## The Natural Course of Infantile Pompe's Disease: 20 Ori Cases From the Literature

Pediatrics 112, 332-340 DOI: 10.1542/peds.112.2.332

Citation Report

#	Article	IF	CITATIONS
1	Metabolic and inherited connective tissue disorders involving the lung. , 0, , 409-438.		0
2	Conjugation of Mannose 6-Phosphate-containing Oligosaccharides to Acid α-Glucosidase Improves the Clearance of Glycogen in Pompe Mice. Journal of Biological Chemistry, 2004, 279, 50336-50341.	1.6	79
3	Nephrotic Syndrome Complicating Â-Glucosidase Replacement Therapy for Pompe Disease. Pediatrics, 2004, 114, e532-e535.	1.0	83
4	Late-onset Pompe disease primarily affects quality of life in physical health domains. Neurology, 2004, 63, 1688-1692.	1.5	94
5	Enzyme replacement therapy in late-onset Pompe's disease: A three-year follow-up. Annals of Neurology, 2004, 55, 495-502.	2.8	221
6	A case of childhood Pompe disease demonstrating phenotypic variability of p.Asp645Asn. Neuromuscular Disorders, 2004, 14, 371-374.	0.3	18
7	Hearing loss in infantile Pompe's disease and determination of underlying pathology in the knockout mouse. Neurobiology of Disease, 2004, 16, 14-20.	2.1	59
8	Pompe disease in infants and children. Journal of Pediatrics, 2004, 144, S35-S43.	0.9	226
9	Long-Term Intravenous Treatment of Pompe Disease With Recombinant Human Â-Glucosidase From Milk. Pediatrics, 2004, 113, e448-e457.	1.0	326
10	Age-related decline in muscle strength and power output in acid 1-4 α-glucosidase knockout mice. Muscle and Nerve, 2005, 31, 374-381.	1.0	10
11	The natural course of non–classic Pompe's disease; a review of 225 published cases. Journal of Neurology, 2005, 252, 875-884.	1.8	306
12	Enzyme Replacement Therapy in Classical Infantile Pompe Disease: Results of a Ten-Month Follow-Up Study. Neuropediatrics, 2005, 36, 6-11.	0.3	99
13	Infantile onset Pompe disease: A report of physician narratives from an epidemiologic study. Genetics in Medicine, 2005, 7, 147-150.	1.1	34
14	Clinical manifestation and natural course of late-onset Pompe's disease in 54 Dutch patients. Brain, 2005, 128, 671-677.	3.7	310
15	Carbohydrate-remodelled acid α-glucosidase with higher affinity for the cation-independent mannose 6-phosphate receptor demonstrates improved delivery to muscles of Pompe mice. Biochemical Journal, 2005, 389, 619-628.	1.7	96
16	Replacing acid α-glucosidase in Pompe disease: recombinant and transgenic enzymes are equipotent, but neither completely clears glycogen from type II muscle fibers. Molecular Therapy, 2005, 11, 48-56.	3.7	124
17	Pompe disease diagnosis and management guideline. Genetics in Medicine, 2006, 8, 267-288.	1.1	473
18	Physical therapy management of Pompe disease. Genetics in Medicine, 2006, 8, 31 <u>8-327.</u>	1.1	61

ATION REDOL

#	Article	IF	CITATIONS
19	Hyaluronidase increases the biodistribution of acid α-1,4 glucosidase in the muscle of Pompe disease mice: An approach to enhance the efficacy of enzyme replacement therapy. Biochemical and Biophysical Research Communications, 2006, 350, 783-787.	1.0	8
20	A retrospective, multinational, multicenter study on the natural history of infantile-onset Pompe disease. Journal of Pediatrics, 2006, 148, 671-676.e2.	0.9	500
21	Chinese hamster ovary cell-derived recombinant human acid α-glucosidase in infantile-onset Pompe disease. Journal of Pediatrics, 2006, 149, 89-97.	0.9	279
22	The use of acarbose inhibition in the measurement of acid alpha-glucosidase activity in blood lymphocytes for the diagnosis of Pompe disease. Genetics in Medicine, 2006, 8, 307-312.	1.1	39
23	Characterization of pre- and post-treatment pathology after enzyme replacement therapy for pompe disease. Laboratory Investigation, 2006, 86, 1208-1220.	1.7	226
24	Limitations of enzyme replacement therapy: Current and future. Journal of Inherited Metabolic Disease, 2006, 29, 442-447.	1.7	81
25	Sudden deterioration in nonclassical infantile-onset Pompe disease responding to alglucosidase alfa infusion therapy: A case report. Journal of Inherited Metabolic Disease, 2006, 29, 763-763.	1.7	8
26	Mutation profile of theGAA gene in 40 Italian patients with late onset glycogen storage disease type II. Human Mutation, 2006, 27, 999-1006.	1.1	115
27	Dysfunction of endocytic and autophagic pathways in a lysosomal storage disease. Annals of Neurology, 2006, 59, 700-708.	2.8	286
28	Diagnostic challenges for Pompe disease: An under-recognized cause of floppy baby syndrome. Genetics in Medicine, 2006, 8, 289-296.	1.1	52
29	Brain Development in Infantile-Onset Pompe Disease Treated by Enzyme Replacement Therapy. Pediatric Research, 2006, 60, 349-352.	1.1	75
30	Comparison of maltose and acarbose as inhibitors of maltase-glucoamylase activity in assaying acid α-glucosidase activity in dried blood spots for the diagnosis of infantile Pompe disease. Genetics in Medicine, 2006, 8, 302-306.	1.1	74
31	Natural history of Type A Niemann-Pick disease: Possible endpoints for therapeutic trials. Neurology, 2006, 66, 228-232.	1.5	129
32	Electrocardiographic response to enzyme replacement therapy for Pompe disease. Genetics in Medicine, 2006, 8, 297-301.	1.1	55
33	Ambulatory electrocardiogram analysis in infants treated with recombinant human acid α-glucosidase enzyme replacement therapy for Pompe disease. Genetics in Medicine, 2006, 8, 313-317.	1.1	23
34	Diagnosis and early management of inborn errors of metabolism presenting around the time of birth1. Acta Paediatrica, International Journal of Paediatrics, 2006, 95, 6-14.	0.7	63
35	Course of disability and respiratory function in untreated late-onset Pompe disease. Neurology, 2006, 66, 581-583.	1.5	77
36	Autophagy and Mistargeting of Therapeutic Enzyme in Skeletal Muscle in Pompe Disease. Molecular Therapy, 2006, 14, 831-839.	3.7	169

