Glenda M Beaman

List of Publications by Year
in descending order

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136 papers	8,845 citations	94433 37 h-index	53230 85 g-index
139 all docs	139 docs citations	139 times ranked	17163 citing authors

#	Article	IF	Citations
1	High likelihood of actionable pathogenic variant detection in breast cancer genes in women with very early onset breast cancer. Journal of Medical Genetics, 2022, 59, 115-121.	3.2	13
2	Extended gene panel testing in lobular breast cancer. Familial Cancer, 2022, 21, 129-136.	1.9	1
3	Clinical Pharmacogenetics Implementation Consortium Guideline for the Use of Aminoglycosides Based on MTâ€RNR1 Genotype. Clinical Pharmacology and Therapeutics, 2022, 111, 366-372.	4.7	50
4	New insights into Perrault syndrome, a clinically and genetically heterogeneous disorder. Human Genetics, 2022, 141, 805-819.	3.8	19
5	The importance of ethnicity: Are breast cancer polygenic risk scores ready for women who are not of White European origin?. International Journal of Cancer, 2022, 150, 73-79.	5.1	24
6	Dominantâ€negative pathogenic variant <scp>BRIP1</scp> c.<scp>1045G</scp>>C is a highâ€risk allele for nonâ€mucinous epithelial ovarian cancer: A caseâ€control study. Clinical Genetics, 2022, 101, 48-54.	2.0	3
7	Expanding the genotypic spectrum of <scp>TXNL4A</scp> variants in <scp>Burnâ€McKeown</scp> syndrome. Clinical Genetics, 2022, 101, 255-259.	2.0	5
8	Common variants in breast cancer risk loci predispose to distinct tumor subtypes. Breast Cancer Research, 2022, 24, 2.	5.0	15
9	MRSD: A quantitative approach for assessing suitability of RNA-seq in the investigation of mis-splicing in Mendelian disease. American Journal of Human Genetics, 2022, 109, 210-222.	6.2	12
10	Rapid Point-of-Care Genotyping to Avoid Aminoglycoside-Induced Ototoxicity in Neonatal Intensive Care. JAMA Pediatrics, 2022, 176, 486.	6.2	30
11	Genome-wide and transcriptome-wide association studies of mammographic density phenotypes reveal novel loci. Breast Cancer Research, 2022, 24, 27.	5.0	15
12	Characterization of the mechanism by which a nonsense variant in RYR2 leads to disordered calcium handling. Physiological Reports, 2022, 10, e15265.	1.7	7
13	Narrowing the chromosome 22q11.2 locus duplicated in bladder exstrophyâ€“epispadias complex. Journal of Pediatric Urology, 2022, 18, 362.e1-362.e8.	1.1	1
14	Genome-wide interaction analysis of menopausal hormone therapy use and breast cancer risk among 62,370 women. Scientific Reports, 2022, 12, 6199.	3.3	2
15	Breast cancer risk stratification in women of screening age: Incremental effects of adding mammographic density, polygenic risk, and a gene panel. Genetics in Medicine, 2022, 24, 1485-1494.	2.4	23
16	Breast cancer risks associated with missense variants in breast cancer susceptibility genes. Genome Medicine, 2022, 14, 51.	8.2	19
17	Integrating polygenic risk scores in the prediction of type 2 diabetes risk and subtypes in British Pakistanis and Bangladeshis: A population-based cohort study. PLoS Medicine, 2022, 19, e1003981.	8.4	24
18	Combined Associations of a Polygenic Risk Score and Classical Risk Factors With Breast Cancer Risk. Journal of the National Cancer Institute, 2021, 113, 329-337.	6.3	45

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19	CYP3A7*1C allele: linking premenopausal oestrone and progesterone levels with risk of hormone receptor-positive breast cancers. British Journal of Cancer, 2021, 124, 842-854.	6.4	5
20	The rise of point-of-care genetics: how the SARS-CoV-2 pandemic will accelerate adoption of genetic testing in the acute setting. European Journal of Human Genetics, 2021, 29, 891-893.	2.8	12
