Derralynn A Hughes

List of Publications by Year
in descending order

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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	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
2	Treatment of Fabryâ€™s Disease with the Pharmacologic Chaperone Migalastat. New England Journal of Medicine, 2016, 375, 545-555.	27.0	390
3	Divalent cation-independent macrophage adhesion inhibited by monoclonal antibody to murine scavenger receptor. Nature, 1993, 364, 343-346.	27.8	334
4	Cardiovascular magnetic resonance measurement of myocardial extracellular volume in health and disease. Heart, 2012, 98, 1436-1441.	2.9	276
5	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
6	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
7	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
8	Natural history of Fabry disease in females in the Fabry Outcome Survey. Journal of Medical Genetics, 2005, 43, 347-352.	3.2	247
9	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
10	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
11	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
12	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
13	Prevalence and Clinical Significance of Cardiac Arrhythmia in Anderson-Fabry Disease. American Journal of Cardiology, 2005, 96, 842-846.	1.6	180
14	Evolution of Prodromal Clinical Markers of Parkinson Disease in aGBAMutationâ€“Positive Cohort. JAMA Neurology, 2015, 72, 201.	9.0	180
15	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
16	Glucocerebrosidase inhibition causes mitochondrial dysfunction and free radical damage. Neurochemistry International, 2013, 62, 1-7.	3.8	166
17	Prevalence of Anderson-Fabry disease in patients with hypertrophic cardiomyopathy: the European Anderson-Fabry Disease Survey. Heart, 2011, 97, 1957-1960.	2.9	163
18	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

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19	Fabry disease and the skin: data from FOS, the Fabry outcome survey. British Journal of Dermatology, 2007, 157, 331-337.	1.5	140
20	Macrophage-colony-stimulating factor selectively enhances macrophage scavenger receptor expression and function.. Journal of Experimental Medicine, 1994, 180, 705-709.	8.5	126
21	Multiple myeloma: causes and consequences of delay in diagnosis. QJM - Monthly Journal of the Association of Physicians, 2007, 100, 635-640.	0.5	124
22	European expert consensus statement on therapeutic goals in Fabry disease. Molecular Genetics and Metabolism, 2018, 124, 189-203.	1.1	122
23	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
24	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
25	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
26	Hyposmia and cognitive impairment in Gaucher disease patients and carriers. Movement Disorders, 2012, 27, 526-532.	3.9	108
27	Osseous Manifestations of Adult Gaucher Disease in the Era of Enzyme Replacement Therapy. Medicine (United States), 2011, 90, 52-60.	1.0	104
28	Belgian Fabry Study. Stroke, 2010, 41, 863-868.	2.0	99
29	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
30	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
31	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
32	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
33	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
34	Gaucher Disease in Bone: From Pathophysiology to Practice. Journal of Bone and Mineral Research, 2019, 34, 996-1013.	2.8	94
35	Depression in adults with Fabry disease: A common and underâ€diagnosed problem. Journal of Inherited Metabolic Disease, 2007, 30, 943-951.	3.6	93
36	Proposed Stages of Myocardial Phenotype Development in FabryÂDisease. JACC: Cardiovascular Imaging, 2019, 12, 1673-1683.	5.3	91

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37	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
38	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
39	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
40	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
41	Gaucher disease: haematological presentations and complications. British Journal of Haematology, 2014, 165, 427-440.	2.5	81
42	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
43	Clinical and genetic predictors of major cardiac events in patients with Andersonâ€“Fabry Disease. Heart, 2015, 101, 961-966.	2.9	78
44	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
45	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
46	Visual short-term memory deficits associated with GBA mutation and Parkinsonâ€™s disease. Brain, 2014, 137, 2303-2311.	7.6	77
47	Macrophages in tissues and in vitro. Current Opinion in Immunology, 1992, 4, 25-32.	5.5	75
48	An overview on bone manifestations in Gaucher disease. Wiener Medizinische Wochenschrift, 2010, 160, 609-624.	1.1	75
49	Peripheral neuropathy in adult type 1 Gaucher disease: a 2-year prospective observational study. Brain, 2010, 133, 2909-2919.	7.6	73
50	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
51	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
52	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
53	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
54	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

