Frances M Platt

List of Publications by Year in Descending Order

205 papers	14,629 citations	62 h-index	118 g-index
222 ext. papers	16,333 ext. citations	9.2 avg, IF	6.52 L-index

#	Paper	IF	Citations
205	Correlation of age of onset and clinical severity in Niemann-Pick disease type C1 with lysosomal abnormalities and gene expression.. *Scientific Reports*, **2022**, 12, 2162	4.9	0
204	Current methods to analyse lysosome morphology, positioning, motility and function.. *Traffic*, **2022**,	5.7	3
203	Vesicle cholesterol controls exocytotic fusion pore. *Cell Calcium*, **2021**, 101, 102503	4	3
202	Glycosphingolipid metabolism and its role in ageing and Parkinson's disease. *Glycoconjugate Journal*, **2021**, 1	3	2
201	International consensus on clinical severity scale use in evaluating Niemann-Pick disease Type C in paediatric and adult patients: results from a Delphi Study. *Orphanet Journal of Rare Diseases*, **2021**, 16, 482	4.2	0
200	Lipid-mediated motor-adaptor sequestration impairs axonal lysosome delivery leading to autophagic stress and dystrophy in Niemann-Pick type C. *Developmental Cell*, **2021**, 56, 1452-1468.e8	10.2	15
199	Lipid-mediated impairment of axonal lysosome transport contributing to autophagic stress. *Autophagy*, **2021**, 17, 1796-1798	10.2	1
198	Transcriptome of HP□CD-treated Niemann-Pick disease type C1 cells highlights GPNMB as a biomarker for therapeutics. *Human Molecular Genetics*, **2021**, 30, 2456-2468	5.6	1
197	Acetyl-leucine slows disease progression in lysosomal storage disorders. *Brain Communications*, **2021**, 3, fcaa148	4.5	12
196	An iPSC model of hereditary sensory neuropathy-1 reveals L-serine-responsive deficits in neuronal ganglioside composition and axoglial interactions. *Cell Reports Medicine*, **2021**, 2, 100345	18	2
195	Acetylation turns leucine into a drug by membrane transporter switching. *Scientific Reports*, **2021**, 11, 15812	4.9	0
194	A modified density gradient proteomic-based method to analyze endolysosomal proteins in cardiac tissue. *IScience*, **2021**, 24, 102949	6.1	0
193	GM1 Gangliosidosis-A Mini-Review. *Frontiers in Genetics*, **2021**, 12, 734878	4.5	5
192	Selective estrogen receptor modulators (SERMs) affect cholesterol homeostasis through the master regulators SREBP and LXR. *Biomedicine and Pharmacotherapy*, **2021**, 141, 111871	7.5	5
191	Identification of genetic modifiers of murine hepatic □-glucocerebrosidase activity. *Biochemistry and Biophysics Reports*, **2021**, 28, 101105	2.2	0
190	Defective platelet function in Niemann-Pick disease type C1. *JIMD Reports*, **2020**, 56, 46-57	1.9	2
189	Unexpected differences in the pharmacokinetics of N-acetyl-DL-leucine enantiomers after oral dosing and their clinical relevance. *PLoS ONE*, **2020**, 15, e0229585	3.7	10

188	Brain Pathology in Mucopolysaccharidoses (MPS) Patients with Neurological Forms. *Journal of Clinical Medicine*, **2020**, 9,	5.1	20
187	Beneficial Effects of Acetyl-DL-Leucine (ADLL) in a Mouse Model of Sandhoff Disease. *Journal of Clinical Medicine*, **2020**, 9,	5.1	11
186	Genetic background modifies phenotypic severity and longevity in a mouse model of Niemann-Pick disease type C1. *DMM Disease Models and Mechanisms*, **2020**, 13,	4.1	10
185	Unbiased yeast screens identify cellular pathways affected in Niemann-Pick disease type C. *Life Science Alliance*, **2020**, 3,	5.8	2
184	Mechanistic convergence and shared therapeutic targets in Niemann-Pick disease. *Journal of Inherited Metabolic Disease*, **2020**, 43, 574-585	5.4	7
