John David Brook

List of Publications by Year in Descending Order

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	Splicing in two skeletal muscle transcripts correlates with clinical phenotype in myotonic dystrophy type 1 patients.. *Journal of Neurology*, **2022**, 1	5.5	
89	Integrative analysis of genomic variants reveals new associations of candidate haploinsufficient genes with congenital heart disease. *PLoS Genetics*, **2021**, 17, e1009679	6	1
88	Differential fates of introns in gene expression due to global alternative splicing.. *Human Genetics*, **2021**, 141, 31	6.3	2
87	CDK12 inhibition reduces abnormalities in cells from patients with myotonic dystrophy and in a mouse model. *Science Translational Medicine*, **2020**, 12,	17.5	7
86	Myotonic dystrophy **2020**, 61-83		
85	Systems genetics analysis identifies calcium-signaling defects as novel cause of congenital heart disease. *Genome Medicine*, **2020**, 12, 76	14.4	5
84	Global Increase in Circular RNA Levels in Myotonic Dystrophy. *Frontiers in Genetics*, **2019**, 10, 649	4.5	14
83	HDAC4 and 5 repression of TBX5 is relieved by protein kinase D1. *Scientific Reports*, **2019**, 9, 17992	4.9	3
82	Whole Exome Sequencing Reveals the Major Genetic Contributors to Nonsyndromic Tetralogy of Fallot. *Circulation Research*, **2019**, 124, 553-563	15.7	62
81	Acetylation of TBX5 by KAT2B and KAT2A regulates heart and limb development. *Journal of Molecular and Cellular Cardiology*, **2018**, 114, 185-198	5.8	20
80	Quantitative Methods to Monitor RNA Biomarkers in Myotonic Dystrophy. *Scientific Reports*, **2018**, 8, 5885	4.9	18
79	Small Molecules Which Improve Pathogenesis of Myotonic Dystrophy Type 1. *Frontiers in Neurology*, **2018**, 9, 349	4.1	23
78	Body composition and clinical outcome measures in patients with myotonic dystrophy type 1. *Neuromuscular Disorders*, **2017**, 27, 286-289	2.9	8
77	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	5.8	24
76	T-Box Genes in Human Development and Disease. *Current Topics in Developmental Biology*, **2017**, 122, 383-415	5.3	16
75	Recent advances in congenital heart disease genomics. *F1000Research*, **2017**, 6, 869	3.6	4
74	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	5.6	18

73	Distinct genetic architectures for syndromic and nonsyndromic congenital heart defects identified by exome sequencing. *Nature Genetics*, **2016**, 48, 1060-5	36.3	200
72	Rare variants in NR2F2 cause congenital heart defects in humans. *American Journal of Human Genetics*, **2014**, 94, 574-85	11	115
71	Studies of Genes Involved in Congenital Heart Disease. *Journal of Cardiovascular Development and Disease*, **2014**, 1, 134-145	4.2	2
70	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, 1551-62	5.6	48
69	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, 822-4	36.3	91
68	Low-frequency intermediate penetrance variants in the ROCK1 gene predispose to Tetralogy of Fallot. *BMC Genetics*, **2013**, 14, 57	2.6	9
67	Association between C677T polymorphism of methylene tetrahydrofolate reductase and congenital heart disease: meta-analysis of 7697 cases and 13,125 controls. *Circulation: Cardiovascular Genetics*, **2013**, 6, 347-53		26
66	Genome-wide association study identifies loci on 12q24 and 13q32 associated with tetralogy of Fallot. *Human Molecular Genetics*, **2013**, 22, 1473-81	5.6	68
65	The miR-30 microRNA family targets smoothened to regulate hedgehog signalling in zebrafish early muscle development. *PLoS ONE*, **2013**, 8, e65170	3.7	29
64	Combined mutation screening of NKX2-5, GATA4, and TBX5 in congenital heart disease: multiple heterozygosity and novel mutations. *Congenital Heart Disease*, **2012**, 7, 151-9	3.1	62
63	The impact of mechanical forces in heart morphogenesis. *Circulation: Cardiovascular Genetics*, **2012**, 5, 132-42		56
62	Contribution of global rare copy-number variants to the risk of sporadic congenital heart disease. *American Journal of Human Genetics*, **2012**, 91, 489-501	11	213
61	A common variant in the PTPN11 gene contributes to the risk of tetralogy of Fallot. *Circulation: Cardiovascular Genetics*, **2012**, 5, 287-92		25
60	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, 1513-20	5.6	83
59	Formation, contraction, and mechanotransduction of myofribrils in cardiac development: clues from genetics. *Biochemistry Research International*, **2012**, 2012, 504906	2.4	6
58	Expanded CUG repeats Dysregulate RNA splicing by altering the stoichiometry of the muscleblind 1 complex. *Journal of Biological Chemistry*, **2011**, 286, 38427-38438	5.4	45
57	Zebrafish deficient for Muscleblind-like 2 exhibit features of myotonic dystrophy. *DMM Disease Models and Mechanisms*, **2011**, 4, 381-92	4.1	29
56	Alpha-cardiac myosin heavy chain (MYH6) mutations affecting myofibril formation are associated with congenital heart defects. *Human Molecular Genetics*, **2010**, 19, 4007-16	5.6	104

