

Robert P Erickson

List of Publications by Year in descending order

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164
papers

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117453

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73
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168
all docs

168
docs citations

168
times ranked

6599
citing authors

#	ARTICLE	IF	CITATIONS
1	A Polymorphism* in the 5' Flanking Region of the CD14 Gene Is Associated with Circulating Soluble CD14 Levels and with Total Serum Immunoglobulin E. <i>American Journal of Respiratory Cell and Molecular Biology</i> , 1999, 20, 976-983.	1.4	785
2	Mutations in FOXC2 (MFH-1), a Forkhead Family Transcription Factor, Are Responsible for the Hereditary Lymphedema-Distichiasis Syndrome. <i>American Journal of Human Genetics</i> , 2000, 67, 1382-1388.	2.6	549
3	De Novo Pathogenic SCN8A Mutation Identified by Whole-Genome Sequencing of a Family Quartet Affected by Infantile Epileptic Encephalopathy and SUDEP. <i>American Journal of Human Genetics</i> , 2012, 90, 502-510.	2.6	365
4	Homozygous HOXA1 mutations disrupt human brainstem, inner ear, cardiovascular and cognitive development. <i>Nature Genetics</i> , 2005, 37, 1035-1037.	9.4	267
5	Exome sequencing reveals new causal mutations in children with epileptic encephalopathies. <i>Epilepsia</i> , 2013, 54, 1270-1281.	2.6	250
6	Cyclodextrins in the treatment of a mouse model of Niemann-Pick C disease. <i>Life Sciences</i> , 2001, 70, 131-142.	2.0	180
7	Somatic gene mutation and human disease other than cancer: An update. <i>Mutation Research - Reviews in Mutation Research</i> , 2010, 705, 96-106.	2.4	167
8	Microarray-based CGH detects chromosomal mosaicism not revealed by conventional cytogenetics. <i>American Journal of Medical Genetics, Part A</i> , 2007, 143A, 1679-1686.	0.7	158
9	Transcription of paternal Y-linked genes in the human zygote as early as the pronucleate stage. <i>Zygote</i> , 1994, 2, 281-287.	0.5	135
10	A novel mouse model of Niemann-Pick type C disease carrying a D1005G-Npc1 mutation comparable to commonly observed human mutations. <i>Human Molecular Genetics</i> , 2012, 21, 730-750.	1.4	111
11	The clinical spectrum of homozygous HOXA1 mutations. <i>American Journal of Medical Genetics, Part A</i> , 2008, 146A, 1235-1240.	0.7	101
12	Somatic gene mutation and human disease other than cancer. <i>Mutation Research - Reviews in Mutation Research</i> , 2003, 543, 125-136.	2.4	99
13	Localization of the murine Niemann-Pick C1 protein to two distinct intracellular compartments. <i>Journal of Lipid Research</i> , 2000, 41, 673-687.	2.0	96
14	Mouse models of human genetic disease: Which mouse is more like a man?. <i>BioEssays</i> , 1996, 18, 993-998.	1.2	89
15	Clinical heterogeneity in lymphoedema-distichiasis with FOXC2 truncating mutations. <i>Journal of Medical Genetics</i> , 2001, 38, 761-766.	1.5	88
16	Astrocyte-only Npc1 reduces neuronal cholesterol and triples life span of Npc1 ^{-/-} mice. <i>Journal of Neuroscience Research</i> , 2008, 86, 2848-2856.	1.3	83
17	Navajo microvillous inclusion disease is due to a mutation in MYO5B. <i>American Journal of Medical Genetics, Part A</i> , 2008, 146A, 3117-3119.	0.7	82
18	Allopregnanolone treatment, both as a single injection or repetitively, delays demyelination and enhances survival of Niemann-Pick C mice. <i>Journal of Neuroscience Research</i> , 2005, 82, 811-821.	1.3	77

