John David Brook

List of Publications by Year
in descending order

92 papers	10,666 citations	101543 36 h-index	43889 91 g-index
97 all docs	97 docs citations	97 times ranked	9367 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.	28.9	2,464
2	Campomelic dysplasia and autosomal sex reversal caused by mutations in an SRY-related gene. Nature, 1994, 372, 525-530.	27.8	1,476
3	Holt-Oram syndrome is caused by mutations in TBX5, a member of the Brachyury (T) gene family. Nature Genetics, 1997, 15, 21-29.	21.4	859
4	Expansion of an unstable DNA region and phenotypic variation in myotonic dystrophy. Nature, 1992, 355, 545-546.	27.8	746
5	Three proteins, MBNL, MBLL and MBXL, co-localize in vivo with nuclear foci of expanded-repeat transcripts in DM1 and DM2 cells. Human Molecular Genetics, 2002, 11, 805-814.	2.9	401
6	Distinct genetic architectures for syndromic and nonsyndromic congenital heart defects identified by exome sequencing. Nature Genetics, 2016, 48, 1060-1065.	21.4	351
7	Contribution of Global Rare Copy-Number Variants to the Risk of Sporadic Congenital Heart Disease. American Journal of Human Genetics, 2012, 91, 489-501.	6.2	272
8	Mutation in myosin heavy chain 6 causes atrial septal defect. Nature Genetics, 2005, 37, 423-428.	21.4	243
9	Clinical and molecular aspects of the myotonic dystrophies: A review. Muscle and Nerve, 2005, 32, 1-18.	2.2	208
10	In vivo co-localisation of MBNL protein with DMPK expanded-repeat transcripts. Nucleic Acids Research, 2001, 29, 2766-2771.	14.5	190
11	Unstable DNA sequence in myotonic dystrophy. Lancet, The, 1992, 339, 1125-1128.	13.7	189
12	T-box genes in human disorders. Human Molecular Genetics, 2003, 12, 37R-44.	2.9	182
13	Rare Variants in NR2F2 Cause Congenital Heart Defects in Humans. American Journal of Human Genetics, 2014, 94, 574-585.	6.2	146
14	Î±-Cardiac myosin heavy chain (MYH6) mutations affecting myofibril formation are associated with congenital heart defects. Human Molecular Genetics, 2010, 19, 4007-4016.	2.9	131
15	Transcriptional abnormality in myotonic dystrophy affects DMPK but not neighboring genes. Proceedings of the National Academy of Sciences of the United States of America, 1997, 94, 7394-7399.	7.1	130
16	Alpha-cardiac actin mutations produce atrial septal defects. Human Molecular Genetics, 2008, 17, 256-265.	2.9	128
17	Genome-wide association study of multiple congenital heart disease phenotypes identifies a susceptibility locus for atrial septal defect at chromosome 4p16. Nature Genetics, 2013, 45, 822-824.	21.4	123
18	Whole Exome Sequencing Reveals the Major Genetic Contributors to Nonsyndromic Tetralogy of Fallot. Circulation Research, 2019, 124, 553-563.	4.5	118

