

Keith J Johnson

List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/6500673/publications.pdf>

Version: 2024-02-01

75
papers

6,388
citations

172457

29
h-index

102487

66
g-index

77
all docs

77
docs citations

77
times ranked

4062
citing authors

#	ARTICLE	IF	CITATIONS
1	Molecular basis of myotonic dystrophy: Expansion of a trinucleotide (CTG) repeat at the 3' end of a transcript encoding a protein kinase family member. <i>Cell</i> , 1992, 68, 799-808.	28.9	2,464
2	Detection of an unstable fragment of DNA specific to individuals with myotonic dystrophy. <i>Nature</i> , 1992, 355, 547-548.	27.8	622
3	Cloning of the essential myotonic dystrophy region and mapping of the putative defect. <i>Nature</i> , 1992, 355, 548-551.	27.8	498
4	Localization of the malignant hyperthermia susceptibility locus to human chromosome 19q13.2. <i>Nature</i> , 1990, 343, 562-564.	27.8	416
5	Myotonic dystrophy patients have larger CTG expansions in skeletal muscle than in leukocytes. <i>Annals of Neurology</i> , 1994, 35, 104-107.	5.3	259
6	Origin of the expansion mutation in myotonic dystrophy. <i>Nature Genetics</i> , 1993, 4, 72-76.	21.4	215
7	Genetics and Physiology of the Myotonic Muscle Disorders. <i>New England Journal of Medicine</i> , 1993, 328, 482-489.	27.0	154
8	Unstable DNA may be responsible for the incomplete penetrance of the myotonic dystrophy phenotype. <i>Human Molecular Genetics</i> , 1992, 1, 467-473.	2.9	115
9	Characterization of the expression of DMPK and SIX5 in the human eye and implications for pathogenesis in myotonic dystrophy. <i>Human Molecular Genetics</i> , 1999, 8, 481-492.	2.9	88
10	Myotonic dystrophy: absence of CTG enlarged transcript in congenital forms, and low expression of the normal allele. <i>Human Molecular Genetics</i> , 1993, 2, 1263-1266.	2.9	86
11	LMX1B Mutations Cause Hereditary FSGS without Extrarenal Involvement. <i>Journal of the American Society of Nephrology: JASN</i> , 2013, 24, 1216-1222.	6.1	83
12	Myotonic dystrophy: will the real gene please step forward!. <i>Human Molecular Genetics</i> , 1996, 5, 1417-1423.	2.9	75
13	Direct Diagnosis of Myotonic Dystrophy with a Disease-Specific DNA Marker. <i>New England Journal of Medicine</i> , 1993, 328, 471-475.	27.0	74
14	Somatic Instability of the Myotonic Dystrophy (CTG) _n Repeat during Human Fetal Development. <i>Human Molecular Genetics</i> , 1997, 6, 877-880.	2.9	66
15	PhRMA White Paper on ADME Pharmacogenomics. <i>Journal of Clinical Pharmacology</i> , 2008, 48, 849-889.	2.0	62
16	Further Evidence for Clustering of Human GABAA Receptor Subunit Genes: Localization of the δ -Subunit Gene (GABRA6) to Distal Chromosome 5q by Linkage Analysis. <i>Genomics</i> , 1994, 20, 285-288.	2.9	53
17	Meiotic drive at the myotonic dystrophy locus?. <i>Nature Genetics</i> , 1994, 6, 117-118.	21.4	51
18	Drosophila homolog of the myotonic dystrophy-associated gene, SIX5, is required for muscle and gonad development. <i>Current Biology</i> , 2001, 11, 1044-1049.	3.9	49

