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	Cardiac responses in paediatric Pompe disease in the ADVANCE patient cohort. Cardiology in the Young, 2022, 32, 364-373.	0.8	7
2	Early clinical phenotype of late onset Pompe disease: Lessons learned from newborn screening. Molecular Genetics and Metabolism, 2022, 135, 179-185.	1.1	17
3	Screening, patient identification, evaluation, and treatment in patients with Gaucher disease: Results from a Delphi consensus. Molecular Genetics and Metabolism, 2022, 135, 154-162.	1.1	10
4	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
5	Beyond predicting diagnosis: Is there a role for measuring biotinidase activity in liver glycogen storage diseases?. Molecular Genetics and Metabolism Reports, 2022, 31, 100856.	1.1	2
6	Very early-onset inflammatory bowel disease: Novel description in glycogen storage disease type Ia. Molecular Genetics and Metabolism Reports, 2022, 31, 100848.	1.1	2
7	Whatâ€™s new and whatâ€™s next for gene therapy in Pompe disease?. Expert Opinion on Biological Therapy, 2022, 22, 1117-1135.	3.1	3
8	A favorable outcome in an infantile-onset Pompe patient with cross reactive immunological material (CRIM) negative disease with high dose enzyme replacement therapy and adjusted immunomodulation. Molecular Genetics and Metabolism Reports, 2022, 32, 100893.	1.1	2
9	Pharmacodynamics of asfotase alfa in adults with pediatric-onset hypophosphatasia. Bone, 2021, 142, 115664.	2.9	15
10	Quantitative wholeâ€•body magnetic resonance imaging in children with Pompe disease: Clinical tools to evaluate severity of muscle disease. JIMD Reports, 2021, 57, 94-101.	1.5	4
11	Diurnal variability of glucose tetrasaccharide (Glc 4) excretion in patients with glycogen storage disease type III. JIMD Reports, 2021, 58, 37-43.	1.5	4
12	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
13	Late onset Pompe Disease in India â€“ Beyond the Caucasian phenotype. Neuromuscular Disorders, 2021, 31, 431-441.	0.6	6
14	"Bullâ€™s eyeâ€•appearance of hepatocellular adenomas in patients with glycogen storage disease type I â€” atypical magnetic resonance imaging findings: Two case reports. World Journal of Clinical Cases, 2021, 9, 871-877.	0.8	2
15	Case Report: Improvement Following Immunotherapy in an Individual With Seronegative Down Syndrome Disintegrative Disorder. Frontiers in Neurology, 2021, 12, 621637.	2.4	12
16	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
17	Three-dimensional tissue-engineered human skeletal muscle model of Pompe disease. Communications Biology, 2021, 4, 524.	4.4	17
18	Investigation of ALPL variant states and clinical outcomes: An analysis of adults and adolescents with hypophosphatasia treated with asfotase alfa. Molecular Genetics and Metabolism, 2021, 133, 113-121.	1.1	3

