# Ans T Van Der Ploeg

List of Publications by Year
in descending order



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|---|---|---|---|
| 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 | A Randomized Study of Alglucosidase Alfa in Late-Onset Pompe's Disease. New England Journal of Medicine, 2010, 362, 1396-1406. | 13.9 | 674 |
| 2 | Pompe's disease. Lancet, The, 2008, 372, 1342-1353. | 6.3 | 669 |
| 3 | 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 |
| 4 | Long-Term Intravenous Treatment of Pompe Disease With Recombinant Human Â-Glucosidase From Milk. Pediatrics, 2004, 113, e448-e457. | 1.0 | 326 |
| 5 | Recombinant human Î±-glucosidase from rabbit milk in Pompe patients. Lancet, The, 2000, 356, 397-398. | 6.3 | 321 |
| 6 | Clinical manifestation and natural course of late-onset Pompe's disease in 54 Dutch patients. Brain, 2005, 128, 671-677. | 3.7 | 310 |
| 7 | 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 |
| 8 | Early Treatment With Alglucosidase Alfa Prolongs Long-Term Survival of Infants With Pompe Disease. Pediatric Research, 2009, 66, 329-335. | 1.1 | 277 |
| 9 | 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 |
| 10 | Enzyme replacement therapy in late-onset Pompe's disease: A three-year follow-up. Annals of Neurology, 2004, 55, 495-502. | 2.8 | 221 |
| 11 | Disease severity in children and adults with Pompe disease related to age and disease duration. Neurology, 2005, 64, 2139-2141. | 1.5 | 159 |
| 12 | Broad spectrum of Pompe disease in patients with the same c.-32-13T->G haplotype. Neurology, 2007, 68, 110-115. | 1.5 | 154 |
| 13 | Glycogenosis type II (acid maltase deficiency). Muscle and Nerve, 1995, 18, S61-S69. | 1.0 | 149 |
| 14 | Pompe disease: Design, methodology, and early findings from the Pompe Registry. Molecular Genetics and Metabolism, 2011, 103, 1-11. | 0.5 | 130 |
| 15 | 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 |
| 16 | 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 |
| 17 | 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 |
| 18 | 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 |

| # | Article | IF | Citations |
|---|---|---|---|
| 19 | 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 |
| 20 | Late-onset Pompe disease primarily affects quality of life in physical health domains. Neurology, 2004, 63, 1688-1692. | 1.5 | 94 |
| 21 | 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 |
| 22 | 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 |
| 23 | Long-term benefit of enzyme replacement therapy in Pompe disease. Neurology, 2017, 89, 2365-2373. | 1.5 | 93 |
| 24 | 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 |
| 25 | 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 |
| 26 | 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 |
| 27 | 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 |
| 28 | 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 |
| 29 | 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 |
| 30 | 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 |
| 31 | 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 |
| 32 | Sharpening the Molecular Scissors: Advances in Gene-Editing Technology. IScience, 2020, 23, 100789. | 1.9 | 81 |
| 33 | 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 |
| 34 | Large variation in effects during 10 years of enzyme therapy in adults with Pompe disease. Neurology, 2019, 93, e1756-e1767. | 1.5 | 70 |
| 35 | Fatigue in neuromuscular disorders: focus on Guillainâ€"BarrÃ© syndrome and Pompe disease. Cellular and Molecular Life Sciences, 2010, 67, 701-713. | 2.4 | 68 |
| 36 | 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 |

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|---|---|---|---|
| 37 | 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 |
| 38 | 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 |
| 39 | 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 |
| 40 | 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 |
| 41 | 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 |
| 42 | 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 |
| 43 | Fatigue: an important feature of late-onset Pompe disease. Journal of Neurology, 2007, 254, 941-945. | 1.8 | 55 |
| 44 | Cardiac involvement in adults with Pompe disease. Journal of Internal Medicine, 2008, 264, 333-339. | 2.7 | 54 |
| 45 | 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 |
| 46 | Long-term outcomes of systemic therapies for Hurler syndrome: an international multicenter comparison. Genetics in Medicine, 2018, 20, 1423-1429. | 1.1 | 54 |
| 47 | Low bone mass in Pompe disease. Bone, 2010, 47, 643-649. | 1.4 | 53 |
| 48 | Pathological features of glycogen storage disease type II highlighted in the knockout mouse model. Journal of Pathology, 1999, 189, 416-424. | 2.1 | 52 |
| 49 | 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 |
| 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 variants and phenotypes among 1,079 patients with Pompe disease: Data from the Pompe Registry. Human Mutation, 2019, 40, 2146-2164. | 1.1 | 51 |
| 52 | 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 |
| 53 | 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 |
| 54 | Extension of the Pompe mutation database by linking diseaseâ€associated variants to clinical severity. Human Mutation, 2019, 40, 1954-1967. | 1.1 | 47 |

