Raphael Schiffmann

List of Publications by Year
in descending order

216 papers	16,394 citations	10979 71 h-index	18115 120 g-index
230 all docs	230 docs citations	230 times ranked	12224 citing authors

#	Article	IF	Citations
1	Enzyme Replacement Therapy in Fabry Disease. JAMA - Journal of the American Medical Association, 2001, 285, 2743.	3.8	1,141
2	Invited Article: An MRI-based approach to the diagnosis of white matter disorders. Neurology, 2009, 72, 750-759.	1.5	486
3	Mitochondrial aspartyl-tRNA synthetase deficiency causes leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation. Nature Genetics, 2007, 39, 534-539.	9.4	415
4	Neuropathology provides clues to the pathophysiology of Gaucher disease. Molecular Genetics and Metabolism, 2004, 82, 192-207.	0.5	405
5	Natural History of Fabry Renal Disease. Medicine (United States), 2002, 81, 122-138.	0.4	400
6	Treatment of Fabryâ€™s Disease with the Pharmacologic Chaperone Migalastat. New England Journal of Medicine, 2016, 375, 545-555.	13.9	390
7	Clinical and Molecular Phenotype of Aicardi-GoutiÃ¨res Syndrome. American Journal of Human Genetics, 2007, 81, 713-725.	2.6	375
8	Lamin B1 duplications cause autosomal dominant leukodystrophy. Nature Genetics, 2006, 38, 1114-1123.	9.4	365
9	Fabry disease: progression of nephropathy, and prevalence of cardiac and cerebrovascular events before enzyme replacement therapy. Nephrology Dialysis Transplantation, 2009, 24, 2102-2111.	0.4	297
10	Regional Cerebral Hyperperfusion and Nitric Oxide Pathway Dysregulation in Fabry Disease. Circulation, 2001, 104, 1506-1512.	1.6	264
11	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.	1.5	262
12	Childhood ataxia with diffuse central nervous system hypomyelination. Annals of Neurology, 1994, 35, 331-340.	2.8	253
13	Long-term therapy with agalsidase alfa for Fabry disease: safety and effects on renal function in a home infusion setting. Nephrology Dialysis Transplantation, 2006, 21, 345-354.	0.4	246
14	Mutations in CTC1, encoding conserved telomere maintenance component 1, cause Coats plus. Nature Genetics, 2012, 44, 338-342.	9.4	234
15	Randomized, controlled trial of miglustat in Gaucher's disease type 3. Annals of Neurology, 2008, 64, 514-522.	2.8	223
16	Mutations of POLR3A Encoding a Catalytic Subunit of RNA Polymerase Pol III Cause a Recessive Hypomyelinating Leukodystrophy. American Journal of Human Genetics, 2011, 89, 415-423.	2.6	219
17	Fabry disease. , 2009, 122, 65-77.		202
18	Enzyme replacement therapy improves peripheral nerve and sweat function in Fabry disease. Muscle and Nerve, 2003, 28, 703-710.	1.0	195

#	Article	IF	Citations
19	Globotriaosylceramide induces oxidative stress and up-regulates cell adhesion molecule expression in Fabry disease endothelial cells. Molecular Genetics and Metabolism, 2008, 95, 163-168.	0.5	193
20	Case definition and classification of leukodystrophies and leukoencephalopathies. Molecular Genetics and Metabolism, 2015, 114, 494-500.	0.5	185
21	Profile of endothelial and leukocyte activation in fabry patients. Annals of Neurology, 2000, 47, 229-233.	2.8	184
22	A De Novo Mutation in the Î²-Tubulin Gene TUBB4A Results in the Leukoencephalopathy Hypomyelination with Atrophy of the Basal Ganglia and Cerebellum. American Journal of Human Genetics, 2013, 92, 767-773.	2.6	174
23	Ovarian Failure Related to Eukaryotic Initiation Factor 2B Mutations. American Journal of Human Genetics, 2003, 72, 1544-1550.	2.6	172
24	Pediatric Fabry Disease. Pediatrics, 2005, 115, e344-e355.	1.0	171
25	Clinical spectrum of 4H leukodystrophy caused by POLR3A and POLR3B mutations. Neurology, 2014, 83, 1898-1905.	1.5	170
26	Elevated Cerebral Blood Flow Velocities in Fabry Disease With Reversal After Enzyme Replacement. Stroke, 2002, 33, 525-531.	1.0	161