183	Systemic AAV9 gene therapy using the synapsin I promoter rescues a mouse model of neuronopathic Gaucher disease but with limited cross-correction potential to astrocytes. *Human Molecular Genetics*, **2020**, 29, 1933-1949	5.6	13
182	Sandhoff Disease: Improvement of Gait by Acetyl-DL-Leucine: A Case Report. *Neuropediatrics*, **2020**, 51, 450-452	1.6	5
181	Upregulating □-hexosaminidase activity in rodents prevents □synuclein lipid associations and protects dopaminergic neurons from □synuclein-mediated neurotoxicity. *Acta Neuropathologica Communications*, **2020**, 8, 127	7.3	6
180	Investigating the Mechanism of Cyclodextrins in the Treatment of Niemann-Pick Disease Type C Using Crosslinked 2-Hydroxypropyl-□-cyclodextrin. *Small*, **2020**, 16, e2004735	11	8
179	c-Abl Inhibition Activates TFEB and Promotes Cellular Clearance in a Lysosomal Disorder. *IScience*, **2020**, 23, 101691	6.1	10
178	Molecular basis for a new bovine model of Niemann-Pick type C disease. *PLoS ONE*, **2020**, 15, e0238697	3.7	2
177	Metabolomic Studies of Lipid Storage Disorders, with Special Reference to Niemann-Pick Type C Disease: A Critical Review with Future Perspectives. *International Journal of Molecular Sciences*, **2020**, 21,	6.3	7
176	Unexpected differences in the pharmacokinetics of N-acetyl-DL-leucine enantiomers after oral dosing and their clinical relevance **2020**, 15, e0229585		
175	Unexpected differences in the pharmacokinetics of N-acetyl-DL-leucine enantiomers after oral dosing and their clinical relevance **2020**, 15, e0229585		
174	Unexpected differences in the pharmacokinetics of N-acetyl-DL-leucine enantiomers after oral dosing and their clinical relevance **2020**, 15, e0229585		
173	Unexpected differences in the pharmacokinetics of N-acetyl-DL-leucine enantiomers after oral dosing and their clinical relevance **2020**, 15, e0229585		
172	Combined Anti-inflammatory and Neuroprotective Treatments Have the Potential to Impact Disease Phenotypes in Mice. *Frontiers in Neurology*, **2019**, 10, 963	4.1	7
171	Drug-induced increase in lysobisphosphatidic acid reduces the cholesterol overload in Niemann-Pick type C cells and mice. *EMBO Reports*, **2019**, 20, e47055	6.5	18

170	Synthesis and Study of Multifunctional Cyclodextrin-Deferasirox Hybrids. *ChemMedChem*, **2019**, 14, 1484-1492	3.7	4
169	TLR9-mediated dendritic cell activation uncovers mammalian ganglioside species with specific ceramide backbones that activate invariant natural killer T cells. *PLoS Biology*, **2019**, 17, e3000169	9.7	11
168	Age-related gait standards for healthy children and young people: the GOS-ICH paediatric gait centiles. *Archives of Disease in Childhood*, **2019**, 104, 755-760	2.2	7
167	Imaging of changes in copper trafficking and redistribution in a mouse model of Niemann-Pick C disease using positron emission tomography. *BioMetals*, **2019**, 32, 293-306	3.4	3
166	Sterile activation of invariant natural killer T cells by ER-stressed antigen-presenting cells. *Proceedings of the National Academy of Sciences of the United States of America*, **2019**, 116, 23671-23681	11.5	9
165	Reduced sphingolipid hydrolase activities, substrate accumulation and ganglioside decline in Parkinson's disease. *Molecular Neurodegeneration*, **2019**, 14, 40	19	56
164	NPC1 Deficiency in Mice is Associated with Fetal Growth Restriction, Neonatal Lethality and Abnormal Lung Pathology. *Journal of Clinical Medicine*, **2019**, 9,	5.1	6
