

# Incidence of the mucopolysaccharidoses in Northern Ireland

Human Genetics

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Citation Report

#	ARTICLE	IF	CITATIONS
1	The frequency of lysosomal storage diseases in The Netherlands. <i>Human Genetics</i> , 1999, 105, 151-156.	3.8	615
2	Clinical and laboratorial study of 19 cases of mucopolysaccharidoses. <i>Revista Do Hospital Das Clinicas</i> , 2000, 55, 213-218.	0.5	21
4	Lysosomal storage disorders: Diagnostic dilemmas and prospects for therapy. <i>Genetics in Medicine</i> , 2002, 4, 412-419.	2.4	32
5	Left ventricular aneurysm, aortic valve disease and coronary narrowing in a patient with Hunter's syndrome. <i>Cardiovascular Pathology</i> , 2002, 11, 94-96.	1.6	19
6	Direct estimates of human per nucleotide mutation rates at 20 loci causing mendelian diseases. <i>Human Mutation</i> , 2003, 21, 12-27.	2.5	308
7	Incidence of the mucopolysaccharidoses in Western Australia. <i>American Journal of Medical Genetics Part A</i> , 2003, 123A, 310-313.	2.4	185
8	Sleep Disorders in Sanfilippo Syndrome: A Polygraphic Study. <i>Clinical EEG (electroencephalography)</i> , 2003, 34, 18-22.	0.9	26
9	Enzyme replacement therapy in mucopolysaccharidosis VI (Maroteaux-Lamy syndrome). <i>Journal of Pediatrics</i> , 2004, 144, 574-580.	1.8	267
10	Mutation and polymorphism spectrum of the GALNS gene in mucopolysaccharidosis IVA (Morquio A). <i>Human Mutation</i> , 2005, 26, 500-512.	2.5	140
11	Threshold effect of urinary glycosaminoglycans and the walk test as indicators of disease progression in a survey of subjects with Mucopolysaccharidosis VI (Maroteaux-Lamy syndrome). , 2005, 134A, 144-150.		130
12	Sanfilippo type B syndrome: five patients with an R565P homozygous mutation in the Î±-N-acetylglucosaminidase gene from the Okinawa islands in Japan. <i>Journal of Human Genetics</i> , 2005, 50, 357-359.	2.3	15
13	Cumulative incidence rates of the mucopolysaccharidoses in Germany. <i>Journal of Inherited Metabolic Disease</i> , 2005, 28, 1011-1017.	3.6	330
14	Mucopolysaccharidosis I: Î±-L-Iduronidase mutations in three Tunisian families. <i>Journal of Inherited Metabolic Disease</i> , 2005, 28, 1019-1026.	3.6	21
15	Neurological Manifestations in Lysosomal Storage Disorders - From Pathology to First Therapeutic Possibilities. <i>Neuropediatrics</i> , 2005, 36, 285-289.	0.6	31
16	Mucopolisacaridosis. <i>EMC Pediatria</i> , 2006, 41, 1-12.	0.0	0
17	Neurophysiologic assessment of mucopolysaccharidosis III. <i>Clinical Neurophysiology</i> , 2006, 117, 2059-2063.	1.5	10
18	Mucopolysaccharidosis type IV: N-Acetylgalactosamine-6-sulfatase mutations in Tunisian patients. <i>Molecular Genetics and Metabolism</i> , 2006, 87, 213-218.	1.1	23
20	Second-trimester diagnosis of mucopolysaccharidosis type IV a presenting as hydrops fetalis. <i>Prenatal Diagnosis</i> , 2006, 26, 750-752.	2.3	5

