

David E Barton

List of Publications by Year in descending order

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121
papers

5,680
citations

61857

43
h-index

85405

71
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127
all docs

127
docs citations

127
times ranked

6509
citing authors

#	ARTICLE	IF	CITATIONS
1	Copy Number Variant Analysis and Genome-wide Association Study Identify Loci with Large Effect for Vesicoureteral Reflux. <i>Journal of the American Society of Nephrology: JASN</i> , 2021, 32, 805-820.	3.0	17
2	National Newborn Screening for cystic fibrosis in the Republic of Ireland: genetic data from the first 6.5 years. <i>European Journal of Human Genetics</i> , 2020, 28, 1669-1674.	1.4	4
3	Investigation of DNA variants specific to ROBO2 Isoform α ™ in Irish vesicoureteric reflux patients reveals marked CpG island variation. <i>Scientific Reports</i> , 2020, 10, 2265.	1.6	2
4	Managing uncertainty in inherited cardiac pathologies—an international multidisciplinary survey. <i>European Journal of Human Genetics</i> , 2019, 27, 1178-1185.	1.4	6
5	The copy number variation landscape of congenital anomalies of the kidney and urinary tract. <i>Nature Genetics</i> , 2019, 51, 117-127.	9.4	144
6	Estimating the age of p.(Phe508del) with family studies of geographically distinct European populations and the early spread of cystic fibrosis. <i>European Journal of Human Genetics</i> , 2018, 26, 1832-1839.	1.4	45
7	Genetic Drivers of Kidney Defects in the DiGeorge Syndrome. <i>New England Journal of Medicine</i> , 2017, 376, 742-754.	13.9	120
8	Incidence of Fragile X syndrome in Ireland. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 678-683.	0.7	4
9	Genome-wide linkage and association study implicates the 10q26 region as a major genetic contributor to primary nonsyndromic vesicoureteric reflux. <i>Scientific Reports</i> , 2017, 7, 14595.	1.6	17
10	Reply to Sajantila and Budowle. <i>European Journal of Human Genetics</i> , 2016, 24, 330-330.	1.4	1
11	Urinary Tract Effects of HPSE2 Mutations. <i>Journal of the American Society of Nephrology: JASN</i> , 2015, 26, 797-804.	3.0	31
12	Development and validation of a novel PCR-RFLP based method for the detection of 3 primary mitochondrial mutations in Leber's hereditary optic neuropathy patients. <i>Eye and Vision (London,)</i> 10 Tf 50		
13	Recommendations for reporting results of diagnostic genetic testing (biochemical, cytogenetic and) 10 Tf 109	1.4	109
14	A new genome scan for primary nonsyndromic vesicoureteric reflux emphasizes high genetic heterogeneity and shows linkage and association with various genes already implicated in urinary tract development. <i>Molecular Genetics & Genomic Medicine</i> , 2014, 2, 7-29.	0.6	23
15	Identification of a BRCA2-Specific Modifier Locus at 6p24 Related to Breast Cancer Risk. <i>PLoS Genetics</i> , 2013, 9, e1003173.	1.5	105
16	Genome-Wide Association Study in BRCA1 Mutation Carriers Identifies Novel Loci Associated with Breast and Ovarian Cancer Risk. <i>PLoS Genetics</i> , 2013, 9, e1003212.	1.5	244
17	Heterozygous non-synonymous ROBO2 variants are unlikely to be sufficient to cause familial vesicoureteric reflux. <i>Kidney International</i> , 2013, 84, 327-337.	2.6	12
18	EMQN/CMGS best practice guidelines for the molecular genetic testing of Huntington disease. <i>European Journal of Human Genetics</i> , 2013, 21, 480-486.	1.4	59

