Forbes D Porter

List of Publications by Year
in descending order

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144 papers	7,262 citations	47006 47 h-index	62596 80 g-index
145 all docs	145 docs citations	145 times ranked	6166 citing authors

#	Article	IF	Citations
1	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
2	Phenotype assessment for neurodegenerative murine models with ataxia and application to Niemannâ€“Pick disease, type C1. Biology Open, 2022, 11, .	1.2	4
3	Complex N-Linked Glycosylation: A Potential Modifier of Niemannâ€“Pick Disease, Type C1 Pathology. International Journal of Molecular Sciences, 2022, 23, 5082.	4.1	2
4	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.	2.5	6
5	Oxidative phosphorylation in creatine transporter deficiency. NMR in Biomedicine, 2021, 34, e4419.	2.8	4
6	Auditory phenotype of <scp>Smithâ€“Lemliâ€“Opitz</scp> syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 1131-1141.	1.2	3
7	The role of Niemann-Pick type C2 in zebrafish embryonic development. Development (Cambridge), 2021, 148, dev.194258.	2.5	7
8	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.	2.4	8
9	Hepatocellular carcinoma as a complication of Niemannâ€Pick disease type C1. American Journal of Medical Genetics, Part A, 2021, 185, 3111-3117.	1.2	8
10	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
11	Sterol and lipid analyses identifies hypolipidemia and apolipoprotein disorders in autism associated with adaptive functioning deficits. Translational Psychiatry, 2021, 11, 471.	4.8	9
12	A human iPSC-derived inducible neuronal model of Niemann-Pick disease, type C1. BMC Biology, 2021, 19, 218.	3.8	7
13	Reduction of glutamate neurotoxicity: A novel therapeutic approach for Niemann-Pick disease, type C1. Molecular Genetics and Metabolism, 2021, 134, 330-336.	1.1	8
14	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.	2.1	1
15	Mechanistic convergence and shared therapeutic targets in Niemannâ€Pick disease. Journal of Inherited Metabolic Disease, 2020, 43, 574-585.	3.6	13
16	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.	4.1	18
17	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
18	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.	1.1	5

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19	Neurodevelopmental Characterization of Young Children Diagnosed with Niemann-Pick Disease, Type C1. Journal of Developmental and Behavioral Pediatrics, 2020, 41, 388-396.	1.1	6
20	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
21	Prevalence of Diabetes and Hypertension and Their Associated Risks for Poor Outcomes in Covid-19 Patients. Journal of the Endocrine Society, 2020, 4, bvaa102.	0.2	56
22	Single Cell Transcriptome Analysis of Niemannâ€“Pick Disease, Type C1 Cerebella. International Journal of Molecular Sciences, 2020, 21, 5368.	4.1	20
23	Toll-like receptor mediated lysozyme expression in Niemann-pick disease, type C1. Molecular Genetics and Metabolism, 2020, 131, 364-366.	1.1	2
24	Association of Miglustat With Swallowing Outcomes in Niemann-Pick Disease, Type C1. JAMA Neurology, 2020, 77, 1564.	9.0	20
25	Defective platelet function in <scp>Niemannâ€Pick</scp> disease type <scp>C1</scp>. JIMD Reports, 2020, 56, 46-57.	1.5	9
26	Endocrine Conditions and COVID-19. Hormone and Metabolic Research, 2020, 52, 471-484.	1.5	34
27	Statins for Smith-Lemli-Opitz syndrome. The Cochrane Library, 2020, 2020, .	2.8	8
28	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.	0.7	4
29	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.	11.4	62
30	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.	4.1	7
31	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
32	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
33	Abnormal LAMP1 glycosylation may play a role in Niemann-Pick disease, type C pathology. PLoS ONE, 2020, 15, e0227829.	2.5	21
34	Unbiased yeast screens identify cellular pathways affected in Niemannâ€“Pick disease type C. Life Science Alliance, 2020, 3, e201800253.	2.8	10
35	Abnormal LAMP1 glycosylation may play a role in Niemann-Pick disease, type C pathology. , 2020, 15, e0227829.		0
36	Abnormal LAMP1 glycosylation may play a role in Niemann-Pick disease, type C pathology. , 2020, 15, e0227829.		0

