Keith J Johnson

List of Publications by Year
in descending order

75 papers	6,388 citations	172207 29 h-index	102304 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 3â€² end of a transcript encoding a protein kinase family member. Cell, 1992, 68, 799-808.	13.5	2,464
2	Detection of an unstable fragment of DNA specific to individuals with myotonic dystrophy. Nature, 1992, 355, 547-548.	13.7	622
3	Cloning of the essential myotonic dystrophy region and mapping of the putative defect. Nature, 1992, 355, 548-551.	13.7	498
4	Localization of the malignant hyperthermia susceptibility locus to human chromosome 19ql2â€“13.2. Nature, 1990, 343, 562-564.	13.7	416
5	Myotonic dystrophy patients have larger CTG expansions in skeletal muscle than in leukocytes. Annals of Neurology, 1994, 35, 104-107.	2.8	259
6	Origin of the expansion mutation in myotonic dystrophy. Nature Genetics, 1993, 4, 72-76.	9.4	215
7	Genetics and Physiology of the Myotonic Muscle Disorders. New England Journal of Medicine, 1993, 328, 482-489.	13.9	154
8	Unstable DNA may be responsible for the incomplete penetrance of the myotonic dystrophy phenotype. Human Molecular Genetics, 1992, 1, 467-473.	1.4	115
9	Characterization of the expression of DMPK and SIX5 in the human eye and implications for pathogenesis in myotonic dystrophy. Human Molecular Genetics, 1999, 8, 481-492.	1.4	88
10	Myotonic dystrophy: absence of CTG enlarged transcript in congenital forms, and low expression of the normal allele. Human Molecular Genetics, 1993, 2, 1263-1266.	1.4	86
11	LMX1B Mutations Cause Hereditary FSGS without Extrarenal Involvement. Journal of the American Society of Nephrology: JASN, 2013, 24, 1216-1222.	3.0	83
12	Myotonic dystrophy: will the real gene pleasestep forward!. Human Molecular Genetics, 1996, 5, 1417-1423.	1.4	75
13	Direct Diagnosis of Myotonic Dystrophy with a Disease-Specific DNA Marker. New England Journal of Medicine, 1993, 328, 471-475.	13.9	74
14	Somatic Instability of the Myotonic Dystrophy (CTG)n Repeat during Human Fetal Development. Human Molecular Genetics, 1997, 6, 877-880.	1.4	66
15	PhRMA White Paper on ADME Pharmacogenomics. Journal of Clinical Pharmacology, 2008, 48, 849-889.	1.0	62
16	Further Evidence for Clustering of Human GABAA Receptor Subunit Genes: Localization of the Î±6-Subunit Gene (GABRA6) to Distal Chromosome 5q by Linkage Analysis. Genomics, 1994, 20, 285-288.	1.3	53
17	Meiotic drive at the myotonic dystrophy locus?. Nature Genetics, 1994, 6, 117-118.	9.4	51
18	Drosophila homolog of the myotonic dystrophy-associated gene, SIX5, is required for muscle and gonad development. Current Biology, 2001, 11, 1044-1049.	1.8	49

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19	Genetic diversity in black South Africans from Soweto. BMC Genomics, 2013, 14, 644.	1.2	49
20	Cloning of the HumanSIX1Gene and Its Assignment to Chromosome 14. Genomics, 1996, 33, 140-142.	1.3	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. Revista De Biologia Tropical, 2008, 56, 1-11.	0.1	48
22	Confirmation of the localization of the human GABAA receptor Î±1-subunit gene (GABRA1) to distal 5q by linkage analysis. Genomics, 1992, 14, 745-748.	1.3	45
23	Expansion of unstable DNA region in Japanese myotonic dystrophy patients. Lancet, The, 1992, 339, 692.	6.3	44
24	Genetics (molecular biology) and Meniere's disease. Otolaryngologic Clinics of North America, 2002, 35, 497-516.	0.5	41
25	Characterisation of Expression of mDMAHP, a Homeodomain-Encoding Gene at the Murine DM Locus. Human Molecular Genetics, 1997, 6, 651-657.	1.4	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. Clinical Rheumatology, 2019, 38, 2967-2976.	1.0	37
27	Genetic linkage and radiation hybrid mapping of the three human GABAC receptor Ï•subunit genes: GABRR1, GABRR2 and GABRR3. Biochimica Et Biophysica Acta Gene Regulatory Mechanisms, 1999, 1447, 307-312.	2.4	33
28	Robust and tissue-independent gender-specific transcript biomarkers. Biomarkers, 2013, 18, 436-445.	0.9	32
29	Genomic mapping and evolution of human GABA A receptor subunit gene clusters. Mammalian Genome, 1999, 10, 839-843.	1.0	30
30	Sex-related difference in intergenerational expansion of myotonic dystrophy gene. Lancet, The, 1993, 341, 1159-1160.	6.3	29
31	Structure, mapping and expression of the human gene encoding the homeodomain protein, SIX2. Gene, 2000, 247, 145-151.	1.0	29
32	The role of ADME pharmacogenomics in early clinical trials: perspective of the Industry Pharmacogenomics Working Group (I-PWG). Pharmacogenomics, 2015, 16, 2055-2067.	0.6	28
33	Dinucleotide repeat polymorphism in the human X-linked GABAAreceptora3-subunit gene. Nucleic Acids Research, 1991, 19, 4016-4016.	6.5	27
34	So Many Studies, Too Few Subjects: Establishing Functional Relevance of Genetic Polymorphisms on Pharmacokinetics. Journal of Clinical Pharmacology, 2006, 46, 258-264.	1.0	26
35	Clinical Validation of a Blood-Based Predictive Test for Stratification of Response to Tumor Necrosis Factor Inhibitor Therapies in Rheumatoid Arthritis Patients. Network and Systems Medicine, 2020, 3, 91-104.	2.7	26
36	Functional analysis of the homeodomain protein SIX5. Nucleic Acids Research, 2000, 28, 1871-1878.	6.5	25

