David E Barton

List of Publications by Year
in descending order

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121 papers	5,680 citations	61984 43 h-index	82547 72 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.	7.1	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.	3.5	244
3	Mutations at the mitochondrial DNA polymerase (POLG) locus associated with male infertility. Nature Genetics, 2001, 29, 261-262.	21.4	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.	2.9	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.	7.1	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.	14.5	154
7	The copy number variation landscape of congenital anomalies of the kidney and urinary tract. Nature Genetics, 2019, 51, 117-127.	21.4	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.	21.4	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.	2.9	125
10	Quality control in molecular genetic testing. Nature Reviews Genetics, 2001, 2, 717-723.	16.3	120
11	Genetic Drivers of Kidney Defects in the DiGeorge Syndrome. New England Journal of Medicine, 2017, 376, 742-754.	27.0	120
12	The detection of large deletions or duplications in genomic DNA. Human Mutation, 2002, 20, 325-337.	2.5	110
13	Recommendations for reporting results of diagnostic genetic testing (biochemical, cytogenetic and) Tj ETQq1 1 0.784314 rgBT /Over	2.8	109
14	Identification of a BRCA2-Specific Modifier Locus at 6p24 Related to Breast Cancer Risk. PLoS Genetics, 2013, 9, e1003173.	3.5	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.	7.8	105
16	Somatic NF2 gene mutations in familial and non-familial vestibular schwannoma. Human Molecular Genetics, 1994, 3, 347-350.	2.9	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.	6.2	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.	2.9	95

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19	Stickler syndrome: Correlation between vitreoretinal phenotypes and linkage to COL 2A1. Eye, 1994, 8, 609-614.	2.1	90
20	Chromosome mapping of the growth hormone receptor gene in man and mouse. Cytogenetic and Genome Research, 1989, 50, 137-141.	1.1	88
21	Mosaic uniparental disomy in Beckwith-Wiedemann syndrome.. Journal of Medical Genetics, 1994, 31, 749-753.	3.2	83
22	Hyperinsulinism caused by paternal-specific inheritance of a recessive mutation in the sulfonylurea-receptor gene. Diabetes, 1999, 48, 1652-1657.	0.6	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. Genomics, 1988, 3, 224-229.	2.9	78
24	Benchmarks for Cystic Fibrosis carrier screening: A European consensus document. Journal of Cystic Fibrosis, 2010, 9, 165-178.	0.7	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.. Journal of Experimental Medicine, 1989, 169, 1087-1099.	8.5	69
26	Incidence and expression of the N1303K mutation of the cystic fibrosis (CFTR) gene. Human Genetics, 1992, 89, 653-658.	3.8	69
27	Molecular genetic investigation of the neurofibromatosis type 2 tumor suppressor gene in sporadic meningioma. Journal of Neurosurgery, 1996, 84, 847-851.	1.6	69
28	The myelin-associated glycoprotein gene: Mapping to human chromosome 19 and mouse chromosome 7 and expression in quivering mice. Genomics, 1987, 1, 107-112.	2.9	68
29	Common alleles at 6q25.1 and 1p11.2 are associated with breast cancer risk for BRCA1 and BRCA2 mutation carriers. Human Molecular Genetics, 2011, 20, 3304-3321.	2.9	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. Oncogene Research, 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. Cancer Epidemiology Biomarkers and Prevention, 2007, 16, 740-746.	2.5	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. Genomics, 1987, 1, 77-86.	2.9	62
33	Hyperinsulinism: molecular aetiology of focal disease. Archives of Disease in Childhood, 1998, 79, 445-447.	1.9	62
34	EMQN/CMGS best practice guidelines for the molecular genetic testing of Huntington disease. European Journal of Human Genetics, 2013, 21, 480-486.	2.8	59
35	Isolation, Chromosomal Mapping, and Expression of the Mouse Tyrosinase Gene. Journal of Investigative Dermatology, 1989, 93, 589-594.	0.7	58
36	Fragile X-associated tremor/ataxia syndrome presenting in a woman after chemotherapy. Neurology, 2005, 65, 331-332.	1.1	49

