Chris j Hendriksz

List of Publications by Year
in descending order

102 papers	4,120 citations	117625 34 h-index	128289 60 g-index
117 all docs	117 docs citations	117 times ranked	3812 citing authors

#	Article	IF	Citations
1	Recommendations for the diagnosis and management of Niemannâ€“Pick disease type C: An update. Molecular Genetics and Metabolism, 2012, 106, 330-344.	1.1	465
2	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.	2.7	239
3	Consensus clinical management guidelines for Niemann-Pick disease type C. Orphanet Journal of Rare Diseases, 2018, 13, 50.	2.7	200
4	Niemann-Pick disease type C symptomatology: an expert-based clinical description. Orphanet Journal of Rare Diseases, 2013, 8, 166.	2.7	176
5	Efficacy and safety of enzyme replacement therapy with BMN 110 (elosulfase alfa) for Morquio A syndrome (mucopolysaccharidosis IVA): a phase 3 randomised placeboâ€controlled study. Journal of Inherited Metabolic Disease, 2014, 37, 979-990.	3.6	176
6	Review of clinical presentation and diagnosis of mucopolysaccharidosis IVA. Molecular Genetics and Metabolism, 2013, 110, 54-64.	1.1	140
7	A phase I/II study of intrathecal idursulfase-IT in children with severe mucopolysaccharidosis II. Genetics in Medicine, 2016, 18, 73-81.	2.4	138
8	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.	1.1	117
9	Diagnostic tests for Niemann-Pick disease type C (NP-C): A critical review. Molecular Genetics and Metabolism, 2016, 118, 244-254.	1.1	114
10	International guidelines for the management and treatment of Morquio A syndrome. American Journal of Medical Genetics, Part A, 2015, 167, 11-25.	1.2	104
11	Spinal involvement in mucopolysaccharidosis IVA (Morquioâ€Brailsford or Morquio A syndrome): presentation, diagnosis and management. Journal of Inherited Metabolic Disease, 2013, 36, 339-355.	3.6	100
12	Clinical overview and treatment options for nonâ€skeletal manifestations of mucopolysaccharidosis type IVA. Journal of Inherited Metabolic Disease, 2013, 36, 309-322.	3.6	79
13	Mortality in Patients with Morquio Syndrome A. JIMD Reports, 2014, 15, 59-66.	1.5	78
14	Diagnosing mucopolysaccharidosis IVA. Journal of Inherited Metabolic Disease, 2013, 36, 293-307.	3.6	77
15	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.	2.7	74
16	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.	2.7	72
17	Gastrointestinal disturbances and their management in miglustatâ€treated patients. Journal of Inherited Metabolic Disease, 2011, 34, 991-1001.	3.6	69
18	Rapid progression and mortality of lysosomal acid lipase deficiency presenting in infants. Genetics in Medicine, 2016, 18, 452-458.	2.4	67

