Neal Sondheimer

List of Publications by Year
in descending order

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70	3,407	201674	149698
papers	citations	27	56
		h-index	g-index

78	78	78	5509
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Rnq1. Molecular Cell, 2000, 5, 163-172.	9.7	492
2	Improved diagnostic yield compared with targeted gene sequencing panels suggests a role for whole-genome sequencing as a first-tier genetic test. Genetics in Medicine, 2018, 20, 435-443.	2.4	404
3	Polyglutamine aggregates alter protein folding homeostasis in Caenorhabditis elegans. Proceedings of the National Academy of Sciences of the United States of America, 2000, 97, 5750-5755.	7.1	358
4	The role of Sis1 in the maintenance of the [RNQ+] prion. EMBO Journal, 2001, 20, 2435-2442.	7.8	188
5	Mutations in FBXL4, Encoding a Mitochondrial Protein, Cause Early-Onset Mitochondrial Encephalomyopathy. American Journal of Human Genetics, 2013, 93, 482-495.	6.2	138
6	Changes in the middle region of Sup35 profoundly alter the nature of epigenetic inheritance for the yeast prion [PSI ⁺]. Proceedings of the National Academy of Sciences of the United States of America, 2002, 99, 16446-16453.	7.1	133
7	Modeling kinetic rate variation in third generation DNA sequencing data to detect putative modifications to DNA bases. Genome Research, 2013, 23, 129-141.	5.5	99
8	Predicting the pathogenicity of novel variants in mitochondrial tRNA with MitoTIP. PLoS Computational Biology, 2017, 13, e1005867.	3.2	93
9	Red Blood Cells Homeostatically Bind Mitochondrial DNA through TLR9 to Maintain Quiescence and to Prevent Lung Injury. American Journal of Respiratory and Critical Care Medicine, 2018, 197, 470-480.	5.6	90
10	Periodic reanalysis of whole-genome sequencing data enhances the diagnostic advantage over standard clinical genetic testing. European Journal of Human Genetics, 2018, 26, 740-744.	2.8	88
11	Specifications of the ACMG/AMP standards and guidelines for mitochondrial DNA variant interpretation. Human Mutation, 2020, 41, 2028-2057.	2.5	84
12	Neutral mitochondrial heteroplasmy and the influence of aging. Human Molecular Genetics, 2011, 20, 1653-1659.	2.9	82
13	High-dose continuous renal replacement therapy for neonatal hyperammonemia. Pediatric Nephrology, 2013, 28, 983-986.	1.7	68
14	G-quadruplex dynamics contribute to regulation of mitochondrial gene expression. Scientific Reports, 2019, 9, 5605.	3.3	65
15	The Personal Genome Project Canada: findings from whole genome sequences of the inaugural 56 participants. Cmaj, 2018, 190, E126-E136.	2.0	57
16	A Distinctive Physiological Role for IÎºBÎ² in the Propagation of Mitochondrial Respiratory Stress Signaling. Journal of Biological Chemistry, 2008, 283, 12586-12594.	3.4	56
17	The mitochondrial phosphate carrier: Role in oxidative metabolism, calcium handling and mitochondrial disease. Biochemical and Biophysical Research Communications, 2015, 464, 369-375.	2.1	52
18	Mitochondrial respiratory chain disease discrimination by retrospective cohort analysis of blood metabolites. Molecular Genetics and Metabolism, 2013, 110, 145-152.	1.1	49