21	Breast Cancer Risk Genes â€" Association Analysis in More than 113,000 Women. New England Journal of Medicine, 2021, 384, 428-439.	27.0	532
22	Impaired eIF5A function causes a Mendelian disorder that is partially rescued in model systems by spermidine. Nature Communications, 2021, 12, 833.	12.8	41
23	Genomic and healthcare dynamics of nosocomial SARS-CoV-2 transmission. ELife, 2021, 10, .	6.0	35
24	Enzyme replacement therapy and hematopoietic stem cell transplant: a new paradigm of treatment in Wolman disease. Orphanet Journal of Rare Diseases, 2021, 16, 235.	2.7	18
25	A Nonadaptive Combinatorial Group Testing Strategy to Facilitate Health Care Worker Screening during the Severe Acute Respiratory Syndrome Coronavirus-2 (SARS-CoV-2) Outbreak. Journal of Molecular Diagnostics, 2021, 23, 532-540.	2.8	7
26	Clinical utility of testing for PALB2 and CHEK2 c.1100delC in breast and ovarian cancer. Genetics in Medicine, 2021, 23, 1969-1976.	2.4	8
27	Pathogenic Intronic Splice-Affecting Variants in MYBPC3 in Three Patients with Hypertrophic Cardiomyopathy. Neurology International, 2021, 11, 73-83.	0.5	1
28	Pharmacogenetics to Avoid Loss of Hearing (PALOH) trial: a protocol for a prospective observational implementation trial. BMJ Open, 2021, 11, e044457.	1.9	9
29	Bi-allelic premature truncating variants in LTBP1 cause cutis laxa syndrome. American Journal of Human Genetics, 2021, 108, 1095-1114.	6.2	19
30	The diagnostic utility of clinical exome sequencing in 60 patients with hearing loss disorders: A singleâ€institution experience. Clinical Otolaryngology, 2021, 46, 1257-1262.	1.2	3
31	The Genomic Architecture of Bladder Exstrophy Epispadias Complex. Genes, 2021, 12, 1149.	2.4	8
32	Biallelic loss of function variants in STAG3 result in primary ovarian insufficiency. Reproductive BioMedicine Online, 2021, 43, 899-902.	2.4	5
33	Association of germline genetic variants with breast cancer-specific survival in patient subgroups defined by clinic-pathological variables related to tumor biology and type of systemic treatment. Breast Cancer Research, 2021, 23, 86.	5.0	7
34	Mendelian randomisation study of smoking exposure in relation to breast cancer risk. British Journal of Cancer, 2021, 125, 1135-1145.	6.4	9
35	Gene Panel Testing for Breast Cancer Reveals Differential Effect of Prior BRCA1/2 Probability. Cancers, 2021, 13, 4154.	3.7	5
36	Breast Cancer Risk Factors and Survival by Tumor Subtype: Pooled Analyses from the Breast Cancer Association Consortium. Cancer Epidemiology Biomarkers and Prevention, 2021, 30, 623-642.	2.5	19

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37	Mosaic Fabry Disease in a Male Presenting as Hypertrophic Cardiomyopathy. Neurology International, 2021, 11, 1-9.	0.5	1
38	Comparison of in silico strategies to prioritize rare genomic variants impacting RNA splicing for the diagnosis of genomic disorders. Scientific Reports, 2021, 11, 20607.	3.3	37
39	Quantifying the Impact of Capacity Constraints in Economic Evaluations: An Application in Precision Medicine. Medical Decision Making, 2021, , 0272989X2110537.	2.4	1
40	Bi-allelic variants in the mitochondrial RNase P subunit PRORP cause mitochondrial tRNA processing defects and pleiotropic multisystem presentations. American Journal of Human Genetics, 2021, 108, 2195-2204.	6.2	26
41	Characterising a homozygous twoâ€exon deletion in UQCRH : comparing human and mouse phenotypes. EMBO Molecular Medicine, 2021, 13, e14397.	6.9	5
