

Chris j Hendriksz

List of Publications by Citations

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

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	Recommendations for the diagnosis and management of Niemann-Pick disease type C: an update. <i>Molecular Genetics and Metabolism</i> , 2012 , 106, 330-44	3.7	319
89	Recommendations for initiation and cessation of enzyme replacement therapy in patients with Fabry disease: the European Fabry Working Group consensus document. <i>Orphanet Journal of Rare Diseases</i> , 2015 , 10, 36	4.2	173
88	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. <i>Journal of Inherited Metabolic Disease</i> , 2014 , 37, 979-90	5.4	152
87	Consensus clinical management guidelines for Niemann-Pick disease type C. <i>Orphanet Journal of Rare Diseases</i> , 2018 , 13, 50	4.2	124
86	Review of clinical presentation and diagnosis of mucopolysaccharidosis IVA. <i>Molecular Genetics and Metabolism</i> , 2013 , 110, 54-64	3.7	117
85	A phase I/II study of intrathecal idursulfase-IT in children with severe mucopolysaccharidosis II. <i>Genetics in Medicine</i> , 2016 , 18, 73-81	8.1	111
84	The Morquio A Clinical Assessment Program: baseline results illustrating progressive, multisystemic clinical impairments in Morquio A subjects. <i>Molecular Genetics and Metabolism</i> , 2013 , 109, 54-61	3.7	102
83	Diagnostic tests for Niemann-Pick disease type C (NP-C): A critical review. <i>Molecular Genetics and Metabolism</i> , 2016 , 118, 244-54	3.7	97
82	Niemann-Pick disease type C symptomatology: an expert-based clinical description. <i>Orphanet Journal of Rare Diseases</i> , 2013 , 8, 166	4.2	89
81	International guidelines for the management and treatment of Morquio A syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2015 , 167A, 11-25	2.5	86
80	Spinal involvement in mucopolysaccharidosis IVA (Morquio-Brailsford or Morquio A syndrome): presentation, diagnosis and management. <i>Journal of Inherited Metabolic Disease</i> , 2013 , 36, 339-55	5.4	78
79	Enzyme replacement therapy for late-onset Pompe disease. <i>The Cochrane Library</i> , 2018 ,	5.2	78
78	Diagnosing mucopolysaccharidosis IVA. <i>Journal of Inherited Metabolic Disease</i> , 2013 , 36, 293-307	5.4	66
77	Mortality in patients with morquio syndrome a. <i>JIMD Reports</i> , 2015 , 15, 59-66	1.9	65
76	Clinical overview and treatment options for non-skeletal manifestations of mucopolysaccharidosis type IVA. <i>Journal of Inherited Metabolic Disease</i> , 2013 , 36, 309-22	5.4	62
75	Burden of disease in patients with Morquio A syndrome: results from an international patient-reported outcomes survey. <i>Orphanet Journal of Rare Diseases</i> , 2014 , 9, 32	4.2	59
74	Multi-domain impact of elosufase alfa in Morquio A syndrome in the pivotal phase III trial. <i>Molecular Genetics and Metabolism</i> , 2015 , 114, 178-85	3.7	57