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19	Frequency of Ectopic Calcifications Among Patients With Hypophosphatasia in the Global HPP Registry. Journal of the Endocrine Society, 2021, 5, A261-A261.	0.2	0
20	The role of glucosylsphingosine as an early indicator of disease progression in early symptomatic type 1 Gaucher disease. Molecular Genetics and Metabolism Reports, 2021, 27, 100729.	1.1	4
21	Immune Tolerance-Adjusted Personalized Immunogenicity Prediction for Pompe Disease. Frontiers in Immunology, 2021, 12, 636731.	4.8	10
22	Characterization of liver GSD IX Î³2 pathophysiology in a novel Phkg2/ mouse model. Molecular Genetics and Metabolism, 2021, 133, 269-276.	1.1	4
23	Tongue weakness and atrophy differentiates late-onset Pompe disease from other forms of acquired/hereditary myopathy. Molecular Genetics and Metabolism, 2021, 133, 261-268.	1.1	12
24	New Insights into Gastrointestinal Involvement in Late-Onset Pompe Disease: Lessons Learned from Bench and Bedside. Journal of Clinical Medicine, 2021, 10, 3395.	2.4	8
25	Interpretation of Incidental Genetic Findings Localizing to Genes Associated With Cardiac Channelopathies and Cardiomyopathies. Circulation Genomic and Precision Medicine, 2021, 14, e003200.	3.6	8
26	Assessment of Dysphonia in Children with Pompe Disease Using Auditory-Perceptual and Acoustic/Physiologic Methods. Journal of Clinical Medicine, 2021, 10, 3617.	2.4	2
27	Targeted long-read sequencing identifies missing disease-causing variation. American Journal of Human Genetics, 2021, 108, 1436-1449.	6.2	105
28	Quantitative muscle ultrasound and electrical impedance myography in late onset Pompe disease: A pilot study of reliability, longitudinal change and correlation with function. Molecular Genetics and Metabolism Reports, 2021, 28, 100785.	1.1	6
29	Disorders of Carbohydrate Metabolism. , 2021, , 105-156.		1
30	Physical therapy assessment and whole-body magnetic resonance imaging findings in children with glycogen storage disease type IIIa: A clinical study and review of the literature. Molecular Genetics and Metabolism, 2021, 134, 223-234.	1.1	3
31	A retrospective longitudinal study and comprehensive review of adult patients with glycogen storage disease type III. Molecular Genetics and Metabolism Reports, 2021, 29, 100821.	1.1	7
32	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
33	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
34	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
35	Evaluation of antihypertensive drugs in combination with enzyme replacement therapy in mice with Pompe disease. Molecular Genetics and Metabolism, 2020, 129, 73-79.	1.1	2
36	Detection of iron deficiency in children with Down syndrome. Genetics in Medicine, 2020, 22, 317-325.	2.4	10

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37	Response to Zhang et al.. Genetics in Medicine, 2020, 22, 662-662.	2.4	0
38	Wholeâ€body magnetic resonance imaging in lateâ€onset Pompe disease: Clinical utility and correlation with functional measures. Journal of Inherited Metabolic Disease, 2020, 43, 549-557.	3.6	14
39	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
40	Improved muscle function in a phase I/II clinical trial of albuterol in Pompe disease. Molecular Genetics and Metabolism, 2020, 129, 67-72.	1.1	13
41	Unexplained regression in Down syndrome: 35 cases from an international Down syndrome database. Genetics in Medicine, 2020, 22, 767-776.	2.4	36
42	Pulmonary outcome measures in longâ€term survivors of infantile Pompe disease on enzyme replacement therapy: A case series. Pediatric Pulmonology, 2020, 55, 674-681.	2.0	11
43	Pregnancy Outcomes in Late Onset Pompe Disease. Life, 2020, 10, 194.	2.4	4
44	Behavioral, social and school functioning in children with Pompe disease. Molecular Genetics and Metabolism Reports, 2020, 25, 100635.	1.1	5
45	The potential impact of timing of IVIG administration on the efficacy of rituximab for immune tolerance induction for patients with Pompe disease. Clinical Immunology, 2020, 219, 108541.	3.2	3
46	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
47	Respiratory muscle training in late-onset Pompe disease: Results of a sham-controlled clinical trial. Neuromuscular Disorders, 2020, 30, 904-914.	0.6	10
48	A Novel Gene Therapy Approach for GSD III Using an AAV Vector Encoding a Bacterial Glycogen Debranching Enzyme. Molecular Therapy - Methods and Clinical Development, 2020, 18, 240-249.	4.1	11
49	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
50	Multigenerational case examples of hypophosphatasia: Challenges in genetic counseling and disease management. Molecular Genetics and Metabolism Reports, 2020, 25, 100661.	1.1	12
51	Benign or not benign? Deep phenotyping of liver Glycogen Storage Disease IX. Molecular Genetics and Metabolism, 2020, 131, 299-305.	1.1	19
52	Response to Heiner-Fokkema et al.. Genetics in Medicine, 2020, 22, 1917-1918.	2.4	1
53	A Race Against Timeâ€"Changing the Natural History of CRIM Negative Infantile Pompe Disease. Frontiers in Immunology, 2020, 11, 1929.	4.8	6
54	Training, detraining, and retraining: Two 12-week respiratory muscle training regimens in a child with infantile-onset Pompe disease. Journal of Pediatric Rehabilitation Medicine, 2020, 13, 71-80.	0.5	4

