Priya S Kishnani

List of Publications by Year
in descending order

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268 papers	11,297 citations	30070 54 h-index	42399 92 g-index
277 all docs	277 docs citations	277 times ranked	6060 citing authors

#	Article	IF	Citations
1	A Randomized Study of Alglucosidase Alfa in Late-Onset Pompe's Disease. New England Journal of Medicine, 2010, 362, 1396-1406.	27.0	674
2	A retrospective, multinational, multicenter study on the natural history of infantile-onset Pompe disease. Journal of Pediatrics, 2006, 148, 671-676.e2.	1.8	500
3	Pompe disease diagnosis and management guideline. Genetics in Medicine, 2006, 8, 267-288.	2.4	473
4	Cross-reactive immunologic material status affects treatment outcomes in Pompe disease infants. Molecular Genetics and Metabolism, 2010, 99, 26-33.	1.1	348
5	Early Treatment With Alglucosidase Alfa Prolongs Long-Term Survival of Infants With Pompe Disease. Pediatric Research, 2009, 66, 329-335.	2.3	277
6	Recombinant human acid Î±-glucosidase enzyme therapy for infantile glycogen storage disease type II: Results of a phase I/II clinical trial. Genetics in Medicine, 2001, 3, 132-138.	2.4	276
7	Clinical outcomes after long-term treatment with alglucosidase alfa in infants and children with advanced Pompe disease. Genetics in Medicine, 2009, 11, 210-219.	2.4	259
8	Glycogen Storage Disease Type III diagnosis and management guidelines. Genetics in Medicine, 2010, 12, 446-463.	2.4	236
9	Characterization of pre- and post-treatment pathology after enzyme replacement therapy for pompe disease. Laboratory Investigation, 2006, 86, 1208-1220.	3.7	226
10	The impact of antibodies on clinical outcomes in diseases treated with therapeutic protein: Lessons learned from infantile Pompe disease. Genetics in Medicine, 2011, 13, 729-736.	2.4	216
11	Successful immune tolerance induction to enzyme replacement therapy in CRIM-negative infantile Pompe disease. Genetics in Medicine, 2012, 14, 135-142.	2.4	183
12	The emerging phenotype of long-term survivors with infantile Pompe disease. Genetics in Medicine, 2012, 14, 800-810.	2.4	163
13	The emerging phenotype of late-onset Pompe disease: A systematic literature review. Molecular Genetics and Metabolism, 2017, 120, 163-172.	1.1	140
14	Elimination of Antibodies to Recombinant Enzyme in Pompe's Disease. New England Journal of Medicine, 2009, 360, 194-195.	27.0	136
15	Pompe disease: Design, methodology, and early findings from the Pompe Registry. Molecular Genetics and Metabolism, 2011, 103, 1-11.	1.1	130
16	Glycogen storage disease type III-hepatocellular carcinoma a long-term complication?. Journal of Hepatology, 2007, 46, 492-498.	3.7	124
17	Enhanced Response to Enzyme Replacement Therapy in Pompe Disease after the Induction of Immune Tolerance. American Journal of Human Genetics, 2007, 81, 1042-1049.	6.2	118
18	Predicting crossâ€reactive immunological material (CRIM) status in Pompe disease using GAA mutations: Lessons learned from 10 years of clinical laboratory testing experience. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2012, 160C, 40-49.	1.6	110

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19	Liver transplantation for pediatric metabolic disease. Molecular Genetics and Metabolism, 2014, 111, 418-427.	1.1	105
20	Targeted long-read sequencing identifies missing disease-causing variation. American Journal of Human Genetics, 2021, 108, 1436-1449.	6.2	105
21	Deconstructing Pompe Disease by Analyzing Single Muscle Fibers: â€œTo See a World in a Grain of Sandâ€¦â€• Autophagy, 2007, 3, 546-552.	9.1	102
22	Five-year efficacy and safety of asfotase alfa therapy for adults and adolescents with hypophosphatasia. Bone, 2019, 121, 149-162.	2.9	99
