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List of Publications by Year in Descending Order

90 papers	3,068 citations	29 h-index	53 g-index
117 ext. papers	3,700 ext. citations	3.1 avg, IF	5.04 L-index

#	Paper	IF	Citations
90	Addressing the need for patient-friendly medical communications: adaptation of the 2019 recommendations for the management of MPS VI and MPS IVA.. *Orphanet Journal of Rare Diseases*, **2022**, 17, 91	4.2	0
89	An Overview of Benefits and Challenges of Rare Disease Biobanking in Africa, Focusing on South Africa. *Biopreservation and Biobanking*, **2021**, 19, 143-150	2.1	3
88	Are there common walking gait characteristics in patients diagnosed with late-onset Pompe disease?. *Human Movement Science*, **2021**, 77, 102777	2.4	1
87	Clinical outcomes in an adult patient with mannose phosphate isomerase-congenital disorder of glycosylation who discontinued mannose therapy. *Molecular Genetics and Metabolism Reports*, **2020** , 25, 100646	1.8	0
86	Toileting Abilities Survey as a surrogate outcome measure for cognitive function: Findings from neuronopathic mucopolysaccharidosis II patients treated with idursulfase and intrathecal idursulfase. *Molecular Genetics and Metabolism Reports*, **2020**, 25, 100669	1.8	1
85	Critical clinical situations in adult patients with Mucopolysaccharidoses (MPS). *Orphanet Journal of Rare Diseases*, **2020**, 15, 114	4.2	6
84	Cardiac rhythm abnormalities - An underestimated cardiovascular risk in adult patients with Mucopolysaccharidoses. *Molecular Genetics and Metabolism*, **2020**, 130, 133-139	3.7	2
83	Mechanisms of Mitochondrial Dysfunction in Lysosomal Storage Disorders: A Review. *Journal of Clinical Medicine*, **2020**, 9,	5.1	18
82	Disease progression of alpha-mannosidosis and impact on patients and carers - A UK natural history survey. *Molecular Genetics and Metabolism Reports*, **2019**, 20, 100480	1.8	8
81	Recommendations for the management of MPS VI: systematic evidence- and consensus-based guidance. *Orphanet Journal of Rare Diseases*, **2019**, 14, 118	4.2	18
80	Recommendations for the management of MPS IVA: systematic evidence- and consensus-based guidance. *Orphanet Journal of Rare Diseases*, **2019**, 14, 137	4.2	36
79	Recommendations for patient screening in ultra-rare inherited metabolic diseases: what have we learned from Niemann-Pick disease type C?. *Orphanet Journal of Rare Diseases*, **2019**, 14, 20	4.2	11
78	Challenges in diagnosing and managing adult patients with urea cycle disorders. *Journal of Inherited Metabolic Disease*, **2019**, 42, 1136-1146	5.4	15
77	Potential benefits of Fitbit device in managing a patient with mucopolysaccharidosis. *Molecular Genetics and Metabolism*, **2019**, 126, S111	3.7	3
76	Transition of patients with mucopolysaccharidosis from paediatric to adult care. *Molecular Genetics and Metabolism Reports*, **2019**, 21, 100508	1.8	9
75	Phenotype, treatment practice and outcome in the cobalamin-dependent remethylation disorders and MTHFR deficiency: Data from the E-HOD registry. *Journal of Inherited Metabolic Disease*, **2019**, 42, 333-352	5.4	28
74	Safety, immunogenicity, and clinical outcomes in patients with Morquio A syndrome participating in 2 sequential open-label studies of elosulfase alfa enzyme replacement therapy (MOR-002/MOR-100), representing 5 years of treatment. *Molecular Genetics and Metabolism*, **2018**, 123, 479-487	3.7	19