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55	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
56	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
57	Incidence and predictors of anti-bradycardia pacing in patients with Anderson-Fabry disease. Europace, 2011, 13, 1781-1788.	1.7	63
58	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
59	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
60	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
61	Intravenous enzyme replacement therapy: better in home or hospital?. British Journal of Nursing, 2006, 15, 330-333.	0.7	58
62	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
63	Clinical prodromes of neurodegeneration in Anderson-Fabry disease. Neurology, 2015, 84, 1454-1464.	1.1	58
64	Cardiac Phenotype of Prehypertrophic Fabry Disease. Circulation: Cardiovascular Imaging, 2018, 11, e007168.	2.6	58
65	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
66	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
67	Diagnosing Gaucher disease: An on-going need for increased awareness amongst haematologists. Blood Cells, Molecules, and Diseases, 2013, 50, 212-217.	1.4	52
68	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
69	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
70	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
71	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
72	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

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73	Potential biomarkers of osteonecrosis in Gaucher disease. Blood Cells, Molecules, and Diseases, 2011, 46, 27-33.	1.4	47
74	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
75	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
76	Global longitudinal strain, myocardial storage and hypertrophy in Fabry disease. Heart, 2019, 105, 470-476.	2.9	45
77	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
78	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
79	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
80	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
81	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
82	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
83	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
84	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
85	Prevalence of CADASIL and Fabry Disease in a Cohort of MRI Defined Younger Onset Lacunar Stroke. PLoS ONE, 2015, 10, e0136352.	2.5	38
86	Lysosomal Acid Lipase Deficiency: Therapeutic Options . Drug Design, Development and Therapy, 2020, Volume 14, 591-601.	4.3	38
87	The myocardial phenotype of Fabry disease pre-hypertrophy and pre-detectable storage. European Heart Journal Cardiovascular Imaging, 2021, 22, 790-799.	1.2	35
88	Myocardial Edema, Myocyte Injury, and Disease Severity in Fabry Disease. Circulation: Cardiovascular Imaging, 2020, 13, e010171.	2.6	35
89	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
90	Cystatin C and NT-proBNP as prognostic biomarkers in Fabry disease. Molecular Genetics and Metabolism, 2011, 104, 301-307.	1.1	33

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91	Enhanced differentiation of osteoclasts from mononuclear precursors in patients with Gaucher disease. Blood Cells, Molecules, and Diseases, 2013, 51, 185-194.	1.4	33
92	Evolution and clustering of prodromal parkinsonian features in GBA1 carriers. Movement Disorders, 2019, 34, 1365-1373.	3.9	33
93	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
94	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
95	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
96	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
97	The natural history of left ventricular systolic function in Anderson-Fabry disease. Heart, 2005, 91, 533-534.	2.9	30
98	Extracellular matrix turnover and disease severity in Andersonâ€“Fabry disease. Journal of Inherited Metabolic Disease, 2007, 30, 88-95.	3.6	30
99	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
100	Retinal thinning in Gaucher disease patients and carriers: Results of a pilot study. Molecular Genetics and Metabolism, 2013, 109, 221-223.	1.1	28
101	Murine MÏ† scavenger receptor: Adhesion function and expression. Immunology Letters, 1994, 43, 7-14.	2.5	27
102	<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
103	Home therapy for lysosomal storage disorders. British Journal of Nursing, 2007, 16, 1384-1389.	0.7	25
104	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
105	Globotriaosylsphingosine (Lysoâ€Gb₃) as a biomarker for cardiac variant (N215S) Fabry disease. Journal of Inherited Metabolic Disease, 2018, 41, 239-247.	3.6	25
106	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
107	Muscle MRI findings in siblings with juvenile-onset acid maltase deficiency (Pompe disease). Neuromuscular Disorders, 2008, 18, 408-409.	0.6	24
108	Fabry International Prognostic Index: a predictive severity score for Anderson-Fabry disease. Journal of Medical Genetics, 2012, 49, 212-220.	3.2	24