#	Article	IF	CITATIONS
37	N-glycans of recombinant human acid α-glucosidase expressed in the milk of transgenic rabbits. Glycobiology, 2007, 17, 600-619.	1.3	20
39	Newborn Screening for Pompe Disease: Synthesis of the Evidence and Development of Screening Recommendations. Pediatrics, 2007, 120, e1327-e1334.	1.0	53
40	Long-Term Enzyme Replacement Therapy for Pompe Disease With Recombinant Human Alpha-glucosidase Derived From Chinese Hamster Ovary Cells. Journal of Child Neurology, 2007, 22, 565-573.	0.7	50
41	Recombinant human acid Â-glucosidase: Major clinical benefits in infantile-onset Pompe disease. Neurology, 2007, 68, 99-109.	1.5	696
42	The Liver in Lysosomal Storage Diseases. , 0, , 714-735.		0
43	Rapid diagnosis of late-onset Pompe disease by fluorometric assay of α-glucosidase activities in dried blood spots. Molecular Genetics and Metabolism, 2007, 90, 449-452.	0.5	53
44	Pompe Disease: A Continuum of Clinical Phenotypes. Clinical Therapeutics, 2007, 29, S103-S104.	1.1	0
45	Lysosomal Storage Disorders. , 2007, , .		12
46	Pompe Disease-Glycogenosis Type II: Acid Maltase Deficiency. , 2007, , 473-498.		2
47	The heart in Anderson-Fabry disease and other lysosomal storage disorders. Heart, 2007, 93, 528-535.	1.2	211
47 48	The heart in Anderson-Fabry disease and other lysosomal storage disorders. Heart, 2007, 93, 528-535. Broad spectrum of Pompe disease in patients with the same c32-13T->G haplotype. Neurology, 2007, 68, 110-115.	1.2 1.5	211 154
47 48 49	The heart in Anderson-Fabry disease and other lysosomal storage disorders. Heart, 2007, 93, 528-535. Broad spectrum of Pompe disease in patients with the same c32-13T->G haplotype. Neurology, 2007, 68, 110-115. Treatment of Lysosomal Storage Disorders. Drugs, 2007, 67, 2697-2716.	1.2 1.5 4.9	211 154 133
47 48 49 50	The heart in Anderson-Fabry disease and other lysosomal storage disorders. Heart, 2007, 93, 528-535.   Broad spectrum of Pompe disease in patients with the same c32-13T->G haplotype. Neurology, 2007, 68, 110-115.   Treatment of Lysosomal Storage Disorders. Drugs, 2007, 67, 2697-2716.   METABOLIC MYOPATHIES (INCLUDING MITOCHONDRIAL DISEASES). , 2007, , 1195-1209.	1.2 1.5 4.9	211 154 133 0
47 48 49 50 51	The heart in Anderson-Fabry disease and other lysosomal storage disorders. Heart, 2007, 93, 528-535.   Broad spectrum of Pompe disease in patients with the same c32-13T->G haplotype. Neurology, 2007, 68, 110-115.   Treatment of Lysosomal Storage Disorders. Drugs, 2007, 67, 2697-2716.   METABOLIC MYOPATHIES (INCLUDING MITOCHONDRIAL DISEASES). , 2007, , 1195-1209.   Sibling phenotype concordance in classical infantile Pompe disease. American Journal of Medical Genetics, Part A, 2007, 143A, 2493-2501.	1.2 1.5 4.9 0.7	211 154 133 0 16
47 48 49 50 51 52	The heart in Anderson-Fabry disease and other lysosomal storage disorders. Heart, 2007, 93, 528-535.Broad spectrum of Pompe disease in patients with the same c32-13T->G haplotype. Neurology, 2007, 68, 110-115.Treatment of Lysosomal Storage Disorders. Drugs, 2007, 67, 2697-2716.METABOLIC MYOPATHIES (INCLUDING MITOCHONDRIAL DISEASES). , 2007, , 1195-1209.Sibling phenotype concordance in classical infantile Pompe disease. American Journal of Medical Cenetics, Part A, 2007, 143A, 2493-2501.Cardiac arrhythmias following anesthesia induction in infantile-onset Pompe disease: a case series. Paediatric Anaesthesia, 2007, 17, 738-748.	1.2 1.5 4.9 0.7 0.6	211 154 133 0 16 54
47 48 49 50 51 52 53	The heart in Anderson-Fabry disease and other lysosomal storage disorders. Heart, 2007, 93, 528-535.   Broad spectrum of Pompe disease in patients with the same c32-13T->G haplotype. Neurology, 2007, 68, 110-115.   Treatment of Lysosomal Storage Disorders. Drugs, 2007, 67, 2697-2716.   METABOLIC MYOPATHIES (INCLUDING MITOCHONDRIAL DISEASES)., 2007, , 1195-1209.   Sibling phenotype concordance in classical infantile Pompe disease. American Journal of Medical Genetics, Part A, 2007, 143A, 2493-2501.   Cardiac arrhythmias following anesthesia induction in infantile-onset Pompe disease: a case series. Paediatric Anaesthesia, 2007, 17, 738-748.   Diagnosis and early management of inborn errors of metabolism presenting around the time of birth. Acta Paediatrica, International Journal of Paediatrics, 2006, 95, 6-14.	1.2 1.5 4.9 0.7 0.6	211 154 133 0 16 54 4
47 48 49 50 51 52 53 54	The heart in Anderson-Fabry disease and other lysosomal storage disorders. Heart, 2007, 93, 528-535.   Broad spectrum of Pompe disease in patients with the same c32-13T->G haplotype. Neurology, 2007, 68, 110-115.   Treatment of Lysosomal Storage Disorders. Drugs, 2007, 67, 2697-2716.   METABOLIC MYOPATHIES (INCLUDING MITOCHONDRIAL DISEASES)., 2007, , 1195-1209.   Sibling phenotype concordance in classical infantile Pompe disease. American Journal of Medical Genetics, Part A, 2007, 143A, 2493-2501.   Cardiac arrhythmias following anesthesia induction in infantile-onset Pompe disease: a case series. Paediatric Anaesthesia, 2007, 17, 738-748.   Diagnosis and early management of inborn errors of metabolism presenting around the time of birth. Acta Paediatrica, International Journal of Paediatrics, 2006, 95, 6-14.   Lysosomal dysfunction, cellular pathology and clinical symptoms: basic principles. Acta Paediatrica, International Journal of Paediatrics, 2006, 95, 77-82.	1.2 1.5 4.9 0.7 0.6 0.7	211 154 133 0 16 54 4 4

ARTICLE IF CITATIONS # Neuromuscular disorders associated with cerebral malformations. Handbook of Clinical Neurology / 1.0 0 56 Edited By PJ Vinken and GW Bruyn, 2007, 87, 409-431. Fractures in children with Pompe disease: a potentiallong-term complication. Pediatric Radiology, 1.1 28 2007, 37, 437-445. A severity scoring tool to assess the neurological features of neuronopathic Gaucher disease. 58 1.7 28 Journal of Inherited Metabolic Disease, 2007, 30, 768-782. Isolated elevated serum transaminases leading to the diagnosis of asymptomatic Pompe disease. 59 European Journal of Pediatrics, 2007, 166, 871-874. New therapeutic options for lysosomal storage disorders: enzyme replacement, small molecules and 60 1.8 171 gene therapy. Human Genetics, 2007, 121, 1-22. Fatigue: an important feature of late-onset Pompe disease. Journal of Neurology, 2007, 254, 941-945. 1.8 Acid alpha-glucosidase deficiency (Pompe disease). Current Neurology and Neuroscience Reports, 2007, 62 2.0 44 7,71-77. Early administration of enzyme replacement therapy for Pompe disease: Shortâ€term followâ€up results. Journal of Inherited Metabolic Disease, 2008, 31, 431-436. 1.7 Cardiac Remodeling After Enzyme Replacement Therapy with Acid α-Glucosidase for Infants with Pompe 64 0.6 51 Disease. Pediatric Cardiology, 2008, 29, 1033-1042. Both type 1 and type 2a muscle fibers can respond to enzyme therapy in Pompe disease. Muscle and 1.0 Nerve, 2008, 37, 251-255. Clinical features of lateâ€onset Pompe disease: A prospective cohort study. Muscle and Nerve, 2008, 38, 1.0 200 66 1236-1245. Update of the Pompe disease mutation database with 107 sequence variants and a format for severity 1.1 186 rating. Human Mutation, 2008, 29, E13-E26. Molecular and functional characterization of eight novel GAA mutations in Italian infants with 68 1.1 51 Pompe disease. Human Mutation, 2008, 29, E27-E36. Cardiac involvement in adults with Pompe disease. Journal of Internal Medicine, 2008, 264, 333-339. 2.7 54 Cardiac evaluation in children and adults with Pompe disease sharing the common c.â<sup>^3</sup>32-13T&gt;G 70 0.3 33 genotype rarely reveals abnormalities. Journal of the Neurological Sciences, 2008, 275, 46-50. Eight years experience with enzyme replacement therapy in two children and one adult with Pompe disease. Neuromuscular Disorders, 2008, 18, 447-452. Methods for a prompt and reliable laboratory diagnosis of Pompe disease: Report from an 73 0.5 133 international consensus meeting. Molecular Genetics and Metabolism, 2008, 93, 275-281. Improvement with ongoing Enzyme Replacement Therapy in advanced late-onset Pompe disease: A case 74 study. Molecular Genetics and Metabolism, 2008, 95, 233-235.

	CITATION RE	CITATION REPORT	
#	Article	IF	CITATIONS
75	Cardiomyopathy in Pompe's disease. European Journal of Internal Medicine, 2008, 19, 57-59.	1.0	18
76	Pompe's disease. Lancet, The, 2008, 372, 1342-1353.	6.3	669
77	The role of current biomarkers in the management of lysosomal storage disorders. Clinical Therapeutics, 2008, 30, S90-S91.	1.1	1
78	Biomarkers for Pompe Disease. Clinical Therapeutics, 2008, 30, S4-S5.	1.1	0
79	Natural Course and Effects of Enzyme Therapy in Adults with Pompe Disease. Clinical Therapeutics, 2008, 30, S15-S16.	1.1	0
80	Aspectos respiratórios da doença de Pompe: Relato de caso. Revista Portuguesa De Pneumologia, 2008, 14, 159-164.	0.7	0
81	STIFF EYES IN STIFF-PERSON SYNDROME. Neurology, 2008, 71, 378-380.	1.5	29
82	Management and treatment of glycogenosis type II. Neurology, 2008, 71, S12-36.	1.5	52
83	Diagnosis of glycogenosis type II. Neurology, 2008, 71, S4-11.	1.5	87
84	Arrhythmias in patients receiving enzyme replacement therapy for infantile Pompe disease. Genetics in Medicine, 2008, 10, 758-762.	1.1	23
85	Ability of Adeno-Associated Virus Serotype 8-Mediated Hepatic Expression of Acid α-Glucosidase to Correct the Biochemical and Motor Function Deficits of Presymptomatic and Symptomatic Pompe Mice. Human Gene Therapy, 2008, 19, 609-621.	1.4	61
87	Pompe Disease: A Review of the Current Diagnosis and Treatment Recommendations in the Era of Enzyme Replacement Therapy. Journal of Clinical Neuromuscular Disease, 2008, 9, 421-431.	0.3	61
88	Alglucosidase alfa: Long term use in the treatment of patients with Pompe disease. Therapeutics and Clinical Risk Management, 2009, 5, 767.	0.9	18
89	La maladie de PompeÂ: une myopathie métabolique à l'ère de l'enzymothérapie. Neurologie Com, 2009, 1, 012-015.	0.0	0
90	Treatment of Gastroesophageal Reflux with Nissen Fundoplication and Gastrostomy Tube Insertion in Infantile Pompe's Disease. Neuropediatrics, 2009, 40, 28-31.	0.3	7
91	Glycoengineered Acid α-Glucosidase With Improved Efficacy at Correcting the Metabolic Aberrations and Motor Function Deficits in a Mouse Model of Pompe Disease. Molecular Therapy, 2009, 17, 954-963.	3.7	137
92	The Pharmacological Chaperone N-butyldeoxynojirimycin Enhances Enzyme Replacement Therapy in Pompe Disease Fibroblasts. Molecular Therapy, 2009, 17, 964-971.	3.7	130
93	Reversal of Cardiac Dysfunction after Enzyme Replacement in Patients with Infantile-Onset Pompe Disease. Journal of Pediatrics, 2009, 155, 271-275.e2.	0.9	56

