Ans T Van Der Ploeg

List of Publications by Year
in descending order

143 papers	9,182 citations	44042 48 h-index	45285 90 g-index
144 all docs	144 docs citations	144 times ranked	4253 citing authors

#	Article	IF	Citations
1	Antibodies against recombinant human alpha-glucosidase do not seem to affect clinical outcome in childhood onset Pompe disease. Orphanet Journal of Rare Diseases, 2022, 17, 31.	1.2	5
2	Mild disease course of SARS-CoV-2 infections and mild side effects of vaccination in Pompe disease: a cohort description. Orphanet Journal of Rare Diseases, 2022, 17, 102.	1.2	5
3	Lentiviral gene therapy prevents anti-human acid Î±-glucosidase antibody formation in murine Pompe disease. Molecular Therapy - Methods and Clinical Development, 2022, 25, 520-532.	1.8	9
4	Effect of anti-iduronidase sulfatase in patients with Mucopolysaccharidosis type II treated with enzyme replacement therapy. Journal of Pediatrics, 2022, , .	0.9	1
5	Hip disease in Mucopolysaccharidoses and Mucolipidoses: A review of mechanisms, interventions and future perspectives. Bone, 2021, 143, 115729.	1.4	10
6	Enzymatic diagnosis of Pompe disease: lessons from 28 years of experience. European Journal of Human Genetics, 2021, 29, 434-446.	1.4	13
7	Can serial cerebral <scp>MRIs</scp> predict the neuronopathic phenotype of <scp>MPS II</scp>?. Journal of Inherited Metabolic Disease, 2021, 44, 751-762.	1.7	3
8	Update of the Pompe variant database for the prediction of clinical phenotypes: Novel diseaseâ€associated variants, common sequence variants, and results from newborn screening. Human Mutation, 2021, 42, 119-134.	1.1	19
9	â€œ<scp>Building bridges</scp>â€â€”An opportunity to connect, inspire, and innovate. <scp>SSIEM</scp> 2019 Annual Symposium in Rotterdam, The Netherlands. Journal of Inherited Metabolic Disease, 2021, 44, 1-1.	1.7	1
10	Chest MRI to diagnose early diaphragmatic weakness in Pompe disease. Orphanet Journal of Rare Diseases, 2021, 16, 21.	1.2	7
11	P515 A 12-week tailored physical training program including dietary advice in children with Inflammatory Bowel Disease: a randomized crossover trial. Journal of Crohn's and Colitis, 2021, 15, S493-S494.	0.6	0
12	Presymptomatic treatment of classic late-infantile neuronal ceroid lipofuscinosis with cerliponase alfa. Orphanet Journal of Rare Diseases, 2021, 16, 221.	1.2	8
13	Mucolipidosis type II and type III: a systematic review of 843 published cases. Genetics in Medicine, 2021, 23, 2047-2056.	1.1	12
14	Broad variation in phenotypes for common GAA genotypes in Pompe disease. Human Mutation, 2021, 42, 1461-1472.	1.1	4
15	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.	4.9	42
16	Is the brain involved in patients with lateâ€onset Pompe disease?. Journal of Inherited Metabolic Disease, 2021, , .	1.7	3
17	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.	4.9	59
18	Sharpening the Molecular Scissors: Advances in Gene-Editing Technology. IScience, 2020, 23, 100789.	1.9	81

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19	Discontinuation of enzyme replacement therapy in adults with Pompe disease: Evaluating the European POmpe Consortium stop criteria. Neuromuscular Disorders, 2020, 30, 59-66.	0.3	8
20	Positive association between physical outcomes and patient-reported outcomes in late-onset Pompe disease: a cross sectional study. Orphanet Journal of Rare Diseases, 2020, 15, 232.	1.2	9
21	Distal muscle weakness is a common and early feature in long-term enzyme-treated classic infantile Pompe patients. Orphanet Journal of Rare Diseases, 2020, 15, 247.	1.2	8
22	Effects of higher and more frequent dosing of alglucosidase alfa and immunomodulation on longâ€term clinical outcome of classic infantile Pompe patients. Journal of Inherited Metabolic Disease, 2020, 43, 1243-1253.	1.7	22
23	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.	1.8	19
