Frances M Platt

List of Publications by Year
in descending order

205 papers	17,781 citations	13099 68 h-index	14759 127 g-index
222 all docs	222 docs citations	222 times ranked	18101 citing authors

#	Article	IF	Citations
1	bcl-2-Immunoglobulin transgenic mice demonstrate extended B cell survival and follicular lymphoproliferation. Cell, 1989, 57, 79-88.	28.9	1,181
2	Autophagy Induction and Autophagosome Clearance in Neurons: Relationship to Autophagic Pathology in Alzheimer's Disease. Journal of Neuroscience, 2008, 28, 6926-6937.	3.6	979
3	Niemann-Pick disease type C1 is a sphingosine storage disease that causes deregulation of lysosomal calcium. Nature Medicine, 2008, 14, 1247-1255.	30.7	730
4	Novel oral treatment of Gaucher's disease with N-butyldeoxynojirimycin (OGT 918) to decrease substrate biosynthesis. Lancet, The, 2000, 355, 1481-1485.	13.7	700
5	Lysosomal storage disorders: The cellular impact of lysosomal dysfunction. Journal of Cell Biology, 2012, 199, 723-734.	5.2	579
6	Lysosomal storage diseases. Nature Reviews Disease Primers, 2018, 4, 27.	30.5	563
7	Targeting glycosylation as a therapeutic approach. Nature Reviews Drug Discovery, 2002, 1, 65-75.	46.4	409
8	Infantile-onset symptomatic epilepsy syndrome caused by a homozygous loss-of-function mutation of GM3 synthase. Nature Genetics, 2004, 36, 1225-1229.	21.4	359
9	Glycosphingolipid synthesis requires FAPP2 transfer of glucosylceramide. Nature, 2007, 449, 62-67.	27.8	359
10	Molecular mechanisms of endolysosomal Ca2+ signalling in health and disease. Biochemical Journal, 2011, 439, 349-378.	3.7	329
11	Invariant NKT cells reduce the immunosuppressive activity of influenza A virusâ€“induced myeloid-derived suppressor cells in mice and humans. Journal of Clinical Investigation, 2008, 118, 4036-4048.	8.2	299
12	Common and Uncommon Pathogenic Cascades in Lysosomal Storage Diseases. Journal of Biological Chemistry, 2010, 285, 20423-20427.	3.4	298
13	Stem cells act through multiple mechanisms to benefit mice with neurodegenerative metabolic disease. Nature Medicine, 2007, 13, 439-447.	30.7	293
14	Inhibition of Glycosphingolipid Biosynthesis:Â Application to Lysosomal Storage Disorders. Chemical Reviews, 2000, 100, 4683-4696.	47.7	290
15	Sphingolipid lysosomal storage disorders. Nature, 2014, 510, 68-75.	27.8	270
16	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.	14.3	243
17	Purified TPC Isoforms Form NAADP Receptors with Distinct Roles for Ca2+ Signaling and Endolysosomal Trafficking. Current Biology, 2010, 20, 703-709.	3.9	234
18	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, 1435-1445.	4.2	230

#	Article	IF	Citations
19	Imino sugar inhibitors for treating the lysosomal glycosphingolipidoses. Glycobiology, 2005, 15, 43R-52R.	2.5	204
20	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, 5977-5982.	7.1	198
21	Emptying the stores: lysosomal diseases and therapeutic strategies. Nature Reviews Drug Discovery, 2018, 17, 133-150.	46.4	191
22	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, 20490-20495.	7.1	173
23	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, 41-48.	2.4	171
24	Lipids on Trial: The Search for the Offending Metabolite in Niemann-Pick type C Disease. Traffic, 2010, 11, 419-428.	2.7	170
25	Accumulation of Glycosphingolipids in Niemann-Pick C Disease Disrupts Endosomal Transport. Journal of Biological Chemistry, 2004, 279, 26167-26175.	3.4	167
26	Analysis of fluorescently labeled glycosphingolipid-derived oligosaccharides following ceramide glycanase digestion and anthranilic acid labeling. Analytical Biochemistry, 2004, 331, 275-282.	2.4	162
