William G Newman

List of Publications by Year
in descending order

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252 papers	22,780 citations	25014 57 h-index	10152 140 g-index
278 all docs	278 docs citations	278 times ranked	34061 citing authors

#	Article	IF	Citations
1	Hostâ€“microbe interactions have shaped the genetic architecture of inflammatory bowel disease. Nature, 2012, 491, 119-124.	13.7	4,038
2	Genome-wide meta-analysis increases to 71 the number of confirmed Crohn's disease susceptibility loci. Nature Genetics, 2010, 42, 1118-1125.	9.4	2,284
3	Association analyses identify 38 susceptibility loci for inflammatory bowel disease and highlight shared genetic risk across populations. Nature Genetics, 2015, 47, 979-986.	9.4	1,965
4	Meta-analysis identifies 29 additional ulcerative colitis risk loci, increasing the number of confirmed associations to 47. Nature Genetics, 2011, 43, 246-252.	9.4	1,201
5	Genome-wide association study implicates immune activation of multiple integrin genes in inflammatory bowel disease. Nature Genetics, 2017, 49, 256-261.	9.4	943
6	Genome-wide association study of CNVs in 16,000 cases of eight common diseases and 3,000 shared controls. Nature, 2010, 464, 713-720.	13.7	737
7	Polygenic Risk Scores for Prediction of Breast Cancer and Breast Cancer Subtypes. American Journal of Human Genetics, 2019, 104, 21-34.	2.6	711
8	Breast Cancer Risk Genes â€" Association Analysis in More than 113,000 Women. New England Journal of Medicine, 2021, 384, 428-439.	13.9	532
9	Genome-wide association study of ulcerative colitis identifies three new susceptibility loci, including the HNF4A region. Nature Genetics, 2009, 41, 1330-1334.	9.4	483
10	100,000 Genomes Pilot on Rare-Disease Diagnosis in Health Care â€" Preliminary Report. New England Journal of Medicine, 2021, 385, 1868-1880.	13.9	352
11	Heterozygous Mutations in TREX1 Cause Familial Chilblain Lupus and Dominant Aicardi-GoutiÃ¨res Syndrome. American Journal of Human Genetics, 2007, 80, 811-815.	2.6	339
12	Genome-wide association study identifies 32 novel breast cancer susceptibility loci from overall and subtype-specific analyses. Nature Genetics, 2020, 52, 572-581.	9.4	265
13	Dominant Mutations in the Autoimmune Regulator AIRE Are Associated with Common Organ-Specific Autoimmune Diseases. Immunity, 2015, 42, 1185-1196.	6.6	246
14	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.	0.8	236
15	Loss-of-function mutations in SMARCE1 cause an inherited disorder of multiple spinal meningiomas. Nature Genetics, 2013, 45, 295-298.	9.4	208
16	Histone Lysine Methylases and Demethylases in the Landscape of Human Developmental Disorders. American Journal of Human Genetics, 2018, 102, 175-187.	2.6	204
17	Perrault Syndrome Is Caused by Recessive Mutations in CLPP, Encoding a Mitochondrial ATP-Dependent Chambered Protease. American Journal of Human Genetics, 2013, 92, 605-613.	2.6	186
18	Molecular characterisation of SMARCB1 and NF2 in familial and sporadic schwannomatosis. Journal of Medical Genetics, 2008, 45, 332-339.	1.5	179

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19	Germline selection shapes human mitochondrial DNA diversity. Science, 2019, 364, .	6.0	178
20	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.	0.6	169
21	Iron overload in the Asian community. Blood, 2009, 114, 20-25.	0.6	164
22	Exploring the genetic architecture of inflammatory bowel disease by whole-genome sequencing identifies association at ADCY7. Nature Genetics, 2017, 49, 186-192.	9.4	153
23	CYP2D6 Genotype and Adjuvant Tamoxifen: Meta-Analysis of Heterogeneous Study Populations. Clinical Pharmacology and Therapeutics, 2014, 95, 216-227.	2.3	150