163	Glycosphingolipid levels and glucocerebrosidase activity are altered in normal aging of the mouse brain. *Neurobiology of Aging*, **2018**, 67, 189-200	5.6	50
162	Altered Expression of Ganglioside Metabolizing Enzymes Results in GM3 Ganglioside Accumulation in Cerebellar Cells of a Mouse Model of Juvenile Neuronal Ceroid Lipofuscinosis. *International Journal of Molecular Sciences*, **2018**, 19,	6.3	11
161	Fetal gene therapy for neurodegenerative disease of infants. *Nature Medicine*, **2018**, 24, 1317-1323	50.5	76
160	Annual severity increment score as a tool for stratifying patients with Niemann-Pick disease type C and for recruitment to clinical trials. *Orphanet Journal of Rare Diseases*, **2018**, 13, 143	4.2	23
159	GM1 ganglioside-independent intoxication by Cholera toxin. *PLoS Pathogens*, **2018**, 14, e1006862	7.6	39
158	AAV9 intracerebroventricular gene therapy improves lifespan, locomotor function and pathology in a mouse model of Niemann-Pick type C1 disease. *Human Molecular Genetics*, **2018**, 27, 3079-3098	5.6	37
157	Haematopoietic Stem Cell Transplantation Arrests the Progression of Neurodegenerative Disease in Late-Onset Tay-Sachs Disease. *JIMD Reports*, **2018**, 41, 17-23	1.9	12
156	Emptying the stores: lysosomal diseases and therapeutic strategies. *Nature Reviews Drug Discovery*, **2018**, 17, 133-150	64.1	118
155	Lysosomal storage diseases. *Nature Reviews Disease Primers*, **2018**, 4, 27	51.1	304
154	A novel approach to analyze lysosomal dysfunctions through subcellular proteomics and lipidomics: the case of NPC1 deficiency. *Scientific Reports*, **2017**, 7, 41408	4.9	63
153	FTY720/fingolimod increases NPC1 and NPC2 expression and reduces cholesterol and sphingolipid accumulation in Niemann-Pick type C mutant fibroblasts. *FASEB Journal*, **2017**, 31, 1719-1730	0.9	30

152	Impaired antibacterial autophagy links granulomatous intestinal inflammation in Niemann-Pick disease type C1 and XIAP deficiency with NOD2 variants in Crohn's disease. *Gut*, **2017**, 66, 1060-1073	19.2	89
151	Neuraminidases 3 and 4 regulate neuronal function by catabolizing brain gangliosides. *FASEB Journal*, **2017**, 31, 3467-3483	0.9	35
150	The metabolism of glucocerebrosides - From 1965 to the present. *Molecular Genetics and Metabolism*, **2017**, 120, 22-26	3.7	16
149	Inhibition of β-Glucocerebrosidase Activity Preserves Motor Unit Integrity in a Mouse Model of Amyotrophic Lateral Sclerosis. *Scientific Reports*, **2017**, 7, 5235	4.9	28
148	NMR analysis reveals significant differences in the plasma metabolic profiles of Niemann Pick C1 patients, heterozygous carriers, and healthy controls. *Scientific Reports*, **2017**, 7, 6320	4.9	12
147	N-Butyl-l-deoxynojirimycin (l-NBDNJ): Synthesis of an Allosteric Enhancer of β-Glucosidase Activity for the Treatment of Pompe Disease. *Journal of Medicinal Chemistry*, **2017**, 60, 9462-9469	8.3	20
146	Case Report: Ursodeoxycholic acid treatment in Niemann-Pick disease type C; clinical experience in four cases. *Wellcome Open Research*, **2017**, 2, 75	4.8	9
145	Differential response of the liver to bile acid treatment in a mouse model of Niemann-Pick disease type C. *Wellcome Open Research*, **2017**, 2, 76	4.8	2
144	Heat shock protein-based therapy as a potential candidate for treating the sphingolipidoses. *Science Translational Medicine*, **2016**, 8, 355ra118	17.5	96
143	High incidence of unrecognized visceral/neurological late-onset Niemann-Pick disease, type C1, predicted by analysis of massively parallel sequencing data sets. *Genetics in Medicine*, **2016**, 18, 41-8	8.1	138