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19	Autosomal-Recessive Mutations in AP3B2, Adaptor-Related Protein Complex 3 Beta 2 Subunit, Cause an Early-Onset Epileptic Encephalopathy with Optic Atrophy. American Journal of Human Genetics, 2016, 99, 1368-1376.	6.2	46
20	Transcriptional requirements of the distal heavy-strand promoter of mtDNA. Proceedings of the National Academy of Sciences of the United States of America, 2012, 109, 6508-6512.	7.1	45
21	Role of calcineurin, hnRNPA2 and Akt in mitochondrial respiratory stress-mediated transcription activation of nuclear gene targets. Biochimica Et Biophysica Acta - Bioenergetics, 2010, 1797, 1055-1065.	1.0	44
22	Mitochondrial genetic diseases. Current Opinion in Pediatrics, 2010, 22, 711-716.	2.0	41
23	Leucine-Rich Pentatricopeptide-Repeat Containing Protein Regulates Mitochondrial Transcription. Biochemistry, 2010, 49, 7467-7473.	2.5	40
24	Precision therapy for a new disorder of AMPA receptor recycling due to mutations in ATAD1. Neurology: Genetics, 2017, 3, e130.	1.9	40
25	Investigating protein conformationâ€“based inheritance and disease in yeast. Philosophical Transactions of the Royal Society B: Biological Sciences, 2001, 356, 169-176.	4.0	37
26	Mutation in the mitochondrial tRNAVal causes mitochondrial encephalopathy, lactic acidosis and stroke-like episodes. Mitochondrion, 2011, 11, 615-619.	3.4	33
27	Pathologic Variants of the Mitochondrial Phosphate Carrier SLC25A3: Two New Patients and Expansion of the Cardiomyopathy/Skeletal Myopathy Phenotype With and Without Lactic Acidosis. JIMD Reports, 2014, 19, 59-66.	1.5	31
28	Mitochondrial tRNAPhe mutation as a cause of end-stage renal disease in childhood. Pediatric Nephrology, 2013, 28, 515-519.	1.7	29
29	The value of the metabolic autopsy in the pediatric hospital setting. Journal of Pediatrics, 2006, 148, 779-783.	1.8	27
30	Novel recessive mutations in COQ4 cause severe infantile cardiomyopathy and encephalopathy associated with CoQ 10 deficiency. Molecular Genetics and Metabolism Reports, 2017, 12, 23-27.	1.1	27
31	Complex management of a patient with a contiguous Xp11.4 gene deletion involving ornithine transcarbamylase: A role for detailed molecular analysis in complex presentations of classical diseases. Molecular Genetics and Metabolism, 2008, 94, 498-502.	1.1	25
32	DNM1L Variant Alters Baseline Mitochondrial Function and Response to Stress in a Patient with Severe Neurological Dysfunction. Biochemical Genetics, 2018, 56, 56-77.	1.7	24
33	Hepatic oxidant injury and glutathione depletion during total parenteral nutrition in weanling rats. American Journal of Physiology - Renal Physiology, 1996, 270, G691-G700.	3.4	20
34	DNAJC12-associated developmental delay, movement disorder, and mild hyperphenylalaninemia identified by whole-exome sequencing re-analysis. European Journal of Human Genetics, 2018, 26, 1867-1870.	2.8	19
35	Natural and Induced Mitochondrial Phosphate Carrier Loss. Journal of Biological Chemistry, 2016, 291, 26126-26137.	3.4	18
36	Divergent Patterns of Mitochondrial and Nuclear Ancestry Are Associated with the Risk for Preterm Birth. Journal of Pediatrics, 2018, 194, 40-46.e4.	1.8	18