27	Hypomyelination with atrophy of the basal ganglia and cerebellum: further delineation of the phenotype and genotypeâ€“phenotype correlation. Brain, 2014, 137, 1921-1930.	3.7	161
28	The validation of pharmacogenetics for the identification of Fabry patients to be treated with migalastat. Genetics in Medicine, 2017, 19, 430-438.	1.1	157
29	Enzyme-Replacement Therapy With Agalsidase Alfa in Children With Fabry Disease. Pediatrics, 2006, 118, 924-932.	1.0	156
30	The efficacy of enzyme replacement therapy in patients with chronic neuronopathic Gaucherâ€™s disease. Journal of Pediatrics, 2001, 138, 539-547.	0.9	151
31	Glucosylceramide and Glucosylsphingosine Modulate Calcium Mobilization from Brain Microsomes via Different Mechanisms. Journal of Biological Chemistry, 2003, 278, 23594-23599.	1.6	151
32	Agalsidase Alfa and Kidney Dysfunction in Fabry Disease. Journal of the American Society of Nephrology: JASN, 2009, 20, 1132-1139.	3.0	148
33	Retroviral Transfer of the Glucocerebrosidase Gene into CD34⁺Cells from Patients with Gaucher Disease:In VivoDetection of Transduced Cells without Myeloablation. Human Gene Therapy, 1998, 9, 2629-2640.	1.4	144
34	Phenotypic continuum in neuronopathic gaucher disease: an intermediate phenotype between type 2 and type 3. Journal of Pediatrics, 2003, 143, 273-276.	0.9	140
35	Recessive Mutations in POLR3B, Encoding the Second Largest Subunit of Pol III, Cause a Rare Hypomyelinating Leukodystrophy. American Journal of Human Genetics, 2011, 89, 652-655.	2.6	139
36	The pharmacological chaperone isofagomine increases the activity of the Gaucher disease L444P mutant form of Î²â€glucosidase. FEBS Journal, 2010, 277, 1618-1638.	2.2	135

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37	Enhanced calcium release in the acute neuronopathic form of Gaucher disease. Neurobiology of Disease, 2005, 18, 83-88.	2.1	134
38	The cerebral vasculopathy of Fabry disease. Journal of the Neurological Sciences, 2007, 257, 258-263.	0.3	134
39	Adult polyglucosan body disease: Natural History and Key Magnetic Resonance Imaging Findings. Annals of Neurology, 2012, 72, 433-441.	2.8	125
40	Increased signal intensity in the pulvinar on T1-weighted images: a pathognomonic MR imaging sign of Fabry disease. American Journal of Neuroradiology, 2003, 24, 1096-101.	1.2	124
41	Enhanced Endothelium-Dependent Vasodilation in Fabry Disease. Stroke, 2001, 32, 1559-1562.	1.0	119
42	Weekly Enzyme Replacement Therapy May Slow Decline of Renal Function in Patients with Fabry Disease Who Are on Long-Term Biweekly Dosing. Journal of the American Society of Nephrology: JASN, 2007, 18, 1576-1583.	3.0	116
43	Whole exome sequencing in patients with white matter abnormalities. Annals of Neurology, 2016, 79, 1031-1037.	2.8	116
44	Mutations in SNORD118 cause the cerebral microangiopathy leukoencephalopathy with calcifications and cysts. Nature Genetics, 2016, 48, 1185-1192.	9.4	114
45	Retroviral Transfer of the Glucocerebrosidase Gene into CD34+ Cells from Patients with Gaucher Disease: In Vivo Detection of Transduced Cells without Myeloablation. Human Gene Therapy, 1998, 9, 2629-2640.	1.4	112
46	Gaucher disease: Progress and ongoing challenges. Molecular Genetics and Metabolism, 2017, 120, 8-21.	0.5	112
47	The Relationship of Vascular Glycolipid Storage to Clinical Manifestations of Fabry Disease. Medicine (United States), 2005, 84, 261-268.	0.4	111
48	Prospective study of neurological responses to treatment with macrophage-targeted glucocerebrosidase in patients with type 3 Gaucher's disease. Annals of Neurology, 1997, 42, 613-621.	2.8	109
49	Diagnosis, prognosis, and treatment of leukodystrophies. Lancet Neurology, The, 2019, 18, 962-972.	4.9	106