55	Genetic variation in VEGF does not contribute significantly to the risk of congenital cardiovascular malformation. *PLoS ONE*, **2009**, 4, e4978	3.7	13
54	Physical interaction between TBX5 and MEF2C is required for early heart development. *Molecular and Cellular Biology*, **2009**, 29, 2205-18	4.8	97
53	Knockdown of alpha myosin heavy chain disrupts the cytoskeleton and leads to multiple defects during chick cardiogenesis. *Journal of Anatomy*, **2009**, 214, 905-15	2.9	19
52	Muscleblind-like proteins: similarities and differences in normal and myotonic dystrophy muscle. *American Journal of Pathology*, **2009**, 174, 216-27	5.8	58
51	Alpha-cardiac actin mutations produce atrial septal defects. *Human Molecular Genetics*, **2008**, 17, 256-65	5.6	104
50	Defective mRNA in myotonic dystrophy accumulates at the periphery of nuclear splicing speckles. *Genes To Cells*, **2007**, 12, 1035-48	2.3	71
49	Flies deficient in Muscleblind protein model features of myotonic dystrophy with altered splice forms of Z-band associated transcripts. *Human Genetics*, **2006**, 120, 487-99	6.3	36
48	Mutation in myosin heavy chain 6 causes atrial septal defect. *Nature Genetics*, **2005**, 37, 423-8	36.3	215
47	Clinical and molecular aspects of the myotonic dystrophies: a review. *Muscle and Nerve*, **2005**, 32, 1-18	3.4	179
46	Interaction makes the heart grow stronger. *Trends in Molecular Medicine*, **2003**, 9, 407-9	11.5	5
45	T-box genes in human disorders. *Human Molecular Genetics*, **2003**, 12 Spec No 1, R37-44	5.6	161
44	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, 805-14	5.6	346
43	In vivo co-localisation of MBNL protein with DMPK expanded-repeat transcripts. *Nucleic Acids Research*, **2001**, 29, 2766-71	20.1	165
42	Loss of heterozygosity in bilateral breast cancer. *Breast Cancer Research and Treatment*, **2000**, 64, 241-51	4.4	32
41	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, 425-9	4.3	3
40	Virtual cloning and physical mapping of a human T-box gene, TBX4. *Genomics*, **2000**, 67, 92-5	4.3	20
39	Myotonic dystrophy is associated with a reduced level of RNA from the DMWD allele adjacent to the expanded repeat. *Human Molecular Genetics*, **1999**, 8, 1491-7	5.6	69
38	Identification, mapping, and phylogenomic analysis of four new human members of the T-box gene family: EOMES, TBX6, TBX18, and TBX19. *Genomics*, **1999**, 55, 10-20	4.3	52