27	Storage solutions: treating lysosomal disorders of the brain. Nature Reviews Neuroscience, 2005, 6, 713-725.	10.2	159
28	Therapeutic Applications of Imino Sugars in Lysosomal Storage Disorders. Current Topics in Medicinal Chemistry, 2003, 3, 561-574.	2.1	156
29	Treatment with miglustat reverses the lipid-trafficking defect in Niemannâ€“Pick disease type C. Neurobiology of Disease, 2004, 16, 654-658.	4.4	147
30	RIPK3 as a potential therapeutic target for Gaucher's disease. Nature Medicine, 2014, 20, 204-208.	30.7	147
31	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, 5971-5976.	7.1	145
32	Expression of Ca ²⁺ â€permeable twoâ€pore channels rescues <scp>NAADP</scp> signalling in <scp>TPC</scp> â€deficient cells. EMBO Journal, 2015, 34, 1743-1758.	7.8	144
33	Substrate reduction therapy. Acta Paediatrica, International Journal of Paediatrics, 2008, 97, 88-93.	1.5	139
34	N-butyldeoxygalactonojirimycin: a more selective inhibitor of glycosphingolipid biosynthesis than N-butyldeoxynojirimycin, in vitro and in vivo. Biochemical Pharmacology, 2000, 59, 821-829.	4.4	137
35	Heat shock proteinâ€“based therapy as a potential candidate for treating the sphingolipidoses. Science Translational Medicine, 2016, 8, 355ra118.	12.4	137
36	Vesicular and non-vesicular transport feed distinct glycosylation pathways in the Golgi. Nature, 2013, 501, 116-120.	27.8	136

#	Article	IF	Citations
37	Beneficial effects of anti-inflammatory therapy in a mouse model of Niemann-Pick disease type C1. Neurobiology of Disease, 2009, 36, 242-251.	4.4	132
38	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, 29496-29501.	3.4	129
39	Impaired selection of invariant natural killer T cells in diverse mouse models of glycosphingolipid lysosomal storage diseases. Journal of Experimental Medicine, 2006, 203, 2293-2303.	8.5	127
40	Impaired antibacterial autophagy links granulomatous intestinal inflammation in Niemannâ€“Pick disease type C1 and XIAP deficiency with NOD2 variants in Crohn's disease. Gut, 2017, 66, 1060-1073.	12.1	126
41	Lysosomal Ca2+ homeostasis: Role in pathogenesis of lysosomal storage diseases. Cell Calcium, 2011, 50, 200-205.	2.4	122
42	Glucosylceramide modulates membrane traffic along the endocytic pathway. Journal of Lipid Research, 2002, 43, 1837-1845.	4.2	120
43	Enhanced survival in Sandhoff disease mice receiving a combination of substrate deprivation therapy and bone marrow transplantation. Blood, 2001, 97, 327-329.	1.4	118
44	Fetal gene therapy for neurodegenerative disease of infants. Nature Medicine, 2018, 24, 1317-1323.	30.7	117
45	NSAIDs increase survival in the Sandhoff disease mouse: Synergy withN-butyldeoxynojirimycin. Annals of Neurology, 2004, 56, 642-649.	5.3	116
46	Extensive Glycosphingolipid Depletion in the Liver and Lymphoid Organs of Mice Treated with N-Butyldeoxynojirimycin. Journal of Biological Chemistry, 1997, 272, 19365-19372.	3.4	115
47	Mutations in B4GALNT1 (GM2 synthase) underlie a new disorder of ganglioside biosynthesis. Brain, 2013, 136, 3618-3624.	7.6	115
48	Intracellular sphingosine releases calcium from lysosomes. ELife, 2015, 4, .	6.0	115
49	A novel, highly sensitive and specific biomarker for Niemann-Pick type C1 disease. Orphanet Journal of Rare Diseases, 2015, 10, 78.	2.7	105
50	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, 17173-17178.	7.1	104
51	Male Germ Cells Require Polyenoic Sphingolipids with Complex Glycosylation for Completion of Meiosis. Journal of Biological Chemistry, 2008, 283, 13357-13369.	3.4	100
52	Reduced sphingolipid hydrolase activities, substrate accumulation and ganglioside decline in Parkinsonâ€™s disease. Molecular Neurodegeneration, 2019, 14, 40.	10.8	100
