Raphael Schiffmann

List of Publications by Year
in descending order

216	16,394	10986	18130
papers	citations	71 h-index	120 g-index

230	230	230	12224
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Brain pathology and cerebellar purkinje cell loss in a mouse model of chronic neuronopathic Gaucher disease. Progress in Neurobiology, 2021, 197, 101939.	5.7	6
2	Cerebral Microangiopathy in Leukoencephalopathy With Cerebral Calcifications and Cysts: A Pathological Description. Journal of Child Neurology, 2021, 36, 133-140.	1.4	3
3	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.	3.6	26
4	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.	2.4	26
5	Migalastat Tissue Distribution: Extrapolation From Mice to Humans Using Pharmacokinetic Modeling and Comparison With Agalsidase Beta Tissue Distribution in Mice. Clinical Pharmacology in Drug Development, 2021, 10, 1075-1088.	1.6	4
6	White matter abnormalities and iron deposition in prenatal mucolipidosis IV- fetal imaging and pathology. Metabolic Brain Disease, 2021, 36, 2155-2167.	2.9	6
7	Expanded phenotype of AARS1-related white matter disease. Genetics in Medicine, 2021, 23, 2352-2359.	2.4	8
8	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
9	Investigation of a dysmorphic facial phenotype in patients with Gaucher disease types 2 and 3. Molecular Genetics and Metabolism, 2021, 134, 274-280.	1.1	3
10	Fabry disease genotype, phenotype, and migalastat amenability: Insights from a national cohort. Journal of Inherited Metabolic Disease, 2020, 43, 326-333.	3.6	22
11	Genome sequencing in persistently unsolved white matter disorders. Annals of Clinical and Translational Neurology, 2020, 7, 144-152.	3.7	26
12	EEG abnormalities in patients with chronic neuronopathic Gaucher disease: A retrospective review. Molecular Genetics and Metabolism, 2020, 131, 358-363.	1.1	7
13	Assessing the role of glycosphingolipids in the phenotype severity of Fabry disease mouse model. Journal of Lipid Research, 2020, 61, 1410-1423.	4.2	7
14	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
15	Randomized Clinical Trial of <scp>First‐Line</scp> Genome Sequencing in Pediatric White Matter Disorders. Annals of Neurology, 2020, 88, 264-273.	5.3	17
16	The definition of neuronopathic Gaucher disease. Journal of Inherited Metabolic Disease, 2020, 43, 1056-1059.	3.6	51
17	Gaucher diseaseâ€"neuronopathic forms. , 2020, , 439-449.		0
18	The migalastat GLP-HEK assay is the gold standard for determining amenability in patients with Fabry disease. Molecular Genetics and Metabolism Reports, 2019, 20, 100494.	1.1	5