23	Open-label extension study following the Late-Onset Treatment Study (LOTS) of alglucosidase alfa. Molecular Genetics and Metabolism, 2012, 107, 456-461.	1.1	93
24	Algorithm for the Early Diagnosis and Treatment of Patients with Cross Reactive Immunologic Material-Negative Classic Infantile Pompe Disease: A Step towards Improving the Efficacy of ERT. PLoS ONE, 2013, 8, e67052.	2.5	93
25	AAV Vector-mediated Reversal of Hypoglycemia in Canine and Murine Glycogen Storage Disease Type Ia. Molecular Therapy, 2008, 16, 665-672.	8.2	85
26	Monitoring guidance for patients with hypophosphatasia treated with asfotase alfa. Molecular Genetics and Metabolism, 2017, 122, 4-17.	1.1	84
27	Nephrotic Syndrome Complicating Â-Glucosidase Replacement Therapy for Pompe Disease. Pediatrics, 2004, 114, e532-e535.	2.1	83
28	Diagnosis and management of glycogen storage diseases type VI and IX: a clinical practice resource of the American College of Medical Genetics and Genomics (ACMG). Genetics in Medicine, 2019, 21, 772-789.	2.4	81
29	Oropharyngeal Dysphagia in Infants and Children with Infantile Pompe Disease. Dysphagia, 2010, 25, 277-283.	1.8	80
30	Immunomodulatory Gene Therapy Prevents Antibody Formation and Lethal Hypersensitivity Reactions in Murine Pompe Disease. Molecular Therapy, 2010, 18, 353-360.	8.2	80
31	Glucose tetrasaccharide as a biomarker for monitoring the therapeutic response to enzyme replacement therapy for Pompe disease. Molecular Genetics and Metabolism, 2005, 85, 247-254.	1.1	79
32	Long-term monitoring of patients with infantile-onset Pompe disease on enzyme replacement therapy using a urinary glucose tetrasaccharide biomarker. Genetics in Medicine, 2009, 11, 536-541.	2.4	79
33	Autopsy findings in late-onset Pompe disease: A case report and systematic review of the literature. Molecular Genetics and Metabolism, 2012, 106, 462-469.	1.1	76
34	Pompe Disease Results in a Golgi-based Glycosylation Deficit in Human Induced Pluripotent Stem Cell-derived Cardiomyocytes. Journal of Biological Chemistry, 2015, 290, 3121-3136.	3.4	76
35	Bortezomib in the rapid reduction of high sustained antibody titers in disorders treated with therapeutic protein: lessons learned from Pompe disease. Genetics in Medicine, 2013, 15, 123-131.	2.4	75
36	Comparison of maltose and acarbose as inhibitors of maltase-glucoamylase activity in assaying acid Î±-glucosidase activity in dried blood spots for the diagnosis of infantile Pompe disease. Genetics in Medicine, 2006, 8, 302-306.	2.4	74

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37	Donepezil for treatment of cognitive dysfunction in children with Down syndrome aged 10â€“17. American Journal of Medical Genetics, Part A, 2010, 152A, 3028-3035.	1.2	71
38	Screening for pompe disease using a rapid dried blood spot method: Experience of a clinical diagnostic laboratory. Muscle and Nerve, 2009, 40, 32-36.	2.2	69
39	Diagnostic delay is common among patients with hypophosphatasia: initial findings from a longitudinal, prospective, global registry. BMC Musculoskeletal Disorders, 2019, 20, 80.	1.9	69
40	The new era of Pompe disease: Advances in the detection, understanding of the phenotypic spectrum, pathophysiology, and management. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2012, 160C, 1-7.	1.6	68
41	Burden of disease in adult patients with hypophosphatasia: Results from two patient-reported surveys. Metabolism: Clinical and Experimental, 2016, 65, 1522-1530.	3.4	68
42	Infantile Pompe disease on ERTâ€”Update on clinical presentation, musculoskeletal management, and exercise considerations. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2012, 160C, 69-79.	1.6	67
