William G Newman

List of Publications by Year in Descending Order

246 papers	16,252 citations	52 h-index	125 g-index
278 ext. papers	19,901 ext. citations	7.5 avg, IF	5.61 L-index

#	Paper	IF	Citations
246	Common variants in breast cancer risk loci predispose to distinct tumor subtypes.. *Breast Cancer Research*, **2022**, 24, 2	8.3	3
245	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**,	11	2
244	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	7.5	1
243	Dominant-negative pathogenic variant BRIP1 c.1045G>C is a high-risk allele for non-mucinous epithelial ovarian cancer: A case-control study. *Clinical Genetics*, **2022**, 101, 48-54	4	0
242	Rapid Point-of-Care Genotyping to Avoid Aminoglycoside-Induced Ototoxicity in Neonatal Intensive Care.. *JAMA Pediatrics*, **2022**,	8.3	6
241	Characterization of the mechanism by which a nonsense variant in RYR2 leads to disordered calcium handling.. *Physiological Reports*, **2022**, 10, e15265	2.6	1
240	Breast cancer risks associated with missense variants in breast cancer susceptibility genes.. *Genome Medicine*, **2022**, 14, 51	14.4	0
239	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	11.6	1
238	Characterising a homozygous two-exon deletion in UQCRH: comparing human and mouse phenotypes. *EMBO Molecular Medicine*, **2021**, 13, e14397	12	0
237	100,000 Genomes Pilot on Rare-Disease Diagnosis in Health Care - Preliminary Report. *New England Journal of Medicine*, **2021**, 385, 1868-1880	59.2	34
236	Mosaic Fabry Disease in a Male Presenting as Hypertrophic Cardiomyopathy. *Neurology International*, **2021**, 11, 1-9	0	1
235	Comparison of in silico strategies to prioritize rare genomic variants impacting RNA splicing for the diagnosis of genomic disorders. *Scientific Reports*, **2021**, 11, 20607	4.9	5
234	Quantifying the Impact of Capacity Constraints in Economic Evaluations: An Application in Precision Medicine. *Medical Decision Making*, **2021**, 272989X211053792	2.5	1
233	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	11	3
232	Using a biomarker acutely to identify babies at risk of serious adverse effects from antibiotics: where is the 'terrible Moral and Medical Dilemma'?. *Journal of Medical Ethics*, **2021**, 47, 117-118	2.5	
231	Extended gene panel testing in lobular breast cancer. *Familial Cancer*, **2021**, 1	3	
230	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*, **2021**,	7.8	6

229	Personalised virtual gene panels reduce interpretation workload and maintain diagnostic rates of proband-only clinical exome sequencing for rare disorders. *Journal of Medical Genetics*, **2021**,	5.8	2
228	Enzyme replacement therapy and hematopoietic stem cell transplant: a new paradigm of treatment in Wolman disease. *Orphanet Journal of Rare Diseases*, **2021**, 16, 235	4.2	3
227	Clinical utility of testing for PALB2 and CHEK2 c.1100delC in breast and ovarian cancer. *Genetics in Medicine*, **2021**, 23, 1969-1976	8.1	1
226	Pathogenic Intronic Splice-Affecting Variants in MYBPC3 in Three Patients with Hypertrophic Cardiomyopathy. *Neurology International*, **2021**, 11, 73-83	0	1
225	Pharmacogenetics to Avoid Loss of Hearing (PALOH) trial: a protocol for a prospective observational implementation trial. *BMJ Open*, **2021**, 11, e044457	3	5
224	Bi-allelic premature truncating variants in LTBP1 cause cutis laxa syndrome. *American Journal of Human Genetics*, **2021**, 108, 1095-1114	11	1
223	Clinical Pharmacogenetics Implementation Consortium Guideline for the Use of Aminoglycosides Based on MT-RNR1 Genotype. *Clinical Pharmacology and Therapeutics*, **2021**,	6.1	18
222	The diagnostic utility of clinical exome sequencing in 60 patients with hearing loss disorders: A single-institution experience. *Clinical Otolaryngology*, **2021**, 46, 1257-1262	1.8	0
221	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	9.7	14
220	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	8.7	2
219	The Role of the U5 snRNP in Genetic Disorders and Cancer. *Frontiers in Genetics*, **2021**, 12, 636620	4.5	3
218	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	5.3	6
217	Breast Cancer Risk Genes - Association Analysis in More than 113,000 Women. *New England Journal of Medicine*, **2021**, 384, 428-439	59.2	143
216	Impaired eIF5A function causes a Mendelian disorder that is partially rescued in model systems by spermidine. *Nature Communications*, **2021**, 12, 833	17.4	13
215	High likelihood of actionable pathogenic variant detection in breast cancer genes in women with very early onset breast cancer. *Journal of Medical Genetics*, **2021**,	5.8	3
214	Genomic and healthcare dynamics of nosocomial SARS-CoV-2 transmission. *ELife*, **2021**, 10,	8.9	15
213	The Genomic Architecture of Bladder Exstrophy Epispadias Complex. *Genes*, **2021**, 12,	4.2	2
212	Biallelic loss of function variants in STAG3 result in primary ovarian insufficiency. *Reproductive BioMedicine Online*, **2021**, 43, 899-902	4	2