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19	Rescue of neurodegeneration in Niemann-Pick C mice by a prion-promoter-driven Npc1 cDNA transgene. <i>Human Molecular Genetics</i> , 2002, 11, 3107-3114.	1.4	73
20	Athabaskan brainstem dysgenesis syndrome. <i>American Journal of Medical Genetics Part A</i> , 2003, 120A, 169-173.	2.4	66
21	The pathogenesis of lysosomal storage disorders: beyond the engorgement of lysosomes to abnormal development and neuroinflammation. <i>Human Molecular Genetics</i> , 2018, 27, R119-R129.	1.4	59
22	Shortened primary cilium length and dysregulated Sonic hedgehog signaling in Niemann-Pick C1 disease. <i>Human Molecular Genetics</i> , 2017, 26, 2277-2289.	1.4	57
23	True hermaphroditism with partial duplication of chromosome 22 and without SRY. , 1999, 85, 2-4.		55
24	Altered Regulation of Hepatic Efflux Transporters Disrupts Acetaminophen Disposition in Pediatric Nonalcoholic Steatohepatitis. <i>Drug Metabolism and Disposition</i> , 2015, 43, 829-835.	1.7	55
25	A 122.5-Kilobase Deletion of the P Gene Underlies the High Prevalence of Oculocutaneous Albinism Type 2 in the Navajo Population. <i>American Journal of Human Genetics</i> , 2003, 72, 62-72.	2.6	53
26	Southwestern Athabaskan (Navajo and Apache) genetic diseases. <i>Genetics in Medicine</i> , 1999, 1, 151-157.	1.1	47
27	Studies on neuronal death in the mouse model of Niemann-Pick C disease. <i>Journal of Neuroscience Research</i> , 2002, 68, 738-744.	1.3	46
28	Changes in polyadenylation of lactate dehydrogenase-X mRNA during spermatogenesis in mice. <i>Molecular Reproduction and Development</i> , 1988, 1, 27-34.	1.0	44
29	Understanding Niemann-Pick type C disease: a fat problem. <i>Current Opinion in Neurology</i> , 2003, 16, 155-161.	1.8	43
30	Does sex determination start at conception?. <i>BioEssays</i> , 1997, 19, 1027-1032.	1.2	42
31	Variable presentation of Rothmund-Thomson syndrome. <i>American Journal of Medical Genetics Part A</i> , 2000, 95, 204-207.	2.4	41
32	Regional localization of sex-specific Bkm-related sequences on proximal chromosome 17 of mice. <i>Nature</i> , 1984, 310, 579-581.	13.7	40
33	Hepatic glucocorticoid receptors and the H α 2 locus. <i>Nature</i> , 1978, 275, 136-138.	13.7	39
34	Decreased <i>Npc1</i> Gene Dosage in Mice Is Associated With Weight Gain. <i>Obesity</i> , 2010, 18, 1457-1459.	1.5	38
35	Mice deleted for heart-type cytochrome c oxidase subunit 7a1 develop dilated cardiomyopathy. <i>Mitochondrion</i> , 2012, 12, 294-304.	1.6	37
36	Recent advances in the study of somatic mosaicism and diseases other than cancer. <i>Current Opinion in Genetics and Development</i> , 2014, 26, 73-78.	1.5	37

