Derralynn A Hughes

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/9351238/publications.pdf

Version: 2024-02-01

206 papers	10,977 citations	23567 58 h-index	37204 96 g-index
220 all docs	220 docs citations	220 times ranked	8610 citing authors

#	Article	IF	Citations
1	Development and validation of Gaucher disease type 1 (GD1)-specific patient-reported outcome measures (PROMs) for clinical monitoring and for clinical trials. Orphanet Journal of Rare Diseases, 2022, 17, 9.	2.7	10
2	Cardiac device implantation and device usage in Fabry and hypertrophic cardiomyopathy. Orphanet Journal of Rare Diseases, 2022, 17, 6.	2.7	3
3	Cognitive dysfunction and white matter hyperintensities in Fabry disease. Journal of Inherited Metabolic Disease, 2022, 45, 782-795.	3.6	1
4	Do clinical guidelines facilitate or impede drivers of treatment in Fabry disease?. Orphanet Journal of Rare Diseases, 2022, 17, 42.	2.7	6
5	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
6	Twenty years of the Fabry Outcome Survey (FOS): insights, achievements, and lessons learned from a global patient registry. Orphanet Journal of Rare Diseases, 2022, 17, .	2.7	14
7	The myocardial phenotype of Fabry disease pre-hypertrophy and pre-detectable storage. European Heart Journal Cardiovascular Imaging, 2021, 22, 790-799.	1.2	35
8	New drugs for the treatment of Andersonâ€“Fabry disease. Journal of Nephrology, 2021, 34, 221-230.	2.0	17
9	Pregnancy outcome in women with Gaucher disease type 1 who had unplanned pregnancies during eliglustat clinical trials. JIMD Reports, 2021, 57, 76-84.	1.5	7
10	Impact of long-term elosulfase alfa treatment on clinical and patient-reported outcomes in patients with mucopolysaccharidosis type IVA: results from a Managed Access Agreement in England. Orphanet Journal of Rare Diseases, 2021, 16, 38.	2.7	12
11	Rare but important haematological conditions: Gaucher disease. Medicine, 2021, 49, 248-251.	0.4	0
12	Prompt Agalsidase Alfa Therapy Initiation is Associated with Improved Renal and Cardiovascular Outcomes in a Fabry Outcome Survey Analysis. Drug Design, Development and Therapy, 2021, Volume 15, 3561-3572.	4.3	8
13	Long-term follow-up of renal function in patients treated with migalastat for Fabry disease. Molecular Genetics and Metabolism Reports, 2021, 28, 100786.	1.1	14
14	Impact of <scp>SARSâ€CoV</scp>â€2 (COVIDâ€19) pandemic on patients with lysosomal storage disorders and restoration of services: experience from a specialist centre. Internal Medicine Journal, 2021, 51, 1580-1593.	0.8	5
15	In-depth phenotyping for clinical stratification of Gaucher disease. Orphanet Journal of Rare Diseases, 2021, 16, 431.	2.7	11
16	Estimated impact of the COVID-19 pandemic on cancer services and excess 1-year mortality in people with cancer and multimorbidity: near real-time data on cancer care, cancer deaths and a population-based cohort study. BMJ Open, 2020, 10, e043828.	1.9	233
17	Effects of Baseline Left Ventricular Hypertrophy and Decreased Renal Function on Cardiovascular and Renal Outcomes in Patients with Fabry Disease Treated with Agalsidase Alfa: A Fabry Outcome Survey Study. Clinical Therapeutics, 2020, 42, 2321-2330.e0.	2.5	10
18	Long-term efficacy and safety of migalastat treatment in Fabry disease: 30-month results from the open-label extension of the randomized, phase 3 ATTRACT study. Molecular Genetics and Metabolism, 2020, 131, 219-228.	1.1	44

