W W M Pim Pijnappel

List of Publications by Year
in descending order

45 papers	1,235 citations	331670 21 h-index	377865 34 g-index
45 all docs	45 docs citations	45 times ranked	1825 citing authors

#	Article	IF	Citations
1	Antibodies against recombinant human alpha-glucosidase do not seem to affect clinical outcome in childhood onset Pompe disease. Orphanet Journal of Rare Diseases, 2022, 17, 31.	2.7	5
2	Generation of Human iPSC-Derived Myotubes to Investigate RNA-Based Therapies In Vitro. Methods in Molecular Biology, 2022, 2434, 235-243.	0.9	2
3	Lentiviral gene therapy prevents anti-human acid Î±-glucosidase antibody formation in murine Pompe disease. Molecular Therapy - Methods and Clinical Development, 2022, 25, 520-532.	4.1	9
4	Effect of anti-iduronidase sulfatase in patients with Mucopolysaccharidosis type II treated with enzyme replacement therapy. Journal of Pediatrics, 2022, , .	1.8	1
5	Opportunities and challenges for antisense oligonucleotide therapies. Journal of Inherited Metabolic Disease, 2021, 44, 72-87.	3.6	86
6	Hip disease in Mucopolysaccharidoses and Mucolipidoses: A review of mechanisms, interventions and future perspectives. Bone, 2021, 143, 115729.	2.9	10
7	A generic assay for the identification of splicing variants that induce nonsense-mediated decay in Pompe disease. European Journal of Human Genetics, 2021, 29, 422-433.	2.8	6
8	Enzymatic diagnosis of Pompe disease: lessons from 28 years of experience. European Journal of Human Genetics, 2021, 29, 434-446.	2.8	13
9	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.	2.5	19
10	CRISPR-Cas9-Mediated Gene Editing in Human Induced Pluripotent Stem Cells. Springer Protocols, 2021, , 235-264.	0.3	1
11	An in vitro assay to quantify satellite cell activation using isolated mouse myofibers. STAR Protocols, 2021, 2, 100482.	1.2	0
12	Broad variation in phenotypes for common GAA genotypes in Pompe disease. Human Mutation, 2021, 42, 1461-1472.	2.5	4
13	Sharpening the Molecular Scissors: Advances in Gene-Editing Technology. IScience, 2020, 23, 100789.	4.1	81
14	Coupling 3D Printing and Novel Replica Molding for In House Fabrication of Skeletal Muscle Tissue Engineering Devices. Advanced Materials Technologies, 2020, 5, 2000344.	5.8	28
15	Generation of genomic-integration-free human induced pluripotent stem cells and the derived cardiomyocytes of X-linked dilated cardiomyopathy from DMD gene mutation. Stem Cell Research, 2020, 49, 102040.	0.7	2
16	A Generic Assay to Detect Aberrant ARSB Splicing and mRNA Degradation for the Molecular Diagnosis of MPS VI. Molecular Therapy - Methods and Clinical Development, 2020, 19, 174-185.	4.1	7
17	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.	3.6	22
18	Ready for Repair? Gene Editing Enters the Clinic for the Treatment of Human Disease. Molecular Therapy - Methods and Clinical Development, 2020, 18, 532-557.	4.1	67

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19	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.	4.1	15
20	Extension of the Pompe mutation database by linking diseaseâ€associated variants to clinical severity. Human Mutation, 2019, 40, 1954-1967.	2.5	47
21	Modelling the neuropathology of lysosomal storage disorders through disease-specific human induced pluripotent stem cells. Experimental Cell Research, 2019, 380, 216-233.	2.6	28
22	A genetic modifier of symptom onset in Pompe disease. EBioMedicine, 2019, 43, 553-561.	6.1	32
23	Segmental and total uniparental isodisomy (UPiD) as a disease mechanism in autosomal recessive lysosomal disorders: evidence from SNP arrays. European Journal of Human Genetics, 2019, 27, 919-927.	2.8	8
24	Front Cover, Volume 40, Issue 11. Human Mutation, 2019, 40, i.	2.5	0
25	Restoring the regenerative balance in neuromuscular disorders: satellite cell activation as therapeutic target in Pompe disease. Annals of Translational Medicine, 2019, 7, 280-280.	1.7	8
26	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.	1.8	27
27	Satellite cells maintain regenerative capacity but fail to repair disease-associated muscle damage in mice with Pompe disease. Acta Neuropathologica Communications, 2018, 6, 119.	5.2	28
28	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.	2.5	9
29	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.	4.8	81
30	Alternative Splicing in Genetic Diseases: Improved Diagnosis and Novel Treatment Options. International Review of Cell and Molecular Biology, 2018, 335, 85-141.	3.2	23
31	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
32	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.	2.1	28
33	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.	5.1	52
34	GAA Deficiency in Pompe Disease Is Alleviated by Exon Inclusion in iPSC-Derived Skeletal Muscle Cells. Molecular Therapy - Nucleic Acids, 2017, 7, 101-115.	5.1	56
35	Commentary. Clinical Chemistry, 2017, 63, 48-48.	3.2	0
36	Response to Herbert et al.. Genetics in Medicine, 2017, 19, 1282-1283.	2.4	0

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37	Pompe disease in adulthood: effects of antibody formation on enzyme replacement therapy. Genetics in Medicine, 2017, 19, 90-97.	2.4	41
38	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.	5.1	29
39	Elevated Plasma Cardiac Troponin T Levels Caused by Skeletal Muscle Damage in Pompe Disease. Circulation: Cardiovascular Genetics, 2016, 9, 6-13.	5.1	70
40	Identification and Characterization of AberrantGAAPre-mRNA Splicing in Pompe Disease Using a Generic Approach. Human Mutation, 2015, 36, 57-68.	2.5	28
41	Lack of robust satellite cell activation and muscle regeneration during the progression of Pompe disease. Acta Neuropathologica Communications, 2015, 3, 65.	5.2	32
42	Exercise Training in Adults With Pompe Disease: TheÂEffects on Pain, Fatigue, and Functioning. Archives of Physical Medicine and Rehabilitation, 2015, 96, 817-822.	0.9	30
43	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.	6.5	20
44	Epigenetic Characterization of the FMR1 Promoter in Induced Pluripotent Stem Cells from Human Fibroblasts Carrying an Unmethylated Full Mutation. Stem Cell Reports, 2014, 3, 548-555.	4.8	54
45	Engineering the mouse genome with bacterial artificial chromosomes to create multipurpose alleles. Nature Biotechnology, 2003, 21, 443-447.	17.5	126