73	Impact of long-term elosulfase alfa on activities of daily living in patients with Morquio A syndrome in an open-label, multi-center, phase 3 extension study. *Molecular Genetics and Metabolism*, **2018**, 123, 127-134	3.7	18
72	Enzyme replacement therapy with velmanase alfa (human recombinant alpha-mannosidase): Novel global treatment response model and outcomes in patients with alpha-mannosidosis. *Molecular Genetics and Metabolism*, **2018**, 124, 152-160	3.7	19
71	Risks of long-term port use in enzyme replacement therapy for lysosomal storage disorders. *Molecular Genetics and Metabolism Reports*, **2018**, 15, 71-73	1.8	4
70	Consensus clinical management guidelines for Niemann-Pick disease type C. *Orphanet Journal of Rare Diseases*, **2018**, 13, 50	4.2	124
69	Haematopoietic Stem Cell Transplantation Arrests the Progression of Neurodegenerative Disease in Late-Onset Tay-Sachs Disease. *JIMD Reports*, **2018**, 41, 17-23	1.9	12
68	Healthcare resource use and costs of managing children and adults with lysosomal acid lipase deficiency at a tertiary referral centre in the United Kingdom. *PLoS ONE*, **2018**, 13, e0191945	3.7	2
67	Oculomotor abnormalities in children with Niemann-Pick type C. *Molecular Genetics and Metabolism* , **2018**, 123, 159-168	3.7	11
66	Enzyme replacement therapy for late-onset Pompe disease. *The Cochrane Library*, **2018**,	5.2	78
65	Markers of cognitive function in individuals with metabolic disease: Morquio syndrome and tyrosinemia type III. *Cognitive Neuropsychology*, **2018**, 35, 120-147	2.3	6
64	Differences in Niemann-Pick disease Type C symptomatology observed in patients of different ages. *Molecular Genetics and Metabolism*, **2017**, 120, 180-189	3.7	24
63	Evaluation of impact of anti-idursulfase antibodies during long-term idursulfase enzyme replacement therapy in mucopolysaccharidosis II patients. *Molecular Genetics and Metabolism Reports*, **2017**, 12, 2-7	1.8	10
62	The hidden Niemann-Pick type C patient: clinical niches for a rare inherited metabolic disease. *Current Medical Research and Opinion*, **2017**, 33, 877-890	2.5	18
61	Diagnosing childhood-onset inborn errors of metabolism by next-generation sequencing. *Archives of Disease in Childhood*, **2017**, 102, 1019-1029	2.2	24
60	The effect of galsulfase enzyme replacement therapy on the growth of patients with mucopolysaccharidosis VI (Maroteaux-Lamy syndrome). *Molecular Genetics and Metabolism*, **2017**, 122, 107-112	3.7	29
59	The factors affecting lipid profile in adult patients with Mucopolysaccharidosis. *Molecular Genetics and Metabolism Reports*, **2017**, 12, 35-40	1.8	4
58	Impact of elosulfase alfa in patients with morquio A syndrome who have limited ambulation: An open-label, phase 2 study. *American Journal of Medical Genetics, Part A*, **2017**, 173, 375-383	2.5	17
57	The use of port-a-caths in adult patients with Lysosomal Storage Disorders receiving Enzyme Replacement Therapy-one centre experience. *Molecular Genetics and Metabolism Reports*, **2017**, 13, 111-114	1.8	4
56	Mortality in patients with Sanfilippo syndrome. *Orphanet Journal of Rare Diseases*, **2017**, 12, 168	4.2	29