43	The impact of antibodies in late-onset Pompe disease: A case series and literature review. Molecular Genetics and Metabolism, 2012, 106, 301-309.	1.1	66
44	Immune response to enzyme replacement therapies in lysosomal storage diseases and the role of immune tolerance induction. Molecular Genetics and Metabolism, 2016, 117, 66-83.	1.1	64
45	Benzoate therapy and carnitine deficiency in non-ketotic hyperglycinemia. American Journal of Medical Genetics Part A, 1995, 59, 444-453.	2.4	63
46	Chromosomal and genetic alterations in human hepatocellular adenomas associated with type Ia glycogen storage disease. Human Molecular Genetics, 2009, 18, 4781-4790.	2.9	63
47	Expanding the phenotype of lateâ€onset pompe disease: Tongue weakness: A new clinical observation. Muscle and Nerve, 2011, 44, 897-901.	2.2	63
48	Assessing disease severity in Pompe disease: The roles of a urinary glucose tetrasaccharide biomarker and imaging techniques. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2012, 160C, 50-58.	1.6	60
49	Timing of diagnosis of patients with pompe disease: Data from the pompe registry. American Journal of Medical Genetics, Part A, 2013, 161, 2431-2443.	1.2	60
50	Persistence of high sustained antibodies to enzyme replacement therapy despite extensive immunomodulatory therapy in an infant with Pompe disease: Need for agents to target antibody-secreting plasma cells. Molecular Genetics and Metabolism, 2012, 105, 677-680.	1.1	59
51	Eliglustat maintains long-term clinical stability in patients with Gaucher disease type 1 stabilized on enzyme therapy. Blood, 2017, 129, 2375-2383.	1.4	59
52	Safety and efficacy of avalglucosidase alfa versus alglucosidase alfa in patients with late-onset Pompe disease (COMET): a phase 3, randomised, multicentre trial. Lancet Neurology, The, 2021, 20, 1012-1026.	10.2	59
53	Skeletal muscle pathology of infantile Pompe disease during long-term enzyme replacement therapy. Orphanet Journal of Rare Diseases, 2013, 8, 90.	2.7	58
54	Recommendations for the use of eliglustat in the treatment of adults with Gaucher disease type 1 in the United States. Molecular Genetics and Metabolism, 2016, 117, 95-103.	1.1	57

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55	The value of muscle biopsies in Pompe disease: identifying lipofuscin inclusions in juvenile- and adult-onset patients. Acta Neuropathologica Communications, 2014, 2, 2.	5.2	55
56	Velaglucerase alfa enzyme replacement therapy compared with imiglucerase in patients with Gaucher disease. American Journal of Hematology, 2013, 88, 179-184.	4.1	54
57	CRIM-negative infantile Pompe disease: characterization of immune responses in patients treated with ERT monotherapy. Genetics in Medicine, 2015, 17, 912-918.	2.4	54
58	Rapid diagnosis of late-onset Pompe disease by fluorometric assay of Î±-glucosidase activities in dried blood spots. Molecular Genetics and Metabolism, 2007, 90, 449-452.	1.1	53
59	Diagnostic challenges for Pompe disease: An under-recognized cause of floppy baby syndrome. Genetics in Medicine, 2006, 8, 289-296.	2.4	52
60	The clinical and electrodiagnostic characteristics of Pompe disease with post-enzyme replacement therapy findings. Clinical Neurophysiology, 2011, 122, 2312-2317.	1.5	52
61	Further understanding the connection between Alzheimer's disease and Down syndrome. Alzheimer's and Dementia, 2020, 16, 1065-1077.	0.8	52
62	GAA variants and phenotypes among 1,079 patients with Pompe disease: Data from the Pompe Registry. Human Mutation, 2019, 40, 2146-2164.	2.5	51