24	Ready for Repair? Gene Editing Enters the Clinic for the Treatment of Human Disease. Molecular Therapy - Methods and Clinical Development, 2020, 18, 532-557.	1.8	67
25	Novel GAA Variants and Mosaicism in Pompe Disease Identified by Extended Analyses of Patients with an Incomplete DNA Diagnosis. Molecular Therapy - Methods and Clinical Development, 2020, 17, 337-348.	1.8	15
26	microRNAs as biomarkers in Pompe disease. Genetics in Medicine, 2019, 21, 591-600.	1.1	22
27	GAA variants and phenotypes among 1,079 patients with Pompe disease: Data from the Pompe Registry. Human Mutation, 2019, 40, 2146-2164.	1.1	51
28	Extension of the Pompe mutation database by linking diseaseâ€associated variants to clinical severity. Human Mutation, 2019, 40, 1954-1967.	1.1	47
29	Effects of immunomodulation in classic infantile Pompe patients with high antibody titers. Orphanet Journal of Rare Diseases, 2019, 14, 71.	1.2	21
30	Delayed Diagnosis of Danon Disease in Patients Presenting With Isolated Cardiomyopathy. Circulation Genomic and Precision Medicine, 2019, 12, e002395.	1.6	3
31	A genetic modifier of symptom onset in Pompe disease. EBioMedicine, 2019, 43, 553-561.	2.7	32
32	Large variation in effects during 10 years of enzyme therapy in adults with Pompe disease. Neurology, 2019, 93, e1756-e1767.	1.5	70
33	Safety, tolerability, pharmacokinetics, pharmacodynamics, and exploratory efficacy of the novel enzyme replacement therapy avalglucosidase alfa (neoGAA) in treatment-naÃ¯ve and alglucosidase alfa-treated patients with late-onset Pompe disease: A phase 1, open-label, multicenter, multinational, ascending dose study. Neuromuscular Disorders, 2019, 29, 167-186.	0.3	59
34	Long-term outcomes of systemic therapies for Hurler syndrome: an international multicenter comparison. Genetics in Medicine, 2018, 20, 1423-1429.	1.1	54
35	High Sustained Antibody Titers in Patients with Classic Infantile Pompe Disease Following Immunomodulation at Start of Enzyme Replacement Therapy. Journal of Pediatrics, 2018, 195, 236-243.e3.	0.9	27
36	Mucolipidosis type III, a series of adult patients. Journal of Inherited Metabolic Disease, 2018, 41, 839-848.	1.7	14

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37	Longâ€term followâ€up of 17 patients with childhood Pompe disease treated with enzyme replacement therapy. Journal of Inherited Metabolic Disease, 2018, 41, 1205-1214.	1.7	28
38	Imaging of respiratory muscles in neuromuscular disease: A review. Neuromuscular Disorders, 2018, 28, 246-256.	0.3	21
39	Satellite cells maintain regenerative capacity but fail to repair disease-associated muscle damage in mice with Pompe disease. Acta Neuropathologica Communications, 2018, 6, 119.	2.4	28
40	The ACE I/D polymorphism does not explain heterogeneity of natural course and response to enzyme replacement therapy in Pompe disease. PLoS ONE, 2018, 13, e0208854.	1.1	9
41	Association of Muscle Strength and Walking Performance in Adult Patients With Pompe Disease. Physical Therapy, 2018, 98, 925-931.	1.1	4
42	Enzyme replacement therapy reduces the risk for wheelchair dependency in adult Pompe patients. Orphanet Journal of Rare Diseases, 2018, 13, 82.	1.2	11
43	Cardiac outcome in classic infantile Pompe disease after 13â€¯years of treatment with recombinant human acid alpha-glucosidase. International Journal of Cardiology, 2018, 269, 104-110.	0.8	32
44	The impact of metabolic control and tetrahydrobiopterin treatment on health related quality of life of patients with early-treated phenylketonuria: A PKU-COBESO study. Molecular Genetics and Metabolism, 2018, 125, 96-103.	0.5	16
45	Large-Scale Expansion of Human iPSC-Derived Skeletal Muscle Cells for Disease Modeling and Cell-Based Therapeutic Strategies. Stem Cell Reports, 2018, 10, 1975-1990.	2.3	81
46	Alternative Splicing in Genetic Diseases: Improved Diagnosis and Novel Treatment Options. International Review of Cell and Molecular Biology, 2018, 335, 85-141.	1.6	23
