Raphael Schiffmann

List of Publications by Citations

205 papers	13,095 citations	67 h-index	109 g-index
230 ext. papers	14,852 ext. citations	6.7 avg, IF	6.03 L-index

#	Paper	IF	Citations
205	Enzyme replacement therapy in Fabry disease: a randomized controlled trial. *JAMA - Journal of the American Medical Association*, **2001**, 285, 2743-9	27.4	930
204	Invited article: an MRI-based approach to the diagnosis of white matter disorders. *Neurology*, **2009**, 72, 750-9	6.5	395
203	Mitochondrial aspartyl-tRNA synthetase deficiency causes leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation. *Nature Genetics*, **2007**, 39, 534-9	36.3	363
202	Neuropathology provides clues to the pathophysiology of Gaucher disease. *Molecular Genetics and Metabolism*, **2004**, 82, 192-207	3.7	360
201	Lamin B1 duplications cause autosomal dominant leukodystrophy. *Nature Genetics*, **2006**, 38, 1114-23	36.3	312
200	Clinical and molecular phenotype of Aicardi-Goutieres syndrome. *American Journal of Human Genetics*, **2007**, 81, 713-25	11	310
199	Natural history of Fabry renal disease: influence of alpha-galactosidase A activity and genetic mutations on clinical course. *Medicine (United States)*, **2002**, 81, 122-38	1.8	307
198	Treatment of Fabry's Disease with the Pharmacologic Chaperone Migalastat. *New England Journal of Medicine*, **2016**, 375, 545-55	59.2	254
197	Fabry disease: progression of nephropathy, and prevalence of cardiac and cerebrovascular events before enzyme replacement therapy. *Nephrology Dialysis Transplantation*, **2009**, 24, 2102-11	4.3	252
196	Regional cerebral hyperperfusion and nitric oxide pathway dysregulation in Fabry disease: reversal by enzyme replacement therapy. *Circulation*, **2001**, 104, 1506-12	16.7	240
195	Childhood ataxia with diffuse central nervous system hypomyelination. *Annals of Neurology*, **1994**, 35, 331-40	9.4	219
194	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, 345-54	4.3	207
193	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	5.8	193
192	Mutations in CTC1, encoding conserved telomere maintenance component 1, cause Coats plus. *Nature Genetics*, **2012**, 44, 338-42	36.3	186
191	Randomized, controlled trial of miglustat in Gaucher's disease type 3. *Annals of Neurology*, **2008**, 64, 514-22	9.4	177
190	Enzyme replacement therapy improves peripheral nerve and sweat function in Fabry disease. *Muscle and Nerve*, **2003**, 28, 703-10	3.4	177
189	Fabry disease. *Pharmacology & Therapeutics*, **2009**, 122, 65-77	13.9	169

188	Mutations of POLR3A encoding a catalytic subunit of RNA polymerase Pol III cause a recessive hypomyelinating leukodystrophy. *American Journal of Human Genetics*, **2011**, 89, 415-23	11	163
187	Profile of endothelial and leukocyte activation in fabry patients. *Annals of Neurology*, **2000**, 47, 229-233	9.4	159
186	Globotriaosylceramide induces oxidative stress and up-regulates cell adhesion molecule expression in Fabry disease endothelial cells. *Molecular Genetics and Metabolism*, **2008**, 95, 163-8	3.7	155
185	Ovarian failure related to eukaryotic initiation factor 2B mutations. *American Journal of Human Genetics*, **2003**, 72, 1544-50	11	150
184	Pediatric Fabry disease. *Pediatrics*, **2005**, 115, e344-55	7.4	144
183	Enzyme-replacement therapy with agalsidase alfa in children with Fabry disease. *Pediatrics*, **2006**, 118, 924-32	7.4	140
182	Elevated cerebral blood flow velocities in Fabry disease with reversal after enzyme replacement. *Stroke*, **2002**, 33, 525-31	6.7	138