50	Early Alterations of Brain Cellular Energy Homeostasis in Huntington Disease Models. Journal of Biological Chemistry, 2012, 287, 1361-1370.	1.6	104
51	Physiological characterization of neuropathy in Fabry's disease. Muscle and Nerve, 2002, 26, 622-629.	1.0	102
52	White matter lesions in Fabry disease occur in â€˜priorâ€™ selectively hypometabolic and hyperperfused brain regions. Brain Research Bulletin, 2003, 62, 231-240.	1.4	102
53	Myoclonic Epilepsy in Gaucher Disease: Genotype-Phenotype Insights from a Rare Patient Subgroup. Pediatric Research, 2003, 53, 387-395.	1.1	100
54	Cellular and tissue localization of globotriaosylceramide in Fabry disease. Virchows Archiv Fur Pathologische Anatomie Und Physiologie Und Fur Klinische Medizin, 2007, 451, 823-834.	1.4	96

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55	Bi-allelic CSF1R Mutations Cause Skeletal Dysplasia of Dysosteosclerosis-Pyle Disease Spectrum and Degenerative Encephalopathy with Brain Malformation. American Journal of Human Genetics, 2019, 104, 925-935.	2.6	92
56	New Prospects for the Treatment of Lysosomal Storage Diseases. Drugs, 2002, 62, 733-742.	4.9	91
57	Foamy cells with oligodendroglial phenotype in childhood ataxia with diffuse central nervous system hypomyelination syndrome. Acta Neuropathologica, 2000, 100, 635-646.	3.9	90
58	The latest on leukodystrophies. Current Opinion in Neurology, 2004, 17, 187-192.	1.8	89
59	Effect of genetic modifiers on cerebral lesions in Fabry disease. Neurology, 2005, 64, 2148-2150.	1.5	88
60	Update on Leukodystrophies: A Historical Perspective and Adapted Definition. Neuropediatrics, 2016, 47, 349-354.	0.3	88
61	Mapping of the Mucolipidosis Type IV Gene to Chromosome 19p and Definition of Founder Haplotypes. American Journal of Human Genetics, 1999, 65, 773-778.	2.6	87
62	Decreased guanine nucleotide exchange factor activity in eIF2B-mutated patients. European Journal of Human Genetics, 2004, 12, 561-566.	1.4	87
63	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.	1.7	86
64	New syndrome characterized by hypomyelination with atrophy of the basal ganglia and cerebellum. American Journal of Neuroradiology, 2002, 23, 1466-74.	1.2	85
65	Enzyme-replacement therapy for metabolic storage disorders. Lancet Neurology, The, 2004, 3, 752-756.	4.9	84
66	Enzyme replacement therapy and intraepidermal innervation density in Fabry disease. Muscle and Nerve, 2006, 34, 53-56.	1.0	83
67	A pharmacogenetic approach to identify mutant forms of Î±â€galactosidase a that respond to a pharmacological chaperone for Fabry disease. Human Mutation, 2011, 32, 965-977.	1.1	81
68	Pathological findings in a patient with Fabry disease who died after 2.5 years of enzyme replacement. Virchows Archiv Fur Pathologische Anatomie Und Physiologie Und Fur Klinische Medizin, 2006, 448, 337-343.	1.4	80
69	Characterization of tiger tail banding and hair shaft abnormalities in trichothiodystrophy. Journal of the American Academy of Dermatology, 2005, 52, 224-232.	0.6	79
70	Heightened stress response in primary fibroblasts expressing mutant eIF2B genes from CACH/VWM leukodystrophy patients. Human Genetics, 2005, 118, 99-106.	1.8	77
71	Mannose receptorâ€mediated delivery of mossâ€made Î±â€galactosidase A efficiently corrects enzyme deficiency in Fabry mice. Journal of Inherited Metabolic Disease, 2016, 39, 293-303.	1.7	76
72	Transfer of a mitochondrial DNA fragment toMCOLN1 causes an inherited case of mucolipidosis IV. Human Mutation, 2004, 24, 460-465.	1.1	74

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73	Leukodystrophy in patients with ovarian dysgenesis. Annals of Neurology, 1997, 41, 654-661.	2.8	73