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109	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
110	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
111	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
112	Vascular complications of Fabry disease: enzyme replacement and other therapies. Acta Paediatrica, International Journal of Paediatrics, 2005, 94, 28-33.	1.5	22
113	Long-Term Outcomes of Liver Transplantation in Type 1 Gaucher Disease. American Journal of Transplantation, 2010, 10, 1934-1939.	4.7	22
114	Age adjusting severity scores for Andersonâ€“Fabry Disease. Molecular Genetics and Metabolism, 2010, 101, 219-227.	1.1	22
115	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
116	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
117	Gaucher Disease and Myeloma. Critical Reviews in Oncogenesis, 2013, 18, 247-268.	0.4	22
118	Early therapeutic intervention in females with Fabry disease?. Acta Paediatrica, International Journal of Paediatrics, 2008, 97, 41-47.	1.5	21
119	Impact of sphingolipids on osteoblast and osteoclast activity in Gaucher disease. Molecular Genetics and Metabolism, 2018, 124, 278-286.	1.1	21
120	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
121	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
122	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
123	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
124	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
125	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
126	Does geographical location influence the phenotype of Fabry disease in women in Europe?. Clinical Genetics, 2010, 77, 131-140.	2.0	17

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127	The cognitive profile of type 1 Gaucher disease patients. Journal of Inherited Metabolic Disease, 2012, 35, 1093-1099.	3.6	17
128	New drugs for the treatment of Andersonâ€“Fabry disease. Journal of Nephrology, 2021, 34, 221-230.	2.0	17
129	Use of surface molecules and receptors for studying macrophages and mononuclear phagocytes. Journal of Immunological Methods, 1994, 174, 95-102.	1.4	15
130	Fabry in the older patient: Clinical consequences and possibilities for treatment. Molecular Genetics and Metabolism, 2016, 118, 319-325.	1.1	15
131	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
132	Study of indications for cardiac device implantation and utilisation in Fabry cardiomyopathy. Heart, 2019, 105, 1825-1831.	2.9	15
133	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
134	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
135	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
136	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
137	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
138	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
139	Haematological manifestations and complications of Gaucher disease. Current Opinion in Hematology, 2013, 20, 41-47.	2.5	13
140	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
141	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
142	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
143	White matter integrity correlates with cognition and disease severity in Fabry disease. Brain, 2020, 143, 3331-3342.	7.6	12
144	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

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145	Lysosomal Storage Disorders and Malignancy. Diseases (Basel, Switzerland), 2017, 5, 8.	2.5	11
146	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
147	In-depth phenotyping for clinical stratification of Gaucher disease. Orphanet Journal of Rare Diseases, 2021, 16, 431.	2.7	11
148	Enzyme, substrate, and myeloma in Gaucher disease. American Journal of Hematology, 2009, 84, 199-201.	4.1	10
149	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
150	Fabry disease and incidence of cancer. Orphanet Journal of Rare Diseases, 2017, 12, 150.	2.7	10
151	The Influence of Patient-Reported Joint Manifestations on Quality of Life in Fabry Patients. JIMD Reports, 2018, 41, 37-45.	1.5	10
152	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
153	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
154	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
155	Pseudoacromegalic facial features in Fabry disease. Clinical and Experimental Dermatology, 2013, 38, 137-139.	1.3	9
156	Fabry disease. Current Opinion in Cardiology, 2016, 31, 434-439.	1.8	9
157	A 15-Year Perspective of the Fabry Outcome Survey. FIRE Forum for International Research in Education, 2016, 4, 232640981666629.	0.7	9
158	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
159	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
160	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
161	The Role of Heparin in Alleviating Complement-Mediated Acute Intravascular Haemolysis. Acta Haematologica, 2008, 119, 166-168.	1.4	7
162	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

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163	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
164	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
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