#	Article	IF	CITATIONS
94	Diagnostic criteria for lateâ€onset (childhood and adult) pompe disease. Muscle and Nerve, 2009, 40, 149-160.	1.0	100
95	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
96	Pompe disease: A neuromuscular disease with respiratory muscle involvement. Respiratory Medicine, 2009, 103, 477-484.	1.3	95
97	Clinical outcomes after long-term treatment with alglucosidase alfa in infants and children with advanced Pompe disease. Genetics in Medicine, 2009, 11, 210-219.	1.1	259
98	Early Treatment With Alglucosidase Alfa Prolongs Long-Term Survival of Infants With Pompe Disease. Pediatric Research, 2009, 66, 329-335.	1.1	277
99	Pompe Disease in Infants: Improving the Prognosis by Newborn Screening and Early Treatment. Pediatrics, 2009, 124, e1116-e1125.	1.0	185
100	Phenotype Variations in Early Onset Pompe Disease: Diagnosis and Treatment Results with Myozyme®. Advances in Experimental Medicine and Biology, 2009, 652, 39-46.	0.8	11
101	Clinical and Histologic Ocular Findings in Pompe Disease. Journal of Pediatric Ophthalmology and Strabismus, 2010, 47, 34-40.	0.3	41
102	Lentiviral gene therapy of murine hematopoietic stem cells ameliorates the Pompe disease phenotype. Blood, 2010, 115, 5329-5337.	0.6	81
103	Fatigue in neuromuscular disorders: focus on Guillain–Barré syndrome and Pompe disease. Cellular and Molecular Life Sciences, 2010, 67, 701-713.	2.4	68
104	Oropharyngeal Dysphagia in Infants and Children with Infantile Pompe Disease. Dysphagia, 2010, 25, 277-283.	1.0	80
105	Combined general and epidural anesthesia for major abdominal surgery in a patient with Pompe disease. Journal of Anesthesia, 2010, 24, 768-773.	0.7	12
106	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
107	Treatment of infantile Pompe disease with alglucosidase alpha: the UK experience. Journal of Inherited Metabolic Disease, 2010, 33, 747-750.	1.7	77
109	Fiber Type Conversion by PGC-1α Activates Lysosomal and Autophagosomal Biogenesis in Both Unaffected and Pompe Skeletal Muscle. PLoS ONE, 2010, 5, e15239.	1.1	48
110	Metabolic and endocrine conditions. , 0, , 463-490.		0
111	A Severe Form of Non-Classic Pompe's Disease with Normal Creatinine Kinase Level. Neuropediatrics, 2010, 41, 193-195.	0.3	0
112	A Randomized Study of Alglucosidase Alfa in Late-Onset Pompe's Disease. New England Journal of Medicine, 2010, 362, 1396-1406.	13.9	674

#	Article	IF	CITATIONS
113	Chorionic villi ultrastructure in the prenatal diagnosis of glycogenosis type II. Journal of Inherited Metabolic Disease, 2010, 33, 105-111.	1.7	46
114	Antenatal diagnosis of pompe disease by fetal echocardiography: impact on outcome after early initiation of enzyme replacement therapy. Journal of Inherited Metabolic Disease, 2010, 33, 333-339.	1.7	29
115	Longâ€ŧerm followâ€up results in enzyme replacement therapy for Pompe disease: a case report. Journal of Inherited Metabolic Disease, 2010, 33, 389-393.	1.7	10
116	Low bone mass in Pompe disease. Bone, 2010, 47, 643-649.	1.4	53
117	Acute Progression of Neuromuscular Findings in Infantile Pompe Disease. Pediatric Neurology, 2010, 42, 455-458.	1.0	42
118	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
119	Cross-reactive immunologic material status affects treatment outcomes in Pompe disease infants. Molecular Genetics and Metabolism, 2010, 99, 26-33.	0.5	348
120	Prognostic factors for the late onset Pompe disease with enzyme replacement therapy: From our experience of 4 cases including an autopsy case. Molecular Genetics and Metabolism, 2010, 100, 14-19.	0.5	55
121	Pompe disease: Dramatic improvement in gastrointestinal function following enzyme replacement therapy. A report of three later-onset patients. Molecular Genetics and Metabolism, 2010, 101, 130-133.	0.5	52
122	Use of cardiac magnetic resonance imaging to evaluate cardiac structure, function and fibrosis in children with infantile Pompe disease on enzyme replacement therapy. Molecular Genetics and Metabolism, 2010, 101, 332-337.	0.5	26
123	Differences in the predominance of lysosomal and autophagic pathologies between infants and adults with Pompe disease: implications for therapy. Molecular Genetics and Metabolism, 2010, 101, 324-331.	0.5	69
124	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
125	Tijdige diagnostiek en behandeling van de ziekte van Pompe kan irreversibele orgaanschade voorkomen. Tijdschrift Voor Kindergeneeskunde, 2010, 78, 81-88.	0.0	0
126	Glycogen Storage Diseases: A Brief Review and Update on Clinical Features, Genetic Abnormalities, Pathologic Features, and Treatment. Ultrastructural Pathology, 2011, 35, 183-196.	0.4	110
127	2011 ACCF/AHA Guideline for the Diagnosis and Treatment of Hypertrophic Cardiomyopathy. Journal of the American College of Cardiology, 2011, 58, e212-e260.	1.2	984
128	2011 ACCF/AHA Guideline for the Diagnosis and Treatment of Hypertrophic Cardiomyopathy: Executive Summary. Journal of the American College of Cardiology, 2011, 58, 2703-2738.	1.2	252
129	2011 ACCF/AHA Guideline for the Diagnosis and Treatment of Hypertrophic Cardiomyopathy. Circulation, 2011, 124, e783-831.	1.6	1,039
130	Proteasome inhibitors improve the function of mutant lysosomal α-glucosidase in fibroblasts from Pompe disease patient carrying c.546G>T mutation. Biochemical and Biophysical Research Communications, 2011, 415, 274-278.	1.0	19

