Joseph Muenzer

List of Publications by Year
in descending order

52	5,661	147566	189595
papers	citations	31 h-index	50 g-index

54	54	54	2775
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Long-term open-label phase I/II extension study of intrathecal idursulfase-IT in the treatment of neuronopathic mucopolysaccharidosis II. Genetics in Medicine, 2022, 24, 1437-1448.	1.1	6
2	Improvement in time to treatment, but not time to diagnosis, in patients with mucopolysaccharidosis type I. Archives of Disease in Childhood, 2021, 106, 674-679.	1.0	13
3	A multicenter open-label extension study of intrathecal heparan-N-sulfatase in patients with Sanfilippo syndrome type A. Molecular Genetics and Metabolism, 2021, 134, 175-181.	0.5	4
4	Evaluation of the long-term treatment effects of intravenous idursulfase in patients with mucopolysaccharidosis II (MPS II) using statistical modeling: data from the Hunter Outcome Survey (HOS). Orphanet Journal of Rare Diseases, 2021, 16, 456.	1.2	15
5	Mucopolysaccharidosis Type I Diagnosed by Aortic and Mitral Valve Replacement. JACC: Case Reports, 2021, 3, 1891-1894.	0.3	3
6	Therapy development for the mucopolysaccharidoses: Updated consensus recommendations for neuropsychological endpoints. Molecular Genetics and Metabolism, 2020, 131, 181-196.	0.5	26
7	Idursulfase pharmacokinetics, cellular uptake, and pharmacodynamics: Effect of sialylation and manufacturing process. Engineering Reports, 2020, 2, e12271.	0.9	0
8	Metabonomics reveals altered metabolites related to inflammation and energy utilization at recovery of cystic fibrosis lung exacerbation. Metabolism Open, 2019, 3, 100010.	1.4	8
9	Growth patterns for untreated individuals with MPS I: Report from the international MPS I registry. American Journal of Medical Genetics, Part A, 2019, 179, 2425-2432.	0.7	16
10	The North Carolina Experience with Mucopolysaccharidosis Type I Newborn Screening. Journal of Pediatrics, 2019, 211, 193-200.e2.	0.9	22
11	Recommendations for the management of MPS VI: systematic evidence- and consensus-based guidance. Orphanet Journal of Rare Diseases, 2019, 14, 118.	1.2	30
12	Recommendations for the management of MPS IVA: systematic evidence- and consensus-based guidance. Orphanet Journal of Rare Diseases, 2019, 14, 137.	1.2	62
13	Genotypeâ€phenotype relationships in mucopolysaccharidosis type I (MPS I): Insights from the International MPS I Registry. Clinical Genetics, 2019, 96, 281-289.	1.0	54
14	Intrathecal heparan-N-sulfatase in patients with Sanfilippo syndrome type A: A phase IIb randomized trial. Molecular Genetics and Metabolism, 2019, 126, 121-130.	0.5	37
15	Targeting Root Cause by Systemic scAAV9-hIDS Gene Delivery: Functional Correction and Reversal of Severe MPS II in Mice. Molecular Therapy - Methods and Clinical Development, 2018, 10, 327-340.	1.8	14
16	Cognitive endpoints for therapy development for neuronopathic mucopolysaccharidoses: Results of a consensus procedure. Molecular Genetics and Metabolism, 2017, 121, 70-79.	0.5	35
17	Mucopolysaccharidosis Type I Newborn Screening: Best Practices for Diagnosis and Management. Journal of Pediatrics, 2017, 182, 363-370.	0.9	65
18	Carpal tunnel syndrome in mucopolysaccharidosis I: a registryâ€based cohort study. Developmental Medicine and Child Neurology, 2017, 59, 1269-1275.	1.1	24

