Forbes D Porter

List of Publications by Year
in descending order

144 papers	7,262 citations	46918 47 h-index	62479 80 g-index
145 all docs	145 docs citations	145 times ranked	6166 citing authors

#	Article	IF	Citations
1	Mutations in the Human Sterol Î”7-Reductase Gene at 11q12-13 Cause Smith-Lemli-Opitz Syndrome. American Journal of Human Genetics, 1998, 63, 55-62.	2.6	405
2	Malformation syndromes caused by disorders of cholesterol synthesis. Journal of Lipid Research, 2011, 52, 6-34.	2.0	375
3	A defective response to Hedgehog signaling in disorders of cholesterol biosynthesis. Nature Genetics, 2003, 33, 508-513.	9.4	363
4	Cholesterol Oxidation Products Are Sensitive and Specific Blood-Based Biomarkers for Niemann-Pick C1 Disease. Science Translational Medicine, 2010, 2, 56ra81.	5.8	302
5	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.	6.3	275
6	Smithâ€“Lemliâ€“Opitz syndrome: pathogenesis, diagnosis and management. European Journal of Human Genetics, 2008, 16, 535-541.	1.4	264
7	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.	2.0	230
8	Behavior phenotype in the RSH/Smith-Lemli-Opitz syndrome. American Journal of Medical Genetics Part A, 2001, 98, 191-200.	2.4	202
9	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.	1.1	171
10	Lathosterolosis: an inborn error of human and murine cholesterol synthesis due to lathosterol 5-desaturase deficiency. Human Molecular Genetics, 2003, 12, 1631-1641.	1.4	153
11	Linear clinical progression, independent of age of onset, in Niemannâ€“Pick disease, type C. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2010, 153B, 132-140.	1.1	145
12	Human malformation syndromes due to inborn errors of cholesterol synthesis. Current Opinion in Pediatrics, 2003, 15, 607-613.	1.0	115
13	Oxidative stress in Niemannâ€“Pick disease, type C. Molecular Genetics and Metabolism, 2010, 101, 214-218.	0.5	113
14	Collaborative Development of 2-Hydroxypropyl-β-Cyclodextrin for the Treatment of Niemann-Pick Type C1 Disease. Current Topics in Medicinal Chemistry, 2014, 14, 330-339.	1.0	108
15	Î´-Tocopherol Reduces Lipid Accumulation in Niemann-Pick Type C1 and Wolman Cholesterol Storage Disorders. Journal of Biological Chemistry, 2012, 287, 39349-39360.	1.6	107
16	A novel, highly sensitive and specific biomarker for Niemann-Pick type C1 disease. Orphanet Journal of Rare Diseases, 2015, 10, 78.	1.2	105
17	RSH/Smithâ€“Lemliâ€“Opitz Syndrome: A Multiple Congenital Anomaly/Mental Retardation Syndrome due to an Inborn Error of Cholesterol Biosynthesis. Molecular Genetics and Metabolism, 2000, 71, 163-174.	0.5	102
18	Trifunctional lipid probes for comprehensive studies of single lipid species in living cells. Proceedings of the National Academy of Sciences of the United States of America, 2017, 114, 1566-1571.	3.3	100

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19	3Î²-Hydroxysterol Î”7-reductase and the Smithâ€“Lemliâ€“Opitz syndrome. Molecular Genetics and Metabolism, 2005, 84, 112-126.	0.5	92
20	Development of a bile acidâ€“based newborn screen for Niemann-Pick disease type C. Science Translational Medicine, 2016, 8, 337ra63.	5.8	89
21	Identification of Niemann-Pick C1 disease biomarkers through sphingolipid profiling. Journal of Lipid Research, 2013, 54, 2800-2814.	2.0	88
22	Malformation syndromes due to inborn errors of cholesterol synthesis. Journal of Clinical Investigation, 2002, 110, 715-724.	3.9	88
23	Abnormal sterols in cholesterol-deficiency diseases cause secretory granule malformation and decreased membrane curvature. Journal of Cell Science, 2006, 119, 1876-1885.	1.2	84
24	Microarray expression analysis and identification of serum biomarkers for Niemannâ€“Pick disease, type C1. Human Molecular Genetics, 2012, 21, 3632-3646.	1.4	84