53	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, 1375-1387.	2.1	93
54	A novel approach to analyze lysosomal dysfunctions through subcellular proteomics and lipidomics: the case of NPC1 deficiency. Scientific Reports, 2017, 7, 41408.	3.3	93

#	Article	IF	Citations
55	Early glial activation, synaptic changes and axonal pathology in the thalamocortical system of Niemannâ€“Pick type C1 mice. Neurobiology of Disease, 2012, 45, 1086-1100.	4.4	84
56	Amyotrophic lateral sclerosis and denervation alter sphingolipids and up-regulate glucosylceramide synthase. Human Molecular Genetics, 2015, 24, 7390-7405.	2.9	84
57	Identification of novel bile acids as biomarkers for the early diagnosis of Niemannâ€Pick C disease. FEBS Letters, 2016, 590, 1651-1662.	2.8	82
58	Macroautophagy Is Not Directly Involved in the Metabolism of Amyloid Precursor Protein. Journal of Biological Chemistry, 2010, 285, 37415-37426.	3.4	81
59	Improved outcome of N-butyldeoxygalactonojirimycin-mediated substrate reduction therapy in a mouse model of Sandhoff disease. Neurobiology of Disease, 2004, 16, 506-515.	4.4	79
60	Activation of Invariant NKT Cells by the Helminth Parasite Schistosoma mansoni. Journal of Immunology, 2006, 176, 2476-2485.	0.8	78
61	A comparative study on fluorescent cholesterol analogs as versatile cellular reporters. Journal of Lipid Research, 2016, 57, 299-309.	4.2	78
62	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, 15796-15803.	3.4	76
63	Treating lysosomal storage disorders: Current practice and future prospects. Biochimica Et Biophysica Acta - Molecular Cell Research, 2009, 1793, 737-745.	4.1	76
64	Preparation, biochemical characterization and biological properties of radiolabelled N-alkylated deoxynojirimycins. Biochemical Journal, 2002, 366, 225-233.	3.7	75
65	Beneficial effects of substrate reduction therapy in a mouse model of GM1 gangliosidosis. Molecular Genetics and Metabolism, 2008, 94, 204-211.	1.1	75
66	Cyclodextrin alleviates neuronal storage of cholesterol in Niemannâ€Pick C disease without evidence of detectable bloodâ€“brain barrier permeability. Journal of Inherited Metabolic Disease, 2013, 36, 491-498.	3.6	74
67	Improved neuroprotection using miglustat, curcumin and ibuprofen as a triple combination therapy in Niemannâ€“Pick disease type C1 mice. Neurobiology of Disease, 2014, 67, 9-17.	4.4	74
68	Disorders of Cholesterol Metabolism and Their Unanticipated Convergent Mechanisms of Disease. Annual Review of Genomics and Human Genetics, 2014, 15, 173-194.	6.2	73
69	Substrate reduction therapy for glycosphingolipid storage disorders. Expert Opinion on Investigational Drugs, 2001, 10, 455-466.	4.1	72
70	Î²-Glucosidase 2 (GBA2) Activity and Imino Sugar Pharmacology. Journal of Biological Chemistry, 2013, 288, 26052-26066.	3.4	69
71	N-butyldeoxygalactonojirimycin reduces neonatal brain ganglioside content in a mouse model of GM1 gangliosidosis. Journal of Neurochemistry, 2004, 89, 645-653.	3.9	68
72	Glycosphingolipid levels and glucocerebrosidase activity are altered in normal aging of the mouse brain. Neurobiology of Aging, 2018, 67, 189-200.	3.1	66

#	Article	IF	Citations
73	The postacrosomal assembly of sperm head protein, PAWP, is independent of acrosome formation and dependent on microtubular manchette transport. Developmental Biology, 2007, 312, 471-483.	2.0	64
74	Accumulation of Glucosylceramide in Murine Testis, Caused by Inhibition of Î²-Glucosidase 2. Journal of Biological Chemistry, 2007, 282, 32655-32664.	3.4	63