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37	Deletion mapping of the t complex of chromosome 17 of the mouse. <i>Nature</i> , 1978, 274, 163-164.	13.7	36
38	Absence of RECQL4 mutations in poikiloderma with neutropenia in Navajo and non-Navajo patients. <i>American Journal of Medical Genetics Part A</i> , 2003, 118A, 299-301.	2.4	36
39	Identification of a novel <i>C16orf57</i> mutation in Athabaskan patients with Poikiloderma with Neutropenia. <i>American Journal of Medical Genetics, Part A</i> , 2011, 155, 337-342.	0.7	34
40	Primary lymphoedema. <i>Nature Reviews Disease Primers</i> , 2021, 7, 77.	18.1	33
41	Evidence that the serological determinant of H ₂ Y antigen is carbohydrate. <i>Nature</i> , 1981, 290, 503-505.	13.7	32
42	Tamoxifen and vitamin E treatments delay symptoms in the mouse model of Niemann-Pick C. <i>Journal of Applied Genetics</i> , 2004, 45, 461-7.	1.0	32
43	Developmental delay in motor skill acquisition in Niemann-Pick C1 mice reveals abnormal cerebellar morphogenesis. <i>Acta Neuropathologica Communications</i> , 2016, 4, 94.	2.4	31
44	Segregation analyses and a genome-wide linkage search confirm genetic heterogeneity and suggest oligogenic inheritance in some Milroy congenital primary lymphedema families. <i>American Journal of Medical Genetics Part A</i> , 2001, 98, 303-312.	2.4	30
45	Further evidence for haploid gene expression during spermatogenesis: Heterogeneous, poly(A)-containing RNA is synthesized post-meiotically. <i>The Journal of Experimental Zoology</i> , 1980, 214, 13-19.	1.4	29
46	Increased Expression of Caveolin-1 in Heterozygous Niemann-Pick Type II Human Fibroblasts. <i>Biochemical and Biophysical Research Communications</i> , 1997, 236, 189-193.	1.0	29
47	Mannosidosis: two brothers with different degrees of disease severity. <i>Clinical Genetics</i> , 1981, 20, 191-202.	1.0	29
48	Variation in NPC1, the gene encoding Niemann-Pick C1, a protein involved in intracellular cholesterol transport, is associated with Alzheimer disease and/or aging in the Polish population. <i>Neuroscience Letters</i> , 2008, 447, 153-157.	1.0	29
49	Niemann-Pick C1 Mice, a Model of Juvenile Alzheimer's Disease, with Normal Gene Expression in Neurons and Fibrillary Astrocytes Show Long Term Survival and Delayed Neurodegeneration. <i>Journal of Alzheimer's Disease</i> , 2012, 30, 875-887.	1.2	29
50	The separation of lactate dehydrogenase X from other lactate dehydrogenase isozymes of mouse testes by affinity chromatography. <i>FEBS Letters</i> , 1973, 35, 19-23.	1.3	28
51	Liver disease with altered bile acid transport in Niemann-Pick C mice on a high-fat, 1% cholesterol diet. <i>American Journal of Physiology - Renal Physiology</i> , 2005, 289, G300-G307.	1.6	25
52	Neural stem cell implantation extends life in Niemann-Pick C1 mice. <i>Journal of Applied Genetics</i> , 2007, 48, 269-272.	1.0	25
53	A familial form of convulsive disorder with or without mental retardation limited to females: extension of a pedigree limits possible genetic mechanisms. <i>Clinical Genetics</i> , 1990, 38, 353-358.	1.0	25
54	Pulmonary function and pathology in hydroxypropyl-beta-cyclodextrin-treated and untreated <i>Npc1</i> mice. <i>Molecular Genetics and Metabolism</i> , 2011, 103, 142-147.	0.5	25

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55	Expression of Niemann-Pick type C transcript in rodent cerebellum in vivo and in vitro. <i>Brain Research</i> , 1999, 839, 49-57.	1.1	24
56	Diffusion Tensor Imaging in Niemann-Pick Type C Disease. <i>Pediatric Neurology</i> , 2005, 33, 325-330.	1.0	24
57	Current controversies in Niemann-Pick C1 disease: steroids or gangliosides; neurons or neurons and glia. <i>Journal of Applied Genetics</i> , 2013, 54, 215-224.	1.0	23
58	Visual evoked potentials of Niemann-Pick type C1 mice reveal an impairment of the visual pathway that is rescued by 2-hydroxypropyl- β -cyclodextrin. <i>Orphanet Journal of Rare Diseases</i> , 2015, 10, 133.	1.2	21
59	Understanding Niemann-Pick type C disease: a fat problem. <i>Current Opinion in Neurology</i> , 2003, 16, 155-161.	1.8	21
60	Electron microscopy of t-allele synaptonemal complexes discloses no inversions. <i>Nature</i> , 1982, 299, 752-754.	13.7	20
61	Creating a conditional mutation of Wnt-1 by antisense transgenesis provides evidence that Wnt-1 is not essential for spermatogenesis. <i>Genesis</i> , 1993, 14, 274-281.	3.1	20
62	Molecular analysis in true hermaphrodites with different karyotypes and similar phenotypes. , 1996, 63, 348-355.		20
63	GENETIC DIFFERENCES AMONG THE A/J x C57BL/6J RECOMBINANT INBRED MOUSE LINES AND THEIR DEGREE OF ASSOCIATION WITH GLUCOCORTICOID-INDUCED CLEFT PALATE. <i>Genetics</i> , 1986, 113, 745-754.	1.2	20
64	Female pseudohermaphroditism with multiple caudal anomalies: Absence of Y-specific DNA sequences as pathogenetic factors. <i>American Journal of Medical Genetics Part A</i> , 1994, 51, 16-21.	2.4	19
65	Agnesis of tibia with bifid femur, congenital heart disease, and cleft lip with cleft palate or tracheoesophageal fistula: Possible variants of Gollop-Wolfgang complex. <i>American Journal of Medical Genetics, Part A</i> , 2005, 134A, 315-317.	0.7	19
66	MRI detects therapeutic effects in weanling Niemann-Pick type C mice. <i>Journal of Neuroscience Research</i> , 2008, 86, 2802-2807.	1.3	19
67	Apparently changing patterns of inheritance in Alport's hereditary nephritis: Genetic heterogeneity versus altered diagnostic criteria. <i>Clinical Genetics</i> , 1980, 17, 285-292.	1.0	19
68	Pediatric Cytochrome P450 Activity Alterations in Nonalcoholic Steatohepatitis. <i>Drug Metabolism and Disposition</i> , 2017, 45, 1317-1325.	1.7	19
69	Sex-limited penetrance of lymphedema to females with <i>CELSR1</i> haploinsufficiency: A second family. <i>Clinical Genetics</i> , 2019, 96, 478-482.	1.0	19
70	Lack of efficacy of curcumin on neurodegeneration in the mouse model of Niemann-Pick C1. <i>Pharmacology Biochemistry and Behavior</i> , 2012, 101, 125-131.	1.3	18
71	Immunological relatedness of two isozymes of 3-phosphoglycerate kinase from the mouse. <i>FEBS Letters</i> , 1978, 95, 371-374.	1.3	17
72	Phenotypic heterogeneity in cystic fibrosis. <i>American Journal of Medical Genetics Part A</i> , 1982, 13, 179-195.	2.4	17