#	Article	IF	Citations
19	Predicting the Development of Anti-Drug Antibodies against Recombinant alpha-Galactosidase A in Male Patients with Classical Fabry Disease. International Journal of Molecular Sciences, 2020, 21, 5784.	4.1	9
20	White matter integrity correlates with cognition and disease severity in Fabry disease. Brain, 2020, 143, 3331-3342.	7.6	12
21	Early indicators of disease progression in Fabry disease that may indicate the need for disease-specific treatment initiation: findings from the opinion-based PREDICT-FD modified Delphi consensus initiative. BMJ Open, 2020, 10, e035182.	1.9	20
22	Coombs-positive Paroxysmal Nocturnal Haemoglobinuria. Oxford Medical Case Reports, 2020, 2020, omz125.	0.4	2
23	Myocardial Edema, Myocyte Injury, and Disease Severity in Fabry Disease. Circulation: Cardiovascular Imaging, 2020, 13, e010171.	2.6	35
24	An expert consensus document on the management of cardiovascular manifestations of Fabry disease. European Journal of Heart Failure, 2020, 22, 1076-1096.	7.1	96
25	Cardiomyopathy and kidney function in agalsidase betaâ€treated female Fabry patients: a preâ€treatment vs. postâ€treatment analysis. ESC Heart Failure, 2020, 7, 825-834.	3.1	13
26	Lysosomal Acid Lipase Deficiency: Therapeutic Options . Drug Design, Development and Therapy, 2020, Volume 14, 591-601.	4.3	38
27	Proposed Stages of Myocardial Phenotype Development in FabryÂDisease. JACC: Cardiovascular Imaging, 2019, 12, 1673-1683.	5.3	91
28	Evolution and clustering of prodromal parkinsonian features in GBA1 carriers. Movement Disorders, 2019, 34, 1365-1373.	3.9	33
29	Evolution of prodromal parkinsonian features in a cohort ofGBAmutation-positive individuals: a 6-year longitudinal study. Journal of Neurology, Neurosurgery and Psychiatry, 2019, 90, 1091-1097.	1.9	44
30	Gaucher Disease in Bone: From Pathophysiology to Practice. Journal of Bone and Mineral Research, 2019, 34, 996-1013.	2.8	94
31	A randomised controlled trial evaluating arrhythmia burden, risk of sudden cardiac death and stroke in patients with Fabry disease: the role of implantable loop recorders (RaILRoAD) compared with current standard practice. Trials, 2019, 20, 314.	1.6	6
32	Pegunigalsidase alfa, a novel PEGylated enzyme replacement therapy for Fabry disease, provides sustained plasma concentrations and favorable pharmacodynamics: A 1â€year Phase 1/2 clinical trial. Journal of Inherited Metabolic Disease, 2019, 42, 534-544.	3.6	86
33	Safety of switching to Migalastat from enzyme replacement therapy in Fabry disease: Experience from the Phase 3 ATTRACT study. American Journal of Medical Genetics, Part A, 2019, 179, 1069-1073.	1.2	13
34	Efficacy of the pharmacologic chaperone migalastat in a subset of male patients with the classic phenotype of Fabry disease and migalastat-amenable variants: data from the phase 3 randomized, multicenter, double-blind clinical trial and extension study. Genetics in Medicine, 2019, 21, 1987-1997.	2.4	66
35	Study of indications for cardiac device implantation and utilisation in Fabry cardiomyopathy. Heart, 2019, 105, 1825-1831.	2.9	15
36	Cardio- Renal Outcomes With Long- Term Agalsidase Alfa Enzyme Replacement Therapy: A 10-Year Fabry Outcome Survey (FOS) Analysis . Drug Design, Development and Therapy, 2019, Volume 13, 3705-3715.	4.3	19

#	Article	IF	Citations
37	Myocardial Storage, Inflammation, and Cardiac Phenotype in Fabry Disease After One Year of Enzyme Replacement Therapy. Circulation: Cardiovascular Imaging, 2019, 12, e009430.	2.6	47
38	Presenting signs and patient coâ€variables in Gaucher disease: outcome of the Gaucher Earlier Diagnosis Consensus (GEDâ€C) Delphi initiative. Internal Medicine Journal, 2019, 49, 578-591.	0.8	39
39	Bortezomib consolidation postâ€ASCT as frontline therapy for multiple myeloma deepens disease response and MRDâ€negative rate whilst maintaining QOL and response to reâ€treatment at relapse. British Journal of Haematology, 2019, 185, 948-951.	2.5	3
40	Evaluation of the detection of GBA missense mutations and other variants using the Oxford Nanopore MinION. Molecular Genetics & Genomic Medicine, 2019, 7, e564.	1.2	65
41	The effect of enzyme replacement therapy on clinical outcomes in female patients with Fabry disease â€“ A systematic literature review by a European panel of experts. Molecular Genetics and Metabolism, 2019, 126, 224-235.	1.1	65
42	Global longitudinal strain, myocardial storage and hypertrophy in Fabry disease. Heart, 2019, 105, 470-476.	2.9	45
43	Agalsidase alfa versus agalsidase beta for the treatment of Fabry disease: an international cohort study. Journal of Medical Genetics, 2018, 55, 351-358.	3.2	72
44	The Influence of Patient-Reported Joint Manifestations on Quality of Life in Fabry Patients. JIMD Reports, 2018, 41, 37-45.	1.5	10
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. Molecular Genetics and Metabolism, 2018, 123, 127-134.	1.1	25
46	Globotriaosylsphingosine (Lysoâ€Gb₃) as a biomarker for cardiac variant (N215S) Fabry disease. Journal of Inherited Metabolic Disease, 2018, 41, 239-247.	3.6	25
47	Biomarkers of Myocardial Fibrosis: Revealing the Natural History of Fibrogenesis in Fabry Disease Cardiomyopathy. Journal of the American Heart Association, 2018, 7, .	3.7	15
48	Management goals for type 1 Gaucher disease: An expert consensus document from the European working group on Gaucher disease. Blood Cells, Molecules, and Diseases, 2018, 68, 203-208.	1.4	82
49	Phenotype, disease severity and pain are major determinants of quality of life in Fabry disease: results from a large multicenter cohort study. Journal of Inherited Metabolic Disease, 2018, 41, 141-149.	3.6	31
50	The utility of the FIPI score in predicting long-term clinical outcomes in patients with Fabry disease receiving enzyme replacement therapy with agalsidase alfa. Molecular Genetics and Metabolism, 2018, 123, 154-158.	1.1	2
51	Long-term outcomes with agalsidase alfa enzyme replacement therapy: Analysis using deconstructed composite events. Molecular Genetics and Metabolism Reports, 2018, 14, 31-35.	1.1	11
52	54â€…Characterisation of systolic myocardial strain in patients with fabry disease. , 2018, , .		0
53	Renal outcomes with up to 9 years of migalastat in patients with Fabry disease: Results from an open-label extension study. Molecular Genetics and Metabolism, 2018, 123, S105-S106.	1.1	2
54	Cardiac Phenotype of Prehypertrophic Fabry Disease. Circulation: Cardiovascular Imaging, 2018, 11, e007168.	2.6	58