181	Case definition and classification of leukodystrophies and leukoencephalopathies. *Molecular Genetics and Metabolism*, **2015**, 114, 494-500	3.7	137
180	A de novo mutation in the β-tubulin gene TUBB4A results in the leukoencephalopathy hypomyelination with atrophy of the basal ganglia and cerebellum. *American Journal of Human Genetics*, **2013**, 92, 767-73	11	133
179	Glucosylceramide and glucosylsphingosine modulate calcium mobilization from brain microsomes via different mechanisms. *Journal of Biological Chemistry*, **2003**, 278, 23594-9	5.4	132
178	The efficacy of enzyme replacement therapy in patients with chronic neuronopathic Gaucher's disease. *Journal of Pediatrics*, **2001**, 138, 539-47	3.6	132
177	Agalsidase alfa and kidney dysfunction in Fabry disease. *Journal of the American Society of Nephrology: JASN*, **2009**, 20, 1132-9	12.7	127
176	Enhanced calcium release in the acute neuronopathic form of Gaucher disease. *Neurobiology of Disease*, **2005**, 18, 83-8	7.5	125
175	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, 2629-40	4.8	122
174	Clinical spectrum of 4H leukodystrophy caused by POLR3A and POLR3B mutations. *Neurology*, **2014**, 83, 1898-905	6.5	118
173	The pharmacological chaperone isofagomine increases the activity of the Gaucher disease L444P mutant form of beta-glucosidase. *FEBS Journal*, **2010**, 277, 1618-38	5.7	117
172	Phenotypic continuum in neuronopathic Gaucher disease: an intermediate phenotype between type 2 and type 3. *Journal of Pediatrics*, **2003**, 143, 273-6	3.6	112
171	The validation of pharmacogenetics for the identification of Fabry patients to be treated with migalastat. *Genetics in Medicine*, **2017**, 19, 430-438	8.1	110

170	The cerebral vasculopathy of Fabry disease. *Journal of the Neurological Sciences*, **2007**, 257, 258-63	3.2	109
169	Recessive mutations in POLR3B, encoding the second largest subunit of Pol III, cause a rare hypomyelinating leukodystrophy. *American Journal of Human Genetics*, **2011**, 89, 652-5	11	107
168	Enhanced endothelium-dependent vasodilation in Fabry disease. *Stroke*, **2001**, 32, 1559-62	6.7	107
167	Prospective study of neurological responses to treatment with macrophage-targeted glucocerebrosidase in patients with type 3 Gaucher disease. *Annals of Neurology*, **1997**, 42, 613-21	9.4	103
166	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	4.8	103
165	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	4.4	103
164	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, 1576-83	12.7	101
163	Early alterations of brain cellular energy homeostasis in Huntington disease models. *Journal of Biological Chemistry*, **2012**, 287, 1361-70	5.4	92
162	The relationship of vascular glycolipid storage to clinical manifestations of Fabry disease: a cross-sectional study of a large cohort of clinically affected heterozygous women. *Medicine (United States)*, **2005**, 84, 261-268	1.8	91
161	Adult polyglucosan body disease: Natural History and Key Magnetic Resonance Imaging Findings. *Annals of Neurology*, **2012**, 72, 433-41	9.4	88
160	Physiological characterization of neuropathy in Fabry disease. *Muscle and Nerve*, **2002**, 26, 622-9	3.4	88
159	White matter lesions in Fabry disease occur in prior selectively hypometabolic and hyperperfused brain regions. *Brain Research Bulletin*, **2003**, 62, 231-40	3.9	87
158	Whole exome sequencing in patients with white matter abnormalities. *Annals of Neurology*, **2016**, 79, 1031-1037	9.4	86