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73	Magnetization transfer contrast imaging in Niemann pick type C mouse liver. <i>Journal of Magnetic Resonance Imaging</i> , 2003, 18, 321-327.	1.9	17
74	Mosaic tetrasomy 12p with triplication of 12p detected by array-based comparative genomic hybridization of peripheral blood DNA. <i>American Journal of Medical Genetics, Part A</i> , 2007, 143A, 2910-2915.	0.7	17
75	Interactions of Npc1 and amyloid accumulation/deposition in the APP/PS1 mouse model of Alzheimer's. <i>Journal of Applied Genetics</i> , 2011, 52, 213-218.	1.0	17
76	In vivo assessment of neurodegeneration in niemann-pick type C mice by quantitative T2 mapping and diffusion tensor imaging. <i>Journal of Magnetic Resonance Imaging</i> , 2012, 35, 528-536.	1.9	17
77	In Niemann-Pick C1 mouse models, glial-only expression of the normal gene extends survival much further than do changes in genetic background or treatment with hydroxypropyl-beta-cyclodextrin. <i>Gene</i> , 2018, 643, 117-123.	1.0	17
78	Genetic aspects of the effects of methylmercury in mice: The incidence of cleft palate and concentrations of adenosine 3'-5' cyclic monophosphate in tongue and palatal shelf. <i>Teratology</i> , 1981, 23, 397-401.	1.8	16
79	A modifier of Niemann Pick C 1 maps to mouse Chromosome 19. <i>Mammalian Genome</i> , 2000, 11, 69-71.	1.0	16
80	Axenfeld-Rieger anomaly, hypertelorism, clinodactyly, and cardiac anomalies in sibs with an unbalanced translocation der(6)t(6;8). <i>American Journal of Medical Genetics Part A</i> , 2001, 100, 187-190.	2.4	16
81	Confirmation of the role of N-acetyltransferase 2 in teratogen-induced cleft palate using transgenics and knockouts. <i>Molecular Reproduction and Development</i> , 2008, 75, 1071-1076.	1.0	16
82	Amelioration of enteric neuropathology in a mouse model of Niemann-Pick C by Npc1 expression in enteric glia. <i>Journal of Neuroscience Research</i> , 2009, 87, 2994-3001.	1.3	16
83	Dysregulation of Testicular Cholesterol Metabolism Following Spontaneous Mutation of the Niemann-Pick C1 Gene in Mice1. <i>Biology of Reproduction</i> , 2014, 91, 42.	1.2	16
84	The endogenous heat-stable glucocorticoid receptor stabilizing factor and the H-2 locus. <i>The Journal of Steroid Biochemistry</i> , 1982, 17, 121-123.	1.3	15
85	Re-evaluation of new X-linked syndrome for evidence of CHARGE syndrome or association. <i>American Journal of Medical Genetics Part A</i> , 1989, 34, 397-400.	2.4	15
86	Erythrocytic nicotinamide-adenine dinucleotide phosphate levels and the genetic regulation of erythrocytic glucose 6-phosphate dehydrogenase activity in the inbred mouse. <i>Biochemical Genetics</i> , 1974, 11, 33-40.	0.8	14
87	N-acetyltransferase 2 activity and folate levels. <i>Life Sciences</i> , 2010, 86, 103-106.	2.0	14
88	Distribution and Diffusion of Macromolecule Delivery to the Brain via Focused Ultrasound using Magnetic Resonance and Multispectral Fluorescence Imaging. <i>Ultrasound in Medicine and Biology</i> , 2020, 46, 122-136.	0.7	14
89	IDENTIFICATION OF AN AUTOSOMAL LOCUS AFFECTING STEROID SULFATASE ACTIVITY AMONG INBRED STRAINS OF MICE. <i>Genetics</i> , 1983, 105, 181-189.	1.2	14
90	Variation among inbred strains of mice in adenosine 3'-5' cyclic monophosphate levels of spermatozoa. <i>Genetical Research</i> , 1979, 33, 129-136.	0.3	13