#	Article	IF	Citations
55	Impact of sphingolipids on osteoblast and osteoclast activity in Gaucher disease. Molecular Genetics and Metabolism, 2018, 124, 278-286.	1.1	21
56	Migalastat improves diarrhea in patients with Fabry disease: clinical-biomarker correlations from the phase 3 FACETS trial. Orphanet Journal of Rare Diseases, 2018, 13, 68.	2.7	23
57	Generation of osteoclasts from type 1 Gaucher patients and correlation with clinical and genetic features of disease. Gene, 2018, 678, 196-206.	2.2	9
58	European expert consensus statement on therapeutic goals in Fabry disease. Molecular Genetics and Metabolism, 2018, 124, 189-203.	1.1	122
59	Oral pharmacological chaperone migalastat compared with enzyme replacement therapy in Fabry disease: 18-month results from the randomised phase III ATTRACT study. Journal of Medical Genetics, 2017, 54, 288-296.	3.2	262
60	Favourable effect of early versus late start of enzyme replacement therapy on plasma globotriaosylsphingosine levels in men with classical Fabry disease. Molecular Genetics and Metabolism, 2017, 121, 157-161.	1.1	64
61	New biomarkers defining a novel early stage of Fabry nephropathy: A diagnostic test study. Molecular Genetics and Metabolism, 2017, 121, 162-169.	1.1	14
62	Rare but important haematological conditions: Gaucher disease. Medicine, 2017, 45, 256-259.	0.4	2
63	Maximum inspiratory pressure as a clinically meaningful trial endpoint for neuromuscular diseases: a comprehensive review of the literature. Orphanet Journal of Rare Diseases, 2017, 12, 52.	2.7	69
64	Characterization of Classical and Nonclassical Fabry Disease: A Multicenter Study. Journal of the American Society of Nephrology: JASN, 2017, 28, 1631-1641.	6.1	244
65	Gaucher disease: risk stratification and comorbidities. Expert Opinion on Orphan Drugs, 2017, 5, 839-846.	0.8	0
66	Exploring the patient journey to diagnosis of Gaucher disease from the perspective of 212 patients with Gaucher disease and 16 Gaucher expert physicians. Molecular Genetics and Metabolism, 2017, 122, 122-129.	1.1	51
67	Clinical outcomes in a subpopulation of adults with Morquio A syndrome: results from a long-term extension study of elosulfase alfa. Orphanet Journal of Rare Diseases, 2017, 12, 98.	2.7	24
68	Management and monitoring recommendations for the use of eliglustat in adults with type 1 Gaucher disease in Europe. European Journal of Internal Medicine, 2017, 37, 25-32.	2.2	60
69	Social preference weights for treatments in Fabry disease in the UK: a discrete choice experiment. Current Medical Research and Opinion, 2017, 33, 23-29.	1.9	5
70	Insight into hypertrophied hearts: a cardiovascular magnetic resonance study of papillary muscle mass and T1 mapping. European Heart Journal Cardiovascular Imaging, 2017, 18, 1034-1040.	1.2	31
71	The validation of pharmacogenetics for the identification of Fabry patients to be treated with migalastat. Genetics in Medicine, 2017, 19, 430-438.	2.4	157
72	Lysosomal Storage Disorders and Malignancy. Diseases (Basel, Switzerland), 2017, 5, 8.	2.5	11

