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	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
2	Extended gene panel testing in lobular breast cancer. Familial Cancer, 2022, 21, 129-136.	0.9	1
3	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
4	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
5	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
6	New insights into Perrault syndrome, a clinically and genetically heterogeneous disorder. Human Genetics, 2022, 141, 805-819.	1.8	19
7	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
8	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.	1.0	3
9	Expanding the genotypic spectrum of <scp>TXNL4A</scp> variants in <scp>Burnâ€McKeown</scp> syndrome. Clinical Genetics, 2022, 101, 255-259.	1.0	5
10	Common variants in breast cancer risk loci predispose to distinct tumor subtypes. Breast Cancer Research, 2022, 24, 2.	2.2	15
11	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.	2.6	12
12	Rapid Point-of-Care Genotyping to Avoid Aminoglycoside-Induced Ototoxicity in Neonatal Intensive Care. JAMA Pediatrics, 2022, 176, 486.	3.3	30
13	Characterization of the mechanism by which a nonsense variant in RYR2 leads to disordered calcium handling. Physiological Reports, 2022, 10, e15265.	0.7	7
14	Narrowing the chromosome 22q11.2 locus duplicated in bladder exstrophyâ€“epispadias complex. Journal of Pediatric Urology, 2022, 18, 362.e1-362.e8.	0.6	1
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.	1.1	23
16	Homozygous missense variants in BMPR15 can result in primary ovarian insufficiency. Reproductive BioMedicine Online, 2022, , .	1.1	1
17	Breast cancer risks associated with missense variants in breast cancer susceptibility genes. Genome Medicine, 2022, 14, 51.	3.6	19
18	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

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19	Recommendations for clinical interpretation of variants found in non-coding regions of the genome. Genome Medicine, 2022, 14, .	3.6	65
20	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
21	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.	2.9	5
22	The Role of the U5 snRNP in Genetic Disorders and Cancer. Frontiers in Genetics, 2021, 12, 636620.	1.1	25
23	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.	1.4	12
24	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
25	Impaired eIF5A function causes a Mendelian disorder that is partially rescued in model systems by spermidine. Nature Communications, 2021, 12, 833.	5.8	41
26	Genomic and healthcare dynamics of nosocomial SARS-CoV-2 transmission. ELife, 2021, 10, .	2.8	35
27	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
28	Clinical utility of testing for PALB2 and CHEK2 c.1100delC in breast and ovarian cancer. Genetics in Medicine, 2021, 23, 1969-1976.	1.1	8
29	Pathogenic Intronic Splice-Affecting Variants in MYBPC3 in Three Patients with Hypertrophic Cardiomyopathy. Neurology International, 2021, 11, 73-83.	0.2	1
30	Pharmacogenetics to Avoid Loss of Hearing (PALOH) trial: a protocol for a prospective observational implementation trial. BMJ Open, 2021, 11, e044457.	0.8	9
31	Bi-allelic premature truncating variants in LTBP1 cause cutis laxa syndrome. American Journal of Human Genetics, 2021, 108, 1095-1114.	2.6	19
32	The diagnostic utility of clinical exome sequencing in 60 patients with hearing loss disorders: A singleâ€institution experience. Clinical Otolaryngology, 2021, 46, 1257-1262.	0.6	3
33	The Genomic Architecture of Bladder Exstrophy Epispadias Complex. Genes, 2021, 12, 1149.	1.0	8
34	Biallelic loss of function variants in STAG3 result in primary ovarian insufficiency. Reproductive BioMedicine Online, 2021, 43, 899-902.	1.1	5
35	Pharmacogenomic testing to support prescribing in primary care: a structured review of implementation models. Pharmacogenomics, 2021, 22, 761-776.	0.6	13
36	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.	2.2	7