211	New insights into Perrault syndrome, a clinically and genetically heterogeneous disorder. *Human Genetics*, **2021**, 1	6.3	4
210	Pharmacogenomic testing to support prescribing in primary care: a structured review of implementation models. *Pharmacogenomics*, **2021**, 22, 761-776	2.6	1
209	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	8.3	1
208	Mendelian randomisation study of smoking exposure in relation to breast cancer risk. *British Journal of Cancer*, **2021**, 125, 1135-1145	8.7	0
207	Gene Panel Testing for Breast Cancer Reveals Differential Effect of Prior Probability. *Cancers*, **2021**, 13,	6.6	1
206	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	4	4
205	Genome-wide association study identifies 32 novel breast cancer susceptibility loci from overall and subtype-specific analyses. *Nature Genetics*, **2020**, 52, 572-581	36.3	76
204	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	4.3	12
203	Risk of Contralateral Breast Cancer in Women with and without Pathogenic Variants in , and Genes in Women with Very Early-Onset (. *Cancers*, **2020**, 12,	6.6	11
202	A network analysis to identify mediators of germline-driven differences in breast cancer prognosis. *Nature Communications*, **2020**, 11, 312	17.4	20
201	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	3.7	3
200	EFTUD2 missense variants disrupt protein function and splicing in mandibulofacial dysostosis Guion-Almeida type. *Human Mutation*, **2020**, 41, 1372-1382	4.7	6
199	Pharmacogenomics in the UK National Health Service: opportunities and challenges. *Pharmacogenomics*, **2020**, 21, 1237-1246	2.6	5
198	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	5.6	2
197	Refusal of viral testing during the SARS-CoV-2 pandemic. *Clinical Medicine*, **2020**, 20, e163-e164	1.9	13
196	A recurrent missense variant in HARS2 results in variable sensorineural hearing loss in three unrelated families. *Journal of Human Genetics*, **2020**, 65, 305-311	4.3	4
195	Early B-cell Factor 3-Related Genetic Disease Can Mimic Urofacial Syndrome. *Kidney International Reports*, **2020**, 5, 1823-1827	4.1	3
194	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	2.6	2