#	Article	IF	Citations
73	A study on the safety and efficacy of reveglucosidase alfa in patients with late-onset Pompe disease. Orphanet Journal of Rare Diseases, 2017, 12, 144.	2.7	29
74	Fabry disease and incidence of cancer. Orphanet Journal of Rare Diseases, 2017, 12, 150.	2.7	10
75	Retrospective study of long-term outcomes of enzyme replacement therapy in Fabry disease: Analysis of prognostic factors. PLoS ONE, 2017, 12, e0182379.	2.5	83
76	Fabry disease. Current Opinion in Cardiology, 2016, 31, 434-439.	1.8	9
77	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
78	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
79	Cardiac Fabry Disease With Late Gadolinium Enhancement Is a Chronic Inflammatory Cardiomyopathy. Journal of the American College of Cardiology, 2016, 68, 1707-1708.	2.8	78
80	Treatment of Fabryâ€™s Disease with the Pharmacologic Chaperone Migalastat. New England Journal of Medicine, 2016, 375, 545-555.	27.0	390
81	Pregnancy and associated events in women receiving enzyme replacement therapy for lateâ€onset glycogen storage disease type II (Pompe disease). Journal of Obstetrics and Gynaecology Research, 2016, 42, 1263-1271.	1.3	10
82	A 15-Year Perspective of the Fabry Outcome Survey. FIRE Forum for International Research in Education, 2016, 4, 232640981666629.	0.7	9
83	A Novel Rapid MALDI-TOF-MS-Based Method for Measuring Urinary Globotriaosylceramide in Fabry Patients. Journal of the American Society for Mass Spectrometry, 2016, 27, 719-725.	2.8	7
84	Fabry in the older patient: Clinical consequences and possibilities for treatment. Molecular Genetics and Metabolism, 2016, 118, 319-325.	1.1	15
85	Bortezomib Consolidation Following Upfront ASCT for Multiple Myeloma Deepens Disease Response and MRD-Negative Rate without Compromising Response to Subsequent Bortezomib Salvage: Results of a Phase II Study. Blood, 2016, 128, 4508-4508.	1.4	3
86	Lysosomal Storage Disorders: Haematology Perspective. , 2016, , 619-631.		0
87	Safety and efficacy results of switch from imiglucerase to velaglucerase alfa treatment in patients with type 1 G aucher disease. American Journal of Hematology, 2015, 90, 592-597.	4.1	14
88	Results from a 9â€year Intensive Safety Surveillance Scheme (IS³) in miglustat (Zavesca^{Â®})â€treated patients. Pharmacoepidemiology and Drug Safety, 2015, 24, 329-333.	1.9	13
89	Prevalence of CADASIL and Fabry Disease in a Cohort of MRI Defined Younger Onset Lacunar Stroke. PLoS ONE, 2015, 10, e0136352.	2.5	38
90	Chronic kidney disease and an uncertain diagnosis of Fabry disease: Approach to a correct diagnosis. Molecular Genetics and Metabolism, 2015, 114, 242-247.	1.1	51

#	Article	IF	Citations
91	Long-term effectiveness of agalsidase alfa enzyme replacement in Fabry disease: A Fabry Outcome Survey analysis. Molecular Genetics and Metabolism Reports, 2015, 3, 21-27.	1.1	97
92	Eliglustat for Gaucher's disease: trippingly on the tongue. Lancet, The, 2015, 385, 2328-2330.	13.7	6
93	Clinical and genetic predictors of major cardiac events in patients with Andersonâ€“Fabry Disease. Heart, 2015, 101, 961-966.	2.9	78
94	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
95	Evolution of Prodromal Clinical Markers of Parkinson Disease in aGBAMutationâ€“Positive Cohort. JAMA Neurology, 2015, 72, 201.	9.0	180
96	Clinical prodromes of neurodegeneration in Anderson-Fabry disease. Neurology, 2015, 84, 1454-1464.	1.1	58
97	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
98	Non-neuronopathic lysosomal storage disorders: Disease spectrum and treatments. Best Practice and Research in Clinical Endocrinology and Metabolism, 2015, 29, 173-182.	4.7	20
99	Velaglucerase alfa (VPRIV) enzyme replacement therapy in patients with Gaucher disease: Longâ€term data from phase III clinical trials. American Journal of Hematology, 2015, 90, 584-591.	4.1	39
100	Clinicopathologic characteristics and outcomes of patients experiencing severe pyrimethamine poisoning. Leukemia and Lymphoma, 2014, 55, 2410-2412.	1.3	1
101	Visual short-term memory deficits associated with GBA mutation and Parkinsonâ€™s disease. Brain, 2014, 137, 2303-2311.	7.6	77
102	Outcome of pregnancies in women receiving velaglucerase alfa for <scp>G</scp>aucher disease. Journal of Obstetrics and Gynaecology Research, 2014, 40, 968-975.	1.3	22
103	Eliglustat tartrate: an oral therapeutic option for Gaucher disease type 1. Clinical Investigation, 2014, 4, 45-53.	0.0	2
104	Gaucher disease: haematological presentations and complications. British Journal of Haematology, 2014, 165, 427-440.	2.5	81
105	Uncertain diagnosis of Fabry disease: Consensus recommendation on diagnosis in adults with left ventricular hypertrophy and genetic variants of unknown significance. International Journal of Cardiology, 2014, 177, 400-408.	1.7	119
106	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
107	Longâ€term effectiveness of enzyme replacement therapy in Fabry disease: results from the NCSâ€LSD cohort study. Journal of Inherited Metabolic Disease, 2014, 37, 969-978.	3.6	38
108	Effectiveness of enzyme replacement therapy in adults with lateâ€onset Pompe disease: results from the NCSâ€LSD cohort study. Journal of Inherited Metabolic Disease, 2014, 37, 945-952.	3.6	51

