Daniel S Ory

List of Publications by Year
in descending order

76 papers	3,929 citations	136950 32 h-index	128289 60 g-index
78 all docs	78 docs citations	78 times ranked	5715 citing authors

#	Article	IF	Citations
1	Lysosomal cholesterol activates mTORC1 via an SLC38A9â€“Niemann-Pick C1 signaling complex. Science, 2017, 355, 1306-1311.	12.6	386
2	Intrathecal 2-hydroxypropyl-Î²-cyclodextrin decreases neurological disease progression in Niemann-Pick disease, type C1: a non-randomised, open-label, phase 1â€“2 trial. Lancet, The, 2017, 390, 1758-1768.	13.7	275
3	Fatty acid synthesis configures the plasma membrane for inflammation in diabetes. Nature, 2016, 539, 294-298.	27.8	213
4	ERâ€“lysosome contacts enable cholesterol sensing by mTORC1 and drive aberrant growth signalling in Niemannâ€“Pick type C. Nature Cell Biology, 2019, 21, 1206-1218.	10.3	193
5	A New Glucocerebrosidase Chaperone Reduces Â-Synuclein and Glycolipid Levels in iPSC-Derived Dopaminergic Neurons from Patients with Gaucher Disease and Parkinsonism. Journal of Neuroscience, 2016, 36, 7441-7452.	3.6	189
6	Intracisternal cyclodextrin prevents cerebellar dysfunction and Purkinje cell death in feline Niemann-Pick type C1 disease. Science Translational Medicine, 2015, 7, 276ra26.	12.4	174
7	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
8	Nuclear Receptor Signaling in the Control of Cholesterol Homeostasis. Circulation Research, 2004, 95, 660-670.	4.5	111
9	A Murine Niemann-Pick C1 I1061T Knock-In Model Recapitulates the Pathological Features of the Most Prevalent Human Disease Allele. Journal of Neuroscience, 2015, 35, 8091-8106.	3.6	97
10	Effectors of Rapid Homeostatic Responses of Endoplasmic Reticulum Cholesterol and 3-Hydroxy-3-methylglutaryl-CoA Reductase. Journal of Biological Chemistry, 2008, 283, 1445-1455.	3.4	91
11	Development of a bile acidâ€“based newborn screen for Niemann-Pick disease type C. Science Translational Medicine, 2016, 8, 337ra63.	12.4	89
12	An optical nanoreporter of endolysosomal lipid accumulation reveals enduring effects of diet on hepatic macrophages in vivo. Science Translational Medicine, 2018, 10, .	12.4	80
13	Lysosomal Acid Lipase Deficiency Impairs Regulation of ABCA1 Gene and Formation of High Density Lipoproteins in Cholesteryl Ester Storage Disease. Journal of Biological Chemistry, 2011, 286, 30624-30635.	3.4	79
14	Intrathecal 2-hydroxypropyl-beta-cyclodextrin in a single patient with Niemannâ€“Pick C1. Molecular Genetics and Metabolism, 2015, 116, 75-79.	1.1	76
15	Reduction of TMEM97 increases NPC1 protein levels and restores cholesterol trafficking in Niemann-pick type C1 disease cells. Human Molecular Genetics, 2016, 25, 3588-3599.	2.9	74
16	Methyl-Î²-cyclodextrin restores impaired autophagy flux in Niemann-Pick C1-deficient cells through activation of AMPK. Autophagy, 2017, 13, 1435-1451.	9.1	73
17	The Niemann-Pick Disease Genes Regulators of Cellular Cholesterol Homeostasis. Trends in Cardiovascular Medicine, 2004, 14, 66-72.	4.9	68
18	Psychosine, the cytotoxic sphingolipid that accumulates in globoid cell leukodystrophy, alters membrane architecture. Journal of Lipid Research, 2013, 54, 3303-3311.	4.2	61

