

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
g-index

127
all docs

127
docs citations

127
times ranked

6509
citing authors

#	ARTICLE	IF	CITATIONS
1	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
2	Genome-Wide Association Study in BRCA1 Mutation Carriers Identifies Novel Loci Associated with Breast and Ovarian Cancer Risk. PLoS Genetics, 2013, 9, e1003212.	1.5	244
3	Mutations at the mitochondrial DNA polymerase (POLG) locus associated with male infertility. Nature Genetics, 2001, 29, 261-262.	9.4	173
4	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
5	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.. Proceedings of the National Academy of Sciences of the United States of America, 1991, 88, 9228-9232.	3.3	164
6	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
7	The copy number variation landscape of congenital anomalies of the kidney and urinary tract. Nature Genetics, 2019, 51, 117-127.	9.4	144
8	Mutational bias provides a model for the evolution of Huntington's disease and predicts a general increase in disease prevalence. Nature Genetics, 1994, 7, 525-530.	9.4	141
9	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. Human Molecular Genetics, 1993, 2, 1713-1715.	1.4	125
10	Quality control in molecular genetic testing. Nature Reviews Genetics, 2001, 2, 717-723.	7.7	120
11	Genetic Drivers of Kidney Defects in the DiGeorge Syndrome. New England Journal of Medicine, 2017, 376, 742-754.	13.9	120
12	The detection of large deletions or duplications in genomic DNA. Human Mutation, 2002, 20, 325-337.	1.1	110
13	Recommendations for reporting results of diagnostic genetic testing (biochemical, cytogenetic and) Tj ETQq1 1 0.784314 rgBT /Over 1.4 109	1.4	109
14	Identification of a BRCA2-Specific Modifier Locus at 6p24 Related to Breast Cancer Risk. PLoS Genetics, 2013, 9, e1003173.	1.5	105
15	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
16	Somatic NF2 gene mutations in familial and non-familial vestibular schwannoma. Human Molecular Genetics, 1994, 3, 347-350.	1.4	102
17	The Exon 13 Duplication in the BRCA1 Gene Is a Founder Mutation Present in Geographically Diverse Populations. American Journal of Human Genetics, 2000, 67, 207-212.	2.6	100
18	Mapping of genes for inhibin subunits Î±, Î²A, and Î²B on human and mouse chromosomes and studies of jsd mice. Genomics, 1989, 5, 91-99.	1.3	95

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19	Stickler syndrome: Correlation between vitreoretinal phenotypes and linkage to COL 2A1. <i>Eye</i> , 1994, 8, 609-614.	1.1	90
20	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
21	Mosaic uniparental disomy in Beckwith-Wiedemann syndrome.. <i>Journal of Medical Genetics</i> , 1994, 31, 749-753.	1.5	83
22	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
23	Chromosomal mapping of the gene for the type II insulin-like growth factor receptor/cation-independent mannose 6-phosphate receptor in man and mouse. <i>Genomics</i> , 1988, 3, 224-229.	1.3	78
24	Benchmarks for Cystic Fibrosis carrier screening: A European consensus document. <i>Journal of Cystic Fibrosis</i> , 2010, 9, 165-178.	0.3	75
25	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
26	Incidence and expression of the N1303K mutation of the cystic fibrosis (CFTR) gene. <i>Human Genetics</i> , 1992, 89, 653-658.	1.8	69
27	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
28	The myelin-associated glycoprotein gene: Mapping to human chromosome 19 and mouse chromosome 7 and expression in quivering mice. <i>Genomics</i> , 1987, 1, 107-112.	1.3	68
29	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
30	Chromosomal mapping of genes for transforming growth factors beta 2 and beta 3 in man and mouse: dispersion of TGF-beta gene family. <i>Oncogene Research</i> , 1988, 3, 323-31.	1.2	64
31	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
32	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. <i>Genomics</i> , 1987, 1, 77-86.	1.3	62
33	Hyperinsulinism: molecular aetiology of focal disease. <i>Archives of Disease in Childhood</i> , 1998, 79, 445-447.	1.0	62
34	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
35	Isolation, Chromosomal Mapping, and Expression of the Mouse Tyrosinase Gene. <i>Journal of Investigative Dermatology</i> , 1989, 93, 589-594.	0.3	58
36	Fragile X-associated tremor/ataxia syndrome presenting in a woman after chemotherapy. <i>Neurology</i> , 2005, 65, 331-332.	1.5	49

