117, 129-136.	0.8	169
112	Hermansky-Pudlak Syndrome and Related Disorders of Organelle Formation. Traffic, 2000, 1, 823-835.	2.7	135
113	Ocular Nonnephropathic Cystinosis: Clinical, Biochemical, and Molecular Correlations. Pediatric Research, 2000, 47, 17-17.	2.3	103
114	Identification and Detection of the Common 65-kb Deletion Breakpoint in the Nephropathic Cystinosis Gene (CTNS). Molecular Genetics and Metabolism, 1999, 66, 111-116.	1.1	81
115	Sialuria in a Portuguese Girl: Clinical, Biochemical, and Molecular Characteristics. Molecular Genetics and Metabolism, 1999, 67, 131-137.	1.1	20