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19	Long-Term Treatment of Niemann-Pick Type C1 Disease With Intrathecal 2-Hydroxypropyl-Î²-Cyclodextrin. Pediatric Neurology, 2018, 80, 24-34.	2.1	60
20	AAVrh10 Gene Therapy Ameliorates Central and Peripheral Nervous System Disease in Canine Globoid Cell Leukodystrophy (Krabbe Disease). Human Gene Therapy, 2018, 29, 785-801.	2.7	56
21	A validated LC-MS/MS assay for quantification of 24(S)-hydroxycholesterol in plasma and cerebrospinal fluid. Journal of Lipid Research, 2015, 56, 1222-1233.	4.2	54
22	A characterization of Gaucher iPS-derived astrocytes: Potential implications for Parkinson's disease. Neurobiology of Disease, 2020, 134, 104647.	4.4	50
23	snoRNA U17 Regulates Cellular Cholesterol Trafficking. Cell Metabolism, 2015, 21, 855-867.	16.2	49
24	Improved Coarse-Grained Modeling of Cholesterol-Containing Lipid Bilayers. Journal of Chemical Theory and Computation, 2014, 10, 2137-2150.	5.3	48
25	Bone marrow transplantation increases efficacy of central nervous system-directed enzyme replacement therapy in the murine model of globoid cell leukodystrophy. Molecular Genetics and Metabolism, 2012, 107, 186-196.	1.1	47
26	Krabbe disease successfully treated via monotherapy of intrathecal gene therapy. Journal of Clinical Investigation, 2020, 130, 4906-4920.	8.2	41
27	A Diet Rich in Medium-Chain Fatty Acids Improves Systolic Function and Alters the Lipidomic Profile in Patients With Type 2 Diabetes: A Pilot Study. Journal of Clinical Endocrinology and Metabolism, 2016, 101, 504-512.	3.6	39
28	Metabolism of Non-Enzymatically Derived Oxysterols: Clues from sterol metabolic disorders. Free Radical Biology and Medicine, 2019, 144, 124-133.	2.9	39
29	A novel gene editing system to treat both Tayâ€“Sachs and Sandhoff diseases. Gene Therapy, 2020, 27, 226-236.	4.5	39
30	Niemann-Pick Type C Disease Reveals a Link between Lysosomal Cholesterol and PtdIns(4,5)P2 That Regulates Neuronal Excitability. Cell Reports, 2019, 27, 2636-2648.e4.	6.4	38
31	Fourier Transform Infrared Microscopy Enables Guidance of Automated Mass Spectrometry Imaging to Predefined Tissue Morphologies. Scientific Reports, 2018, 8, 313.	3.3	37
32	Cholesterol homeostatic responses provide biomarkers for monitoring treatment for the neurodegenerative disease Niemannâ€“Pick C1 (NPC1). Human Molecular Genetics, 2014, 23, 6022-6033.	2.9	36
33	Neural stem cells for disease modeling and evaluation of therapeutics for Tay-Sachs disease. Orphanet Journal of Rare Diseases, 2018, 13, 152.	2.7	34
34	Disease-associated mutations in Niemann-Pick type C1 alter ER calcium signaling and neuronal plasticity. Journal of Cell Biology, 2019, 218, 4141-4156.	5.2	32
35	Establishing the precise evolutionary history of a gene improves prediction of disease-causing missense mutations. Genetics in Medicine, 2016, 18, 1029-1036.	2.4	31
36	N-acyl-O-phosphocholineserines: structures of a novel class of lipids that are biomarkers for Niemann-Pick C1 disease. Journal of Lipid Research, 2019, 60, 1410-1424.	4.2	31