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19	Quality assurance practices in Europe: a survey of molecular genetic testing laboratories. <i>European Journal of Human Genetics</i> , 2012, 20, 1118-1126.	1.4	24
20	Clinical correlation and molecular evaluation confirm that the MLH1 p.Arg182Gly (c.544A>G) mutation is pathogenic and causes Lynch syndrome. <i>Familial Cancer</i> , 2012, 11, 509-518.	0.9	4
21	Multiallelic Synthetic Quality Control Material: Lessons Learned from the Cystic Fibrosis External Quality Assessment Scheme. <i>Genetic Testing and Molecular Biomarkers</i> , 2011, 15, 579-586.	0.3	1
22	Preparation and validation of the first WHO international genetic reference panel for Fragile X syndrome. <i>European Journal of Human Genetics</i> , 2011, 19, 10-17.	1.4	20
23	Establishment of the first WHO international genetic reference panel for Prader Willi and Angelman syndromes. <i>European Journal of Human Genetics</i> , 2011, 19, 857-864.	1.4	12
24	Common alleles at 6q25.1 and 1p11.2 are associated with breast cancer risk for BRCA1 and BRCA2 mutation carriers. <i>Human Molecular Genetics</i> , 2011, 20, 3304-3321.	1.4	68
25	Common variants of the BRCA1 wild-type allele modify the risk of breast cancer in BRCA1 mutation carriers. <i>Human Molecular Genetics</i> , 2011, 20, 4732-4747.	1.4	32
26	Genetics of vesicoureteral reflux. <i>Nature Reviews Urology</i> , 2011, 8, 539-552.	1.9	33
27	Alterations in the steroid hormone receptor co-chaperone FKBPL are associated with male infertility: a case-control study. <i>Reproductive Biology and Endocrinology</i> , 2010, 8, 22.	1.4	31
28	EMQN Best Practice Guidelines for molecular genetic testing of SCAs. <i>European Journal of Human Genetics</i> , 2010, 18, 1173-1176.	1.4	41
29	Benchmarks for Cystic Fibrosis carrier screening: A European consensus document. <i>Journal of Cystic Fibrosis</i> , 2010, 9, 165-178.	0.3	75
30	All azoospermic males should be screened for cystic fibrosis mutations before intracytoplasmic sperm injection. <i>Fertility and Sterility</i> , 2010, 94, 2448-2450.	0.5	14
31	Cross-border genetic testing. <i>Orphanet Journal of Rare Diseases</i> , 2010, 5, .	1.2	0
32	The increased incidence of the RET p.Gly691Ser variant in French-Canadian vesicoureteric reflux patients is not replicated by a larger study in Ireland. <i>Human Mutation</i> , 2009, 30, E612-E617.	1.1	10
33	When good CF tests go bad. <i>European Journal of Human Genetics</i> , 2009, 17, 403-405.	1.4	2
34	Linkage Analysis of Candidate Genes in Families With Vesicoureteral Reflux. <i>Journal of Urology</i> , 2009, 182, 1669-1672.	0.2	11
35	Evaluation and use of a synthetic quality control material, included in the European external quality assessment scheme for cystic fibrosis. <i>Human Mutation</i> , 2008, 29, 1063-1070.	1.1	10
36	Emerging technologies for point-of-care genetic testing. <i>Expert Review of Molecular Diagnostics</i> , 2007, 7, 359-370.	1.5	44

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37	A genome-wide scan for genes involved in primary vesicoureteric reflux. <i>Journal of Medical Genetics</i> , 2007, 44, 710-717.	1.5	39
38	Age at Menarche and Menopause and Breast Cancer Risk in the International BRCA1/2 Carrier Cohort Study. <i>Cancer Epidemiology Biomarkers and Prevention</i> , 2007, 16, 740-746.	1.1	63
39	Report of an International Survey of Molecular Genetic Testing Laboratories. <i>Public Health Genomics</i> , 2007, 10, 123-131.	0.6	20
40	Targeted versus Whole-Genome Array Comparative Genome Hybridization. <i>Journal of Molecular Diagnostics</i> , 2007, 9, 278.	1.2	1
41	Familial Vesicoureteral Reflux: Influence of Sex on Prevalence and Expression. <i>Journal of Urology</i> , 2006, 176, 1776-1780.	0.2	16
42	Best practice guidelines for the molecular genetic diagnosis of Type 1 (HFE-related) hereditary haemochromatosis. <i>BMC Medical Genetics</i> , 2006, 7, 81.	2.1	20
43	Uroplakin III is not a major candidate gene for primary vesicoureteral reflux. <i>European Journal of Human Genetics</i> , 2005, 13, 500-502.	1.4	32
44	Fragile X-associated tremor/ataxia syndrome presenting in a woman after chemotherapy. <i>Neurology</i> , 2005, 65, 331-332.	1.5	49
45	Developing a Sustainable Process to Provide Quality Control Materials for Genetic Testing. <i>Genetics in Medicine</i> , 2005, 7, 534-549.	1.1	49
46	Detection of Five Common CFTR Mutations by Rapid-Cycle Real-Time Amplification Refractory Mutation System PCR. <i>Clinical Chemistry</i> , 2004, 50, 773-775.	1.5	6
47	Certified Reference Materials for Genetic Testing. , 2004, , 226-231.		2
48	Predictive testing for BRCA1 and 2 mutations: a male contribution. <i>Annals of Oncology</i> , 2003, 14, 549-553.	0.6	33
49	Intrafamilial Phenotypic Variability in Friedreich Ataxia Associated With a G130V Mutation in the FRDA Gene. <i>Archives of Neurology</i> , 2002, 59, 296.	4.9	17
50	Angiotensin II Type 2 Receptor Gene is Not Responsible For Familial Vesicoureteral Reflux. <i>Journal of Urology</i> , 2002, 168, 1138-1141.	0.2	33
51	The detection of large deletions or duplications in genomic DNA. <i>Human Mutation</i> , 2002, 20, 325-337.	1.1	110
52	Angiotensin II type 2 receptor gene is not responsible for familial vesicoureteral reflux. <i>Journal of Urology</i> , 2002, 168, 1138-41.	0.2	17
53	The relationship between Y chromosome DNA haplotypes and Y chromosome deletions leading to male infertility. <i>Human Genetics</i> , 2001, 108, 55-58.	1.8	36
54	Mutations at the mitochondrial DNA polymerase (POLG) locus associated with male infertility. <i>Nature Genetics</i> , 2001, 29, 261-262.	9.4	173

