Frances M Platt

List of Publications by Citations

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	bcl-2-immunoglobulin transgenic mice demonstrate extended B cell survival and follicular lymphoproliferation. *Cell*, **1989**, 57, 79-88	56.2	1062
204	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
203	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
202	Novel oral treatment of Gaucher's disease with N-butyldeoxynojirimycin (OGT 918) to decrease substrate biosynthesis. *Lancet, The*, **2000**, 355, 1481-5	40	628
201	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
200	Targeting glycosylation as a therapeutic approach. *Nature Reviews Drug Discovery*, **2002**, 1, 65-75	64.1	348
199	Glycosphingolipid synthesis requires FAPP2 transfer of glucosylceramide. *Nature*, **2007**, 449, 62-7	50.4	327
198	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
197	Lysosomal storage diseases. *Nature Reviews Disease Primers*, **2018**, 4, 27	51.1	304
196	Molecular mechanisms of endolysosomal Ca2+ signalling in health and disease. *Biochemical Journal*, **2011**, 439, 349-74	3.8	278
195	Inhibition of glycosphingolipid biosynthesis: application to lysosomal storage disorders. *Chemical Reviews*, **2000**, 100, 4683-96	68.1	270
194	Stem cells act through multiple mechanisms to benefit mice with neurodegenerative metabolic disease. *Nature Medicine*, **2007**, 13, 439-47	50.5	264
193	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
192	Common and uncommon pathogenic cascades in lysosomal storage diseases. *Journal of Biological Chemistry*, **2010**, 285, 20423-7	5.4	255
191	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
190	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
189	Sphingolipid lysosomal storage disorders. *Nature*, **2014**, 510, 68-75	50.4	211

188	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
187	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
186	Imino sugar inhibitors for treating the lysosomal glycosphingolipidoses. *Glycobiology*, **2005**, 15, 43R-52R	5.8	183
185	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
184	Therapeutic applications of imino sugars in lysosomal storage disorders. *Current Topics in Medicinal Chemistry*, **2003**, 3, 561-74	3	152
183	Analysis of fluorescently labeled glycosphingolipid-derived oligosaccharides following ceramide glycanase digestion and anthranilic acid labeling. *Analytical Biochemistry*, **2004**, 331, 275-82	3.1	149
182	Storage solutions: treating lysosomal disorders of the brain. *Nature Reviews Neuroscience*, **2005**, 6, 713-25	33.5	149
181	Accumulation of glycosphingolipids in Niemann-Pick C disease disrupts endosomal transport. *Journal of Biological Chemistry*, **2004**, 279, 26167-75	5.4	148
180	Lipids on trial: the search for the offending metabolite in Niemann-Pick type C disease. *Traffic*, **2010**, 11, 419-28	5.7	144
179	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
178	Treatment with miglustat reverses the lipid-trafficking defect in Niemann-Pick disease type C. *Neurobiology of Disease*, **2004**, 16, 654-8	7.5	138
177	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
176	N-butyldeoxygalactonojirimycin: a more selective inhibitor of glycosphingolipid biosynthesis than N-butyldeoxynojirimycin, in vitro and in vivo. *Biochemical Pharmacology*, **2000**, 59, 821-9	6	125
175	RIPK3 as a potential therapeutic target for Gaucher□ disease. *Nature Medicine*, **2014**, 20, 204-8	50.5	122
174	Emptying the stores: lysosomal diseases and therapeutic strategies. *Nature Reviews Drug Discovery*, **2018**, 17, 133-150	64.1	118
173	Vesicular and non-vesicular transport feed distinct glycosylation pathways in the Golgi. *Nature*, **2013**, 501, 116-20	50.4	117
172	Inhibition of calcium uptake via the sarco/endoplasmic reticulum Ca2+-ATPase in a mouse model of Sandhoff disease and prevention by treatment with N-butyldeoxynojirimycin. *Journal of Biological Chemistry*, **2003**, 278, 29496-501	5.4	115
171	Expression of Ca□+-permeable two-pore channels rescues NAADP signalling in TPC-deficient cells. *EMBO Journal*, **2015**, 34, 1743-58	13	114