63	Treatment of pyruvate carboxylase deficiency with high doses of citrate and aspartate. American Journal of Medical Genetics Part A, 1999, 87, 331-338.	2.4	49
64	Variability of disease spectrum in children with liver phosphorylase kinase deficiency caused by mutations in the PHKG2 gene. Molecular Genetics and Metabolism, 2014, 111, 309-313.	1.1	48
65	Oropharyngeal dysphagia may occur in late-onset Pompe disease, implicating bulbar muscle involvement. Neuromuscular Disorders, 2013, 23, 319-323.	0.6	47
66	Sustained immune tolerance induction in enzyme replacement therapyâ€“treated CRIM-negative patients with infantile Pompe disease. JCI Insight, 2017, 2, .	5.0	47
67	The efficacy, safety, and tolerability of donepezil for the treatment of young adults with Down syndrome. American Journal of Medical Genetics, Part A, 2009, 149A, 1641-1654.	1.2	46
68	Improvement of Bilateral Ptosis on Higher Dose Enzyme Replacement Therapy in Pompe Disease. Journal of Neuro-Ophthalmology, 2010, 30, 165-166.	0.8	46
69	Neutropenia in glycogen storage disease Ib: outcomes for patients treated with granulocyte colony-stimulating factor. Current Opinion in Hematology, 2019, 26, 16-21.	2.5	46
70	Expanding the clinical spectrum of late-onset Pompe disease: Dilated arteriopathy involving the thoracic aorta, a novel vascular phenotype uncovered. Molecular Genetics and Metabolism, 2011, 103, 362-366.	1.1	44
71	The prevalence and impact of scoliosis in Pompe disease: Lessons learned from the Pompe Registry. Molecular Genetics and Metabolism, 2011, 104, 574-582.	1.1	44
72	Anaesthetic management of infants with glycogen storage disease type II: a physiological approach. Paediatric Anaesthesia, 2004, 14, 514-519.	1.1	43

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73	Safety and efficacy of alternative alglucosidase alfa regimens in Pompe disease. Neuromuscular Disorders, 2015, 25, 321-332.	0.6	43
74	Case series: Odontohypophosphatasia or missed diagnosis of childhood/adult-onset hypophosphatasia? â€“ Call for a long-term follow-up of premature loss of primary teeth. Bone Reports, 2016, 5, 228-232.	0.4	43
75	Management of Confirmed Newborn-Screened Patients With Pompe Disease Across the Disease Spectrum. Pediatrics, 2017, 140, S24-S45.	2.1	43
76	Diagnosis of late-onset Pompe disease and other muscle disorders by next-generation sequencing. Orphanet Journal of Rare Diseases, 2016, 11, 8.	2.7	42
77	An emerging phenotype of central nervous system involvement in Pompe disease: from bench to bedside and beyond. Annals of Translational Medicine, 2019, 7, 289-289.	1.7	42
78	Safety and efficacy of cipaglucosidase alfa plus miglustat versus alglucosidase alfa plus placebo in late-onset Pompe disease (PROPEL): an international, randomised, double-blind, parallel-group, phase 3 trial. Lancet Neurology, The, 2021, 20, 1027-1037.	10.2	42
79	Clinical and Histologic Ocular Findings in Pompe Disease. Journal of Pediatric Ophthalmology and Strabismus, 2010, 47, 34-40.	0.7	41
80	Molecular analysis of the AGL gene: Identification of 25 novel mutations and evidence of genetic heterogeneity in patients with Glycogen Storage Disease Type III. Genetics in Medicine, 2010, 12, 424-430.	2.4	41
81	Gene therapy for glycogen storage diseases. Human Molecular Genetics, 2019, 28, R31-R41.	2.9	40
82	Higher dosing of alglucosidase alfa improves outcomes in children with Pompe disease: a clinical study and review of the literature. Genetics in Medicine, 2020, 22, 898-907.	2.4	40
83	Cardiovascular abnormalities in late-onset Pompe disease and response to enzyme replacement therapy. Genetics in Medicine, 2011, 13, 625-631.	2.4	39
84	Correlation between quantitative wholeâ€body muscle magnetic resonance imaging and clinical muscle weakness in pompe disease. Muscle and Nerve, 2015, 51, 722-730.	2.2	39