74	Triheptanoin dramatically reduces paroxysmal motor disorder in patients with GLUT1 deficiency. Journal of Neurology, Neurosurgery and Psychiatry, 2016, 87, 550-553.	0.9	73
75	Ascorbate decreases Fabry cerebral hyperperfusion suggesting a reactive oxygen species abnormality: An arterial spin tagging study. Journal of Magnetic Resonance Imaging, 2004, 20, 674-683.	1.9	71
76	Is it Fabry disease?. Genetics in Medicine, 2016, 18, 1181-1185.	1.1	70
77	Fabry disease. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2015, 132, 231-248.	1.0	65
78	5 Neuronopathic forms of Gaucher's disease. Best Practice and Research: Clinical Haematology, 1997, 10, 711-723.	1.1	62
79	Childhood ataxia with CNS hypomyelination/vanishing white matter diseaseâ€"A common leukodystrophy caused by abnormal control of protein synthesis. Molecular Genetics and Metabolism, 2006, 88, 7-15.	0.5	62
80	The Saccadic and Neurological Deficits in Type 3 Gaucher Disease. PLoS ONE, 2011, 6, e22410.	1.1	62
81	Selective Arterial Distribution of Cerebral Hyperperfusion in Fabry Disease. Journal of Neuroimaging, 2001, 11, 303-307.	1.0	61
82	GnRH-Deficient Phenotypes in Humans and Mice with Heterozygous Variants inKISS1/Kiss1. Journal of Clinical Endocrinology and Metabolism, 2011, 96, E1771-E1781.	1.8	59
83	TUBB4A mutations result in specific neuronal and oligodendrocytic defects that closely match clinically distinct phenotypes. Human Molecular Genetics, 2017, 26, 4506-4518.	1.4	59
84	Biomarkers of Fabry Disease Nephropathy. Clinical Journal of the American Society of Nephrology: CJASN, 2010, 5, 360-364.	2.2	58
85	Decreased Bone Density in Splenectomized Gaucher Patients Receiving Enzyme Replacement Therapy. Blood Cells, Molecules, and Diseases, 2002, 28, 288-296.	0.6	57
86	4H Syndrome With Late-Onset Growth Hormone Deficiency Caused by POLR3A Mutations. Archives of Neurology, 2012, 69, 920-3.	4.9	56
87	Four-Year Prospective Clinical Trial of Agalsidase Alfa in Children with Fabry Disease. Journal of Pediatrics, 2010, 156, 832-837.e1.	0.9	54
88	Proteomics of specific treatment-related alterations in Fabry disease: A strategy to identify biological abnormalities. Proceedings of the National Academy of Sciences of the United States of America, 2007, 104, 2873-2878.	3.3	53
89	Identification of a Biomarker in Cerebrospinal Fluid for Neuronopathic Forms of Gaucher Disease. PLoS ONE, 2015, 10, e0120194.	1.1	53
90	Noninvasive diagnosis and ophthalmic features of mucolipidosis type IV. Ophthalmology, 2002, 109, 588-594.	2.5	52

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91	The definition of neuronopathic Gaucher disease. Journal of Inherited Metabolic Disease, 2020, 43, 1056-1059.	1.7	51
92	Auditory analysis of xeroderma pigmentosum 1971â€“2012: hearing function, sun sensitivity and DNA repair predict neurological degeneration. Brain, 2013, 136, 194-208.	3.7	50
93	A Founder Mutation in VPS11 Causes an Autosomal Recessive Leukoencephalopathy Linked to Autophagic Defects. PLoS Genetics, 2016, 12, e1005848.	1.5	50
94	The glycosylation design space for recombinant lysosomal replacement enzymes produced in CHO cells. Nature Communications, 2019, 10, 1785.	5.8	49
95	Screening for pharmacological chaperones in Fabry disease. Biochemical and Biophysical Research Communications, 2007, 359, 168-173.	1.0	47
96	Genetic and Clinical Heterogeneity in eIF2B-Related Disorder. Journal of Child Neurology, 2008, 23, 205-215.	0.7	46
97	Eukaryotic Initiation Factor 2B (eIF2B) GEF Activity as a Diagnostic Tool for EIF2B-Related Disorders. PLoS ONE, 2009, 4, e8318.	1.1	45
98	Altered Dopamine and Serotonin Metabolism in Motorically Asymptomatic R6/2 Mice. PLoS ONE, 2011, 6, e18336.	1.1	45
99	Disease specific therapies in leukodystrophies and leukoencephalopathies. Molecular Genetics and Metabolism, 2015, 114, 527-536.	0.5	45