#	Article	IF	CITATIONS
131	Human Pompe disease-induced pluripotent stem cells for pathogenesis modeling, drug testing and disease marker identification. Human Molecular Genetics, 2011, 20, 4851-4864.	1.4	129
132	Oral administration of recombinant human acid α-glucosidase reduces specific antibody formation against enzyme in mouse. Molecular Genetics and Metabolism, 2011, 103, 98-100.	0.5	13
133	Pompe disease: Design, methodology, and early findings from the Pompe Registry. Molecular Genetics and Metabolism, 2011, 103, 1-11.	0.5	130
134	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
135	Atypical immunologic response in a patient with CRIM-negative Pompe disease. Molecular Genetics and Metabolism, 2011, 104, 583-586.	0.5	29
136	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
137	Endoplasmic reticulum stress induces autophagy through activation of p38 MAPK in fibroblasts from Pompe disease patients carrying c.546G>T mutation. Molecular Genetics and Metabolism, 2011, 104, 566-573.	0.5	39
138	From symptoms to causes: progress in the treatment of neurological disease. British Journal of Hospital Medicine (London, England: 2005), 2011, 72, 350-351.	0.2	2
139	Hypoglossal Neuropathology and Respiratory Activity in Pompe Mice. Frontiers in Physiology, 2011, 2, 31.	1.3	46
140	Pompe Disease: From New Views on Pathophysiology to Innovative Therapeutic Strategies. Current Pharmaceutical Biotechnology, 2011, 12, 902-915.	0.9	33
142	Splicing mutations in glycogen-storage disease type II: evaluation of the full spectrum of mutations and their relation to patients' phenotypes. European Journal of Human Genetics, 2011, 19, 422-431.	1.4	29
143	Gingival Overgrowth in Pompe Disease: A Case Report. Journal of Oral and Maxillofacial Surgery, 2011, 69, 2186-2190.	0.5	9
144	2011 ACCF/AHA guideline for the diagnosis and treatment of hypertrophic cardiomyopathy: Executive summary. Journal of Thoracic and Cardiovascular Surgery, 2011, 142, 1303-1338.	0.4	73
145	2011 ACCF/AHA guideline for the diagnosis and treatment of hypertrophic cardiomyopathy. Journal of Thoracic and Cardiovascular Surgery, 2011, 142, e153-e203.	0.4	260
146	Burden of illness of Pompe disease in patients only receiving supportive care. Journal of Inherited Metabolic Disease, 2011, 34, 1045-1052.	1.7	29
147	Clinical consequences of reduced dosing schedule during treatment of a patient with Pompe's disease. Biologics in Therapy, 2011, 1, 1.	1.8	2
148	Lysosomal storage diseases: Diagnostic confirmation and management of presymptomatic individuals. Genetics in Medicine, 2011, 13, 457-484.	1.1	188
149	CEREBRAL DIFFUSION TENSOR IMAGES IN INFANTS AND NEONATES WITH INFANTILE ONSET POMPE DISEASE. Biomedical Engineering - Applications, Basis and Communications, 2011, 23, 205-213.	0.3	2

#	Article	IF	CITATIONS
150	Cardiovascular abnormalities in late-onset Pompe disease and response to enzyme replacement therapy. Genetics in Medicine, 2011, 13, 625-631.	1.1	39
151	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.	1.1	216
152	2011 ACCF/AHA Guideline for the Diagnosis and Treatment of Hypertrophic Cardiomyopathy: Executive Summary. Circulation, 2011, 124, 2761-2796.	1.6	725
153	Cognitive outcome of patients with classic infantile Pompe disease receiving enzyme therapy. Neurology, 2012, 78, 1512-1518.	1.5	67
154	Identification and Functional Characterization of GAA Mutations in Colombian Patients Affected by Pompe Disease. JIMD Reports, 2012, 7, 39-48.	0.7	10
155	The effectiveness and cost-effectiveness of enzyme and substrate replacement therapies: a longitudinal cohort study of people with lysosomal storage disorders Health Technology Assessment, 2012, 16, 1-543.	1.3	94
156	Infantile Pompe Disease: Clinical and Genetic Characteristics With an Experience of Enzyme Replacement Therapy. Journal of Child Neurology, 2012, 27, 319-324.	0.7	10
157	The emerging phenotype of long-term survivors with infantile Pompe disease. Genetics in Medicine, 2012, 14, 800-810.	1.1	163
158	CRIM-Negative Pompe Disease Patients with Satisfactory Clinical Outcomes on Enzyme Replacement Therapy. JIMD Reports, 2012, 9, 133-137.	0.7	10
159	In Vivo Bone Architecture in Pompe Disease Using High-Resolution Peripheral Computed Tomography. JIMD Reports, 2012, 7, 81-88.	0.7	11
160	The Changing Face of Infantile Pompe Disease: A Report of Five Patients from the UAE. JIMD Reports, 2012, 8, 7-10.	0.7	1
162	Akt inactivation induces endoplasmic reticulum stress-independent autophagy in fibroblasts from patients with Pompe disease. Molecular Genetics and Metabolism, 2012, 107, 490-495.	0.5	19
163	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
164	Two cases of Pompe's disease: case report and review of literature. Indian Heart Journal, 2012, 64, 214-216.	0.2	8
165	Myalgic phenotype and preserved muscle strength in adult-onset acid maltase deficiency. Neuromuscular Disorders, 2012, 22, 763-766.	0.3	5
166	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.	0.5	59
167	Transcriptional response to GAA deficiency (Pompe disease) in infantile-onset patients. Molecular Genetics and Metabolism, 2012, 106, 287-300.	0.5	20
168	Public support for neonatal screening for Pompe disease, a broad-phenotype condition. Orphanet Journal of Rare Diseases, 2012, 7, 15.	1.2	19

#	Article	IF	CITATIONS
169	A cross-sectional single-centre study on the spectrum of Pompe disease, German patients: molecular analysis of the GAA gene, manifestation and genotype-phenotype correlations. Orphanet Journal of Rare Diseases, 2012, 7, 35.	1.2	73
170	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
171	The Glycogen Storage Diseases and Related Disorders. , 2012, , 115-139.		18
172	Update of the pompe disease mutation database with 60 novel GAA sequence variants and additional studies on the functional effect of 34 previously reported variants. Human Mutation, 2012, 33, 1161-1165.	1.1	67
173	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
174	High-density CT of muscle and liver may allow early diagnosis of childhood-onset Pompe disease. Brain and Development, 2012, 34, 103-106.	0.6	6
175	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.	0.7	60
176	Infantile Pompe disease on ERT—Update on clinical presentation, musculoskeletal management, and exercise considerations. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2012, 160C, 69-79.	0.7	67
177	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
178	Improvement of dysphagia in a child affected by Pompe disease treated with enzyme replacement therapy. Italian Journal of Pediatrics, 2013, 39, 30.	1.0	8
179	Hypertransaminasemia and fatal lung disease: a case report. Italian Journal of Pediatrics, 2013, 39, 9.	1.0	1
180	Muscle fiberâ€ŧype distribution, fiberâ€ŧypeâ€specific damage, and the Pompe disease phenotype. Journal of Inherited Metabolic Disease, 2013, 36, 787-794.	1.7	18
181	Enzyme Replacement Therapy Improves Respiratory Outcomes in Patients with Late-Onset Type II Glycogenosis and High Ventilator Dependency. Lung, 2013, 191, 537-544.	1.4	24
182	Enzyme Replacement in Neuronal Storage Disorders in the Pediatric Population. Current Treatment Options in Neurology, 2013, 15, 634-651.	0.7	4
183	Perioperative respiratory management of pediatric patients with neuromuscular disease. Paediatric Anaesthesia, 2013, 23, 770-776.	0.6	12
184	Intrapleural Administration of AAV9 Improves Neural and Cardiorespiratory Function in Pompe Disease. Molecular Therapy, 2013, 21, 1661-1667.	3.7	63
185	Phenotypical variation within 22 families with Pompe disease. Orphanet Journal of Rare Diseases, 2013, 8, 182.	1.2	45
186	Prevalence of hearing loss in patients with late-onset Pompe disease: Audiological and otological consequences. International Journal of Audiology, 2013, 52, 816-823.	0.9	18

#	Article	IF	CITATIONS
187	Respiratory manifestations in patients with inherited metabolic diseases. European Respiratory Review, 2013, 22, 437-453.	3.0	18
188	Pharmacotherapy of Pompe disease. Expert Opinion on Orphan Drugs, 2013, 1, 457-471.	0.5	1
189	Detection of c. â^'32T>G (IVS1â^'13T>G) mutation of Pompe disease by real-time PCR in dried blood spot specimen. Clinica Chimica Acta, 2013, 418, 107-108.	0.5	6
191	The respiratory neuromuscular system in Pompe disease. Respiratory Physiology and Neurobiology, 2013, 189, 241-249.	0.7	97
192	The Rasch-built Pompe-specific Activity (R-PAct) scale. Neuromuscular Disorders, 2013, 23, 256-264.	0.3	45
193	Pharmacological Chaperones as Therapeutics for Lysosomal Storage Diseases. Journal of Medicinal Chemistry, 2013, 56, 2705-2725.	2.9	182
194	Timing of diagnosis of patients with pompe disease: Data from the pompe registry. American Journal of Medical Genetics, Part A, 2013, 161, 2431-2443.	0.7	60
195	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.	1.1	75
196	A 16-Week-Old Infant With Failure to Thrive and Hypotonia. Clinical Pediatrics, 2013, 52, 1075-1078.	0.4	0
197	Successful Desensitisation in a Patient with CRIM-Positive Infantile-Onset Pompe Disease. JIMD Reports, 2013, 12, 99-102.	0.7	17
198	Molecular basis and clinical management of Pompe disease. Neurology International, 2013, 3, .	0.2	2
199	Polysomnographic findings in infantile Pompe disease. American Journal of Medical Genetics, Part A, 2013, 161, 3196-3200.	0.7	28
200	Autophagy in GNE Myopathy. , 2013, , .		3
201	Lysosomal storage disorders. , 0, , 546-566.		1
202	Two novel mutations in acid $\hat{l}\pm$ -glucosidase gene in two patients with Pompe disease. Journal of Pediatric Endocrinology and Metabolism, 2014, 27, 1265-7.	0.4	2
203	Pompe disease: from pathophysiology to therapy and back again. Frontiers in Aging Neuroscience, 2014, 6, 177.	1.7	147
204	Non-depleting anti-CD4 monoclonal antibody induces immune tolerance to ERT in a murine model of Pompe disease. Molecular Genetics and Metabolism Reports, 2014, 1, 446-450.	0.4	13
205	Outcome of Patients with Classical Infantile Pompe Disease Receiving Enzyme Replacement Therapy in Germany. JIMD Reports, 2014, 20, 65-75.	0.7	47