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19	Physical Interaction between TBX5 and MEF2C Is Required for Early Heart Development. Molecular and Cellular Biology, 2009, 29, 2205-2218.	2.3	115
20	Phenotype-specific effect of chromosome 1q21.1 rearrangements and GJA5 duplications in 2436 congenital heart disease patients and 6760 controls. Human Molecular Genetics, 2012, 21, 1513-1520.	2.9	101
21	Holtâ€“Oram syndrome is a genetically heterogeneous disease with one locus mapping to human chromosome 12q. Nature Genetics, 1994, 6, 401-404.	21.4	87
22	Genome-wide association study identifies loci on 12q24 and 13q32 associated with Tetralogy of Fallot. Human Molecular Genetics, 2013, 22, 1473-1481.	2.9	82
23	Defective mRNA in myotonic dystrophy accumulates at the periphery of nuclear splicing speckles. Genes To Cells, 2007, 12, 1035-1048.	1.2	80
24	Genomic organization and transcriptional units at the myotonic dystrophy locus. Genomics, 1993, 18, 673-679.	2.9	77
25	Myotonic Dystrophy Is Associated with a Reduced Level of RNA from the DMWD Allele Adjacent to the Expanded Repeat. Human Molecular Genetics, 1999, 8, 1491-1497.	2.9	76
26	Stem cell factor (SCF), a novel hematopoietic growth factor and ligand for c-kit tyrosine kinase receptor, maps on human chromosome 12 between 12q14.3 and 12qter. Somatic Cell and Molecular Genetics, 1991, 17, 207-214.	0.7	75
27	Combined Mutation Screening of NKX2-5, GATA4, and TBX5 in Congenital Heart Disease: Multiple Heterozygosity and Novel Mutations. Congenital Heart Disease, 2012, 7, 151-159.	0.2	73
28	High-content screening identifies small molecules that remove nuclear foci, affect MBNL distribution and CELF1 protein levels via a PKC-independent pathway in myotonic dystrophy cell lines. Human Molecular Genetics, 2014, 23, 1551-1562.	2.9	69
29	The Impact of Mechanical Forces in Heart Morphogenesis. Circulation: Cardiovascular Genetics, 2012, 5, 132-142.	5.1	67
30	Muscleblind-Like Proteins. American Journal of Pathology, 2009, 174, 216-227.	3.8	65
31	Expanded CUG Repeats Dysregulate RNA Splicing by Altering the Stoichiometry of the Muscleblind 1 Complex. Journal of Biological Chemistry, 2011, 286, 38427-38438.	3.4	58
32	A novel human cytochrome P4S0 gene (P450IIB): chromosomal localization and evidence for alternative splicing. Nucleic Acids Research, 1988, 16, 5783-5795.	14.5	57
33	Identification, Mapping, and Phylogenomic Analysis of Four New Human Members of the T-box Gene Family:EOMES, TBX6, TBX18,andTBX19. Genomics, 1999, 55, 10-20.	2.9	57
34	The Amyloid Precursor-like Protein (APLP) Gene Maps to the Long Arm of Human Chromosome 19. Genomics, 1993, 15, 237-239.	2.9	52
35	Close linkage of the human cytochrome P450IIA and P450IIB gene subfamilies: implications for the assignment of substrate specificity. Nucleic Acids Research, 1989, 17, 2907-2917.	14.5	47
36	Tropomyosin 1: Multiple roles in the developing heart and in the formation of congenital heart defects. Journal of Molecular and Cellular Cardiology, 2017, 106, 1-13.	1.9	40