42	Inactivity of Peptidase ClpP Causes Primary Accumulation of Mitochondrial Disaggregase ClpX with Its Interacting Nucleoid Proteins, and of mtDNA. Cells, 2021, 10, 3354.	4.1	4
43	A caseâ€“control evaluation of 143 single nucleotide polymorphisms for breast cancer risk stratification with classical factors and mammographic density. International Journal of Cancer, 2020, 146, 2122-2129.	5.1	38
44	A recurrent missense variant in HARS2 results in variable sensorineural hearing loss in three unrelated families. Journal of Human Genetics, 2020, 65, 305-311.	2.3	5
45	Early B-cell Factor 3â€“Related Genetic Disease Can Mimic Urofacial Syndrome. Kidney International Reports, 2020, 5, 1823-1827.	0.8	7
46	Ligase IV syndrome can present with microcephaly and radial ray anomalies similar to Fanconi anaemia plus fatal kidney malformations. European Journal of Medical Genetics, 2020, 63, 103974.	1.3	5
47	SLC20A1 Is Involved in Urinary Tract and Urorectal Development. Frontiers in Cell and Developmental Biology, 2020, 8, 567.	3.7	22
48	Genetic testing in the acute setting: a round table discussion. Journal of Medical Ethics, 2020, 46, 533-533.	1.8	4
49	Breast Cancer Polygenic Risk Score and Contralateral Breast Cancer Risk. American Journal of Human Genetics, 2020, 107, 837-848.	6.2	39
50	Genome-wide association study identifies 32 novel breast cancer susceptibility loci from overall and subtype-specific analyses. Nature Genetics, 2020, 52, 572-581.	21.4	265
51	Classification and correlation of RYR2 missense variants in individuals with catecholaminergic polymorphic ventricular tachycardia reveals phenotypic relationships. Journal of Human Genetics, 2020, 65, 531-539.	2.3	20
52	Risk of Contralateral Breast Cancer in Women with and without Pathogenic Variants in BRCA1, BRCA2, and TP53 Genes in Women with Very Early-Onset (<36 Years) Breast Cancer. Cancers, 2020, 12, 378.	3.7	21
53	A network analysis to identify mediators of germline-driven differences in breast cancer prognosis. Nature Communications, 2020, 11, 312.	12.8	30
54	Genetic polymorphism in C3 is associated with progression in chronic kidney disease (CKD) patients with IgA nephropathy but not in other causes of CKD. PLoS ONE, 2020, 15, e0228101.	2.5	6

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55	EFTUD2 missense variants disrupt protein function and splicing in mandibulofacial dysostosis Guionâ€Almeida type. Human Mutation, 2020, 41, 1372-1382.	2.5	15
56	Pharmacogenomics in the UK National Health Service: opportunities and challenges. Pharmacogenomics, 2020, 21, 1237-1246.	1.3	15
57	Refusal of viral testing during the SARS-CoV-2 pandemic. Clinical Medicine, 2020, 20, e163-e164.	1.9	20
58	Title is missing!. , 2020, 15, e0228101.		0
59	Title is missing!. , 2020, 15, e0228101.		0
60	Title is missing!. , 2020, 15, e0228101.		0
61	Title is missing!. , 2020, 15, e0228101.		0
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64	Title is missing!. , 2020, 15, e0233582.		0
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66	Title is missing!. , 2020, 15, e0233582.		0
67	Title is missing!. , 2020, 15, e0233582.		0
68	Diagnosing and Preventing Hearing Loss in the Genomic Age. Trends in Hearing, 2019, 23, 233121651987898.	1.3	16
69	A homozygous missense variant in CHRM3 associated with familial urinary bladder disease. Clinical Genetics, 2019, 96, 515-520.	2.0	9
70	Two truncating variants in FANCC and breast cancer risk. Scientific Reports, 2019, 9, 12524.	3.3	5
71	Rare Variants in BNC2 Are Implicated in Autosomal-Dominant Congenital Lower Urinary-Tract Obstruction. American Journal of Human Genetics, 2019, 104, 994-1006.	6.2	47
