List of Publications by Year in descending order

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#	Article	IF	CITATIONS
1	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.	2.5	26
2	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.	2.2	37
3	The role of ADME pharmacogenomics in early clinical trials: perspective of the Industry Pharmacogenomics Working Group (I-PWG). Pharmacogenomics, 2015, 16, 2055-2067.	1.3	28
4	Identifying genetic risk variants for coronary heart disease in familial hypercholesterolemia: an extreme genetics approach. European Journal of Human Genetics, 2015, 23, 381-387.	2.8	15
5	Gene expression profiling of immunomagnetically separated cells directly from stabilized whole blood for multicenter clinical trials. Clinical and Translational Medicine, 2014, 3, 36.	4.0	9
6	Genetic diversity in black South Africans from Soweto. BMC Genomics, 2013, 14, 644.	2.8	49
7	Pharmacogenomics in Drug Discovery and Development. , 2013, , 353-361.		0
8	Robust and tissue-independent gender-specific transcript biomarkers. Biomarkers, 2013, 18, 436-445.	1.9	32
9	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
10	LMX1B Mutations Cause Hereditary FSCS without Extrarenal Involvement. Journal of the American Society of Nephrology: JASN, 2013, 24, 1216-1222.	6.1	83
11	The virtuous technology cycle concept and its application in next-generation sequencing. Drug Discovery Today, 2012, 17, 1015-1022.	6.4	0
12	PhRMA White Paper on ADME Pharmacogenomics. Journal of Clinical Pharmacology, 2008, 48, 849-889.	2.0	62
13	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.	0.4	48
14	Novel human pathological mutations. Human Genetics, 2007, 122, 413-420.	3.8	0
15	So Many Studies, Too Few Subjects: Establishing Functional Relevance of Genetic Polymorphisms on Pharmacokinetics. Journal of Clinical Pharmacology, 2006, 46, 258-264.	2.0	26
16	Race and Ethnicity in the Era of Emerging Pharmacogenomics. Journal of Clinical Pharmacology, 2006, 46, 405-407.	2.0	22
17	Characterisation of the transcription factor, SIX5, using a new panel of monoclonal antibodies. Journal of Cellular Biochemistry, 2005, 95, 990-1001.	2.6	4
18	Pharmacogenomics: Integration into Drug Discovery and Development. Current Topics in Medicinal Chemistry, 2005, 5, 1039-1046.	2.1	2

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19	A physical map of the genomic region on mouse chromosome 3 containing the hindshaker (hsh) mutation. Genomics, 2004, 83, 225-230.	2.9	2
20	Abnormal contractile activity and calcium cycling in cardiac myocytes isolated from <i>dmpk</i> knockout mice. Physiological Genomics, 2003, 13, 139-146.	2.3	22
21	Mapping of the Dysmyelinating Murine Hindshaker Mutation to a 1.2-cM Interval on Chromosome 3. Genomics, 2002, 80, 126-128.	2.9	2
22	Genetics (molecular biology) and Meniere's disease. Otolaryngologic Clinics of North America, 2002, 35, 497-516.	1.1	41
23	Drosophila homolog of the myotonic dystrophy-associated gene, SIX5, is required for muscle and gonad development. Current Biology, 2001, 11, 1044-1049.	3.9	49
24	Functional analysis of the homeodomain protein SIX5. Nucleic Acids Research, 2000, 28, 1871-1878.	14.5	25
25	Structure, mapping and expression of the human gene encoding the homeodomain protein, SIX2. Gene, 2000, 247, 145-151.	2.2	29
26	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
27	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.	2.9	88
28	Genomic mapping and evolution of human GABA A receptor subunit gene clusters. Mammalian Genome, 1999, 10, 839-843.	2.2	30
29	Genetic linkage and radiation hybrid mapping of the three human GABAC receptor ϕsubunit genes: GABRR1, GABRR2 and GABRR3. Biochimica Et Biophysica Acta Gene Regulatory Mechanisms, 1999, 1447, 307-312.	2.4	33
30	Cloning and Chromosomal Localization of Human Cdc42-Binding Protein Kinase β. Genomics, 1999, 57, 297-300.	2.9	17
31	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.	2.3	9
32	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.	2.1	11
33	Somatic Instability of the Myotonic Dystrophy (CTG)n Repeat during Human Fetal Development. Human Molecular Genetics, 1997, 6, 877-880.	2.9	66
34	Characterisation of Expression of mDMAHP, a Homeodomain-Encoding Gene at the Murine DM Locus. Human Molecular Genetics, 1997, 6, 651-657.	2.9	39
35	Waiting for frataxin. Nature Genetics, 1997, 16, 323-325.	21.4	10
36	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.	1.9	24