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37	Abnormal LAMP1 glycosylation may play a role in Niemann-Pick disease, type C pathology. , 2020, 15, e0227829.		0
38	Abnormal LAMP1 glycosylation may play a role in Niemann-Pick disease, type C pathology. , 2020, 15, e0227829.		0
39	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
40	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.	5.9	28
41	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
42	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.	3.8	10
43	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.	1.1	22
44	Quantitating the epigenetic transformation contributing to cholesterol homeostasis using Gaussian process. Nature Communications, 2019, 10, 5052.	12.8	18
45	Unique molecular signature in mucolipidosis type IV microglia. Journal of Neuroinflammation, 2019, 16, 276.	7.2	17
46	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
47	Gastrointestinal Tract Pathology in a BALB/c Niemannâ€“Pick Disease Type C1 Null Mouse Model. Digestive Diseases and Sciences, 2018, 63, 870-880.	2.3	10
48	Gait, Balance, and Coordination Impairments in Niemann Pick Disease, Type C1. Journal of Child Neurology, 2018, 33, 114-124.	1.4	2
49	Long-Term Treatment of Niemann-Pick Type C1 Disease With Intrathecal 2-Hydroxypropyl-Î²-Cyclodextrin. Pediatric Neurology, 2018, 80, 24-34.	2.1	60
50	Microglia activation in Niemannâ€“Pick disease, type C1 is amendable to therapeutic intervention. Human Molecular Genetics, 2018, 27, 2076-2089.	2.9	54
51	Spontaneously regressing brain lesions in Smithâ€“Lemliâ€“Opitz syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 386-390.	1.2	4
52	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, .	2.4	38
53	Systemic AAV9 gene therapy improves the lifespan of mice with Niemann-Pick disease, type C1. Human Molecular Genetics, 2017, 26, ddw367.	2.9	50
54	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.	7.1	100

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55	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
56	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.	1.2	3
57	Normal IQ is possible in Smithâ€Lemliâ€Opitz syndrome. American Journal of Medical Genetics, Part A, 2017, 173, 2097-2100.	1.2	7
58	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
59	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
60	Vitamin D levels in Smithâ€Lemliâ€Opitz syndrome. American Journal of Medical Genetics, Part A, 2017, 173, 2577-2583.	1.2	8
61	A placebo-controlled trial of simvastatin therapy in Smith-Lemli-Opitz syndrome. Genetics in Medicine, 2017, 19, 297-305.	2.4	34
62	Role of Diffusion Tensor Imaging in Prognostication and Treatment Monitoring in Niemann-Pick Disease Type C1. Diseases (Basel, Switzerland), 2016, 4, 29.	2.5	10
63	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
64	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.	2.1	13
65	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.	4.5	39
66	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.	2.5	29
67	Development of a bile acidâ€“based newborn screen for Niemann-Pick disease type C. Science Translational Medicine, 2016, 8, 337ra63.	12.4	89
68	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
69	Altered cerebrospinal fluid proteins in Smithâ€“Lemliâ€“Opitz syndrome patients. American Journal of Medical Genetics, Part A, 2016, 170, 2060-2068.	1.2	2
70	Development, behavior, and biomarker characterization of Smith-Lemli-Opitz syndrome: an update. Journal of Neurodevelopmental Disorders, 2016, 8, 12.	3.1	45
71	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
72	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.	30.7	46

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73	Defective Cytochrome P450-Catalysed Drug Metabolism in Niemann-Pick Type C Disease. PLoS ONE, 2016, 11, e0152007.	2.5	22
74	A novel, highly sensitive and specific biomarker for Niemann-Pick type C1 disease. Orphanet Journal of Rare Diseases, 2015, 10, 78.	2.7	105
75	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.	3.3	48
76	Intrathecal 2-hydroxypropyl-beta-cyclodextrin in a single patient with Niemannâ€“Pick C1. Molecular Genetics and Metabolism, 2015, 116, 75-79.	1.1	76
77	Pathogenesis, epidemiology, diagnosis and clinical aspects of Smithâ€“Lemliâ€“Opitz syndrome. Expert Opinion on Orphan Drugs, 2015, 3, 267-280.	0.8	42
78	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
79	Cholesterol Biosynthesis and Trafficking in Cortisol-Producing Lesions of the Adrenal Cortex. Journal of Clinical Endocrinology and Metabolism, 2015, 100, 3660-3667.	3.6	7
80	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.	3.7	5
81	Variations in EEG discharges predict ADHD severity within individual Smith-Lemli-Opitz patients. Neurology, 2014, 83, 151-159.	1.1	15
82	Auditory Phenotype of Niemann-Pick Disease, Type C1. Ear and Hearing, 2014, 35, 110-117.	2.1	60
83	Human and mouse neuroinflammation markers in Niemannâ€Pick disease, type C1. Journal of Inherited Metabolic Disease, 2014, 37, 83-92.	3.6	71
84	Altered transition metal homeostasis in Niemannâ€“Pick disease, type C1. Metallomics, 2014, 6, 542-553.	2.4	26
85	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
86	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
87	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.	2.1	15
88	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
89	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.	1.8	24
90	Relative acidic compartment volume as a lysosomal storage disorderâ€“associated biomarker. Journal of Clinical Investigation, 2014, 124, 1320-1328.	8.2	63