47	GAA deficiency in Pompe disease is alleviated by exon inclusion in iPS cell-derived skeletal muscle cells. Proceedings for Annual Meeting of the Japanese Pharmacological Society, 2018, WCP2018, SY30-2.	0.0	0
48	European consensus for starting and stopping enzyme replacement therapy in adult patients with Pompe disease: a 10â€year experience. European Journal of Neurology, 2017, 24, 768.	1.7	118
49	Genotypeâ€“phenotype relationship in mucopolysaccharidosis <scp>II</scp>: predictive power of IDS variants for the neuronopathic phenotype. Developmental Medicine and Child Neurology, 2017, 59, 1063-1070.	1.1	28
50	Antisense Oligonucleotides Promote Exon Inclusion and Correct the Common c.-32-13T>G GAA Splicing Variant in Pompe Disease. Molecular Therapy - Nucleic Acids, 2017, 7, 90-100.	2.3	52
51	GAA Deficiency in Pompe Disease Is Alleviated by Exon Inclusion in iPSC-Derived Skeletal Muscle Cells. Molecular Therapy - Nucleic Acids, 2017, 7, 101-115.	2.3	56
52	Commentary. Clinical Chemistry, 2017, 63, 48-48.	1.5	0
53	The Dilemma of Two Innovative Therapies for Spinal Muscular Atrophy. New England Journal of Medicine, 2017, 377, 1786-1787.	13.9	17
54	Long-Term Follow-Up of Cognition and Mental Health in Adult Phenylketonuria: A PKU-COBESO Study. Behavior Genetics, 2017, 47, 486-497.	1.4	31

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55	Long-term benefit of enzyme replacement therapy in Pompe disease. Neurology, 2017, 89, 2365-2373.	1.5	93
56	A long term follow-up study of the development of hip disease in Mucopolysaccharidosis type VI. Molecular Genetics and Metabolism, 2017, 121, 241-251.	0.5	11
57	Cost-effectiveness of enzyme replacement therapy with alglucosidase alfa in adult patients with Pompe disease. Orphanet Journal of Rare Diseases, 2017, 12, 179.	1.2	15
58	Cognitive profile and mental health in adult phenylketonuria: A PKU-COBESO study.. Neuropsychology, 2017, 31, 437-447.	1.0	46
59	Pompe disease in adulthood: effects of antibody formation on enzyme replacement therapy. Genetics in Medicine, 2017, 19, 90-97.	1.1	41
60	Quantification of Diaphragm Mechanics in Pompe Disease Using Dynamic 3D MRI. PLoS ONE, 2016, 11, e0158912.	1.1	30
61	From Cryptic Toward Canonical Pre-mRNA Splicing in Pompe Disease: a Pipeline for the Development of Antisense Oligonucleotides. Molecular Therapy - Nucleic Acids, 2016, 5, e361.	2.3	29
62	Residual Nâ€acetylâ€Îâ€glucosaminidase activity in fibroblasts correlates with disease severity in patients with mucopolysaccharidosis type IIIB. Journal of Inherited Metabolic Disease, 2016, 39, 437-445.	1.7	18
63	Childhood Pompe disease: clinical spectrum and genotype in 31 patients. Orphanet Journal of Rare Diseases, 2016, 11, 65.	1.2	45
64	Effects of a higher dose of alglucosidase alfa on ventilatorâ€free survival and motor outcome in classic infantile Pompe disease: an openâ€label singleâ€center study. Journal of Inherited Metabolic Disease, 2016, 39, 383-390.	1.7	62
65	Longâ€term cognitive followâ€up in children treated for Maroteauxâ€Lamy syndrome. Journal of Inherited Metabolic Disease, 2016, 39, 285-292.	1.7	14
66	Severe tracheal and bronchial collapse in adults with type II mucopolysaccharidosis. Orphanet Journal of Rare Diseases, 2016, 11, 50.	1.2	24
67	Quality of life and participation in daily life of adults with Pompe disease receiving enzyme replacement therapy: 10 years of international followâ€up. Journal of Inherited Metabolic Disease, 2016, 39, 253-260.	1.7	43
68	Socialâ€cognitive functioning and social skills in patients with early treated phenylketonuria: a PKUâ€COBESO study. Journal of Inherited Metabolic Disease, 2016, 39, 355-362.	1.7	57
69	Identification and Characterization of AberrantGAAPre-mRNA Splicing in Pompe Disease Using a Generic Approach. Human Mutation, 2015, 36, 57-68.	1.1	28
