Sandra T Cooper

List of Publications by Year
in descending order

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74 papers	4,180 citations	94381 37 h-index	118793 62 g-index
76 all docs	76 docs citations	76 times ranked	6506 citing authors

#	Article	IF	Citations
1	Standardized practices for RNA diagnostics using clinically accessible specimens reclassifies 75% of putative splicing variants. Genetics in Medicine, 2022, 24, 130-145.	1.1	45
2	Neonatal-lethal dilated cardiomyopathy due to a homozygous LMOD2 donor splice-site variant. European Journal of Human Genetics, 2022, 30, 450-457.	1.4	9
3	Empirical prediction of variant-activated cryptic splice donors using population-based RNA-Seq data. Nature Communications, 2022, 13, 1655.	5.8	16
4	Prevalence, parameters, and pathogenic mechanisms for splice-altering acceptor variants that disrupt the AG exclusion zone. Human Genetics and Genomics Advances, 2022, 3, 100125.	1.0	2
5	Pathogenic deep intronic MTM1 variant activates a pseudo-exon encoding a nonsense codon resulting in severe X-linked myotubular myopathy. European Journal of Human Genetics, 2021, 29, 61-66.	1.4	10
6	WGS and RNA Studies Diagnose Noncoding DMD Variants in Males With High Creatine Kinase. Neurology: Genetics, 2021, 7, e554.	0.9	21
7	Biallelic and monoallelic variants in PLXNA1 are implicated in a novel neurodevelopmental disorder with variable cerebral and eye anomalies. Genetics in Medicine, 2021, 23, 1715-1725.	1.1	22
8	A Pathway to Precision Medicine for Aboriginal Australians: A Study Protocol. Methods and Protocols, 2021, 4, 42.	0.9	8
9	Recurrent TTN metatranscriptâ€only c.39974â€“11T>G splice variant associated with autosomal recessive arthrogryposis multiplex congenita and myopathy. Human Mutation, 2020, 41, 403-411.	1.1	28
10	Rapid exome sequencing and adjunct RNA studies confirm the pathogenicity of a novel homozygous ASNS splicing variant in a critically ill neonate. Human Mutation, 2020, 41, 1884-1891.	1.1	8
11	Two novel B9D1 variants causing Joubert syndrome: Utility of mRNA and splicing studies. European Journal of Medical Genetics, 2020, 63, 104000.	0.7	1
12	Expanding the disease phenotype of ADSSL1-associated myopathy in non-Korean patients. Neuromuscular Disorders, 2020, 30, 310-314.	0.3	12
13	Loss of calpains-1 and -2 prevents repair of plasma membrane scrape injuries, but not small pores, and induces a severe muscular dystrophy. American Journal of Physiology - Cell Physiology, 2020, 318, C1226-C1237.	2.1	12
14	Pathogenic Abnormal Splicing Due to Intronic Deletions that Induce Biophysical Space Constraint for Spliceosome Assembly. American Journal of Human Genetics, 2019, 105, 573-587.	2.6	25
15	Importance of muscle biopsy to establish pathogenicity of DMD missense and splice variants. Neuromuscular Disorders, 2019, 29, 913-919.	0.3	19
16	Mice with myocyte deletion of vitamin D receptor have sarcopenia and impaired muscle function. Journal of Cachexia, Sarcopenia and Muscle, 2019, 10, 1228-1240.	2.9	79
17	Gene discovery informatics toolkit defines candidate genes for unexplained infertility and prenatal or infantile mortality. Npj Genomic Medicine, 2019, 4, 8.	1.7	31
18	Recessive DES cardio/myopathy without myofibrillar aggregates: intronic splice variant silences one allele leaving only missense L190P-desmin. European Journal of Human Genetics, 2019, 27, 1267-1273.	1.4	14