73	A systematic review of the prevalence of Morquio A syndrome: challenges for study reporting in rare diseases. <i>Orphanet Journal of Rare Diseases</i> , 2014 , 9, 173	4.2	57
72	Gastrointestinal disturbances and their management in miglustat-treated patients. <i>Journal of Inherited Metabolic Disease</i> , 2011 , 34, 991-1001	5.4	55
71	Relative acidic compartment volume as a lysosomal storage disorder-associated biomarker. <i>Journal of Clinical Investigation</i> , 2014 , 124, 1320-8	15.9	51
70	Design, baseline characteristics, and early findings of the MPS VI (mucopolysaccharidosis VI) Clinical Surveillance Program (CSP). <i>Journal of Inherited Metabolic Disease</i> , 2013 , 36, 373-84	5.4	43
69	Rapid progression and mortality of lysosomal acid lipase deficiency presenting in infants. <i>Genetics in Medicine</i> , 2016 , 18, 452-8	8.1	42
68	Long-term endurance and safety of elosulfase alfa enzyme replacement therapy in patients with Morquio A syndrome. <i>Molecular Genetics and Metabolism</i> , 2016 , 119, 131-43	3.7	42
67	Niemann-Pick type C Suspicion Index tool: analyses by age and association of manifestations. <i>Journal of Inherited Metabolic Disease</i> , 2014 , 37, 93-101	5.4	40
66	Recommendations for the management of MPS IVA: systematic evidence- and consensus-based guidance. <i>Orphanet Journal of Rare Diseases</i> , 2019 , 14, 137	4.2	36
65	Observational clinical study of 22 adult-onset Pompe disease patients undergoing enzyme replacement therapy over 5years. <i>Molecular Genetics and Metabolism</i> , 2016 , 117, 413-8	3.7	31
64	Diagnostic evaluation, monitoring, and perioperative management of spinal cord compression in patients with Morquio syndrome. <i>Molecular Genetics and Metabolism</i> , 2015 , 114, 11-8	3.7	31
63	Cervical cord compression in mucopolysaccharidosis VI (MPS VI): Findings from the MPS VI Clinical Surveillance Program (CSP). <i>Molecular Genetics and Metabolism</i> , 2016 , 118, 310-8	3.7	31
62	The effect of galsulfase enzyme replacement therapy on the growth of patients with mucopolysaccharidosis VI (Maroteaux-Lamy syndrome). <i>Molecular Genetics and Metabolism</i> , 2017 , 122, 107-112	3.7	29
61	Mortality in patients with Sanfilippo syndrome. <i>Orphanet Journal of Rare Diseases</i> , 2017 , 12, 168	4.2	29
60	Longitudinal analysis of endurance and respiratory function from a natural history study of Morquio A syndrome. <i>Molecular Genetics and Metabolism</i> , 2015 , 114, 186-94	3.7	29
59	Phenotype, treatment practice and outcome in the cobalamin-dependent remethylation disorders and MTHFR deficiency: Data from the E-HOD registry. <i>Journal of Inherited Metabolic Disease</i> , 2019 , 42, 333-352	5.4	28
58	Health-related quality of life in mucopolysaccharidosis: looking beyond biomedical issues. <i>Orphanet Journal of Rare Diseases</i> , 2016 , 11, 119	4.2	25
57	Altered distribution and function of natural killer cells in murine and human Niemann-Pick disease type C1. <i>Blood</i> , 2014 , 123, 51-60	2.2	25
56	Differences in Niemann-Pick disease Type C symptomatology observed in patients of different ages. <i>Molecular Genetics and Metabolism</i> , 2017 , 120, 180-189	3.7	24

55	Diagnosing childhood-onset inborn errors of metabolism by next-generation sequencing. <i>Archives of Disease in Childhood</i> , 2017 , 102, 1019-1029	2.2	24
54	Niemann-Pick type C disease - the tip of the iceberg? A review of neuropsychiatric presentation, diagnosis and treatment. <i>BJPsych Bulletin</i> , 2017 , 41, 109-114	1.2	24
53	Growth Charts for Individuals with Mucopolysaccharidosis VI (Maroteaux-Lamy Syndrome). <i>JIMD Reports</i> , 2015 , 18, 1-11	1.9	22
52	MRI morphometric characterisation of the paediatric cervical spine and spinal cord in children with MPS IVA (Morquio-Brailsford syndrome). <i>Journal of Inherited Metabolic Disease</i> , 2013 , 36, 329-37	5.4	21
51	Elosulfase alfa (BMN 110) for the treatment of mucopolysaccharidosis IVA (Morquio A Syndrome). <i>Expert Review of Clinical Pharmacology</i> , 2016 , 9, 1521-1532	3.8	20
50	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. <i>Molecular Genetics and Metabolism</i> , 2018 , 123, 176-187	3.7	19
49	Enzyme replacement therapy with velmanase alfa (human recombinant alpha-mannosidase): Novel global treatment response model and outcomes in patients with alpha-mannosidosis. <i>Molecular Genetics and Metabolism</i> , 2018 , 124, 152-160	3.7	19
48	Glutaric aciduria type 1 presenting with epilepsy. <i>Developmental Medicine and Child Neurology</i> , 2009 , 51, 235-9	3.3	19
47	The hidden Niemann-Pick type C patient: clinical niches for a rare inherited metabolic disease. <i>Current Medical Research and Opinion</i> , 2017 , 33, 877-890	2.5	18
46	Recommendations for the management of MPS VI: systematic evidence- and consensus-based guidance. <i>Orphanet Journal of Rare Diseases</i> , 2019 , 14, 118	4.2	18
45	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. <i>Molecular Genetics and Metabolism</i> , 2018 , 123, 127-134	3.7	18
44	Levels of glycosaminoglycans in the cerebrospinal fluid of healthy young adults, surrogate-normal children, and Hunter syndrome patients with and without cognitive impairment. <i>Molecular Genetics and Metabolism Reports</i> , 2015 , 5, 103-106	1.8	18
43	Mechanisms of Mitochondrial Dysfunction in Lysosomal Storage Disorders: A Review. <i>Journal of Clinical Medicine</i> , 2020 , 9,	5.1	18
42	Impact of long-term elosulfase alfa treatment on respiratory function in patients with Morquio A syndrome. <i>Journal of Inherited Metabolic Disease</i> , 2016 , 39, 839-847	5.4	18
41	Impact of elosulfase alfa in patients with morquio A syndrome who have limited ambulation: An open-label, phase 2 study. <i>American Journal of Medical Genetics, Part A</i> , 2017 , 173, 375-383	2.5	17
40	Subjective and Objective Assessment of Hand Function in Mucopolysaccharidosis IVa Patients. <i>JIMD Reports</i> , 2013 , 9, 59-65	1.9	17
39	Challenges in diagnosing and managing adult patients with urea cycle disorders. <i>Journal of Inherited Metabolic Disease</i> , 2019 , 42, 1136-1146	5.4	15
38	Disruption of enamel crystal formation quantified by synchrotron microdiffraction. <i>Journal of Dentistry</i> , 2012 , 40, 1074-80	4.8	15