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21	Prenatal diagnosis of congenital chylothorax associated with de novo partial trisomy 12q (12q21.2â††qter). <i>Prenatal Diagnosis</i> , 2006, 26, 752-755.	2.3	5
22	Hurler's syndrome: dental findings in a case treated with bone marrow transplantation in infancy. <i>International Journal of Paediatric Dentistry</i> , 2006, 16, 207-212.	1.8	21
23	Recurrent Fetal Hydrops due to Mucopolysaccharidoses Type VII. <i>Fetal Diagnosis and Therapy</i> , 2006, 21, 250-254.	1.4	24
24	The MPS I registry: Design, methodology, and early findings of a global disease registry for monitoring patients with Mucopolysaccharidosis Type I. <i>Molecular Genetics and Metabolism</i> , 2007, 91, 37-47.	1.1	172
26	Management Guidelines for Mucopolysaccharidosis VI. <i>Pediatrics</i> , 2007, 120, 405-418.	2.1	214
27	Treatment of Lysosomal Storage Disorders. <i>Drugs</i> , 2007, 67, 2697-2716.	10.9	133
28	Mucopolysaccharidosis II (Hunter Syndrome). , 2007, , 407-414.		0
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30	A clinical study of 77 patients with mucopolysaccharidosis type II. <i>Acta Paediatrica, International Journal of Paediatrics</i> , 2007, 96, 63-70.	1.5	112
31	International Morquio A Registry: Clinical manifestation and natural course of Morquio A disease. <i>Journal of Inherited Metabolic Disease</i> , 2007, 30, 165-174.	3.6	278
32	Sanfilippo syndrome: A miniâ€”review. <i>Journal of Inherited Metabolic Disease</i> , 2008, 31, 240-252.	3.6	303
33	Growth charts for patients affected with Morquio A disease. <i>American Journal of Medical Genetics, Part A</i> , 2008, 146A, 1286-1295.	1.2	122
34	Mucopolysaccharidoses in Brazil: What happens from birth to biochemical diagnosis?. <i>American Journal of Medical Genetics, Part A</i> , 2008, 146A, 1741-1747.	1.2	63
35	The prevalence of and survival in Mucopolysaccharidosis I: Hurler, Hurler-Scheie and Scheie syndromes in the UK. <i>Orphanet Journal of Rare Diseases</i> , 2008, 3, 24.	2.7	129
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37	The Clinical Outcome of Hurler Syndrome after Stem Cell Transplantation. <i>Biology of Blood and Marrow Transplantation</i> , 2008, 14, 485-498.	2.0	161
38	Idursulfase for the treatment of mucopolysaccharidosis II. <i>Expert Opinion on Pharmacotherapy</i> , 2008, 9, 311-317.	1.8	23
39	Recognition and Diagnosis of Mucopolysaccharidosis II (Hunter Syndrome). <i>Pediatrics</i> , 2008, 121, e377-e386.	2.1	260