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19	Congenital Titinopathy: Comprehensive characterization and pathogenic insights. Annals of Neurology, 2018, 83, 1105-1124.	2.8	93
20	Nemaline myopathy and distal arthrogryposis associated with an autosomal recessiveÂTNNT3 Âsplice variant. Human Mutation, 2018, 39, 383-388.	1.1	48
21	Dietary intervention rescues myopathy associated with neurofibromatosis type 1. Human Molecular Genetics, 2018, 27, 577-588.	1.4	21
22	Enzymatic cleavage of myoferlin releases a dual C2-domain module linked to ERK signalling. Cellular Signalling, 2017, 33, 30-40.	1.7	15
23	A â€˜limb-girdle muscular dystrophyâ€™ responsive to asthma therapy. Practical Neurology, 2017, 17, 327-331.	0.5	3
24	Improving genetic diagnosis in Mendelian disease with transcriptome sequencing. Science Translational Medicine, 2017, 9, .	5.8	516
25	Limited proteolysis as a tool to probe the tertiary conformation of dysferlin and structural consequences of patient missense variant L344P. Journal of Biological Chemistry, 2017, 292, 18577-18591.	1.6	6
26	Ca ²⁺ and mitochondrial ROS: Both hero and villain in membrane repair. Science Signaling, 2017, 10, .	1.6	6
27	Cost-effectiveness of massively parallel sequencing for diagnosis of paediatric muscle diseases. Npj Genomic Medicine, 2017, 2, .	1.7	67
28	The Effects of Disease Models of Nuclear Actin Polymerization on the Nucleus. Frontiers in Physiology, 2016, 7, 454.	1.3	18
29	TOR1AIP1 as a cause of cardiac failure and recessive limb-girdle muscular dystrophy. Neuromuscular Disorders, 2016, 26, 500-503.	0.3	38
30	Variants in SLC18A3 , vesicular acetylcholine transporter, cause congenital myasthenic syndrome. Neurology, 2016, 87, 1442-1448.	1.5	46
31	Variants in the Oxidoreductase PYROXD1 Cause Early-Onset Myopathy with Internalized Nuclei and Myofibrillar Disorganization. American Journal of Human Genetics, 2016, 99, 1086-1105.	2.6	45
32	Dopamine-2 receptor extracellular N-terminus regulates receptor surface availability and is the target of human pathogenic antibodies from children with movement and psychiatric disorders. Acta Neuropathologica Communications, 2016, 4, 126.	2.4	28
33	Ferlins Show Tissueâ€Specific Expression and Segregate as Plasma Membrane/Late Endosomal or Transâ€Golgi/Recycling Ferlins. Traffic, 2016, 17, 245-266.	1.3	42
34	Diagnosis and etiology of congenital muscular dystrophy: We are halfway there. Annals of Neurology, 2016, 80, 101-111.	2.8	57
35	Lack of MG53 in human heart precludes utility as a biomarker of myocardial injury or endogenous cardioprotective factor. Cardiovascular Research, 2016, 110, 178-187.	1.8	46
36	Zebrafish models for nemaline myopathy reveal a spectrum of nemaline bodies contributing to reduced muscle function. Acta Neuropathologica, 2015, 130, 389-406.	3.9	47

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37	Mutation in mitochondrial ribosomal protein S7 (MRPS7) causes congenital sensorineural deafness, progressive hepatic and renal failure and lactic acidemia. Human Molecular Genetics, 2015, 24, 2297-2307.	1.4	64
38	Use of Whole-Exome Sequencing for Diagnosis of Limb-Girdle Muscular Dystrophy. JAMA Neurology, 2015, 72, 1424.	4.5	164
39	Muscle weakness inTPM3-myopathy is due to reduced Ca²⁺-sensitivity and impaired acto-myosin cross-bridge cycling in slow fibres. Human Molecular Genetics, 2015, 24, 6278-6292.	1.4	38
40	Membrane Repair: Mechanisms and Pathophysiology. Physiological Reviews, 2015, 95, 1205-1240.	13.1	273
41	LARS2 Variants Associated with Hydrops, Lactic Acidosis, Sideroblastic Anemia, and Multisystem Failure. JIMD Reports, 2015, 28, 49-57.	0.7	48
42	Membrane Injury and Repair in the Muscular Dystrophies. Neuroscientist, 2015, 21, 653-668.	2.6	40
43	Rapid Identification of a Novel Complex I MT-ND3 m.10134C>A Mutation in a Leigh Syndrome Patient. PLoS ONE, 2014, 9, e104879.	1.1	5
44	Alternate Splicing of Dysferlin C2A Confers Ca2+-Dependent and Ca2+-Independent Binding for Membrane Repair. Structure, 2014, 22, 104-115.	1.6	47
45	Calpain cleavage within dysferlin exon 40a releases a synaptotagmin-like module for membrane repair. Molecular Biology of the Cell, 2014, 25, 3037-3048.	0.9	62
46	Calpains, Cleaved Mini-Dysferlin_{C72}, and L-Type Channels Underpin Calcium-Dependent Muscle Membrane Repair. Journal of Neuroscience, 2013, 33, 5085-5094.	1.7	93
47	Mutations in CYC1, Encoding Cytochrome c1 Subunit of Respiratory Chain Complex III, Cause Insulin-Responsive Hyperglycemia. American Journal of Human Genetics, 2013, 93, 384-389.	2.6	61
48	Phenotypic variability and identification of novel YARS2 mutations in YARS2 mitochondrial myopathy, lactic acidosis and sideroblastic anaemia. Orphanet Journal of Rare Diseases, 2013, 8, 193.	1.2	49
49	Ferlins: Regulators of Vesicle Fusion for Auditory Neurotransmission, Receptor Trafficking and Membrane Repair. Traffic, 2012, 13, 185-194.	1.3	119
50	Mosaic caveolin-3 expression in acquired rippling muscle disease without evidence of myasthenia gravis or acetylcholine receptor autoantibodies. Neuromuscular Disorders, 2011, 21, 194-203.	0.3	16
51	Dysferlin, Annexin A1, and Mitsugumin 53 Are Upregulated in Muscular Dystrophy and Localize to Longitudinal Tubules of the T-System With Stretch. Journal of Neuropathology and Experimental Neurology, 2011, 70, 302-313.	0.9	77
52	Using complementary DNA from MyoDâ€transduced fibroblasts to sequence large muscle genes. Muscle and Nerve, 2011, 44, 280-282.	1.0	3
53	In Vitro Analysis of Rod Composition and Actin Dynamics in Inherited Myopathies. Journal of Neuropathology and Experimental Neurology, 2010, 69, 429-441.	0.9	24
54	Mutation of the Mitochondrial Tyrosyl-tRNA Synthetase Gene, YARS2, Causes Myopathy, Lactic Acidosis, and Sideroblastic Anemiaâ€"MLASA Syndrome. American Journal of Human Genetics, 2010, 87, 52-59.	2.6	211