#	Article	IF	CITATIONS
206	Proteasome Inhibitor Bortezomib Enhances the Activity of Multiple Mutant Forms of Lysosomal α-Glucosidase in Pompe Disease. JIMD Reports, 2014, 18, 33-39.	0.7	24
207	Radiological and clinical characterization of the lysosomal storage disorders: non-lipid disorders. British Journal of Radiology, 2014, 87, 20130467.	1.0	18
208	A largeâ€scale nationwide newborn screening program for pompe disease in Taiwan: Towards effective diagnosis and treatment. American Journal of Medical Genetics, Part A, 2014, 164, 54-61.	0.7	59
209	Immune Tolerance Induced Using Plasma Exchange and Rituximab in an Infantile Pompe Disease Patient. Journal of Child Neurology, 2014, 29, 850-854.	0.7	11
210	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.	0.3	22
211	Functional characterization of the common c32-13T>G mutation of GAA gene: identification of potential therapeutic agents. Nucleic Acids Research, 2014, 42, 1291-1302.	6.5	40
213	Increased aortic stiffness and blood pressure in nonâ€classic Pompe disease. Journal of Inherited Metabolic Disease, 2014, 37, 391-397.	1.7	12
214	Enzyme replacement therapy in juvenile glycogenosis type II: a longitudinal study. European Journal of Pediatrics, 2014, 173, 805-813.	1.3	18
215	Lentiviral Hematopoietic Stem Cell Gene Therapy in Inherited Metabolic Disorders. Human Gene Therapy, 2014, 25, 862-865.	1.4	15
216	Methods of diagnosis of patients with Pompe disease: Data from the Pompe Registry. Molecular Genetics and Metabolism, 2014, 113, 84-91.	0.5	38
217	Pompe Disease. Neurologic Clinics, 2014, 32, 751-776.	0.8	104
218	Newborn screening for pompe disease? a qualitative study exploring professional views. BMC Pediatrics, 2014, 14, 203.	0.7	11
219	Effectiveness of enzyme replacement therapy in adults with lateâ€onset Pompe disease: results from the NCSâ€LSD cohort study. Journal of Inherited Metabolic Disease, 2014, 37, 945-952.	1.7	51
220	Cost-effectiveness of enzyme replacement therapy with alglucosidase alfa in classic-infantile patients with Pompe disease. Orphanet Journal of Rare Diseases, 2014, 9, 75.	1.2	26
221	Sustained Correction of Motoneuron Histopathology Following Intramuscular Delivery of AAV in Pompe Mice. Molecular Therapy, 2014, 22, 702-712.	3.7	69
222	Clinical and molecular genetic study of infantile-onset Pompe disease in Chinese patients: Identification of 6 novel mutations. Gene, 2014, 535, 53-59.	1.0	20
224	Novel GAA sequence variant c.1211 A>G reduces enzyme activity but not protein expression in in infantile and adult onset Pompe disease. Gene, 2014, 537, 41-45.	1.0	6
225	Ten Years of The International Pompe Survey: Patient Reported Outcomes As A Reliable Tool for Studying Treated and Untreated Children and Adults With Non-Classic Pompe Disease. Value in Health, 2015, 18, A673.	0.1	2

#	Article	IF	CITATIONS
226	Longitudinal polysomnographic findings in infantile Pompe disease. American Journal of Medical Genetics, Part A, 2015, 167, 858-861.	0.7	13
227	Late onset form of Pompe disease. Bratislava Medical Journal, 2015, 116, 502-505.	0.4	2
228	Metabolic muscle disorders in infants and children. Journal of Pediatric Biochemistry, 2015, 04, 231-248.	0.2	0
229	Lysosomal Storage Diseases-Regulating Neurodegeneration. Journal of Experimental Neuroscience, 2015, 9s2, JEN.S25475.	2.3	27
230	Long-Term Prognosis of Patients with Infantile-Onset Pompe Disease Diagnosed by Newborn Screening and Treated since Birth. Journal of Pediatrics, 2015, 166, 985-991.e2.	0.9	113
231	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
232	Late-Onset Pompe Disease With Left-Sided Bronchomalacia. Respiratory Care, 2015, 60, e26-e29.	0.8	20
233	Immune tolerance strategies in siblings with infantile Pompe disease — Advantages for a preemptive approach to high-sustained antibody titers. Molecular Genetics and Metabolism Reports, 2015, 4, 30-34.	0.4	24
234	Cardiomyopathy in a Dish: Using Human Inducible Pluripotent Stem Cells to Model Inherited Cardiomyopathies. Journal of Cardiac Failure, 2015, 21, 761-770.	0.7	28
235	Structural and functional cardiac analyses using modern and sensitive myocardial techniques in adult Pompe disease. International Journal of Cardiovascular Imaging, 2015, 31, 947-956.	0.7	15
236	Lung MRI and impairment of diaphragmatic function in Pompe disease. BMC Pulmonary Medicine, 2015, 15, 54.	0.8	42
237	Stimulation of Respiratory Motor Output and Ventilation in a Murine Model of Pompe Disease by Ampakines. American Journal of Respiratory Cell and Molecular Biology, 2015, 53, 326-335.	1.4	39
238	Enzyme replacement therapy for infantile-onset Pompe disease. The Cochrane Library, 2015, , .	1.5	2
239	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
240	Pompe disease: clinical perspectives. Orphan Drugs: Research and Reviews, 0, Volume 7, 1-10.	0.6	5
241	Inherited Wolff–Parkinson–White Syndrome. Cardiovascular Innovations and Applications, 2016, 1, .	0.1	1
242	Lysosomal Diseases. , 2016, , 763-788.		1
243	Childhood Pompe disease: clinical spectrum and genotype in 31 patients. Orphanet Journal of Rare Diseases, 2016, 11, 65.	1.2	45

#	Article	IF	CITATIONS
244	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
245	Pompe Disease: Diagnosis and Management. Evidence-Based Guidelines from a Canadian Expert Panel. Canadian Journal of Neurological Sciences, 2016, 43, 472-485.	0.3	54
246	Cognitive Development in Infantile-Onset Pompe Disease Under Very Early Enzyme Replacement Therapy. Journal of Child Neurology, 2016, 31, 1617-1621.	0.7	11
247	Meta-opinion: from screening to diagnosis of Pompe disease – a European perspective. Expert Opinion on Orphan Drugs, 2016, 4, 1075-1078.	0.5	1
248	Clinical manifestation of late onset Pompe disease patients in Hong Kong. Neuromuscular Disorders, 2016, 26, 873-879.	0.3	9
249	Prospective exploratory muscle biopsy, imaging, and functional assessment in patients with late-onset Pompe disease treated with alglucosidase alfa: The EMBASSY Study. Molecular Genetics and Metabolism, 2016, 119, 115-123.	0.5	49
250	Diagnosis of late-onset Pompe disease and other muscle disorders by next-generation sequencing. Orphanet Journal of Rare Diseases, 2016, 11, 8.	1.2	42
251	Elevated Plasma Cardiac Troponin T Levels Caused by Skeletal Muscle Damage in Pompe Disease. Circulation: Cardiovascular Genetics, 2016, 9, 6-13.	5.1	70
252	Advances in newborn screening for Pompe disease and resulting clinical outcomes. Expert Opinion on Orphan Drugs, 2016, 4, 21-29.	0.5	0
253	Response of 33 UK patients with infantileâ€onset Pompe disease to enzyme replacement therapy. Journal of Inherited Metabolic Disease, 2016, 39, 261-271.	1.7	54
254	Very Early Treatment for Infantile-Onset Pompe Disease Contributes toÂBetter Outcomes. Journal of Pediatrics, 2016, 169, 174-180.e1.	0.9	85
255	Neuropathology in respiratory-related motoneurons in young Pompe (Gaa) mice. Respiratory Physiology and Neurobiology, 2016, 227, 48-55.	0.7	41
256	Longitudinal follow-up to evaluate speech disorders in early-treated patients with infantile-onset Pompe disease. European Journal of Paediatric Neurology, 2017, 21, 485-493.	0.7	14
257	Clinical and Molecular Characterization of Infantile-Onset Pompe Disease in Mainland Chinese Patients: Identification of Two Common Mutations. Genetic Testing and Molecular Biomarkers, 2017, 21, 391-396.	0.3	12
258	Duvoglustat HCl Increases Systemic and Tissue Exposure of Active Acid α-Glucosidase in Pompe Patients Co-administered with Alglucosidase α. Molecular Therapy, 2017, 25, 1199-1208.	3.7	36
259	Glycogen Reduction in Myotubes of Late-Onset Pompe Disease Patients Using Antisense Technology. Molecular Therapy, 2017, 25, 2117-2128.	3.7	24
260	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.	0.5	7
261	Investigation on acute effects of enzyme replacement therapy and influence of clinical severity on physiological variables related to exercise tolerance in patients with late onset Pompe disease. Neuromuscular Disorders, 2017, 27, 542-549.	0.3	3