85	Neuroimaging findings in infantile Pompe patients treated with enzyme replacement therapy. Molecular Genetics and Metabolism, 2018, 123, 85-91.	1.1	39
86	Methods of diagnosis of patients with Pompe disease: Data from the Pompe Registry. Molecular Genetics and Metabolism, 2014, 113, 84-91.	1.1	38
87	Starch Binding Domain-containing Protein 1 Plays a Dominant Role in Glycogen Transport to Lysosomes in Liver. Journal of Biological Chemistry, 2016, 291, 16479-16484.	3.4	38
88	Intravenous Injection of an AAV-PHP.B Vector Encoding Human Acid Î±-Glucosidase Rescues Both Muscle and CNS Defects in Murine Pompe Disease. Molecular Therapy - Methods and Clinical Development, 2019, 12, 233-245.	4.1	38
89	Burden of Illness in Adults With Hypophosphatasia: Data From the Global Hypophosphatasia Patient Registry. Journal of Bone and Mineral Research, 2020, 35, 2171-2178.	2.8	38
90	Immunological challenges and approaches to immunomodulation in Pompe disease: a literature review. Annals of Translational Medicine, 2019, 7, 285-285.	1.7	38

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91	Pharmacological interventions to improve cognition and adaptive functioning in Down syndrome: Strides to date. American Journal of Medical Genetics, Part A, 2017, 173, 3029-3041.	1.2	37
92	Correction of glycogen storage disease type III with rapamycin in a canine model. Journal of Molecular Medicine, 2014, 92, 641-650.	3.9	36
93	Duvoglustat HCl Increases Systemic and Tissue Exposure of Active Acid Î±-Glucosidase in Pompe Patients Co-administered with Alglucosidase Î±. Molecular Therapy, 2017, 25, 1199-1208.	8.2	36
94	Unexplained regression in Down syndrome: 35 cases from an international Down syndrome database. Genetics in Medicine, 2020, 22, 767-776.	2.4	36
95	Transformation in pretreatment manifestations of Gaucher disease type 1 during two decades of alglucerase/imiglucerase enzyme replacement therapy in the International Collaborative Gaucher Group (ICGG) Gaucher Registry. American Journal of Hematology, 2017, 92, 929-939.	4.1	35
96	The electrodiagnostic characteristics of Glycogen Storage Disease Type III. Genetics in Medicine, 2010, 12, 440-445.	2.4	34
97	Characterization of a canine model of glycogen storage disease type IIIa. DMM Disease Models and Mechanisms, 2012, 5, 804-11.	2.4	34
98	Postmortem Findings and Clinical Correlates in Individuals with Infantile-Onset Pompe Disease. JIMD Reports, 2015, 23, 45-54.	1.5	34
99	Thyroid dysfunction in patients with Down syndrome: Results from a multiâ€institutional registry study. American Journal of Medical Genetics, Part A, 2017, 173, 1539-1545.	1.2	34
100	Improvement with ongoing Enzyme Replacement Therapy in advanced late-onset Pompe disease: A case study. Molecular Genetics and Metabolism, 2008, 95, 233-235.	1.1	33
101	Echocardiographic manifestations of Glycogen Storage Disease III: Increase in wall thickness and left ventricular mass over time. Genetics in Medicine, 2010, 12, 413-423.	2.4	33
102	Increased inspiratory and expiratory muscle strength following respiratory muscle strength training (RMST) in two patients with late-onset Pompe disease. Molecular Genetics and Metabolism, 2011, 104, 417-420.	1.1	33
103	Respiratory muscle training (RMT) in late-onset Pompe disease (LOPD): Effects of training and detraining. Molecular Genetics and Metabolism, 2016, 117, 120-128.	1.1	31
104	Immunotherapy in selected patients with Down syndrome disintegrative disorder. Developmental Medicine and Child Neurology, 2019, 61, 847-851.	2.1	31