55	The Challenges of a Successful Pregnancy in a Patient with Adult Refsum's Disease due to Phytanoyl-CoA Hydroxylase Deficiency. *JIMD Reports*, **2017**, 33, 49-53	1.9	3
54	Lipid profile in adult patients with Fabry disease - Ten-year follow up. *Molecular Genetics and Metabolism Reports*, **2017**, 13, 3-6	1.8	4
53	Niemann-Pick type C disease - the tip of the iceberg? A review of neuropsychiatric presentation, diagnosis and treatment. *BJPsych Bulletin*, **2017**, 41, 109-114	1.2	24
52	Synchrotron X-ray diffraction to understand crystallographic texture of enamel affected by Hunter syndrome. *Archives of Oral Biology*, **2017**, 80, 193-196	2.8	2
51	Rapid progression and mortality of lysosomal acid lipase deficiency presenting in infants. *Genetics in Medicine*, **2016**, 18, 452-8	8.1	42
50	A phase I/II study of intrathecal idursulfase-IT in children with severe mucopolysaccharidosis II. *Genetics in Medicine*, **2016**, 18, 73-81	8.1	111
49	Health-related quality of life in mucopolysaccharidosis: looking beyond biomedical issues. *Orphanet Journal of Rare Diseases*, **2016**, 11, 119	4.2	25
48	Synchrotron X-ray diffraction and scanning electron microscopy to understand enamel affected by metabolic disorder mucopolysaccharidosis. *Micron*, **2016**, 83, 48-53	2.3	3
47	Observational clinical study of 22 adult-onset Pompe disease patients undergoing enzyme replacement therapy over 5years. *Molecular Genetics and Metabolism*, **2016**, 117, 413-8	3.7	31
46	Diagnostic tests for Niemann-Pick disease type C (NP-C): A critical review. *Molecular Genetics and Metabolism*, **2016**, 118, 244-54	3.7	97
45	Elosulfase alfa (BMN 110) for the treatment of mucopolysaccharidosis IVA (Morquio A Syndrome). *Expert Review of Clinical Pharmacology*, **2016**, 9, 1521-1532	3.8	20
44	Impact of long-term elosulfase alfa treatment on respiratory function in patients with Morquio A syndrome. *Journal of Inherited Metabolic Disease*, **2016**, 39, 839-847	5.4	18
43	Long-term endurance and safety of elosulfase alfa enzyme replacement therapy in patients with Morquio A syndrome. *Molecular Genetics and Metabolism*, **2016**, 119, 131-43	3.7	42
42	Cervical cord compression in mucopolysaccharidosis VI (MPS VI): Findings from the MPS VI Clinical Surveillance Program (CSP). *Molecular Genetics and Metabolism*, **2016**, 118, 310-8	3.7	31
41	Recommendations for initiation and cessation of enzyme replacement therapy in patients with Fabry disease: the European Fabry Working Group consensus document. *Orphanet Journal of Rare Diseases*, **2015**, 10, 36	4.2	173
40	Objective results of median nerve decompression and tenosynovectomy for carpal tunnel syndrome in patients with mucopolysaccharidoses Types I and II. *Journal of Hand Surgery: European Volume*, **2015**, 40, 216-8	1.4	11
39	Glycogen storage disease. *Paediatrics and Child Health (United Kingdom)*, **2015**, 25, 139-144	0.6	4
38	International guidelines for the management and treatment of Morquio A syndrome. *American Journal of Medical Genetics, Part A*, **2015**, 167A, 11-25	2.5	86

37	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	1.8	18
36	A Cerebrospinal Fluid Collection Study in Pediatric and Adult Patients With Hunter Syndrome. *FIRE Forum for International Research in Education*, **2015**, 3, 232640981559582	1.4	5
35	Longitudinal analysis of endurance and respiratory function from a natural history study of Morquio A syndrome. *Molecular Genetics and Metabolism*, **2015**, 114, 186-94	3.7	29
34	Diagnostic evaluation, monitoring, and perioperative management of spinal cord compression in patients with Morquio syndrome. *Molecular Genetics and Metabolism*, **2015**, 114, 11-8	3.7	31
33	Multi-domain impact of elosufase alfa in Morquio A syndrome in the pivotal phase III trial. *Molecular Genetics and Metabolism*, **2015**, 114, 178-85	3.7	57
32	Growth Charts for Individuals with Mucopolysaccharidosis VI (Maroteaux-Lamy Syndrome). *JIMD Reports*, **2015**, 18, 1-11	1.9	22
31	Mortality in patients with morquio syndrome a. *JIMD Reports*, **2015**, 15, 59-66	1.9	65
30	A Comparison of Gait Patterns between Late-Onset Pompe Disease and Age-Matched Healthy Individuals: Does Late-Onset Pompe Disease have a Typical Gait Pattern?. *Journal of Neuromuscular Diseases*, **2015**, 2, S31	5	1
29	Efficacy and safety of enzyme replacement therapy with BMN 110 (elosulfase alfa) for Morquio A syndrome (mucopolysaccharidosis IVA): a phase 3 randomised placebo-controlled study. *Journal of Inherited Metabolic Disease*, **2014**, 37, 979-90	5.4	152
28	Burden of disease in patients with Morquio A syndrome: results from an international patient-reported outcomes survey. *Orphanet Journal of Rare Diseases*, **2014**, 9, 32	4.2	59
27	Altered distribution and function of natural killer cells in murine and human Niemann-Pick disease type C1. *Blood*, **2014**, 123, 51-60	2.2	25
26	A systematic review of the prevalence of Morquio A syndrome: challenges for study reporting in rare diseases. *Orphanet Journal of Rare Diseases*, **2014**, 9, 173	4.2	57
25	Niemann-Pick type C Suspicion Index tool: analyses by age and association of manifestations. *Journal of Inherited Metabolic Disease*, **2014**, 37, 93-101	5.4	40
24	Relative acidic compartment volume as a lysosomal storage disorder-associated biomarker. *Journal of Clinical Investigation*, **2014**, 124, 1320-8	15.9	51
23	Design, baseline characteristics, and early findings of the MPS VI (mucopolysaccharidosis VI) Clinical Surveillance Program (CSP). *Journal of Inherited Metabolic Disease*, **2013**, 36, 373-84	5.4	43
22	Review of clinical presentation and diagnosis of mucopolysaccharidosis IVA. *Molecular Genetics and Metabolism*, **2013**, 110, 54-64	3.7	117
21	Niemann-Pick disease type C symptomatology: an expert-based clinical description. *Orphanet Journal of Rare Diseases*, **2013**, 8, 166	4.2	89
20	The Morquio A Clinical Assessment Program: baseline results illustrating progressive, multisystemic clinical impairments in Morquio A subjects. *Molecular Genetics and Metabolism*, **2013**, 109, 54-61	3.7	102