70	Safety and efficacy of exercise training in adults with Pompe disease: evalution of endurance, muscle strength and core stability before and after a 12Âweek training program. Orphanet Journal of Rare Diseases, 2015, 10, 87.	1.2	41
71	A conceptual disease model for adult Pompe disease. Orphanet Journal of Rare Diseases, 2015, 10, 112.	1.2	10
72	Lack of robust satellite cell activation and muscle regeneration during the progression of Pompe disease. Acta Neuropathologica Communications, 2015, 3, 65.	2.4	32

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73	A Multiplex Assay for the Diagnosis of Mucopolysaccharidoses and Mucolipidoses. PLoS ONE, 2015, 10, e0138622.	1.1	35
74	Is BRIEF a useful instrument in day to day care of patients with phenylketonuria?. Molecular Genetics and Metabolism, 2015, 114, 425-430.	0.5	9
75	Pain: a prevalent feature in patients with mucopolysaccharidosis. Results of a crossâ€sectional national survey. Journal of Inherited Metabolic Disease, 2015, 38, 323-331.	1.7	34
76	Ten years of the international Pompe survey: patient reported outcomes as a reliable tool for studying treated and untreated children and adults with nonâ€classic Pompe disease. Journal of Inherited Metabolic Disease, 2015, 38, 495-503.	1.7	25
77	208th ENMC International Workshop: Formation of a European Network to develop a European data sharing model and treatment guidelines for Pompe disease Naarden, The Netherlands, 26â€“28 September 2014. Neuromuscular Disorders, 2015, 25, 674-678.	0.3	24
78	Exercise Training in Adults With Pompe Disease: TheÂEffects on Pain, Fatigue, and Functioning. Archives of Physical Medicine and Rehabilitation, 2015, 96, 817-822.	0.5	30
79	Absolute Quantification of the Total and Antidrug Antibody-Bound Concentrations of Recombinant Human Î±-Glucosidase in Human Plasma Using Protein G Extraction and LC-MS/MS. Analytical Chemistry, 2015, 87, 4394-4401.	3.2	20
80	Lung MRI and impairment of diaphragmatic function in Pompe disease. BMC Pulmonary Medicine, 2015, 15, 54.	0.8	42
81	Enzyme therapy and immune response in relation to CRIM status: the Dutch experience in classic infantile Pompe disease. Journal of Inherited Metabolic Disease, 2015, 38, 305-314.	1.7	84
82	Increased aortic stiffness and blood pressure in nonâ€classic Pompe disease. Journal of Inherited Metabolic Disease, 2014, 37, 391-397.	1.7	12
83	Impact of enzyme replacement therapy on survival in adults with Pompe disease: results from a prospective international observational study. Orphanet Journal of Rare Diseases, 2013, 8, 49.	1.2	87
84	Muscle fiberâ€type distribution, fiberâ€typeâ€specific damage, and the Pompe disease phenotype. Journal of Inherited Metabolic Disease, 2013, 36, 787-794.	1.7	18
85	Phenotypical variation within 22 families with Pompe disease. Orphanet Journal of Rare Diseases, 2013, 8, 182.	1.2	45
86	Pain in adult patients with Pompe disease. Molecular Genetics and Metabolism, 2013, 109, 371-376.	0.5	31
87	The impact of informal care for patients with Pompe disease: An application of the CarerQol instrument. Molecular Genetics and Metabolism, 2013, 110, 281-286.	0.5	18
88	The Rasch-built Pompe-specific Activity (R-PAct) scale. Neuromuscular Disorders, 2013, 23, 256-264.	0.3	45
89	Enzyme replacement therapy and fatigue in adults with Pompe disease. Molecular Genetics and Metabolism, 2013, 109, 174-178.	0.5	28
90	Up to five years experience with 11 mucopolysaccharidosis type VI patients. Molecular Genetics and Metabolism, 2013, 109, 70-76.	0.5	35

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91	Mucopolysaccharidosis: Cardiologic features and effects of enzymeâ€replacement therapy in 24 children with MPS I, II and VI. Journal of Inherited Metabolic Disease, 2013, 36, 227-234.	1.7	52
92	Remarkably low fibroblast acid Î±-glucosidase activity in three adults with Pompe disease. Molecular Genetics and Metabolism, 2012, 107, 485-489.	0.5	6
93	Open-label extension study following the Late-Onset Treatment Study (LOTS) of alglucosidase alfa. Molecular Genetics and Metabolism, 2012, 107, 456-461.	0.5	93