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37	Quantitative Methods to Monitor RNA Biomarkers in Myotonic Dystrophy. Scientific Reports, 2018, 8, 5885.	3.3	39
38	Physical and genetic characterization of the distal segment of the myotonic dystrophy area on 19q. Genomics, 1992, 13, 509-517.	2.9	38
39	Flies deficient in Muscleblind protein model features of myotonic dystrophy with altered splice forms of Z-band associated transcripts. Human Genetics, 2006, 120, 487-499.	3.8	38
40	Loss of Heterozygosity in Bilateral Breast Cancer. Breast Cancer Research and Treatment, 2000, 64, 241-251.	2.5	35
41	Acetylation of TBX5 by KAT2B and KAT2A regulates heart and limb development. Journal of Molecular and Cellular Cardiology, 2018, 114, 185-198.	1.9	35
42	Localisation of the myotonic dystrophy locus to 19q13.2?19q13.3 and its relationship to twelve polymorphic loci on 19q. Human Genetics, 1991, 87, 73-80.	3.8	34
43	Zebrafish deficient for Muscleblind-like 2 exhibit features of myotonic dystrophy. DMM Disease Models and Mechanisms, 2011, 4, 381-392.	2.4	34
44	A Common Variant in the PTPN11 Gene Contributes to the Risk of Tetralogy of Fallot. Circulation: Cardiovascular Genetics, 2012, 5, 287-292.	5.1	34
45	Small Molecules Which Improve Pathogenesis of Myotonic Dystrophy Type 1. Frontiers in Neurology, 2018, 9, 349.	2.4	32
46	Further mapping of markers around the centromere of human chromosome 19. Genomics, 1987, 1, 320-328.	2.9	31
47	Human glandular kallikrein genes: Genetic and physical mapping of the KLK1 locus using a highly polymorphic microsatellite PCR marker. Genomics, 1991, 11, 77-82.	2.9	31
48	Association Between C677T Polymorphism of Methylene Tetrahydrofolate Reductase and Congenital Heart Disease. Circulation: Cardiovascular Genetics, 2013, 6, 347-353.	5.1	31
49	A genome-wide association study of congenital cardiovascular left-sided lesions shows association with a locus on chromosome 20. Human Molecular Genetics, 2016, 25, 2331-2341.	2.9	31
50	Regional localisations and linkage relationships of seven RFLPs and myotonic dystrophy on chromosome 19. Human Genetics, 1986, 74, 262-6.	3.8	30
51	The miR-30 MicroRNA Family Targets smoothened to Regulate Hedgehog Signalling in Zebrafish Early Muscle Development. PLoS ONE, 2013, 8, e65170.	2.5	30
52	T-Box Genes in Human Development and Disease. Current Topics in Developmental Biology, 2017, 122, 383-415.	2.2	30
53	Localization of a human Na+, K+-ATPase ? subunit gene to chromosome 19q12ï¿½q13.2 and linkage to the myotonic dystrophy locus. Genomics, 1988, 3, 380-384.	2.9	28
54	Testing for the chemical induction of aneuploidy in the male mouse. Mutation Research - Environmental Mutagenesis and Related Subjects Including Methodology, 1986, 164, 117-125.	0.4	27

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55	Retreat of the triplet repeat?. Nature Genetics, 1993, 3, 279-281.	21.4	26
56	Virtual Cloning and Physical Mapping of a Human T-Box Gene, TBX4. Genomics, 2000, 67, 92-95.	2.9	25
57	Global Increase in Circular RNA Levels in Myotonic Dystrophy. Frontiers in Genetics, 2019, 10, 649.	2.3	24
58	Long-range restriction map of a region of human chromosome 19 containing the apolipoprotein genes, a CLL-associated translocation breakpoint, and two polymorphic MluI sites. Human Genetics, 1989, 83, 71-74.	3.8	23
59	Knockdown of alpha myosin heavy chain disrupts the cytoskeleton and leads to multiple defects during chick cardiogenesis. Journal of Anatomy, 2009, 214, 905-915.	1.5	22
60	Linkage relationships of the insulin receptor gene with the complement component 3, LDL receptor, apolipoprotein C2 and myotonic dystrophy loci on chromosome 19. Human Genetics, 1986, 74, 267-9.	3.8	20
61	Assignment of two of the translation initiation factor-4E (EIF4EL1 and EIF4EL2) genes to human chromosomes 4 and 20. Genomics, 1991, 10, 1079-1082.	2.9	19
62	Genetic Variation in VEGF Does Not Contribute Significantly to the Risk of Congenital Cardiovascular Malformation. PLoS ONE, 2009, 4, e4978.	2.5	19
63	Radiation-reduced hybrids for the myotonic dystrophy locus. Genomics, 1992, 13, 243-250.	2.9	17
64	A High-Resolution Whole Genome Radiation Hybrid Map of Human Chromosome 17q22â€“q25.3 across the Genes forGHandTK. Genomics, 1996, 33, 185-192.	2.9	17
65	Integrative analysis of genomic variants reveals new associations of candidate haploinsufficient genes with congenital heart disease. PLoS Genetics, 2021, 17, e1009679.	3.5	17
66	Body composition and clinical outcome measures in patients with myotonic dystrophy type 1. Neuromuscular Disorders, 2017, 27, 286-289.	0.6	15
67	Systems genetics analysis identifies calcium-signaling defects as novel cause of congenital heart disease. Genome Medicine, 2020, 12, 76.	8.2	15
68	Differential fates of introns in gene expression due to global alternative splicing. Human Genetics, 2022, 141, 31-47.	3.8	14
69	The physical map of chromosome arm 19q: some new assignments, confirmations and re-assessments. Human Genetics, 1991, 87, 65-72.	3.8	13
70	Low-frequency intermediate penetrance variants in the ROCK1 gene predispose to Tetralogy of Fallot. BMC Genetics, 2013, 14, 57.	2.7	12
71	CDK12 inhibition reduces abnormalities in cells from patients with myotonic dystrophy and in a mouse model. Science Translational Medicine, 2020, 12, .	12.4	12
72	Chromosomal assignment of c-MEL, a human transforming oncogene, to chromosome 19 (p13.2-q13.2). Somatic Cell and Molecular Genetics, 1986, 12, 637-640.	0.7	11