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37	Mendelian randomisation study of smoking exposure in relation to breast cancer risk. British Journal of Cancer, 2021, 125, 1135-1145.	2.9	9
38	Gene Panel Testing for Breast Cancer Reveals Differential Effect of Prior BRCA1/2 Probability. Cancers, 2021, 13, 4154.	1.7	5
39	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
40	Mosaic Fabry Disease in a Male Presenting as Hypertrophic Cardiomyopathy. Neurology International, 2021, 11, 1-9.	0.2	1
41	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
42	Quantifying the Impact of Capacity Constraints in Economic Evaluations: An Application in Precision Medicine. Medical Decision Making, 2021, , 0272989X2110537.	1.2	1
43	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
44	Using a biomarker acutely to identify babies at risk of serious adverse effects from antibiotics: where is the â€˜Terrible Moral and Medical Dilemmaâ€™?. Journal of Medical Ethics, 2021, 47, 117-118.	1.0	0
45	Characterising a homozygous twoâ€exon deletion in UQCRH : comparing human and mouse phenotypes. EMBO Molecular Medicine, 2021, 13, e14397.	3.3	5
46	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
47	Inactivity of Peptidase ClpP Causes Primary Accumulation of Mitochondrial Disaggregase ClpX with Its Interacting Nucleoid Proteins, and of mtDNA. Cells, 2021, 10, 3354.	1.8	4
48	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
49	A recurrent missense variant in HARS2 results in variable sensorineural hearing loss in three unrelated families. Journal of Human Genetics, 2020, 65, 305-311.	1.1	5
50	Early B-cell Factor 3â€"Related Genetic Disease Can Mimic Urofacial Syndrome. Kidney International Reports, 2020, 5, 1823-1827.	0.4	7
51	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.	0.7	5
52	SLC20A1 Is Involved in Urinary Tract and Urorectal Development. Frontiers in Cell and Developmental Biology, 2020, 8, 567.	1.8	22
53	Genetic testing in the acute setting: a round table discussion. Journal of Medical Ethics, 2020, 46, 533-533.	1.0	4
54	Breast Cancer Polygenic Risk Score and Contralateral Breast Cancer Risk. American Journal of Human Genetics, 2020, 107, 837-848.	2.6	39

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55	Modelling the developmental spliceosomal craniofacial disorder Burn-McKeown syndrome using induced pluripotent stem cells. PLoS ONE, 2020, 15, e0233582.	1.1	17
56	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
57	Assessment of Disease-Associated Missense Variants in RYR2 on Transcript Splicing. Neurology International, 2020, 10, 8637.	0.2	0
58	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
59	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
60	A network analysis to identify mediators of germline-driven differences in breast cancer prognosis. Nature Communications, 2020, 11, 312.	5.8	30
61	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.	1.1	6
62	EFTUD2 missense variants disrupt protein function and splicing in mandibulofacial dysostosis Guionâ€Almeida type. Human Mutation, 2020, 41, 1372-1382.	1.1	15
63	Pharmacogenomics in the UK National Health Service: opportunities and challenges. Pharmacogenomics, 2020, 21, 1237-1246.	0.6	15
64	Refractory very early-onset inflammatory bowel disease associated with cytosolic isoleucyl-tRNA synthetase deficiency: A case report. World Journal of Gastroenterology, 2020, 26, 1841-1846.	1.4	6
65	Refusal of viral testing during the SARS-CoV-2 pandemic. Clinical Medicine, 2020, 20, e163-e164.	0.8	20
66	Abstract B49: â€œTriple wild-typeâ€•co-mutational profile in early-stage KRAS-mutant lung cancer. , 2020, , .		0
67	Abstract 3810: Persistence of smoking signature 4 in the non-small cell lung cancer genome. , 2020, , .		1
68	Disease modeling of core pre-mRNA splicing factor haploinsufficiency. Human Molecular Genetics, 2019, 28, 3704-3723.	1.4	24
69	Diagnosing and Preventing Hearing Loss in the Genomic Age. Trends in Hearing, 2019, 23, 233121651987898.	0.7	16
70	The FANCM:p.Arg658* truncating variant is associated with risk of triple-negative breast cancer. Npj Breast Cancer, 2019, 5, 38.	2.3	28
71	A homozygous missense variant in CHRM3 associated with familial urinary bladder disease. Clinical Genetics, 2019, 96, 515-520.	1.0	9
72	Two truncating variants in FANCC and breast cancer risk. Scientific Reports, 2019, 9, 12524.	1.6	5