25	Identification of novel bile acids as biomarkers for the early diagnosis of Niemannâ€Pick C disease. FEBS Letters, 2016, 590, 1651-1662.	1.3	82
26	Intrathecal 2-hydroxypropyl-beta-cyclodextrin in a single patient with Niemannâ€“Pick C1. Molecular Genetics and Metabolism, 2015, 116, 75-79.	0.5	76
27	Disorders of Cholesterol Metabolism and Their Unanticipated Convergent Mechanisms of Disease. Annual Review of Genomics and Human Genetics, 2014, 15, 173-194.	2.5	73
28	Human and mouse neuroinflammation markers in Niemannâ€Pick disease, type C1. Journal of Inherited Metabolic Disease, 2014, 37, 83-92.	1.7	71
29	Development and characterization of a hypomorphic Smithâ€“Lemliâ€“Opitz syndrome mouse model and efficacy of simvastatin therapy. Human Molecular Genetics, 2006, 15, 839-851.	1.4	67
30	Cholesterol deficiency in a mouse model of Smith-Lemli-Opitz syndrome reveals increased mast cell responsiveness. Journal of Experimental Medicine, 2006, 203, 1161-1171.	4.2	65
31	Characterization of placental cholesterol transport: ABCA1 is a potential target for in utero therapy of Smith-Lemli-Opitz syndrome. Human Molecular Genetics, 2008, 17, 3806-3813.	1.4	63
32	Relative acidic compartment volume as a lysosomal storage disorderâ€“associated biomarker. Journal of Clinical Investigation, 2014, 124, 1320-1328.	3.9	63
33	Residual cholesterol synthesis and simvastatin induction of cholesterol synthesis in Smithâ€“Lemliâ€“Opitz syndrome fibroblasts. Molecular Genetics and Metabolism, 2005, 85, 96-107.	0.5	62
34	Disruption of Dhcr7 and Insig1/2 in cholesterol metabolism causes defects in bone formation and homeostasis through primary cilium formation. Bone Research, 2020, 8, 1.	5.4	62
35	Auditory Phenotype of Niemann-Pick Disease, Type C1. Ear and Hearing, 2014, 35, 110-117.	1.0	60
36	Long-Term Treatment of Niemann-Pick Type C1 Disease With Intrathecal 2-Hydroxypropyl-Î²-Cyclodextrin. Pediatric Neurology, 2018, 80, 24-34.	1.0	60

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37	Identification of 7(8) and 8(9) unsaturated adrenal steroid metabolites produced by patients with 7-dehydrosterol-Î”7-reductase deficiency (Smithâ€“Lemliâ€“Opitz syndrome). Journal of Steroid Biochemistry and Molecular Biology, 2002, 82, 225-232.	1.2	59
38	Quantitative Proteomic Analysis of Niemann-Pick Disease, Type C1 Cerebellum Identifies Protein Biomarkers and Provides Pathological Insight. PLoS ONE, 2012, 7, e47845.	1.1	59
39	Mutation analysis and description of sixteen RSH/Smith-Lemli-Opitz syndrome patients: Polymerase chain reaction-based assays to simplify genotyping. American Journal of Medical Genetics Part A, 2000, 94, 214-227.	2.4	58
40	HEM dysplasia and ichthyosis are likely laminopathies and not due to 3Î²-hydroxysterol Î”14-reductase deficiency. Human Molecular Genetics, 2007, 16, 1176-1187.	1.4	56
41	Prevalence of Diabetes and Hypertension and Their Associated Risks for Poor Outcomes in Covid-19 Patients. Journal of the Endocrine Society, 2020, 4, bvaa102.	0.1	56
42	Analysis of shortâ€term behavioral effects of dietary cholesterol supplementation in Smithâ€“Lemliâ€“Opitz syndrome. American Journal of Medical Genetics, Part A, 2010, 152A, 91-95.	0.7	54
43	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.	2.0	54
44	Microglia activation in Niemannâ€“Pick disease, type C1 is amendable to therapeutic intervention. Human Molecular Genetics, 2018, 27, 2076-2089.	1.4	54
45	Cholesterol storage defect in RSH/Smithâ€“Lemliâ€“Opitz syndrome fibroblasts. Molecular Genetics and Metabolism, 2002, 75, 325-334.	0.5	52
46	Systemic AAV9 gene therapy improves the lifespan of mice with Niemann-Pick disease, type C1. Human Molecular Genetics, 2017, 26, ddw367.	1.4	50