100	Obstructive hypertrophic adenoids and tonsils as a cause of infantile failure to thrive: Reversed by tonsillectomy and adenoidectomy. International Journal of Pediatric Otorhinolaryngology, 1985, 9, 183-187.	0.4	43
101	An update on the leukodsytrophies. Current Opinion in Neurology, 2001, 14, 789-794.	1.8	43
102	Decreased Asialotransferrin in Cerebrospinal Fluid of Patients with Childhood-Onset Ataxia and Central Nervous System Hypomyelination/Vanishing White Matter Disease. Clinical Chemistry, 2005, 51, 2031-2042.	1.5	43
103	Quantitative dysmorphology assessment in Fabry disease. Genetics in Medicine, 2006, 8, 96-101.	1.1	43
104	Parapelvic kidney cysts: A distinguishing feature with high prevalence in Fabry disease. Kidney International, 2004, 66, 978-982.	2.6	41
105	Effect of agalsidase alfa replacement therapy on fabry diseaseâ€"related hypertrophic cardiomyopathy: A 12- to 36-month, retrospective, blinded echocardiographic pooled analysis. Clinical Therapeutics, 2009, 31, 1966-1976.	1.1	39
106	Leukodystrophy-associated POLR3A mutations down-regulate the RNA polymerase III transcript and important regulatory RNA BC200. Journal of Biological Chemistry, 2019, 294, 7445-7459.	1.6	39
107	Enzymatic and functional correction along with long-term enzyme secretion from transduced bone marrow hematopoietic stem/progenitor and stromal cells derived from patients with Fabry disease. Experimental Hematology, 1999, 27, 1149-1159.	0.2	38
108	The pharmacology of multiple regimens of agalsidase alfa enzyme replacement therapy for Fabry disease. Genetics in Medicine, 2007, 9, 504-509.	1.1	38

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109	Fabry Disease: A Disorder of Childhood Onset. Pediatric Neurology, 2016, 64, 10-20.	1.0	38
110	Enzyme Replacement in Fabry Disease: Pharmacokinetics and Pharmacodynamics of Agalsidase Alfa in Children and Adolescents. Journal of Clinical Pharmacology, 2007, 47, 1222-1230.	1.0	37
111	Therapeutic approaches for neuronopathic lysosomal storage disorders. Journal of Inherited Metabolic Disease, 2010, 33, 373-379.	1.7	37
112	Cellular and tissue distribution of intravenously administered agalsidase alfa. Molecular Genetics and Metabolism, 2007, 90, 307-312.	0.5	35
113	Time Series Proteome Profiling To Study Endoplasmic Reticulum Stress Response. Journal of Proteome Research, 2008, 7, 2435-2444.	1.8	35
114	XPC initiation codon mutation in xeroderma pigmentosum patients with and without neurological symptoms. DNA Repair, 2009, 8, 114-125.	1.3	35
115	Fabry's diseaseâ€"an important risk factor for stroke. Lancet, The, 2005, 366, 1754-1756.	6.3	34
116	Genotype-phenotype evaluation of MED13L defects in the light of a novel truncating and a recurrent missense mutation. European Journal of Medical Genetics, 2017, 60, 451-464.	0.7	34
117	Deep IntronicGBE1Mutation in Manifesting Heterozygous Patients With Adult Polyglucosan Body Disease. JAMA Neurology, 2015, 72, 441.	4.5	33
118	A prospective 10â€year study of individualized, intensified enzyme replacement therapy in advanced Fabry disease. Journal of Inherited Metabolic Disease, 2015, 38, 1129-1136.	1.7	33
119	Skin ultrastructural findings in type 2 Gaucher disease: Diagnostic implications. Molecular Genetics and Metabolism, 2011, 104, 631-636.	0.5	32
120	Arterial Wall Properties and Womersley Flow in Fabry Disease. BMC Cardiovascular Disorders, 2002, 2, 1.	0.7	31
121	Agalsidase alfa in pediatric patients with Fabry disease: a 6.5-year open-label follow-up study. Orphanet Journal of Rare Diseases, 2014, 9, 169.	1.2	31
122	Diffuse Neuroaxonal Involvement in Mucolipidosis IV as Assessed by Proton Magnetic Resonance Spectroscopic Imaging. Journal of Child Neurology, 2003, 18, 443-449.	0.7	30