24	Gerodermia osteodysplastica is caused by mutations in SCYL1BP1, a Rab-6 interacting golgin. Nature Genetics, 2008, 40, 1410-1412.	9.4	138
25	Whole Genome Sequencing Increases Molecular Diagnostic Yield Compared with Current Diagnostic Testing for Inherited Retinal Disease. Ophthalmology, 2016, 123, 1143-1150.	2.5	122
26	Whole Exome Sequencing Reveals the Major Genetic Contributors to Nonsyndromic Tetralogy of Fallot. Circulation Research, 2019, 124, 553-563.	2.0	118
27	Epidermal Growth Factor Receptor in Pancreatic Cancer. Cancers, 2011, 3, 1513-1526.	1.7	114
28	Current use of pharmacogenetic testing: a national survey of thiopurine methyltransferase testing prior to azathioprine prescription. Journal of Clinical Pharmacy and Therapeutics, 2007, 32, 187-195.	0.7	109
29	Use of Single-Nucleotide Polymorphisms and Mammographic Density Plus Classic Risk Factors for Breast Cancer Risk Prediction. JAMA Oncology, 2018, 4, 476.	3.4	109
30	The genetic basis of DOORS syndrome: an exome-sequencing study. Lancet Neurology, The, 2014, 13, 44-58.	4.9	108
31	Germline <scp>SMARCE1</scp> mutations predispose to both spinal and cranial clear cell meningiomas. Journal of Pathology, 2014, 234, 436-440.	2.1	108
32	Impaired Tamoxifen Metabolism Reduces Survival in Familial Breast Cancer Patients. Clinical Cancer Research, 2008, 14, 5913-5918.	3.2	107
33	The MDM2 Promoter SNP285C/309G Haplotype Diminishes Sp1 Transcription Factor Binding and Reduces Risk for Breast and Ovarian Cancer in Caucasians. Cancer Cell, 2011, 19, 273-282.	7.7	104
34	Assessing Individual Breast Cancer Risk within the U.K. National Health Service Breast Screening Program: A New Paradigm for Cancer Prevention. Cancer Prevention Research, 2012, 5, 943-951.	0.7	104
35	Heimler Syndrome Is Caused by Hypomorphic Mutations in the Peroxisome-Biogenesis Genes PEX1 and PEX6. American Journal of Human Genetics, 2015, 97, 535-545.	2.6	103
36	Review of biomarkers in colorectal cancer. Colorectal Disease, 2012, 14, 3-17.	0.7	100

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37	Patientsâ€™ and healthcare professionalsâ€™ views on pharmacogenetic testing and its future delivery in the NHS. Pharmacogenomics, 2007, 8, 1511-1519.	0.6	99
38	Rates of loss of heterozygosity and mitotic recombination in NF2 schwannomas, sporadic vestibular schwannomas and schwannomatosis schwannomas. Oncogene, 2010, 29, 6216-6221.	2.6	91
39	Identification and Characterization of an Inborn Error of Metabolism Caused by Dihydrofolate Reductase Deficiency. American Journal of Human Genetics, 2011, 88, 216-225.	2.6	90
40	Muscarinic Acetylcholine Receptor M3 Mutation Causes Urinary Bladder Disease and a Prune-Belly-like Syndrome. American Journal of Human Genetics, 2011, 89, 668-674.	2.6	89
41	Mutations in HPSE2 Cause Urofacial Syndrome. American Journal of Human Genetics, 2010, 86, 963-969.	2.6	88
42	Irbesartan in Marfan syndrome (AIMS): a double-blind, placebo-controlled randomised trial. Lancet, The, 2019, 394, 2263-2270.	6.3	88
43	Are patients with intermediate TPMT activity at increased risk of myelosuppression when taking thiopurine medications?. Pharmacogenomics, 2010, 11, 177-188.	0.6	86
44	The mutational spectrum of brachydactyly type C. American Journal of Medical Genetics Part A, 2002, 112, 291-296.	2.4	82
45	Comprehensive CYP2D6 genotype and adherence affect outcome in breast cancer patients treated with tamoxifen monotherapy. Breast Cancer Research and Treatment, 2011, 125, 279-287.	1.1	80