#	Article	IF	CITATIONS
262	Cardiac response to enzyme replacement therapy in infantile Pompe disease with severe hypertrophic cardiomyopathy. Echocardiography, 2017, 34, 621-624.	0.3	8
263	Insight into the phenotype of infants with Pompe disease identified by newborn screening with the common c32-13T > G "late-onset―GAA variant. Molecular Genetics and Metabolism, 2017, 122, 99-107.	0.5	26
264	A Skeletal Muscle Model of Infantile-onset Pompe Disease with Patient-specific iPS Cells. Scientific Reports, 2017, 7, 13473.	1.6	45
265	Biochemical study of glycogen storage disease type II (Pompe disease) in Egyptian infants. Middle East Journal of Medical Genetics, 2017, 6, 75-81.	0.0	1
266	Rapidly Progressive White Matter Involvement in Early Childhood: The Expanding Phenotype of Infantile Onset Pompe?. JIMD Reports, 2017, 39, 55-62.	0.7	19
267	Infantile Pompe Disease and Enzyme Replacement Therapy. Journal of Paediatrics and Child Health, 2017, 53, 1242-1243.	0.4	1
268	Selective Spinal Fusion for Neuromuscular Scoliosis in a Patient with Pompe Disease. JBJS Case Connector, 2017, 7, e15.	0.1	2
269	Enzyme replacement therapy for infantile-onset Pompe disease. The Cochrane Library, 2017, 2017, CD011539.	1.5	30
270	Long-term benefit of enzyme replacement therapy in Pompe disease. Neurology, 2017, 89, 2365-2373.	1.5	93
271	The Initial Evaluation of Patients After Positive Newborn Screening: Recommended Algorithms Leading to a Confirmed Diagnosis of Pompe Disease. Pediatrics, 2017, 140, S14-S23.	1.0	26
272	Bent spine syndrome as the initial symptom of lateâ€onset Pompe disease. Muscle and Nerve, 2017, 56, 167-170.	1.0	9
273	Quantification of muscle pathology in infantile Pompe disease. Neuromuscular Disorders, 2017, 27, 141-152.	0.3	18
274	Inspiratory muscle conditioning exercise and diaphragm gene therapy in Pompe disease: Clinical evidence of respiratory plasticity. Experimental Neurology, 2017, 287, 216-224.	2.0	37
275	Lipid Involvement in Neurodegenerative Diseases of the Motor System: Insights from Lysosomal Storage Diseases. Frontiers in Molecular Neuroscience, 2017, 10, 356.	1.4	32
276	Clinical Analysis of Algerian Patients with Pompe Disease. Journal of Neurodegenerative Diseases, 2017, 2017, 1-7.	1.1	3
277	The humanistic burden of Pompe disease: are there still unmet needs? A systematic review. BMC Neurology, 2017, 17, 202.	0.8	31
278	Pompe Disease: Cyanosed Hypotonic Infant with Normal Respiratory Rate. Kathmandu University Medical Journal, 2017, 13, 172-174.	0.1	1
279	Pompe disease, a storage cardiomyopathy. Neurology International, 2017, 7, .	0.2	5

#	Article	IF	CITATIONS
280	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
281	Perioperative management of children with glycogen storage disease type <scp>II</scp> —Pompe disease. Paediatric Anaesthesia, 2018, 28, 428-435.	0.6	5
282	Classic infantile Pompe patients approaching adulthood: a cohort study on consequences for the brain. Developmental Medicine and Child Neurology, 2018, 60, 579-586.	1.1	77
283	The phenotype, genotype, and outcome of infantile-onset Pompe disease in 18 Saudi patients. Molecular Genetics and Metabolism Reports, 2018, 15, 50-54.	0.4	9
284	Levels Above Lower Motor Neuron to Neuromuscular Junction. , 2018, , 887-921.e11.		0
285	Enzyme replacement therapy with alglucosidase alfa in Pompe disease: Clinical experience with rate escalation. Molecular Genetics and Metabolism, 2018, 123, 92-96.	0.5	12
286	Incidence of infantile Pompe disease in the Maroon population of French Guiana. BMJ Paediatrics Open, 2018, 2, e000182.	0.6	14
287	Longâ€ŧerm 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
288	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.	1.1	22
289	Neuroimaging findings in infantile Pompe patients treated with enzyme replacement therapy. Molecular Genetics and Metabolism, 2018, 123, 85-91.	0.5	39
290	Pompe disease in Austria: clinical, genetic and epidemiological aspects. Journal of Neurology, 2018, 265, 159-164.	1.8	29
291	The impact of Pompe disease on smooth muscle: a review. Journal of Smooth Muscle Research, 2018, 54, 100-118.	0.7	26
292	Enzyme replacement therapy for late-onset Pompe disease. The Cochrane Library, 0, , .	1.5	1
293	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
294	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
295	Satellite cells fail to contribute to muscle repair but are functional in Pompe disease (glycogenosis) Tj ETQq1 1	0.784314 r 2.4	gBT /Overloc
296	Post-mortem diagnosis of Pompe disease by exome sequencing in a Moroccan family: a case report. Journal of Medical Case Reports, 2018, 12, 322.	0.4	3
297	Preventing or Eradicating Factor VIII Antibody Formation in Patients with Hemophilia A: What Can We Learn from Other Disorders?. Seminars in Thrombosis and Hemostasis, 2018, 44, 531-543.	1.5	9

		CITATION REPORT		
#	ARTICLE Cardiac outcome in classic infantile Pompe disease after 13†years of treatment with i	recombinant	IF	CITATIONS
298	human acid alpha-glucosidase. International Journal of Cardiology, 2018, 269, 104-110.		0.8	32
299	Impact, Characterization, and Rescue of Pre-mRNA Splicing Mutations in Lysosomal Sto Genes, 2018, 9, 73.	rage Disorders.	1.0	11
300	Pompe Disease: From Basic Science to Therapy. Neurotherapeutics, 2018, 15, 928-942.		2.1	127
301	Neurometabolic Hereditary Diseases of Adults. , 2018, , .			2
302	Pompe Disease. , 2018, , 99-120.			0
303	microRNAs as biomarkers in Pompe disease. Genetics in Medicine, 2019, 21, 591-600.		1.1	22
304	Engineered skeletal muscles for disease modeling and drug discovery. Biomaterials, 201	9, 221, 119416.	5.7	74
305	<i>GAA</i> variants and phenotypes among 1,079 patients with Pompe disease: Data f Registry. Human Mutation, 2019, 40, 2146-2164.	rom the Pompe	1.1	51
306	Infantile-onset Pompe disease: Diagnosis and management. Archivos Argentinos De Pec 271-278.	diatria, 2019, 117,	0.3	13
307	Neuromuscular Diseases of the Newborn. Seminars in Pediatric Neurology, 2019, 32, 10	00771.	1.0	6
308	Clinical course, mutations and its functional characteristics of infantile-onset Pompe dis Thailand. BMC Medical Genetics, 2019, 20, 156.	sease in	2.1	7
309	Long-Term Observation of the Safety and Effectiveness of Enzyme Replacement Therap Patients with Pompe Disease: Results From the Post-marketing Surveillance. Neurology 2019, 8, 397-409.	y in Japanese and Therapy,	1.4	6
310	Characterization of immune response in Cross-Reactive Immunological Material (CRIM) infantile Pompe disease patients treated with enzyme replacement therapy. Molecular ( Metabolism Reports, 2019, 20, 100475.	-positive Genetics and	0.4	25
311	Advances in imaging of brain abnormalities in neuromuscular disease. Therapeutic Adva Neurological Disorders, 2019, 12, 175628641984556.	nces in	1.5	22
312	Clinical Gene Therapy Trials for Pompe Disease. , 2019, , 759-774.			0
313	Effects of immunomodulation in classic infantile Pompe patients with high antibody tite Journal of Rare Diseases, 2019, 14, 71.	ers. Orphanet	1.2	21
314	Characteristics of Pompe disease in China: a report from the Pompe registry. Orphanet Diseases, 2019, 14, 78.	Journal of Rare	1.2	14
315	Respiratory complications of metabolic disease in the paediatric population: A review of diagnosis and therapeutic options. Paediatric Respiratory Reviews, 2019, 32, 55-65.	presentation,	1.2	5