142	Urinary excretion and metabolism of miglustat and valproate in patients with Niemann-Pick type C1 disease: One- and two-dimensional solution-state (1)H NMR studies. *Journal of Pharmaceutical and Biomedical Analysis*, **2016**, 117, 276-88	3.5	3
141	A comparative study on fluorescent cholesterol analogs as versatile cellular reporters. *Journal of Lipid Research*, **2016**, 57, 299-309	6.3	56
140	Pathogenic mycobacteria achieve cellular persistence by inhibiting the Niemann-Pick Type C disease cellular pathway. *Wellcome Open Research*, **2016**, 1, 18	4.8	13
139	Defective Cytochrome P450-Catalysed Drug Metabolism in Niemann-Pick Type C Disease. *PLoS ONE* , **2016**, 11, e0152007	3.7	17
138	Silencing the porcine iGb3s gene does not affect Galα3Gal levels or measures of anticipated pig-to-human and pig-to-primate acute rejection. *Xenotransplantation*, **2016**, 23, 106-16	2.8	17
137	Identification of novel bile acids as biomarkers for the early diagnosis of Niemann-Pick C disease. *FEBS Letters*, **2016**, 590, 1651-62	3.8	69
136	Chemoenzymatic Synthesis of a Phosphorylated Glycoprotein. *Angewandte Chemie - International Edition*, **2016**, 55, 5058-61	16.4	37
135	Chemoenzymatic Synthesis of a Phosphorylated Glycoprotein. *Angewandte Chemie*, **2016**, 128, 5142-5145	3.6	6

134	An anecdotal report by an Oxford basic neuroscientist: effects of acetyl-DL-leucine on cognitive function and mobility in the elderly. *Journal of Neurology*, **2016**, 263, 1239-40	5.5	6
133	Circadian profiling in two mouse models of lysosomal storage disorders; Niemann Pick type-C and Sandhoff disease. *Behavioural Brain Research*, **2016**, 297, 213-23	3.4	6
132	H NMR-Linked Metabolomics Analysis of Liver from a Mouse Model of NP-C1 Disease. *Journal of Proteome Research*, **2016**, 15, 3511-3527	5.6	9
131	Immune dysfunction in Niemann-Pick disease type C. *Journal of Neurochemistry*, **2016**, 136 Suppl 1, 74-86	6	40
130	Measuring relative lysosomal volume for monitoring lysosomal storage diseases. *Methods in Cell Biology*, **2015**, 126, 331-47	1.8	4
129	Guidelines for incorporating scientific knowledge and practice on rare diseases into higher education: neuronal ceroid lipofuscinoses as a model disorder. *Biochimica Et Biophysica Acta - Molecular Basis of Disease*, **2015**, 1852, 2316-23	6.9	8
128	Amyotrophic lateral sclerosis and denervation alter sphingolipids and up-regulate glucosylceramide synthase. *Human Molecular Genetics*, **2015**, 24, 7390-405	5.6	59
127	Biomarkers for disease progression and AAV therapeutic efficacy in feline Sandhoff disease. *Experimental Neurology*, **2015**, 263, 102-12	5.7	21
126	Bridging the age spectrum of neurodegenerative storage diseases. *Best Practice and Research in Clinical Endocrinology and Metabolism*, **2015**, 29, 127-43	6.5	13
125	Expression of Ca□+-permeable two-pore channels rescues NAADP signalling in TPC-deficient cells. *EMBO Journal*, **2015**, 34, 1743-58	13	114
124	Preferential Coupling of the NAADP Pathway to Exocytosis in T-Cells. *Messenger (Los Angeles, Calif: Print)*, **2015**, 4, 53-66		8
123	A novel, highly sensitive and specific biomarker for Niemann-Pick type C1 disease. *Orphanet Journal of Rare Diseases*, **2015**, 10, 78	4.2	84
122	Intracellular sphingosine releases calcium from lysosomes. *ELife*, **2015**, 4,	8.9	90
121	Hepatic metabolic response to restricted copper intake in a Niemann-Pick C murine model. *Metallomics*, **2014**, 6, 1527-39	4.5	6