47	The implications of 7-dehydrosterol-7-reductase deficiency (Smithâ€“Lemliâ€“Opitz syndrome) to neurosteroid production. Steroids, 2004, 69, 51-60.	0.8	48
48	Rescue of an In Vitro Neuron Phenotype Identified in Niemann-Pick Disease, Type C1 Induced Pluripotent Stem Cell-Derived Neurons by Modulating the WNT Pathway and Calcium Signaling. Stem Cells Translational Medicine, 2015, 4, 230-238.	1.6	48
49	Modeling Smith-Lemli-Opitz syndrome with induced pluripotent stem cells reveals a causal role for Wnt/Î²-catenin defects in neuronal cholesterol synthesis phenotypes. Nature Medicine, 2016, 22, 388-396.	15.2	46
50	Incidence of Smith-Lemli-Opitz syndrome in Ontario, Canada. American Journal of Medical Genetics Part A, 2001, 102, 18-20.	2.4	45
51	27-Hydroxylation of 7- and 8-dehydrocholesterol in Smithâ€“Lemliâ€“Opitz syndrome: a novel metabolic pathway. Steroids, 2003, 68, 497-502.	0.8	45
52	Development, behavior, and biomarker characterization of Smith-Lemli-Opitz syndrome: an update. Journal of Neurodevelopmental Disorders, 2016, 8, 12.	1.5	45
53	Malformation syndromes due to inborn errors of cholesterol synthesis. Journal of Clinical Investigation, 2002, 110, 715-724.	3.9	43
54	Activation of Rho GTPases in Smithâ€“Lemliâ€“Opitz syndrome: pathophysiological and clinical implications. Human Molecular Genetics, 2010, 19, 1347-1357.	1.4	42

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55	Pathogenesis, epidemiology, diagnosis and clinical aspects of Smithâ€“Lemliâ€“Opitz syndrome. Expert Opinion on Orphan Drugs, 2015, 3, 267-280.	0.5	42
56	Smith-Lemli-Opitz (RHS) syndrome: holoprosencephaly and homozygous IVS8-1G?C genotype. American Journal of Medical Genetics Part A, 2001, 103, 75-80.	2.4	41
57	Frequency and ethnic distribution of the commonDHCR7 mutation in Smith-Lemli-Opitz syndrome. American Journal of Medical Genetics Part A, 2001, 102, 383-386.	2.4	40
58	Quantitative Proteomics Analysis of Inborn Errors of Cholesterol Synthesis. Molecular and Cellular Proteomics, 2010, 9, 1461-1475.	2.5	40
59	Fusion of lysosomes with secretory organelles leads to uncontrolled exocytosis in the lysosomal storage disease mucolipidosis type <scp>IV</scp>. EMBO Reports, 2016, 17, 266-278.	2.0	39
60	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.	0.2	39
61	Modeling Niemann-Pick disease type C1 in zebrafish: a robust platform for in vivo screening of candidate therapeutic compounds. DMM Disease Models and Mechanisms, 2018, 11, .	1.2	38
62	Cholesterol homeostatic responses provide biomarkers for monitoring treatment for the neurodegenerative disease Niemannâ€“Pick C1 (NPC1). Human Molecular Genetics, 2014, 23, 6022-6033.	1.4	36
63	Apolipoprotein E genotype and neurological disease onset in Niemannâ€“Pick disease, type C1. American Journal of Medical Genetics, Part A, 2012, 158A, 2775-2780.	0.7	34
64	A placebo-controlled trial of simvastatin therapy in Smith-Lemli-Opitz syndrome. Genetics in Medicine, 2017, 19, 297-305.	1.1	34
65	Endocrine Conditions and COVID-19. Hormone and Metabolic Research, 2020, 52, 471-484.	0.7	34
66	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.	2.0	31
67	Cholesterol precursors and facial clefting. Journal of Clinical Investigation, 2006, 116, 2322-2325.	3.9	31
68	Cerebrospinal Fluid Calbindin D Concentration as a Biomarker of Cerebellar Disease Progression in Niemann-Pick Type C1 Disease. Journal of Pharmacology and Experimental Therapeutics, 2016, 358, 254-261.	1.3	29
69	Long-Term Neuropsychological Outcomes from an Open-Label Phase I/IIa Trial of 2-Hydroxypropyl-î²-Cyclodextrins (VTS-270) in Niemann-Pick Disease, Type C1. CNS Drugs, 2019, 33, 677-683.	2.7	28
70	Cholesterol biosynthesis from birth to adulthood in a mouse model for 7-dehydrosterol reductase deficiency (Smithâ€“Lemliâ€“Opitz syndrome). Steroids, 2007, 72, 802-808.	0.8	27