46	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.	2.6	78
47	ORAI1 Mutations with Distinct Channel Gating Defects in Tubular Aggregate Myopathy. Human Mutation, 2017, 38, 426-438.	1.1	75
48	Validation of copy number variation analysis for next-generation sequencing diagnostics. European Journal of Human Genetics, 2017, 25, 719-724.	1.4	72
49	The Cost-Effectiveness of a Pharmacogenetic Test: A Trial-Based Evaluation of TPMT Genotyping for Azathioprine. Value in Health, 2014, 17, 22-33.	0.1	71
50	A pragmatic randomized controlled trial of thiopurine methyltransferase genotyping prior to azathioprine treatment: the TARGET study. Pharmacogenomics, 2011, 12, 815-826.	0.6	69
51	Expanding the genotypic spectrum of Perrault syndrome. Clinical Genetics, 2017, 91, 302-312.	1.0	68
52	Extended Spectrum of Human Glucose-6-Phosphatase Catalytic Subunit 3 Deficiency: Novel Genotypes and Phenotypic Variability in Severe Congenital Neutropenia. Journal of Pediatrics, 2012, 160, 679-683.e2.	0.9	67
53	A clinical and molecular review of ubiquitous glucose-6-phosphatase deficiency caused by G6PC3 mutations. Orphanet Journal of Rare Diseases, 2013, 8, 84.	1.2	67
54	Genetics of mitochondrial dysfunction and infertility. Clinical Genetics, 2017, 91, 199-207.	1.0	66

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55	The Contribution of Whole Gene Deletions and Large Rearrangements to the Mutation Spectrum in Inherited Tumor Predisposing Syndromes. Human Mutation, 2016, 37, 250-256.	1.1	65
56	Recommendations for clinical interpretation of variants found in non-coding regions of the genome. Genome Medicine, 2022, 14, .	3.6	65
57	CTLA4/ICOS gene variants and haplotypes are associated with rheumatoid arthritis and primary biliary cirrhosis in the Canadian population. Arthritis and Rheumatism, 2009, 60, 931-937.	6.7	63
58	LRIG2 Mutations Cause Urofacial Syndrome. American Journal of Human Genetics, 2013, 92, 259-264.	2.6	63
59	Exome sequencing identifies ATP4A gene as responsible of an atypical familial type I gastric neuroendocrine tumour. Human Molecular Genetics, 2015, 24, 2914-2922.	1.4	60
60	Valuing pharmacogenetic testing services: A comparison of patients' and health care professionals' preferences. Value in Health, 2011, 14, 121-134.	0.1	58
61	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.	0.8	58
62	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.	1.5	56
63	First evidence of genotypeâ€“phenotype correlations in Gorlin syndrome. Journal of Medical Genetics, 2017, 54, 530-536.	1.5	56
64	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.	1.1	56
65	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.	2.6	55
66	Management of craniopharyngioma: the Liverpool experience following the introduction of the CCLG guidelines. Introducing a new risk assessment grading system. Child's Nervous System, 2012, 28, 1181-1192.	0.6	53
67	Rheumatoid arthritis association with theFCRL3 â€“169C polymorphism is restricted toPTPN22 1858Tâ€“homozygous individuals in a Canadian population. Arthritis and Rheumatism, 2006, 54, 3820-3827.	6.7	51
68	COMP mutation screening as an aid for the clinical diagnosis and counselling of patients with a suspected diagnosis of pseudoachondroplasia or multiple epiphyseal dysplasia. European Journal of Human Genetics, 2005, 13, 547-555.	1.4	50
69	Further delineation of the phenotype of severe congenital neutropenia type 4 due to mutations in G6PC3. European Journal of Human Genetics, 2011, 19, 18-22.	1.4	50
70	Clinical Pharmacogenetics Implementation Consortium Guideline for the Use of Aminoglycosides Based on MTâ€RNR1 Genotype. Clinical Pharmacology and Therapeutics, 2022, 111, 366-372.	2.3	50