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37	Regional Workload Induced Changes in Electrophysiology and Immediate Early Gene Expression in IntactIn SituPorcine Heart. Journal of Molecular and Cellular Cardiology, 1997, 29, 3147-3155.	0.9	24
38	Detection of a premutation in Japanese myotonic dystrophy. Human Molecular Genetics, 1994, 3, 819-820.	1.4	23
39	Abnormal contractile activity and calcium cycling in cardiac myocytes isolated from dmpk knockout mice. Physiological Genomics, 2003, 13, 139-146.	1.0	22
40	Race and Ethnicity in the Era of Emerging Pharmacogenomics. Journal of Clinical Pharmacology, 2006, 46, 405-407.	1.0	22
41	Association of CTG repeats and the 1-kbAlu insertion/deletion polymorphism at the myotonin protein kinase gene in the Japanese population suggests a common Eurasian origin of the myotonic dystrophy mutation. Human Genetics, 1996, 97, 145-147.	1.8	21
42	Recombination events that locate myotonic dystrophy distal to APOC2 on 19q. Genomics, 1989, 5, 746-751.	1.3	19
43	Myotonic dystrophy: Another case of too many repeats?. Human Mutation, 1992, 1, 183-189.	1.1	18
44	Cloning and Chromosomal Localization of Human Cdc42-Binding Protein Kinase Î². Genomics, 1999, 57, 297-300.	1.3	17
45	Identifying genetic risk variants for coronary heart disease in familial hypercholesterolemia: an extreme genetics approach. European Journal of Human Genetics, 2015, 23, 381-387.	1.4	15
46	Exclusion of linkage between idiopathic generalized epilepsies and the GABAA receptor Î±1 and Î³2 subunit gene cluster on chromosome 5. Epilepsy Research, 1996, 23, 235-244.	0.8	12
47	Over Expression of the Murine Myotonic Dystrophy Protein Kinase in the Mouse Myogenic C2C12 Cell Line Leads to Inhibition of Terminal Differentiation. Biochemical and Biophysical Research Communications, 1998, 246, 905-911.	1.0	11
48	Genetic Evidence That the Gene Controlling Au^b Is Located on Chromosome 19. Vox Sanguinis, 1990, 58, 126-128.	0.7	10
49	Waiting for frataxin. Nature Genetics, 1997, 16, 323-325.	9.4	10
50	Further evidence for a major ancient mutation underlying myotonic dystrophy from linkage disequilibrium studies in the Japanese population. Journal of Human Genetics, 1998, 43, 246-249.	1.1	9
51	Gene expression profiling of immunomagnetically separated cells directly from stabilized whole blood for multicenter clinical trials. Clinical and Translational Medicine, 2014, 3, 36.	1.7	9
52	Chronic myopathy in a patient suspected of carrying two malignant hyperthermia susceptibility (MHS) mutations. Neuromuscular Disorders, 1992, 2, 389-396.	0.3	8
53	Characterization of a YAC and cosmid contig containing markers tightly linked to the myotonic dystrophy locus on chromosome 19. Genomics, 1992, 13, 526-531.	1.3	8
54	Malignant hyperthermia hots up!. Human Molecular Genetics, 1993, 2, 849-849.	1.4	8

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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.	1.8	6
57	Localization of the malignant hyperthermia susceptibility locus to human chromosome 19q12-q13.2. Biochemical Society Transactions, 1990, 18, 326-326.	1.6	4
58	Characterisation of the transcription factor, SIX5, using a new panel of monoclonal antibodies. Journal of Cellular Biochemistry, 2005, 95, 990-1001.	1.2	4
59	Preparative Scale, High Resolution Purification of Low Molecular Weight DNA Fragments. Preparative Biochemistry and Biotechnology, 1982, 12, 429-443.	0.4	3
60	Advances in Gene Technology: Molecular biology of the endocrine system. FEBS Letters, 1987, 214, 200-200.	1.3	3
61	Reply to â€œMeiotic drive and myotonic dystrophyâ€. Nature Genetics, 1995, 10, 133-133.	9.4	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.	1.6	2
63	Mapping of the Dysmyelinating Murine Hindshaker Mutation to a 1.2-cM Interval on Chromosome 3. Genomics, 2002, 80, 126-128.	1.3	2
64	A physical map of the genomic region on mouse chromosome 3 containing the hindshaker (hsh) mutation. Genomics, 2004, 83, 225-230.	1.3	2
65	Pharmacogenomics: Integration into Drug Discovery and Development. Current Topics in Medicinal Chemistry, 2005, 5, 1039-1046.	1.0	2
66	A c-DNA probe for the oncogene c-MEL (pC7â€“1) recognises a polymorphism with NcoI. Nucleic Acids Research, 1987, 15, 3940-3940.	6.5	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.	1.6	0
69	Isolation and ordering of bacteriophage genomic clones corresponding to two YACs from 19q13.3. Molecular and Cellular Probes, 1993, 7, 75-80.	0.9	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 Â£67.50.. Genetical Research, 1999, 73, 275-277.	0.3	0
71	Novel human pathological mutations. Human Genetics, 2007, 122, 413-420.	1.8	0
72	The virtuous technology cycle concept and its application in next-generation sequencing. Drug Discovery Today, 2012, 17, 1015-1022.	3.2	0

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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.	0.6	0
75	Rearrangement of the Human mel Gene, the rab 8 Homologue, in Human Malignant Melanomas. , 1991, , 81-88.		0