75	Relative acidic compartment volume as a lysosomal storage disorderâ€“associated biomarker. Journal of Clinical Investigation, 2014, 124, 1320-1328.	8.2	63
76	The Yeast P5 Type ATPase, Spf1, Regulates Manganese Transport into the Endoplasmic Reticulum. PLoS ONE, 2013, 8, e85519.	2.5	62
77	Therapy of Niemannâ€“Pick disease, type C. Biochimica Et Biophysica Acta - Molecular and Cell Biology of Lipids, 2004, 1685, 77-82.	2.4	60
78	Smallâ€“molecule therapeutics for the treatment of glycolipid lysosomal storage disorders. Philosophical Transactions of the Royal Society B: Biological Sciences, 2003, 358, 927-945.	4.0	58
79	GM1 ganglioside-independent intoxication by Cholera toxin. PLoS Pathogens, 2018, 14, e1006862.	4.7	57
80	Endolysosomal calcium regulation and disease. Biochemical Society Transactions, 2010, 38, 1458-1464.	3.4	56
81	Increased glycosphingolipid levels in serum and aortae of apolipoprotein E gene knockout mice. Journal of Lipid Research, 2002, 43, 205-214.	4.2	56
82	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 Deformities1. Biology of Reproduction, 2005, 72, 805-813.	2.7	55
83	Immune dysfunction in Niemannâ€Pick disease type C. Journal of Neurochemistry, 2016, 136, 74-80.	3.9	55
84	Membrane disruption and cytotoxicity of hydrophobic N-alkylated imino sugars is independent of the inhibition of protein and lipid glycosylation. Biochemical Journal, 2003, 374, 307-314.	3.7	54
85	Diverse Endogenous Antigens for Mouse NKT Cells: Self-Antigens That Are Not Glycosphingolipids. Journal of Immunology, 2011, 186, 1348-1360.	0.8	54
86	Inhibition of Î²-Glucocerebrosidase Activity Preserves Motor Unit Integrity in a Mouse Model of Amyotrophic Lateral Sclerosis. Scientific Reports, 2017, 7, 5235.	3.3	53
87	AAV9 intracerebroventricular gene therapy improves lifespan, locomotor function and pathology in a mouse model of Niemannâ€“Pick type C1 disease. Human Molecular Genetics, 2018, 27, 3079-3098.	2.9	51
88	Substrate reduction therapy in mouse models of the glycosphingolipidoses. Philosophical Transactions of the Royal Society B: Biological Sciences, 2003, 358, 947-954.	4.0	50
89	Increased glycosphingolipid levels in serum and aortae of apolipoprotein E gene knockout mice. Journal of Lipid Research, 2002, 43, 205-14.	4.2	50
90	Inhibition of Glucosylceramide Synthase Does Not Reverse Drug Resistance in Cancer Cells. Journal of Biological Chemistry, 2004, 279, 40412-40418.	3.4	48

#	Article	IF	Citations
91	Chemoenzymatic Synthesis of a Phosphorylated Glycoprotein. Angewandte Chemie - International Edition, 2016, 55, 5058-5061.	13.8	46
92	Neuraminidases 3 and 4 regulate neuronal function by catabolizing brain gangliosides. FASEB Journal, 2017, 31, 3467-3483.	0.5	46
93	Cellular effects of deoxynojirimycin analogues: uptake, retention and inhibition of glycosphingolipid biosynthesis. Biochemical Journal, 2004, 381, 861-866.	3.7	45
94	N-butyldeoxygalactonojirimycin reduces brain ganglioside and GM2 content in neonatal Sandhoff disease mice. Neurochemistry International, 2008, 52, 1125-1133.	3.8	44
95	Neural Stem Cell Transplantation Benefits a Monogenic Neurometabolic Disorder During the Symptomatic Phase of Disease. Stem Cells, 2009, 27, 2362-2370.	3.2	44
96	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.	2.7	41
97	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.	7.0	41
98	Brain Pathology in Mucopolysaccharidoses (MPS) Patients with Neurological Forms. Journal of Clinical Medicine, 2020, 9, 396.	2.4	40
99	Glycolipid depletion in antimicrobial therapy. Molecular Microbiology, 2003, 47, 453-461.	2.5	39