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91	Post-meiotic transcription of phosphoglycerate-kinase 2 in mouse testes. <i>Bioscience Reports</i> , 1985, 5, 1087-1091.	1.1	13
92	The use of antisense approaches to study development. <i>Genesis</i> , 1993, 14, 251-257.	3.1	13
93	Gene expression, X-inactivation, and methylation during spermatogenesis: The case of Zfa, Zfx, and Zfy in mice. <i>Molecular Reproduction and Development</i> , 1993, 35, 114-120.	1.0	13
94	Mutagenesis and human genetic disease: An introduction. <i>Environmental and Molecular Mutagenesis</i> , 1995, 25, 2-6.	0.9	13
95	From "magic bullet" to "especially engineered shotgun loads" the new genetics and the need for individualized pharmacotherapy. <i>BioEssays</i> , 1998, 20, 683-685.	1.2	13
96	Fine linkage and physical mapping suggests cross-over suppression with a retroposon insertion at the npc1 mutation. <i>Mammalian Genome</i> , 2000, 11, 774-778.	1.0	13
97	mdr1a deficiency corrects sterility in Niemann-Pick C1 protein deficient female mice. <i>Molecular Reproduction and Development</i> , 2002, 62, 167-173.	1.0	13
98	The effects of genetic variation in N-acetyltransferases on 4-aminobiphenyl genotoxicity in mouse liver. <i>Chemico-Biological Interactions</i> , 2003, 146, 51-60.	1.7	13
99	SRY alone can induce normal male sexual differentiation. <i>American Journal of Medical Genetics Part A</i> , 1995, 55, 356-358.	2.4	12
100	Molecular and clinical studies of three cases of female pseudohermaphroditism with caudal dysplasia suggest multiple etiologies. <i>Clinical Genetics</i> , 1997, 51, 331-337.	1.0	12
101	Susceptibility to phenytoin-induced cleft lip with or without cleft palate: many genes are involved. <i>Genetical Research</i> , 1987, 49, 43-49.	0.3	11
102	Genetics of susceptibility to 6-aminonicotinamide-induced cleft palate in the mouse: Studies in congenic and recombinant inbred strains. <i>Teratology</i> , 1988, 37, 283-287.	1.8	11
103	Genes, Environment, and Orofacial Clefting. <i>Journal of Craniofacial Surgery</i> , 2010, 21, 1384-1387.	0.3	11
104	Does chromosome 22 have anything to do with sex determination: Further studies on a 46,XX,22q11.2 del male. <i>American Journal of Medical Genetics Part A</i> , 2003, 123A, 64-67.	2.4	10
105	The role of multiple drug resistance proteins in fetal and/or placental protection against teratogen-induced orofacial clefting. <i>Molecular Reproduction and Development</i> , 2007, 74, 1483-1489.	1.0	10
106	Autosomal recessive diseases among the Athabaskans of the Southwestern United States: Recent advances and implications for the future. <i>American Journal of Medical Genetics, Part A</i> , 2009, 149A, 2602-2611.	0.7	10
107	Genetic variation in the mouse model of Niemann Pick C1 affects female, as well as male, adiposity, and hepatic bile transporters but has indeterminate effects on caveolae. <i>Gene</i> , 2012, 491, 128-134.	1.0	10
108	The low frequency of recessive disease: insights from ENU mutagenesis, severity of disease phenotype, GWAS associations, and demography: an analytical review. <i>Journal of Applied Genetics</i> , 2014, 55, 319-327.	1.0	10