157	Myoclonic epilepsy in Gaucher disease: genotype-phenotype insights from a rare patient subgroup. *Pediatric Research*, **2003**, 53, 387-95	3.2	85
156	Mapping of the mucolipidosis type IV gene to chromosome 19p and definition of founder haplotypes. *American Journal of Human Genetics*, **1999**, 65, 773-8	11	83
155	Cellular and tissue localization of globotriaosylceramide in Fabry disease. *Virchows Archiv Fur Pathologische Anatomie Und Physiologie Und Fur Klinische Medizin*, **2007**, 451, 823-34	5.1	81
154	Effect of genetic modifiers on cerebral lesions in Fabry disease. *Neurology*, **2005**, 64, 2148-50	6.5	79
153	Foamy cells with oligodendroglial phenotype in childhood ataxia with diffuse central nervous system hypomyelination syndrome. *Acta Neuropathologica*, **2000**, 100, 635-46	14.3	79

152	Enzyme replacement therapy and intraepidermal innervation density in Fabry disease. *Muscle and Nerve*, **2006**, 34, 53-6	3.4	77
151	Decreased guanine nucleotide exchange factor activity in eIF2B-mutated patients. *European Journal of Human Genetics*, **2004**, 12, 561-6	5.3	75
150	Enzyme-replacement therapy for metabolic storage disorders. *Lancet Neurology, The*, **2004**, 3, 752-6	24.1	75
149	Mutations in SNORD118 cause the cerebral microangiopathy leukoencephalopathy with calcifications and cysts. *Nature Genetics*, **2016**, 48, 1185-92	36.3	74
148	New prospects for the treatment of lysosomal storage diseases. *Drugs*, **2002**, 62, 733-42	12.1	74
147	The latest on leukodystrophies. *Current Opinion in Neurology*, **2004**, 17, 187-92	7.1	73
146	New syndrome characterized by hypomyelination with atrophy of the basal ganglia and cerebellum. *American Journal of Neuroradiology*, **2002**, 23, 1466-74	4.4	73
145	Update on Leukodystrophies: A Historical Perspective and Adapted Definition. *Neuropediatrics*, **2016**, 47, 349-354	1.6	72
144	Hypomyelination with atrophy of the basal ganglia and cerebellum: further delineation of the phenotype and genotype-phenotype correlation. *Brain*, **2014**, 137, 1921-30	11.2	72
143	Gaucher disease: Progress and ongoing challenges. *Molecular Genetics and Metabolism*, **2017**, 120, 8-21	3.7	72
142	A pharmacogenetic approach to identify mutant forms of -galactosidase A that respond to a pharmacological chaperone for Fabry disease. *Human Mutation*, **2011**, 32, 965-77	4.7	69
141	Heightened stress response in primary fibroblasts expressing mutant eIF2B genes from CACH/VWM leukodystrophy patients. *Human Genetics*, **2005**, 118, 99-106	6.3	69
140	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, 337-43	5.1	67
139	Leukodystrophy in patients with ovarian dysgenesis. *Annals of Neurology*, **1997**, 41, 654-61	9.4	64
138	Transfer of a mitochondrial DNA fragment to MCOLN1 causes an inherited case of mucolipidosis IV. *Human Mutation*, **2004**, 24, 460-5	4.7	61
137	Ascorbate decreases Fabry cerebral hyperperfusion suggesting a reactive oxygen species abnormality: an arterial spin tagging study. *Journal of Magnetic Resonance Imaging*, **2004**, 20, 674-83	5.6	61
136	Triheptanoin dramatically reduces paroxysmal motor disorder in patients with GLUT1 deficiency. *Journal of Neurology, Neurosurgery and Psychiatry*, **2016**, 87, 550-3	5.5	60
135	Characterization of tiger-tail banding and hair shaft abnormalities in trichothiodystrophy. *Journal of the American Academy of Dermatology*, **2005**, 52, 224-32	4.5	59

