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List of Publications by Year in descending order

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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	Retro-orbital injections in mice. Lab Animal, 2011, 40, 155-160.	0.4	398
2	Disorders of Lysosome-Related Organelle Biogenesis: Clinical and Molecular Genetics. Annual Review of Genomics and Human Genetics, 2008, 9, 359-386.	6.2	349
3	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
4	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
5	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
6	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
7	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
8	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
9	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
10	AP-3 Mediates Tyrosinase but Not TRP-1 Trafficking in Human Melanocytes. Molecular Biology of the Cell, 2001, 12, 2075-2085.	2.1	138
11	Hermansky-Pudlak syndrome typeÂ4 (HPS-4): clinical and molecular characteristics. Human Genetics, 2003, 113, 10-17.	3.8	138
12	Gray platelet syndrome: natural history of a large patient cohort and locus assignment to chromosome 3p. Blood, 2010, 116, 4990-5001.	1.4	137
13	Hermansky-Pudlak Syndrome and Related Disorders of Organelle Formation. Traffic, 2000, 1, 823-835.	2.7	135
14	Hermansky-Pudlak Syndrome and Chediak-Higashi Syndrome: Disorders of Vesicle Formation and Trafficking. Thrombosis and Haemostasis, 2001, 86, 233-245.	3.4	115
15	Molecular Defects that Affect Platelet Dense Granules. Seminars in Thrombosis and Hemostasis, 2004, 30, 537-547.	2.7	113
16	Disorders of Vesicles of Lysosomal Lineage: The Hermansky- Pudlak Syndromes. Current Molecular Medicine, 2002, 2, 451-467.	1.3	109
17	Hypoglycosylation of Î±-dystroglycan in patients with hereditary IBM due to GNE mutations. Molecular Genetics and Metabolism, 2004, 81, 196-202.	1.1	107
18	Ocular Nonnephropathic Cystinosis: Clinical, Biochemical, and Molecular Correlations. Pediatric Research, 2000, 47, 17-17.	2.3	103

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19	Hermanskyâ€“Pudlak Syndrome: Vesicle Formation from Yeast to Man. Pigment Cell & Melanoma Research, 2002, 15, 405-419.	3.6	102
20	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
21	Mutation Update forGNEGene Variants Associated with GNE Myopathy. Human Mutation, 2014, 35, 915-926.	2.5	90
22	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
23	Molecular cloning and characterization of human VPS18, VPS 11, VPS16, and VPS33. Gene, 2001, 264, 241-247.	2.2	83
24	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
25	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
26	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
27	Hereditary Inclusion Body Myopathy: A decade of progress. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2009, 1792, 881-887.	3.8	78
28	Alveolar Macrophage Dysregulation in Hermansky-Pudlak Syndrome Type 1. American Journal of Respiratory and Critical Care Medicine, 2009, 180, 1114-1121.	5.6	71
29	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
30	Hermanskyâ€“Pudlak syndrome: Mutation update. Human Mutation, 2020, 41, 543-580.	2.5	65
31	Cellular, Molecular and Clinical Characterization of Patients with Hermanskyâ€“Pudlak Syndrome Type 5. Traffic, 2004, 5, 711-722.	2.7	64
32	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
33	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
34	GNE Myopathy: Etiology, Diagnosis, and Therapeutic Challenges. Neurotherapeutics, 2018, 15, 900-914.	4.4	63
35	GNE myopathy: New name and new mutation nomenclature. Neuromuscular Disorders, 2014, 24, 387-389.	0.6	61
36	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

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37	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
38	Intestinal Disease in Hermansky-Pudlak Syndrome: Occurrence of Colitis and Relation to Genotype. Clinical Gastroenterology and Hepatology, 2006, 4, 73-80.	4.4	51
39	Intravenous immune globulin in hereditary inclusion body myopathy: a pilot study. BMC Neurology, 2007, 7, 3.	1.8	49
40	Dominant Inheritance of Sialuria, an Inborn Error of Feedback Inhibition. American Journal of Human Genetics, 2001, 68, 1419-1427.	6.2	48
41	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
42	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
43	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
44	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
45	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
46	The Slc35d3 gene, encoding an orphan nucleotide sugar transporter, regulates platelet-dense granules. Blood, 2007, 109, 1533-1540.	1.4	42
47	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
48	Improper Trafficking of Melanocyte-Specific Proteins in Hermanskyâ€“Pudlak Syndrome Type-5. Journal of Investigative Dermatology, 2007, 127, 1471-1478.	0.7	41
49	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
50	Hereditary Inclusion Body Myopathy: Single Patient Response to Intravenous Dosing of GNE Gene Lipoplex. Human Gene Therapy, 2011, 22, 1331-1341.	2.7	40
51	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
52	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
53	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
54	A model of Costeff Syndrome reveals metabolic and protective functions of mitochondrial OPA3. Development (Cambridge), 2010, 137, 2587-2596.	2.5	36