193	Is Involved in Urinary Tract and Urorectal Development. *Frontiers in Cell and Developmental Biology*, **2020**, 8, 567	5.7	6
192	Genetic testing in the acute setting: a round table discussion. *Journal of Medical Ethics*, **2020**, 46, 533	2.5	4
191	Breast Cancer Polygenic Risk Score and Contralateral Breast Cancer Risk. *American Journal of Human Genetics*, **2020**, 107, 837-848	11	12
190	Modelling the developmental spliceosomal craniofacial disorder Burn-McKeown syndrome using induced pluripotent stem cells. *PLoS ONE*, **2020**, 15, e0233582	3.7	6
189	A case-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	7.5	19
188	A homozygous missense variant in CHRM3 associated with familial urinary bladder disease. *Clinical Genetics*, **2019**, 96, 515-520	4	4
187	Two truncating variants in FANCC and breast cancer risk. *Scientific Reports*, **2019**, 9, 12524	4.9	2
186	Germline selection shapes human mitochondrial DNA diversity. *Science*, **2019**, 364,	33.3	105
185	Accounting for Capacity Constraints in Economic Evaluations of Precision Medicine: A Systematic Review. *Pharmacoeconomics*, **2019**, 37, 1011-1027	4.4	7
184	Rare Variants in BNC2 Are Implicated in Autosomal-Dominant Congenital Lower Urinary-Tract Obstruction. *American Journal of Human Genetics*, **2019**, 104, 994-1006	11	11
183	Lrig2 and Hpse2, mutated in urofacial syndrome, pattern nerves in the urinary bladder. *Kidney International*, **2019**, 95, 1138-1152	9.9	15
182	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	4.4	30
181	Disease modeling of core pre-mRNA splicing factor haploinsufficiency. *Human Molecular Genetics*, **2019**, 28, 3704-3723	5.6	12
180	Diagnosing and Preventing Hearing Loss in the Genomic Age. *Trends in Hearing*, **2019**, 23, 2331216519878983	3.2	9
179	The :p.Arg658* truncating variant is associated with risk of triple-negative breast cancer. *Npj Breast Cancer*, **2019**, 5, 38	7.8	12
178	Loss-of-function variants in myocardin cause congenital megabladder in humans and mice. *Journal of Clinical Investigation*, **2019**, 129, 5374-5380	15.9	11
177	Irbesartan in Marfan syndrome (AIMS): a double-blind, placebo-controlled randomised trial. *Lancet, The*, **2019**, 394, 2263-2270	40	46
176	Clinical and genetic heterogeneity in Melkersson-Rosenthal Syndrome. *European Journal of Medical Genetics*, **2019**, 62, 103536	2.6	6

175	Polygenic Risk Scores for Prediction of Breast Cancer and Breast Cancer Subtypes. *American Journal of Human Genetics*, **2019**, 104, 21-34	11	363
174	A comparative analysis of KMT2D missense variants in Kabuki syndrome, cancers and the general population. *Journal of Human Genetics*, **2019**, 64, 161-170	4.3	16
173	22q11.2 duplications in a UK cohort with bladder exstrophy-epispadias complex. *American Journal of Medical Genetics, Part A*, **2019**, 179, 404-409	2.5	6
172	Whole Exome Sequencing Reveals the Major Genetic Contributors to Nonsyndromic Tetralogy of Fallot. *Circulation Research*, **2019**, 124, 553-563	15.7	62
171	A known pathogenic variant in the essential mitochondrial translation gene RMND1 causes a Perrault-like syndrome with renal defects. *Clinical Genetics*, **2018**, 94, 276-277	4	4
170	Use of Single-Nucleotide Polymorphisms and Mammographic Density Plus Classic Risk Factors for Breast Cancer Risk Prediction. *JAMA Oncology*, **2018**, 4, 476-482	13.4	72
169	Histone Lysine Methylases and Demethylases in the Landscape of Human Developmental Disorders. *American Journal of Human Genetics*, **2018**, 102, 175-187	11	108
168	A Dominantly Inherited 5PUTR Variant Causing Methylation-Associated Silencing of BRCA1 as a Cause of Breast and Ovarian Cancer. *American Journal of Human Genetics*, **2018**, 103, 213-220	11	43
167	Structural insight into the human mitochondrial tRNA purine N1-methyltransferase and ribonuclease P complexes. *Journal of Biological Chemistry*, **2018**, 293, 12862-12876	5.4	13
166	Understanding barriers to the introduction of precision medicines in non-small cell lung cancer: A qualitative interview protocol. *Wellcome Open Research*, **2018**, 3, 24	4.8	8
165	Expanding the genotypic spectrum of Perrault syndrome. *Clinical Genetics*, **2017**, 91, 302-312	4	54
164	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	2.2	51
163	ORAI1 Mutations with Distinct Channel Gating Defects in Tubular Aggregate Myopathy. *Human Mutation*, **2017**, 38, 426-438	4.7	54
162	Genome-wide association study implicates immune activation of multiple integrin genes in inflammatory bowel disease. *Nature Genetics*, **2017**, 49, 256-261	36.3	462
161	Exploring the genetic architecture of inflammatory bowel disease by whole-genome sequencing identifies association at ADCY7. *Nature Genetics*, **2017**, 49, 186-192	36.3	104
160	ISL1 is a major susceptibility gene for classic bladder exstrophy and a regulator of urinary tract development. *Scientific Reports*, **2017**, 7, 42170	4.9	16
159	The impact of a panel of 18 SNPs on breast cancer risk in women attending a UK familial screening clinic: a case-control study. *Journal of Medical Genetics*, **2017**, 54, 111-113	5.8	42
158	Severe intellectual disability in a patient with Burn-McKeown syndrome. *Clinical Dysmorphology*, **2017**, 26, 193-194	0.9	3