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|---|---|---|---|
| 55 | Cognitive profile and mental health in adult phenylketonuria: A PKU-COBESO study.. Neuropsychology, 2017, 31, 437-447. | 1.0 | 46 |
| 56 | Phenotypical variation within 22 families with Pompe disease. Orphanet Journal of Rare Diseases, 2013, 8, 182. | 1.2 | 45 |
| 57 | The Rasch-built Pompe-specific Activity (R-PAct) scale. Neuromuscular Disorders, 2013, 23, 256-264. | 0.3 | 45 |
| 58 | Childhood Pompe disease: clinical spectrum and genotype in 31 patients. Orphanet Journal of Rare Diseases, 2016, 11, 65. | 1.2 | 45 |
| 59 | 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 |
| 60 | Prospect for enzyme therapy in glycogenosis II variants: a study on cultured muscle cells. Journal of Neurology, 1988, 235, 392-396. | 1.8 | 43 |
| 61 | Receptor-Mediated Uptake of Acid Î±-Glucosidase Corrects Lysosomal Glycogen Storage in Cultured Skeletal Muscle. Pediatric Research, 1988, 24, 90-94. | 1.1 | 43 |
| 62 | 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 |
| 63 | Lung MRI and impairment of diaphragmatic function in Pompe disease. BMC Pulmonary Medicine, 2015, 15, 54. | 0.8 | 42 |
| 64 | 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 |
| 65 | 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 |
| 66 | Pompe disease in adulthood: effects of antibody formation on enzyme replacement therapy. Genetics in Medicine, 2017, 19, 90-97. | 1.1 | 41 |
| 67 | 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 |
| 68 | Up to five years experience with 11 mucopolysaccharidosis type VI patients. Molecular Genetics and Metabolism, 2013, 109, 70-76. | 0.5 | 35 |
| 69 | A Multiplex Assay for the Diagnosis of Mucopolysaccharidoses and Mucolipidoses. PLoS ONE, 2015, 10, e0138622. | 1.1 | 35 |
| 70 | 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 |
| 71 | 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 |
| 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 | 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 |
| 74 | A genetic modifier of symptom onset in Pompe disease. EBioMedicine, 2019, 43, 553-561. | 2.7 | 32 |
| 75 | Pain in adult patients with Pompe disease. Molecular Genetics and Metabolism, 2013, 109, 371-376. | 0.5 | 31 |
| 76 | 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 |
| 77 | 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 |
| 78 | Quantification of Diaphragm Mechanics in Pompe Disease Using Dynamic 3D MRI. PLoS ONE, 2016, 11, e0158912. | 1.1 | 30 |
| 79 | First experience with enzyme replacement therapy during pregnancy and lactation in Pompe disease. Molecular Genetics and Metabolism, 2011, 104, 552-555. | 0.5 | 29 |
| 80 | 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 |
| 81 | Enzyme replacement therapy and fatigue in adults with Pompe disease. Molecular Genetics and Metabolism, 2013, 109, 174-178. | 0.5 | 28 |
| 82 | Identification and Characterization of AberrantGAAPre-mRNA Splicing in Pompe Disease Using a Generic Approach. Human Mutation, 2015, 36, 57-68. | 1.1 | 28 |
| 83 | 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 |
| 84 | 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 |
| 85 | 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 |
| 86 | 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 |
| 87 | 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 |
| 88 | 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 |
| 89 | 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 |
| 90 | Severe tracheal and bronchial collapse in adults with type II mucopolysaccharidosis. Orphanet Journal of Rare Diseases, 2016, 11, 50. | 1.2 | 24 |