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41	Mobility in Hurler Syndrome. <i>Journal of Pediatric Orthopaedics</i> , 2008, 28, 163-168.	1.2	59
42	Avaliaço da motricidade orofacial em pacientes com mucopolissacaridose: um estudo transversal. <i>Jornal De Pediatria</i> , 2009, 85, 254-260.	2.0	2
43	Mutational Analysis of the Î±-L-Iduronidase Gene in Three Egyptian Families: Identification of Three Novel Mutations and Five Novel Polymorphisms. <i>Genetic Testing and Molecular Biomarkers</i> , 2009, 13, 761-764.	0.7	3
44	The management of thoracic inlet syndrome associated with Hurler's syndrome: a novel surgical technique. <i>European Journal of Cardio-thoracic Surgery</i> , 2009, 36, 1081-1083.	1.4	2
45	Incidence and prevalence of mucopolysaccharidosis type 1 in the Irish republic. <i>Archives of Disease in Childhood</i> , 2009, 94, 52-54.	1.9	35
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47	Clinical and biochemical studies in mucopolysaccharidosis type II carriers. <i>Journal of Inherited Metabolic Disease</i> , 2009, 32, 732-738.	3.6	12
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51	Anaesthetic considerations of adults with Morquio's syndrome - a case report. <i>BMC Anesthesiology</i> , 2010, 10, 2.	1.8	18
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53	Orthopedic management of mucopolysaccharide disease. <i>Journal of Pediatric Rehabilitation Medicine</i> , 2010, 3, 47-56.	0.5	45
55	Mucopolysaccharidosis VI. <i>Orphanet Journal of Rare Diseases</i> , 2010, 5, 5.	2.7	246
56	Morquio disease: The role of cartilage canals in the pathogenesis of chondrogenic dwarfism. <i>Medical Hypotheses</i> , 2010, 75, 642-644.	1.5	10
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63	Hurler disease (mucopolysaccharidosis type IH): clinical features and consanguinity in Tunisian population. <i>Diagnostic Pathology</i> , 2011, 6, 113.	2.0	12
64	Molecular analysis of mucopolysaccharidosis type I in Tunisia: identification of novel mutation and eight Novel polymorphisms. <i>Diagnostic Pathology</i> , 2011, 6, 39.	2.0	20
65	Molecular analysis of iduronate -2- sulfatase gene in Tunisian patients with mucopolysaccharidosis type II. <i>Diagnostic Pathology</i> , 2011, 6, 42.	2.0	9
66	Mucopolysaccharidosis type I: molecular characteristics of two novel alpha-L-iduronidase mutations in Tunisian patients. <i>Diagnostic Pathology</i> , 2011, 6, 47.	2.0	14
67	Genomic instability in blood cells from murine model of mucopolysaccharidosis type I. <i>Journal of Molecular Histology</i> , 2011, 42, 575-578.	2.2	5
68	Mucopolysaccharidosis type II: European recommendations for the diagnosis and multidisciplinary management of a rare disease. <i>Orphanet Journal of Rare Diseases</i> , 2011, 6, 72.	2.7	183
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72	Histopathologic Changes of the Ear in Canine Models of Mucopolysaccharidosis Types I and VII. <i>Veterinary Pathology</i> , 2011, 48, 616-626.	1.7	5
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76	Spinal cord infarction remote from maximal compression in a patient with Morquio syndrome. <i>Journal of Neurosurgery: Pediatrics</i> , 2012, 9, 608-612.	1.3	52
77	Subjective and Objective Assessment of Hand Function in Mucopolysaccharidosis IVa Patients. <i>JIMD Reports</i> , 2012, 9, 59-65.	1.5	21
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80	The Live-Birth Prevalence of Mucopolysaccharidoses in Estonia. <i>Genetic Testing and Molecular Biomarkers</i> , 2012, 16, 846-849.	0.7	18
81	Morquio A syndrome due to Maternal Uniparental Isodisomy of the telomeric end of chromosome 16. <i>Molecular Genetics and Metabolism</i> , 2012, 105, 438-442.	1.1	17
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85	Transplantation of human umbilical mesenchymal stem cells cures the corneal defects of mucopolysaccharidosis VII mice. <i>Stem Cells</i> , 2013, 31, 2116-2126.	3.2	95
87	Review of clinical presentation and diagnosis of mucopolysaccharidosis IVA. <i>Molecular Genetics and Metabolism</i> , 2013, 110, 54-64.	1.1	140
88	Newborn screening and diagnosis of mucopolysaccharidoses. <i>Molecular Genetics and Metabolism</i> , 2013, 110, 42-53.	1.1	131
89	A pilot newborn screening program for Mucopolysaccharidosis type I in Taiwan. <i>Orphanet Journal of Rare Diseases</i> , 2013, 8, 147.	2.7	69
90	Mucopolysaccharidosis IVA: Correlation between genotype, phenotype and keratan sulfate levels. <i>Molecular Genetics and Metabolism</i> , 2013, 110, 129-138.	1.1	54
91	Clinical overview and treatment options for non-skeletal manifestations of mucopolysaccharidosis type IVA. <i>Journal of Inherited Metabolic Disease</i> , 2013, 36, 309-322.	3.6	79
92	Spinal involvement in mucopolysaccharidosis IVA (Morquio-Brilsford or Morquio A syndrome): presentation, diagnosis and management. <i>Journal of Inherited Metabolic Disease</i> , 2013, 36, 339-355.	3.6	100
93	Foot and Ankle Abnormalities in the Hurler Syndrome. <i>Journal of Pediatric Orthopaedics</i> , 2013, 33, 558-562.	1.2	13
94	Dental findings and oral health status in patients with mucopolysaccharidosis: a case series. <i>Acta Odontologica Scandinavica</i> , 2013, 71, 157-167.	1.6	18
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100	Pharmacokinetic and Pharmacodynamic Evaluation of Elosulfase Alfa, an Enzyme Replacement Therapy in Patients with Morquio A Syndrome. <i>Clinical Pharmacokinetics</i> , 2014, 53, 1137-1147.	3.5	32
101	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.	2.7	72
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108	Current and potential therapeutic strategies for mucopolysaccharidoses. <i>Journal of Clinical Pharmacy and Therapeutics</i> , 2014, 39, 215-224.	1.5	82
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110	Mortality in Patients with Morquio Syndrome A. <i>JIMD Reports</i> , 2014, 15, 59-66.	1.5	78
111	Morquio disease with CNS involvement: a rare association. <i>Journal of Nepal Paediatric Society</i> , 2014, 34, 157-159.	0.1	0
112	Natural history and clinical assessment of Taiwanese patients with mucopolysaccharidosis IVA. <i>Orphanet Journal of Rare Diseases</i> , 2014, 9, 21.	2.7	25
113	Brazilian reference values for MPS II screening in dried blood spots – A fluorimetric assay. <i>Clinical Biochemistry</i> , 2014, 47, 1297-1299.	1.9	4
114	Novel heparan sulfate assay by using automated high-throughput mass spectrometry: Application to monitoring and screening for mucopolysaccharidoses. <i>Molecular Genetics and Metabolism</i> , 2014, 113, 92-99.	1.1	30
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117	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, 979-990.	3.6	176