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19	Symptoms and Quality of Life in Patients with Fabry Disease: Results from an International Patient Survey. Advances in Therapy, 2019, 36, 2866-2880.	2.9	17
20	Diagnosis, prognosis, and treatment of leukodystrophies. Lancet Neurology, The, 2019, 18, 962-972.	10.2	106
21	The glycosylation design space for recombinant lysosomal replacement enzymes produced in CHO cells. Nature Communications, 2019, 10, 1785.	12.8	49
22	GJA1 Variants Cause Spastic Paraplegia Associated with Cerebral Hypomyelination. American Journal of Neuroradiology, 2019, 40, 788-791.	2.4	4
23	Leukodystrophy-associated POLR3A mutations down-regulate the RNA polymerase III transcript and important regulatory RNA BC200. Journal of Biological Chemistry, 2019, 294, 7445-7459.	3.4	39
24	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
25	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.	6.2	92
26	Effects of genetic background on disease phenotypes in a mouse model of Fabry disease. Molecular Genetics and Metabolism, 2019, 126, S79.	1.1	0
27	Dysregulated DNA methylation in the pathogenesis of Fabry disease. Molecular Genetics and Metabolism, 2019, 126, S134.	1.1	0
28	Once every 4 weeks - 2 mg/kg of pegunigalsidase alfa for treating Fabry disease Preliminary results of a phase 3 study. Molecular Genetics and Metabolism, 2019, 126, S73.	1.1	5
29	Venglustat in adult Gaucher disease type 3: Preliminary safety, pharmacology, and exploratory efficacy from a phase 2 trial in combination with imiglucerase (LEAP). Molecular Genetics and Metabolism, 2019, 126, S131.	1.1	5
30	Variation in cognitive function over time in Gaucher disease type 3. Neurology, 2019, 93, e2272-e2283.	1.1	13
31	Unique molecular signature in mucolipidosis type IV microglia. Journal of Neuroinflammation, 2019, 16, 276.	7.2	17
32	Cerebrotendinous xanthomatosis. Neurology, 2019, 92, 61-62.	1.1	12
33	Dystonia in RNA Polymerase IIIâ€Related Leukodystrophy. Movement Disorders Clinical Practice, 2019, 6, 155-159.	1.5	14
34	The natural history of cognition in Gaucher disease type 3. Molecular Genetics and Metabolism, 2019, 126, S139.	1.1	0
35	Enhanced pharmacokinetics profile of pegunigalsidase alfa (PRX-102) supports once-monthly 2mg/kg dosing for the treatment of Fabry disease. Molecular Genetics and Metabolism, 2018, 123, S145-S146.	1.1	1
36	Health-Related Quality of Life for Patients With Genetically Determined Leukoencephalopathy. Pediatric Neurology, 2018, 84, 21-26.	2.1	9

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37	Low frequency of Fabry disease in patients with common heart disease. Genetics in Medicine, 2018, 20, 754-759.	2.4	7
38	Prognostic value of urinary 11â€dehydroâ€thromboxane B₂ for mortality: A cohort study of stable coronary artery disease patients treated with aspirin. Catheterization and Cardiovascular Interventions, 2018, 92, 653-658.	1.7	6
39	Priapism in a Fabry disease mouse model is associated with upregulated penile nNOS and eNOS expression. Journal of Inherited Metabolic Disease, 2018, 41, 231-238.	3.6	8
40	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.	3.6	17
41	Priapism in a Fabry disease mouse model is associated with upregulated penile nNOS and eNOS expression. Molecular Genetics and Metabolism, 2018, 123, S130.	1.1	0
42	Migalastat for the treatment of Fabry disease. Expert Opinion on Orphan Drugs, 2018, 6, 301-309.	0.8	9
43	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
44	Roscoe Owen Brady, MD: Remembrances of co-investigators and colleagues. Molecular Genetics and Metabolism, 2017, 120, 1-7.	1.1	3
45	Urinary 11-Dehydro-Thromboxane B 2 and Mortality in Patients With Stable Coronary Artery Disease. American Journal of Cardiology, 2017, 119, 972-977.	1.6	20
46	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
47	Residual thromboxane activity and oxidative stress. Coronary Artery Disease, 2017, 28, 287-293.	0.7	8
48	Tetrahydrobiopterin deficiency in the pathogenesis of Fabry disease. Human Molecular Genetics, 2017, 26, 1182-1192.	2.9	16
49	One-year follow up of Fabry disease patients treated by IV administration of a plant derived alpha-Gal-A enzyme: safety and efficacy. Molecular Genetics and Metabolism, 2017, 120, S68.	1.1	1
50	TUBB4A mutations result in specific neuronal and oligodendrocytic defects that closely match clinically distinct phenotypes. Human Molecular Genetics, 2017, 26, 4506-4518.	2.9	59
51	Functionally pathogenic EARS2 variants in vitro may not manifest a phenotype in vivo. Neurology: Genetics, 2017, 3, e162.	1.9	11
52	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.	1.3	34
53	Gaucher disease: Progress and ongoing challenges. Molecular Genetics and Metabolism, 2017, 120, 8-21.	1.1	112
54	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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55	Oculomotor and Vestibular Findings in Gaucher Disease Type 3 and Their Correlation with Neurological Findings. Frontiers in Neurology, 2017, 8, 711.	2.4	20
56	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.	4.3	15
57	Whole exome sequencing in patients with white matter abnormalities. Annals of Neurology, 2016, 79, 1031-1037.	5.3	116
58	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.	1.7	10
59	Is it Fabry disease?. Genetics in Medicine, 2016, 18, 1181-1185.	2.4	70
60	Long-term follow-up and sudden unexpected death in Gaucher disease type 3 in Egypt. Neurology: Genetics, 2016, 2, e55.	1.9	17
61	Treatment of Fabryâ€™s Disease with the Pharmacologic Chaperone Migalastat. New England Journal of Medicine, 2016, 375, 545-555.	27.0	390
62	Update on Leukodystrophies: A Historical Perspective and Adapted Definition. Neuropediatrics, 2016, 47, 349-354.	0.6	88
63	Fabry Disease: A Disorder of Childhood Onset. Pediatric Neurology, 2016, 64, 10-20.	2.1	38
64	Mutations in SNORD118 cause the cerebral microangiopathy leukoencephalopathy with calcifications and cysts. Nature Genetics, 2016, 48, 1185-1192.	21.4	114
65	Brain MRI and motor function in leukodystrophies. Neurology, 2016, 87, 748-749.	1.1	0
66	Molecular basis for globotriaosylceramide regulation and enzyme uptake in immortalized aortic endothelial cells from Fabry mice. Journal of Inherited Metabolic Disease, 2016, 39, 447-455.	3.6	1
67	Triheptanoin dramatically reduces paroxysmal motor disorder in patients with GLUT1 deficiency. Journal of Neurology, Neurosurgery and Psychiatry, 2016, 87, 550-553.	1.9	73
68	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.	3.6	76
69	A Founder Mutation in VPS11 Causes an Autosomal Recessive Leukoencephalopathy Linked to Autophagic Defects. PLoS Genetics, 2016, 12, e1005848.	3.5	50
70	Gaucher Disease. , 2015, , 301-311.		0
71	Fabry disease. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2015, 132, 231-248.	1.8	65
72	Identification of a Biomarker in Cerebrospinal Fluid for Neuronopathic Forms of Gaucher Disease. PLoS ONE, 2015, 10, e0120194.	2.5	53