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73	Germline selection shapes human mitochondrial DNA diversity. Science, 2019, 364, .	6.0	178
74	Accounting for Capacity Constraints in Economic Evaluations of Precision Medicine: A Systematic Review. Pharmacoeconomics, 2019, 37, 1011-1027.	1.7	9
75	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
76	Lrig2 and Hpse2, mutated in urofacial syndrome, pattern nerves in the urinary bladder. Kidney International, 2019, 95, 1138-1152.	2.6	25
77	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
78	Câ€...Identification of the major genetic contributors to tetralogy of fallot. , 2019, , .		0
79	Irbesartan in Marfan syndrome (AIMS): a double-blind, placebo-controlled randomised trial. Lancet, The, 2019, 394, 2263-2270.	6.3	88
80	Clinical and genetic heterogeneity in Melkersson-Rosenthal Syndrome. European Journal of Medical Genetics, 2019, 62, 103536.	0.7	8
81	Polygenic Risk Scores for Prediction of Breast Cancer and Breast Cancer Subtypes. American Journal of Human Genetics, 2019, 104, 21-34.	2.6	711
82	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
83	22q11.2 duplications in a UK cohort with bladder exstrophyâ€"epispadias complex. American Journal of Medical Genetics, Part A, 2019, 179, 404-409.	0.7	11
84	Whole Exome Sequencing Reveals the Major Genetic Contributors to Nonsyndromic Tetralogy of Fallot. Circulation Research, 2019, 124, 553-563.	2.0	118
85	Loss-of-function variants in myocardin cause congenital megabladder in humans and mice. Journal of Clinical Investigation, 2019, 129, 5374-5380.	3.9	27
86	Abstract 1763: Tumor mutational burden, mutational signatures and copy number variation in lung cancer driven by the Ras-Raf-MEK-ERK pathway. , 2019, , .		0
87	Abstract 1763: Tumor mutational burden, mutational signatures and copy number variation in lung cancer driven by the Ras-Raf-MEK-ERK pathway. , 2019, , .		0
88	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
89	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
90	Histone Lysine Methylases and Demethylases in the Landscape of Human Developmental Disorders. American Journal of Human Genetics, 2018, 102, 175-187.	2.6	204

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91	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
92	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
93	Understanding barriers to the introduction of precision medicines in non-small cell lung cancer: A qualitative interview protocol. Wellcome Open Research, 2018, 3, 24.	0.9	9
94	Expanding the genotypic spectrum of Perrault syndrome. Clinical Genetics, 2017, 91, 302-312.	1.0	68
95	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
96	ORAI1 Mutations with Distinct Channel Gating Defects in Tubular Aggregate Myopathy. Human Mutation, 2017, 38, 426-438.	1.1	75
97	Genome-wide association study implicates immune activation of multiple integrin genes in inflammatory bowel disease. Nature Genetics, 2017, 49, 256-261.	9.4	943
98	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
99	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
100	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
101	Severe intellectual disability in a patient with Burnâ€“McKeown syndrome. Clinical Dysmorphology, 2017, 26, 193-194.	0.1	5
102	First evidence of genotypeâ€“phenotype correlations in Gorlin syndrome. Journal of Medical Genetics, 2017, 54, 530-536.	1.5	56
103	Validation of copy number variation analysis for next-generation sequencing diagnostics. European Journal of Human Genetics, 2017, 25, 719-724.	1.4	72
104	Homozygous mutation in PTRH2 gene causes progressive sensorineural deafness and peripheral neuropathy. American Journal of Medical Genetics, Part A, 2017, 173, 1051-1055.	0.7	9
105	Marfanoid habitus is a nonspecific feature of Perrault syndrome. Clinical Dysmorphology, 2017, 26, 200-204.	0.1	7
106	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
107	Genetics of mitochondrial dysfunction and infertility. Clinical Genetics, 2017, 91, 199-207.	1.0	66
108	Chronic Kidney Disease in a Boy with Enuresis: The Diagnosis Behind the Smile. Turkish Nephrology, Dialysis and Transplantation Journal, 2017, 26, 347-350.	0.0	0