#	Article	IF	Citations
109	Pseudoacromegalic facial features in Fabry disease. Clinical and Experimental Dermatology, 2013, 38, 137-139.	1.3	9
110	The glucocerobrosidase E326K variant predisposes to Parkinson's disease, but does not cause Gaucher's disease. Movement Disorders, 2013, 28, 232-236.	3.9	121
111	Glucocerebrosidase inhibition causes mitochondrial dysfunction and free radical damage. Neurochemistry International, 2013, 62, 1-7.	3.8	166
112	Diagnosing Gaucher disease: An on-going need for increased awareness amongst haematologists. Blood Cells, Molecules, and Diseases, 2013, 50, 212-217.	1.4	52
113	Miglustat therapy in type 1 Gaucher disease: Clinical and safety outcomes in a multicenter retrospective cohort study. Blood Cells, Molecules, and Diseases, 2013, 51, 116-124.	1.4	60
114	Phenotypical characterization of Î±-galactosidase A gene mutations identified in a large Fabry disease screening program in stroke in the young. Clinical Neurology and Neurosurgery, 2013, 115, 1088-1093.	1.4	31
115	Enhanced differentiation of osteoclasts from mononuclear precursors in patients with Gaucher disease. Blood Cells, Molecules, and Diseases, 2013, 51, 185-194.	1.4	33
116	Rare but important haematological conditions: Gaucher disease. Medicine, 2013, 41, 252-254.	0.4	0
117	A randomised, double-blind, placebo-controlled, crossover study to assess the efficacy and safety of three dosing schedules of agalsidase alfa enzyme replacement therapy for Fabry disease. Molecular Genetics and Metabolism, 2013, 109, 269-275.	1.1	20
118	Retinal thinning in Gaucher disease patients and carriers: Results of a pilot study. Molecular Genetics and Metabolism, 2013, 109, 221-223.	1.1	28
119	Role of Serum N-Terminal Pro-Brain Natriuretic Peptide Measurement in Diagnosis of Cardiac Involvement in Patients With Anderson-Fabry Disease. American Journal of Cardiology, 2013, 111, 111-117.	1.6	54
120	Identification and Assessment of Anderson-Fabry Disease by Cardiovascular Magnetic Resonance Noncontrast Myocardial T1 Mapping. Circulation: Cardiovascular Imaging, 2013, 6, 392-398.	2.6	399
121	Haematological manifestations and complications of Gaucher disease. Current Opinion in Hematology, 2013, 20, 41-47.	2.5	13
122	Migalastat HCl Reduces Globotriaosylsphingosine (Lyso-Gb3) in Fabry Transgenic Mice and in the Plasma of Fabry Patients. PLoS ONE, 2013, 8, e57631.	2.5	40
123	Safety and efficacy of velaglucerase alfa in Gaucher disease type 1 patients previously treated with imiglucerase. American Journal of Hematology, 2013, 88, 172-178.	4.1	50
124	Gaucher Disease and Myeloma. Critical Reviews in Oncogenesis, 2013, 18, 247-268.	0.4	22
125	Female Andersonâ€“Fabry disease mimicking hypertrophic cardiomyopathy: Figure 1. Journal of Clinical Pathology, 2012, 65, 377-378.	2.0	0
126	The effectiveness and cost-effectiveness of enzyme and substrate replacement therapies: a longitudinal cohort study of people with lysosomal storage disorders.. Health Technology Assessment, 2012, 16, 1-543.	2.8	94