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37	Normalization of Hepatic Homeostasis in the Npc1 Mouse Model of Niemann-Pick Type C Disease Treated with the Histone Deacetylase Inhibitor Vorinostat. Journal of Biological Chemistry, 2017, 292, 4395-4410.	3.4	28
38	Glucocerebrosidase haploinsufficiency in A53T Î±-synuclein mice impacts disease onset and course. Molecular Genetics and Metabolism, 2017, 122, 198-208.	1.1	28
39	Monitoring the itinerary of lysosomal cholesterol in Niemann-Pick Type C1-deficient cells after cyclodextrin treatment. Journal of Lipid Research, 2020, 61, 403-412.	4.2	28
40	Genetic and pharmacological evidence implicates cathepsins in Niemann-Pick C cerebellar degeneration. Human Molecular Genetics, 2016, 25, 1434-1446.	2.9	27
41	Enhanced Efficacy and Increased Long-Term Toxicity of CNS-Directed, AAV-Based Combination Therapy for Krabbe Disease. Molecular Therapy, 2021, 29, 691-701.	8.2	27
42	Pathogenic mycobacteria achieve cellular persistence by inhibiting the Niemann-Pick Type C disease cellular pathway. Wellcome Open Research, 2016, 1, 18.	1.8	26
43	Circulating ceramide ratios and risk of vascular brain aging and dementia. Annals of Clinical and Translational Neurology, 2020, 7, 160-168.	3.7	25
44	Application of N-palmitoyl-O-phosphocholineserine for diagnosis and assessment of response to treatment in Niemann-Pick type C disease. Molecular Genetics and Metabolism, 2020, 129, 292-302.	1.1	24
45	Clinical, electrophysiological, and biochemical markers of peripheral and central nervous system disease in canine globoid cell leukodystrophy (<scp>K</scp>rabbe'sÂdisease). Journal of Neuroscience Research, 2016, 94, 1007-1017.	2.9	23
46	High-content screen for modifiers of Niemann-Pick type C disease in patient cells. Human Molecular Genetics, 2018, 27, 2101-2112.	2.9	23
47	Oxysterol Signatures Distinguish Age-Related Macular Degeneration from Physiologic Aging. EBioMedicine, 2018, 32, 9-20.	6.1	23
48	Prevention of hepatic fibrosis with liver microsomal triglyceride transfer protein deletion in liver fatty acid binding protein null mice. Hepatology, 2017, 65, 836-852.	7.3	22
49	Diagnosis of niemann-pick C1 by measurement of bile acid biomarkers in archived newborn dried blood spots. Molecular Genetics and Metabolism, 2019, 126, 183-187.	1.1	21
50	IP ₃ R-driven increases in mitochondrial Ca ²⁺ promote neuronal death in NPC disease. Proceedings of the National Academy of Sciences of the United States of America, 2021, 118, .	7.1	21
51	Selective Aster inhibitors distinguish vesicular and nonvesicular sterol transport mechanisms. Proceedings of the National Academy of Sciences of the United States of America, 2021, 118, .	7.1	21
52	A new glucocerebrosidase deficient neuronal cell model provides a tool to probe pathophysiology and therapeutics for Gaucher disease. DMM Disease Models and Mechanisms, 2016, 9, 769-78.	2.4	20
53	Comprehensive behavioral and biochemical outcomes of novel murine models of GM1-gangliosidosis and Morquio syndrome type B. Molecular Genetics and Metabolism, 2019, 126, 139-150.	1.1	20
54	Neurologic Abnormalities in Mouse Models of the Lysosomal Storage Disorders Mucolipidosis II and Mucolipidosis III Î³. PLoS ONE, 2014, 9, e109768.	2.5	20