120	Disorders of cholesterol metabolism and their unanticipated convergent mechanisms of disease. *Annual Review of Genomics and Human Genetics*, **2014**, 15, 173-94	9.7	47
119	RIPK3 as a potential therapeutic target for Gaucher□ disease. *Nature Medicine*, **2014**, 20, 204-8	50.5	122
118	Sphingolipid lysosomal storage disorders. *Nature*, **2014**, 510, 68-75	50.4	211
117	Altered distribution and function of natural killer cells in murine and human Niemann-Pick disease type C1. *Blood*, **2014**, 123, 51-60	2.2	25

116	A novel mouse model of a patient mucolipidosis II mutation recapitulates disease pathology. *Journal of Biological Chemistry*, **2014**, 289, 26709-26721	5.4	14
115	Effects of miglustat treatment in a patient affected by an atypical form of Tangier disease. *Orphanet Journal of Rare Diseases*, **2014**, 9, 143	4.2	9
114	Improved neuroprotection using miglustat, curcumin and ibuprofen as a triple combination therapy in Niemann-Pick disease type C1 mice. *Neurobiology of Disease*, **2014**, 67, 9-17	7.5	53
113	Relative acidic compartment volume as a lysosomal storage disorder-associated biomarker. *Journal of Clinical Investigation*, **2014**, 124, 1320-8	15.9	51
112	1H NMR-Linked Urinary Metabolic Profiling of Niemann-Pick Class C1 (NPC1) Disease: Identification of Potential New Biomarkers using Correlated Component Regression (CCR) and Genetic Algorithm (GA) Analysis Strategies. *Current Metabolomics*, **2014**, 2, 88-121	1	11
111	Glycomimetic affinity-enrichment proteomics identifies partners for a clinically-utilized iminosugar. *Chemical Science*, **2013**, 4, 3442-3446	9.4	7
110	Vesicular and non-vesicular transport feed distinct glycosylation pathways in the Golgi. *Nature*, **2013**, 501, 116-20	50.4	117
109	-Glucosidase 2 (GBA2) activity and imino sugar pharmacology. *Journal of Biological Chemistry*, **2013**, 288, 26052-26066	5.4	58
108	Cyclodextrin alleviates neuronal storage of cholesterol in Niemann-Pick C disease without evidence of detectable blood-brain barrier permeability. *Journal of Inherited Metabolic Disease*, **2013**, 36, 491-8	5.4	62
107	Mutations in B4GALNT1 (GM2 synthase) underlie a new disorder of ganglioside biosynthesis. *Brain*, **2013**, 136, 3618-24	11.2	100
106	The yeast p5 type ATPase, spf1, regulates manganese transport into the endoplasmic reticulum. *PLoS ONE*, **2013**, 8, e85519	3.7	48
105	Early glial activation, synaptic changes and axonal pathology in the thalamocortical system of Niemann-Pick type C1 mice. *Neurobiology of Disease*, **2012**, 45, 1086-100	7.5	68
104	Globosides but not isoglobosides can impact the development of invariant NKT cells and their interaction with dendritic cells. *Journal of Immunology*, **2012**, 189, 3007-17	5.3	31
103	The cell biology of disease: lysosomal storage disorders: the cellular impact of lysosomal dysfunction. *Journal of Cell Biology*, **2012**, 199, 723-34	7.3	470
102	Invariant natural killer T cells are not affected by lysosomal storage in patients with Niemann-Pick disease type C. *European Journal of Immunology*, **2012**, 42, 1886-92	6.1	11
101	Lysosomal Ca(2+) homeostasis: role in pathogenesis of lysosomal storage diseases. *Cell Calcium*, **2011**, 50, 200-5	4	108
100	Molecular mechanisms of endolysosomal Ca2+ signalling in health and disease. *Biochemical Journal*, **2011**, 439, 349-74	3.8	278
99	A sensitive and specific LC-MS/MS method for rapid diagnosis of Niemann-Pick C1 disease from human plasma. *Journal of Lipid Research*, **2011**, 52, 1435-45	6.3	191