72	Lrig2 and Hpse2, mutated in urofacial syndrome, pattern nerves in the urinary bladder. Kidney International, 2019, 95, 1138-1152.	5.2	25

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73	Breast cancer pathology and stage are better predicted by risk stratification models that include mammographic density and common genetic variants. Breast Cancer Research and Treatment, 2019, 176, 141-148.	2.5	56
74	Clinical and genetic heterogeneity in Melkersson-Rosenthal Syndrome. European Journal of Medical Genetics, 2019, 62, 103536.	1.3	8
75	Polygenic Risk Scores for Prediction of Breast Cancer and Breast Cancer Subtypes. American Journal of Human Genetics, 2019, 104, 21-34.	6.2	711
76	A comparative analysis of KMT2D missense variants in Kabuki syndrome, cancers and the general population. Journal of Human Genetics, 2019, 64, 161-170.	2.3	26
77	22q11.2 duplications in a UK cohort with bladder exstrophyâ€“epispadias complex. American Journal of Medical Genetics, Part A, 2019, 179, 404-409.	1.2	11
78	Whole Exome Sequencing Reveals the Major Genetic Contributors to Nonsyndromic Tetralogy of Fallot. Circulation Research, 2019, 124, 553-563.	4.5	118
79	Loss-of-function variants in myocardin cause congenital megabladder in humans and mice. Journal of Clinical Investigation, 2019, 129, 5374-5380.	8.2	27
80	Use of Single-Nucleotide Polymorphisms and Mammographic Density Plus Classic Risk Factors for Breast Cancer Risk Prediction. JAMA Oncology, 2018, 4, 476.	7.1	109
81	Histone Lysine Methylases and Demethylases in the Landscape of Human Developmental Disorders. American Journal of Human Genetics, 2018, 102, 175-187.	6.2	204
82	A Dominantly Inherited 5â€² UTR Variant Causing Methylation-Associated Silencing of BRCA1 as a Cause of Breast and Ovarian Cancer. American Journal of Human Genetics, 2018, 103, 213-220.	6.2	78
83	Structural insight into the human mitochondrial tRNA purine N1-methyltransferase and ribonuclease P complexes. Journal of Biological Chemistry, 2018, 293, 12862-12876.	3.4	28
84	Impact of a Panel of 88 Single Nucleotide Polymorphisms on the Risk of Breast Cancer in High-Risk Women: Results From Two Randomized Tamoxifen Prevention Trials. Journal of Clinical Oncology, 2017, 35, 743-750.	1.6	58
85	Genome-wide association study implicates immune activation of multiple integrin genes in inflammatory bowel disease. Nature Genetics, 2017, 49, 256-261.	21.4	943
86	Exploring the genetic architecture of inflammatory bowel disease by whole-genome sequencing identifies association at ADCY7. Nature Genetics, 2017, 49, 186-192.	21.4	153
87	ISL1 is a major susceptibility gene for classic bladder exstrophy and a regulator of urinary tract development. Scientific Reports, 2017, 7, 42170.	3.3	41
88	The impact of a panel of 18 SNPs on breast cancer risk in women attending a UK familial screening clinic: a caseâ€“control study. Journal of Medical Genetics, 2017, 54, 111-113.	3.2	56
89	First evidence of genotypeâ€“phenotype correlations in Gorlin syndrome. Journal of Medical Genetics, 2017, 54, 530-536.	3.2	56
90	Validation of copy number variation analysis for next-generation sequencing diagnostics. European Journal of Human Genetics, 2017, 25, 719-724.	2.8	72

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91	Response to: â€˜Mutation in MMP2 gene may result in scleroderma-like skin thickeningâ€™ by Bader-Meunieret al. Annals of the Rheumatic Diseases, 2016, 75, e2-e2.	0.9	1
92	Non lethal Raine syndrome and differential diagnosis. European Journal of Medical Genetics, 2016, 59, 577-583.	1.3	33