#	Article	IF	Citations
127	Recommendations on Reintroduction of Agalsidase Beta for Patients with Fabry Disease in Europe, Following a Period of Shortage. JIMD Reports, 2012, 8, 51-56.	1.5	9
128	Fabry International Prognostic Index: a predictive severity score for Anderson-Fabry disease. Journal of Medical Genetics, 2012, 49, 212-220.	3.2	24
129	A variant of unknown significance in the GLA gene causing diagnostic uncertainty in a young female with isolated hypertrophic cardiomyopathy. Gene, 2012, 497, 320-322.	2.2	6
130	The cognitive profile of type 1 Gaucher disease patients. Journal of Inherited Metabolic Disease, 2012, 35, 1093-1099.	3.6	17
131	Novel pathogenic mutations in the glucocerebrosidase locus. Molecular Genetics and Metabolism, 2012, 106, 495-497.	1.1	5
132	Cardiovascular magnetic resonance measurement of myocardial extracellular volume in health and disease. Heart, 2012, 98, 1436-1441.	2.9	276
133	Safety and pharmacodynamic effects of a pharmacological chaperone on Î±-galactosidase A activity and globotriaosylceramide clearance in Fabry disease: report from two phase 2 clinical studies. Orphanet Journal of Rare Diseases, 2012, 7, 91.	2.7	95
134	In utero administration of Ad5 and AAV pseudotypes to the fetal brain leads to efficient, widespread and long-term gene expression. Gene Therapy, 2012, 19, 936-946.	4.5	31
135	A clinical and family history study of Parkinson's disease in heterozygousglucocerebrosidasemutation carriers. Journal of Neurology, Neurosurgery and Psychiatry, 2012, 83, 853-854.	1.9	99
136	<scp>F</scp>abry disease in unselected patients with <scp>TIA</scp> or stroke: populationâ€based study. European Journal of Neurology, 2012, 19, 1427-1432.	3.3	27
137	Hyposmia and cognitive impairment in Gaucher disease patients and carriers. Movement Disorders, 2012, 27, 526-532.	3.9	108
138	Functional analysis of variant lysosomal acid glycosidases of Andersonâ€Fabry and Pompe disease in a human embryonic kidney epithelial cell line (HEK 293 T). Journal of Inherited Metabolic Disease, 2012, 35, 325-334.	3.6	6
139	Prevalence of Anderson-Fabry disease in patients with hypertrophic cardiomyopathy: the European Anderson-Fabry Disease Survey. Heart, 2011, 97, 1957-1960.	2.9	163
140	Clinico-pathologic characteristics of patients with hepatic lymphoma diagnosed using image-guided liver biopsy techniques. Leukemia and Lymphoma, 2011, 52, 2130-2134.	1.3	14
141	Exercise-Induced Left Ventricular Outflow Tract Obstruction in Symptomatic Patients With Anderson-Fabry Disease. Journal of the American College of Cardiology, 2011, 58, 88-89.	2.8	34
142	Potential biomarkers of osteonecrosis in Gaucher disease. Blood Cells, Molecules, and Diseases, 2011, 46, 27-33.	1.4	47
143	Early access experience with VPRIVÂ®: Recommendations for â€˜core dataâ€™ collection. Blood Cells, Molecules, and Diseases, 2011, 47, 140-142.	1.4	4
144	Expert opinion on temporary treatment recommendations for Fabry disease during the shortage of enzyme replacement therapy (ERT). Molecular Genetics and Metabolism, 2011, 102, 99-102.	1.1	43