94	Severely impaired health status at diagnosis of Pompe disease: A cross-sectional analysis to explore the potential utility of neonatal screening. Molecular Genetics and Metabolism, 2012, 107, 448-455.	0.5	13
95	Effect of enzyme therapy and prognostic factors in 69 adults with Pompe disease: an open-label single-center study. Orphanet Journal of Rare Diseases, 2012, 7, 73.	1.2	86
96	Clinical features and predictors for disease natural progression in adults with Pompe disease: a nationwide prospective observational study. Orphanet Journal of Rare Diseases, 2012, 7, 88.	1.2	112
97	Treatment options for lysosomal storage disorders: developing insights. Expert Opinion on Pharmacotherapy, 2012, 13, 2281-2299.	0.9	22
98	Facialâ€muscle weakness, speech disorders and dysphagia are common in patients with classic infantile Pompe disease treated with enzyme therapy. Journal of Inherited Metabolic Disease, 2012, 35, 505-511.	1.7	81
99	The genotypeâ€“phenotype correlation in Pompe disease. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2012, 160C, 59-68.	0.7	102
100	The quick motor function test: a new tool to rate clinical severity and motor function in Pompe patients. Journal of Inherited Metabolic Disease, 2012, 35, 317-323.	1.7	49
101	Reply to the letter to the editor by Papadimas et al.: â€œBone mineral density in adult patients with Pompe diseaseâ€• Bone, 2011, 48, 418-419.	1.4	1
102	Bone, joint and tooth development in mucopolysaccharidoses: Relevance to therapeutic options. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2011, 1812, 1542-1556.	1.8	75
103	Pompe disease: Design, methodology, and early findings from the Pompe Registry. Molecular Genetics and Metabolism, 2011, 103, 1-11.	0.5	130
104	Rate of progression and predictive factors for pulmonary outcome in children and adults with Pompe disease. Molecular Genetics and Metabolism, 2011, 104, 129-136.	0.5	82
105	The prevalence and impact of scoliosis in Pompe disease: Lessons learned from the Pompe Registry. Molecular Genetics and Metabolism, 2011, 104, 574-582.	0.5	44
106	First experience with enzyme replacement therapy during pregnancy and lactation in Pompe disease. Molecular Genetics and Metabolism, 2011, 104, 552-555.	0.5	29
107	A case of adult Pompe disease presenting with severe fatigue and selective involvement of type 1 muscle fibers. Neuromuscular Disorders, 2011, 21, 232-234.	0.3	7
108	Fatigue in neuromuscular disorders: focus on Guillainâ€“BarrÃ© syndrome and Pompe disease. Cellular and Molecular Life Sciences, 2010, 67, 701-713.	2.4	68

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109	PASâ€positive lymphocyte vacuoles can be used as diagnostic screening test for Pompe disease. Journal of Inherited Metabolic Disease, 2010, 33, 133-139.	1.7	32
110	Hearing loss in Pompe disease revisited: results from a study of 24 children. Journal of Inherited Metabolic Disease, 2010, 33, 597-602.	1.7	50
111	A Randomized Study of Alglucosidase Alfa in Late-Onset Pompe's Disease. New England Journal of Medicine, 2010, 362, 1396-1406.	13.9	674
112	Low bone mass in Pompe disease. Bone, 2010, 47, 643-649.	1.4	53
113	Effect of enzyme therapy in juvenile patients with Pompe disease: A three-year open-label study. Neuromuscular Disorders, 2010, 20, 775-782.	0.3	54
114	Home treatment with intravenous enzyme replacement therapy with idursulfase for mucopolysaccharidosis type II â€" data from the Hunter Outcome Survey. Molecular Genetics and Metabolism, 2010, 101, 123-129.	0.5	26
115	High antibody titer in an adult with Pompe disease affects treatment with alglucosidase alfa. Molecular Genetics and Metabolism, 2010, 101, 338-345.	0.5	93
116	Rate of disease progression during long-term follow-up of patients with late-onset Pompe disease. Neuromuscular Disorders, 2009, 19, 113-117.	0.3	95