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37	Developing a Sustainable Process to Provide Quality Control Materials for Genetic Testing. Genetics in Medicine, 2005, 7, 534-549.	2.4	49
38	Confirmation of FWT1 as a Wilmsâ€™ tumour susceptibility gene and phenotypic characteristics of Wilmsâ€™ tumour attributable to FWT1. Human Genetics, 1998, 103, 547-556.	3.8	48
39	The human tyrosine aminotransferase gene mapped to the long arm of chromosome 16 (region) Tj ETQq1 1 0.784314 rgBT /Overlock	3.8	45
40	Estimating the age of p.(Phe508del) with family studies of geographically distinct European populations and the early spread of cystic fibrosis. European Journal of Human Genetics, 2018, 26, 1832-1839.	2.8	45
41	Mapping of Von Hippel-Lindau disease to chromosome 3p confirmed by genetic linkage analysis. Journal of the Neurological Sciences, 1990, 100, 27-30.	0.6	44
42	Direct, non-radioactive detection of mutations in Multiple Endocrine Neoplasia Type 2A families. Human Molecular Genetics, 1994, 3, 643-646.	2.9	44
43	Haplotype analysis of the Î”2642 and (CAG)n polymorphisms in the Huntington's disease (HD) gene provides an explanation for an apparent â€˜founderâ€™ HD haplotype. Human Molecular Genetics, 1995, 4, 203-206.	2.9	44
44	Emerging technologies for point-of-care genetic testing. Expert Review of Molecular Diagnostics, 2007, 7, 359-370.	3.1	44
45	A frequent hMSH2 mutation in hereditary non-polyposis colon cancer syndrome. Lancet, The, 1995, 345, 727.	13.7	42
46	Typical Friedreich's ataxia without GAA expansions and GAA expansions without typical Friedreich's ataxia. Journal of Neurology, 2000, 247, 346-355.	3.6	41
47	EMQN Best Practice Guidelines for molecular genetic testing of SCAs. European Journal of Human Genetics, 2010, 18, 1173-1176.	2.8	41
48	A genome-wide scan for genes involved in primary vesicoureteric reflux. Journal of Medical Genetics, 2007, 44, 710-717.	3.2	39
49	Cystic fibrosis identified by neonatal screening: incidence, genotype, and early natural history.. Archives of Disease in Childhood, 1993, 68, 464-467.	1.9	38
50	Cystic fibrosis mutation analysis: Report from 22 U.K. regional genetics laboratories. Human Mutation, 1995, 6, 326-333.	2.5	38
51	The relationship between Y chromosome DNA haplotypes and Y chromosome deletions leading to male infertility. Human Genetics, 2001, 108, 55-58.	3.8	36
52	Site of (CCG) polymorphism in the HD gene. Nature Genetics, 1993, 5, 214-215.	21.4	35
53	Fine-resolution mapping by haplotype evaluation: the examples of PFIC1 and BRIC. Human Genetics, 1999, 104, 241-248.	3.8	33
54	Angiotensin II Type 2 Receptor Gene is Not Responsible For Familial Vesicoureteral Reflux. Journal of Urology, 2002, 168, 1138-1141.	0.4	33

