

John David Brook

List of Publications by Citations

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

90
papers

8,914
citations

36
h-index

94
g-index

97
ext. papers

9,923
ext. citations

10.6
avg, IF

4.91
L-index

#	Paper	IF	Citations
90	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	56.2	2045
89	Campomelic dysplasia and autosomal sex reversal caused by mutations in an SRY-related gene. <i>Nature</i> , 1994 , 372, 525-30	50.4	1340
88	Holt-Oram syndrome is caused by mutations in TBX5, a member of the Brachyury (T) gene family. <i>Nature Genetics</i> , 1997 , 15, 21-9	36.3	755
87	Expansion of an unstable DNA region and phenotypic variation in myotonic dystrophy. <i>Nature</i> , 1992 , 355, 545-6	50.4	656
86	Three proteins, MBNL, MBLL and MBXL, co-localize in vivo with nuclear foci of expanded-repeat transcripts in DM1 and DM2 cells. <i>Human Molecular Genetics</i> , 2002 , 11, 805-14	5.6	346
85	Mutation in myosin heavy chain 6 causes atrial septal defect. <i>Nature Genetics</i> , 2005 , 37, 423-8	36.3	215
84	Contribution of global rare copy-number variants to the risk of sporadic congenital heart disease. <i>American Journal of Human Genetics</i> , 2012 , 91, 489-501	11	213
83	Distinct genetic architectures for syndromic and nonsyndromic congenital heart defects identified by exome sequencing. <i>Nature Genetics</i> , 2016 , 48, 1060-5	36.3	200
82	Clinical and molecular aspects of the myotonic dystrophies: a review. <i>Muscle and Nerve</i> , 2005 , 32, 1-18	3.4	179
81	Unstable DNA sequence in myotonic dystrophy. <i>Lancet, The</i> , 1992 , 339, 1125-8	40	169
80	In vivo co-localisation of MBNL protein with DMPK expanded-repeat transcripts. <i>Nucleic Acids Research</i> , 2001 , 29, 2766-71	20.1	165
79	T-box genes in human disorders. <i>Human Molecular Genetics</i> , 2003 , 12 Spec No 1, R37-44	5.6	161
78	Transcriptional abnormality in myotonic dystrophy affects DMPK but not neighboring genes. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 1997 , 94, 7394-9	11.5	122
77	Rare variants in NR2F2 cause congenital heart defects in humans. <i>American Journal of Human Genetics</i> , 2014 , 94, 574-85	11	115
76	Alpha-cardiac myosin heavy chain (MYH6) mutations affecting myofibril formation are associated with congenital heart defects. <i>Human Molecular Genetics</i> , 2010 , 19, 4007-16	5.6	104
75	Alpha-cardiac actin mutations produce atrial septal defects. <i>Human Molecular Genetics</i> , 2008 , 17, 256-65	5.6	104
74	Physical interaction between TBX5 and MEF2C is required for early heart development. <i>Molecular and Cellular Biology</i> , 2009 , 29, 2205-18	4.8	97