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119	JIMD Reports, Volume 19. <i>JIMD Reports</i> , 2015, , .	1.5	0
120	Cervical Instability in Young Adults. <i>Operative Techniques in Orthopaedics</i> , 2015, 25, 202-208.	0.1	2
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122	Multi-domain impact of elosulfase alfa in Morquio A syndrome in the pivotal phase III trial. <i>Molecular Genetics and Metabolism</i> , 2015, 114, 178-185.	1.1	65
123	Immunogenicity of Elosulfase Alfa, an Enzyme Replacement Therapy in Patients With Morquio A Syndrome: Results From MOR-004, a Phase III Trial. <i>Clinical Therapeutics</i> , 2015, 37, 1012-1021.e6.	2.5	43
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132	Lower Extremity Deformity Management in MPS IVA, Morquio-Brailsford Syndrome: Preliminary Report of Hemiepiphysiodesis Correction of Genu Valgum. <i>Journal of Pediatric Orthopaedics</i> , 2016, 36, 376-381.	1.2	23
133	Mucopolisacaridosis. <i>EMC Pediatria</i> , 2016, 51, 1-14.	0.0	1
134	Causes of death and clinical characteristics of 34 patients with Mucopolysaccharidosis II in Taiwan from 1995â€“2012. <i>Orphanet Journal of Rare Diseases</i> , 2016, 11, 85.	2.7	26
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148	Long-term Immunogenicity of Elosulfase Alfa in the Treatment of Morquio A Syndrome: Results From MOR-005, a Phase III Extension Study. Clinical Therapeutics, 2017, 39, 118-129.e3.	2.5	24
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150	Bio-Plex immunoassay measuring the quantity of lysosomal <i>N</i> -acetylgalactosamine-6-sulfatase protein in dried blood spots for the screening of mucopolysaccharidosis IVA in newborn: a pilot study. BMJ Open, 2017, 7, e014410.	1.9	7
151	Oxidative profile exhibited by Mucopolysaccharidosis type IVA patients at diagnosis: Increased keratan urinary levels. Molecular Genetics and Metabolism Reports, 2017, 11, 46-53.	1.1	17
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153	A Blood-Brain-Barrier-Penetrating Anti-human Transferrin Receptor Antibody Fusion Protein for Neuronopathic Mucopolysaccharidosis II. Molecular Therapy, 2018, 26, 1366-1374.	8.2	141
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157	A 16-year-old Girl with Morquio Syndrome: A Case Report. <i>BIRDEM Medical Journal</i> , 2018, 8, 266-269.	0.1	0
158	Status of newborn screening and follow up investigations for Mucopolysaccharidoses I and II in Taiwan. <i>Orphanet Journal of Rare Diseases</i> , 2018, 13, 84.	2.7	50
159	Quality of life in mucopolysaccharidoses: construction of a specific measure using the focus group technique. <i>BMC Research Notes</i> , 2018, 11, 28.	1.4	7
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161	Clinical, biochemical and genetic profiles of patients with mucopolysaccharidosis type IVA (Morquio A) Tj ETQq1 1 0.784314 rgBT /Over Diseases, 2019, 14, 143.	2.7	9
162	Oral manifestation and root canal therapy of the patient with mucopolysaccharidosis. <i>Restorative Dentistry &amp; Endodontics</i> , 2019, 44, e14.	1.5	3
163	Effect of enzyme replacement therapy on the growth of patients with Morquio A. <i>Journal of Human Genetics</i> , 2019, 64, 625-635.	2.3	32
164	Hematopoietic Stem Cell Transplantation for Mucopolysaccharidoses: Past, Present, and Future. <i>Biology of Blood and Marrow Transplantation</i> , 2019, 25, e226-e246.	2.0	110
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