157	First evidence of genotype-phenotype correlations in Gorlin syndrome. *Journal of Medical Genetics*, **2017**, 54, 530-536	5.8	34
156	Validation of copy number variation analysis for next-generation sequencing diagnostics. *European Journal of Human Genetics*, **2017**, 25, 719-724	5.3	53
155	Homozygous mutation in PTRH2 gene causes progressive sensorineural deafness and peripheral neuropathy. *American Journal of Medical Genetics, Part A*, **2017**, 173, 1051-1055	2.5	7
154	Marfanoid habitus is a nonspecific feature of Perrault syndrome. *Clinical Dysmorphology*, **2017**, 26, 200-204	0.9	6
153	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	11	18
152	Genetics of mitochondrial dysfunction and infertility. *Clinical Genetics*, **2017**, 91, 199-207	4	48
151	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, 391-7	4	13
150	Relationship of ZNF423 and CTSO with breast cancer risk in two randomised tamoxifen prevention trials. *Breast Cancer Research and Treatment*, **2016**, 158, 591-6	4.4	4
149	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, 507-11	2.6	15
148	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	5.3	21
147	Spectrum of PEX1 and PEX6 variants in Heimler syndrome. *European Journal of Human Genetics*, **2016**, 24, 1565-1571	5.3	34
146	The Contribution of Whole Gene Deletions and Large Rearrangements to the Mutation Spectrum in Inherited Tumor Predisposing Syndromes. *Human Mutation*, **2016**, 37, 250-6	4.7	44
145	Whole Genome Sequencing Increases Molecular Diagnostic Yield Compared with Current Diagnostic Testing for Inherited Retinal Disease. *Ophthalmology*, **2016**, 123, 1143-50	7.3	82
144	Response to: Mutation in MMP2 gene may result in scleroderma-like skin thickening by Bader-Meunier et al. *Annals of the Rheumatic Diseases*, **2016**, 75, e2	2.4	1
143	A Method to Exploit the Structure of Genetic Ancestry Space to Enhance Case-Control Studies. *American Journal of Human Genetics*, **2016**, 98, 857-868	11	14
142	Non lethal Raine syndrome and differential diagnosis. *European Journal of Medical Genetics*, **2016**, 59, 577-583	2.6	24
141	Dominant Mutations in the Autoimmune Regulator AIRE Are Associated with Common Organ-Specific Autoimmune Diseases. *Immunity*, **2015**, 42, 1185-96	32.3	156
140	Leri's pleonosteosis, a congenital rheumatic disease, results from microduplication at 8q22.1 encompassing GDF6 and SDC2 and provides insight into systemic sclerosis pathogenesis. *Annals of the Rheumatic Diseases*, **2015**, 74, 1249-56	2.4	19