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55	Phylogenetic analysis of ferlin genes reveals ancient eukaryotic origins. BMC Evolutionary Biology, 2010, 10, 231.	3.2	49
56	Recessive mutations in RYR1 are a common cause of congenital fiber type disproportion. Human Mutation, 2010, 31, E1544-E1550.	1.1	153
57	Reduced Plasma Membrane Expression of Dysferlin Mutants Is Attributed to Accelerated Endocytosis via a Syntaxin-4-associated Pathway. Journal of Biological Chemistry, 2010, 285, 28529-28539.	1.6	37
58	Myocardial membrane injury in pediatric cardiac surgery: An animal model. Journal of Thoracic and Cardiovascular Surgery, 2009, 137, 1154-1162.	0.4	9
59	Changes in skeletal muscle expression of AQP1 and AQP4 in dystrophinopathy and dysferlinopathy patients. Acta Neuropathologica, 2008, 116, 235-246.	3.9	27
60	Mutations in Contactin-1, a Neural Adhesion and Neuromuscular Junction Protein, Cause a Familial Form of Lethal Congenital Myopathy. American Journal of Human Genetics, 2008, 83, 714-724.	2.6	72
61	Limbâ€“girdle muscular dystrophy: Diagnostic evaluation, frequency and clues to pathogenesis. Neuromuscular Disorders, 2008, 18, 34-44.	0.3	99
62	Disease Severity and Thin Filament Regulation in M9RTPM3Nemaline Myopathy. Journal of Neuropathology and Experimental Neurology, 2008, 67, 867-877.	0.9	27
63	Mechanisms underlying intranuclear rod formation. Brain, 2007, 130, 3275-3284.	3.7	63
64	Dystrophinopathy carrier determination and detection of protein deficiencies in muscular dystrophy using lentiviral MyoD-forced myogenesis. Neuromuscular Disorders, 2007, 17, 276-284.	0.3	29
65	The pathogenesis ofACTA1-related congenital fiber type disproportion. Annals of Neurology, 2007, 61, 552-561.	2.8	63
66	Intranuclear rod myopathy: molecular pathogenesis and mechanisms of weakness. Annals of Neurology, 2007, 62, 597-608.	2.8	39
67	Aberrant dysferlin trafficking in cells lacking caveolin or expressing dystrophy mutants of caveolin-3. Human Molecular Genetics, 2006, 15, 129-142.	1.4	66
68	An ?tropomyosin mutation alters dimer preference in nemaline myopathy. Annals of Neurology, 2005, 57, 42-49.	2.8	62
69	The Syntrophin-Dystrobrevin Subcomplex in Human Neuromuscular Disorders. Journal of Neuropathology and Experimental Neurology, 2005, 64, 350-361.	0.9	30
70	Defining Î±-skeletal and Î±-cardiac actin expression in human heart and skeletal muscle explains the absence of cardiac involvement in ACTA1 nemaline myopathy. Neuromuscular Disorders, 2005, 15, 829-835.	0.3	73
71	Evidence for a dominant-negative effect in ACTA1 nemaline myopathy caused by abnormal folding, aggregation and altered polymerization of mutant actin isoforms. Human Molecular Genetics, 2004, 13, 1727-1743.	1.4	89
72	Expression of aquaporin 1Bin human cardiac and skeletal muscle. Journal of Molecular and Cellular Cardiology, 2004, 36, 655-662.	0.9	63

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73	Single section Western blot. Neurology, 2003, 61, 93-97.	1.5	86
74	Nemaline Myopathy Caused by Mutations in the Muscle Î±-Skeletal-Actin Gene. American Journal of Human Genetics, 2001, 68, 1333-1343.	2.6	144