105	Hemangioma, supraumbilical midline raphÃ©, and coarctation of the aorta with a right aortic arch: Single causal entity?. American Journal of Medical Genetics Part A, 1995, 59, 44-48.	2.4	30
106	Atypical immunologic response in a patient with CRIM-negative Pompe disease. Molecular Genetics and Metabolism, 2011, 104, 583-586.	1.1	29
107	Baseline Urinary Glucose Tetrasaccharide Concentrations in Patients with Infantile- and Late-Onset Pompe Disease Identified by Newborn Screening. JIMD Reports, 2014, 19, 67-73.	1.5	29
108	Cognitive and academic outcomes in long-term survivors of infantile-onset Pompe disease: A longitudinal follow-up. Molecular Genetics and Metabolism, 2017, 121, 127-137.	1.1	28

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109	Liver fibrosis during clinical ascertainment of glycogen storage disease type III: a need for improved and systematic monitoring. Genetics in Medicine, 2019, 21, 2686-2694.	2.4	28
110	An immune tolerance approach using transient low-dose methotrexate in the ERT-naÃ¨ve setting of patients treated with a therapeutic protein: experience in infantile-onset Pompe disease. Genetics in Medicine, 2019, 21, 887-895.	2.4	28
111	Immunomodulatory Gene Therapy in Lysosomal Storage Disorders. Current Gene Therapy, 2009, 9, 503-510.	2.0	27
112	Safety, efficacy, and authorization of eliglustat as a first-line therapy in Gaucher disease type 1. Blood Cells, Molecules, and Diseases, 2018, 71, 71-74.	1.4	27
113	New therapeutic approaches for Pompe disease: enzyme replacement therapy and beyond. Pediatric Endocrinology Reviews, 2014, 12 Suppl 1, 114-24.	1.2	27
114	Insight into the phenotype of infants with Pompe disease identified by newborn screening with the common c.-32-13T > G â€œlate-onsetâ€•GAA variant. Molecular Genetics and Metabolism, 2017, 122, 99-107.	1.1	26
115	The Initial Evaluation of Patients After Positive Newborn Screening: Recommended Algorithms Leading to a Confirmed Diagnosis of Pompe Disease. Pediatrics, 2017, 140, S14-S23.	2.1	26
116	Correction of Biochemical Abnormalities and Improved Muscle Function in a Phase I/II Clinical Trial of Clenbuterol in Pompe Disease. Molecular Therapy, 2018, 26, 2304-2314.	8.2	26
117	Transforming the clinical outcome in CRIM-negative infantile Pompe disease identified via newborn screening: the benefits of early treatment with enzyme replacement therapy and immune tolerance induction. Genetics in Medicine, 2021, 23, 845-855.	2.4	26
118	Realâ€•world effectiveness of eliglustat in treatmentâ€•naÃ¨ve and switch patients enrolled in the International Collaborative Gaucher Group Gaucher Registry. American Journal of Hematology, 2020, 95, 1038-1046.	4.1	26
119	Prevalence of Iron Deficiency in Children with Down Syndrome. Journal of Pediatrics, 2010, 157, 967-971.e1.	1.8	25
120	Molecular analysis and protein processing in lateâ€•onset pompe disease patients with low levels of acid Î±â€•glucosidase activity. Muscle and Nerve, 2011, 43, 665-670.	2.2	25
121	Quantitative assessment of lingual strength in lateâ€•onset Pompe disease. Muscle and Nerve, 2015, 51, 731-735.	2.2	25
122	Characterization of immune response in Cross-Reactive Immunological Material (CRIM)-positive infantile Pompe disease patients treated with enzyme replacement therapy. Molecular Genetics and Metabolism Reports, 2019, 20, 100475.	1.1	25
123	Gaucher disease and SARS-CoV-2 infection: Emerging management challenges. Molecular Genetics and Metabolism, 2020, 130, 164-169.	1.1	25
124	Immune modulation in Pompe disease treated with enzyme replacement therapy. Expert Review of Clinical Immunology, 2012, 8, 497-499.	3.0	24