71	Efficacy of N-acetylcysteine in phenotypic suppression of mouse models of Niemannâ€“Pick disease, type C1. Human Molecular Genetics, 2013, 22, 3508-3523.	1.4	27
72	Altered transition metal homeostasis in Niemannâ€“Pick disease, type C1. Metallomics, 2014, 6, 542-553.	1.0	26

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73	Miglustat Treatment May Reduce Cerebrospinal Fluid Levels of the Axonal Degeneration Marker Tau in Niemannâ€“Pick Type C. JIMD Reports, 2011, 3, 45-52.	0.7	25
74	Hearing Loss is an Early Consequence of Npc1 Gene Deletion in the Mouse Model of Niemannâ€“Pick Disease, Type C. JARO - Journal of the Association for Research in Otolaryngology, 2014, 15, 529-541.	0.9	24
75	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.	0.5	24
76	Carrier frequency of the RSH/Smith-Lemli-Opitz IVS8-1G>C mutation in African Americans. American Journal of Medical Genetics Part A, 2003, 120A, 139-141.	2.4	22
77	Evaluation of age of death in Niemann-Pick disease, type C: Utility of disease support group websites to understand natural history. Molecular Genetics and Metabolism, 2019, 126, 466-469.	0.5	22
78	Defective Cytochrome P450-Catalysed Drug Metabolism in Niemann-Pick Type C Disease. PLoS ONE, 2016, 11, e0152007.	1.1	22
79	SMITH-LEMLI-OPITZ SYNDROME. Journal of the American Academy of Child and Adolescent Psychiatry, 2001, 40, 506-507.	0.3	21
80	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.	0.5	21
81	Abnormal LAMP1 glycosylation may play a role in Niemann-Pick disease, type C pathology. PLoS ONE, 2020, 15, e0227829.	1.1	21
82	Single Cell Transcriptome Analysis of Niemannâ€“Pick Disease, Type C1 Cerebella. International Journal of Molecular Sciences, 2020, 21, 5368.	1.8	20
83	Association of Miglustat With Swallowing Outcomes in Niemann-Pick Disease, Type C1. JAMA Neurology, 2020, 77, 1564.	4.5	20
84	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.	1.2	19
85	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.	2.0	18
86	Quantitating the epigenetic transformation contributing to cholesterol homeostasis using Gaussian process. Nature Communications, 2019, 10, 5052.	5.8	18
87	Identification of Novel Pathways Associated with Patterned Cerebellar Purkinje Neuron Degeneration in Niemann-Pick Disease, Type C1. International Journal of Molecular Sciences, 2020, 21, 292.	1.8	18
88	Discordant phenotype and sterol biochemistry in Smithâ€“Lemliâ€“Opitz syndrome. American Journal of Medical Genetics, Part A, 2010, 152A, 2094-2098.	0.7	17
89	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.	1.6	17
90	Unique molecular signature in mucolipidosis type IV microglia. Journal of Neuroinflammation, 2019, 16, 276.	3.1	17

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91	Genetic background modifies phenotypic severity and longevity in a mouse model of Niemann-Pick disease type C1. DMM Disease Models and Mechanisms, 2020, 13, .	1.2	17
92	NPC1 Deficiency in Mice is Associated with Fetal Growth Restriction, Neonatal Lethality and Abnormal Lung Pathology. Journal of Clinical Medicine, 2020, 9, 12.	1.0	16
93	Variations in EEG discharges predict ADHD severity within individual Smith-Lemli-Opitz patients. Neurology, 2014, 83, 151-159.	1.5	15
94	Corpus Callosum Diffusion Tensor Imaging and Volume Measures Are Associated With Disease Severity in Pediatric Niemann-Pick Disease Type C1. Pediatric Neurology, 2014, 51, 669-674.e5.	1.0	15
95	Transcriptome of HPÎ²CD-treated Niemann-Pick disease type C1 cells highlights GPNMB as a biomarker for therapeutics. Human Molecular Genetics, 2021, 30, 2456-2468.	1.4	15
96	Growth charts for individuals with Smithâ€“Lemliâ€“Opitz syndrome. American Journal of Medical Genetics, Part A, 2012, 158A, 2707-2713.	0.7	13