98	Diverse endogenous antigens for mouse NKT cells: self-antigens that are not glycosphingolipids. *Journal of Immunology*, **2011**, 186, 1348-60	5.3	49
97	Restricted ketogenic diet enhances the therapeutic action of N-butyldeoxynojirimycin towards brain GM2 accumulation in adult Sandhoff disease mice. *Journal of Neurochemistry*, **2010**, 113, 1525-35	6	19
96	Lipids on trial: the search for the offending metabolite in Niemann-Pick type C disease. *Traffic*, **2010**, 11, 419-28	5.7	144
95	Macroautophagy is not directly involved in the metabolism of amyloid precursor protein. *Journal of Biological Chemistry*, **2010**, 285, 37415-26	5.4	78
94	Common and uncommon pathogenic cascades in lysosomal storage diseases. *Journal of Biological Chemistry*, **2010**, 285, 20423-7	5.4	255
93	Endolysosomal calcium regulation and disease. *Biochemical Society Transactions*, **2010**, 38, 1458-64	5.1	48
92	Glycosphingolipid storage leads to the enhanced degradation of the B cell receptor in Sandhoff disease mice. *Journal of Inherited Metabolic Disease*, **2010**, 33, 261-70	5.4	12
91	Purified TPC isoforms form NAADP receptors with distinct roles for Ca(2+) signaling and endolysosomal trafficking. *Current Biology*, **2010**, 20, 703-9	6.3	213
90	Critical role of iron in the pathogenesis of the murine gangliosidoses. *Neurobiology of Disease*, **2009**, 34, 406-16	7.5	25
89	Beneficial effects of anti-inflammatory therapy in a mouse model of Niemann-Pick disease type C1. *Neurobiology of Disease*, **2009**, 36, 242-51	7.5	110
88	Treating lysosomal storage disorders: current practice and future prospects. *Biochimica Et Biophysica Acta - Molecular Cell Research*, **2009**, 1793, 737-45	4.9	60
87	Neural stem cell transplantation benefits a monogenic neurometabolic disorder during the symptomatic phase of disease. *Stem Cells*, **2009**, 27, 2362-70	5.8	38
86	A new surrogate marker for CNS pathology in Niemann-Pick disease type C?. *Molecular Genetics and Metabolism*, **2009**, 96, 53-4	3.7	3
85	CD1d presentation of glycolipids. *Immunology and Cell Biology*, **2008**, 86, 588-97	5	19
84	Niemann-Pick disease type C1 is a sphingosine storage disease that causes deregulation of lysosomal calcium. *Nature Medicine*, **2008**, 14, 1247-55	50.5	632
83	Substrate reduction therapy. *Acta Paediatrica, International Journal of Paediatrics*, **2008**, 97, 88-93	3.1	113
82	Glycosphingolipid depletion in PC12 cells using iminosugars protects neuronal membranes from anti-ganglioside antibody mediated injury. *Journal of Neuroimmunology*, **2008**, 203, 33-8	3.5	3
81	Autophagy induction and autophagosome clearance in neurons: relationship to autophagic pathology in Alzheimer's disease. *Journal of Neuroscience*, **2008**, 28, 6926-37	6.6	837

80	N-butyldeoxygalactonojirimycin reduces brain ganglioside and GM2 content in neonatal Sandhoff disease mice. *Neurochemistry International*, **2008**, 52, 1125-33	4.4	42
79	Beneficial effects of substrate reduction therapy in a mouse model of GM1 gangliosidosis. *Molecular Genetics and Metabolism*, **2008**, 94, 204-11	3.7	67
78	Male germ cells require polyenoic sphingolipids with complex glycosylation for completion of meiosis: a link to ceramide synthase-3. *Journal of Biological Chemistry*, **2008**, 283, 13357-69	5.4	79
77	Differential sensitivity of mouse strains to an N-alkylated imino sugar: glycosphingolipid metabolism and acrosome formation. *Pharmacogenomics*, **2008**, 9, 717-31	2.6	7