134	Childhood ataxia with CNS hypomyelination/vanishing white matter disease--a common leukodystrophy caused by abnormal control of protein synthesis. *Molecular Genetics and Metabolism*, **2006**, 88, 7-15	3.7	57
133	Mannose receptor-mediated delivery of moss-made α-galactosidase A efficiently corrects enzyme deficiency in Fabry mice. *Journal of Inherited Metabolic Disease*, **2016**, 39, 293-303	5.4	56
132	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	11	56
131	Selective arterial distribution of cerebral hyperperfusion in Fabry disease. *Journal of Neuroimaging*, **2001**, 11, 303-7	2.8	56
130	The saccadic and neurological deficits in type 3 Gaucher disease. *PLoS ONE*, **2011**, 6, e22410	3.7	52
129	GnRH-deficient phenotypes in humans and mice with heterozygous variants in KISS1/Kiss1. *Journal of Clinical Endocrinology and Metabolism*, **2011**, 96, E1771-81	5.6	51
128	Neuronopathic forms of Gaucher's disease. *Best Practice and Research: Clinical Haematology*, **1997**, 10, 711-23		49
127	Diagnosis, prognosis, and treatment of leukodystrophies. *Lancet Neurology, The*, **2019**, 18, 962-972	24.1	48
126	Noninvasive diagnosis and ophthalmic features of mucolipidosis type IV. *Ophthalmology*, **2002**, 109, 588-94	7.3	47
125	Biomarkers of Fabry disease nephropathy. *Clinical Journal of the American Society of Nephrology: CJASN*, **2010**, 5, 360-4	6.9	46
124	Four-year prospective clinical trial of agalsidase alfa in children with Fabry disease. *Journal of Pediatrics*, **2010**, 156, 832-7, 837.e1	3.6	46
123	Decreased bone density in splenectomized Gaucher patients receiving enzyme replacement therapy. *Blood Cells, Molecules, and Diseases*, **2002**, 28, 288-96	2.1	45
122	Is it Fabry disease?. *Genetics in Medicine*, **2016**, 18, 1181-1185	8.1	45
121	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, 2873-8	11.5	44
120	Fabry disease. *Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn*, **2015**, 132, 231-48	3	42
119	Genetic and clinical heterogeneity in eIF2B-related disorder. *Journal of Child Neurology*, **2008**, 23, 205-15	2.5	42
118	Screening for pharmacological chaperones in Fabry disease. *Biochemical and Biophysical Research Communications*, **2007**, 359, 168-73	3.4	42
117	Altered dopamine and serotonin metabolism in motorically asymptomatic R6/2 mice. *PLoS ONE*, **2011**, 6, e18336	3.7	41

116	A Founder Mutation in VPS11 Causes an Autosomal Recessive Leukoencephalopathy Linked to Autophagic Defects. *PLoS Genetics*, **2016**, 12, e1005848	6	41
115	Decreased asialotransferrin in cerebrospinal fluid of patients with childhood-onset ataxia and central nervous system hypomyelination/vanishing white matter disease. *Clinical Chemistry*, **2005**, 51, 2031-42	5.5	40
114	Auditory analysis of xeroderma pigmentosum 1971-2012: hearing function, sun sensitivity and DNA repair predict neurological degeneration. *Brain*, **2013**, 136, 194-208	11.2	39
113	Pegunigalsidase alfa, a novel PEGylated enzyme replacement therapy for Fabry disease, provides sustained plasma concentrations and favorable pharmacodynamics: A 1-year Phase 1/2 clinical trial. *Journal of Inherited Metabolic Disease*, **2019**, 42, 534-544	5.4	38
112	4H syndrome with late-onset growth hormone deficiency caused by POLR3A mutations. *Archives of Neurology*, **2012**, 69, 920-3		37
111	Eukaryotic initiation factor 2B (eIF2B) GEF activity as a diagnostic tool for EIF2B-related disorders. *PLoS ONE*, **2009**, 4, e8318	3.7	37
110	Identification of a biomarker in cerebrospinal fluid for neuronopathic forms of Gaucher disease. *PLoS ONE*, **2015**, 10, e0120194	3.7	36