139	Association analyses identify 38 susceptibility loci for inflammatory bowel disease and highlight shared genetic risk across populations. *Nature Genetics*, **2015**, 47, 979-986	36.3	1278
138	SMARCE1 mutations in pediatric clear cell meningioma: case report. *Journal of Neurosurgery: Pediatrics*, **2015**, 16, 296-300	2.1	23
137	Identification of Patients With Variants in TPMT and Dose Reduction Reduces Hematologic Events During Thiopurine Treatment of Inflammatory Bowel Disease. *Gastroenterology*, **2015**, 149, 907-17.e7	13.3	121
136	Exome sequencing identifies ATP4A gene as responsible of an atypical familial type I gastric neuroendocrine tumour. *Human Molecular Genetics*, **2015**, 24, 2914-22	5.6	49
135	Heimler Syndrome Is Caused by Hypomorphic Mutations in the Peroxisome-Biogenesis Genes PEX1 and PEX6. *American Journal of Human Genetics*, **2015**, 97, 535-45	11	75
134	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, 558-63	1.5	30
133	Common variants modify the age of onset for basal cell carcinomas in Gorlin syndrome. *European Journal of Human Genetics*, **2015**, 23, 708-10	5.3	6
132	Urinary tract effects of HPSE2 mutations. *Journal of the American Society of Nephrology: JASN*, **2015**, 26, 797-804	12.7	25
131	The cost-effectiveness of a pharmacogenetic test: a trial-based evaluation of TPMT genotyping for azathioprine. *Value in Health*, **2014**, 17, 22-33	3.3	56
130	Germline SMARCE1 mutations predispose to both spinal and cranial clear cell meningiomas. *Journal of Pathology*, **2014**, 234, 436-40	9.4	81
129	Intronic splicing mutations in PTCH1 cause Gorlin syndrome. *Familial Cancer*, **2014**, 13, 477-80	3	22
128	Exome Sequencing Identifies a Dominant TNNT3 Mutation in a Large Family with Distal Arthrogryposis. *Molecular Syndromology*, **2014**, 5, 218-28	1.5	6
127	CYP2D6 genotype and adjuvant tamoxifen: meta-analysis of heterogeneous study populations. *Clinical Pharmacology and Therapeutics*, **2014**, 95, 216-27	6.1	131
126	Foramen ovale closure is a process of endothelial-to-mesenchymal transition leading to fibrosis. *PLoS ONE*, **2014**, 9, e107175	3.7	19
125	Delivery of a clinical genomics service. *Genes*, **2014**, 5, 1001-17	4.2	17
124	A single nucleotide polymorphism of bone morphogenic protein-15 is not associated with ovarian reserve or response to ovarian stimulation. *Human Reproduction*, **2014**, 29, 2832-7	5.7	5
123	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	11	45
122	Germline mutations in SUFU cause Gorlin syndrome-associated childhood medulloblastoma and redefine the risk associated with PTCH1 mutations. *Journal of Clinical Oncology*, **2014**, 32, 4155-61	2.2	185

121	Identification of a novel familial FGF16 mutation in metacarpal 4-5 fusion. *Clinical Dysmorphology*, **2014**, 23, 95-97	0.9	5
120	Chapter 2:Diagnosis of Rare Inherited Diseases. *RSC Drug Discovery Series*, **2014**, 35-49	0.6	1
119	Heparanase 2, mutated in urofacial syndrome, mediates peripheral neural development in Xenopus. *Human Molecular Genetics*, **2014**, 23, 4302-14	5.6	24
118	The genetic basis of DOORS syndrome: an exome-sequencing study. *Lancet Neurology, The*, **2014**, 13, 44-58	24.1	96
117	Urofacial syndrome: a genetic and congenital disease of aberrant urinary bladder innervation. *Pediatric Nephrology*, **2014**, 29, 513-8	3.2	20
116	Genetics of human congenital urinary bladder disease. *Pediatric Nephrology*, **2014**, 29, 353-60	3.2	15
115	A clinical and molecular review of ubiquitous glucose-6-phosphatase deficiency caused by G6PC3 mutations. *Orphanet Journal of Rare Diseases*, **2013**, 8, 84	4.2	53
114	Perrault syndrome is caused by recessive mutations in CLPP, encoding a mitochondrial ATP-dependent chambered protease. *American Journal of Human Genetics*, **2013**, 92, 605-13	11	152
113	Spontaneous ovarian hyperstimulation syndrome: case report, pathophysiological classification and diagnostic algorithm. *European Journal of Obstetrics, Gynecology and Reproductive Biology*, **2013**, 169, 143-8	2.4	11
112	Loss-of-function mutations in SMARCE1 cause an inherited disorder of multiple spinal meningiomas. *Nature Genetics*, **2013**, 45, 295-8	36.3	162
111	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	2.8	22
110	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	4.8	29
109	FSH receptor genotype does not predict metaphase-II oocyte output or fertilization rates in ICSI patients. *Reproductive BioMedicine Online*, **2013**, 27, 305-9	4	5
108	G6PC3 mutations cause non-syndromic severe congenital neutropenia. *Molecular Genetics and Metabolism*, **2013**, 108, 138-41	3.7	16
107	Haplotype analysis of the 185delAG BRCA1 mutation in ethnically diverse populations. *European Journal of Human Genetics*, **2013**, 21, 212-6	5.3	32
106	LRIG2 mutations cause urofacial syndrome. *American Journal of Human Genetics*, **2013**, 92, 259-64	11	55
105	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, 2269-76	4	12
104	Can manipulation of splicing offer gene therapy possibilities to those with tumour-prone disorders?. *European Journal of Human Genetics*, **2013**, 21, 701-2	5.3	1