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109	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.	0.5	1
110	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
111	Non lethal Raine syndrome and differential diagnosis. European Journal of Medical Genetics, 2016, 59, 577-583.	0.7	33
112	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
113	Relationship of ZNF423 and CTSO with breast cancer risk in two randomised tamoxifen prevention trials. Breast Cancer Research and Treatment, 2016, 158, 591-596.	1.1	5
114	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
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	Spectrum of PEX1 and PEX6 variants in Heimler syndrome. European Journal of Human Genetics, 2016, 24, 1565-1571.	1.4	49
117	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
118	Whole Genome Sequencing Increases Molecular Diagnostic Yield Compared with Current Diagnostic Testing for Inherited Retinal Disease. Ophthalmology, 2016, 123, 1143-1150.	2.5	122
119	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
120	Common variants modify the age of onset for basal cell carcinomas in Gorlin syndrome. European Journal of Human Genetics, 2015, 23, 708-710.	1.4	10
121	Urinary Tract Effects of HPSE2 Mutations. Journal of the American Society of Nephrology: JASN, 2015, 26, 797-804.	3.0	31
122	Dominant Mutations in the Autoimmune Regulator AIRE Are Associated with Common Organ-Specific Autoimmune Diseases. Immunity, 2015, 42, 1185-1196.	6.6	246
123	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
124	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
125	SMARCE1 mutations in pediatric clear cell meningioma: case report. Journal of Neurosurgery: Pediatrics, 2015, 16, 296-300.	0.8	26
126	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

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127	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
128	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
129	Foramen Ovale Closure Is a Process of Endothelial-to-Mesenchymal Transition Leading to Fibrosis. PLoS ONE, 2014, 9, e107175.	1.1	28
130	Delivery of a Clinical Genomics Service. Genes, 2014, 5, 1001-1017.	1.0	21
131	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.	0.4	5
132	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
133	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
134	Identification of a novel familial FGF16 mutation in metacarpal 4â€"5 fusion. Clinical Dysmorphology, 2014, 23, 95-97.	0.1	8
135	Diagnosis of Rare Inherited Diseases. RSC Drug Discovery Series, 2014, , 35-49.	0.2	1
136	Heparanase 2, mutated in urofacial syndrome, mediates peripheral neural development in Xenopus. Human Molecular Genetics, 2014, 23, 4302-4314.	1.4	27
137	The genetic basis of DOORS syndrome: an exome-sequencing study. Lancet Neurology, The, 2014, 13, 44-58.	4.9	108
138	Urofacial syndrome: a genetic and congenital disease of aberrant urinary bladder innervation. Pediatric Nephrology, 2014, 29, 513-518.	0.9	22
139	Genetics of human congenital urinary bladder disease. Pediatric Nephrology, 2014, 29, 353-360.	0.9	18
140	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
141	Germline <scp>SMARCE1</scp> mutations predispose to both spinal and cranial clear cell meningiomas. Journal of Pathology, 2014, 234, 436-440.	2.1	108
142	Intronic splicing mutations in PTCH1 cause Gorlin syndrome. Familial Cancer, 2014, 13, 477-480.	0.9	27
143	Exome Sequencing Identifies a Dominant <i>TNNT3</i> Mutation in a Large Family with Distal Arthrogryposis. Molecular Syndromology, 2014, 5, 218-228.	0.3	11
144	CYP2D6 Genotype and Adjuvant Tamoxifen: Meta-Analysis of Heterogeneous Study Populations. Clinical Pharmacology and Therapeutics, 2014, 95, 216-227.	2.3	150