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55	2-Hydroxypropyl-Î²-cyclodextrin is the active component in a triple combination formulation for treatment of Niemann-Pick C1 disease. Biochimica Et Biophysica Acta - Molecular and Cell Biology of Lipids, 2019, 1864, 1545-1561.	2.4	19
56	Development and validation of sensitive LC-MS/MS assays for quantification of HP-#x03B2;-CD in human plasma and CSF. Journal of Lipid Research, 2014, 55, 1537-1548.	4.2	18
57	Phenotypic divergence in two lines ofL-Fabp^{â^'/â^'}mice reflects substrain differences and environmental modifiers. American Journal of Physiology - Renal Physiology, 2015, 309, G648-G661.	3.4	17
58	Analytical Characterization of Methyl-Î²-Cyclodextrin for Pharmacological Activity to Reduce Lysosomal Cholesterol Accumulation in Niemann-Pick Disease Type C1 Cells. Assay and Drug Development Technologies, 2017, 15, 154-166.	1.2	17
59	Lipidomic Evaluation of Feline Neurologic Disease after AAV Gene Therapy. Molecular Therapy - Methods and Clinical Development, 2017, 6, 135-142.	4.1	17
60	Cerebrospinal fluid and serum glycosphingolipid biomarkers in canine globoid cell leukodystrophy (Krabbe Disease). Molecular and Cellular Neurosciences, 2020, 102, 103451.	2.2	16
61	NPC1 regulates the distribution of phosphatidylinositol 4â€kinases at Golgi and lysosomal membranes. EMBO Journal, 2021, 40, e105990.	7.8	14
62	Fostering collaborative research for rare genetic disease: the example of niemann-pick type C disease. Orphanet Journal of Rare Diseases, 2016, 11, 161.	2.7	13
63	A HILICâ€MS/MS method for simultaneous quantification of the lysosomal disease markers galactosylsphingosine and glucosylsphingosine in mouse serum. Biomedical Chromatography, 2018, 32, e4235.	1.7	12
64	Chylomicrons and Lipoprotein Lipase at the Endothelial Surface: Bound and GAG-ged?. Cell Metabolism, 2007, 5, 229-231.	16.2	11
65	A novel intrinsically fluorescent probe for study of uptake and trafficking of 25-hydroxycholesterol. Journal of Lipid Research, 2015, 56, 2408-2419.	4.2	11
66	Structural design of intrinsically fluorescent oxysterols. Chemistry and Physics of Lipids, 2018, 212, 26-34.	3.2	11
67	Alterations in plasma triglycerides and ceramides: links with cardiac function in humans with type 2 diabetes. Journal of Lipid Research, 2020, 61, 1065-1074.	4.2	11
68	Application of a glycinated bile acid biomarker for diagnosis and assessment of response to treatment in Niemann-pick disease type C1. Molecular Genetics and Metabolism, 2020, 131, 405-417.	1.1	11
69	4Î²-Hydroxycholesterol is a prolipogenic factor that promotes SREBP1c expression and activity through the liver X receptor. Journal of Lipid Research, 2021, 62, 100051.	4.2	10
70	ApoA-1 in Diabetes: Damaged Goods: FIG. 1.. Diabetes, 2010, 59, 2358-2359.	0.6	7
71	A human iPSC-derived inducible neuronal model of Niemann-Pick disease, type C1. BMC Biology, 2021, 19, 218.	3.8	7
72	Improved systemic AAV gene therapy with a neurotrophic capsid in Niemannâ€“Pick disease type C1 mice. Life Science Alliance, 2021, 4, e202101040.	2.8	6

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73	19q13.12 microdeletion syndrome fibroblasts display abnormal storage of cholesterol and sphingolipids in the endo-lysosomal system. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2018, 1864, 2108-2118.	3.8	4
74	Getting a â€œHoldâ€•on NPC2. Cell Metabolism, 2009, 10, 161-162.	16.2	2
75	Whole exome sequencing and functional characterization increase diagnostic yield in siblings with a 46, XY difference of sexual development (DSD). Journal of Steroid Biochemistry and Molecular Biology, 2021, 212, 105908.	2.5	1
76	The nonâ€coding RNA gadd7 is a regulator of lipotoxicâ€induced ROS and ER stress. FASEB Journal, 2008, 22, 1034.1.	0.5	0