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73	Leukoencephalopathy: "Before concluding treatment efficacy...". Neurology, 2015, 84, 218-219.	1.1	3
74	A genetic form of achlorhydria and gastritis. American Journal of Clinical Nutrition, 2015, 102, 1615.	4.7	0
75	Disease specific therapies in leukodystrophies and leukoencephalopathies. Molecular Genetics and Metabolism, 2015, 114, 527-536.	1.1	45
76	Case definition and classification of leukodystrophies and leukoencephalopathies. Molecular Genetics and Metabolism, 2015, 114, 494-500.	1.1	185
77	Deep IntronicGBE1Mutation in Manifesting Heterozygous Patients With Adult Polyglucosan Body Disease. JAMA Neurology, 2015, 72, 441.	9.0	33
78	A prospective 10â€year study of individualized, intensified enzyme replacement therapy in advanced Fabry disease. Journal of Inherited Metabolic Disease, 2015, 38, 1129-1136.	3.6	33
79	The consequences of genetic and pharmacologic reduction in sphingolipid synthesis. Journal of Inherited Metabolic Disease, 2015, 38, 77-84.	3.6	8
80	Neurogenic bladder and neuroendocrine abnormalities in Pol III-related leukodystrophy. BMC Neurology, 2015, 15, 22.	1.8	6
81	Blocking hyperactive androgen receptor signaling ameliorates cardiac and renal hypertrophy in Fabry mice. Human Molecular Genetics, 2015, 24, 3181-3191.	2.9	21
82	CSF and Blood Levels of GFAP in Alexander Disease. ENeuro, 2015, 2, ENEURO.0080-15.2015.	1.9	30
83	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.	2.7	31
84	Risk of Death in Heart Disease is Associated With Elevated Urinary Globotriaosylceramide. Journal of the American Heart Association, 2014, 3, e000394.	3.7	22
85	Quantitative neuroimaging in mucolipidosis type IV. Molecular Genetics and Metabolism, 2014, 111, 147-151.	1.1	29
86	Clinical spectrum of 4H leukodystrophy caused by POLR3A and POLR3B mutations. Neurology, 2014, 83, 1898-1905.	1.1	170
87	Hypomyelination with atrophy of the basal ganglia and cerebellum: further delineation of the phenotype and genotypeâ€“phenotype correlation. Brain, 2014, 137, 1921-1930.	7.6	161
88	Abnormal glycogen in astrocytes is sufficient to cause adult polyglucosan body disease. Gene, 2013, 515, 376-379.	2.2	26
89	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.	6.2	174
90	Auditory analysis of xeroderma pigmentosum 1971â€“2012: hearing function, sun sensitivity and DNA repair predict neurological degeneration. Brain, 2013, 136, 194-208.	7.6	50