71	Identification of a mutation in the ubiquitin-fold modifier 1-specific peptidase 2 gene, UFSP2, in an extended South African family with Beukes hip dysplasia. South African Medical Journal, 2015, 105, 558.	0.2	49
72	Spectrum of PEX1 and PEX6 variants in Heimler syndrome. European Journal of Human Genetics, 2016, 24, 1565-1571.	1.4	49

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73	Gender-stratified analysis of DLG5 R30Q in 4707 patients with Crohn disease and 4973 controls from 12 Caucasian cohorts. Journal of Medical Genetics, 2007, 45, 36-42.	1.5	47
74	Rare Variants in BNC2 Are Implicated in Autosomal-Dominant Congenital Lower Urinary-Tract Obstruction. American Journal of Human Genetics, 2019, 104, 994-1006.	2.6	47
75	EGFR and KRAS Mutational Analysis and Their Correlation to Survival in Pancreatic and Periampullary Cancer. Pancreas, 2012, 41, 428-434.	0.5	45
76	Follicle-stimulating hormone receptor gene polymorphisms are not associated with ovarian reserve markers. Fertility and Sterility, 2012, 97, 677-681.	0.5	45
77	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.	3.0	45
78	Haplotype analysis of the 185delAG BRCA1 mutation in ethnically diverse populations. European Journal of Human Genetics, 2013, 21, 212-216.	1.4	44
79	Improving pharmacovigilance in Europe: TPMT genotyping and phenotyping in the UK and Spain. European Journal of Human Genetics, 2009, 17, 991-998.	1.4	43
80	Prospective evaluation of a breast-cancer risk model integrating classical risk factors and polygenic risk in 15 cohorts from six countries. International Journal of Epidemiology, 2022, 50, 1897-1911.	0.9	43
81	Pharmacogenetics education in British medical schools. Genomic Medicine, 2008, 2, 101-105.	0.6	41
82	ISL1 is a major susceptibility gene for classic bladder exstrophy and a regulator of urinary tract development. Scientific Reports, 2017, 7, 42170.	1.6	41
83	Impaired eIF5A function causes a Mendelian disorder that is partially rescued in model systems by spermidine. Nature Communications, 2021, 12, 833.	5.8	41
84	Current status and future potential of somatic mutation testing from circulating free DNA in patients with solid tumours. The HUGO Journal, 2010, 4, 11-21.	4.1	40
85	Allele and genotype frequencies of the polymorphic cytochrome P450 genes (CYP1A1, CYP3A4, CYP3A5,) Tj ETQq1 1 0.784314 rgBT	1.0	39
86	Breast Cancer Polygenic Risk Score and Contralateral Breast Cancer Risk. American Journal of Human Genetics, 2020, 107, 837-848.	2.6	39
87	Expanding the clinical spectrum of SLC29A3 gene defects. European Journal of Medical Genetics, 2010, 53, 309-313.	0.7	38
88	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.	2.3	38
89	Mutations in the G6PC3 gene cause Dursun syndrome. American Journal of Medical Genetics, Part A, 2010, 152A, 2609-2611.	0.7	37
90	Comparison of in silico strategies to prioritize rare genomic variants impacting RNA splicing for the diagnosis of genomic disorders. Scientific Reports, 2021, 11, 20607.	1.6	37

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91	Polymorphisms of CYP19A1 and response to aromatase inhibitors in metastatic breast cancer patients. Breast Cancer Research and Treatment, 2012, 133, 1191-1198.	1.1	36
92	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.	0.5	36
93	Breast cancer susceptibility variants alter risks in familial disease. Journal of Medical Genetics, 2010, 47, 126-131.	1.5	35
94	Genomic and healthcare dynamics of nosocomial SARS-CoV-2 transmission. ELife, 2021, 10, .	2.8	35