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19	Multi-domain impact of elosulfase alfa in Morquio A syndrome in the pivotal phase III trial. Molecular Genetics and Metabolism, 2015, 114, 178-185.	1.1	65
20	Relative acidic compartment volume as a lysosomal storage disorderâ€“associated biomarker. Journal of Clinical Investigation, 2014, 124, 1320-1328.	8.2	63
21	Recommendations for the management of MPS IVA: systematic evidence- and consensus-based guidance. Orphanet Journal of Rare Diseases, 2019, 14, 137.	2.7	62
22	Design, baseline characteristics, and early findings of the MPS VI (mucopolysaccharidosis VI) Clinical Surveillance Program (CSP). Journal of Inherited Metabolic Disease, 2013, 36, 373-384.	3.6	57
23	Mechanisms of Mitochondrial Dysfunction in Lysosomal Storage Disorders: A Review. Journal of Clinical Medicine, 2020, 9, 2596.	2.4	55
24	Mortality in patients with Sanfilippo syndrome. Orphanet Journal of Rare Diseases, 2017, 12, 168.	2.7	53
25	Phenotype, treatment practice and outcome in the cobalaminâ€dependent remethylation disorders and MTHFR deficiency: Data from the Eâ€HOD registry. Journal of Inherited Metabolic Disease, 2019, 42, 333-352.	3.6	53
26	Niemann-Pick type C Suspicion Index tool: analyses by age and association of manifestations. Journal of Inherited Metabolic Disease, 2014, 37, 93-101.	3.6	48
27	Long-term endurance and safety of elosulfase alfa enzyme replacement therapy in patients with Morquio A syndrome. Molecular Genetics and Metabolism, 2016, 119, 131-143.	1.1	47
28	Observational clinical study of 22 adult-onset Pompe disease patients undergoing enzyme replacement therapy over 5years. Molecular Genetics and Metabolism, 2016, 117, 413-418.	1.1	45
29	Diagnosing childhood-onset inborn errors of metabolism by next-generation sequencing. Archives of Disease in Childhood, 2017, 102, 1019-1029.	1.9	43
30	Health-related quality of life in mucopolysaccharidosis: looking beyond biomedical issues. Orphanet Journal of Rare Diseases, 2016, 11, 119.	2.7	41
31	Niemann-Pick type C disease â€“ the tip of the iceberg? A review of neuropsychiatric presentation, diagnosis and treatment. BJPsych Bulletin, 2017, 41, 109-114.	1.1	41
32	Challenges in diagnosing and managing adult patients with urea cycle disorders. Journal of Inherited Metabolic Disease, 2019, 42, 1136-1146.	3.6	41
33	Diagnostic evaluation, monitoring, and perioperative management of spinal cord compression in patients with Morquio syndrome. Molecular Genetics and Metabolism, 2015, 114, 11-18.	1.1	39
34	Altered distribution and function of natural killer cells in murine and human Niemann-Pick disease type C1. Blood, 2014, 123, 51-60.	1.4	38
35	Cervical cord compression in mucopolysaccharidosis VI (MPS VI): Findings from the MPS VI Clinical Surveillance Program (CSP). Molecular Genetics and Metabolism, 2016, 118, 310-318.	1.1	38
36	Longitudinal analysis of endurance and respiratory function from a natural history study of Morquio A syndrome. Molecular Genetics and Metabolism, 2015, 114, 186-194.	1.1	33

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37	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.	1.1	33
38	Differences in Niemann-Pick disease Type C symptomatology observed in patients of different ages. Molecular Genetics and Metabolism, 2017, 120, 180-189.	1.1	32
39	Growth Charts for Individuals with Mucopolysaccharidosis VI (Maroteauxâ€“Lamy Syndrome). JIMD Reports, 2014, 18, 1-11.	1.5	31
40	Recommendations for the management of MPS VI: systematic evidence- and consensus-based guidance. Orphanet Journal of Rare Diseases, 2019, 14, 118.	2.7	30
41	Elosulfase alfa (BMN 110) for the treatment of mucopolysaccharidosis IVA (Morquio A Syndrome). Expert Review of Clinical Pharmacology, 2016, 9, 1521-1532.	3.1	27
42	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.	1.1	27
43	The hidden Niemann-Pick type C patient: clinical niches for a rare inherited metabolic disease. Current Medical Research and Opinion, 2017, 33, 877-890.	1.9	25
44	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.	1.1	25
45	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.	1.1	24
46	Impact of longâ€term elosulfase alfa treatment on respiratory function in patients with Morquio A syndrome. Journal of Inherited Metabolic Disease, 2016, 39, 839-847.	3.6	24
47	Impact of elosulfase alfa in patients with morquio A syndrome who have limited ambulation: An openâ€label, phase 2 study. American Journal of Medical Genetics, Part A, 2017, 173, 375-383.	1.2	24
48	Glutaric aciduria type 1 presenting with epilepsy. Developmental Medicine and Child Neurology, 2009, 51, 235-239.	2.1	23
49	Disruption of enamel crystal formation quantified by synchrotron microdiffraction. Journal of Dentistry, 2012, 40, 1074-1080.	4.1	23
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 5â€¯years of treatment. Molecular Genetics and Metabolism, 2018, 123, 479-487.	1.1	23
51	MRI morphometric characterisation of the paediatric cervical spine and spinal cord in children with MPS IVA (Morquioâ€Brailsford syndrome). Journal of Inherited Metabolic Disease, 2013, 36, 329-337.	3.6	22
52	Subjective and Objective Assessment of Hand Function in Mucopolysaccharidosis IVa Patients. JIMD Reports, 2012, 9, 59-65.	1.5	21
53	Haematopoietic Stem Cell Transplantation Arrests theÂProgression of Neurodegenerative Disease in Late-Onset Tay-Sachs Disease. JIMD Reports, 2017, 41, 17-23.	1.5	18
54	Design and validation of a metabolic disorder resequencing microarray (BRUM1). Human Mutation, 2010, 31, 858-865.	2.5	16