#	Article	IF	Citations
145	Response of women with Fabry disease to enzyme replacement therapy: Comparison with men, using data from FOSâ€"the Fabry Outcome Survey. Molecular Genetics and Metabolism, 2011, 103, 207-214.	1.1	48
146	Cystatin C and NT-proBNP as prognostic biomarkers in Fabry disease. Molecular Genetics and Metabolism, 2011, 104, 301-307.	1.1	33
147	Osseous Manifestations of Adult Gaucher Disease in the Era of Enzyme Replacement Therapy. Medicine (United States), 2011, 90, 52-60.	1.0	104
148	Toward a consensus in the laboratory diagnostics of Fabry disease â€•recommendations of a European expert group. Journal of Inherited Metabolic Disease, 2011, 34, 509-514.	3.6	77
149	Incidence and predictors of anti-bradycardia pacing in patients with Anderson-Fabry disease. Europace, 2011, 13, 1781-1788.	1.7	63
150	Gaucher Disease: Outcome following Total Hip Replacements and Effect of Enzyme Replacement Therapy in a Cohort of Uk Patients. HIP International, 2011, 21, 665-671.	1.7	5
151	A Distinct Urinary Biomarker Pattern Characteristic of Female Fabry Patients That Mirrors Response to Enzyme Replacement Therapy. PLoS ONE, 2011, 6, e20534.	2.5	22
152	An overview on bone manifestations in Gaucher disease. Wiener Medizinische Wochenschrift, 2010, 160, 609-624.	1.1	75
153	The pathophysiology of GD â€“ current understanding and rationale for existing and emerging therapeutic approaches. Wiener Medizinische Wochenschrift, 2010, 160, 594-599.	1.1	19
154	Editorial overview: lysosomal storage disorders with primary neurological involvement. Journal of Inherited Metabolic Disease, 2010, 33, 311-312.	3.6	0
155	Long-Term Outcomes of Liver Transplantation in Type 1 Gaucher Disease. American Journal of Transplantation, 2010, 10, 1934-1939.	4.7	22
156	Patients undergoing high dose chemotherapy for primary CNS lymphoma should receive prophylactic thiamine to prevent Wernikeâ€™s encephalopathy. British Journal of Haematology, 2010, 149, 899-901.	2.5	4
157	Does geographical location influence the phenotype of Fabry disease in women in Europe?. Clinical Genetics, 2010, 77, 131-140.	2.0	17
158	Peripheral neuropathy in adult type 1 Gaucher disease: a 2-year prospective observational study. Brain, 2010, 133, 2909-2919.	7.6	73
159	Belgian Fabry Study. Stroke, 2010, 41, 863-868.	2.0	99
160	Guidelines for the restart of imiglucerase in patients with Gaucher disease: Recommendations from the European Working Group on Gaucher disease. Blood Cells, Molecules, and Diseases, 2010, 44, 86-87.	1.4	5
161	Age adjusting severity scores for Andersonâ€"Fabry Disease. Molecular Genetics and Metabolism, 2010, 101, 219-227.	1.1	22
162	Fabry Disease in Females. , 2010, , 339-351.		1

#	Article	IF	Citations
163	Enzyme-replacement therapy for Pompe disease. Pediatric Health, 2009, 3, 41-49.	0.3	0
164	To see a world in a grain of sand: elucidating the pathophysiology of Andersonâ€“Fabry disease through investigations of a cellular model. Kidney International, 2009, 75, 351-353.	5.2	7
165	The management of Gaucher disease as a chronic disorder. Clinical Therapeutics, 2009, 31, S182.	2.5	0
166	Enzyme, substrate, and myeloma in Gaucher disease. American Journal of Hematology, 2009, 84, 199-201.	4.1	10
167	Miglustat (ZavescaÂ®) in type 1 Gaucher disease: 5â€year results of a postâ€authorisation safety surveillance programme. Pharmacoepidemiology and Drug Safety, 2009, 18, 770-777.	1.9	79
168	The female Gaucher patient: The impact of enzyme replacement therapy around key reproductive events (menstruation, pregnancy and menopause). Blood Cells, Molecules, and Diseases, 2009, 43, 264-288.	1.4	53
169	Patients with Gaucher disease living in England show a high prevalence of vitamin D insufficiency with correlation to osteodensitometry. Molecular Genetics and Metabolism, 2009, 96, 113-120.	1.1	17
170	Gaucher disease and pregnancy. Journal of Obstetrics and Gynaecology, 2009, 29, 240-242.	0.9	1
171	Changes of Bone Metabolism in Seven Patients with Gaucher Disease Treated Consecutively with Imiglucerase and Miglustat. Calcified Tissue International, 2008, 83, 43-54.	3.1	24
172	Early therapeutic intervention in females with Fabry disease?. Acta Paediatrica, International Journal of Paediatrics, 2008, 97, 41-47.	1.5	21
173	The Binary Endocardial Appearance Is a Poor Discriminator of Anderson-Fabry Disease From Familial Hypertrophic Cardiomyopathy. Journal of the American College of Cardiology, 2008, 51, 2058-2061.	2.8	44
174	Muscle MRI findings in siblings with juvenile-onset acid maltase deficiency (Pompe disease). Neuromuscular Disorders, 2008, 18, 408-409.	0.6	24
175	Effective treatment of an elderly patient with Gaucher's disease and Parkinsonism: a case report of 24 monthsâ€™ oral substrate reduction therapy with miglustat.. Parkinsonism and Related Disorders, 2008, 14, 83.	2.2	0
176	The Role of Heparin in Alleviating Complement-Mediated Acute Intravascular Haemolysis. Acta Haematologica, 2008, 119, 166-168.	1.4	7
177	Effects of enzyme replacement therapy on the cardiomyopathy of Anderson Fabry disease: a randomised, double-blind, placebo-controlled clinical trial of agalsidase alfa. Heart, 2008, 94, 153-158.	2.9	269
178	Home therapy for lysosomal storage disorders. British Journal of Nursing, 2007, 16, 1384-1389.	0.7	25
179	Single Nucleotide Polymorphisms in the NOD2/CARD15 Gene Are Associated With an Increased Risk of Relapse and Death for Patients With Acute Leukemia After Hematopoietic Stem-Cell Transplantation With Unrelated Donors. Journal of Clinical Oncology, 2007, 25, 4262-4269.	1.6	58
180	Multiple myeloma: causes and consequences of delay in diagnosis. QJM - Monthly Journal of the Association of Physicians, 2007, 100, 635-640.	0.5	124