19	Clinical overview and treatment options for non-skeletal manifestations of mucopolysaccharidosis type IVA. *Journal of Inherited Metabolic Disease*, **2013**, 36, 309-22	5.4	62
18	MRI morphometric characterisation of the paediatric cervical spine and spinal cord in children with MPS IVA (Morquio-Brailsford syndrome). *Journal of Inherited Metabolic Disease*, **2013**, 36, 329-37	5.4	21
17	Spinal involvement in mucopolysaccharidosis IVA (Morquio-Brailsford or Morquio A syndrome): presentation, diagnosis and management. *Journal of Inherited Metabolic Disease*, **2013**, 36, 339-55	5.4	78
16	Diagnosing mucopolysaccharidosis IVA. *Journal of Inherited Metabolic Disease*, **2013**, 36, 293-307	5.4	66
15	Subjective and Objective Assessment of Hand Function in Mucopolysaccharidosis IVa Patients. *JIMD Reports*, **2013**, 9, 59-65	1.9	17
14	Mutation detection in cholestatic patients using microarray resequencing of ATP8B1 and ABCB11. *F1000Research*, **2013**, 2, 32	3.6	3
13	Recommendations for the diagnosis and management of Niemann-Pick disease type C: an update. *Molecular Genetics and Metabolism*, **2012**, 106, 330-44	3.7	319
12	Dietary practices in glutaric aciduria type 1 over 16 years. *Journal of Human Nutrition and Dietetics*, **2012**, 25, 514-9	3.1	13
11	Disruption of enamel crystal formation quantified by synchrotron microdiffraction. *Journal of Dentistry*, **2012**, 40, 1074-80	4.8	15
10	Invariant natural killer T cells are not affected by lysosomal storage in patients with Niemann-Pick disease type C. *European Journal of Immunology*, **2012**, 42, 1886-92	6.1	11
9	The oral health needs of children, adolescents and young adults affected by a mucopolysaccharide disorder. *JIMD Reports*, **2012**, 2, 51-8	1.9	11
8	Glycogen storage disease. *Paediatrics and Child Health (United Kingdom)*, **2011**, 21, 84-89	0.6	4
7	Improved diagnostic procedures in attenuated mucopolysaccharidosis. *British Journal of Hospital Medicine (London, England: 2005)*, **2011**, 72, 91-5	0.8	5
6	Gastrointestinal disturbances and their management in miglustat-treated patients. *Journal of Inherited Metabolic Disease*, **2011**, 34, 991-1001	5.4	55
5	Quantitative in vivo brain magnetic resonance spectroscopic monitoring of neurological involvement in mucopolysaccharidosis type II (Hunter Syndrome). *Journal of Inherited Metabolic Disease*, **2010**, 33 Suppl 3, S395-9	5.4	13
4	Design and validation of a metabolic disorder resequencing microarray (BRUM1). *Human Mutation*, **2010**, 31, 858-65	4.7	14
3	Inborn errors of metabolism for the diagnostic radiologist. *Pediatric Radiology*, **2009**, 39, 211-20	2.8	8
2	Glutaric aciduria type 1 presenting with epilepsy. *Developmental Medicine and Child Neurology*, **2009** , 51, 235-9	3.3	19

1	The management of children with Hunter syndrome - a case study. *British Journal of Nursing*, **2009**, 18, 321-2	0.7	5