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37	Cloning of the HumanSIX1Gene and Its Assignment to Chromosome 14. Genomics, 1996, 33, 140-142.	2.9	48
38	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.	3.8	21
39	Myotonic dystrophy: will the real gene pleasestep forward!. Human Molecular Genetics, 1996, 5, 1417-1423.	2.9	75
40	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
41	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
42	Exclusion of linkage between idiopathic generalized epilepsies and the GABAA receptor α1 and γ2 subunit gene cluster on chromosome 5. Epilepsy Research, 1996, 23, 235-244.	1.6	12
43	Reply to "Meiotic drive and myotonic dystrophy― Nature Genetics, 1995, 10, 133-133.	21.4	2
44	Detection of a premutation in Japanese myotonic dystrophy. Human Molecular Genetics, 1994, 3, 819-820.	2.9	23
45	Myotonic dystrophy patients have larger CTG expansions in skeletal muscle than in leukocytes. Annals of Neurology, 1994, 35, 104-107.	5.3	259
46	Meiotic drive at the myotonic dystrophy locus?. Nature Genetics, 1994, 6, 117-118.	21.4	51
47	Further Evidence for Clustering of Human GABAA Receptor Subunit Genes: Localization of the α6-Subunit Gene (GABRA6) to Distal Chromosome 5q by Linkage Analysis. Genomics, 1994, 20, 285-288.	2.9	53
48	Origin of the expansion mutation in myotonic dystrophy. Nature Genetics, 1993, 4, 72-76.	21.4	215
49	Isolation and ordering of bacteriophage genomic clones corresponding to two YACs from 19q13.3. Molecular and Cellular Probes, 1993, 7, 75-80.	2.1	Ο
50	Genetics and Physiology of the Myotonic Muscle Disorders. New England Journal of Medicine, 1993, 328, 482-489.	27.0	154
51	Sex-related difference in intergenerational expansion of myotonic dystrophy gene. Lancet, The, 1993, 341, 1159-1160.	13.7	29
52	Malignant hyperthermia hots up!. Human Molecular Genetics, 1993, 2, 849-849.	2.9	8
53	Direct Diagnosis of Myotonic Dystrophy with a Disease-Specific DNA Marker. New England Journal of Medicine, 1993, 328, 471-475.	27.0	74
54	Myotonic dystrophy: absence of CTG enlarged transcript in congenital forms, and low expression of the normal allele. Human Molecular Genetics, 1993, 2, 1263-1266.	2.9	86

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55	Inheritance and pathogenicity of myotonic dystrophy. , 1993, 3, 85-110.		1
56	Chronic myopathy in a patient suspected of carrying two malignant hyperthermia susceptibility (MHS) mutations. Neuromuscular Disorders, 1992, 2, 389-396.	0.6	8
57	Unstable DNA may be responsible for the incomplete penetrance of the myotonic dystrophy phenotype. Human Molecular Genetics, 1992, 1, 467-473.	2.9	115
58	Characterization of a YAC and cosmid contig containing markers tightly linked to the myotonic dystrophy locus on chromosome 19. Genomics, 1992, 13, 526-531.	2.9	8
59	Expansion of unstable DNA region in Japanese myotonic dystrophy patients. Lancet, The, 1992, 339, 692.	13.7	44
60	Confirmation of the localization of the human GABAA receptor α1-subunit gene (GABRA1) to distal 5q by linkage analysis. Genomics, 1992, 14, 745-748.	2.9	45
61	Molecular basis of myotonic dystrophy: Expansion of a trinucleotide (CTG) repeat at the 3′ end of a transcript encoding a protein kinase family member. Cell, 1992, 68, 799-808.	28.9	2,464
62	Detection of an unstable fragment of DNA specific to individuals with myotonic dystrophy. Nature, 1992, 355, 547-548.	27.8	622
63	Cloning of the essential myotonic dystrophy region and mapping of the putative defect. Nature, 1992, 355, 548-551.	27.8	498
64	Myotonic dystrophy: Another case of too many repeats?. Human Mutation, 1992, 1, 183-189.	2.5	18
65	Dinucleotide repeat polymorphism in the human X-linked GABAAreceptora3-subunit gene. Nucleic Acids Research, 1991, 19, 4016-4016.	14.5	27
66	Rearrangement of the Human mel Gene, the rab 8 Homologue, in Human Malignant Melanomas. , 1991, , 81-88.		0
67	Localization of the malignant hyperthermia susceptibility locus to human chromosome 19q12-q13.2. Biochemical Society Transactions, 1990, 18, 326-326.	3.4	4
68	Localization of the malignant hyperthermia susceptibility locus to human chromosome 19ql2–13.2. Nature, 1990, 343, 562-564.	27.8	416
69	Genetic Evidence That the Gene Controlling Au ^b Is Located on Chromosome 19. Vox Sanguinis, 1990, 58, 126-128.	1.5	10
70	Linkage disequilibrium detected between dystrophia myotonica and APOC2 locus in the Finnish population. Human Genetics, 1990, 85, 541-5.	3.8	6
71	Recombination events that locate myotonic dystrophy distal to APOC2 on 19q. Genomics, 1989, 5, 746-751.	2.9	19
72	A c-DNA probe for the oncogene c-MEL (pC7–1) recognises a polymorphism with Ncol. Nucleic Acids Research, 1987, 15, 3940-3940.	14.5	1

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73	Advances in Gene Technology: Molecular biology of the endocrine system. FEBS Letters, 1987, 214, 200-200.	2.8	3
74	Reconstitution of dinucleosomes on restriction fragments. Biochemical Society Transactions, 1983, 11, 370-370.	3.4	0
75	Preparative Scale, High Resolution Purification of Low Molecular Weight DNA Fragments. Preparative Biochemistry and Biotechnology, 1982, 12, 429-443.	0.5	3