93	Association of a promoter polymorphism in FSHR with ovarian reserve and response to ovarian stimulation in women undergoing assisted reproductive treatment. Reproductive BioMedicine Online, 2016, 33, 391-397.	2.4	14
94	Maternal mosaicism for IDUA deletion clarifies recurrence risk in MPS I. Human Genome Variation, 2016, 3, 16031.	0.7	7
95	Relationship of ZNF423 and CTSO with breast cancer risk in two randomised tamoxifen prevention trials. Breast Cancer Research and Treatment, 2016, 158, 591-596.	2.5	5
96	Severe early onset retinitis pigmentosa in a Moroccan patient with Heimler syndrome due to novel homozygous mutation of PEX1 gene. European Journal of Medical Genetics, 2016, 59, 507-511.	1.3	21
97	Sensitivity of BRCA1/2 testing in high-risk breast/ovarian/male breast cancer families: little contribution of comprehensive RNA/NGS panel testing. European Journal of Human Genetics, 2016, 24, 1591-1597.	2.8	26
98	AMH type II receptor and AMH gene polymorphisms are not associated with ovarian reserve, response, or outcomes in ovarian stimulation. Journal of Assisted Reproduction and Genetics, 2016, 33, 1085-1091.	2.5	11
99	The Contribution of Whole Gene Deletions and Large Rearrangements to the Mutation Spectrum in Inherited Tumor Predisposing Syndromes. Human Mutation, 2016, 37, 250-256.	2.5	65
100	Whole Genome Sequencing Increases Molecular Diagnostic Yield Compared with Current Diagnostic Testing for Inherited Retinal Disease. Ophthalmology, 2016, 123, 1143-1150.	5.2	122
101	Mosaic CREBBP mutation causes overlapping clinical features of Rubinsteinâ€“Taybi and Filippi syndromes. European Journal of Human Genetics, 2016, 24, 1363-1366.	2.8	8
102	Effective cascade screening through identification of a mutation in RYR2 in a large family with a history of sudden death. Journal of Cardiology Cases, 2016, 13, 9-13.	0.5	1
103	Common variants modify the age of onset for basal cell carcinomas in Gorlin syndrome. European Journal of Human Genetics, 2015, 23, 708-710.	2.8	10
104	Mutations in LZTR1 add to the complex heterogeneity of schwannomatosis. Neurology, 2015, 84, 141-147.	1.1	90
105	Urinary Tract Effects of HPSE2 Mutations. Journal of the American Society of Nephrology: JASN, 2015, 26, 797-804.	6.1	31
106	Diagnostic Mutation Profiling and Validation of Nonâ€“Small-Cell Lung Cancer Small Biopsy Samples using a High Throughput Platform. Journal of Thoracic Oncology, 2015, 10, 784-792.	1.1	16
107	Dominant Mutations in the Autoimmune Regulator AIRE Are Associated with Common Organ-Specific Autoimmune Diseases. Immunity, 2015, 42, 1185-1196.	14.3	246
108	Oculo-auriculo-vertebral spectrum: Clinical and molecular analysis of 51 patients. European Journal of Medical Genetics, 2015, 58, 455-465.	1.3	83

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109	Leriâ€™s pleonosteosis, a congenital rheumatic disease, results from microduplication at 8q22.1 encompassingGDF6andSDC2and provides insight into systemic sclerosis pathogenesis. Annals of the Rheumatic Diseases, 2015, 74, 1249-1256.	0.9	22
110	Association analyses identify 38 susceptibility loci for inflammatory bowel disease and highlight shared genetic risk across populations. Nature Genetics, 2015, 47, 979-986.	21.4	1,965
111	SMARCE1 mutations in pediatric clear cell meningioma: case report. Journal of Neurosurgery: Pediatrics, 2015, 16, 296-300.	1.3	26
112	Mutations of Human NARS2, Encoding the Mitochondrial Asparaginyl-tRNA Synthetase, Cause Nonsyndromic Deafness and Leigh Syndrome. PLoS Genetics, 2015, 11, e1005097.	3.5	97
113	Identification of Patients With Variants in TPMT and Dose Reduction Reduces Hematologic Events During Thiopurine Treatment of Inflammatory Bowel Disease. Gastroenterology, 2015, 149, 907-917.e7.	1.3	169