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37	Developing a Sustainable Process to Provide Quality Control Materials for Genetic Testing. <i>Genetics in Medicine</i> , 2005, 7, 534-549.	1.1	49
38	Confirmation of FWT1 as a Wilms's tumour susceptibility gene and phenotypic characteristics of Wilms's tumour attributable to FWT1. <i>Human Genetics</i> , 1998, 103, 547-556.	1.8	48
39	The human tyrosine aminotransferase gene mapped to the long arm of chromosome 16 (region Tj ETQq1 1 0.784314 rgBT /Overlock	1.8	45
40	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
41	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
42	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
43	Haplotype analysis of the \hat{r}^{2642} and (CAC) $_n$ 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
44	Emerging technologies for point-of-care genetic testing. <i>Expert Review of Molecular Diagnostics</i> , 2007, 7, 359-370.	1.5	44
45	A frequent hMSH2 mutation in hereditary non-polyposis colon cancer syndrome. <i>Lancet, The</i> , 1995, 345, 727.	6.3	42
46	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
47	EMQN Best Practice Guidelines for molecular genetic testing of SCAs. <i>European Journal of Human Genetics</i> , 2010, 18, 1173-1176.	1.4	41
48	A genome-wide scan for genes involved in primary vesicoureteric reflux. <i>Journal of Medical Genetics</i> , 2007, 44, 710-717.	1.5	39
49	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
50	Cystic fibrosis mutation analysis: Report from 22 U.K. regional genetics laboratories. <i>Human Mutation</i> , 1995, 6, 326-333.	1.1	38
51	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
52	Site of (CCG) polymorphism in the HD gene. <i>Nature Genetics</i> , 1993, 5, 214-215.	9.4	35
53	Fine-resolution mapping by haplotype evaluation: the examples of PFIC1 and BRIC. <i>Human Genetics</i> , 1999, 104, 241-248.	1.8	33
54	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

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55	Predictive testing for BRCA1 and 2 mutations: a male contribution. <i>Annals of Oncology</i> , 2003, 14, 549-553.	0.6	33
56	Genetics of vesicoureteral reflux. <i>Nature Reviews Urology</i> , 2011, 8, 539-552.	1.9	33
57	Human SSAV-related endogenous retroviral element: LTR-like sequence and chromosomal localization to 18q21. <i>Genomics</i> , 1989, 4, 68-75.	1.3	32
58	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
59	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
60	Alterations in the steroid hormone receptor co-chaperone FKBP1 are associated with male infertility: a case-control study. <i>Reproductive Biology and Endocrinology</i> , 2010, 8, 22.	1.4	31
61	Urinary Tract Effects of HPSE2 Mutations. <i>Journal of the American Society of Nephrology: JASN</i> , 2015, 26, 797-804.	3.0	31
62	Genotype analysis in cystic fibrosis in relation to the occurrence of diabetes mellitus. <i>Clinical Genetics</i> , 1993, 43, 186-189.	1.0	30
63	Chimaerism shown by cytogenetics and DNA polymorphism analysis.. <i>Journal of Medical Genetics</i> , 1994, 31, 816-817.	1.5	29
64	Assignment of the T-cell differentiation gene MAL to human chromosome 2, region cen?q13. <i>Immunogenetics</i> , 1988, 27, 91-95.	1.2	28
65	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
66	Genetic linkage analysis in hereditary non-polyposis colon cancer syndrome.. <i>Journal of Medical Genetics</i> , 1995, 32, 352-357.	1.5	26
67	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
68	Isolation of cDNA clones coding for rat isovaleryl-CoA dehydrogenase and assignment of the gene to human chromosome 15. <i>Genomics</i> , 1987, 1, 264-269.	1.3	24
69	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
70	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
71	Presymptomatic diagnosis of von Hippel-Lindau disease with flanking DNA markers.. <i>Journal of Medical Genetics</i> , 1992, 29, 902-905.	1.5	22
72	Isolation of a polymorphic DNA segment unique to human chromosome 7 by molecular cloning of hybrid cell DNA. <i>Molecular Genetics and Genomics</i> , 1983, 190, 143-149.	2.4	21

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73	Genetic Mapping of X-Linked Ocular Albinism: Linkage Analysis in a Large Newfoundland Kindred. <i>Genomics</i> , 1993, 16, 259-261.	1.3	21
74	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
75	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
76	Report of an International Survey of Molecular Genetic Testing Laboratories. <i>Public Health Genomics</i> , 2007, 10, 123-131.	0.6	20
77	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
78	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
79	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
80	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
81	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
82	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
83	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
84	Familial Vesicoureteral Reflux: Influence of Sex on Prevalence and Expression. <i>Journal of Urology</i> , 2006, 176, 1776-1780.	0.2	16
85	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
86	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
87	Improved genetic mapping of X linked retinoschisis.. <i>Journal of Medical Genetics</i> , 1996, 33, 919-922.	1.5	13
88	B37 repeats are normal in most schizophrenic patients. <i>British Journal of Psychiatry</i> , 1994, 164, 851-852.	1.7	12
89	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
90	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