73	Genome-wide association study of multiple congenital heart disease phenotypes identifies a susceptibility locus for atrial septal defect at chromosome 4p16. <i>Nature Genetics</i> , 2013 , 45, 822-4	36.3	91
72	Phenotype-specific effect of chromosome 1q21.1 rearrangements and GJA5 duplications in 2436 congenital heart disease patients and 6760 controls. <i>Human Molecular Genetics</i> , 2012 , 21, 1513-20	5.6	83
71	Holt-Oram syndrome is a genetically heterogeneous disease with one locus mapping to human chromosome 12q. <i>Nature Genetics</i> , 1994 , 6, 401-4	36.3	73
70	Defective mRNA in myotonic dystrophy accumulates at the periphery of nuclear splicing speckles. <i>Genes To Cells</i> , 2007 , 12, 1035-48	2.3	71
69	Myotonic dystrophy is associated with a reduced level of RNA from the DMWD allele adjacent to the expanded repeat. <i>Human Molecular Genetics</i> , 1999 , 8, 1491-7	5.6	69
68	Genome-wide association study identifies loci on 12q24 and 13q32 associated with tetralogy of Fallot. <i>Human Molecular Genetics</i> , 2013 , 22, 1473-81	5.6	68
67	Genomic organization and transcriptional units at the myotonic dystrophy locus. <i>Genomics</i> , 1993 , 18, 673-9	4.3	67
66	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. <i>Somatic Cell and Molecular Genetics</i> , 1991 , 17, 207-14		67
65	Combined mutation screening of NKX2-5, GATA4, and TBX5 in congenital heart disease: multiple heterozygosity and novel mutations. <i>Congenital Heart Disease</i> , 2012 , 7, 151-9	3.1	62
64	Whole Exome Sequencing Reveals the Major Genetic Contributors to Nonsyndromic Tetralogy of Fallot. <i>Circulation Research</i> , 2019 , 124, 553-563	15.7	62
63	Muscleblind-like proteins: similarities and differences in normal and myotonic dystrophy muscle. <i>American Journal of Pathology</i> , 2009 , 174, 216-27	5.8	58
62	The impact of mechanical forces in heart morphogenesis. <i>Circulation: Cardiovascular Genetics</i> , 2012 , 5, 132-42		56
61	A novel human cytochrome P450 gene (P450IIB): chromosomal localization and evidence for alternative splicing. <i>Nucleic Acids Research</i> , 1988 , 16, 5783-95	20.1	54
60	Identification, mapping, and phylogenomic analysis of four new human members of the T-box gene family: EOMES, TBX6, TBX18, and TBX19. <i>Genomics</i> , 1999 , 55, 10-20	4.3	52
59	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. <i>Human Molecular Genetics</i> , 2014 , 23, 1551-62	5.6	48
58	The amyloid precursor-like protein (APLP) gene maps to the long arm of human chromosome 19. <i>Genomics</i> , 1993 , 15, 237-9	4.3	47
57	Expanded CUG repeats Dysregulate RNA splicing by altering the stoichiometry of the muscleblind 1 complex. <i>Journal of Biological Chemistry</i> , 2011 , 286, 38427-38438	5.4	45
56	Close linkage of the human cytochrome P450IIA and P450IIB gene subfamilies: implications for the assignment of substrate specificity. <i>Nucleic Acids Research</i> , 1989 , 17, 2907-17	20.1	45