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73	Insertion/deletion polymorphism at D19S95 associated with the myotonic dystrophy CTG repeat. Human Molecular Genetics, 1992, 1, 451-451.	2.9	10
74	The Identification of Exons from the MED/PSACH Region of Human Chromosome 19. Genomics, 1996, 32, 218-224.	2.9	9
75	Six transcripts map within 200 kilobases of the myotonic dystrophy expanded repeat. Mammalian Genome, 1998, 9, 485-487.	2.2	9
76	Formation, Contraction, and Mechanotransduction of Myofribrils in Cardiac Development: Clues from Genetics. Biochemistry Research International, 2012, 2012, 1-6.	3.3	9
77	Confirmation and refinement of the localisation of the c-MEL locus on chromosome 19 by physical and genetic mapping. Human Genetics, 1989, 81, 382-384.	3.8	8
78	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
79	HDAC4 and 5 repression of TBX5 is relieved by protein kinase D1. Scientific Reports, 2019, 9, 17992.	3.3	7
80	Recent advances in congenital heart disease genomics. F1000Research, 2017, 6, 869.	1.6	7
81	A polymorphic DNA clone which maps to 19q13.2 - 19qter (D19S62). Nucleic Acids Research, 1990, 18, 1086-1086.	14.5	6
82	Linkage relationships of the apolipoprotein C1 gene and a cytochrome P450 gene (CYP2A) to myotonic dystrophy. Human Genetics, 1990, 85, 305-10.	3.8	5
83	Interaction makes the heart grow stronger. Trends in Molecular Medicine, 2003, 9, 407-409.	6.7	5
84	Disrupting the Molecular Pathway in Myotonic Dystrophy. International Journal of Molecular Sciences, 2021, 22, 13225.	4.1	5
85	Mapping human chromosomes in somatic cell hybrids using a low-copy-number repetitive sequence. Somatic Cell and Molecular Genetics, 1986, 12, 333-337.	0.7	3
86	A Transcript Map of a 10-Mb Region of Chromosome 19: A Source of Genes for Human Disorders, Including Candidates for Genes Involved in Asthma, Heart Defects, and Eye Development. Genomics, 2000, 63, 425-429.	2.9	3
87	Studies of Genes Involved in Congenital Heart Disease. Journal of Cardiovascular Development and Disease, 2014, 1, 134-145.	1.6	2
88	A polymorphic DNA clone which maps to 19p13.2 â†' 19q12 (D19S27). Nucleic Acids Research, 1988, 16, 7751-7751.	14.5	1
89	A HindIII polymorphism identified by a DNA clone which maps to chromosome 17 (D17S245). Nucleic Acids Research, 1990, 18, 1085-1085.	14.5	0
90	DMPK. , 1995, , 122-123.		0

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91	Myotonic dystrophy. , 2020, , 61-83.		0
92	Splicing in two skeletal muscle transcripts correlates with clinical phenotype in myotonic dystrophy type 1 patients. Journal of Neurology, 2022, , 1.	3.6	0