95	UGT1A1*28genotype predicts gastrointestinal toxicity in patients treated with intermediate-dose irinotecan. Pharmacogenomics, 2009, 10, 733-739.	0.6	34
96	Pernicious anemia â€“ Genetic insights. Autoimmunity Reviews, 2011, 10, 455-459.	2.5	33
97	Non lethal Raine syndrome and differential diagnosis. European Journal of Medical Genetics, 2016, 59, 577-583.	0.7	33
98	Genetic Variants in IL-23R and ATG16L1 Independently Predispose to Increased Susceptibility to Crohn's Disease in a Canadian Population. Journal of Clinical Gastroenterology, 2009, 43, 444-447.	1.1	32
99	Prevention of breast cancer in the context of a national breast screening programme. Journal of Internal Medicine, 2012, 271, 321-330.	2.7	31
100	Urinary Tract Effects of HPSE2 Mutations. Journal of the American Society of Nephrology: JASN, 2015, 26, 797-804.	3.0	31
101	A network analysis to identify mediators of germline-driven differences in breast cancer prognosis. Nature Communications, 2020, 11, 312.	5.8	30
102	Rapid Point-of-Care Genotyping to Avoid Aminoglycoside-Induced Ototoxicity in Neonatal Intensive Care. JAMA Pediatrics, 2022, 176, 486.	3.3	30
103	Foramen Ovale Closure Is a Process of Endothelial-to-Mesenchymal Transition Leading to Fibrosis. PLoS ONE, 2014, 9, e107175.	1.1	28
104	Structural insight into the human mitochondrial tRNA purine N1-methyltransferase and ribonuclease P complexes. Journal of Biological Chemistry, 2018, 293, 12862-12876.	1.6	28
105	The FANCM:p.Arg658* truncating variant is associated with risk of triple-negative breast cancer. Npj Breast Cancer, 2019, 5, 38.	2.3	28
106	DLG5 variants contribute to Crohn disease risk in a Canadian population. Human Mutation, 2006, 27, 353-358.	1.1	27
107	Microfluidic Platform for Single Nucleotide Polymorphism Genotyping of the Thiopurine S-Methyltransferase Gene to Evaluate Risk for Adverse Drug Events. Journal of Molecular Diagnostics, 2007, 9, 521-529.	1.2	27
108	Chiari I malformation without hydrocephalus: acute intracranial hypertension managed with endoscopic third ventriculostomy (ETV). Child's Nervous System, 2008, 24, 1493-1497.	0.6	27

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109	The clinical management of relatives of young sudden unexplained death victims; implantable defibrillators are rarely indicated. Heart, 2012, 98, 631-636.	1.2	27
110	Perrault syndrome: further evidence for genetic heterogeneity. Journal of Neurology, 2012, 259, 974-976.	1.8	27
111	Heparanase 2, mutated in urofacial syndrome, mediates peripheral neural development in Xenopus. Human Molecular Genetics, 2014, 23, 4302-4314.	1.4	27
112	Intronic splicing mutations in PTCH1 cause Gorlin syndrome. Familial Cancer, 2014, 13, 477-480.	0.9	27
113	Loss-of-function variants in myocardin cause congenital megabladder in humans and mice. Journal of Clinical Investigation, 2019, 129, 5374-5380.	3.9	27
114	SMARCE1 mutations in pediatric clear cell meningioma: case report. Journal of Neurosurgery: Pediatrics, 2015, 16, 296-300.	0.8	26
115	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.	1.4	26
116	A comparative analysis of KMT2D missense variants in Kabuki syndrome, cancers and the general population. Journal of Human Genetics, 2019, 64, 161-170.	1.1	26
117	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.	2.6	26
118	Lrig2 and Hpse2, mutated in urofacial syndrome, pattern nerves in the urinary bladder. Kidney International, 2019, 95, 1138-1152.	2.6	25
119	The Role of the U5 snRNP in Genetic Disorders and Cancer. Frontiers in Genetics, 2021, 12, 636620.	1.1	25