#	Article	IF	CITATIONS
316	Intravenous Injection of an AAV-PHP.B Vector Encoding Human Acid α-Glucosidase Rescues Both Muscle and CNS Defects in Murine Pompe Disease. Molecular Therapy - Methods and Clinical Development, 2019, 12, 233-245.	1.8	38
317	Challenges in treating Pompe disease: an industry perspective. Annals of Translational Medicine, 2019, 7, 291-291.	0.7	38
318	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.	1.1	28
319	Vacuolar hydrolysis and efflux: current knowledge and unanswered questions. Autophagy, 2019, 15, 212-227.	4.3	26
320	Skeletal alterations, developmental delay and new mutations in juvenile-onset Pompe disease. Neuromuscular Disorders, 2019, 29, 192-197.	0.3	8
321	Clinical and Molecular Disease Spectrum and Outcomes in Patients with Infantile-Onset Pompe Disease. Journal of Pediatrics, 2020, 216, 44-50.e5.	0.9	22
322	Molecular Approaches for the Treatment of Pompe Disease. Molecular Neurobiology, 2020, 57, 1259-1280.	1.9	17
323	An integrative correlation of myopathology, phenotype and genotype in late onset Pompe disease. Neuropathology and Applied Neurobiology, 2020, 46, 359-374.	1.8	13
324	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.	1.1	40
325	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
326	Advancements in AAV-mediated Gene Therapy for Pompe Disease. Journal of Neuromuscular Diseases, 2020, 7, 15-31.	1.1	41
327	Pulmonary outcome measures in longâ€ŧerm survivors of infantile Pompe disease on enzyme replacement therapy: A case series. Pediatric Pulmonology, 2020, 55, 674-681.	1.0	11
328	Lentiviral Hematopoietic Stem Cell Gene Therapy Corrects Murine Pompe Disease. Molecular Therapy - Methods and Clinical Development, 2020, 17, 1014-1025.	1.8	26
329	Orofacial features and pediatric dentistry in the long-term management of Infantile Pompe Disease children. Orphanet Journal of Rare Diseases, 2020, 15, 329.	1.2	3
330	Clinical and GAA gene mutation analysis in 21 Chinese patients with classic infantile pompe disease. European Journal of Medical Genetics, 2020, 63, 103997.	0.7	3
331	A Race Against Time—Changing the Natural History of CRIM Negative Infantile Pompe Disease. Frontiers in Immunology, 2020, 11, 1929.	2.2	6
332	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
333	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

#	ARTICLE	IF	CITATIONS
334	disease: results of a cross over study. Orphanet Journal of Rare Diseases, 2020, 15, 143.	1.2	15
335	Rescue of Advanced Pompe Disease in Mice with Hepatic Expression of Secretable Acid α-Glucosidase. Molecular Therapy, 2020, 28, 2056-2072.	3.7	16
336	The First Year Experience of Newborn Screening for Pompe Disease in California. International Journal of Neonatal Screening, 2020, 6, 9.	1.2	29
337	Diagnosis and Care of Infants and Children with Pompe Disease. Klinische Padiatrie, 2020, 232, 55-61.	0.2	3
338	The Respiratory Phenotype of Pompe Disease Mouse Models. International Journal of Molecular Sciences, 2020, 21, 2256.	1.8	8
339	Pulmonary Manifestations of Endocrine and Metabolic Diseases in Children. Pediatric Clinics of North America, 2021, 68, 81-102.	0.9	0
340	Cardiovascular disease in non-classic Pompe disease: A systematic review. Neuromuscular Disorders, 2021, 31, 79-90.	0.3	9
341	Deferoxamine mesylate improves splicing and GAA activity of the common c32-13T>G allele in late-onset PD patient fibroblasts. Molecular Therapy - Methods and Clinical Development, 2021, 20, 227-236.	1.8	5
342	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.	1.1	26
343	Endolysosomal N-glycan processing is critical to attain the most active form of the enzyme acid alpha-glucosidase. Journal of Biological Chemistry, 2021, 296, 100769.	1.6	5
344	Glycogen accumulation in smooth muscle of a Pompe disease mouse model. Journal of Smooth Muscle Research, 2021, 57, 8-18.	0.7	7
345	Chest MRI to diagnose early diaphragmatic weakness in Pompe disease. Orphanet Journal of Rare Diseases, 2021, 16, 21.	1.2	7
347	Uptake of mossâ€derived human recombinant GAA in Gaa â^'/â^' mice. JIMD Reports, 2021, 59, 81-89.	0.7	3
348	Variable Genotype–Phenotype Correlation of Pompe's Disease Caused by a c.2015 G > A (p.Arg672Gln Mutation in the GAA Gene. Neuropediatrics, 2021, 52, 475-479.	) <sub>0.3</sub>	0
349	Hypertrophic Cardiomyopathy in Children: Pathophysiology, Diagnosis, and Treatment of Non-sarcomeric Causes. Frontiers in Pediatrics, 2021, 9, 632293.	0.9	43
351	Lysosomal Storage Disorders in Children. , 2021, , 570-592.		0
353	An Overview of Benefits and Challenges of Rare Disease Biobanking in Africa, Focusing on South Africa. Biopreservation and Biobanking, 2021, 19, 143-150.	0.5	7
354	Genotype–phenotype correlation of 17 cases of Pompe disease in Spanish patients and identification of 4 novel GAA variants. Orphanet Journal of Rare Diseases, 2021, 16, 233.	1.2	4

ARTICLE IF CITATIONS # Experience with the Urinary Tetrasaccharide Metabolite for Pompe Disease in the Diagnostic 355 1.3 5 Laboratory. Metabolites, 2021, 11, 446. Hearing characteristics of infantile-onset Pompe disease after early enzyme-replacement therapy. 1.2 Orphanet Journal of Rare Diseases, 2021, 16, 348. Broad variation in phenotypes for common <i>GAA</i> genotypes in Pompe disease. Human Mutation, 357 1.1 4 2021, 42, 1461-1472. Genetic analysis of 76 Spanish Pompe disease patients: Identification of 12 novel pathogenic GAA 1.0 variants and functional characterization of splicing variants. Gene, 2022, 808, 145967. The Glycogen Storage Diseases and Related Disorders., 2006, , 101-119. 359 16 The Glycogen Storage Diseases and Related Disorders., 2016, , 121-137. 361 Cardiac Involvement by Systemic Diseases., 2008, , 1684-1706. 2 Neuromuscular Disorders: Levels above the Lower Motor Neuron to the Neuromuscular Junction., 2008, , 767-800. 363 Lysosomal Storage Diseases., 2012, , 403-451. 655 Pompe disease in adulthood: effects of antibody formation on enzyme replacement therapy. Genetics in 364 1.1 Medicine, 2017, 19, 90-97. Improved efficacy of a next-generation ERT in murine Pompe disease. JCI Insight, 2019, 4, . 365 2.357 Durable and sustained immune tolerance to ERT in Pompe disease with entrenched immune responses. 2.3 366 JCI Insight, 2016, 1, . The Pharmacological Chaperone AT2220 Increases Recombinant Human Acid α-Glucosidase Uptake and 367 1.1 73 Glycogen Reduction in a Mouse Model of Pompe Disease. PLoS ONE, 2012, 7, e40776. Immune Responses and Hypercoagulation in ERT for Pompe Disease Are Mutation and rhGAA Dose Dependent. PLoS ONE, 2014, 9, e98336. 1.1 The cost-effectiveness of enzyme replacement therapy (ERT) for the infantile form of Pompe disease: comparing a high-income country's approach (England) to that of a middle-income one 370 0.0 11 (Colombia). Revista De Salud Publica, 2012, 14, 143-155. Pompe disease: pathogenesis, molecular genetics and diagnosis. Aging, 2020, 12, 15856-15874. 33 Molecular genetics of Pompe disease: a comprehensive overview. Annals of Translational Medicine, 372 0.7 43 2019, 7, 278-278. An emerging phenotype of central nervous system involvement in Pompe disease: from bench to 373 bedside and beyond. Annals of Translational Medicine, 2019, 7, 289-289.