170	Substrate reduction therapy. *Acta Paediatrica, International Journal of Paediatrics*, **2008**, 97, 88-93	3.1	113
169	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
168	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
167	Enhanced survival in Sandhoff disease mice receiving a combination of substrate deprivation therapy and bone marrow transplantation. *Blood*, **2001**, 97, 327-9	2.2	110
166	Lysosomal Ca(2+) homeostasis: role in pathogenesis of lysosomal storage diseases. *Cell Calcium*, **2011**, 50, 200-5	4	108
165	NSAIDs increase survival in the Sandhoff disease mouse: synergy with N-butyldeoxynojirimycin. *Annals of Neurology*, **2004**, 56, 642-9	9.4	108
164	Extensive glycosphingolipid depletion in the liver and lymphoid organs of mice treated with N-butyldeoxynojirimycin. *Journal of Biological Chemistry*, **1997**, 272, 19365-72	5.4	103
163	Mutations in B4GALNT1 (GM2 synthase) underlie a new disorder of ganglioside biosynthesis. *Brain*, **2013**, 136, 3618-24	11.2	100
162	Glucosylceramide modulates membrane traffic along the endocytic pathway. *Journal of Lipid Research*, **2002**, 43, 1837-45	6.3	98
161	Heat shock protein-based therapy as a potential candidate for treating the sphingolipidoses. *Science Translational Medicine*, **2016**, 8, 355ra118	17.5	96
160	Intracellular sphingosine releases calcium from lysosomes. *ELife*, **2015**, 4,	8.9	90
159	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
158	Reversible infertility in male mice after oral administration of alkylated imino sugars: a nonhormonal approach to male contraception. *Proceedings of the National Academy of Sciences of the United States of America*, **2002**, 99, 17173-8	11.5	89
157	A novel, highly sensitive and specific biomarker for Niemann-Pick type C1 disease. *Orphanet Journal of Rare Diseases*, **2015**, 10, 78	4.2	84
156	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
155	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
154	Macroautophagy is not directly involved in the metabolism of amyloid precursor protein. *Journal of Biological Chemistry*, **2010**, 285, 37415-26	5.4	78
153	Fetal gene therapy for neurodegenerative disease of infants. *Nature Medicine*, **2018**, 24, 1317-1323	50.5	76

152	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
151	Preparation, biochemical characterization and biological properties of radiolabelled N-alkylated deoxynojirimycins. *Biochemical Journal*, **2002**, 366, 225-33	3.8	71
150	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
149	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
148	Beneficial effects of substrate reduction therapy in a mouse model of GM1 gangliosidosis. *Molecular Genetics and Metabolism*, **2008**, 94, 204-11	3.7	67
147	Activation of invariant NKT cells by the helminth parasite schistosoma mansoni. *Journal of Immunology*, **2006**, 176, 2476-85	5.3	67
146	Inhibition of N-glycan processing in B16 melanoma cells results in inactivation of tyrosinase but does not prevent its transport to the melanosome. *Journal of Biological Chemistry*, **1997**, 272, 15796-803	5.4	66
145	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
144	Substrate reduction therapy for glycosphingolipid storage disorders. *Expert Opinion on Investigational Drugs*, **2001**, 10, 455-66	5.9	63
143	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
142	N-butyldeoxygalactonojirimycin reduces neonatal brain ganglioside content in a mouse model of GM1 gangliosidosis. *Journal of Neurochemistry*, **2004**, 89, 645-53	6	62
141	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
140	Treating lysosomal storage disorders: current practice and future prospects. *Biochimica Et Biophysica Acta - Molecular Cell Research*, **2009**, 1793, 737-45	4.9	60
139	Amyotrophic lateral sclerosis and denervation alter sphingolipids and up-regulate glucosylceramide synthase. *Human Molecular Genetics*, **2015**, 24, 7390-405	5.6	59
138	-Glucosidase 2 (GBA2) activity and imino sugar pharmacology. *Journal of Biological Chemistry*, **2013**, 288, 26052-26066	5.4	58
137	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
136	A comparative study on fluorescent cholesterol analogs as versatile cellular reporters. *Journal of Lipid Research*, **2016**, 57, 299-309	6.3	56
135	Reduced sphingolipid hydrolase activities, substrate accumulation and ganglioside decline in Parkinson's disease. *Molecular Neurodegeneration*, **2019**, 14, 40	19	56