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91	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.	2.1	108
92	Corpus Callosum Measurements Correlate With Developmental Delay in Smith-Lemli-Opitz Syndrome. Pediatric Neurology, 2013, 49, 107-112.	2.1	11
93	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.	1.1	7
94	Efficacy of N-acetylcysteine in phenotypic suppression of mouse models of Niemannâ€“Pick disease, type C1. Human Molecular Genetics, 2013, 22, 3508-3523.	2.9	27
95	Identification of Niemann-Pick C1 disease biomarkers through sphingolipid profiling. Journal of Lipid Research, 2013, 54, 2800-2814.	4.2	88
96	Microarray expression analysis and identification of serum biomarkers for Niemannâ€“Pick disease, type C1. Human Molecular Genetics, 2012, 21, 3632-3646.	2.9	84
97	Niemann-Pick Disease Type C: Implications for Sedation and Anesthesia for Diagnostic Procedures. Journal of Child Neurology, 2012, 27, 1541-1546.	1.4	11
98	Î´-Tocopherol Reduces Lipid Accumulation in Niemann-Pick Type C1 and Wolman Cholesterol Storage Disorders. Journal of Biological Chemistry, 2012, 287, 39349-39360.	3.4	107
99	Apolipoprotein E genotype and neurological disease onset in Niemannâ€“Pick disease, type C1. American Journal of Medical Genetics, Part A, 2012, 158A, 2775-2780.	1.2	34
100	Quantitative Proteomic Analysis of Niemann-Pick Disease, Type C1 Cerebellum Identifies Protein Biomarkers and Provides Pathological Insight. PLoS ONE, 2012, 7, e47845.	2.5	59
101	Growth charts for individuals with Smithâ€“Lemliâ€“Opitz syndrome. American Journal of Medical Genetics, Part A, 2012, 158A, 2707-2713.	1.2	13
102	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.	2.5	6
103	Miglustat Treatment May Reduce Cerebrospinal Fluid Levels of the Axonal Degeneration Marker Tau in Niemannâ€“Pick Type C. JIMD Reports, 2011, 3, 45-52.	1.5	25
104	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
105	Malformation syndromes caused by disorders of cholesterol synthesis. Journal of Lipid Research, 2011, 52, 6-34.	4.2	375
106	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.	1.7	145
107	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.	1.2	54
108	Discordant phenotype and sterol biochemistry in Smithâ€“Lemliâ€“Opitz syndrome. American Journal of Medical Genetics, Part A, 2010, 152A, 2094-2098.	1.2	17