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145	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
146	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
147	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
148	Loss-of-function mutations in SMARCE1 cause an inherited disorder of multiple spinal meningiomas. Nature Genetics, 2013, 45, 295-298.	9.4	208
149	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
150	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
151	FSH receptor genotype does not predict metaphase-II oocyte output or fertilization rates in ICSI patients. Reproductive BioMedicine Online, 2013, 27, 305-309.	1.1	6
152	G6PC3 mutations cause non-syndromic severe congenital neutropenia. Molecular Genetics and Metabolism, 2013, 108, 138-141.	0.5	16
153	Haplotype analysis of the 185delAG BRCA1 mutation in ethnically diverse populations. European Journal of Human Genetics, 2013, 21, 212-216.	1.4	44
154	LRIG2 Mutations Cause Urofacial Syndrome. American Journal of Human Genetics, 2013, 92, 259-264.	2.6	63
155	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
156	Can manipulation of splicing offer gene therapy possibilities to those with tumour-prone disorders?. European Journal of Human Genetics, 2013, 21, 701-702.	1.4	1
157	Advantages of a Subcutaneous Implantable Cardioverterâ€Defibrillator in LAMP2 Hypertrophic Cardiomyopathy. Journal of Cardiovascular Electrophysiology, 2013, 24, 1051-1053.	0.8	14
158	Are health technology assessments of pharmacogenetic tests feasible? A case study ofCYP2D6testing in the treatment of breast cancer with tamoxifen. Personalized Medicine, 2013, 10, 601-611.	0.8	4
159	The clinical management of relatives of young sudden unexplained death victims; implantable defibrillators are rarely indicated. Heart, 2012, 98, 631-636.	1.2	27
160	Fine-Mapping CASP8 Risk Variants in Breast Cancer. Cancer Epidemiology Biomarkers and Prevention, 2012, 21, 176-181.	1.1	21
161	EGFR and KRAS Mutational Analysis and Their Correlation to Survival in Pancreatic and Periampullary Cancer. Pancreas, 2012, 41, 428-434.	0.5	45
162	Prevention of breast cancer in the context of a national breast screening programme. Journal of Internal Medicine, 2012, 271, 321-330.	2.7	31

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163	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
164	Follicle-stimulating hormone receptor gene polymorphisms are not associated with ovarian reserve markers. Fertility and Sterility, 2012, 97, 677-681.	0.5	45
165	PCOS and peripheral AMH levels in relation to FSH receptor gene single nucleotide polymorphisms. Gynecological Endocrinology, 2012, 28, 375-377.	0.7	15
166	Developing National Guidance on Genetic Testing for Breast Cancer Predisposition: The Role of Economic Evidence?. Genetic Testing and Molecular Biomarkers, 2012, 16, 580-591.	0.3	10
167	Hostâ€"microbe interactions have shaped the genetic architecture of inflammatory bowel disease. Nature, 2012, 491, 119-124.	13.7	4,038
168	Breast cancer pharmacogenomics: where we are going. Pharmacogenomics, 2012, 13, 629-631.	0.6	6
169	Pharmacogenetics of aromatase inhibitors. Pharmacogenomics, 2012, 13, 699-707.	0.6	12
170	Allele and genotype frequencies of the polymorphic cytochrome P450 genes (CYP1A1, CYP3A4, CYP3A5,) Tj ETQq0 0 0 rgBT /Overlock 1	1.0	39
171	Pharmacogenetics: transforming clinical medicine. Journal of the Royal College of Physicians of Edinburgh, The, 2012, 42, 244-247.	0.2	2
172	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
173	High sensitivity for BRCA1/2 mutations in breast/ovarian kindreds: are there still other breast/ovary genes to be discovered?. Breast Cancer Research and Treatment, 2012, 134, 895-897.	1.1	4
174	Delivering a Pharmacogenetic Service: Is There a Role for Genetic Counselors?. Journal of Genetic Counseling, 2012, 21, 527-535.	0.9	17
175	Perrault syndrome: further evidence for genetic heterogeneity. Journal of Neurology, 2012, 259, 974-976.	1.8	27
176	Lack of caveolin-1 (P132L) somatic mutations in breast cancer. Breast Cancer Research and Treatment, 2012, 132, 1185-1186.	1.1	7
177	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
178	Review of biomarkers in colorectal cancer. Colorectal Disease, 2012, 14, 3-17.	0.7	100
179	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
180	A role for genetic counsellors and clinical geneticists in pharmacogenetics?. Clinical Genetics, 2012, 82, 201-202.	1.0	5