55	Flies deficient in Muscleblind protein model features of myotonic dystrophy with altered splice forms of Z-band associated transcripts. <i>Human Genetics</i> , 2006 , 120, 487-99	6.3	36
54	Physical and genetic characterization of the distal segment of the myotonic dystrophy area on 19q. <i>Genomics</i> , 1992 , 13, 509-17	4.3	36
53	Loss of heterozygosity in bilateral breast cancer. <i>Breast Cancer Research and Treatment</i> , 2000 , 64, 241-51	4.4	32
52	Localisation of the myotonic dystrophy locus to 19q13.2-19q13.3 and its relationship to twelve polymorphic loci on 19q. <i>Human Genetics</i> , 1991 , 87, 73-80	6.3	31
51	The miR-30 microRNA family targets smoothed to regulate hedgehog signalling in zebrafish early muscle development. <i>PLoS ONE</i> , 2013 , 8, e65170	3.7	29
50	Zebrafish deficient for Muscleblind-like 2 exhibit features of myotonic dystrophy. <i>DMM Disease Models and Mechanisms</i> , 2011 , 4, 381-92	4.1	29
49	Further mapping of markers around the centromere of human chromosome 19. <i>Genomics</i> , 1987 , 1, 320-8	4.3	29
48	Localization of a human Na ⁺ ,K ⁺ -ATPase alpha subunit gene to chromosome 19q12----q13.2 and linkage to the myotonic dystrophy locus. <i>Genomics</i> , 1988 , 3, 380-4	4.3	28
47	Regional localisations and linkage relationships of seven RFLPs and myotonic dystrophy on chromosome 19. <i>Human Genetics</i> , 1986 , 74, 262-6	6.3	27
46	Association between C677T polymorphism of methylene tetrahydrofolate reductase and congenital heart disease: meta-analysis of 7697 cases and 13,125 controls. <i>Circulation: Cardiovascular Genetics</i> , 2013 , 6, 347-53		26
45	A common variant in the PTPN11 gene contributes to the risk of tetralogy of Fallot. <i>Circulation: Cardiovascular Genetics</i> , 2012 , 5, 287-92		25
44	Human glandular Kallikrein genes: genetic and physical mapping of the KLK1 locus using a highly polymorphic microsatellite PCR marker. <i>Genomics</i> , 1991 , 11, 77-82	4.3	25
43	Tropomyosin 1: Multiple roles in the developing heart and in the formation of congenital heart defects. <i>Journal of Molecular and Cellular Cardiology</i> , 2017 , 106, 1-13	5.8	24
42	Small Molecules Which Improve Pathogenesis of Myotonic Dystrophy Type 1. <i>Frontiers in Neurology</i> , 2018 , 9, 349	4.1	23
41	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. <i>Human Genetics</i> , 1989 , 83, 71-4	6.3	21
40	Acetylation of TBX5 by KAT2B and KAT2A regulates heart and limb development. <i>Journal of Molecular and Cellular Cardiology</i> , 2018 , 114, 185-198	5.8	20
39	Virtual cloning and physical mapping of a human T-box gene, TBX4. <i>Genomics</i> , 2000 , 67, 92-5	4.3	20
38	Testing for the chemical induction of aneuploidy in the male mouse. <i>Mutation Research - Environmental Mutagenesis and Related Subjects Including Methodology</i> , 1986 , 164, 117-25		20

37	Knockdown of alpha myosin heavy chain disrupts the cytoskeleton and leads to multiple defects during chick cardiogenesis. <i>Journal of Anatomy</i> , 2009 , 214, 905-15	2.9	19
36	Quantitative Methods to Monitor RNA Biomarkers in Myotonic Dystrophy. <i>Scientific Reports</i> , 2018 , 8, 5885	4.9	18
35	A genome-wide association study of congenital cardiovascular left-sided lesions shows association with a locus on chromosome 20. <i>Human Molecular Genetics</i> , 2016 , 25, 2331-2341	5.6	18
34	Linkage relationships of the insulin receptor gene with the complement component 3, LDL receptor, apolipoprotein C2 and myotonic dystrophy loci on chromosome 19. <i>Human Genetics</i> , 1986 , 74, 267-9	6.3	18
33	A high-resolution whole genome radiation hybrid map of human chromosome 17q22-q25.3 across the genes for GH and TK. <i>Genomics</i> , 1996 , 33, 185-92	4.3	17
32	T-Box Genes in Human Development and Disease. <i>Current Topics in Developmental Biology</i> , 2017 , 122, 383-415	5.3	16
31	Radiation-reduced hybrids for the myotonic dystrophy locus. <i>Genomics</i> , 1992 , 13, 243-50	4.3	16
30	Assignment of two of the translation initiation factor-4E (EIF4EL1 and EIF4EL2) genes to human chromosomes 4 and 20. <i>Genomics</i> , 1991 , 10, 1079-82	4.3	15
29	Global Increase in Circular RNA Levels in Myotonic Dystrophy. <i>Frontiers in Genetics</i> , 2019 , 10, 649	4.5	14
28	Genetic variation in VEGF does not contribute significantly to the risk of congenital cardiovascular malformation. <i>PLoS ONE</i> , 2009 , 4, e4978	3.7	13
27	The physical map of chromosome arm 19q: some new assignments, confirmations and re-assessments. <i>Human Genetics</i> , 1991 , 87, 65-72	6.3	12
26	Chromosomal assignment of c-MEL, a human transforming oncogene, to chromosome 19 (p13.2-q13.2). <i>Somatic Cell and Molecular Genetics</i> , 1986 , 12, 637-40		11
25	Low-frequency intermediate penetrance variants in the ROCK1 gene predispose to Tetralogy of Fallot. <i>BMC Genetics</i> , 2013 , 14, 57	2.6	9
24	Insertion/deletion polymorphism at D19S95 associated with the myotonic dystrophy CTG repeat. <i>Human Molecular Genetics</i> , 1992 , 1, 451	5.6	9
23	Body composition and clinical outcome measures in patients with myotonic dystrophy type 1. <i>Neuromuscular Disorders</i> , 2017 , 27, 286-289	2.9	8
22	The identification of exons from the MED/PSACH region of human chromosome 19. <i>Genomics</i> , 1996 , 32, 218-24	4.3	8
21	Characterization of a YAC and cosmid contig containing markers tightly linked to the myotonic dystrophy locus on chromosome 19. <i>Genomics</i> , 1992 , 13, 526-31	4.3	8
20	Confirmation and refinement of the localisation of the c-MEL locus on chromosome 19 by physical and genetic mapping. <i>Human Genetics</i> , 1989 , 81, 382-4	6.3	8