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|---|---|---|---|
| 91 | 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 |
| 92 | 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 |
| 93 | Treatment options for lysosomal storage disorders: developing insights. Expert Opinion on Pharmacotherapy, 2012, 13, 2281-2299. | 0.9 | 22 |
| 94 | microRNAs as biomarkers in Pompe disease. Genetics in Medicine, 2019, 21, 591-600. | 1.1 | 22 |
| 95 | 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 |
| 96 | Imaging of respiratory muscles in neuromuscular disease: A review. Neuromuscular Disorders, 2018, 28, 246-256. | 0.3 | 21 |
| 97 | Effects of immunomodulation in classic infantile Pompe patients with high antibody titers. Orphanet Journal of Rare Diseases, 2019, 14, 71. | 1.2 | 21 |
| 98 | Monitoring of pulmonary function in Pompe disease: a muscle disease with new therapeutic perspectives. European Respiratory Journal, 2005, 26, 984-985. | 3.1 | 20 |
| 99 | 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 |
| 100 | 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 |
| 101 | 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 |
| 102 | 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 |
| 103 | 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 |
| 104 | Residual Nâ€acetylâ€Î±â€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 |
| 105 | The Dilemma of Two Innovative Therapies for Spinal Muscular Atrophy. New England Journal of Medicine, 2017, 377, 1786-1787. | 13.9 | 17 |
| 106 | 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 |
| 107 | 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 |
| 108 | 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 |

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|---|---|---|---|
| 109 | Longâ€term cognitive followâ€up in children treated for Maroteauxâ€Lamy syndrome. Journal of Inherited Metabolic Disease, 2016, 39, 285-292. | 1.7 | 14 |
| 110 | Mucolipidosis type III, a series of adult patients. Journal of Inherited Metabolic Disease, 2018, 41, 839-848. | 1.7 | 14 |
| 111 | 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 |
| 112 | Enzymatic diagnosis of Pompe disease: lessons from 28 years of experience. European Journal of Human Genetics, 2021, 29, 434-446. | 1.4 | 13 |
| 113 | Increased aortic stiffness and blood pressure in nonâ€classic Pompe disease. Journal of Inherited Metabolic Disease, 2014, 37, 391-397. | 1.7 | 12 |
| 114 | Mucolipidosis type II and type III: a systematic review of 843 published cases. Genetics in Medicine, 2021, 23, 2047-2056. | 1.1 | 12 |
| 115 | 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 |
| 116 | Enzyme replacement therapy reduces the risk for wheelchair dependency in adult Pompe patients. Orphanet Journal of Rare Diseases, 2018, 13, 82. | 1.2 | 11 |
| 117 | A conceptual disease model for adult Pompe disease. Orphanet Journal of Rare Diseases, 2015, 10, 112. | 1.2 | 10 |
| 118 | Hip disease in Mucopolysaccharidoses and Mucolipidoses: A review of mechanisms, interventions and future perspectives. Bone, 2021, 143, 115729. | 1.4 | 10 |
| 119 | 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 |
| 120 | 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 |
| 121 | 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 |
| 122 | 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 |
| 123 | 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 |
| 124 | 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 |
| 125 | Presymptomatic treatment of classic late-infantile neuronal ceroid lipofuscinosis with cerliponase alfa. Orphanet Journal of Rare Diseases, 2021, 16, 221. | 1.2 | 8 |
| 126 | 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 |

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|---|---|---|---|
| 127 | Chest MRI to diagnose early diaphragmatic weakness in Pompe disease. Orphanet Journal of Rare Diseases, 2021, 16, 21. | 1.2 | 7 |
| 128 | Remarkably low fibroblast acid Î±-glucosidase activity in three adults with Pompe disease. Molecular Genetics and Metabolism, 2012, 107, 485-489. | 0.5 | 6 |
| 129 | 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 |
| 130 | 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 |
| 131 | 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 |
| 132 | Association of Muscle Strength and Walking Performance in Adult Patients With Pompe Disease. Physical Therapy, 2018, 98, 925-931. | 1.1 | 4 |
| 133 | Broad variation in phenotypes for common GAA genotypes in Pompe disease. Human Mutation, 2021, 42, 1461-1472. | 1.1 | 4 |
| 134 | Delayed Diagnosis of Danon Disease in Patients Presenting With Isolated Cardiomyopathy. Circulation Genomic and Precision Medicine, 2019, 12, e002395. | 1.6 | 3 |
| 135 | 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 |
| 136 | Is the brain involved in patients with lateâ€onset Pompe disease?. Journal of Inherited Metabolic Disease, 2021, , . | 1.7 | 3 |
| 137 | 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 |
| 138 | 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 |
| 139 | Effect of anti-iduronidase sulfatase in patients with Mucopolysaccharidosis type II treated with enzyme replacement therapy. Journal of Pediatrics, 2022, , . | 0.9 | 1 |
| 140 | Pompe Disease: A Continuum of Clinical Phenotypes. Clinical Therapeutics, 2007, 29, S103-S104. | 1.1 | 0 |
| 141 | Commentary. Clinical Chemistry, 2017, 63, 48-48. | 1.5 | 0 |
| 142 | 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 |
| 143 | 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 |