100	Cellular effects of deoxynojirimycin analogues: inhibition of N-linked oligosaccharide processing and generation of free glucosylated oligosaccharides. Biochemical Journal, 2004, 381, 867-875.	3.7	39
101	FTY720/fingolimod increases NPC1 and NPC2 expression and reduces cholesterol and sphingolipid accumulation in Niemannâ€Pick type C mutant fibroblasts. FASEB Journal, 2017, 31, 1719-1730.	0.5	39
102	Globosides but Not Isoglobosides Can Impact the Development of Invariant NKT Cells and Their Interaction with Dendritic Cells. Journal of Immunology, 2012, 189, 3007-3017.	0.8	38
103	Altered distribution and function of natural killer cells in murine and human Niemann-Pick disease type C1. Blood, 2014, 123, 51-60.	1.4	38
104	GM1 Gangliosidosisâ€”A Mini-Review. Frontiers in Genetics, 2021, 12, 734878.	2.3	38
105	Inhibitors of Glycosphingolipid Biosynthesis.. Trends in Glycoscience and Glycotechnology, 1995, 7, 495-511.	0.1	38
106	Acetyl-leucine slows disease progression in lysosomal storage disorders. Brain Communications, 2021, 3, fcaa148.	3.3	37
107	Current methods to analyze lysosome morphology, positioning, motility and function. Traffic, 2022, 23, 238-269.	2.7	37
108	The sensitivity of murine spermiogenesis to miglustat is a quantitative trait: a pharmacogenetic study. Reproductive Biology and Endocrinology, 2007, 5, 1.	3.3	35

#	Article	IF	Citations
109	Analysis and isolation of human transferrin receptor using the OKT-9 monoclonal antibody covalently crosslinked to magnetic beads. Analytical Biochemistry, 1991, 199, 219-222.	2.4	33
110	Drugâ€induced increase in lysobisphosphatidic acid reduces the cholesterol overload in Niemannâ€“Pick type C cells and mice. EMBO Reports, 2019, 20, e47055.	4.5	33
111	Storage diseases: new insights into sphingolipid functions. Trends in Cell Biology, 2003, 13, 195-203.	7.9	32
112	An Inducible Mouse Model of Late Onset Tayâ€“Sachs Disease. Neurobiology of Disease, 2002, 10, 201-210.	4.4	31
113	N-Butyl-<scp>l</scp>-deoxynojirimycin (<scp>l</scp>-NBDNJ): Synthesis of an Allosteric Enhancer of l±-Glucosidase Activity for the Treatment of Pompe Disease. Journal of Medicinal Chemistry, 2017, 60, 9462-9469.	6.4	31
114	Modulation of THP-1 Macrophage and Cholesterol-Loaded Foam Cell Apolipoprotein E Levels by Glycosphingolipids. Biochemical and Biophysical Research Communications, 2002, 290, 1361-1367.	2.1	30
115	c-Abl Inhibition Activates TFEB and Promotes Cellular Clearance in a Lysosomal Disorder. IScience, 2020, 23, 101691.	4.1	30
116	Pathogenic mycobacteria achieve cellular persistence by inhibiting the Niemann-Pick Type C disease cellular pathway. Wellcome Open Research, 0, 1, 18.	1.8	30
117	Critical role of iron in the pathogenesis of the murine gangliosidoses. Neurobiology of Disease, 2009, 34, 406-416.	4.4	29
118	Inhibition of Glycosphingolipid Biosynthesis Does Not Impair Growth or Morphogenesis of the Postimplantation Mouse Embryo. Journal of Neurochemistry, 2002, 70, 871-882.	3.9	28
119	Glycosphingolipids in endocytic membrane transport. Seminars in Cell and Developmental Biology, 2004, 15, 409-416.	5.0	28
120	The metabolism of glucocerebrosides â€” From 1965 to the present. Molecular Genetics and Metabolism, 2017, 120, 22-26.	1.1	28
121	Biomarkers for disease progression and AAV therapeutic efficacy in feline Sandhoff disease. Experimental Neurology, 2015, 263, 102-112.	4.1	26
122	Beneficial Effects of Acetyl-DL-Leucine (ADLL) in a Mouse Model of Sandhoff Disease. Journal of Clinical Medicine, 2020, 9, 1050.	2.4	26
123	Pathogenic mycobacteria achieve cellular persistence by inhibiting the Niemann-Pick Type C disease cellular pathway. Wellcome Open Research, 2016, 1, 18.	1.8	26