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55	Quality control in molecular genetic testing. <i>Nature Reviews Genetics</i> , 2001, 2, 717-723.	7.7	120
56	A comparison of methods for gene dosage analysis in HMSN type 1. <i>Journal of Medical Genetics</i> , 2001, 38, 90-95.	1.5	21
57	Typical Friedreich's ataxia without GAA expansions and GAA expansions without typical Friedreich's ataxia. <i>Journal of Neurology</i> , 2000, 247, 346-355.	1.8	41
58	Retaining the Confidence of the Public in Molecular Genetic Testing " Quality Assurance. <i>Public Health Genomics</i> , 2000, 3, 164-169.	0.6	3
59	The Exon 13 Duplication in the BRCA1 Gene Is a Founder Mutation Present in Geographically Diverse Populations. <i>American Journal of Human Genetics</i> , 2000, 67, 207-212.	2.6	100
60	Hyperinsulinism caused by paternal-specific inheritance of a recessive mutation in the sulfonylurea-receptor gene. <i>Diabetes</i> , 1999, 48, 1652-1657.	0.3	79
61	Fine-resolution mapping by haplotype evaluation: the examples of PFIC1 and BRIC. <i>Human Genetics</i> , 1999, 104, 241-248.	1.8	33
62	National scientific medical meeting 1997 abstracts. <i>Irish Journal of Medical Science</i> , 1998, 167, 1-44.	0.8	0
63	Ret proto-oncogene analysis in medullary thyroid carcinoma. <i>Irish Journal of Medical Science</i> , 1998, 167, 226-230.	0.8	3
64	Sylvester o"halloran surgical scientific meeting. <i>Irish Journal of Medical Science</i> , 1998, 167, 1-16.	0.8	0
65	Confirmation of FWT1 as a Wilms" tumour susceptibility gene and phenotypic characteristics of Wilms" tumour attributable to FWT1. <i>Human Genetics</i> , 1998, 103, 547-556.	1.8	48
66	Hyperinsulinism: molecular aetiology of focal disease. <i>Archives of Disease in Childhood</i> , 1998, 79, 445-447.	1.0	62
67	Instability of normal (CTG) _n alleles in the DM kinase gene.. <i>Journal of Medical Genetics</i> , 1997, 34, 871-873.	1.5	4
68	Mosaicism for trisomy 3q arising from an unbalanced, de novo t(3;15).. <i>Journal of Medical Genetics</i> , 1997, 34, 512-514.	1.5	11
69	Diagnostic molecular genetics comes of age. <i>Trends in Genetics</i> , 1997, 13, 249-250.	2.9	9
70	Genetic Services in Ireland. <i>European Journal of Human Genetics</i> , 1997, 5, 100-104.	1.4	0
71	Cystic fibrosis diagnosed by molecular genetic investigation in the mother of a patient with cystic fibrosis. <i>Thorax</i> , 1997, 52, 96-7.	2.7	2
72	Improved genetic mapping of X linked retinoschisis.. <i>Journal of Medical Genetics</i> , 1996, 33, 919-922.	1.5	13