37	Design and validation of a metabolic disorder resequencing microarray (BRUM1). <i>Human Mutation</i> , 2010 , 31, 858-65	4.7	14
36	Dietary practices in glutaric aciduria type 1 over 16 years. <i>Journal of Human Nutrition and Dietetics</i> , 2012 , 25, 514-9	3.1	13
35	Quantitative in vivo brain magnetic resonance spectroscopic monitoring of neurological involvement in mucopolysaccharidosis type II (Hunter Syndrome). <i>Journal of Inherited Metabolic Disease</i> , 2010 , 33 Suppl 3, S395-9	5.4	13
34	Haematopoietic Stem Cell Transplantation Arrests the Progression of Neurodegenerative Disease in Late-Onset Tay-Sachs Disease. <i>JIMD Reports</i> , 2018 , 41, 17-23	1.9	12
33	Recommendations for patient screening in ultra-rare inherited metabolic diseases: what have we learned from Niemann-Pick disease type C?. <i>Orphanet Journal of Rare Diseases</i> , 2019 , 14, 20	4.2	11
32	Objective results of median nerve decompression and tenosynovectomy for carpal tunnel syndrome in patients with mucopolysaccharidoses Types I and II. <i>Journal of Hand Surgery: European Volume</i> , 2015 , 40, 216-8	1.4	11
31	Invariant natural killer T cells are not affected by lysosomal storage in patients with Niemann-Pick disease type C. <i>European Journal of Immunology</i> , 2012 , 42, 1886-92	6.1	11
30	The oral health needs of children, adolescents and young adults affected by a mucopolysaccharide disorder. <i>JIMD Reports</i> , 2012 , 2, 51-8	1.9	11
29	Oculomotor abnormalities in children with Niemann-Pick type C. <i>Molecular Genetics and Metabolism</i> , 2018 , 123, 159-168	3.7	11
28	Evaluation of impact of anti-idursulfase antibodies during long-term idursulfase enzyme replacement therapy in mucopolysaccharidosis II patients. <i>Molecular Genetics and Metabolism Reports</i> , 2017 , 12, 2-7	1.8	10
27	Transition of patients with mucopolysaccharidosis from paediatric to adult care. <i>Molecular Genetics and Metabolism Reports</i> , 2019 , 21, 100508	1.8	9
26	Disease progression of alpha-mannosidosis and impact on patients and carers - A UK natural history survey. <i>Molecular Genetics and Metabolism Reports</i> , 2019 , 20, 100480	1.8	8
25	Inborn errors of metabolism for the diagnostic radiologist. <i>Pediatric Radiology</i> , 2009 , 39, 211-20	2.8	8
24	Critical clinical situations in adult patients with Mucopolysaccharidoses (MPS). <i>Orphanet Journal of Rare Diseases</i> , 2020 , 15, 114	4.2	6
23	Markers of cognitive function in individuals with metabolic disease: Morquio syndrome and tyrosinemia type III. <i>Cognitive Neuropsychology</i> , 2018 , 35, 120-147	2.3	6
22	A Cerebrospinal Fluid Collection Study in Pediatric and Adult Patients With Hunter Syndrome. <i>FIRE Forum for International Research in Education</i> , 2015 , 3, 232640981559582	1.4	5
21	Improved diagnostic procedures in attenuated mucopolysaccharidosis. <i>British Journal of Hospital Medicine (London, England: 2005)</i> , 2011 , 72, 91-5	0.8	5
20	The management of children with Hunter syndrome - a case study. <i>British Journal of Nursing</i> , 2009 , 18, 321-2	0.7	5