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55	The Oral Health Needs of Children, Adolescents and Young Adults Affected by a Mucopolysaccharide Disorder. JIMD Reports, 2011, 2, 51-58.	1.5	16
56	Dietary practices in glutaric aciduria type 1 over 16â€ƒyears. Journal of Human Nutrition and Dietetics, 2012, 25, 514-519.	2.5	15
57	Disease progression of alpha-mannosidosis and impact on patients and carers â€“ A UK natural history survey. Molecular Genetics and Metabolism Reports, 2019, 20, 100480.	1.1	15
58	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.	2.7	15
59	Quantitative in vivo brain magnetic resonance spectroscopic monitoring of neurological involvement in mucopolysaccharidosis type II (Hunter Syndrome). Journal of Inherited Metabolic Disease, 2010, 33, 395-399.	3.6	14
60	Invariant natural killer <scp>T</scp> cells are not affected by lysosomal storage in patients with <scp>N</scp>iemannâ€“<scp>P</scp>ick disease type <scp>C</scp>. European Journal of Immunology, 2012, 42, 1886-1892.	2.9	14
61	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.	1.1	14
62	Oculomotor abnormalities in children with Niemann-Pick type C. Molecular Genetics and Metabolism, 2018, 123, 159-168.	1.1	14
63	Transition of patients with mucopolysaccharidosis from paediatric to adult care. Molecular Genetics and Metabolism Reports, 2019, 21, 100508.	1.1	14
64	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, 216-218.	1.0	13
65	Inborn errors of metabolism for the diagnostic radiologist. Pediatric Radiology, 2009, 39, 211-220.	2.0	12
66	Markers of cognitive function in individuals with metabolic disease: Morquio syndrome and tyrosinemia type III. Cognitive Neuropsychology, 2018, 35, 120-147.	1.1	8
67	Critical clinical situations in adult patients with Mucopolysaccharidoses (MPS). Orphanet Journal of Rare Diseases, 2020, 15, 114.	2.7	8
68	Lipid profile in adult patients with Fabry disease - Ten-year follow up. Molecular Genetics and Metabolism Reports, 2017, 13, 3-6.	1.1	7
69	Cardiac rhythm abnormalities - An underestimated cardiovascular risk in adult patients with Mucopolysaccharidoses. Molecular Genetics and Metabolism, 2020, 130, 133-139.	1.1	7
70	An Overview of Benefits and Challenges of Rare Disease Biobanking in Africa, Focusing on South Africa. Biopreservation and Biobanking, 2021, 19, 143-150.	1.0	7
71	The Genetics of Inherited Cholestatic Disorders in Neonates and Infants: Evolving Challenges. Genes, 2021, 12, 1837.	2.4	7
72	Oral treatment for mucopolysaccharidosis <scp>VI</scp>: Outcomes of the first phase <scp>IIa</scp> study with odiparcil. Journal of Inherited Metabolic Disease, 2022, 45, 340-352.	3.6	7