117	Early Treatment With Alglucosidase Alfa Prolongs Long-Term Survival of Infants With Pompe Disease. Pediatric Research, 2009, 66, 329-335.	1.1	277
118	Cardiac involvement in adults with Pompe disease. Journal of Internal Medicine, 2008, 264, 333-339.	2.7	54
119	Eight years experience with enzyme replacement therapy in two children and one adult with Pompe disease. Neuromuscular Disorders, 2008, 18, 447-452.	0.3	90
120	Pompe's disease. Lancet, The, 2008, 372, 1342-1353.	6.3	669
121	Impact of late-onset Pompe disease on participation in daily life activities: Evaluation of the Rotterdam Handicap Scale. Neuromuscular Disorders, 2007, 17, 537-543.	0.3	37
122	Pompe Disease: A Continuum of Clinical Phenotypes. Clinical Therapeutics, 2007, 29, S103-S104.	1.1	0
123	Broad spectrum of Pompe disease in patients with the same c.-32-13T->G haplotype. Neurology, 2007, 68, 110-115.	1.5	154
124	Fatigue: an important feature of late-onset Pompe disease. Journal of Neurology, 2007, 254, 941-945.	1.8	55
125	The natural course of nonâ€"classic Pompeâ€™s disease; a review of 225 published cases. Journal of Neurology, 2005, 252, 875-884.	1.8	306
126	Monitoring of pulmonary function in Pompe disease: a muscle disease with new therapeutic perspectives. European Respiratory Journal, 2005, 26, 984-985.	3.1	20

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127	Disease severity in children and adults with Pompe disease related to age and disease duration. Neurology, 2005, 64, 2139-2141.	1.5	159
128	Clinical manifestation and natural course of late-onset Pompe's disease in 54 Dutch patients. Brain, 2005, 128, 671-677.	3.7	310
129	Late-onset Pompe disease primarily affects quality of life in physical health domains. Neurology, 2004, 63, 1688-1692.	1.5	94
130	Enzyme replacement therapy in late-onset Pompe's disease: A three-year follow-up. Annals of Neurology, 2004, 55, 495-502.	2.8	221
131	Long-Term Intravenous Treatment of Pompe Disease With Recombinant Human Â-Glucosidase From Milk. Pediatrics, 2004, 113, e448-e457.	1.0	326
132	Morphological changes in muscle tissue of patients with infantile Pompe's disease receiving enzyme replacement therapy. Muscle and Nerve, 2003, 27, 743-751.	1.0	88
133	The Natural Course of Infantile Pompe's Disease: 20 Original Cases Compared With 133 Cases From the Literature. Pediatrics, 2003, 112, 332-340.	1.0	463
134	Recombinant human Î±-glucosidase from rabbit milk in Pompe patients. Lancet, The, 2000, 356, 397-398.	6.3	321
135	Human Acid Â-Glucosidase from Rabbit Milk Has Therapeutic Effect in Mice with Glycogen Storage Disease Type II. Human Molecular Genetics, 1999, 8, 2145-2153.	1.4	125
136	Frequency of glycogen storage disease type II in The Netherlands: implications for diagnosis and genetic counselling. European Journal of Human Genetics, 1999, 7, 713-716.	1.4	260
137	Pathological features of glycogen storage disease type II highlighted in the knockout mouse model. Journal of Pathology, 1999, 189, 416-424.	2.1	52
138	Glycogenosis type II (acid maltase deficiency). Muscle and Nerve, 1995, 18, S61-S69.	1.0	149
139	Intravenous administration of phosphorylated acid alpha-glucosidase leads to uptake of enzyme in heart and skeletal muscle of mice.. Journal of Clinical Investigation, 1991, 87, 513-518.	3.9	63
140	An investigation of the possible influence of neutral ?-glucosidases on the clinical heterogeneity of glycogenosis type II. Annals of Human Genetics, 1989, 53, 185-192.	0.3	4
141	Glycogenosis type II: protein and DNA analysis in five South African families from various ethnic origins. American Journal of Human Genetics, 1989, 44, 787-93.	2.6	24
142	Prospect for enzyme therapy in glycogenosis II variants: a study on cultured muscle cells. Journal of Neurology, 1988, 235, 392-396.	1.8	43
143	Receptor-Mediated Uptake of Acid Î±-Glucosidase Corrects Lysosomal Glycogen Storage in Cultured Skeletal Muscle. Pediatric Research, 1988, 24, 90-94.	1.1	43