#	Article	IF	Citations
181	A DELAYED PRESENTATION OF CONGENITAL GLAUCOMA. Journal of Paediatrics and Child Health, 2012, 48, 541-541.	0.4	0
182	Valuing pharmacogenetic testing services: A comparison of patients' and health care professionals' preferences. Value in Health, 2011, 14, 121-134.	0.1	58
183	Natural history of azathioprine-associated lymphopenia in inflammatory bowel disease patients. European Journal of Gastroenterology and Hepatology, 2011, 23, 153-158.	0.8	18
184	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
185	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
186	Pernicious anemia â€“ Genetic insights. Autoimmunity Reviews, 2011, 10, 455-459.	2.5	33
187	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
188	A pragmatic randomized controlled trial of thiopurine methyltransferase genotyping prior to azathioprine treatment: the TARGET study. Pharmacogenomics, 2011, 12, 815-826.	0.6	69
189	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
190	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
191	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
192	Newly recognized recessive syndrome characterized by dysmorphic features, hypogonadotropic hypogonadism, severe microcephaly, and sensorineural hearing loss maps to 3p21.3. American Journal of Medical Genetics, Part A, 2011, 155, 2910-2915.	0.7	2
193	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
194	Exacerbation of Hereditary Warfarin Resistance by Azathioprine. Clinical and Applied Thrombosis/Hemostasis, 2011, 17, 293-296.	0.7	4
195	Pharmacogenetics and pharmacogenomics: a clinical reality. Annals of Clinical Biochemistry, 2011, 48, 410-417.	0.8	16
196	Epidermal Growth Factor Receptor in Pancreatic Cancer. Cancers, 2011, 3, 1513-1526.	1.7	114
197	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
198	Mutations in HPSE2 Cause Urofacial Syndrome. American Journal of Human Genetics, 2010, 86, 963-969.	2.6	88

#	Article	IF	Citations
199	Mutations in HPSE2 Cause Urofacial Syndrome. American Journal of Human Genetics, 2010, 87, 309.	2.6	1
200	Genetic analysis of thiopurine methyltransferase polymorphism in the Jordanian population. European Journal of Clinical Pharmacology, 2010, 66, 999-1003.	0.8	20
201	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
202	RASSF1A polymorphism in familial breast cancer. Familial Cancer, 2010, 9, 263-265.	0.9	19
203	Breast cancer susceptibility variants alter risk in familial ovarian cancer. Familial Cancer, 2010, 9, 503-506.	0.9	4
204	Isolated unilateral vestibular schwannomas do not harborHRASmutations. American Journal of Medical Genetics, Part A, 2010, 152A, 1586-1587.	0.7	0
205	Mutations in the G6PC3 gene cause Dursun syndrome. American Journal of Medical Genetics, Part A, 2010, 152A, 2609-2611.	0.7	37
206	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
207	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
208	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
209	The Impact of CYP2D6 Genotyping on Tamoxifen Treatment. Pharmaceuticals, 2010, 3, 1122-1138.	1.7	16
210	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
211	Avoidance of CYP2D6 Inhibitors in Patients Receiving Tamoxifen. Journal of Clinical Oncology, 2010, 28, e584-e585.	0.8	12
212	Principles of Cancer Treatment. , 2010, , 1-9.		0
213	Pharmacogenetic Testing in the United Kingdom Genetics and Immunogenetics Laboratories. Genetic Testing and Molecular Biomarkers, 2010, 14, 121-125.	0.3	17
214	Breast cancer susceptibility variants alter risks in familial disease. Journal of Medical Genetics, 2010, 47, 126-131.	1.5	35
215	Expanding the clinical spectrum of SLC29A3 gene defects. European Journal of Medical Genetics, 2010, 53, 309-313.	0.7	38
216	Genetics of patent foramen ovaleâ€"NKX2-5 and beyond. Clinical Neurology and Neurosurgery, 2010, 112, 457-458.	0.6	4