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19	Co-occurring Down syndrome and SUCLA2 -related mitochondrial depletion syndrome. , 2017, 173, 2720-2724.		2
20	Ten years of the Hunter Outcome Survey (HOS): insights, achievements, and lessons learned from a global patient registry. Orphanet Journal of Rare Diseases, 2017, 12, 82.	1.2	48
21	Clinical outcomes in idursulfase-treated patients with mucopolysaccharidosis type II: 3-year data from the hunter outcome survey (HOS). Orphanet Journal of Rare Diseases, 2017, 12, 161.	1.2	48
22	A phase I/II study of intrathecal idursulfase-IT in children with severe mucopolysaccharidosis II. Genetics in Medicine, 2016, 18, 73-81.	1.1	138
23	Progression of Polysomnographic Abnormalities in Mucolipidosis II (I-Cell Disease). Journal of Clinical Sleep Medicine, 2016, 12, 1695-1696.	1.4	5
24	Levels of glycosaminoglycans in the cerebrospinal fluid of healthy young adults, surrogate-normal children, and Hunter syndrome patients with and without cognitive impairment. Molecular Genetics and Metabolism Reports, 2015, 5, 103-106.	0.4	24
25	A Cerebrospinal Fluid Collection Study in Pediatric and Adult Patients With Hunter Syndrome. FIRE Forum for International Research in Education, 2015, 3, 232640981559582.	0.7	6
26	The natural history of MPS I: global perspectives from the MPS I Registry. Genetics in Medicine, 2014, 16, 759-765.	1.1	127
27	Enzyme Replacement Therapy in Mucopolysaccharidosis II Patients Under 1 Year of Age. JIMD Reports, 2014, 14, 99-113.	0.7	40
28	The Hunter Syndrome-Functional Outcomes for Clinical Understanding Scale (HS-FOCUS) Questionnaire: item reduction and further validation. Quality of Life Research, 2014, 23, 2457-2462.	1.5	16
29	Early initiation of enzyme replacement therapy for the mucopolysaccharidoses. Molecular Genetics and Metabolism, 2014, 111, 63-72.	0.5	151
30	Relationship of Sleep to Pulmonary Function in Mucopolysaccharidosis II. Journal of Pediatrics, 2013, 162, 1210-1215.	0.9	18
31	Bronchoscopy and airway management in patients with mucopolysaccharidoses (MPS). Pediatric Pulmonology, 2013, 48, 601-607.	1.0	29
32	Patient-Specific Biomechanical Modeling of Ventricular Enlargement in Hydrocephalus from Longitudinal Magnetic Resonance Imaging. Lecture Notes in Computer Science, 2013, 16, 291-298.	1.0	1
33	The role of enzyme replacement therapy in severe Hunter syndromeâ€"an expert panel consensus. European Journal of Pediatrics, 2012, 171, 181-188.	1.3	86
34	Respiratory Manifestations in Mucopolysaccharidoses. Paediatric Respiratory Reviews, 2011, 12, 133-138.	1.2	96
35	Idursulfase treatment of Hunter syndrome in children younger than 6 years: Results from the Hunter Outcome Survey. Genetics in Medicine, 2011, 13, 102-109.	1.1	86
36	Long-term, open-labeled extension study of idursulfase in the treatment of Hunter syndrome. Genetics in Medicine, 2011, 13, 95-101.	1.1	190

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37	Overview of the mucopolysaccharidoses. Rheumatology, 2011, 50, v4-v12.	0.9	384
38	Mucopolysaccharidosis I: Management and Treatment Guidelines. Pediatrics, 2009, 123, 19-29.	1.0	400
39	Multidisciplinary Management of Hunter Syndrome. Pediatrics, 2009, 124, e1228-e1239.	1.0	159
40	Long-term Efficacy and Safety of Laronidase in the Treatment of Mucopolysaccharidosis I. Pediatrics, 2009, 123, 229-240.	1.0	301
41	Hunter syndrome: to treat or not to treat. Acta Paediatrica, International Journal of Paediatrics, 2008, 97, 55-56.	0.7	47
42	Recognition and Diagnosis of Mucopolysaccharidosis II (Hunter Syndrome). Pediatrics, 2008, 121, e377-e386.	1.0	260
43	Initial report from the Hunter Outcome Survey. Genetics in Medicine, 2008, 10, 508-516.	1.1	160
44	A phase I/II clinical trial of enzyme replacement therapy in mucopolysaccharidosis II (Hunter) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50 462 T	0.5	195
45	The MPS I registry: Design, methodology, and early findings of a global disease registry for monitoring patients with Mucopolysaccharidosis Type I. Molecular Genetics and Metabolism, 2007, 91, 37-47.	0.5	172
46	Preclinical dose ranging studies for enzyme replacement therapy with idursulfase in a knock-out mouse model of MPS II. Molecular Genetics and Metabolism, 2007, 91, 183-190.	0.5	45
47	A phase II/III clinical study of enzyme replacement therapy with idursulfase in mucopolysaccharidosis II (Hunter syndrome). Genetics in Medicine, 2006, 8, 465-473.	1.1	499
48	Advances in the Treatment of Mucopolysaccharidosis Type I. New England Journal of Medicine, 2004, 350, 1932-1934.	13.9	74
49	Enzyme replacement therapy for mucopolysaccharidosis I: a randomized, double-blinded, placebo-controlled, multinational study of recombinant human Î±-L-iduronidase (laronidase). Journal of Pediatrics, 2004, 144, 581-588.	0.9	514
50	The mucopolysaccharidoses: a heterogeneous group of disorders with variable pediatric presentations. Journal of Pediatrics, 2004, 144, S27-S34.	0.9	224
51	Enzyme-Replacement Therapy in Mucopolysaccharidosis I. New England Journal of Medicine, 2001, 344, 182-188.	13.9	619
52	Electroretinographic Findings in the Mucopolysaccharidoses. Ophthalmology, 1986, 93, 1612-1616.	2.5	63