134	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
133	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
132	Therapy of Niemann-Pick disease, type C. *Biochimica Et Biophysica Acta - Molecular and Cell Biology of Lipids*, **2004**, 1685, 77-82	5	52
131	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
130	Relative acidic compartment volume as a lysosomal storage disorder-associated biomarker. *Journal of Clinical Investigation*, **2014**, 124, 1320-8	15.9	51
129	Increased glycosphingolipid levels in serum and aortae of apolipoprotein E gene knockout mice. *Journal of Lipid Research*, **2002**, 43, 205-214	6.3	51
128	Glycosphingolipid levels and glucocerebrosidase activity are altered in normal aging of the mouse brain. *Neurobiology of Aging*, **2018**, 67, 189-200	5.6	50
127	Diverse endogenous antigens for mouse NKT cells: self-antigens that are not glycosphingolipids. *Journal of Immunology*, **2011**, 186, 1348-60	5.3	49
126	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
125	The yeast p5 type ATPase, spf1, regulates manganese transport into the endoplasmic reticulum. *PLoS ONE*, **2013**, 8, e85519	3.7	48
124	Endolysosomal calcium regulation and disease. *Biochemical Society Transactions*, **2010**, 38, 1458-64	5.1	48
123	Increased glycosphingolipid levels in serum and aortae of apolipoprotein E gene knockout mice. *Journal of Lipid Research*, **2002**, 43, 205-14	6.3	48
122	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
121	Substrate reduction therapy in mouse models of the glycosphingolipidoses. *Philosophical Transactions of the Royal Society B: Biological Sciences*, **2003**, 358, 947-54	5.8	45
120	N-butyldeoxygalactonojirimycin reduces brain ganglioside and GM2 content in neonatal Sandhoff disease mice. *Neurochemistry International*, **2008**, 52, 1125-33	4.4	42
119	Inhibition of glucosylceramide synthase does not reverse drug resistance in cancer cells. *Journal of Biological Chemistry*, **2004**, 279, 40412-8	5.4	42
118	Immune dysfunction in Niemann-Pick disease type C. *Journal of Neurochemistry*, **2016**, 136 Suppl 1, 74-86	6	40
117	GM1 ganglioside-independent intoxication by Cholera toxin. *PLoS Pathogens*, **2018**, 14, e1006862	7.6	39

116	Cellular effects of deoxynojirimycin analogues: uptake, retention and inhibition of glycosphingolipid biosynthesis. *Biochemical Journal*, **2004**, 381, 861-6	3.8	39
115	Neural stem cell transplantation benefits a monogenic neurometabolic disorder during the symptomatic phase of disease. *Stem Cells*, **2009**, 27, 2362-70	5.8	38
114	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
113	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
112	Glycolipid depletion in antimicrobial therapy. *Molecular Microbiology*, **2003**, 47, 453-61	4.1	37
111	Chemoenzymatic Synthesis of a Phosphorylated Glycoprotein. *Angewandte Chemie - International Edition*, **2016**, 55, 5058-61	16.4	37
110	Neuraminidases 3 and 4 regulate neuronal function by catabolizing brain gangliosides. *FASEB Journal*, **2017**, 31, 3467-3483	0.9	35
109	Inhibitors of Glycosphingolipid Biosynthesis.. *Trends in Glycoscience and Glycotechnology*, **1995**, 7, 495-510	0.1	32
108	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
107	Analysis and isolation of human transferrin receptor using the OKT-9 monoclonal antibody covalently crosslinked to magnetic beads. *Analytical Biochemistry*, **1991**, 199, 219-22	3.1	31
106	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
105	Storage diseases: new insights into sphingolipid functions. *Trends in Cell Biology*, **2003**, 13, 195-203	18.3	30
104	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
103	The sensitivity of murine spermiogenesis to miglustat is a quantitative trait: a pharmacogenetic study. *Reproductive Biology and Endocrinology*, **2007**, 5, 1	5	28
102	An inducible mouse model of late onset Tay-Sachs disease. *Neurobiology of Disease*, **2002**, 10, 201-10	7.5	28
101	Modulation of THP-1 macrophage and cholesterol-loaded foam cell apolipoprotein E levels by glycosphingolipids. *Biochemical and Biophysical Research Communications*, **2002**, 290, 1361-7	3.4	27
100	Glycosphingolipids in endocytic membrane transport. *Seminars in Cell and Developmental Biology*, **2004**, 15, 409-16	7.5	26
99	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

98	Critical role of iron in the pathogenesis of the murine gangliosidoses. *Neurobiology of Disease*, **2009**, 34, 406-16	7.5	25
97	Inhibition of glycosphingolipid biosynthesis does not impair growth or morphogenesis of the postimplantation mouse embryo. *Journal of Neurochemistry*, **1998**, 70, 871-82	6	25
96	Long-term non-hormonal male contraception in mice using N-butyldeoxynojirimycin. *Human Reproduction*, **2006**, 21, 1309-15	5.7	24
95	Lysosomal Disorders of the Brain **2004**,		24
94	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
93	Biomarkers for disease progression and AAV therapeutic efficacy in feline Sandhoff disease. *Experimental Neurology*, **2015**, 263, 102-12	5.7	21
92	Inhibition of glycogen breakdown by imino sugars in vitro and in vivo. *Biochemical Pharmacology*, **2004**, 67, 697-705	6	21
91	Pathogenic mycobacteria achieve cellular persistence by inhibiting the Niemann-Pick Type C disease cellular pathway. *Wellcome Open Research*,1, 18	4.8	21
90	Lysosomal defects and storage **2004**, 32-49		21
89	Brain Pathology in Mucopolysaccharidoses (MPS) Patients with Neurological Forms. *Journal of Clinical Medicine*, **2020**, 9,	5.1	20
88	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
87	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
86	CD1d presentation of glycolipids. *Immunology and Cell Biology*, **2008**, 86, 588-97	5	19
85	Modulation of cell-surface transferrin receptor by the imino sugar N-butyldeoxynojirimycin. *FEBS Journal*, **1992**, 208, 187-93		19
84	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
83	Defective Cytochrome P450-Catalysed Drug Metabolism in Niemann-Pick Type C Disease. *PLoS ONE*, **2016**, 11, e0152007	3.7	17
82	Silencing the porcine iGb3s gene does not affect Galα1,3Gal levels or measures of anticipated pig-to-human and pig-to-primate acute rejection. *Xenotransplantation*, **2016**, 23, 106-16	2.8	17
81	The metabolism of glucocerebrosides - From 1965 to the present. *Molecular Genetics and Metabolism*, **2017**, 120, 22-26	3.7	16