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55	Gaucher disease and SARS-CoV-2 infection: Emerging management challenges. Molecular Genetics and Metabolism, 2020, 130, 164-169.	1.1	25
56	Immune Modulation for Enzyme Replacement Therapy in A Female Patient With Hunter Syndrome. Frontiers in Immunology, 2020, 11, 1000.	4.8	9
57	Novel approaches to quantify CNS involvement in children with Pompe disease. Neurology, 2020, 95, e718-e732.	1.1	13
58	Further understanding the connection between Alzheimer's disease and Down syndrome. Alzheimer's and Dementia, 2020, 16, 1065-1077.	0.8	52
59	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
60	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
61	Adenotonsillectomy should be avoided whenever possible in infantile-onset Pompe disease. Molecular Genetics and Metabolism Reports, 2020, 23, 100574.	1.1	2
62	Realâ€worldeffectiveness 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
63	Use of the patient-reported outcomes measurement information system (PROMISÂ®) to assess late-onset Pompe disease severity. Journal of Patient-Reported Outcomes, 2020, 4, 83.	1.9	6
64	Early-onset of symptoms and clinical course of Pompe disease associated with the c.-32â€“13â€¯Tâ€¯>â€¯G variant. Molecular Genetics and Metabolism, 2019, 126, 106-116.	1.1	13
65	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
66	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
67	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
68	Gene therapy for glycogen storage diseases. Human Molecular Genetics, 2019, 28, R31-R41.	2.9	40
69	Clinical characteristics and genotypes in the ADVANCE baseline data set, a comprehensive cohort of US children and adolescents with Pompe disease. Genetics in Medicine, 2019, 21, 2543-2551.	2.4	7
70	Addendum to Letter to the Editor: Safety, efficacy, and authorization of eliglustat as a first-line therapy in Gaucher disease type 1. Blood Cells, Molecules, and Diseases, 2019, 77, 101-102.	1.4	1
71	Impact of time from diagnosis to treatment on lung function among patients with late-onset Pompe disease: Data from the Pompe registry. Molecular Genetics and Metabolism, 2019, 126, S140.	1.1	1
72	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

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73	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
74	Front Cover, Volume 40, Issue 11. Human Mutation, 2019, 40, i.	2.5	0
75	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
76	Thenar Hypertrophy and Electrical Myotonia in Pompe Disease. Journal of Clinical Neuromuscular Disease, 2019, 20, 135-137.	0.7	1
77	Ectopic Ocular Surface Calcification in Patients With Hypophosphatasia Treated With Asfotase Alfa. Cornea, 2019, 38, 896-900.	1.7	6
78	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
79	Adaptive behavior in adolescents and adults with Down syndrome: Results from a 6â€month longitudinal study. American Journal of Medical Genetics, Part A, 2019, 179, 85-93.	1.2	5
80	Five-year efficacy and safety of asfotase alfa therapy for adults and adolescents with hypophosphatasia. Bone, 2019, 121, 149-162.	2.9	99
81	Bone manifestations in neuronopathic Gaucher disease while receiving high-dose enzyme replacement therapy. Molecular Genetics and Metabolism, 2019, 126, 157-161.	1.1	7
82	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
83	Immunotherapy in selected patients with Down syndrome disintegrative disorder. Developmental Medicine and Child Neurology, 2019, 61, 847-851.	2.1	31
84	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
85	Liver depot gene therapy for Pompe disease. Annals of Translational Medicine, 2019, 7, 288-288.	1.7	18
86	Immunological challenges and approaches to immunomodulation in Pompe disease: a literature review. Annals of Translational Medicine, 2019, 7, 285-285.	1.7	38
87	SUN-529 Burden of Illness in Adults with Hypophosphatasia: Data From the Global Hypophosphatasia Patient Registry. Journal of the Endocrine Society, 2019, 3, .	0.2	1
88	P2â€030: THE LIFEâ€ÐSR STUDY. Alzheimer's and Dementia, 2019, 15, .	0.8	0
89	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
90	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