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37	Contribution of a mitochondrial tyrosyl-tRNA synthetase mutation to the phenotypic expression of the deafness-associated tRNASer(UCN) 7511A>G mutation. Journal of Biological Chemistry, 2019, 294, 19292-19305.	3.4	17
38	Pyruvate carboxylase deficiency type A and type C: Characterization of five novel pathogenic variants inPCand analysis of the genotypeâ€“phenotype correlation. Human Mutation, 2019, 40, 816-827.	2.5	16
39	Characterization of mitochondrial health from human peripheral blood mononuclear cells to cerebral organoids derived from induced pluripotent stem cells. Scientific Reports, 2021, 11, 4523.	3.3	16
40	Nuclear genome-wide associations with mitochondrial heteroplasmy. Science Advances, 2021, 7, .	10.3	16
41	Heteroplasmy Shifting as Therapy for Mitochondrial Disorders. Advances in Experimental Medicine and Biology, 2019, 1158, 257-267.	1.6	16
42	Lack of relationship between mitochondrial heteroplasmy or variation and childhood obesity. International Journal of Obesity, 2012, 36, 80-83.	3.4	14
43	G-quadruplex-mediated reduction of a pathogenic mitochondrial heteroplasmy. Human Molecular Genetics, 2019, 28, 3163-3174.	2.9	14
44	A pathogenic UFSP2 variant in an autosomal recessive form of pediatric neurodevelopmental anomalies and epilepsy. Genetics in Medicine, 2021, 23, 900-908.	2.4	14
45	Homozygous pathogenic variant in BRAT1 associated with nonprogressive cerebellar ataxia. Neurology: Genetics, 2019, 5, e359.	1.9	13
46	<scp>EGFR</scp> mutations cause a lethal syndrome of epithelial dysfunction with progeroid features. Molecular Genetics & Genomic Medicine, 2015, 3, 452-458.	1.2	12
47	Higher Order Organization of the mtDNA: Beyond Mitochondrial Transcription Factor A. Frontiers in Genetics, 2019, 10, 1285.	2.3	12
48	Improving surveillance for hyperammonemia in the newborn. Molecular Genetics and Metabolism, 2013, 110, 102-105.	1.1	11
49	Evaluation of the quality of clinical data collection for a pan-Canadian cohort of children affected by inherited metabolic diseases: lessons learned from the Canadian Inherited Metabolic Diseases Research Network. Orphanet Journal of Rare Diseases, 2020, 15, 89.	2.7	11
50	Topological requirements of the mitochondrial heavy-strand promoters. Transcription, 2017, 8, 307-312.	3.1	10
51	Increased C3-Carnitine in a Healthy Premature Infant. Clinical Chemistry, 2008, 54, 1914-1917.	3.2	9
52	Health Care for Mitochondrial Disorders in Canada: A Survey of Physicians. Canadian Journal of Neurological Sciences, 2019, 46, 717-726.	0.5	6
53	Mitochondrial DNA, nuclear context, and the risk for carcinogenesis. Environmental and Molecular Mutagenesis, 2019, 60, 455-462.	2.2	6
54	Analysis of cerebrospinal fluid mitochondrial DNA levels in Alzheimer disease. Annals of Neurology, 2014, 75, 458-460.	5.3	5

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55	Homozygous GLUL deletion is embryonically viable and leads to glutamine synthetase deficiency. Clinical Genetics, 2020, 98, 613-619.	2.0	5
56	Reversal of Stroke-Like Episodes With L-Arginine and Meticulous Perioperative Management of Renal Transplantation in a Patient With Mitochondrial Encephalomyopathy, Lactic Acidosis and Stroke-Like Episodes (MELAS) Syndrome. Case Report. Neurohospitalist, The, 2022, 12, 67-73.	0.8	4
57	An Infant Refugee with Anemia and Low Serum Vitamin B12. Clinical Chemistry, 2018, 64, 1567-1570.	3.2	3
58	Liver transplantation for Gaucher disease presenting as neonatal cholestasis: Case report and literature review. Pediatric Transplantation, 2020, 24, e13718.	1.0	3
59	Kidney Transplantation From a Deceased Donor With Metachromatic Leukodystrophy. Transplantation, 2014, 97, e42-e44.	1.0	2
60	Utility of metabolic screening in neurological presentations of infancy. Annals of Clinical and Translational Neurology, 2020, 7, 1132-1140.	3.7	2
61	A recurrent de novo ATP5F1A substitution associated with neonatal complex V deficiency. European Journal of Human Genetics, 2021, 29, 1719-1724.	2.8	2
62	Effect of Vitamin E on Transport Processes in Isolated Rat Hepatocytes. Journal of Pediatric Gastroenterology and Nutrition, 1990, 11, 261-267.	1.8	1
63	Newborn Screening by Sequence and the Road Ahead. Clinical Chemistry, 2013, 59, 1011-1013.	3.2	1
64	Whole-exome sequencing identifies a homozygous pathogenic variant in TAT in a girl with palmoplantar keratoderma. Molecular Genetics and Metabolism Reports, 2019, 21, 100534.	1.1	1
65	The mitochondrial genome of Cavia aperea. Mitochondrial DNA Part B: Resources, 2020, 5, 2147-2148.	0.4	1
66	Novel Mutations in SLC25A3 Encoding the Mitochondrial Phosphate Carrier. Biophysical Journal, 2016, 110, 474a.	0.5	0
67	Reply. Environmental and Molecular Mutagenesis, 2019, 60, 465-465.	2.2	0
68	Rod bipolar cell dysfunction in POLG retinopathy. Documenta Ophthalmologica, 2021, 142, 111-118.	2.2	0
69	Deubiquitylation errors cause disease. Science, 2021, 371, 358.19-360.	12.6	0
70	Characterization of a Novel Missense CXCR4 Mutation in a Patient with WHIM-like Syndrome. Blood, 2021, 138, 4309-4309.	1.4	0