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55	Predictive testing for BRCA1 and 2 mutations: a male contribution. Annals of Oncology, 2003, 14, 549-553.	1.2	33
56	Genetics of vesicoureteral reflux. Nature Reviews Urology, 2011, 8, 539-552.	3.8	33
57	Human SSAV-related endogenous retroviral element: LTR-like sequence and chromosomal localization to 18q21. Genomics, 1989, 4, 68-75.	2.9	32
58	Uroplakin III is not a major candidate gene for primary vesicoureteral reflux. European Journal of Human Genetics, 2005, 13, 500-502.	2.8	32
59	Common variants of the BRCA1 wild-type allele modify the risk of breast cancer in BRCA1 mutation carriers. Human Molecular Genetics, 2011, 20, 4732-4747.	2.9	32
60	Alterations in the steroid hormone receptor co-chaperone FKBPL are associated with male infertility: a case-control study. Reproductive Biology and Endocrinology, 2010, 8, 22.	3.3	31
61	Urinary Tract Effects of HPSE2 Mutations. Journal of the American Society of Nephrology: JASN, 2015, 26, 797-804.	6.1	31
62	Genotype analysis in cystic fibrosis in relation to the occurrence of diabetes mellitus. Clinical Genetics, 1993, 43, 186-189.	2.0	30
63	Chimaerism shown by cytogenetics and DNA polymorphism analysis.. Journal of Medical Genetics, 1994, 31, 816-817.	3.2	29
64	Assignment of the T-cell differentiation gene MAL to human chromosome 2, region cen?q13. Immunogenetics, 1988, 27, 91-95.	2.4	28
65	Evaluation of molecular genetic diagnosis in the management of familial adenomatous polyposis coli: a population based study.. Journal of Medical Genetics, 1993, 30, 675-678.	3.2	27
66	Genetic linkage analysis in hereditary non-polyposis colon cancer syndrome.. Journal of Medical Genetics, 1995, 32, 352-357.	3.2	26
67	Level of expression and chromosome mapping of the mouse cholecystokinin gene: Implications for murine models of genetic obesity. Genomics, 1989, 5, 463-469.	2.9	25
68	Isolation of cDNA clones coding for rat isovaleryl-CoA dehydrogenase and assignment of the gene to human chromosome 15. Genomics, 1987, 1, 264-269.	2.9	24
69	Quality assurance practices in Europe: a survey of molecular genetic testing laboratories. European Journal of Human Genetics, 2012, 20, 1118-1126.	2.8	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. Molecular Genetics & Genomic Medicine, 2014, 2, 7-29.	1.2	23
71	Presymptomatic diagnosis of von Hippel-Lindau disease with flanking DNA markers.. Journal of Medical Genetics, 1992, 29, 902-905.	3.2	22
72	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