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91	Mosaicism for trisomy 3q arising from an unbalanced, de novo t(3;15).. Journal of Medical Genetics, 1997, 34, 512-514.	1.5	11
92	Linkage Analysis of Candidate Genes in Families With Vesicoureteral Reflux. Journal of Urology, 2009, 182, 1669-1672.	0.2	11
93	Evaluation and use of a synthetic quality control material, included in the European external quality assessment scheme for cystic fibrosis. Human Mutation, 2008, 29, 1063-1070.	1.1	10
94	The increased incidence of the RET p.Gly691Ser variant in French-Canadian vesicoureteric reflux patients is not replicated by a larger study in Ireland. Human Mutation, 2009, 30, E612-E617.	1.1	10
95	Activation of human α 1-antitrypsin genes in rat hepatoma \times 1/2 human fibroblast hybrid cell lines is correlated with demethylation. Somatic Cell and Molecular Genetics, 1987, 13, 635-644.	0.7	9
96	Carrier detection in X linked ocular albinism using linked DNA polymorphisms.. British Journal of Ophthalmology, 1994, 78, 539-541.	2.1	9
97	Diagnostic molecular genetics comes of age. Trends in Genetics, 1997, 13, 249-250.	2.9	9
98	Molecular genetic analysis of exons 1 to 6 of the APC gene in non- FAP polyposis familial colorectal cancer. Clinical Genetics, 1995, 48, 299-303.	1.0	8
99	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. Eye and Vision (London, England) 2014; 1(1): 1-14	0.78	14
100	Genetic mapping of the Kallmann syndrome and X linked ocular albinism gene loci.. Journal of Medical Genetics, 1993, 30, 923-925.	1.5	6
101	Detection of Five Common CFTR Mutations by Rapid-Cycle Real-Time Amplification Refractory Mutation System PCR. Clinical Chemistry, 2004, 50, 773-775.	1.5	6
102	Managing uncertainty in inherited cardiac pathologies – an international multidisciplinary survey. European Journal of Human Genetics, 2019, 27, 1178-1185.	1.4	6
103	Normal CAG and CCG repeats in the Huntington's disease genes of Parkinson's disease patients. American Journal of Medical Genetics Part A, 1995, 60, 109-110.	2.4	4
104	Instability of normal (CTG) $_n$ alleles in the DM kinase gene.. Journal of Medical Genetics, 1997, 34, 871-873.	1.5	4
105	Clinical correlation and molecular evaluation confirm that the MLH1 p.Arg182Gly (c.544A>G) mutation is pathogenic and causes Lynch syndrome. Familial Cancer, 2012, 11, 509-518.	0.9	4
106	Incidence of Fragile X syndrome in Ireland. American Journal of Medical Genetics, Part A, 2017, 173, 678-683.	0.7	4
107	National Newborn Screening for cystic fibrosis in the Republic of Ireland: genetic data from the first 6.5 years. European Journal of Human Genetics, 2020, 28, 1669-1674.	1.4	4
108	Ret-proto-oncogene analysis in medullary thyroid carcinoma. Irish Journal of Medical Science, 1998, 167, 226-230.	0.8	3

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109	Retaining the Confidence of the Public in Molecular Genetic Testing – Quality Assurance. Public Health Genomics, 2000, 3, 164-169.	0.6	3
110	Two RFLPs associated with the human endogenous retroviral element S71 on chromosome 18q21. Nucleic Acids Research, 1989, 17, 2367-2367.	6.5	2
111	When good CF tests go bad. European Journal of Human Genetics, 2009, 17, 403-405.	1.4	2
112	Certified Reference Materials for Genetic Testing. , 2004, , 226-231.		2
113	Investigation of DNA variants specific to ROBO2 Isoform –™ in Irish vesicoureteric reflux patients reveals marked CpG island variation. Scientific Reports, 2020, 10, 2265.	1.6	2
114	Cystic fibrosis diagnosed by molecular genetic investigation in the mother of a patient with cystic fibrosis. Thorax, 1997, 52, 96-7.	2.7	2
115	Targeted versus Whole-Genome Array Comparative Genome Hybridization. Journal of Molecular Diagnostics, 2007, 9, 278.	1.2	1
116	Multiallelic Synthetic Quality Control Material: Lessons Learned from the Cystic Fibrosis External Quality Assessment Scheme. Genetic Testing and Molecular Biomarkers, 2011, 15, 579-586.	0.3	1
117	Reply to Sajantila and Budowle. European Journal of Human Genetics, 2016, 24, 330-330.	1.4	1
118	National scientific medical meeting 1997 abstracts. Irish Journal of Medical Science, 1998, 167, 1-44.	0.8	0
119	Sylvester –™halloran surgical scientific meeting. Irish Journal of Medical Science, 1998, 167, 1-16.	0.8	0
120	Cross-border genetic testing. Orphanet Journal of Rare Diseases, 2010, 5, .	1.2	0
121	Genetic Services in Ireland. European Journal of Human Genetics, 1997, 5, 100-104.	1.4	0