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73	The management of children with Hunter syndrome â€“ a case study. British Journal of Nursing, 2009, 18, 321-322.	0.7	6
74	Improved diagnostic procedures in attenuated mucopolysaccharidosis. British Journal of Hospital Medicine (London, England: 2005), 2011, 72, 91-95.	0.5	6
75	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
76	Glycogen storage disease. Paediatrics and Child Health (United Kingdom), 2015, 25, 139-144.	0.4	6
77	The Challenges of a Successful Pregnancy in a Patient with Adult Refsumâ€™s Disease due to Phytanoyl-CoA Hydroxylase Deficiency. JIMD Reports, 2016, 33, 49-53.	1.5	6
78	Risks of long-term port use in enzyme replacement therapy for lysosomal storage disorders. Molecular Genetics and Metabolism Reports, 2018, 15, 71-73.	1.1	6
79	Mutation detection in cholestatic patients using microarray resequencing of ATP8B1 and ABCB11. F1000Research, 2013, 2, 32.	1.6	6
80	Are there common walking gait characteristics in patients diagnosed with late-onset Pompe disease?. Human Movement Science, 2021, 77, 102777.	1.4	5
81	Glycogen storage disease. Paediatrics and Child Health (United Kingdom), 2011, 21, 84-89.	0.4	4
82	The factors affecting lipid profile in adult patients with Mucopolysaccharidosis. Molecular Genetics and Metabolism Reports, 2017, 12, 35-40.	1.1	4
83	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.	1.1	4
84	Potential benefits of Fitbit device in managing a patient with mucopolysaccharidosis. Molecular Genetics and Metabolism, 2019, 126, S111.	1.1	4
85	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.	2.5	4
86	Synchrotron X-ray diffraction and scanning electron microscopy to understand enamel affected by metabolic disorder mucopolysaccharidosis. Micron, 2016, 83, 48-53.	2.2	3
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.	1.1	3
88	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.	1.1	3
89	Synchrotron X-ray diffraction to understand crystallographic texture of enamel affected by Hunter syndrome. Archives of Oral Biology, 2017, 80, 193-196.	1.8	3
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.	2.7	2

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91	Transfer of high cost drugs to NICE risks fragmentation of care of rare diseases. BMJ, The, 2012, 345, e5727-e5727.	6.0	1
92	Mucopolysaccharidoses (MPS). Journal of Inherited Metabolic Disease, 2013, 36, 177-178.	3.6	1
93	Enzyme replacement therapy for mucopolysaccharidosis type IV (Morquio syndrome). The Cochrane Library, 2018, , .	2.8	1
94	Enzyme replacement therapy for late-onset Pompe disease. The Cochrane Library, 0, , .	2.8	1
95	Hormonal dysfunction in adult patients with mucopolysaccharidosis type I post haematopoietic stem cell transplantation. Molecular Genetics and Metabolism, 2019, 126, S138-S139.	1.1	1
96	Low Serum Cholesterol Concentration in Adult Patients with Phenylketonuria- One Centre Experience. Clinical & Medical Biochemistry Open Access, 2017, 03, .	0.1	1
97	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.	2.6	1
98	The evaluation of psychological therapy interventions to improve emotional wellbeing in patients with lysosomal disorders. Molecular Genetics and Metabolism, 2015, 114, S53.	1.1	0
99	Identification of relevant clinical niches in rare inherited metabolic diseases: Niemann-Pick disease type C as a model. Molecular Genetics and Metabolism, 2017, 120, S64.	1.1	0
100	A review of serum lipid profile in patients with mucopolysaccharidoses. Molecular Genetics and Metabolism, 2017, 120, S127.	1.1	0
101	What lies beneath: a case of spontaneous hypoglycaemia or glucose transporter type 1 defect disguised as chronic fatigue?. Endocrine Abstracts, 0, , 1-1.	0.0	0
102	Mucopolysaccharidosis in Adults. , 2016, , .		0