#	Article	IF	CITATIONS
374	Long-term outcome and unmet needs in infantile-onset Pompe disease. Annals of Translational Medicine, 2019, 7, 283-283.	0.7	40
375	Variable clinical features and genotype-phenotype correlations in 18 patients with late-onset Pompe disease. Annals of Translational Medicine, 2019, 7, 276-276.	0.7	13
376	Report of the first Brazilian infantile Pompe disease patient to be treated with recombinant human acid alpha-glucosidase. Jornal De Pediatria, 2008, 84, 272-275.	0.9	3
377	Clinical and molecular characterization of Korean children with infantile and late-onset Pompe disease: 10 years of experience with enzyme replacement therapy at a single center. Korean Journal of Pediatrics, 2019, 62, 224-234.	1.9	5
378	Infantile pompe disease presenting itself with severe hypertrophic cardiomyopathy: three case reports. Gaziantep Medical Journal, 2013, 19, 131.	0.2	1
379	Familial Pompe Disease. Medicinski Arhiv = Medical Archives = Archives De Médecine, 2015, 69, 342.	0.4	1
382	Infantile hypotonia with failure to thrive. American Journal of Case Reports, 2012, 13, 214-217.	0.3	0
385	Ocular Manifestations of Inborn Errors of Metabolism. , 2017, , 359-460.		Ο
387	Assessment of adeno-associated virus gene therapies efficacy on acid alpha-glucosidase restoration and glycogen storage correction in cardiac muscle of Pompe disease mice using synchrotron infrared and ultraviolet microspectroscopies. Journal of Spectral Imaging, 0, , .	0.0	1
388	A Newborn with Infantile-Onset Pompe Disease Improving after Administration of Enzyme Replacement Therapy: Case Report. Journal of Pediatric Intensive Care, 2022, 11, 062-066.	0.4	Ο
389	Cost-effectiveness analysis of enzyme replacement therapy (ERT) for treatment of infantile-onset Pompe disease (IOPD) in the Iranian pharmaceutical market. Intractable and Rare Diseases Research, 2020, 9, 130-136.	0.3	1
390	At-Risk Testing for Pompe Disease Using Dried Blood Spots: Lessons Learned for Newborn Screening. International Journal of Neonatal Screening, 2020, 6, 96.	1.2	1
391	Lysosomal disease. , 2020, , 2121-2156.		0
393	A review of treatment of Pompe disease in infants. Biologics: Targets and Therapy, 2007, 1, 195-201.	3.0	7
394	Glycogen storage disease type II: clinical overview. Acta Myologica, 2007, 26, 42-4.	1.5	24
395	Molecular genetics of late onset glycogen storage disease II in Italy. Acta Myologica, 2007, 26, 67-71.	1.5	10
396	A novel homozygous mutation at the GAA gene in Mexicans with early-onset Pompe disease. Acta Myologica, 2013, 32, 95-9.	1.5	3
397	Treatment strategies for acute metabolic disorders in neonates. Sudanese Journal of Paediatrics, 2011, 11, 6-13.	0.6	6

#	Article	IF	CITATIONS
398	Diagnostic methods for Lysosomal Storage Disease. Reports of Biochemistry and Molecular Biology, 2019, 7, 119-128.	0.5	11
399	Respiratory failure and sleep-disordered breathing in late-onset Pompe disease: a narrative review. Journal of Thoracic Disease, 2020, 12, S235-S247.	0.6	Ο
401	Effect of alglucosidase alfa dosage on survival and walking ability in patients with classic infantile Pompe disease: a multicentre observational cohort study from the European Pompe Consortium. The Lancet Child and Adolescent Health, 2022, 6, 28-37.	2.7	27
402	Respiratory failure and sleep-disordered breathing in late-onset Pompe disease: a narrative review. Journal of Thoracic Disease, 2020, 12, S235-S247.	0.6	11
403	Is the brain involved in patients with lateâ€onset Pompe disease?. Journal of Inherited Metabolic Disease, 2021, , .	1.7	3
404	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
405	Immune responses to alglucosidase in infantile Pompe disease: recommendations from an Italian pediatric expert panel. Italian Journal of Pediatrics, 2022, 48, 41.	1.0	6
406	Metabolic myopathies. Neurologie Pro Praxi, 2022, 23, 24-32.	0.0	1
407	The late-onset form of Pompe disease. Neurologie Pro Praxi, 2021, 22, 325-330.	0.0	0
408	What's new and what's next for gene therapy in Pompe disease?. Expert Opinion on Biological Therapy, 2022, 22, 1117-1135.	1.4	3
411	Advances in diagnosis and management of Pompe disease. Medycyna Wieku Rozwojowego, 2020, 24, 3-8.	0.2	6
412	Classic infantileâ€onset Pompe disease with histopathological neurologic findings linked to a novel <i>GAA</i> gene 4Âbp deletion: A case study. Molecular Genetics & Genomic Medicine, 2022, , e1957.	0.6	1
413	A case series of infantile Pompe disease at the university college hospital Ibadan Nigeria. Progress in Pediatric Cardiology, 2022, , 101538.	0.2	0
414	Lentiviral gene therapy prevents anti-human acid α-glucosidase antibody formation in murine Pompe disease. Molecular Therapy - Methods and Clinical Development, 2022, 25, 520-532.	1.8	9
416	Greater Efficacy of Avalglucosidase vs Alglucosidase Alfa in Adult Pompe Disease?. Neurology, 2022, 99, 183-184.	1.5	1
417	The Glycogen Storage Diseases and Related Disorders. , 2022, , 179-200.		2
418	A Multi-Centre Prospective Study of the Efficacy and Safety of Alglucosidase Alfa in Chinese Patients With Infantile-Onset Pompe Disease. Frontiers in Pharmacology, 0, 13, .	1.6	4
419	The earliest enzyme replacement for infantileâ€onset Pompe disease in Japan. Pediatrics International, 2022, 64, .	0.2	0

#	Article	IF	CITATIONS
420	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.	0.4	2
423	Enfermedad de Pompe de aparición tardÃa: primer reporte de caso en Perú. Revista Médica Basadrina, 2021, 15, 51-56.	0.0	0
424	Genotypic and phenotypic characteristics of 12 chinese children with glycogen storage diseases. Frontiers in Genetics, 0, 13, .	1.1	1
425	Long-term outcomes of very early treated infantile-onset Pompe disease with short-term steroid premedication: experiences from a nationwide newborn screening programme. Journal of Medical Genetics, 2023, 60, 430-439.	1.5	2
426	IGF2-tagging of GAA promotes full correction of murine Pompe disease at a clinically relevant dosage of lentiviral gene therapy. Molecular Therapy - Methods and Clinical Development, 2022, 27, 109-130.	1.8	9
427	A Roadmap for Potential Improvement of Newborn Screening for Inherited Metabolic Diseases Following Recent Developments and Successful Applications of Bivariate Normal Limits for Pre-Symptomatic Detection of MPS I, Pompe Disease, and Krabbe Disease. International Journal of Neonatal Screening, 2022, 8, 61.	1.2	6
428	Evaluating brain white matter hyperintensity, IQ scores, and plasma neurofilament light chain concentration in early-treated patients with infantile-onset Pompe disease. Genetics in Medicine, 2023, 25, 27-36.	1.1	5
429	Lysosomal Diseases. , 2023, , 977-1028.		0
430	Retrospective analysis of prenatal ultrasound of children with Pompe disease. Taiwanese Journal of Obstetrics and Gynecology, 2022, 61, 995-998.	0.5	0
431	Diffusion tensor imaging of the brain in Pompe disease. Journal of Neurology, 2023, 270, 1662-1671.	1.8	2
432	CRISPR-mediated generation and characterization of a Gaa homozygous c.1935C>A (p.D645E) Pompe disease knock-in mouse model recapitulating human infantile onset-Pompe disease. Scientific Reports, 2022, 12, .	1.6	3
433	Mass spectrometry for metabolomics analysis: Applications in neonatal and cancer screening. Mass Spectrometry Reviews, 0, , .	2.8	4
434	Induced pluripotent stem cell for modeling Pompe disease. Frontiers in Cardiovascular Medicine, 0, 9, .	1.1	0
435	Safety and efficacy of avalglucosidase alfa in individuals with infantile-onset Pompe disease enrolled in the phase 2, open-label Mini-COMET study: The 6-month primary analysis report. Genetics in Medicine, 2023, 25, 100328.	1.1	11
436	Blepharoptosis in infantile onset Pompe disease: Histological findings and surgical outcomes. Molecular Genetics and Metabolism Reports, 2023, 35, 100969.	0.4	0
437	Transcriptomic characterization of clinical skeletal muscle biopsy from late-onset Pompe patients. Molecular Genetics and Metabolism, 2023, 138, 107526.	0.5	2
438	Enzyme Replacement Therapy (ERT) on Heart Function Changes the Outcome in Patients with Infantile-Onset Pompe Disease: A Familial History. Case Reports in Pediatrics, 2023, 2023, 1-5.	0.2	0
439	Approved Protein Therapeutics and Their Biochemical Targets. , 2023, , 199-232.		0

#	Article	IF	CITATIONS
440	Unusual Evolution of Hypertrophic Cardiomyopathy in Non-Compaction Myocardium in a Pompe Disease Patient. Journal of Clinical Medicine, 2023, 12, 2365.	1.0	1
441	Combined targeted and untargeted high-resolution mass spectrometry analyses to investigate metabolic alterations in pompe disease. Metabolomics, 2023, 19, .	1.4	2
442	Pompe disease, a rare condition in two patients, case reports. , 2023, 1, .		0