97	Cohort study of neurocognitive functioning and adaptive behaviour in children and adolescents with Niemannâ€Pick Disease type C1. Developmental Medicine and Child Neurology, 2016, 58, 262-269.	1.1	13
98	Fostering collaborative research for rare genetic disease: the example of niemann-pick type C disease. Orphanet Journal of Rare Diseases, 2016, 11, 161.	1.2	13
99	Mechanistic convergence and shared therapeutic targets in Niemannâ€Pick disease. Journal of Inherited Metabolic Disease, 2020, 43, 574-585.	1.7	13
100	Adrenal insufficiency and hypertension in a newborn infant with Smith-Lemli-Opitz syndrome. American Journal of Medical Genetics Part A, 2001, 103, 223-225.	2.4	12
101	Acute postnatal cataract formation in Smithâ€Lemliâ€Opitz syndrome. American Journal of Medical Genetics, Part A, 2008, 146A, 208-211.	0.7	11
102	Increasing cholesterol synthesis in 7-dehydrosterol reductase (DHCR7) deficient mouse models through gene transfer. Journal of Steroid Biochemistry and Molecular Biology, 2010, 122, 303-309.	1.2	11
103	Niemann-Pick Disease Type C: Implications for Sedation and Anesthesia for Diagnostic Procedures. Journal of Child Neurology, 2012, 27, 1541-1546.	0.7	11
104	Corpus Callosum Measurements Correlate With Developmental Delay in Smith-Lemli-Opitz Syndrome. Pediatric Neurology, 2013, 49, 107-112.	1.0	11
105	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.	0.5	11
106	Role of Diffusion Tensor Imaging in Prognostication and Treatment Monitoring in Niemann-Pick Disease Type C1. Diseases (Basel, Switzerland), 2016, 4, 29.	1.0	10
107	Gastrointestinal Tract Pathology in a BALB/c Niemannâ€“Pick Disease Type C1 Null Mouse Model. Digestive Diseases and Sciences, 2018, 63, 870-880.	1.1	10
108	Differential Proteomics Reveals miR-155 as a Novel Indicator of Liver and Spleen Pathology in the Symptomatic Niemann-Pick Disease, Type C1 Mouse Model. Molecules, 2019, 24, 994.	1.7	10

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109	Unbiased yeast screens identify cellular pathways affected in Niemannâ€“Pick disease type C. Life Science Alliance, 2020, 3, e201800253.	1.3	10
110	Defective platelet function in <scp>Niemannâ€Pick</scp> disease type <scp>C1</scp>. JIMD Reports, 2020, 56, 46-57.	0.7	9
111	Sterol and lipid analyses identifies hypolipidemia and apolipoprotein disorders in autism associated with adaptive functioning deficits. Translational Psychiatry, 2021, 11, 471.	2.4	9
112	Vitamin D levels in Smithâ€Lemliâ€Opitz syndrome. American Journal of Medical Genetics, Part A, 2017, 173, 2577-2583.	0.7	8
113	Statins for Smith-Lemli-Opitz syndrome. The Cochrane Library, 2020, 2020, .	1.5	8
114	X-linked creatine transporter deficiency results in prolonged QTc and increased sudden death risk in humans and disease model. Genetics in Medicine, 2021, 23, 1864-1872.	1.1	8
115	Hepatocellular carcinoma as a complication of Niemannâ€Pick disease type C1. American Journal of Medical Genetics, Part A, 2021, 185, 3111-3117.	0.7	8
116	Reduction of glutamate neurotoxicity: A novel therapeutic approach for Niemann-Pick disease, type C1. Molecular Genetics and Metabolism, 2021, 134, 330-336.	0.5	8
117	A somatic cell defect is associated with the onset of neurological symptoms in a lysosomal storage disease. Molecular Genetics and Metabolism, 2013, 110, 188-190.	0.5	7
118	Cholesterol Biosynthesis and Trafficking in Cortisol-Producing Lesions of the Adrenal Cortex. Journal of Clinical Endocrinology and Metabolism, 2015, 100, 3660-3667.	1.8	7
119	Normal IQ is possible in Smithâ€Lemliâ€Opitz syndrome. American Journal of Medical Genetics, Part A, 2017, 173, 2097-2100.	0.7	7