124	Long-term non-hormonal male contraception in mice using N-butyldeoxynojirimycin. Human Reproduction, 2006, 21, 1309-1315.	0.9	25
125	Silencing the porcine iGb3s gene does not affect GalÎ±3Gal levels or measures of anticipated pigâ€toâ€human and pigâ€toâ€primate acute rejection. Xenotransplantation, 2016, 23, 106-116.	2.8	25
126	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.	5.6	24

#	Article	IF	Citations
127	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.	2.9	24
128	Inhibition of glycogen breakdown by imino sugars in vitro and in vivo. Biochemical Pharmacology, 2004, 67, 697-705.	4.4	23
129	Restricted ketogenic diet enhances the therapeutic action of Nâ€butyldeoxynojirimycin towards brain GM2 accumulation in adult Sandhoff disease mice. Journal of Neurochemistry, 2010, 113, 1525-1535.	3.9	23
130	Lysosomal defects and storage. , 2004, , 32-49.		23
131	Defective Cytochrome P450-Catalysed Drug Metabolism in Niemann-Pick Type C Disease. PLoS ONE, 2016, 11, e0152007.	2.5	22
132	CD1d presentation of glycolipids. Immunology and Cell Biology, 2008, 86, 588-597.	2.3	21
133	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.	7.1	21
134	Unexpected differences in the pharmacokinetics of N-acetyl-DL-leucine enantiomers after oral dosing and their clinical relevance. PLoS ONE, 2020, 15, e0229585.	2.5	21
135	Modulation of cell-surface transferrin receptor by the imino sugar N-butyldeoxynojirimycin. FEBS Journal, 1992, 208, 187-193.	0.2	20
136	Glycolipid receptor depletion as an approach to specific antimicrobial therapy. FEMS Microbiology Letters, 2006, 258, 1-8.	1.8	18
137	Bridging the age spectrum of neurodegenerative storage diseases. Best Practice and Research in Clinical Endocrinology and Metabolism, 2015, 29, 127-143.	4.7	18
138	Haematopoietic Stem Cell Transplantation Arrests theÂProgression of Neurodegenerative Disease in Late-Onset Tay-Sachs Disease. JIMD Reports, 2017, 41, 17-23.	1.5	18
139	Glycosphingolipid metabolism and its role in ageing and Parkinsonâ€™s disease. Glycoconjugate Journal, 2022, 39, 39-53.	2.7	18
140	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.	3.3	17
141	Upregulating Î²-hexosaminidase activity in rodents prevents Î±-synuclein lipid associations and protects dopaminergic neurons from Î±-synuclein-mediated neurotoxicity. Acta Neuropathologica Communications, 2020, 8, 127.	5.2	17
142	Genetic background modifies phenotypic severity and longevity in a mouse model of Niemann-Pick disease type C1. DMM Disease Models and Mechanisms, 2020, 13, .	2.4	17
143	A Novel Mouse Model of a Patient Mucolipidosis II Mutation Recapitulates Disease Pathology. Journal of Biological Chemistry, 2014, 289, 26709-26721.	3.4	16
144	NPC1 Deficiency in Mice is Associated with Fetal Growth Restriction, Neonatal Lethality and Abnormal Lung Pathology. Journal of Clinical Medicine, 2020, 9, 12.	2.4	16

#	Article	IF	Citations
145	Investigating the Mechanism of Cyclodextrins in the Treatment of Niemannâ€Pick Disease Type C Using Crosslinked 2â€Hydroxypropylâ€Î²â€cyclodextrin. Small, 2020, 16, e2004735.	10.0	16
146	Acetylation turns leucine into a drug by membrane transporter switching. Scientific Reports, 2021, 11, 15812.	3.3	16
147	Inhibition of Î±-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, 39303-39309.	3.4	15
148	Transcriptome of HPÎ²CD-treated Niemann-Pick disease type C1 cells highlights GPNMB as a biomarker for therapeutics. Human Molecular Genetics, 2021, 30, 2456-2468.	2.9	15
149	Invariant natural killer <scp>T</scp> cells are not affected by lysosomal storage in patients with <scp>N</scp>iemannâ€<scp>P</scp>ick disease type <scp>C</scp>. European Journal of Immunology, 2012, 42, 1886-1892.	2.9	14