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109	Activation of Rho GTPases in Smithâ€“Lemliâ€“Opitz syndrome: pathophysiological and clinical implications. Human Molecular Genetics, 2010, 19, 1347-1357.	2.9	42
110	Cholesterol Oxidation Products Are Sensitive and Specific Blood-Based Biomarkers for Niemann-Pick C1 Disease. Science Translational Medicine, 2010, 2, 56ra81.	12.4	302
111	Quantitative Proteomics Analysis of Inborn Errors of Cholesterol Synthesis. Molecular and Cellular Proteomics, 2010, 9, 1461-1475.	3.8	40
112	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.	2.5	11
113	Oxidative stress in Niemannâ€“Pick disease, type C. Molecular Genetics and Metabolism, 2010, 101, 214-218.	1.1	113
114	Acute postnatal cataract formation in Smithâ€Lemliâ€Opitz syndrome. American Journal of Medical Genetics, Part A, 2008, 146A, 208-211.	1.2	11
115	Smithâ€“Lemliâ€“Opitz syndrome: pathogenesis, diagnosis and management. European Journal of Human Genetics, 2008, 16, 535-541.	2.8	264
116	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.	2.9	63
117	HEM dysplasia and ichthyosis are likely laminopathies and not due to 3Î²-hydroxysterol Î”14-reductase deficiency. Human Molecular Genetics, 2007, 16, 1176-1187.	2.9	56
118	Cholesterol biosynthesis from birth to adulthood in a mouse model for 7-dehydrosterol reductase deficiency (Smithâ€“Lemliâ€“Opitz syndrome). Steroids, 2007, 72, 802-808.	1.8	27
119	Abnormal sterols in cholesterol-deficiency diseases cause secretory granule malformation and decreased membrane curvature. Journal of Cell Science, 2006, 119, 1876-1885.	2.0	84
120	Development and characterization of a hypomorphic Smithâ€“Lemliâ€“Opitz syndrome mouse model and efficacy of simvastatin therapy. Human Molecular Genetics, 2006, 15, 839-851.	2.9	67
121	Cholesterol deficiency in a mouse model of Smith-Lemli-Opitz syndrome reveals increased mast cell responsiveness. Journal of Experimental Medicine, 2006, 203, 1161-1171.	8.5	65
122	Cholesterol precursors and facial clefting. Journal of Clinical Investigation, 2006, 116, 2322-2325.	8.2	31
123	Identification of nine novelDHCR7 missense mutations in patients with Smith-Lemli-Opitz syndrome (SLOS). Human Mutation, 2005, 26, 59-59.	2.5	3
124	3Î²-Hydroxysterol Î”7-reductase and the Smithâ€“Lemliâ€“Opitz syndrome. Molecular Genetics and Metabolism, 2005, 84, 112-126.	1.1	92
125	Residual cholesterol synthesis and simvastatin induction of cholesterol synthesis in Smithâ€“Lemliâ€“Opitz syndrome fibroblasts. Molecular Genetics and Metabolism, 2005, 85, 96-107.	1.1	62
126	The implications of 7-dehydrosterol-7-reductase deficiency (Smithâ€“Lemliâ€“Opitz syndrome) to neurosteroid production. Steroids, 2004, 69, 51-60.	1.8	48

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127	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
128	A defective response to Hedgehog signaling in disorders of cholesterol biosynthesis. Nature Genetics, 2003, 33, 508-513.	21.4	363
129	27-Hydroxylation of 7- and 8-dehydrocholesterol in Smithâ€“Lemliâ€“Opitz syndrome: a novel metabolic pathway. Steroids, 2003, 68, 497-502.	1.8	45
130	Lathosterolosis: an inborn error of human and murine cholesterol synthesis due to lathosterol 5-desaturase deficiency. Human Molecular Genetics, 2003, 12, 1631-1641.	2.9	153
131	Human malformation syndromes due to inborn errors of cholesterol synthesis. Current Opinion in Pediatrics, 2003, 15, 607-613.	2.0	115
132	Cholesterol storage defect in RSH/Smithâ€“Lemliâ€“Opitz syndrome fibroblasts. Molecular Genetics and Metabolism, 2002, 75, 325-334.	1.1	52
133	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.	2.5	59
134	Malformation syndromes due to inborn errors of cholesterol synthesis. Journal of Clinical Investigation, 2002, 110, 715-724.	8.2	88
135	Malformation syndromes due to inborn errors of cholesterol synthesis. Journal of Clinical Investigation, 2002, 110, 715-724.	8.2	43
136	SMITH-LEMLI-OPITZ SYNDROME. Journal of the American Academy of Child and Adolescent Psychiatry, 2001, 40, 506-507.	0.5	21
137	Behavior phenotype in the RSH/Smith-Lemli-Opitz syndrome. American Journal of Medical Genetics Part A, 2001, 98, 191-200.	2.4	202
138	Incidence of Smith-Lemli-Opitz syndrome in Ontario, Canada. American Journal of Medical Genetics Part A, 2001, 102, 18-20.	2.4	45
139	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
140	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
141	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
142	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
143	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.	1.1	102
144	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.	6.2	405