37	Six transcripts map within 200 kilobases of the myotonic dystrophy expanded repeat. *Mammalian Genome*, **1998**, 9, 485-7	3.2	7
36	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, 7394-9	11.5	122
35	Holt-Oram syndrome is caused by mutations in TBX5, a member of the Brachyury (T) gene family. *Nature Genetics*, **1997**, 15, 21-9	36.3	755
34	The identification of exons from the MED/PSACH region of human chromosome 19. *Genomics*, **1996**, 32, 218-24	4.3	8
33	A high-resolution whole genome radiation hybrid map of human chromosome 17q22-q25.3 across the genes for GH and TK. *Genomics*, **1996**, 33, 185-92	4.3	17
32	DMPK: Myotonic distrophy PK (vertebrates) **1995**, 122-123		
31	Holt-Oram syndrome is a genetically heterogeneous disease with one locus mapping to human chromosome 12q. *Nature Genetics*, **1994**, 6, 401-4	36.3	73
30	Campomelic dysplasia and autosomal sex reversal caused by mutations in an SRY-related gene. *Nature*, **1994**, 372, 525-30	50.4	1340
29	The amyloid precursor-like protein (APLP) gene maps to the long arm of human chromosome 19. *Genomics*, **1993**, 15, 237-9	4.3	47
28	Genomic organization and transcriptional units at the myotonic dystrophy locus. *Genomics*, **1993**, 18, 673-9	4.3	67
27	Insertion/deletion polymorphism at D19S95 associated with the myotonic dystrophy CTG repeat. *Human Molecular Genetics*, **1992**, 1, 451	5.6	9
26	Characterization of a YAC and cosmid contig containing markers tightly linked to the myotonic dystrophy locus on chromosome 19. *Genomics*, **1992**, 13, 526-31	4.3	8
25	Radiation-reduced hybrids for the myotonic dystrophy locus. *Genomics*, **1992**, 13, 243-50	4.3	16
24	Physical and genetic characterization of the distal segment of the myotonic dystrophy area on 19q. *Genomics*, **1992**, 13, 509-17	4.3	36
23	Unstable DNA sequence in myotonic dystrophy. *Lancet, The*, **1992**, 339, 1125-8	40	169
22	Molecular basis of myotonic dystrophy: expansion of a trinucleotide (CTG) repeat at the 3Send of a transcript encoding a protein kinase family member. *Cell*, **1992**, 68, 799-808	56.2	2045
21	Expansion of an unstable DNA region and phenotypic variation in myotonic dystrophy. *Nature*, **1992**, 355, 545-6	50.4	656
20	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, 207-14		67

19	The physical map of chromosome arm 19q: some new assignments, confirmations and re-assessments. *Human Genetics*, **1991**, 87, 65-72	6.3	12
18	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	6.3	31
17	Assignment of two of the translation initiation factor-4E (EIF4EL1 and EIF4EL2) genes to human chromosomes 4 and 20. *Genomics*, **1991**, 10, 1079-82	4.3	15
16	Human glandular Kallikrein genes: genetic and physical mapping of the KLK1 locus using a highly polymorphic microsatellite PCR marker. *Genomics*, **1991**, 11, 77-82	4.3	25
15	Linkage relationships of the apolipoprotein C1 gene and a cytochrome P450 gene (CYP2A) to myotonic dystrophy. *Human Genetics*, **1990**, 85, 305-10	6.3	5
14	A polymorphic DNA clone which maps to 19q13.2-19qter (D19S62). *Nucleic Acids Research*, **1990**, 18, 1086	20.1	6
13	Close linkage of the human cytochrome P450IIA and P450IIB gene subfamilies: implications for the assignment of substrate specificity. *Nucleic Acids Research*, **1989**, 17, 2907-17	20.1	45
12	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, 71-4	6.3	21
11	Confirmation and refinement of the localisation of the c-MEL locus on chromosome 19 by physical and genetic mapping. *Human Genetics*, **1989**, 81, 382-4	6.3	8
10	Localization of a human Na+,K+-ATPase alpha subunit gene to chromosome 19q12----q13.2 and linkage to the myotonic dystrophy locus. *Genomics*, **1988**, 3, 380-4	4.3	28
9	A polymorphic DNA clone which maps to 19p13.2----19q12 (D19S27). *Nucleic Acids Research*, **1988**, 16, 7751	20.1	1
8	A novel human cytochrome P450 gene (P450IIB): chromosomal localization and evidence for alternative splicing. *Nucleic Acids Research*, **1988**, 16, 5783-95	20.1	54
7	Further mapping of markers around the centromere of human chromosome 19. *Genomics*, **1987**, 1, 320-8	4.3	29
6	Regional localisations and linkage relationships of seven RFLPs and myotonic dystrophy on chromosome 19. *Human Genetics*, **1986**, 74, 262-6	6.3	27
5	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	6.3	18
4	Testing for the chemical induction of aneuploidy in the male mouse. *Mutation Research - Environmental Mutagenesis and Related Subjects Including Methodology*, **1986**, 164, 117-25		20
3	Mapping human chromosomes in somatic cell hybrids using a low-copy-number repetitive sequence. *Somatic Cell and Molecular Genetics*, **1986**, 12, 333-7		3
2	Chromosomal assignment of c-MEL, a human transforming oncogene, to chromosome 19 (p13.2-q13.2). *Somatic Cell and Molecular Genetics*, **1986**, 12, 637-40		11

List of Publications

1 Global increase in circRNA levels in myotonic dystrophy 2