76	Invariant NKT cells reduce the immunosuppressive activity of influenza A virus-induced myeloid-derived suppressor cells in mice and humans. *Journal of Clinical Investigation*, **2008**, 118, 4036-48	15.9	258
75	The sensitivity of murine spermiogenesis to miglustat is a quantitative trait: a pharmacogenetic study. *Reproductive Biology and Endocrinology*, **2007**, 5, 1	5	28
74	Stem cells act through multiple mechanisms to benefit mice with neurodegenerative metabolic disease. *Nature Medicine*, **2007**, 13, 439-47	50.5	264
73	Glycosphingolipid synthesis requires FAPP2 transfer of glucosylceramide. *Nature*, **2007**, 449, 62-7	50.4	327
72	Normal development and function of invariant natural killer T cells in mice with isoglobotrihexosylceramide (iGb3) deficiency. *Proceedings of the National Academy of Sciences of the United States of America*, **2007**, 104, 5977-82	11.5	185
71	Accumulation of glucosylceramide in murine testis, caused by inhibition of beta-glucosidase 2: implications for spermatogenesis. *Journal of Biological Chemistry*, **2007**, 282, 32655-64	5.4	61
70	Modulation of human natural killer T cell ligands on TLR-mediated antigen-presenting cell activation. *Proceedings of the National Academy of Sciences of the United States of America*, **2007**, 104, 20490-5	11.5	160
69	Implications for invariant natural killer T cell ligands due to the restricted presence of isoglobotrihexosylceramide in mammals. *Proceedings of the National Academy of Sciences of the United States of America*, **2007**, 104, 5971-6	11.5	133
68	The postacrosomal assembly of sperm head protein, PAWP, is independent of acrosome formation and dependent on microtubular manchette transport. *Developmental Biology*, **2007**, 312, 471-83	3.1	57
67	Activation of invariant NKT cells by toll-like receptor 9-stimulated dendritic cells requires type I interferon and charged glycosphingolipids. *Immunity*, **2007**, 27, 597-609	32.3	220
66	Substrate Reduction Therapy **2007**, 153-168		4
65	The association of Shiga-like toxin with detergent-resistant membranes is modulated by glucosylceramide and is an essential requirement in the endoplasmic reticulum for a cytotoxic effect. *Molecular Biology of the Cell*, **2006**, 17, 1375-87	3.5	80
64	Long-term non-hormonal male contraception in mice using N-butyldeoxynojirimycin. *Human Reproduction*, **2006**, 21, 1309-15	5.7	24
63	Impaired selection of invariant natural killer T cells in diverse mouse models of glycosphingolipid lysosomal storage diseases. *Journal of Experimental Medicine*, **2006**, 203, 2293-303	16.6	113

62	Activation of invariant NKT cells by the helminth parasite schistosoma mansoni. *Journal of Immunology*, **2006**, 176, 2476-85	5.3	67
61	Glycolipid receptor depletion as an approach to specific antimicrobial therapy. *FEMS Microbiology Letters*, **2006**, 258, 1-8	2.9	14
60	Storage solutions: treating lysosomal disorders of the brain. *Nature Reviews Neuroscience*, **2005**, 6, 713-25	33.5	149
59	Imino sugar inhibitors for treating the lysosomal glycosphingolipidoses. *Glycobiology*, **2005**, 15, 43R-52R	5.8	183
58	Alkylated imino sugars, reversible male infertility-inducing agents, do not affect the genetic integrity of male mouse germ cells during short-term treatment despite induction of sperm deformities. *Biology of Reproduction*, **2005**, 72, 805-13	3.9	51
57	New developments in treating glycosphingolipid storage diseases. *Advances in Experimental Medicine and Biology*, **2005**, 564, 117-26	3.6	10