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73	Molecular genetic investigation of the neurofibromatosis type 2 tumor suppressor gene in sporadic meningioma. <i>Journal of Neurosurgery</i> , 1996, 84, 847-851.	0.9	69
74	Normal CAG and CCG repeats in the Huntington's disease genes of Parkinson's disease patients. <i>American Journal of Medical Genetics Part A</i> , 1995, 60, 109-110.	2.4	4
75	Cystic fibrosis mutation analysis: Report from 22 U.K. regional genetics laboratories. <i>Human Mutation</i> , 1995, 6, 326-333.	1.1	38
76	Haplotype analysis of the \hat{P}^{2642} and (CAG) n polymorphisms in the Huntington's disease (HD) gene provides an explanation for an apparent "founder" HD haplotype. <i>Human Molecular Genetics</i> , 1995, 4, 203-206.	1.4	44
77	Genetic linkage analysis in hereditary non-polyposis colon cancer syndrome.. <i>Journal of Medical Genetics</i> , 1995, 32, 352-357.	1.5	26
78	A frequent hMSH2 mutation in hereditary non-polyposis colon cancer syndrome. <i>Lancet, The</i> , 1995, 345, 727.	6.3	42
79	Molecular genetic analysis of exons 1 to 6 of the APC gene in non-polyposis familial colorectal cancer. <i>Clinical Genetics</i> , 1995, 48, 299-303.	1.0	8
80	B37 repeats are normal in most schizophrenic patients. <i>British Journal of Psychiatry</i> , 1994, 164, 851-852.	1.7	12
81	Direct, non-radioactive detection of mutations in Multiple Endocrine Neoplasia Type 2A families. <i>Human Molecular Genetics</i> , 1994, 3, 643-646.	1.4	44
82	Study of the Huntington's disease (HD) gene CAG repeats in schizophrenic patients shows overlap of the normal and HD affected ranges but absence of correlation with schizophrenia.. <i>Journal of Medical Genetics</i> , 1994, 31, 690-693.	1.5	19
83	Chimaerism shown by cytogenetics and DNA polymorphism analysis.. <i>Journal of Medical Genetics</i> , 1994, 31, 816-817.	1.5	29
84	Stickler syndrome: Correlation between vitreoretinal phenotypes and linkage to COL 2A1. <i>Eye</i> , 1994, 8, 609-614.	1.1	90
85	Mosaic uniparental disomy in Beckwith-Wiedemann syndrome.. <i>Journal of Medical Genetics</i> , 1994, 31, 749-753.	1.5	83
86	Carrier detection in X linked ocular albinism using linked DNA polymorphisms.. <i>British Journal of Ophthalmology</i> , 1994, 78, 539-541.	2.1	9
87	Somatic NF2 gene mutations in familial and non-familial vestibular schwannoma. <i>Human Molecular Genetics</i> , 1994, 3, 347-350.	1.4	102
88	Mutational bias provides a model for the evolution of Huntington's disease and predicts a general increase in disease prevalence. <i>Nature Genetics</i> , 1994, 7, 525-530.	9.4	141
89	Myotonic dystrophy CTG repeats and the associated insertion/deletion polymorphism in human and primate populations. <i>Human Molecular Genetics</i> , 1994, 3, 2031-5.	1.4	17
90	Site of (CCG) polymorphism in the HD gene. <i>Nature Genetics</i> , 1993, 5, 214-215.	9.4	35