150	Effects of miglustat treatment in a patient affected by an atypical form of Tangier disease. Orphanet Journal of Rare Diseases, 2014, 9, 143.	2.7	14
151	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.	0.5	14
152	Carbohydrate Receptor Depletion as an Antimicrobial Strategy for Prevention of Urinary Tract Infection. Journal of Infectious Diseases, 2001, 183, S70-S73.	4.0	13
153	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, 2316-2323.	3.8	13
154	1H NMR-Linked Metabolomics Analysis of Liver from a Mouse Model of NP-C1 Disease. Journal of Proteome Research, 2016, 15, 3511-3527.	3.7	13
155	Combined Anti-inflammatory and Neuroprotective Treatments Have the Potential to Impact Disease Phenotypes in Cln3â^'/â^' Mice. Frontiers in Neurology, 2019, 10, 963.	2.4	13
156	Mechanistic convergence and shared therapeutic targets in Niemannâ€Pick disease. Journal of Inherited Metabolic Disease, 2020, 43, 574-585.	3.6	13
157	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, 2533.	4.1	13
158	Selective estrogen receptor modulators (SERMs) affect cholesterol homeostasis through the master regulators SREBP and LXR. Biomedicine and Pharmacotherapy, 2021, 141, 111871.	5.6	13
159	Vesicle cholesterol controls exocytotic fusion pore. Cell Calcium, 2022, 101, 102503.	2.4	13
160	Substrate deprivation: A new therapeutic approach for the glycosphingolipid lysosomal storage diseases. Expert Reviews in Molecular Medicine, 2000, 2, 1-17.	3.9	12
161	New Developments in Treating Glycosphingolipid Storage Diseases. Advances in Experimental Medicine and Biology, 2005, 564, 117-126.	1.6	12
162	Glycosphingolipid storage leads to the enhanced degradation of the B cell receptor in Sandhoff disease mice. Journal of Inherited Metabolic Disease, 2010, 33, 261-270.	3.6	12

#	Article	IF	Citations
163	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, 625.	4.1	12
164	Hepatic metabolic response to restricted copper intake in a Niemannâ€“Pick C murine model. Metallomics, 2014, 6, 1527-1539.	2.4	11
165	Sandhoff Disease: Improvement of Gait by Acetyl-DL-Leucine: A Case Report. Neuropediatrics, 2020, 51, 450-452.	0.6	11
166	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.	6.5	11
167	Inhibition of substrate synthesis: a pharmacological approach for glycosphingolipid storage disease therapy. , 2004, , 381-408.		11
168	Case Report: Ursodeoxycholic acid treatment in Niemann-Pick disease type C; clinical experience in four cases. Wellcome Open Research, 2017, 2, 75.	1.8	11
169	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.	1.9	10
170	Lipid-mediated impairment of axonal lysosome transport contributing to autophagic stress. Autophagy, 2021, 17, 1796-1798.	9.1	10
171	Unbiased yeast screens identify cellular pathways affected in Niemannâ€“Pick disease type C. Life Science Alliance, 2020, 3, e201800253.	2.8	10
172	Medicinal use of Iminosugars. , 0, , 295-326.		9
173	Defective platelet function in <scp>Niemannâ€Pick</scp> disease type <scp>C1</scp>. JIMD Reports, 2020, 56, 46-57.	1.5	9
174	Differential sensitivity of mouse strains to an N-alkylated imino sugar: glycosphingolipid metabolism and acrosome formation. Pharmacogenomics, 2008, 9, 717-731.	1.3	8
175	Preferential Coupling of the NAADP Pathway to Exocytosis in T-Cells. Messenger (Los Angeles, Calif:) Tj ETQq1 1 0.784314 rgBT /Over	0.3	8
176	Chemoenzymatic Synthesis of a Phosphorylated Glycoprotein. Angewandte Chemie, 2016, 128, 5142-5145.	2.0	8