109	Disease specific therapies in leukodystrophies and leukoencephalopathies. *Molecular Genetics and Metabolism*, **2015**, 114, 527-36	3.7	35
108	Time series proteome profiling to study endoplasmic reticulum stress response. *Journal of Proteome Research*, **2008**, 7, 2435-44	5.6	34
107	An update on the leukodsytrophies. *Current Opinion in Neurology*, **2001**, 14, 789-94	7.1	34
106	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, 1149-59	3.1	34
105	Quantitative dysmorphology assessment in Fabry disease. *Genetics in Medicine*, **2006**, 8, 96-101	8.1	33
104	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, 183-7	1.7	33
103	TUBB4A mutations result in specific neuronal and oligodendrocytic defects that closely match clinically distinct phenotypes. *Human Molecular Genetics*, **2017**, 26, 4506-4518	5.6	32
102	The glycosylation design space for recombinant lysosomal replacement enzymes produced in CHO cells. *Nature Communications*, **2019**, 10, 1785	17.4	32
101	Effect of agalsidase alfa replacement therapy on Fabry disease-related hypertrophic cardiomyopathy: a 12- to 36-month, retrospective, blinded echocardiographic pooled analysis. *Clinical Therapeutics*, **2009**, 31, 1966-76	3.5	32
100	The pharmacology of multiple regimens of agalsidase alfa enzyme replacement therapy for Fabry disease. *Genetics in Medicine*, **2007**, 9, 504-9	8.1	32
99	Enzyme replacement in Fabry disease: pharmacokinetics and pharmacodynamics of agalsidase alpha in children and adolescents. *Journal of Clinical Pharmacology*, **2007**, 47, 1222-30	2.9	32

98	Fabry's disease--an important risk factor for stroke. *Lancet, The*, **2005**, 366, 1754-6	40	32
97	Cellular and tissue distribution of intravenously administered agalsidase alfa. *Molecular Genetics and Metabolism*, **2007**, 90, 307-12	3.7	30
96	A prospective 10-year study of individualized, intensified enzyme replacement therapy in advanced Fabry disease. *Journal of Inherited Metabolic Disease*, **2015**, 38, 1129-36	5.4	29
95	XPC initiation codon mutation in xeroderma pigmentosum patients with and without neurological symptoms. *DNA Repair*, **2009**, 8, 114-25	4.3	29
94	Parapelvic kidney cysts: a distinguishing feature with high prevalence in Fabry disease. *Kidney International*, **2004**, 66, 978-82	9.9	28
93	The definition of neuronopathic Gaucher disease. *Journal of Inherited Metabolic Disease*, **2020**, 43, 1056-1059	5.4	28
92	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	4.2	27
91	Therapeutic approaches for neuronopathic lysosomal storage disorders. *Journal of Inherited Metabolic Disease*, **2010**, 33, 373-9	5.4	27
90	Lysosomal inclusions in gastric parietal cells in mucolipidosis type IV: a novel cause of achlorhydria and hypergastrinemia. *American Journal of Surgical Pathology*, **1999**, 23, 1527-31	6.7	27
89	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	2.6	26
88	Neuropathy and Fabry disease: pathogenesis and enzyme replacement therapy. *Acta Neurologica Belgica*, **2006**, 106, 61-5	1.5	26
87	Deep intronic GBE1 mutation in manifesting heterozygous patients with adult polyglucosan body disease. *JAMA Neurology*, **2015**, 72, 441-5	17.2	25
86	Fabry Disease: A Disorder of Childhood Onset. *Pediatric Neurology*, **2016**, 64, 10-20	2.9	25
85	Skin ultrastructural findings in type 2 Gaucher disease: diagnostic implications. *Molecular Genetics and Metabolism*, **2011**, 104, 631-6	3.7	25