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91	Reply. Annals of Neurology, 2013, 73, 318-318.	5.3	0
92	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.	2.4	21
93	HIV Tat Domain Improves Cross-correction of Human Galactocerebrosidase in a Gene- and Flanking Sequence-dependent Manner. Molecular Therapy - Nucleic Acids, 2013, 2, e130.	5.1	8
94	More Than Hypomyelination in Pol-III Disorder. Journal of Neuropathology and Experimental Neurology, 2013, 72, 67-75.	1.7	27
95	TACH Leukodystrophy: Locus Refinement to Chromosome 10q22.3-23.1. Canadian Journal of Neurological Sciences, 2012, 39, 122-123.	0.5	4
96	GnRH-Deficient Phenotypes in Humans and Mice With Heterozygous Variants in KISS1/Kiss1. Obstetrical and Gynecological Survey, 2012, 67, 546-547.	0.4	0
97	4H Syndrome With Late-Onset Growth Hormone Deficiency Caused by POLR3A Mutations. Archives of Neurology, 2012, 69, 920-3.	4.5	56
98	Adult polyglucosan body disease: Natural History and Key Magnetic Resonance Imaging Findings. Annals of Neurology, 2012, 72, 433-441.	5.3	125
99	Mutations in CTC1, encoding conserved telomere maintenance component 1, cause Coats plus. Nature Genetics, 2012, 44, 338-342.	21.4	234
100	Developmental Splicing Deregulation in Leukodystrophies Related to EIF2B Mutations. PLoS ONE, 2012, 7, e38264.	2.5	14
101	Early Alterations of Brain Cellular Energy Homeostasis in Huntington Disease Models. Journal of Biological Chemistry, 2012, 287, 1361-1370.	3.4	104
102	Characterization of transferrin glycopeptide structures in human cerebrospinal fluid. International Journal of Mass Spectrometry, 2012, 312, 97-106.	1.5	19
103	Skin ultrastructural findings in type 2 Gaucher disease: Diagnostic implications. Molecular Genetics and Metabolism, 2011, 104, 631-636.	1.1	32
104	Altered Dopamine and Serotonin Metabolism in Motorically Asymptomatic R6/2 Mice. PLoS ONE, 2011, 6, e18336.	2.5	45
105	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.	6.2	219
106	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.	6.2	139
107	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.	2.5	81
108	GnRH-Deficient Phenotypes in Humans and Mice with Heterozygous Variants inKISS1/Kiss1. Journal of Clinical Endocrinology and Metabolism, 2011, 96, E1771-E1781.	3.6	59