177	International consensus on clinical severity scale use in evaluating Niemannâ€“Pick disease Type C in paediatric and adult patients: results from a Delphi Study. Orphanet Journal of Rare Diseases, 2021, 16, 482.	2.7	8
178	Monoclonal antibodies specific for novel murine cell surface markers define subpopulations of germinal center cells. Cellular Immunology, 1992, 143, 449-466.	3.0	7
179	Glycomimetic affinity-enrichment proteomics identifies partners for a clinically-utilized iminosugar. Chemical Science, 2013, 4, 3442.	7.4	7
180	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, 1239-1240.	3.6	7

#	Article	IF	Citations
181	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.	4.1	7
182	Glucosamine amends CNS pathology in mucopolysaccharidosis IIIC mouse expressing misfolded HGSNAT. Journal of Experimental Medicine, 2022, 219, .	8.5	7
183	Circadian profiling in two mouse models of lysosomal storage disorders; Niemann Pick type-C and Sandhoff disease. Behavioural Brain Research, 2016, 297, 213-223.	2.2	6
184	Synthesis and Study of Multifunctional Cyclodextrinâ€“Deferasirox Hybrids. ChemMedChem, 2019, 14, 1484-1492.	3.2	5
185	Substrate Reduction Therapy. , 2007, , 153-168.		5
186	Measuring relative lysosomal volume for monitoring lysosomal storage diseases. Methods in Cell Biology, 2015, 126, 331-347.	1.1	4
187	Urinary excretion and metabolism of miglustat and valproate in patients with Niemannâ€“Pick type C1 disease: One- and two-dimensional solution-state 1 H NMR studies. Journal of Pharmaceutical and Biomedical Analysis, 2016, 117, 276-288.	2.8	4
188	Molecular basis for a new bovine model of Niemann-Pick type C disease. PLoS ONE, 2020, 15, e0238697.	2.5	4
189	Identification of genetic modifiers of murine hepatic Î²-glucocerebrosidase activity. Biochemistry and Biophysics Reports, 2021, 28, 101105.	1.3	4
190	Ultrastructural changes in the Golgi apparatus and secretory granules of HL-60 cells treated with the imino sugar N-butyldeoxynojirimycin. Biology of the Cell, 1997, 89, 123-131.	2.0	3
191	Glycosphingolipid depletion in PC12 cells using iminosugars protects neuronal membranes from anti-ganglioside antibody mediated injury. Journal of Neuroimmunology, 2008, 203, 33-38.	2.3	3
192	A new surrogate marker for CNS pathology in Niemannâ€“Pick disease type C?. Molecular Genetics and Metabolism, 2009, 96, 53-54.	1.1	3
193	Anti-TNF therapy for inflammatory bowel disease in patients with neurodegenerative Niemann-Pick disease Type C. Wellcome Open Research, 0, 7, 11.	1.8	3
194	Correlation of age of onset and clinical severity in Niemannâ€“Pick disease type C1 with lysosomal abnormalities and gene expression. Scientific Reports, 2022, 12, 2162.	3.3	3
195	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.	1.8	2
196	Miglustat. Drugs, 2003, 63, 2435-2436.	10.9	1
197	A modified density gradient proteomic-based method to analyze endolysosomal proteins in cardiac tissue. IScience, 2021, 24, 102949.	4.1	1
198	Ultrastructural changes in the Golgi apparatus and secretory granules of HL-60 cells treated with the imino sugar N-butyldeoxynojirimycin. Biology of the Cell, 1997, 89, 123-131.	2.0	1

#	Article	IF	Citations
199	Differential response of the liver to bile acid treatment in a mouse model of Niemann-Pick disease type C. Wellcome Open Research, 0, 2, 76.	1.8	1
200	Miglustat: profile report. Drugs and Therapy Perspectives, 2004, 20, 5-7.	0.6	0
201	Increasing Diversity in Admissions to Postgraduate Study. Journal of Medicinal Chemistry, 2022, 65, 5867-5869.	6.4	0
202	Title is missing!. , 2020, 15, e0229585.		0
203	Title is missing!. , 2020, 15, e0229585.		0
204	Title is missing!. , 2020, 15, e0229585.		0
205	Title is missing!. , 2020, 15, e0229585.		0