19	The factors affecting lipid profile in adult patients with Mucopolysaccharidosis. <i>Molecular Genetics and Metabolism Reports</i> , 2017 , 12, 35-40	1.8	4
18	The use of port-a-caths in adult patients with Lysosomal Storage Disorders receiving Enzyme Replacement Therapy-one centre experience. <i>Molecular Genetics and Metabolism Reports</i> , 2017 , 13, 111-114	1.8	4
17	Glycogen storage disease. <i>Paediatrics and Child Health (United Kingdom)</i> , 2015 , 25, 139-144	0.6	4
16	Risks of long-term port use in enzyme replacement therapy for lysosomal storage disorders. <i>Molecular Genetics and Metabolism Reports</i> , 2018 , 15, 71-73	1.8	4
15	Lipid profile in adult patients with Fabry disease - Ten-year follow up. <i>Molecular Genetics and Metabolism Reports</i> , 2017 , 13, 3-6	1.8	4
14	Glycogen storage disease. <i>Paediatrics and Child Health (United Kingdom)</i> , 2011 , 21, 84-89	0.6	4
13	The Challenges of a Successful Pregnancy in a Patient with Adult Refsum's Disease due to Phytanoyl-CoA Hydroxylase Deficiency. <i>JIMD Reports</i> , 2017 , 33, 49-53	1.9	3
12	Synchrotron X-ray diffraction and scanning electron microscopy to understand enamel affected by metabolic disorder mucopolysaccharidosis. <i>Micron</i> , 2016 , 83, 48-53	2.3	3
11	Potential benefits of Fitbit device in managing a patient with mucopolysaccharidosis. <i>Molecular Genetics and Metabolism</i> , 2019 , 126, S111	3.7	3
10	Mutation detection in cholestatic patients using microarray resequencing of ATP8B1 and ABCB11. <i>F1000Research</i> , 2013 , 2, 32	3.6	3
9	An Overview of Benefits and Challenges of Rare Disease Biobanking in Africa, Focusing on South Africa. <i>Biopreservation and Biobanking</i> , 2021 , 19, 143-150	2.1	3
8	Cardiac rhythm abnormalities - An underestimated cardiovascular risk in adult patients with Mucopolysaccharidoses. <i>Molecular Genetics and Metabolism</i> , 2020 , 130, 133-139	3.7	2
7	Healthcare resource use and costs of managing children and adults with lysosomal acid lipase deficiency at a tertiary referral centre in the United Kingdom. <i>PLoS ONE</i> , 2018 , 13, e0191945	3.7	2
6	Synchrotron X-ray diffraction to understand crystallographic texture of enamel affected by Hunter syndrome. <i>Archives of Oral Biology</i> , 2017 , 80, 193-196	2.8	2
5	Toileting Abilities Survey as a surrogate outcome measure for cognitive function: Findings from neuronopathic mucopolysaccharidosis II patients treated with idursulfase and intrathecal idursulfase. <i>Molecular Genetics and Metabolism Reports</i> , 2020 , 25, 100669	1.8	1
4	Are there common walking gait characteristics in patients diagnosed with late-onset Pompe disease?. <i>Human Movement Science</i> , 2021 , 77, 102777	2.4	1
3	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?. <i>Journal of Neuromuscular Diseases</i> , 2015 , 2, S31	5	1
2	Clinical outcomes in an adult patient with mannose phosphate isomerase-congenital disorder of glycosylation who discontinued mannose therapy. <i>Molecular Genetics and Metabolism Reports</i> , 2020 , 25, 100646	1.8	0

- 1 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 ○