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91	Genotype analysis in cystic fibrosis in relation to the occurrence of diabetes mellitus. <i>Clinical Genetics</i> , 1993, 43, 186-189.	1.0	30
92	Genetic Mapping of X-Linked Ocular Albinism: Linkage Analysis in a Large Newfoundland Kindred. <i>Genomics</i> , 1993, 16, 259-261.	1.3	21
93	Analysis of the huntingtin gene reveals a trinucleotide-length polymorphism in the region of the gene that contains two CCG-rich stretches and a correlation between decreased age of onset of Huntington's disease and CAG repeat number. <i>Human Molecular Genetics</i> , 1993, 2, 1713-1715.	1.4	125
94	Cystic fibrosis identified by neonatal screening: incidence, genotype, and early natural history.. <i>Archives of Disease in Childhood</i> , 1993, 68, 464-467.	1.0	38
95	Evaluation of molecular genetic diagnosis in the management of familial adenomatous polyposis coli: a population based study.. <i>Journal of Medical Genetics</i> , 1993, 30, 675-678.	1.5	27
96	Genetic mapping of the Kallmann syndrome and X linked ocular albinism gene loci.. <i>Journal of Medical Genetics</i> , 1993, 30, 923-925.	1.5	6
97	Presymptomatic diagnosis of von Hippel-Lindau disease with flanking DNA markers.. <i>Journal of Medical Genetics</i> , 1992, 29, 902-905.	1.5	22
98	Incidence and expression of the N1303K mutation of the cystic fibrosis (CFTR) gene. <i>Human Genetics</i> , 1992, 89, 653-658.	1.8	69
99	A melanocyte-specific gene, Pmel 17, maps near the silver coat color locus on mouse chromosome 10 and is in a syntenic region on human chromosome 12.. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 1991, 88, 9228-9232.	3.3	164
100	Mapping of Von Hippel-Lindau disease to chromosome 3p confirmed by genetic linkage analysis. <i>Journal of the Neurological Sciences</i> , 1990, 100, 27-30.	0.3	44
101	Blast-1 possesses a glycosyl-phosphatidylinositol (GPI) membrane anchor, is related to LFA-3 and OX-45, and maps to chromosome 1q21-23.. <i>Journal of Experimental Medicine</i> , 1989, 169, 1087-1099.	4.2	69
102	Two RFLPs associated with the human endogenous retroviral element S71 on chromosome 18q21. <i>Nucleic Acids Research</i> , 1989, 17, 2367-2367.	6.5	2
103	Isolation, Chromosomal Mapping, and Expression of the Mouse Tyrosinase Gene. <i>Journal of Investigative Dermatology</i> , 1989, 93, 589-594.	0.3	58
104	Human SSAV-related endogenous retroviral element: LTR-like sequence and chromosomal localization to 18q21. <i>Genomics</i> , 1989, 4, 68-75.	1.3	32
105	Level of expression and chromosome mapping of the mouse cholecystokinin gene: Implications for murine models of genetic obesity. <i>Genomics</i> , 1989, 5, 463-469.	1.3	25
106	Mapping of genes for inhibin subunits $\hat{I}\pm$, \hat{I}^2A , and \hat{I}^2B on human and mouse chromosomes and studies of jsd mice. <i>Genomics</i> , 1989, 5, 91-99.	1.3	95
107	Chromosome mapping of the growth hormone receptor gene in man and mouse. <i>Cytogenetic and Genome Research</i> , 1989, 50, 137-141.	0.6	88
108	Chromosomal localization of the gene for the human trifunctional enzyme, methylenetetrahydrofolate dehydrogenase-methenyltetrahydrofolate cyclohydrolase-formyltetrahydrofolate synthetase. <i>American Journal of Human Genetics</i> , 1989, 44, 781-6.	2.6	14

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109	Assignment of the T-cell differentiation gene MAL to human chromosome 2, region cen?q13. Immunogenetics, 1988, 27, 91-95.	1.2	28
110	Human tyrosinase gene, mapped to chromosome 11 (q14 â†’ q21), defines second region of homology with mouse chromosome 7. Genomics, 1988, 3, 17-24.	1.3	170
111	Chromosomal mapping of the gene for the type II insulin-like growth factor receptor/cation-independent mannose 6-phosphate receptor in man and mouse. Genomics, 1988, 3, 224-229.	1.3	78
112	Identification of a cDNA encoding a parathyroid hormone-like peptide from a human tumor associated with humoral hypercalcemia of malignancy.. Proceedings of the National Academy of Sciences of the United States of America, 1988, 85, 597-601.	3.3	415
113	Molecular characterization and chromosomal mapping of melanoma growth stimulatory activity, a growth factor structurally related to beta-thromboglobulin. EMBO Journal, 1988, 7, 2025-33.	3.5	105
114	Chromosomal mapping of genes for transforming growth factors beta 2 and beta 3 in man and mouse: dispersion of TGF-beta gene family. Oncogene Research, 1988, 3, 323-31.	1.2	64
115	The human U1-70K snRNP protein: cDNA cloning, chromosomal localization, expression, alternative splicing and RNA-binding. Nucleic Acids Research, 1987, 15, 10373-10391.	6.5	154
116	Ribonucleotide reductase M2 subunit sequences mapped to four different chromosomal sites in humans and mice: Functional locus identified by its amplification in hydroxyurea-resistant cell lines. Genomics, 1987, 1, 77-86.	1.3	62
117	The myelin-associated glycoprotein gene: Mapping to human chromosome 19 and mouse chromosome 7 and expression in quivering mice. Genomics, 1987, 1, 107-112.	1.3	68
118	Isolation of cDNA clones coding for rat isovaleryl-CoA dehydrogenase and assignment of the gene to human chromosome 15. Genomics, 1987, 1, 264-269.	1.3	24
119	Activation of human? 1,-antitrypsin genes in rat hepatoma i;1/2 human fibroblast hybrid cell lines is correlated with demethylation. Somatic Cell and Molecular Genetics, 1987, 13, 635-644.	0.7	9
120	The human tyrosine aminotransferase gene mapped to the long arm of chromosome 16 (region) Tj ETQq0 0 0 rgBT/Qverlock 10 Tf 50 3	1.8	45
121	Isolation of a polymorphic DNA segment unique to human chromosome 7 by molecular cloning of hybrid cell DNA. Molecular Genetics and Genomics, 1983, 190, 143-149.	2.4	21