#	Article	IF	Citations
181	Effective treatment of an elderly patient with Gaucher's disease and Parkinsonism: A case report of 24 monthsâ€™ oral substrate reduction therapy with miglustat. Parkinsonism and Related Disorders, 2007, 13, 365-368.	2.2	13
182	Middelheim Fabry Study (MiFaS): A retrospective Belgian study on the prevalence of Fabry disease in young patients with cryptogenic stroke. Clinical Neurology and Neurosurgery, 2007, 109, 479-484.	1.4	60
183	Hematologic and Hemato-Oncologic Aspects of Gaucher Disease. Clinical Therapeutics, 2007, 29, S88-S90.	2.5	0
184	Fabry disease and the skin: data from FOS, the Fabry outcome survey. British Journal of Dermatology, 2007, 157, 331-337.	1.5	140
185	Recommendations for the management of the haematological and oncoâ€haematological aspects of Gaucher disease¹. British Journal of Haematology, 2007, 138, 676-686.	2.5	81
186	Extracellular matrix turnover and disease severity in Andersonâ€“Fabry disease. Journal of Inherited Metabolic Disease, 2007, 30, 88-95.	3.6	30
187	Depression in adults with Fabry disease: A common and underâ€diagnosed problem. Journal of Inherited Metabolic Disease, 2007, 30, 943-951.	3.6	93
188	Intravenous enzyme replacement therapy: better in home or hospital?. British Journal of Nursing, 2006, 15, 330-333.	0.7	58
189	The use of scoring systems in patients with haematological malignancy. Acta Paediatrica, International Journal of Paediatrics, 2006, 95, 47-51.	1.5	2
190	Retrospective Analysis of Causes and Consequence of Delay in Diagnosis of Multiple Myeloma.. Blood, 2006, 108, 5056-5056.	1.4	0
191	Prevalence and Clinical Significance of Cardiac Arrhythmia in Anderson-Fabry Disease. American Journal of Cardiology, 2005, 96, 842-846.	1.6	180
192	Natural history of Fabry disease in females in the Fabry Outcome Survey. Journal of Medical Genetics, 2005, 43, 347-352.	3.2	247
193	The natural history of left ventricular systolic function in Anderson-Fabry disease. Heart, 2005, 91, 533-534.	2.9	30
194	Vascular complications of Fabry disease: enzyme replacement and other therapies. Acta Paediatrica, International Journal of Paediatrics, 2005, 94, 28-33.	1.5	22
195	High Prevalence of B-Cell Clonality in Patients with Gaucherâ€™s Disease.. Blood, 2004, 104, 2391-2391.	1.4	4
196	Balanced Oral Substrate Reduction Therapy for Patients with Mild to Moderate Gaucherâ€™s Disease: Initial Results in Clinical Practice.. Blood, 2004, 104, 3820-3820.	1.4	1
197	The Macrophage Scavenger Receptor Type A Is Expressed by Activated Macrophages and Protects the Host Against Lethal Endotoxic Shock. Journal of Experimental Medicine, 1997, 186, 1431-1439.	8.5	264
198	Murine macrophage scavenger receptor: in vivo expression and function as receptor for macrophage adhesion in lymphoid and nonâ€lymphoid organs. European Journal of Immunology, 1995, 25, 466-473.	2.9	197

#	Article	IF	Citations
199	Macrophage-colony-stimulating factor selectively enhances macrophage scavenger receptor expression and function.. Journal of Experimental Medicine, 1994, 180, 705-709.	8.5	126
200	Murine MÏ† scavenger receptor: Adhesion function and expression. Immunology Letters, 1994, 43, 7-14.	2.5	27
201	Upregulation of the macrophage scavenger receptor in response to different forms of injury in the CNS. Journal of Neurocytology, 1994, 23, 605-613.	1.5	111
202	Use of surface molecules and receptors for studying macrophages and mononuclear phagocytes. Journal of Immunological Methods, 1994, 174, 95-102.	1.4	15
203	Divalent cation-independent macrophage adhesion inhibited by monoclonal antibody to murine scavenger receptor. Nature, 1993, 364, 343-346.	27.8	334
204	Macrophages in tissues and in vitro. Current Opinion in Immunology, 1992, 4, 25-32.	5.5	75
205	Enzyme replacement therapy for late-onset Pompe disease. The Cochrane Library, 0, , .	2.8	1
206	Lysosomal Storage Disorders. , 0, , 318-329.		0