125	Immune tolerance strategies in siblings with infantile Pompe disease â€" Advantages for a preemptive approach to high-sustained antibody titers. Molecular Genetics and Metabolism Reports, 2015, 4, 30-34.	1.1	24
126	A pilot study on using rapamycin-carrying synthetic vaccine particles (SVP) in conjunction with enzyme replacement therapy to induce immune tolerance in Pompe disease. Molecular Genetics and Metabolism Reports, 2017, 13, 18-22.	1.1	24

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127	Small-Fiber Neuropathy in Pompe Disease: First Reported Cases and Prospective Screening of a Clinic Cohort. American Journal of Case Reports, 2015, 16, 196-201.	0.8	24
128	Ambulatory electrocardiogram analysis in infants treated with recombinant human acid Î±-glucosidase enzyme replacement therapy for Pompe disease. Genetics in Medicine, 2006, 8, 313-317.	2.4	23
129	Arrhythmias in patients receiving enzyme replacement therapy for infantile Pompe disease. Genetics in Medicine, 2008, 10, 758-762.	2.4	23
130	Follow-up of a child with pyruvate dehydrogenase deficiency on a less restrictive ketogenic diet. Molecular Genetics and Metabolism, 2011, 102, 214-215.	1.1	23
131	Cognitive and adaptive functioning of children with infantile Pompe disease treated with enzyme replacement therapy: Longâ€term followâ€up. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2012, 160C, 22-29.	1.6	23
132	Antibody-mediated enzyme replacement therapy targeting both lysosomal and cytoplasmic glycogen in Pompe disease. Journal of Molecular Medicine, 2017, 95, 513-521.	3.9	23
133	Clinical trial of Lâ€Carnitine and valproic acid in spinal muscular atrophy type I. Muscle and Nerve, 2018, 57, 193-199.	2.2	23
134	Dietary lipids in glycogen storage disease type III: A systematic literature study, case studies, and future recommendations. Journal of Inherited Metabolic Disease, 2020, 43, 770-777.	3.6	23
135	Ocular and Histologic Findings in a Series of Children With Infantile Pompe Disease Treated With Enzyme Replacement Therapy. Journal of Pediatric Ophthalmology and Strabismus, 2014, 51, 355-362.	0.7	23
136	Dysregulation of Multiple Facets of Glycogen Metabolism in a Murine Model of Pompe Disease. PLoS ONE, 2013, 8, e56181.	2.5	22
137	Effects of respiratory muscle training (RMT) in children with infantile-onset Pompe disease and respiratory muscle weakness. Journal of Pediatric Rehabilitation Medicine, 2014, 7, 255-265.	0.5	22
138	Efficacy, safety profile, and immunogenicity of alglucosidase alfa produced at the 4,000-liter scale in US children and adolescents with Pompe disease: ADVANCE, a phase IV, open-label, prospective study. Genetics in Medicine, 2018, 20, 1284-1294.	2.4	22
139	Clinical and Molecular Disease Spectrum and Outcomes in Patients with Infantile-Onset Pompe Disease. Journal of Pediatrics, 2020, 216, 44-50.e5.	1.8	22
140	Benefits of Prophylactic Short-Course Immune Tolerance Induction in Patients With Infantile Pompe Disease: Demonstration of Long-Term Safety and Efficacy in an Expanded Cohort. Frontiers in Immunology, 2020, 11, 1727.	4.8	22
141	How common is misdiagnosis in late-onset pompe disease?. Muscle and Nerve, 2012, 45, 301-302.	2.2	21
142	Sensitivity of whole exome sequencing in detecting infantile- and late-onset Pompe disease. Molecular Genetics and Metabolism, 2017, 122, 189-197.	1.1	21
143	Severe Cardiac Involvement Is Rare in Patients with Late-Onset Pompe Disease and the Common c.-32-13T>G Variant: Implications for Newborn Screening. Journal of Pediatrics, 2018, 198, 308-312.	1.8	21
144	Severe Cardiomyopathy as the Isolated Presenting Feature in an Adult with Late-Onset Pompe Disease: A Case Report. JIMD Reports, 2016, 31, 79-83.	1.5	20