19	CDK12 inhibition reduces abnormalities in cells from patients with myotonic dystrophy and in a mouse model. <i>Science Translational Medicine</i> , 2020 , 12,	17.5	7
18	Six transcripts map within 200 kilobases of the myotonic dystrophy expanded repeat. <i>Mammalian Genome</i> , 1998 , 9, 485-7	3.2	7
17	Formation, contraction, and mechanotransduction of myofibrils in cardiac development: clues from genetics. <i>Biochemistry Research International</i> , 2012 , 2012, 504906	2.4	6
16	A polymorphic DNA clone which maps to 19q13.2-19qter (D19S62). <i>Nucleic Acids Research</i> , 1990 , 18, 10860.1	6.1	6
15	Interaction makes the heart grow stronger. <i>Trends in Molecular Medicine</i> , 2003 , 9, 407-9	11.5	5
14	Linkage relationships of the apolipoprotein C1 gene and a cytochrome P450 gene (CYP2A) to myotonic dystrophy. <i>Human Genetics</i> , 1990 , 85, 305-10	6.3	5
13	Systems genetics analysis identifies calcium-signaling defects as novel cause of congenital heart disease. <i>Genome Medicine</i> , 2020 , 12, 76	14.4	5
12	Recent advances in congenital heart disease genomics. <i>F1000Research</i> , 2017 , 6, 869	3.6	4
11	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. <i>Genomics</i> , 2000 , 63, 425-9	4.3	3
10	Mapping human chromosomes in somatic cell hybrids using a low-copy-number repetitive sequence. <i>Somatic Cell and Molecular Genetics</i> , 1986 , 12, 333-7		3
9	HDAC4 and 5 repression of TBX5 is relieved by protein kinase D1. <i>Scientific Reports</i> , 2019 , 9, 17992	4.9	3
8	Studies of Genes Involved in Congenital Heart Disease. <i>Journal of Cardiovascular Development and Disease</i> , 2014 , 1, 134-145	4.2	2
7	Global increase in circRNA levels in myotonic dystrophy		2
6	Differential fates of introns in gene expression due to global alternative splicing.. <i>Human Genetics</i> , 2021 , 141, 31	6.3	2
5	A polymorphic DNA clone which maps to 19p13.2----19q12 (D19S27). <i>Nucleic Acids Research</i> , 1988 , 16, 7751	20.1	1
4	Integrative analysis of genomic variants reveals new associations of candidate haploinsufficient genes with congenital heart disease. <i>PLoS Genetics</i> , 2021 , 17, e1009679	6	1
3	Splicing in two skeletal muscle transcripts correlates with clinical phenotype in myotonic dystrophy type 1 patients.. <i>Journal of Neurology</i> , 2022 , 1	5.5	
2	Myotonic dystrophy 2020 , 61-83		

- 1 DMPK: Myotonic distrophy PK (vertebrates) **1995**, 122-123