84	Diffuse neuroaxonal involvement in mucolipidosis IV as assessed by proton magnetic resonance spectroscopic imaging. *Journal of Child Neurology*, **2003**, 18, 443-9	2.5	25
83	Leukodystrophy-associated mutations down-regulate the RNA polymerase III transcript and important regulatory RNA. *Journal of Biological Chemistry*, **2019**, 294, 7445-7459	5.4	22
82	CSF and Blood Levels of GFAP in Alexander Disease. *ENeuro*, **2015**, 2,	3.9	22
81	Establishment and characterization of Fabry disease endothelial cells with an extended lifespan. *Molecular Genetics and Metabolism*, **2007**, 92, 137-44	3.7	21

80	Improved intracellular delivery of glucocerebrosidase mediated by the HIV-1 TAT protein transduction domain. *Biochemical and Biophysical Research Communications*, **2005**, 337, 701-7	3.4	21
79	Arterial wall properties and Womersley flow in Fabry disease. *BMC Cardiovascular Disorders*, **2002**, 2, 1	2.3	21
78	Elevated endothelial microparticles in Fabry children decreased after enzyme replacement therapy. *Arteriosclerosis, Thrombosis, and Vascular Biology*, **2007**, 27, e138-9	9.4	20
77	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, 31-7	3	20
76	Risk of death in heart disease is associated with elevated urinary globotriaosylceramide. *Journal of the American Heart Association*, **2014**, 3, e000394	6	18
75	Quantitative neuroimaging in mucolipidosis type IV. *Molecular Genetics and Metabolism*, **2014**, 111, 147-51	3.7	18
74	More than hypomyelination in Pol-III disorder. *Journal of Neuropathology and Experimental Neurology*, **2013**, 72, 67-75	3.1	18
73	Blocking hyperactive androgen receptor signaling ameliorates cardiac and renal hypertrophy in Fabry mice. *Human Molecular Genetics*, **2015**, 24, 3181-91	5.6	17
72	Abnormal glycogen in astrocytes is sufficient to cause adult polyglucosan body disease. *Gene*, **2013**, 515, 376-9	3.8	17
71	Gaucher mutation N188S is associated with myoclonic epilepsy. *Human Mutation*, **2005**, 26, 271-3; author reply 274-5	4.7	17
70	Oculomotor and Vestibular Findings in Gaucher Disease Type 3 and Their Correlation with Neurological Findings. *Frontiers in Neurology*, **2017**, 8, 711	4.1	16
69	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	4.2	16
68	Characterization of Transferrin Glycopeptide Structures in Human Cerebrospinal Fluid. *International Journal of Mass Spectrometry*, **2012**, 312, 97-106	1.9	15
67	Changes in plasma and urine globotriaosylceramide levels do not predict Fabry disease progression over 1 year of agalsidase alfa. *Genetics in Medicine*, **2013**, 15, 983-9	8.1	15
66	Sex differences of urinary and kidney globotriaosylceramide and lyso-globotriaosylceramide in Fabry mice. *Journal of Lipid Research*, **2011**, 52, 1742-6	6.3	15
65	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, 8065-70	11.5	14
64	Urinary 11-Dehydro-Thromboxane B and Mortality in Patients With Stable Coronary Artery Disease. *American Journal of Cardiology*, **2017**, 119, 972-977	3	13
63	An autosomal recessive form of benign familial neonatal seizures. *Clinical Genetics*, **1991**, 40, 467-70	4	13

62	Genome sequencing in persistently unsolved white matter disorders. *Annals of Clinical and Translational Neurology*, **2020**, 7, 144-152	5.3	13
61	Long-term follow-up and sudden unexpected death in Gaucher disease type 3 in Egypt. *Neurology: Genetics*, **2016**, 2, e55	3.8	13
60	Developmental splicing deregulation in leukodystrophies related to EIF2B mutations. *PLoS ONE*, **2012**, 7, e38264	3.7	12