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73	Genetic Mapping of X-Linked Ocular Albinism: Linkage Analysis in a Large Newfoundland Kindred. Genomics, 1993, 16, 259-261.	2.9	21
74	A comparison of methods for gene dosage analysis in HMSN type 1. Journal of Medical Genetics, 2001, 38, 90-95.	3.2	21
75	Best practice guidelines for the molecular genetic diagnosis of Type 1 (HFE-related) hereditary haemochromatosis. BMC Medical Genetics, 2006, 7, 81.	2.1	20
76	Report of an International Survey of Molecular Genetic Testing Laboratories. Public Health Genomics, 2007, 10, 123-131.	1.0	20
77	Preparation and validation of the first WHO international genetic reference panel for Fragile X syndrome. European Journal of Human Genetics, 2011, 19, 10-17.	2.8	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.. Journal of Medical Genetics, 1994, 31, 690-693.	3.2	19
79	Intrafamilial Phenotypic Variability in Friedreich Ataxia Associated With a G130V Mutation in the FRDA Gene. Archives of Neurology, 2002, 59, 296.	4.5	17
80	Genome-wide linkage and association study implicates the 10q26 region as a major genetic contributor to primary nonsyndromic vesicoureteric reflux. Scientific Reports, 2017, 7, 14595.	3.3	17
81	Copy Number Variant Analysis and Genome-wide Association Study Identify Loci with Large Effect for Vesicoureteral Reflux. Journal of the American Society of Nephrology: JASN, 2021, 32, 805-820.	6.1	17
82	Myotonic dystrophy CTG repeats and the associated insertion/deletion polymorphism in human and primate populations. Human Molecular Genetics, 1994, 3, 2031-5.	2.9	17
83	Angiotensin II type 2 receptor gene is not responsible for familial vesicoureteral reflux. Journal of Urology, 2002, 168, 1138-41.	0.4	17
84	Familial Vesicoureteral Reflux: Influence of Sex on Prevalence and Expression. Journal of Urology, 2006, 176, 1776-1780.	0.4	16
85	All azoospermic males should be screened for cystic fibrosis mutations before intracytoplasmic sperm injection. Fertility and Sterility, 2010, 94, 2448-2450.	1.0	14
86	Chromosomal localization of the gene for the human trifunctional enzyme, methylenetetrahydrofolate dehydrogenase-methenyltetrahydrofolate cyclohydrolase-formyltetrahydrofolate synthetase. American Journal of Human Genetics, 1989, 44, 781-6.	6.2	14
87	Improved genetic mapping of X linked retinoschisis.. Journal of Medical Genetics, 1996, 33, 919-922.	3.2	13
88	B37 repeats are normal in most schizophrenic patients. British Journal of Psychiatry, 1994, 164, 851-852.	2.8	12
89	Establishment of the first WHO international genetic reference panel for Prader Willi and Angelman syndromes. European Journal of Human Genetics, 2011, 19, 857-864.	2.8	12
90	Heterozygous non-synonymous ROBO2 variants are unlikely to be sufficient to cause familial vesicoureteric reflux. Kidney International, 2013, 84, 327-337.	5.2	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.	3.2	11
92	Linkage Analysis of Candidate Genes in Families With Vesicoureteral Reflux. Journal of Urology, 2009, 182, 1669-1672.	0.4	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.	2.5	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.	2.5	10
95	Activation of human? 1,-antitrypsin genes in rat hepatoma ï¿½ 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.	3.9	9
97	Diagnostic molecular genetics comes of age. Trends in Genetics, 1997, 13, 249-250.	6.7	9
98	Molecular genetic analysis of exons 1 to 6 of the APC gene in nonâ€polyposis familial colorectal cancer. Clinical Genetics, 1995, 48, 299-303.	2.0	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,) Tj ETQq1 1 0.784314 rgBT /Overlock	4.3	7
100	Genetic mapping of the Kallmann syndrome and X linked ocular albinism gene loci.. Journal of Medical Genetics, 1993, 30, 923-925.	3.2	6
101	Detection of Five Common CFTR Mutations by Rapid-Cycle Real-Time Amplification Refractory Mutation System PCR. Clinical Chemistry, 2004, 50, 773-775.	3.2	6
102	Managing uncertainty in inherited cardiac pathologiesâ€"an international multidisciplinary survey. European Journal of Human Genetics, 2019, 27, 1178-1185.	2.8	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.	3.2	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.	1.9	4
106	Incidence of Fragile X syndrome in Ireland. American Journal of Medical Genetics, Part A, 2017, 173, 678-683.	1.2	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.	2.8	4
108	Ret-proto-oncogene analysis in medullary thyroid carcinoma. Irish Journal of Medical Science, 1998, 167, 226-230.	1.5	3

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109	Retaining the Confidence of the Public in Molecular Genetic Testing â€“ Quality Assurance. Public Health Genomics, 2000, 3, 164-169.	1.0	3
110	Two RFLPs associated with the human endogenous retroviral element S71 on chromosome 18q21. Nucleic Acids Research, 1989, 17, 2367-2367.	14.5	2
111	When good CF tests go bad. European Journal of Human Genetics, 2009, 17, 403-405.	2.8	2
112	Certified Reference Materials for Genetic Testing. , 2004, , 226-231.		2
113	Investigation of DNA variants specific to ROBO2 Isoform â€˜aâ€™ in Irish vesicoureteric reflux patients reveals marked CpG island variation. Scientific Reports, 2020, 10, 2265.	3.3	2
114	Cystic fibrosis diagnosed by molecular genetic investigation in the mother of a patient with cystic fibrosis. Thorax, 1997, 52, 96-7.	5.6	2
115	Targeted versus Whole-Genome Array Comparative Genome Hybridization. Journal of Molecular Diagnostics, 2007, 9, 278.	2.8	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.	0.7	1
117	Reply to Sajantila and Budowle. European Journal of Human Genetics, 2016, 24, 330-330.	2.8	1
118	National scientific medical meeting 1997 abstracts. Irish Journal of Medical Science, 1998, 167, 1-44.	1.5	0
119	Sylvester oâ€™halloran surgical scientific meeting. Irish Journal of Medical Science, 1998, 167, 1-16.	1.5	0
120	Cross-border genetic testing. Orphanet Journal of Rare Diseases, 2010, 5, .	2.7	0
121	Genetic Services in Ireland. European Journal of Human Genetics, 1997, 5, 100-104.	2.8	0