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145	Role of continuous glucose monitoring in the management of glycogen storage disorders. Journal of Inherited Metabolic Disease, 2018, 41, 917-927.	3.6	20
146	Durable and sustained immune tolerance to ERT in Pompe disease with entrenched immune responses. JCI Insight, 2016, 1, .	5.0	20
147	National down syndrome patient database: Insights from the development of a multiâ€center registry study. American Journal of Medical Genetics, Part A, 2015, 167, 2520-2526.	1.2	19
148	Diagnosis and Management of Gaucher Disease in India â€“ Consensus Guidelines of the Gaucher Disease Task Force of the Society for Indian Academy of Medical Genetics and the Indian Academy of Pediatrics. Indian Pediatrics, 2018, 55, 143-153.	0.4	19
149	Benign or not benign? Deep phenotyping of liver Glycogen Storage Disease IX. Molecular Genetics and Metabolism, 2020, 131, 299-305.	1.1	19
150	Respiratory function during enzyme replacement therapy in late-onset Pompe disease: longitudinal course, prognostic factors, and the impact of time from diagnosis to treatment start. Journal of Neurology, 2020, 267, 3038-3053.	3.6	19
151	Common mutation in the PHKA2 gene with variable phenotype in patients with liver phosphorylase b kinase deficiency. Molecular Genetics and Metabolism, 2011, 104, 691-694.	1.1	18
152	Response to Reuser. Genetics in Medicine, 2012, 14, 827-828.	2.4	18
153	Early cognitive development in children with infantile Pompe disease. Molecular Genetics and Metabolism, 2012, 105, 428-432.	1.1	18
154	Characteristics of 26 patients with type 3 Gaucher disease: A descriptive analysis from the Gaucher Outcome Survey. Molecular Genetics and Metabolism Reports, 2018, 14, 73-79.	1.1	18
155	Liver depot gene therapy for Pompe disease. Annals of Translational Medicine, 2019, 7, 288-288.	1.7	18
156	The diagnosis and management of Gaucher disease in pediatric patients: Where do we go from here?. Molecular Genetics and Metabolism, 2022, 136, 4-21.	1.1	18
157	Three-dimensional tissue-engineered human skeletal muscle model of Pompe disease. Communications Biology, 2021, 4, 524.	4.4	17
158	Early clinical phenotype of late onset Pompe disease: Lessons learned from newborn screening. Molecular Genetics and Metabolism, 2022, 135, 179-185.	1.1	17
159	Sibling phenotype concordance in classical infantile Pompe disease. American Journal of Medical Genetics, Part A, 2007, 143A, 2493-2501.	1.2	16
160	Alglucosidase alfa enzyme replacement therapy as a therapeutic approach for glycogen storage disease type III. Molecular Genetics and Metabolism, 2013, 108, 145-147.	1.1	16
161	Safety and efficacy of rivastigmine in children with Down syndrome: A double blind placebo controlled trial. American Journal of Medical Genetics, Part A, 2016, 170, 1545-1555.	1.2	16
162	Characterization of gait in late onset Pompe disease. Molecular Genetics and Metabolism, 2015, 116, 152-156.	1.1	15

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163	Pharmacodynamics of asfotase alfa in adults with pediatric-onset hypophosphatasia. Bone, 2021, 142, 115664.	2.9	15
164	Cross-Sectional Exploration of Plasma Biomarkers of Alzheimerâ€™s Disease in Down Syndrome: Early Data from the Longitudinal Investigation for Enhancing Down Syndrome Research (LIFE-DSR) Study. Journal of Clinical Medicine, 2021, 10, 1907.	2.4	15
165	Detecting celiac disease in patients with Down syndrome. American Journal of Medical Genetics, Part A, 2016, 170, 3098-3105.	1.2	14
166	Systemic Correction of Murine Glycogen Storage Disease Type IV by an AAV-Mediated Gene Therapy. Human Gene Therapy, 2017, 28, 286-294.	2.7	14
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