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109	Sex differences of urinary and kidney globotriaosylceramide and lyso-globotriaosylceramide in Fabry mice. Journal of Lipid Research, 2011, 52, 1742-1746.	4.2	21
110	The Saccadic and Neurological Deficits in Type 3 Gaucher Disease. PLoS ONE, 2011, 6, e22410.	2.5	62
111	Therapeutic approaches for neuronopathic lysosomal storage disorders. Journal of Inherited Metabolic Disease, 2010, 33, 373-379.	3.6	37
112	Four-Year Prospective Clinical Trial of Agalsidase Alfa in Children with Fabry Disease. Journal of Pediatrics, 2010, 156, 832-837.e1.	1.8	54
113	The pharmacological chaperone isofagomine increases the activity of the Gaucher disease L444P mutant form of ‹b›Î²‹/b›â€glucosidase. FEBS Journal, 2010, 277, 1618-1638.	4.7	135
114	Use of lissamine rhodamine ceramide trihexoside as a functional assay for alpha-galactosidase A in intact cells. Journal of Lipid Research, 2010, 51, 2808-2817.	4.2	6
115	Biomarkers of Fabry Disease Nephropathy. Clinical Journal of the American Society of Nephrology: CJASN, 2010, 5, 360-364.	4.5	58
116	Agalsidase treatment for Fabry disease: Uses and rivalries. Genetics in Medicine, 2010, 12, 684-685.	2.4	3
117	Unexpected Occurrence of Xeroderma Pigmentosum in an Uncle and Nephew. Archives of Dermatology, 2009, 145, 1285-91.	1.4	13
118	Agalsidase Alfa and Kidney Dysfunction in Fabry Disease. Journal of the American Society of Nephrology: JASN, 2009, 20, 1132-1139.	6.1	148
119	Fabry disease. , 2009, 122, 65-77.		202
120	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.	2.5	39
121	XPC initiation codon mutation in xeroderma pigmentosum patients with and without neurological symptoms. DNA Repair, 2009, 8, 114-125.	2.8	35
122	Fabry disease: progression of nephropathy, and prevalence of cardiac and cerebrovascular events before enzyme replacement therapy. Nephrology Dialysis Transplantation, 2009, 24, 2102-2111.	0.7	297
123	Falsely elevated urinary Gb3 (globotriaosylceramide, CTH, GL3). Molecular Genetics and Metabolism, 2009, 97, 91.	1.1	12
124	Invited Article: An MRI-based approach to the diagnosis of white matter disorders. Neurology, 2009, 72, 750-759.	1.1	486
125	Eukaryotic Initiation Factor 2B (eIF2B) GEF Activity as a Diagnostic Tool for EIF2B-Related Disorders. PLoS ONE, 2009, 4, e8318.	2.5	45
126	The sub-cellular localization globotriaosylceramide in Fabry disease. Virchows Archiv Fur Pathologische Anatomie Und Physiologie Und Fur Klinische Medizin, 2008, 452, 707-708.	2.8	2

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127	Randomized, controlled trial of miglustat in Gaucher's disease type 3. Annals of Neurology, 2008, 64, 514-522.	5.3	223
128	Time Series Proteome Profiling To Study Endoplasmic Reticulum Stress Response. Journal of Proteome Research, 2008, 7, 2435-2444.	3.7	35
129	Globotriaosylceramide induces oxidative stress and up-regulates cell adhesion molecule expression in Fabry disease endothelial cells. Molecular Genetics and Metabolism, 2008, 95, 163-168.	1.1	193
130	Genetic and Clinical Heterogeneity in eIF2B-Related Disorder. Journal of Child Neurology, 2008, 23, 205-215.	1.4	46
131	The pharmacology of multiple regimens of agalsidase alfa enzyme replacement therapy for Fabry disease. Genetics in Medicine, 2007, 9, 504-509.	2.4	38
132	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.	6.1	116
133	Enzyme Replacement in Fabry Disease: Pharmacokinetics and Pharmacodynamics of Agalsidase Alfa in Children and Adolescents. Journal of Clinical Pharmacology, 2007, 47, 1222-1230.	2.0	37
134	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.	7.1	53
135	Elevated Endothelial Microparticles in Fabry Children Decreased After Enzyme Replacement Therapy. Arteriosclerosis, Thrombosis, and Vascular Biology, 2007, 27, e138-9.	2.4	21
136	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.	7.1	15
137	Enzyme Replacement in Fabry Disease: The Essence Is in the Kidney. Annals of Internal Medicine, 2007, 146, 142.	3.9	20
138	Cellular and tissue distribution of intravenously administered agalsidase alfa. Molecular Genetics and Metabolism, 2007, 90, 307-312.	1.1	35
139	Establishment and characterization of Fabry disease endothelial cells with an extended lifespan. Molecular Genetics and Metabolism, 2007, 92, 137-144.	1.1	25
140	Screening for pharmacological chaperones in Fabry disease. Biochemical and Biophysical Research Communications, 2007, 359, 168-173.	2.1	47
141	Clinical and Molecular Phenotype of Aicardi-GoutiÃ¨res Syndrome. American Journal of Human Genetics, 2007, 81, 713-725.	6.2	375
142	The cerebral vasculopathy of Fabry disease. Journal of the Neurological Sciences, 2007, 257, 258-263.	0.6	134
143	Mitochondrial aspartyl-tRNA synthetase deficiency causes leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation. Nature Genetics, 2007, 39, 534-539.	21.4	415
144	Cellular and tissue localization of globotriaosylceramide in Fabry disease. Virchows Archiv Fur Pathologische Anatomie Und Physiologie Und Fur Klinische Medizin, 2007, 451, 823-834.	2.8	96