#	Article	IF	Citations
217	Are patients with intermediate TPMT activity at increased risk of myelosuppression when taking thiopurine medications?. Pharmacogenomics, 2010, 11, 177-188.	0.6	86
218	Future Advances in Cancer Pharmacogenomics. , 2010, , 175-179.		0
219	Principles of Genetics and Pharmacogenetics. , 2010, , 27-44.		0
220	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
221	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
222	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
223	UGT1A1*28genotype predicts gastrointestinal toxicity in patients treated with intermediate-dose irinotecan. Pharmacogenomics, 2009, 10, 733-739.	0.6	34
224	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
225	The SPINK1 N34S variant is associated with acute pancreatitis. European Journal of Gastroenterology and Hepatology, 2009, 21, 485.	0.8	4
226	TPMT testing in azathioprine: a â€˜cost-effective use of healthcare resourcesâ€™?. Personalized Medicine, 2009, 6, 103-113.	0.8	23
227	Iron overload in the Asian community. Blood, 2009, 114, 20-25.	0.6	164
228	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
229	Geroderma osteodysplastica maps to a 4 Mb locus on chromosome 1q24. American Journal of Medical Genetics, Part A, 2008, 146A, 3034-3037.	0.7	9
230	Pharmacogenetics education in British medical schools. Genomic Medicine, 2008, 2, 101-105.	0.6	41
231	Gerodermia osteodysplastica is caused by mutations in SCYL1BP1, a Rab-6 interacting golgin. Nature Genetics, 2008, 40, 1410-1412.	9.4	138
232	Re: Pharmacogenomic Variation of CYP2D6 and the Choice of Optimal Adjuvant Endocrine Therapy for Postmenopausal Breast Cancer: A Modeling Analysis. Journal of the National Cancer Institute, 2008, 100, 1331-1331.	3.0	1
233	Molecular characterisation of SMARCB1 and NF2 in familial and sporadic schwannomatosis. Journal of Medical Genetics, 2008, 45, 332-339.	1.5	179
234	Inflammatory Bowel Disease in the South Asian Population of Northwest England. American Journal of Gastroenterology, 2008, 103, 242-243.	0.2	8

#	Article	IF	Citations
235	Impaired Tamoxifen Metabolism Reduces Survival in Familial Breast Cancer Patients. Clinical Cancer Research, 2008, 14, 5913-5918.	3.2	107
236	Removing barriers to a clinical pharmacogenetics service. Personalized Medicine, 2008, 5, 471-480.	0.8	22
237	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
238	Patientsâ€™ and healthcare professionalsâ€™ views on pharmacogenetic testing and its future delivery in the NHS. Pharmacogenomics, 2007, 8, 1511-1519.	0.6	99
239	A new X-linked mental retardation (XLMR) syndrome with late-onset primary testicular failure, short stature and microcephaly maps to Xq25â€“q26. European Journal of Medical Genetics, 2007, 50, 216-223.	0.7	3
240	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
241	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
242	Array comparative genomic hybridization for diagnosis of developmental delay - an exploratory cost-consequences analysis. Clinical Genetics, 2007, 71, 254-259.	1.0	20
243	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
244	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
245	DLG5 variants contribute to Crohn disease risk in a Canadian population. Human Mutation, 2006, 27, 353-358.	1.1	27
246	Identifying Causal Genetic Factors. , 2006, , 19-26.		1
247	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
248	Genetic dissection of inflammatory bowel disease: unravelling etiology and improving diagnostics. Expert Review of Clinical Immunology, 2005, 1, 609-617.	1.3	2
249	The mutational spectrum of brachydactyly type C. American Journal of Medical Genetics Part A, 2002, 112, 291-296.	2.4	82
250	Understanding barriers to the introduction of precision medicine in non-small cell lung cancer: a qualitative interview study. Wellcome Open Research, 0, 6, 25.	0.9	2
251	Genomic Sequencing and Healthcare Dynamics Track Nosocomial SARS-CoV-2 Transmission. SSRN Electronic Journal, 0, , .	0.4	1
252	Expanding the HPSE2 Genotypic Spectrum in Urofacial Syndrome, A Disease Featuring a Peripheral Neuropathy of the Urinary Bladder. Frontiers in Genetics, 0, 13, .	1.1	4