56	Inhibition of glucosylceramide synthase does not reverse drug resistance in cancer cells. *Journal of Biological Chemistry*, **2004**, 279, 40412-8	5.4	42
55	Accumulation of glycosphingolipids in Niemann-Pick C disease disrupts endosomal transport. *Journal of Biological Chemistry*, **2004**, 279, 26167-75	5.4	148
54	Inhibition of alpha-glucosidases I and II increases the cell surface expression of functional class A macrophage scavenger receptor (SR-A) by extending its half-life. *Journal of Biological Chemistry*, **2004**, 279, 39303-9	5.4	14
53	Infantile-onset symptomatic epilepsy syndrome caused by a homozygous loss-of-function mutation of GM3 synthase. *Nature Genetics*, **2004**, 36, 1225-9	36.3	304
52	Inhibition of glycogen breakdown by imino sugars in vitro and in vivo. *Biochemical Pharmacology*, **2004**, 67, 697-705	6	21
51	Analysis of fluorescently labeled glycosphingolipid-derived oligosaccharides following ceramide glycanase digestion and anthranilic acid labeling. *Analytical Biochemistry*, **2004**, 331, 275-82	3.1	149
50	N-butyldeoxygalactonojirimycin reduces neonatal brain ganglioside content in a mouse model of GM1 gangliosidosis. *Journal of Neurochemistry*, **2004**, 89, 645-53	6	62
49	NSAIDs increase survival in the Sandhoff disease mouse: synergy with N-butyldeoxynojirimycin. *Annals of Neurology*, **2004**, 56, 642-9	9.4	108
48	Miglustat: profile report. *Drugs and Therapy Perspectives*, **2004**, 20, 5-7	1.5	
47	Therapy of Niemann-Pick disease, type C. *Biochimica Et Biophysica Acta - Molecular and Cell Biology of Lipids*, **2004**, 1685, 77-82	5	52
46	Glycosphingolipids in endocytic membrane transport. *Seminars in Cell and Developmental Biology*, **2004**, 15, 409-16	7.5	26
45	Improved outcome of N-butyldeoxygalactonojirimycin-mediated substrate reduction therapy in a mouse model of Sandhoff disease. *Neurobiology of Disease*, **2004**, 16, 506-15	7.5	75

44	Treatment with miglustat reverses the lipid-trafficking defect in Niemann-Pick disease type C. *Neurobiology of Disease*, **2004**, 16, 654-8	7.5	138
43	Cellular effects of deoxynojirimycin analogues: inhibition of N-linked oligosaccharide processing and generation of free glucosylated oligosaccharides. *Biochemical Journal*, **2004**, 381, 867-75	3.8	38
42	Cellular effects of deoxynojirimycin analogues: uptake, retention and inhibition of glycosphingolipid biosynthesis. *Biochemical Journal*, **2004**, 381, 861-6	3.8	39
41	Lysosomal Disorders of the Brain **2004**,		24
40	Lysosomal defects and storage **2004**, 32-49		21
39	Inhibition of substrate synthesis: a pharmacological approach for glycosphingolipid storage disease therapy **2004**, 381-408		6
38	Therapeutic applications of imino sugars in lysosomal storage disorders. *Current Topics in Medicinal Chemistry*, **2003**, 3, 561-74	3	152
37	Membrane disruption and cytotoxicity of hydrophobic N-alkylated imino sugars is independent of the inhibition of protein and lipid glycosylation. *Biochemical Journal*, **2003**, 374, 307-14	3.8	49
36	Storage diseases: new insights into sphingolipid functions. *Trends in Cell Biology*, **2003**, 13, 195-203	18.3	30
35	Glycolipid depletion in antimicrobial therapy. *Molecular Microbiology*, **2003**, 47, 453-61	4.1	37
34	Miglustat. *Drugs*, **2003**, 63, 2435-2436	12.1	1
33	Small-molecule therapeutics for the treatment of glycolipid lysosomal storage disorders. *Philosophical Transactions of the Royal Society B: Biological Sciences*, **2003**, 358, 927-45	5.8	55
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