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109	Orofacial-digital syndrome IX with severe microcephaly: A new variant in a genetically isolated population. <i>American Journal of Medical Genetics, Part A</i> , 2007, 143A, 3309-3313.	0.7	9
110	A five generation family with a novel mutation in FOXC2 and lymphedema worsening to hydrops in the youngest generation. <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 2802-2807.	0.7	9
111	GENETICS OF GLUCOCORTICOID RECEPTOR LEVELS IN RECOMBINANT INBRED LINES OF MICE. <i>Genetics</i> , 1986, 113, 735-744.	1.2	9
112	Quantitative magnetic resonance imaging of brain atrophy in a mouse model of Niemann-Pick type C disease. <i>PLoS ONE</i> , 2017, 12, e0178179.	1.1	9
113	A family with unusual Waardenburg syndrome type I (WSI), cleft lip (palate), and Hirschsprung disease is not linked to <i>PAX3</i> . <i>Clinical Genetics</i> , 1995, 47, 139-143.	1.0	8
114	The role of decreased levels of Niemann-Pick C1 intracellular cholesterol transport on obesity is reversed in the C57BL/6J, metabolic syndrome mouse strain: a metabolic or an inflammatory effect?. <i>Journal of Applied Genetics</i> , 2012, 53, 323-330.	1.0	8
115	Species variation in the testicular angiotensin converting enzyme promoter studied in transgenic mice. <i>Molecular Reproduction and Development</i> , 1996, 44, 324-331.	1.0	7
116	Ascertainment and mutational studies of SRY in nine XY females. , 1999, 83, 138-139.		7
117	A patient with 22q11.2 deletion and Opitz syndrome-like phenotype has the same deletion as velocardiofacial patients. <i>American Journal of Medical Genetics, Part A</i> , 2007, 143A, 3302-3308.	0.7	7
118	An N-ethyl-N-nitrosourea-induced mutation in N-acetyltransferase 1 in mice. <i>Biochemical and Biophysical Research Communications</i> , 2008, 370, 285-288.	1.0	7
119	The importance of de novo mutations for pediatric neurological disease "It is not all in utero or birth trauma. <i>Mutation Research - Reviews in Mutation Research</i> , 2016, 767, 42-58.	2.4	7
120	A single base pair substitution within the paired box of PAX3 in an individual with Waardenburg syndrome type 1 (WS1). <i>Human Mutation</i> , 1994, 4, 227-228.	1.1	6
121	A first therapy for Niemann-Pick C. <i>Lancet Neurology, The</i> , 2007, 6, 748-749.	4.9	6
122	Insulin receptor-related (Irr) is expressed in pre-implantation embryos: A possible relationship to ð growth factor Y and sex determination. <i>Molecular Reproduction and Development</i> , 2011, 78, 552-552.	1.0	6
123	Extensive macrophage accumulation in young and old Niemann-Pick C1 model mice involves the alternative, M2, activation pathway and inhibition of macrophage apoptosis. <i>Gene</i> , 2016, 578, 242-250.	1.0	6
124	A hopeful therapy for Niemann-Pick C diseases. <i>Lancet, The</i> , 2017, 390, 1720-1721.	6.3	6
125	A pilot study of direct delivery of hydroxypropyl-beta-cyclodextrin to the lung by the nasal route in a mouse model of Niemann-Pick C1 disease: motor performance is unaltered and lung disease is worsened. <i>Journal of Applied Genetics</i> , 2018, 59, 187-191.	1.0	6
126	The Cerebellum in Niemann-Pick C1 Disease: Mouse Versus Man. <i>Cerebellum</i> , 2023, 22, 102-119.	1.4	6