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91	Enzyme replacement therapy with alglucosidase alfa in Pompe disease: Clinical experience with rate escalation. Molecular Genetics and Metabolism, 2018, 123, 92-96.	1.1	12
92	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
93	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
94	Neuroimaging findings in infantile Pompe patients treated with enzyme replacement therapy. Molecular Genetics and Metabolism, 2018, 123, 85-91.	1.1	39
95	Clinical trial of Lâ€Carnitine and valproic acid in spinal muscular atrophy type I. Muscle and Nerve, 2018, 57, 193-199.	2.2	23
96	Hepatic Manifestations in Glycogen Storage Disease Type III. Current Pathobiology Reports, 2018, 6, 233-240.	3.4	1
97	Combination of exome sequencing and immune testing confirms Aicardiâ€“GoutiÃ¨res syndrome type 5 in a challenging pediatric neurology case. Journal of Physical Education and Sports Management, 2018, 4, a002758.	1.2	6
98	Letter to the Editors: Concerning â€œLongâ€term safety and efficacy of AAV gene therapy in the canine model of glycogen storage disease type Iaâ€•by Lee et al.. Journal of Inherited Metabolic Disease, 2018, 41, 913-914.	3.6	5
99	Corticobasal syndrome in a man with Gaucher disease type 1: Expansion of the understanding of the neurological spectrum. Molecular Genetics and Metabolism Reports, 2018, 17, 69-72.	1.1	4
100	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
101	Role of continuous glucose monitoring in the management of glycogen storage disorders. Journal of Inherited Metabolic Disease, 2018, 41, 917-927.	3.6	20
102	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
103	Diagnosis and Management of Gaucher Disease in India - 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	6
104	The emerging phenotype of late-onset Pompe disease: A systematic literature review. Molecular Genetics and Metabolism, 2017, 120, 163-172.	1.1	140
105	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
106	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
107	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
108	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

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109	Pulmonary Arterial Hypertension in Glycogen Storage Disease Type I. FIRE Forum for International Research in Education, 2017, 5, 232640981770777.	0.7	1
110	High dose IVIG successfully reduces high rhGAA IgG antibody titers in a CRIM-negative infantile Pompe disease patient. Molecular Genetics and Metabolism, 2017, 122, 76-79.	1.1	7
111	Response to de Vries et al.. Genetics in Medicine, 2017, 19, 1281-1282.	2.4	2
112	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
113	Immunomodulation to enzyme replacement therapy with tolerogenic nanoparticles containing rapamycin in a murine model of Pompe disease. Molecular Genetics and Metabolism, 2017, 120, S83-S84.	1.1	3
114	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
115	Clinical and Molecular Variability in Patients with PHKA2 Variants and Liver Phosphorylase b Kinase Deficiency. JIMD Reports, 2017, 37, 63-72.	1.5	9
116	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
117	Early Diagnosed and Treated Glutaric Acidemia Type 1 Female Presenting with Subependymal Nodules in Adulthood. JIMD Reports, 2017, 40, 85-90.	1.5	4
118	Sensitivity of whole exome sequencing in detecting infantile- and late-onset Pompe disease. Molecular Genetics and Metabolism, 2017, 122, 189-197.	1.1	21
119	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
120	Monitoring guidance for patients with hypophosphatasia treated with asfotase alfa. Molecular Genetics and Metabolism, 2017, 122, 4-17.	1.1	84
121	Challenges in measuring the effects of pharmacological interventions on cognitive and adaptive functioning in individuals with Down syndrome: A systematic review. American Journal of Medical Genetics, Part A, 2017, 173, 3058-3066.	1.2	11
122	Treatment of profound thrombocytopenia in a patient with Gaucher disease type 1: Is there a role for substrate reduction therapy. Molecular Genetics and Metabolism Reports, 2017, 12, 82-84.	1.1	5
123	Three cases of multiâ€generational Pompe disease: Are current practices missing diagnostic and treatment opportunities?. American Journal of Medical Genetics, Part A, 2017, 173, 2628-2634.	1.2	2
124	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
125	PRKAG2 mutations presenting in infancy. Journal of Inherited Metabolic Disease, 2017, 40, 823-830.	3.6	6
126	Introduction to the Newborn Screening, Diagnosis, and Treatment for Pompe Disease Guidance Supplement. Pediatrics, 2017, 140, S1-S3.	2.1	11