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145	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.	1.1	62
146	Lamin B1 duplications cause autosomal dominant leukodystrophy. Nature Genetics, 2006, 38, 1114-1123.	21.4	365
147	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.	1.4	21
148	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.	2.8	80
149	The significance of lysosomal inclusions in Fabry disease. Virchows Archiv Fur Pathologische Anatomie Und Physiologie Und Fur Klinische Medizin, 2006, 449, 134-134.	2.8	1
150	Enzyme replacement therapy and intraepidermal innervation density in Fabry disease. Muscle and Nerve, 2006, 34, 53-56.	2.2	83
151	Quantitative dysmorphology assessment in Fabry disease. Genetics in Medicine, 2006, 8, 96-101.	2.4	43
152	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.	0.7	246
153	Enzyme-Replacement Therapy With Agalsidase Alfa in Children With Fabry Disease. Pediatrics, 2006, 118, 924-932.	2.1	156
154	Elevated Counts of Circulating Endothelial Microparticles in Pediatric Fabry Patients Decreased after Enzyme Replacement Therapy.. Blood, 2006, 108, 1818-1818.	1.4	0
155	Neuropathy and Fabry disease: pathogenesis and enzyme replacement therapy. Acta Neurologica Belgica, 2006, 106, 61-5.	1.1	29
156	Gaucher mutation N188S is associated with myoclonic epilepsy. Human Mutation, 2005, 26, 271-273.	2.5	22
157	Heightened stress response in primary fibroblasts expressing mutant eIF2B genes from CACH/VWM leukodystrophy patients. Human Genetics, 2005, 118, 99-106.	3.8	77
158	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.	3.2	43
159	Pediatric Fabry Disease. Pediatrics, 2005, 115, e344-e355.	2.1	171
160	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.	1.4	11
161	Effect of genetic modifiers on cerebral lesions in Fabry disease. Neurology, 2005, 64, 2148-2150.	1.1	88
162	The Relationship of Vascular Glycolipid Storage to Clinical Manifestations of Fabry Disease. Medicine (United States), 2005, 84, 261-268.	1.0	111

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163	Fabry's diseaseâ€"an important risk factor for stroke. Lancet, The, 2005, 366, 1754-1756.	13.7	34
164	Enhanced calcium release in the acute neuronopathic form of Gaucher disease. Neurobiology of Disease, 2005, 18, 83-88.	4.4	134
165	Characterization of tiger tail banding and hair shaft abnormalities in trichothiodystrophy. Journal of the American Academy of Dermatology, 2005, 52, 224-232.	1.2	79
166	Improved intracellular delivery of glucocerebrosidase mediated by the HIV-1 TAT protein transduction domain. Biochemical and Biophysical Research Communications, 2005, 337, 701-707.	2.1	23
167	Parapelvic kidney cysts: A distinguishing feature with high prevalence in Fabry disease. Kidney International, 2004, 66, 978-982.	5.2	41
168	Decreased guanine nucleotide exchange factor activity in eIF2B-mutated patients. European Journal of Human Genetics, 2004, 12, 561-566.	2.8	87
169	Enzyme-replacement therapy for metabolic storage disorders. Lancet Neurology, The, 2004, 3, 752-756.	10.2	84
170	Transfer of a mitochondrial DNA fragment toMCOLN1 causes an inherited case of mucolipidosis IV. Human Mutation, 2004, 24, 460-465.	2.5	74
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