#	ARTICLE	IF	CITATIONS
19	Genetic diversity in black South Africans from Soweto. <i>BMC Genomics</i> , 2013, 14, 644.	2.8	49
20	Cloning of the HumanSIX1Gene and Its Assignment to Chromosome 14. <i>Genomics</i> , 1996, 33, 140-142.	2.9	48
21	Clinical and molecular diagnosis of a Costa Rican family with autosomal recessive myotonia congenita (Becker disease) carrying a new mutation in the CLCN1 gene. <i>Revista De Biologia Tropical</i> , 2008, 56, 1-11.	0.4	48
22	Confirmation of the localization of the human GABAA receptor $\alpha 1$ -subunit gene (GABRA1) to distal 5q by linkage analysis. <i>Genomics</i> , 1992, 14, 745-748.	2.9	45
23	Expansion of unstable DNA region in Japanese myotonic dystrophy patients. <i>Lancet, The</i> , 1992, 339, 692.	13.7	44
24	Genetics (molecular biology) and Meniere's disease. <i>Otolaryngologic Clinics of North America</i> , 2002, 35, 497-516.	1.1	41
25	Characterisation of Expression of mDMAHP, a Homeodomain-Encoding Gene at the Murine DM Locus. <i>Human Molecular Genetics</i> , 1997, 6, 651-657.	2.9	39
26	Defining response to TNF-inhibitors in rheumatoid arthritis: the negative impact of anti-TNF cycling and the need for a personalized medicine approach to identify primary non-responders. <i>Clinical Rheumatology</i> , 2019, 38, 2967-2976.	2.2	37
27	Genetic linkage and radiation hybrid mapping of the three human GABAC receptor β -subunit genes: CABRR1, CABRR2 and CABRR3. <i>Biochimica Et Biophysica Acta Gene Regulatory Mechanisms</i> , 1999, 1447, 307-312.	2.4	33
28	Robust and tissue-independent gender-specific transcript biomarkers. <i>Biomarkers</i> , 2013, 18, 436-445.	1.9	32
29	Genomic mapping and evolution of human GABA A receptor subunit gene clusters. <i>Mammalian Genome</i> , 1999, 10, 839-843.	2.2	30
30	Sex-related difference in intergenerational expansion of myotonic dystrophy gene. <i>Lancet, The</i> , 1993, 341, 1159-1160.	13.7	29
31	Structure, mapping and expression of the human gene encoding the homeodomain protein, SIX2. <i>Gene</i> , 2000, 247, 145-151.	2.2	29
32	The role of ADME pharmacogenomics in early clinical trials: perspective of the Industry Pharmacogenomics Working Group (I-PWG). <i>Pharmacogenomics</i> , 2015, 16, 2055-2067.	1.3	28
33	Dinucleotide repeat polymorphism in the human X-linked GABA _A receptora3-subunit gene. <i>Nucleic Acids Research</i> , 1991, 19, 4016-4016.	14.5	27
34	So Many Studies, Too Few Subjects: Establishing Functional Relevance of Genetic Polymorphisms on Pharmacokinetics. <i>Journal of Clinical Pharmacology</i> , 2006, 46, 258-264.	2.0	26
35	Clinical Validation of a Blood-Based Predictive Test for Stratification of Response to Tumor Necrosis Factor Inhibitor Therapies in Rheumatoid Arthritis Patients. <i>Network and Systems Medicine</i> , 2020, 3, 91-104.	2.5	26
36	Functional analysis of the homeodomain protein SIX5. <i>Nucleic Acids Research</i> , 2000, 28, 1871-1878.	14.5	25

#	ARTICLE	IF	CITATIONS
37	Regional Workload Induced Changes in Electrophysiology and Immediate Early Gene Expression in Intact In Situ Porcine Heart. <i>Journal of Molecular and Cellular Cardiology</i> , 1997, 29, 3147-3155.	1.9	24
38	Detection of a premutation in Japanese myotonic dystrophy. <i>Human Molecular Genetics</i> , 1994, 3, 819-820.	2.9	23
39	Abnormal contractile activity and calcium cycling in cardiac myocytes isolated from <i>dmpk</i> knockout mice. <i>Physiological Genomics</i> , 2003, 13, 139-146.	2.3	22
40	Race and Ethnicity in the Era of Emerging Pharmacogenomics. <i>Journal of Clinical Pharmacology</i> , 2006, 46, 405-407.	2.0	22
41	Association of CTG repeats and the 1-kb Alu insertion/deletion polymorphism at the myotonin protein kinase gene in the Japanese population suggests a common Eurasian origin of the myotonic dystrophy mutation. <i>Human Genetics</i> , 1996, 97, 145-147.	3.8	21
42	Recombination events that locate myotonic dystrophy distal to APOC2 on 19q. <i>Genomics</i> , 1989, 5, 746-751.	2.9	19
43	Myotonic dystrophy: Another case of too many repeats?. <i>Human Mutation</i> , 1992, 1, 183-189.	2.5	18
44	Cloning and Chromosomal Localization of Human Cdc42-Binding Protein Kinase $\hat{1}^2$. <i>Genomics</i> , 1999, 57, 297-300.	2.9	17
45	Identifying genetic risk variants for coronary heart disease in familial hypercholesterolemia: an extreme genetics approach. <i>European Journal of Human Genetics</i> , 2015, 23, 381-387.	2.8	15
46	Exclusion of linkage between idiopathic generalized epilepsies and the GABAA receptor $\hat{1}\pm 1$ and $\hat{1}^3 2$ subunit gene cluster on chromosome 5. <i>Epilepsy Research</i> , 1996, 23, 235-244.	1.6	12
47	Over Expression of the Murine Myotonic Dystrophy Protein Kinase in the Mouse Myogenic C2C12 Cell Line Leads to Inhibition of Terminal Differentiation. <i>Biochemical and Biophysical Research Communications</i> , 1998, 246, 905-911.	2.1	11
48	Genetic Evidence That the Gene Controlling $Au^{sup}b^{sup}$ Is Located on Chromosome 19. <i>Vox Sanguinis</i> , 1990, 58, 126-128.	1.5	10
49	Waiting for frataxin. <i>Nature Genetics</i> , 1997, 16, 323-325.	21.4	10
50	Further evidence for a major ancient mutation underlying myotonic dystrophy from linkage disequilibrium studies in the Japanese population. <i>Journal of Human Genetics</i> , 1998, 43, 246-249.	2.3	9
51	Gene expression profiling of immunomagnetically separated cells directly from stabilized whole blood for multicenter clinical trials. <i>Clinical and Translational Medicine</i> , 2014, 3, 36.	4.0	9
52	Chronic myopathy in a patient suspected of carrying two malignant hyperthermia susceptibility (MHS) mutations. <i>Neuromuscular Disorders</i> , 1992, 2, 389-396.	0.6	8
53	Characterization of a YAC and cosmid contig containing markers tightly linked to the myotonic dystrophy locus on chromosome 19. <i>Genomics</i> , 1992, 13, 526-531.	2.9	8
54	Malignant hyperthermia hots up!. <i>Human Molecular Genetics</i> , 1993, 2, 849-849.	2.9	8