123	Lysosomal Inclusions in Gastric Parietal Cells in Mucolipidosis Type IV. American Journal of Surgical Pathology, 1999, 23, 1527.	2.1	30
124	CSF and Blood Levels of GFAP in Alexander Disease. ENeuro, 2015, 2, ENEURO.0080-15.2015.	0.9	30
125	Quantitative neuroimaging in mucolipidosis type IV. Molecular Genetics and Metabolism, 2014, 111, 147-151.	0.5	29
126	Neuropathy and Fabry disease: pathogenesis and enzyme replacement therapy. Acta Neurologica Belgica, 2006, 106, 61-5.	0.5	29

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127	More Than Hypomyelination in Pol-III Disorder. Journal of Neuropathology and Experimental Neurology, 2013, 72, 67-75.	0.9	27
128	Abnormal glycogen in astrocytes is sufficient to cause adult polyglucosan body disease. Gene, 2013, 515, 376-379.	1.0	26
129	Genome sequencing in persistently unsolved white matter disorders. Annals of Clinical and Translational Neurology, 2020, 7, 144-152.	1.7	26
130	Endocrine and Growth Abnormalities in 4H Leukodystrophy Caused by Variants in POLR3A, POLR3B, and POLR1C. Journal of Clinical Endocrinology and Metabolism, 2021, 106, e660-e674.	1.8	26
131	Assessment of plasma lyso-Gb3 for clinical monitoring of treatment response in migalastat-treated patients with Fabry disease. Genetics in Medicine, 2021, 23, 192-201.	1.1	26
132	Establishment and characterization of Fabry disease endothelial cells with an extended lifespan. Molecular Genetics and Metabolism, 2007, 92, 137-144.	0.5	25
133	Improved intracellular delivery of glucocerebrosidase mediated by the HIV-1 TAT protein transduction domain. Biochemical and Biophysical Research Communications, 2005, 337, 701-707.	1.0	23
134	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.	1.2	23
135	Gaucher mutation N188S is associated with myoclonic epilepsy. Human Mutation, 2005, 26, 271-273.	1.1	22
136	Risk of Death in Heart Disease is Associated With Elevated Urinary Globotriaosylceramide. Journal of the American Heart Association, 2014, 3, e000394.	1.6	22
137	Fabry disease genotype, phenotype, and migalastat amenability: Insights from a national cohort. Journal of Inherited Metabolic Disease, 2020, 43, 326-333.	1.7	22
138	PLP1 and GPM6B intragenic copy number analysis by MAPH in 262 patients with hypomyelinating leukodystrophies: identification of one partial triplication and two partial deletions of PLP1. Neurogenetics, 2006, 7, 31-37.	0.7	21
139	Elevated Endothelial Microparticles in Fabry Children Decreased After Enzyme Replacement Therapy. Arteriosclerosis, Thrombosis, and Vascular Biology, 2007, 27, e138-9.	1.1	21
140	Sex differences of urinary and kidney globotriaosylceramide and lyso-globotriaosylceramide in Fabry mice. Journal of Lipid Research, 2011, 52, 1742-1746.	2.0	21
141	Changes in plasma and urine globotriaosylceramide levels do not predict Fabry disease progression over 1 year of agalsidase alfa. Genetics in Medicine, 2013, 15, 983-989.	1.1	21
142	Blocking hyperactive androgen receptor signaling ameliorates cardiac and renal hypertrophy in Fabry mice. Human Molecular Genetics, 2015, 24, 3181-3191.	1.4	21
143	Enzyme Replacement in Fabry Disease: The Essence Is in the Kidney. Annals of Internal Medicine, 2007, 146, 142.	2.0	20
144	Urinary 11-Dehydro-Thromboxane B 2 and Mortality in Patients With Stable Coronary Artery Disease. American Journal of Cardiology, 2017, 119, 972-977.	0.7	20

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145	Oculomotor and Vestibular Findings in Gaucher Disease Type 3 and Their Correlation with Neurological Findings. Frontiers in Neurology, 2017, 8, 711.	1.1	20
146	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.	0.8	20