120	Replication and Meta-Analysis of 13,000 Cases Defines the Risk for Interleukin-23 Receptor and Autophagy-Related 16-Like 1 Variants in Crohnâ€™s Disease. Canadian Journal of Gastroenterology & Hepatology, 2010, 24, 297-302.	1.8	24
121	A prospective, randomized, placebo-controlled, double-blind, multicenter study of the effects of irbesartan on aortic dilatation in Marfan syndrome (AIMS trial): study protocol. Trials, 2013, 14, 408.	0.7	24
122	Disease modeling of core pre-mRNA splicing factor haploinsufficiency. Human Molecular Genetics, 2019, 28, 3704-3723.	1.4	24
123	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.	2.3	24
124	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.	3.9	24
125	TPMT testing in azathioprine: a â€˜cost-effective use of healthcare resourcesâ€™?. Personalized Medicine, 2009, 6, 103-113.	0.8	23
126	A Recurrent De Novo Nonsense Variant in ZSWIM6 Results in Severe Intellectual Disability without Frontonasal or Limb Malformations. American Journal of Human Genetics, 2017, 101, 995-1005.	2.6	23

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127	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.	1.1	23
128	Removing barriers to a clinical pharmacogenetics service. Personalized Medicine, 2008, 5, 471-480.	0.8	22
129	Urofacial syndrome: a genetic and congenital disease of aberrant urinary bladder innervation. Pediatric Nephrology, 2014, 29, 513-518.	0.9	22
130	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.	0.5	22
131	SLC20A1 Is Involved in Urinary Tract and Urorectal Development. Frontiers in Cell and Developmental Biology, 2020, 8, 567.	1.8	22
132	Fine-Mapping CASP8 Risk Variants in Breast Cancer. Cancer Epidemiology Biomarkers and Prevention, 2012, 21, 176-181.	1.1	21
133	Delivery of a Clinical Genomics Service. Genes, 2014, 5, 1001-1017.	1.0	21
134	A Method to Exploit the Structure of Genetic Ancestry Space to Enhance Case-Control Studies. American Journal of Human Genetics, 2016, 98, 857-868.	2.6	21
135	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.	0.7	21
136	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.	1.7	21
137	Array comparative genomic hybridization for diagnosis of developmental delay - an exploratory cost-consequences analysis. Clinical Genetics, 2007, 71, 254-259.	1.0	20
138	Genetic analysis of thiopurine methyltransferase polymorphism in the Jordanian population. European Journal of Clinical Pharmacology, 2010, 66, 999-1003.	0.8	20
139	Spontaneous ovarian hyperstimulation syndrome: case report, pathophysiological classification and diagnostic algorithm. European Journal of Obstetrics, Gynecology and Reproductive Biology, 2013, 169, 143-148.	0.5	20
140	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.	1.1	20
141	Refusal of viral testing during the SARS-CoV-2 pandemic. Clinical Medicine, 2020, 20, e163-e164.	0.8	20
142	RASSF1A polymorphism in familial breast cancer. Familial Cancer, 2010, 9, 263-265.	0.9	19
143	Bi-allelic premature truncating variants in LTBP1 cause cutis laxa syndrome. American Journal of Human Genetics, 2021, 108, 1095-1114.	2.6	19
144	New insights into Perrault syndrome, a clinically and genetically heterogeneous disorder. Human Genetics, 2022, 141, 805-819.	1.8	19

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145	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.	1.1	19
146	Breast cancer risks associated with missense variants in breast cancer susceptibility genes. Genome Medicine, 2022, 14, 51.	3.6	19
147	Natural history of azathioprine-associated lymphopenia in inflammatory bowel disease patients. European Journal of Gastroenterology and Hepatology, 2011, 23, 153-158.	0.8	18