120	Evaluation of the Potential Role of Proprotein Convertase Subtilisin/Kexin Type 9 (PCSK9) in Niemannâ€“Pick Disease, Type C1. International Journal of Molecular Sciences, 2020, 21, 2430.	1.8	7
121	The role of Niemann-Pick type C2 in zebrafish embryonic development. Development (Cambridge), 2021, 148, dev.194258.	1.2	7
122	A human iPSC-derived inducible neuronal model of Niemann-Pick disease, type C1. BMC Biology, 2021, 19, 218.	1.7	7
123	Defining Natural History: Assessment of the Ability of College Students to Aid in Characterizing Clinical Progression of Niemann-Pick Disease, Type C. PLoS ONE, 2011, 6, e23666.	1.1	6
124	Neurodevelopmental Characterization of Young Children Diagnosed with Niemann-Pick Disease, Type C1. Journal of Developmental and Behavioral Pediatrics, 2020, 41, 388-396.	0.6	6
125	Glycerophosphoinositol is Elevated in Blood Samples From CLN3^{Î”ex7-8} pigs, Cln3^{Î”ex7-8} Mice, and CLN3-Affected Individuals. Biomarker Insights, 2022, 17, 117727192211077.	1.0	6
126	An Efficient Approach to Evaluate Reporter Ion Behavior from MALDI-MS/MS Data for Quantification Studies Using Isobaric Tags. Journal of Proteome Research, 2015, 14, 4169-4178.	1.8	5

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127	Maternal immune activation modifies the course of Niemann-pick disease, type C1 in a gender specific manner. Molecular Genetics and Metabolism, 2020, 129, 165-170.	0.5	5
128	Spontaneously regressing brain lesions in Smithâ€“Lemliâ€“Opitz syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 386-390.	0.7	4
129	An induced pluripotent stem cell line (TRNDi001-D) from a Niemann-Pick disease type C1 (NPC1) patient carrying a homozygous p. I1061T (c. 3182T>C) mutation in the NPC1 gene. Stem Cell Research, 2020, 44, 101737.	0.3	4
130	Oxidative phosphorylation in creatine transporter deficiency. NMR in Biomedicine, 2021, 34, e4419.	1.6	4
131	Phenotype assessment for neurodegenerative murine models with ataxia and application to Niemannâ€“Pick disease, type C1. Biology Open, 2022, 11, .	0.6	4
132	Identification of nine novelDHCR7 missense mutations in patients with Smith-Lemli-Opitz syndrome (SLOS). Human Mutation, 2005, 26, 59-59.	1.1	3
133	Association of NPC1 variant p.P237S with a pathogenic splice variant in two Niemannâ€“Pick disease type C1 patients. American Journal of Medical Genetics, Part A, 2017, 173, 1038-1040.	0.7	3
134	Auditory phenotype of <scp>Smithâ€“Lemliâ€“Opitz</scp> syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 1131-1141.	0.7	3
135	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.	1.6	3
136	Altered cerebrospinal fluid proteins in Smithâ€“Lemliâ€“Opitz syndrome patients. American Journal of Medical Genetics, Part A, 2016, 170, 2060-2068.	0.7	2
137	Gait, Balance, and Coordination Impairments in Niemann Pick Disease, Type C1. Journal of Child Neurology, 2018, 33, 114-124.	0.7	2
138	Toll-like receptor mediated lysozyme expression in Niemann-pick disease, type C1. Molecular Genetics and Metabolism, 2020, 131, 364-366.	0.5	2
139	Complex N-Linked Glycosylation: A Potential Modifier of Niemannâ€“Pick Disease, Type C1 Pathology. International Journal of Molecular Sciences, 2022, 23, 5082.	1.8	2
140	Consistently high agreement between independent raters of Niemann-Pick type C1 clinical severity scale in phase 2/3 trial. Pediatric Neurology, 2021, 127, 32-38.	1.0	1
141	Abnormal LAMP1 glycosylation may play a role in Niemann-Pick disease, type C pathology. , 2020, 15, e0227829.		0
142	Abnormal LAMP1 glycosylation may play a role in Niemann-Pick disease, type C pathology. , 2020, 15, e0227829.		0
143	Abnormal LAMP1 glycosylation may play a role in Niemann-Pick disease, type C pathology. , 2020, 15, e0227829.		0
144	Abnormal LAMP1 glycosylation may play a role in Niemann-Pick disease, type C pathology. , 2020, 15, e0227829.		0