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127	Management of Confirmed Newborn-Screened Patients With Pompe Disease Across the Disease Spectrum. Pediatrics, 2017, 140, S24-S45.	2.1	43
128	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
129	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
130	Alglucosidase alfa enzyme replacement therapy as a therapeutic approach for a patient presenting with a PRKAG2 mutation. Molecular Genetics and Metabolism, 2017, 120, 96-100.	1.1	10
131	Sustained immune tolerance induction in enzyme replacement therapyâ€“treated CRIM-negative patients with infantile Pompe disease. JCI Insight, 2017, 2, .	5.0	47
132	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
133	Death from supine asphyxia in late onset pompe disease: Two patients. American Journal of Medical Genetics, Part A, 2016, 170, 1928-1929.	1.2	5
134	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
135	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
136	Alglucosidase alfa treatment alleviates liver disease in a mouse model of glycogen storage disease type IV. Molecular Genetics and Metabolism Reports, 2016, 9, 31-33.	1.1	3
137	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
138	Detecting celiac disease in patients with Down syndrome. American Journal of Medical Genetics, Part A, 2016, 170, 3098-3105.	1.2	14
139	Physical therapy management of infants and children with hypophosphatasia. Molecular Genetics and Metabolism, 2016, 119, 14-19.	1.1	12
140	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
141	New observation of sialuria prompts detection of liver tumor in previously reported patient. Molecular Genetics and Metabolism, 2016, 118, 92-99.	1.1	4
142	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
143	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
144	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

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145	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
146	A beta-blocker, propranolol, decreases the efficacy from enzyme replacement therapy in Pompe disease. Molecular Genetics and Metabolism, 2016, 117, 114-119.	1.1	10
147	Durable and sustained immune tolerance to ERT in Pompe disease with entrenched immune responses. JCI Insight, 2016, 1, .	5.0	20
148	Transformation in Pre-Treatment Presentations of Gaucher Disease during the First Two Decades of Imiglucerase Enzyme Replacement Therapy: A Report from the International Collaborative Gaucher Group Gaucher Registry. Blood, 2016, 128, 4877-4877.	1.4	0
149	Natural Progression of Canine Glycogen Storage Disease Type IIIa. Comparative Medicine, 2016, 66, 41-51.	1.0	13
150	Clinical laboratory experience of blood CRIM testing in infantile Pompe disease. Molecular Genetics and Metabolism Reports, 2015, 5, 76-79.	1.1	11
151	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
152	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
153	A Modified Enzymatic Method for Measurement of Glycogen Content in Glycogen Storage Disease Type IV. JIMD Reports, 2015, 30, 89-94.	1.5	4
154	Quantitative assessment of lingual strength in lateâ€onset Pompe disease. Muscle and Nerve, 2015, 51, 731-735.	2.2	25
155	Immunological Factors in Pompe Disease Management: Clinical Experience and Implications for Newborn Screening. Journal of Neuromuscular Diseases, 2015, 2, S7-S7.	2.6	0
156	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
157	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
158	Safety and efficacy of alternative alglucosidase alfa regimens in Pompe disease. Neuromuscular Disorders, 2015, 25, 321-332.	0.6	43
159	Consideration of increased dosing of alglucosidase alfa to achieve improved clinical outcomes in infantile Pompe disease. Molecular Genetics and Metabolism, 2015, 114, S96.	1.1	1
160	Premature Pubarche in Children with Pompe Disease. Journal of Pediatrics, 2015, 166, 1075-1078.e1.	1.8	4
161	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
162	Lower Urinary Tract Symptoms and Incontinence in Children with Pompe Disease. JIMD Reports, 2015, 28, 59-67.	1.5	6

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163	Postmortem Findings and Clinical Correlates in Individuals with Infantile-Onset Pompe Disease. JIMD Reports, 2015, 23, 45-54.	1.5	34
164	Characterization of gait in late onset Pompe disease. Molecular Genetics and Metabolism, 2015, 116, 152-156.	1.1	15
165	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
166	Immunological Factors in Pompe Disease Management: Clinical Experience and Implications for Newborn Screening. Journal of Neuromuscular Diseases, 2015, 2, S7.	2.6	0
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