114	Agnathia-otocephaly complex and asymmetric velopharyngeal insufficiency due to an in-frame duplication in OTX2. Journal of Human Genetics, 2015, 60, 199-202.	2.3	25
115	Exome sequencing identifies ATP4A gene as responsible of an atypical familial type I gastric neuroendocrine tumour. Human Molecular Genetics, 2015, 24, 2914-2922.	2.9	60
116	Heimler Syndrome Is Caused by Hypomorphic Mutations in the Peroxisome-Biogenesis Genes PEX1 and PEX6. American Journal of Human Genetics, 2015, 97, 535-545.	6.2	103
117	Deletion of 19q13 reveals clinical overlap with Dubowitz syndrome. Journal of Human Genetics, 2015, 60, 781-785.	2.3	12
118	Foramen Ovale Closure Is a Process of Endothelial-to-Mesenchymal Transition Leading to Fibrosis. PLoS ONE, 2014, 9, e107175.	2.5	28
119	Delivery of a Clinical Genomics Service. Genes, 2014, 5, 1001-1017.	2.4	21
120	A single nucleotide polymorphism of bone morphogenic protein-15 is not associated with ovarian reserve or response to ovarian stimulation. Human Reproduction, 2014, 29, 2832-2837.	0.9	5
121	Compound Heterozygosity of Low-Frequency Promoter Deletions and Rare Loss-of-Function Mutations in TXNL4A Causes Burn-McKeown Syndrome. American Journal of Human Genetics, 2014, 95, 698-707.	6.2	55
122	Germline Mutations in SUFU Cause Gorlin Syndromeâ€“Associated Childhood Medulloblastoma and Redefine the Risk Associated With PTCH1 Mutations. Journal of Clinical Oncology, 2014, 32, 4155-4161.	1.6	236
123	The genetic basis of DOORS syndrome: an exome-sequencing study. Lancet Neurology, The, 2014, 13, 44-58.	10.2	108
124	Genetics of human congenital urinary bladder disease. Pediatric Nephrology, 2014, 29, 353-360.	1.7	18
125	The Cost-Effectiveness of a Pharmacogenetic Test: A Trial-Based Evaluation of TPMT Genotyping for Azathioprine. Value in Health, 2014, 17, 22-33.	0.3	71
126	Mutations in CKAP2L, the Human Homolog of the Mouse Radmis Gene, Cause Filippi Syndrome. American Journal of Human Genetics, 2014, 95, 622-632.	6.2	34

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127	Exome Sequencing Identifies a Dominant <i>TNNT3</i> Mutation in a Large Family with Distal Arthrogryposis. Molecular Syndromology, 2014, 5, 218-228.	0.8	11
128	A common Asn680Ser polymorphism in the follicle-stimulating hormone receptor gene is not associated with ovarian response to gonadotropin stimulation in patients undergoing in vitro fertilization. Fertility and Sterility, 2013, 99, 149-155.	1.0	36
129	LRIG2 Mutations Cause Urofacial Syndrome. American Journal of Human Genetics, 2013, 92, 259-264.	6.2	63
130	A pragmatic randomized controlled trial of thiopurine methyltransferase genotyping prior to azathioprine treatment: the TARGET study. Pharmacogenomics, 2011, 12, 815-826.	1.3	69
131	Muscarinic Acetylcholine Receptor M3 Mutation Causes Urinary Bladder Disease and a Prune-Belly-like Syndrome. American Journal of Human Genetics, 2011, 89, 668-674.	6.2	89
132	Mutations in HPSE2 Cause Urofacial Syndrome. American Journal of Human Genetics, 2010, 86, 963-969.	6.2	88
133	The Impact of CYP2D6 Genotyping on Tamoxifen Treatment. Pharmaceuticals, 2010, 3, 1122-1138.	3.8	16
134	Geroderma osteodysplastica maps to a 4 Mb locus on chromosome 1q24. American Journal of Medical Genetics, Part A, 2008, 146A, 3034-3037.	1.2	9
135	Impaired Tamoxifen Metabolism Reduces Survival in Familial Breast Cancer Patients. Clinical Cancer Research, 2008, 14, 5913-5918.	7.0	107
136	Options for Detecting Risk of Aminoglycoside-Induced Ototoxicity in Neonatesâ€”Reply. JAMA Pediatrics, 0, , .	6.2	3