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127	Ectopic expression of chloramphenicol acetyltransferase (CAT) in the cerebellum in mice transgenic for a carbonic anhydrase II promoter-CAT construct that is without apparent phenotypic effect. <i>Molecular Reproduction and Development</i> , 1990, 27, 102-109.	1.0	5
128	Use of a probe for the putative sex determining gene, Zinc finger Y, in the study of patients with ambiguous genitalia and XY gonadal dysgenesis. <i>American Journal of Medical Genetics Part A</i> , 1990, 36, 232-236.	2.4	5
129	Correlation of phenotypic and genetic heterogeneity in cystic fibrosis: Variability in sweat electrolyte levels contributes to heterogeneity and is increased with the XV-2c/KM19 B haplotype. <i>American Journal of Medical Genetics Part A</i> , 1991, 39, 137-143.	2.4	5
130	Sites of transcription of the M β 1/4llerian inhibiting substance gene in mouse testis. <i>Molecular Reproduction and Development</i> , 1993, 35, 159-164.	1.0	5
131	Cholesterol Signaling at the Endoplasmic Reticulum Occurs in npc1 Δ but Not in npc1 Δ , LDLR Δ Mice. <i>Biochemical and Biophysical Research Communications</i> , 2001, 284, 326-330.	1.0	5
132	Correlation of susceptibility to 6-aminonicotinamide and hydrocortisone-induced cleft palate. <i>Life Sciences</i> , 2005, 76, 2071-2078.	2.0	5
133	Crowd-Sourcing Syncope Diagnosis: Mobile Smartphone ECG Apps. <i>American Journal of Medicine</i> , 2016, 129, e17-e18.	0.6	5
134	Relative efficacy of nicotinamide treatment of a mouse model of infantile Niemann-Pick C1 disease. <i>Journal of Applied Genetics</i> , 2017, 58, 99-102.	1.0	5
135	Ionic effects on strain differences in hepatic cytosolic glucocorticoid receptor levels in mice. <i>Teratology</i> , 1983, 27, 43-49.	1.8	4
136	Genetic variation in β -adrenergic receptors in mice: A magnesium effect determined by a single gene. <i>Genetical Research</i> , 1983, 42, 159-168.	0.3	4
137	¹ H magnetic resonance spectroscopy of neurodegeneration in a mouse model of niemann-pick type C1 disease. <i>Journal of Magnetic Resonance Imaging</i> , 2013, 37, 1195-1201.	1.9	4
138	Decreased membrane cholesterol in liver mitochondria of the point mutation mouse model of juvenile Niemann-Pick C1, Npc1. <i>Mitochondrion</i> , 2020, 51, 15-21.	1.6	4
139	Cattanach's translocation [Is(7:1>X</i>)Ct] corrects male sterility due to homozygosity for chromosome 7 deletions. <i>Genetical Research</i> , 1984, 43, 35-41.	0.3	3
140	Co-treatment with probucol does not improve lung pathology in hydroxypropyl- β -cyclodextrin-treated Npc1 Δ mice. <i>Journal of Applied Genetics</i> , 2019, 60, 175-178.	1.0	3
141	Haploinsufficiency of tau decreases survival of the mouse model of Niemann-Pick disease type C1 but does not alter tau phosphorylation. <i>Journal of Applied Genetics</i> , 2020, 61, 567-570.	1.0	3
142	Inheritance of the Sex-Determining Factor in the Absence of a Complete Y Chromosome in 46,XX Human Males. <i>Annals of the New York Academy of Sciences</i> , 1987, 513, 505-506.	1.8	2
143	Northern analyses using single-stranded probes do not support a role for GATA/GACA repeats in sex determination in mice and men. <i>Molecular Reproduction and Development</i> , 1989, 1, 116-121.	1.0	2
144	Recent advances in developmental genetics: Growth factors and morphogens. <i>Molecular Reproduction and Development</i> , 1995, 41, 109-125.	1.0	2

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145	Introductory comments: M. Michael Cohen Jr. Festschrift. American Journal of Medical Genetics, Part A, 2007, 143A, 2851-2852.	0.7	2
146	Size of 22q deletions in four previously reported patients with conotruncal anomaly face syndrome. Clinical Genetics, 1996, 50, 545-547.	1.0	2
147	Do GWAS and studies of heterozygotes for NPC1 and/or NPC2 explain why NPC disease cases are so rare?. Journal of Applied Genetics, 2018, 59, 441-447.	1.0	2
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