148	Variability of bone marrow morphology in G6PC3 mutations: Is there a genotypeâ€“phenotype correlation or ageâ€dependent relationship?. American Journal of Hematology, 2011, 86, 235-237.	2.0	18
149	Genetics of human congenital urinary bladder disease. Pediatric Nephrology, 2014, 29, 353-360.	0.9	18
150	Enzyme replacement therapy and hematopoietic stem cell transplant: a new paradigm of treatment in Wolman disease. Orphanet Journal of Rare Diseases, 2021, 16, 235.	1.2	18
151	Early Diagnosis and Treatment of Cobalamin Deficiency of Infancy Owing to Occult Maternal Pernicious Anemia. Journal of Pediatric Hematology/Oncology, 2010, 32, 319-322.	0.3	17
152	Pharmacogenetic Testing in the United Kingdom Genetics and Immunogenetics Laboratories. Genetic Testing and Molecular Biomarkers, 2010, 14, 121-125.	0.3	17
153	Delivering a Pharmacogenetic Service: Is There a Role for Genetic Counselors?. Journal of Genetic Counseling, 2012, 21, 527-535.	0.9	17
154	Modelling the developmental spliceosomal craniofacial disorder Burn-McKeown syndrome using induced pluripotent stem cells. PLoS ONE, 2020, 15, e0233582.	1.1	17
155	The Impact of CYP2D6 Genotyping on Tamoxifen Treatment. Pharmaceuticals, 2010, 3, 1122-1138.	1.7	16
156	Pharmacogenetics and pharmacogenomics: a clinical reality. Annals of Clinical Biochemistry, 2011, 48, 410-417.	0.8	16
157	G6PC3 mutations cause non-syndromic severe congenital neutropenia. Molecular Genetics and Metabolism, 2013, 108, 138-141.	0.5	16
158	Diagnosing and Preventing Hearing Loss in the Genomic Age. Trends in Hearing, 2019, 23, 233121651987898.	0.7	16
159	PCOS and peripheral AMH levels in relation to FSH receptor gene single nucleotide polymorphisms. Gynecological Endocrinology, 2012, 28, 375-377.	0.7	15
160	EFTUD2 missense variants disrupt protein function and splicing in mandibulofacial dysostosis Guionâ€Almeida type. Human Mutation, 2020, 41, 1372-1382.	1.1	15
161	Pharmacogenomics in the UK National Health Service: opportunities and challenges. Pharmacogenomics, 2020, 21, 1237-1246.	0.6	15
162	Common variants in breast cancer risk loci predispose to distinct tumor subtypes. Breast Cancer Research, 2022, 24, 2.	2.2	15

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163	Advantages of a Subcutaneous Implantable Cardioverterâ€Defibrillator in LAMP2 Hypertrophic Cardiomyopathy. Journal of Cardiovascular Electrophysiology, 2013, 24, 1051-1053.	0.8	14
164	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.	1.1	14
165	Personalised virtual gene panels reduce interpretation workload and maintain diagnostic rates of proband-only clinical exome sequencing for rare disorders. Journal of Medical Genetics, 2022, 59, 393-398.	1.5	14
166	Increased Rate of Phenocopies in All Age Groups in BRCA1/BRCA2 Mutation Kindred, but Increased Prospective Breast Cancer Risk Is Confined to BRCA2 Mutation Carriers. Cancer Epidemiology Biomarkers and Prevention, 2013, 22, 2269-2276.	1.1	13
167	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.	1.5	13
168	Pharmacogenomic testing to support prescribing in primary care: a structured review of implementation models. Pharmacogenomics, 2021, 22, 761-776.	0.6	13
169	Avoidance of CYP2D6 Inhibitors in Patients Receiving Tamoxifen. Journal of Clinical Oncology, 2010, 28, e584-e585.	0.8	12
170	Pharmacogenetics of aromatase inhibitors. Pharmacogenomics, 2012, 13, 699-707.	0.6	12
171	A known pathogenic variant in the essential mitochondrial translation gene RMND1 causes a Perraultâ€like syndrome with renal defects. Clinical Genetics, 2018, 94, 276-277.	1.0	12
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