#	ARTICLE	IF	CITATIONS
55	Common subtypes of idiopathic generalized epilepsies: Lack of linkage to D20S19 close to candidate loci (EBN1, EEGV1) on chromosome 20. , 1996, 67, 31-39.		8
56	Linkage disequilibrium detected between dystrophia myotonica and APOC2 locus in the Finnish population. Human Genetics, 1990, 85, 541-5.	3.8	6
57	Localization of the malignant hyperthermia susceptibility locus to human chromosome 19q12-q13.2. Biochemical Society Transactions, 1990, 18, 326-326.	3.4	4
58	Characterisation of the transcription factor, SIX5, using a new panel of monoclonal antibodies. Journal of Cellular Biochemistry, 2005, 95, 990-1001.	2.6	4
59	Preparative Scale, High Resolution Purification of Low Molecular Weight DNA Fragments. Preparative Biochemistry and Biotechnology, 1982, 12, 429-443.	0.5	3
60	Advances in Gene Technology: Molecular biology of the endocrine system. FEBS Letters, 1987, 214, 200-200.	2.8	3
61	Reply to "Meiotic drive and myotonic dystrophy" Nature Genetics, 1995, 10, 133-133.	21.4	2
62	A 3.7kb fragment from the myotonic dystrophy protein kinase promoter directs neural-specific expression in vivo. Biochemical Society Transactions, 1996, 24, 283S-283S.	3.4	2
63	Mapping of the Dysmyelinating Murine Hindshaker Mutation to a 1.2-cM Interval on Chromosome 3. Genomics, 2002, 80, 126-128.	2.9	2
64	A physical map of the genomic region on mouse chromosome 3 containing the hindshaker (hsh) mutation. Genomics, 2004, 83, 225-230.	2.9	2
65	Pharmacogenomics: Integration into Drug Discovery and Development. Current Topics in Medicinal Chemistry, 2005, 5, 1039-1046.	2.1	2
66	A c-DNA probe for the oncogene c-MEL (pC7"1) recognises a polymorphism with NcoI. Nucleic Acids Research, 1987, 15, 3940-3940.	14.5	1
67	Inheritance and pathogenicity of myotonic dystrophy. , 1993, 3, 85-110.		1
68	Reconstitution of dinucleosomes on restriction fragments. Biochemical Society Transactions, 1983, 11, 370-370.	3.4	0
69	Isolation and ordering of bacteriophage genomic clones corresponding to two YACs from 19q13.3. Molecular and Cellular Probes, 1993, 7, 75-80.	2.1	0
70	Analysis of Triplet Repeat Disorders. Edited by D. C. Rubinsztein and M. R. Hayden. Bios Scientific Publishers Ltd. 1998. 352 pages. ISBN 1 85996 266 1. Price £67.50.. Genetical Research, 1999, 73, 275-277.	0.9	0
71	Novel human pathological mutations. Human Genetics, 2007, 122, 413-420.	3.8	0
72	The virtuous technology cycle concept and its application in next-generation sequencing. Drug Discovery Today, 2012, 17, 1015-1022.	6.4	0

#	ARTICLE	IF	CITATIONS
73	Pharmacogenomics in Drug Discovery and Development. , 2013, , 353-361.		0
74	Sa1033 Interferon-Free Alisporivir Treatment Down-Regulates Interferon-Stimulated Genes Suggesting a Unique Antiviral Mechanism of Action for the Cyclophilin Inhibitor Alisporivir. Gastroenterology, 2013, 144, S-977-S-978.	1.3	0
75	Rearrangement of the Human mel Gene, the rab 8 Homologue, in Human Malignant Melanomas. , 1991, , 81-88.		0