80	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
79	A novel mouse model of a patient mucolipidosis II mutation recapitulates disease pathology. *Journal of Biological Chemistry*, **2014**, 289, 26709-26721	5.4	14
78	Glycolipid receptor depletion as an approach to specific antimicrobial therapy. *FEMS Microbiology Letters*, **2006**, 258, 1-8	2.9	14
77	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
76	Bridging the age spectrum of neurodegenerative storage diseases. *Best Practice and Research in Clinical Endocrinology and Metabolism*, **2015**, 29, 127-43	6.5	13
75	Pathogenic mycobacteria achieve cellular persistence by inhibiting the Niemann-Pick Type C disease cellular pathway. *Wellcome Open Research*, **2016**, 1, 18	4.8	13
74	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
73	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
72	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
71	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
70	Acetyl-leucine slows disease progression in lysosomal storage disorders. *Brain Communications*, **2021**, 3, fcaa148	4.5	12
69	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
68	Beneficial Effects of Acetyl-DL-Leucine (ADLL) in a Mouse Model of Sandhoff Disease. *Journal of Clinical Medicine*, **2020**, 9,	5.1	11
67	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
66	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
65	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
64	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
63	New developments in treating glycosphingolipid storage diseases. *Advances in Experimental Medicine and Biology*, **2005**, 564, 117-26	3.6	10

62	Carbohydrate receptor depletion as an antimicrobial strategy for prevention of urinary tract infection. *Journal of Infectious Diseases*, **2001**, 183 Suppl 1, S70-3	7	10
61	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
60	c-Abl Inhibition Activates TFEB and Promotes Cellular Clearance in a Lysosomal Disorder. *IScience*, **2020**, 23, 101691	6.1	10
59	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
58	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
57	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
56	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
55	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
54	Preferential Coupling of the NAADP Pathway to Exocytosis in T-Cells. *Messenger (Los Angeles, Calif: Print)*, **2015**, 4, 53-66		8
53	Substrate deprivation: a new therapeutic approach for the glycosphingolipid lysosomal storage diseases. *Expert Reviews in Molecular Medicine*, **2000**, 2, 1-17	6.7	8
52	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
51	Combined Anti-inflammatory and Neuroprotective Treatments Have the Potential to Impact Disease Phenotypes in Mice. *Frontiers in Neurology*, **2019**, 10, 963	4.1	7
50	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
49	Glycomimetic affinity-enrichment proteomics identifies partners for a clinically-utilized iminosugar. *Chemical Science*, **2013**, 4, 3442-3446	9.4	7
48	Differential sensitivity of mouse strains to an N-alkylated imino sugar: glycosphingolipid metabolism and acrosome formation. *Pharmacogenomics*, **2008**, 9, 717-31	2.6	7
47	Medicinal use of Iminosugars295-326		7
46	Mechanistic convergence and shared therapeutic targets in Niemann-Pick disease. *Journal of Inherited Metabolic Disease*, **2020**, 43, 574-585	5.4	7
45	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

44	Hepatic metabolic response to restricted copper intake in a Niemann-Pick C murine model. *Metallomics*, **2014**, 6, 1527-39	4.5	6
43	Monoclonal antibodies specific for novel murine cell surface markers define subpopulations of germinal center cells. *Cellular Immunology*, **1992**, 143, 449-66	4.4	6
42	Inhibition of substrate synthesis: a pharmacological approach for glycosphingolipid storage disease therapy **2004**, 381-408		6
41	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
40	Upregulating β-hexosaminidase activity in rodents prevents α-synuclein lipid associations and protects dopaminergic neurons from α-synuclein-mediated neurotoxicity. *Acta Neuropathologica Communications*, **2020**, 8, 127	7.3	6
39	Chemoenzymatic Synthesis of a Phosphorylated Glycoprotein. *Angewandte Chemie*, **2016**, 128, 5142-5145	3.6	6
38	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
37	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
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