59	A physical and transcript map of the MCOLN1 gene region on human chromosome 19p13.3-p13.2. *Genomics*, **2001**, 73, 203-10	4.3	12
58	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, 1771-81	4.4	12
57	Myoclonus in Gaucher disease. *Advances in Neurology*, **2002**, 89, 41-8		12
56	Falsely elevated urinary Gb3 (globotriaosylceramide, CTH, GL3). *Molecular Genetics and Metabolism*, **2009**, 97, 91	3.7	10
55	Electroencephalographic findings in patients with mucolipidosis type IV. *Electroencephalography and Clinical Neurophysiology*, **1998**, 106, 400-3		10
54	Variation in cognitive function over time in Gaucher disease type 3. *Neurology*, **2019**, 93, e2272-e2283	6.5	10
53	Unique molecular signature in mucolipidosis type IV microglia. *Journal of Neuroinflammation*, **2019**, 16, 276	10.1	10
52	Fabry disease genotype, phenotype, and migalastat amenability: Insights from a national cohort. *Journal of Inherited Metabolic Disease*, **2020**, 43, 326-333	5.4	10
51	Unexpected occurrence of xeroderma pigmentosum in an uncle and nephew. *Archives of Dermatology*, **2009**, 145, 1285-91		9
50	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	5.6	9
49	A double-blind, placebo-controlled trial of triheptanoin in adult polyglucosan body disease and open-label, long-term outcome. *Journal of Inherited Metabolic Disease*, **2018**, 41, 877-883	5.4	9
48	Tetrahydrobiopterin deficiency in the pathogenesis of Fabry disease. *Human Molecular Genetics*, **2017**, 26, 1182-1192	5.6	8
47	The consequences of genetic and pharmacologic reduction in sphingolipid synthesis. *Journal of Inherited Metabolic Disease*, **2015**, 38, 77-84	5.4	8
46	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	3	8
45	Functionally pathogenic variants in vitro may not manifest a phenotype in vivo. *Neurology: Genetics*, **2017**, 3, e162	3.8	8

44	Oxidative stress reflected by increased F-isoprostanes is associated with increasing urinary 11-dehydro thromboxane B levels in patients with coronary artery disease. *Thrombosis Research*, **2016**, 148, 85-88	8.2	8
43	Randomized Clinical Trial of First-Line Genome Sequencing in Pediatric White Matter Disorders. *Annals of Neurology*, **2020**, 88, 264-273	9.4	7
42	HIV Tat Domain Improves Cross-correction of Human Galactocerebrosidase in a Gene- and Flanking Sequence-dependent Manner. *Molecular Therapy - Nucleic Acids*, **2013**, 2, e130	10.7	7
41	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		7
40	Five novel mutations in fourteen patients with Fabry Disease. *Human Mutation*, **2000**, 15, 207-8	4.7	7
39	Posterior fossa abnormalities in children with infantile spasms. *Journal of Child Neurology*, **1993**, 8, 360-5	2.5	7
38	Dystonia in RNA Polymerase III-Related Leukodystrophy. *Movement Disorders Clinical Practice*, **2019**, 6, 155-159	2.2	7
37	Migalastat for the treatment of Fabry disease. *Expert Opinion on Orphan Drugs*, **2018**, 6, 301-309	1.1	7
36	Residual thromboxane activity and oxidative stress: influence on mortality in patients with stable coronary artery disease. *Coronary Artery Disease*, **2017**, 28, 287-293	1.4	6
35	Symptoms and Quality of Life in Patients with Fabry Disease: Results from an International Patient Survey. *Advances in Therapy*, **2019**, 36, 2866-2880	4.1	6
34	Assessment of plasma lyso-Gb for clinical monitoring of treatment response in migalastat-treated patients with Fabry disease. *Genetics in Medicine*, **2021**, 23, 192-201	8.1	6
33	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	3.7	5
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