147	Characterization of transferrin glycopeptide structures in human cerebrospinal fluid. International Journal of Mass Spectrometry, 2012, 312, 97-106.	0.7	19
148	Long-term follow-up and sudden unexpected death in Gaucher disease type 3 in Egypt. Neurology: Genetics, 2016, 2, e55.	0.9	17
149	A doubleâ€blind, placeboâ€controlled trial of triheptanoin in adult polyglucosan body disease and openâ€label, longâ€term outcome. Journal of Inherited Metabolic Disease, 2018, 41, 877-883.	1.7	17
150	Symptoms and Quality of Life in Patients with Fabry Disease: Results from an International Patient Survey. Advances in Therapy, 2019, 36, 2866-2880.	1.3	17
151	Unique molecular signature in mucolipidosis type IV microglia. Journal of Neuroinflammation, 2019, 16, 276.	3.1	17
152	Randomized Clinical Trial of <scp>Firstâ€Line</scp> Genome Sequencing in Pediatric White Matter Disorders. Annals of Neurology, 2020, 88, 264-273.	2.8	17
153	Tetrahydrobiopterin deficiency in the pathogenesis of Fabry disease. Human Molecular Genetics, 2017, 26, 1182-1192.	1.4	16
154	A Physical and Transcript Map of the MCOLN1 Gene Region on Human Chromosome 19p13.3â€“p13.2. Genomics, 2001, 73, 203-210.	1.3	15
155	Genomic abnormalities of the murine model of Fabry disease after disease-related perturbation, a systems biology approach. Proceedings of the National Academy of Sciences of the United States of America, 2007, 104, 8065-8070.	3.3	15
156	An open-label clinical trial of agalsidase alfa enzyme replacement therapy in children with Fabry disease who are naïve to enzyme replacement therapy. Drug Design, Development and Therapy, 2016, 10, 1771.	2.0	15
157	An autosomal recessive form of benign familial neonatal seizures. Clinical Genetics, 1991, 40, 467-470.	1.0	14
158	Developmental Splicing Deregulation in Leukodystrophies Related to EIF2B Mutations. PLoS ONE, 2012, 7, e38264.	1.1	14
159	Dystonia in RNA Polymerase IIIâ€Related Leukodystrophy. Movement Disorders Clinical Practice, 2019, 6, 155-159.	0.8	14
160	Long-term follow-up of renal function in patients treated with migalastat for Fabry disease. Molecular Genetics and Metabolism Reports, 2021, 28, 100786.	0.4	14
161	Unexpected Occurrence of Xeroderma Pigmentosum in an Uncle and Nephew. Archives of Dermatology, 2009, 145, 1285-91.	1.7	13
162	Variation in cognitive function over time in Gaucher disease type 3. Neurology, 2019, 93, e2272-e2283.	1.5	13

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163	Falsely elevated urinary Gb3 (globotriaosylceramide, CTH, GL3). Molecular Genetics and Metabolism, 2009, 97, 91.	0.5	12
164	Cerebrotendinous xanthomatosis. Neurology, 2019, 92, 61-62.	1.5	12
165	Myoclonus in Gaucher disease. Advances in Neurology, 2002, 89, 41-8.	0.8	12
166	Fabry Disease: Angiokeratoma, Biomarker, and the Effect of Enzyme Replacement Therapy on Kidney Function. Archives of Dermatology, 2005, 141, 904-5; author reply 905-6.	1.7	11
167	Functionally pathogenic EARS2 variants in vitro may not manifest a phenotype in vivo. Neurology: Genetics, 2017, 3, e162.	0.9	11
168	Electroencephalographic findings in patients with mucolipidosis type IV. Electroencephalography and Clinical Neurophysiology, 1998, 106, 400-403.	0.3	10
169	Oxidative stress reflected by increased F2-isoprostanes is associated with increasing urinary 11-dehydro thromboxane B2 levels in patients with coronary artery disease. Thrombosis Research, 2016, 148, 85-88.	0.8	10
170	Five novel mutations in fourteen patients with Fabry disease. Human Mutation, 2000, 15, 207-208.	1.1	9
171	Health-Related Quality of Life for Patients With Genetically Determined Leukoencephalopathy. Pediatric Neurology, 2018, 84, 21-26.	1.0	9
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