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55	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
56	Analysis of Ocular Hypopigmentation inRab38^{cht/cht}Mice. , 2007, 48, 3905.		31
57	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
58	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
59	Mutations in KIAA0753 cause Joubert syndrome associated with growth hormone deficiency. Human Genetics, 2017, 136, 399-408.	3.8	30
60	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
61	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
62	The Gne M712T Mouse as a Model for Human Glomerulopathy. American Journal of Pathology, 2012, 180, 1431-1440.	3.8	27
63	Hereditary inclusion body myopathy: single patient response to GNE gene Lipoplex therapy. Journal of Gene Medicine, 2010, 12, 403-412.	2.8	26
64	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
65	Cellular and clinical report of new Griscelli syndrome type III cases. Pigment Cell and Melanoma Research, 2012, 25, 47-56.	3.3	25
66	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
67	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
68	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
69	Milder ocular findings in Hermanskyâ€“Pudlak syndrome type 3 compared with Hermanskyâ€“Pudlak syndrome type 1. Ophthalmology, 2004, 111, 1599-1603.	5.2	23
70	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
71	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
72	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

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73	Exome analysis of Smithâ€“Magenis-like syndrome cohort identifies de novo likely pathogenic variants. Human Genetics, 2017, 136, 409-420.	3.8	22
74	Platelet alpha granules in BLOC-2 and BLOC-3 subtypes of Hermansky-Pudlak syndrome. Platelets, 2007, 18, 150-157.	2.3	21
75	Atypical presentation of GNE myopathy with asymmetric hand weakness. Neuromuscular Disorders, 2014, 24, 1063-1067.	0.6	21
76	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
77	Neurologic involvement in patients with atypical Chediak-Higashi disease. Neurology, 2016, 86, 1320-1328.	1.1	21
78	Sialuria in a Portuguese Girl: Clinical, Biochemical, and Molecular Characteristics. Molecular Genetics and Metabolism, 1999, 67, 131-137.	1.1	20
79	Association of the Hermansky-Pudlak syndrome type-3 protein with clathrin. BMC Cell Biology, 2005, 6, 33.	3.0	20
80	Neurologic involvement in patients with atypical Chediak-Higashi disease. Neurology, 2017, 88, e57-e65.	1.1	20
81	Optic atrophies in metabolic disorders. Molecular Genetics and Metabolism, 2005, 86, 51-60.	1.1	19
82	Sialylation of Thomsenâ€“Friedenreich antigen is a noninvasive blood-based biomarker for GNE myopathy. Biomarkers in Medicine, 2014, 8, 641-652.	1.4	19
83	Free sialic acid storage disease without sialuria. Annals of Neurology, 2009, 65, 753-757.	5.3	18
84	Inherited disorders of lysosomal membrane transporters. Biochimica Et Biophysica Acta - Biomembranes, 2020, 1862, 183336.	2.6	18
85	An immunoblotting assay to facilitate the molecular diagnosis of Hermanskyâ€“Pudlak syndrome. Molecular Genetics and Metabolism, 2008, 93, 134-144.	1.1	17
86	Hermanskyâ€“Pudlak syndrome type 1 in patients of Indian descent. Molecular Genetics and Metabolism, 2009, 97, 227-233.	1.1	17
87	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
88	A novel mutation in a Turkish patient with Hermanskyâ€“Pudlak syndrome type 5. European Journal of Haematology, 2008, 80, 356-360.	2.2	15
89	Alleleâ€specific silencing of the dominant disease allele in sialuria by RNA interference. FASEB Journal, 2008, 22, 3846-3852.	0.5	15
90	Disease mechanisms associated with mutations of the GNE gene. Drug Discovery Today Disease Mechanisms, 2005, 2, 519-527.	0.8	13

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91	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
92	Quantification of lectin fluorescence in GNE myopathy muscle biopsies. Muscle and Nerve, 2018, 58, 286-292.	2.2	13
93	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
94	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
95	Genetic variants associated with Hermansky-Pudlak syndrome. Platelets, 2020, 31, 544-547.	2.3	12
96	Free sialic acid storage disorder: Progress and promise. Neuroscience Letters, 2021, 755, 135896.	2.1	12
97	Nonâ€specific accumulation of glycosphingolipids in GNE myopathy. Journal of Inherited Metabolic Disease, 2014, 37, 297-308.	3.6	11
98	Ileal Crohn's disease in a woman with Hermansky-Pudlak syndrome. Gastroenterologie Clinique Et Biologique, 2006, 30, 621-624.	0.9	10
99	Two Novel Mutations Identified in an African-American Child with Chediak-Higashi Syndrome. Case Reports in Medicine, 2010, 2010, 1-4.	0.7	10
100	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
101	Hermanskyâ€“Pudlak syndrome in two Africanâ€American brothers. American Journal of Medical Genetics, Part A, 2009, 149A, 987-992.	1.2	9
102	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
103	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
104	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
105	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
106	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
107	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
108	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

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109	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
110	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
111	New observation of sialuria prompts detection of liver tumor in previously reported patient. Molecular Genetics and Metabolism, 2016, 118, 92-99.	1.1	4
112	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
113	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
114	Elevated Plasma Free Sialic Acid Levels in Individuals with Reduced Glomerular Filtration Rates. Kidney360, 2020, 1, 957-961.	2.1	2
115	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