103	Advantages of a subcutaneous implantable cardioverter-defibrillator in LAMP2 hypertrophic cardiomyopathy. *Journal of Cardiovascular Electrophysiology*, **2013**, 24, 1051-3	2.7	11
102	Are health technology assessments of pharmacogenetic tests feasible? A case study of CYP2D6 testing in the treatment of breast cancer with tamoxifen. *Personalized Medicine*, **2013**, 10, 601-611	2.2	2
101	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	3.6	60
100	A role for genetic counsellors and clinical geneticists in pharmacogenetics?. *Clinical Genetics*, **2012**, 82, 201-2; author reply 203	4	5
99	A delayed presentation of congenital glaucoma. *Journal of Paediatrics and Child Health*, **2012**, 48, 541	1.3	
98	Prevention of breast cancer in the context of a national breast screening programme. *Journal of Internal Medicine*, **2012**, 271, 321-30	10.8	24
97	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, 943-51	3.2	79
96	Follicle-stimulating hormone receptor gene polymorphisms are not associated with ovarian reserve markers. *Fertility and Sterility*, **2012**, 97, 677-81	4.8	38
95	PCOS and peripheral AMH levels in relation to FSH receptor gene single nucleotide polymorphisms. *Gynecological Endocrinology*, **2012**, 28, 375-7	2.4	12
94	Developing national guidance on genetic testing for breast cancer predisposition: the role of economic evidence?. *Genetic Testing and Molecular Biomarkers*, **2012**, 16, 580-91	1.6	8
93	Host-microbe interactions have shaped the genetic architecture of inflammatory bowel disease. *Nature*, **2012**, 491, 119-24	50.4	3239
92	Breast cancer pharmacogenomics: where we are going. *Pharmacogenomics*, **2012**, 13, 629-31	2.6	6
91	Pharmacogenetics of aromatase inhibitors. *Pharmacogenomics*, **2012**, 13, 699-707	2.6	11
90	Allele and genotype frequencies of the polymorphic cytochrome P450 genes (CYP1A1, CYP3A4, CYP3A5, CYP2C9 and CYP2C19) in the Jordanian population. *Molecular Biology Reports*, **2012**, 39, 9423-33	2.8	37
89	Pharmacogenetics: transforming clinical medicine. *Journal of the Royal College of Physicians of Edinburgh, The*, **2012**, 42, 244-7	0.9	2
88	Management of craniopharyngioma: the Liverpool experience following the introduction of the CCLG guidelines. Introducing a new risk assessment grading system. *Childjs Nervous System*, **2012**, 28, 1181-92	1.7	41
87	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, 895-7	4.4	4
86	Delivering a pharmacogenetic service: is there a role for genetic counselors?. *Journal of Genetic Counseling*, **2012**, 21, 527-35	2.5	14

85	Perrault syndrome: further evidence for genetic heterogeneity. *Journal of Neurology*, **2012**, 259, 974-6	5.5	24
84	Lack of caveolin-1 (P132L) somatic mutations in breast cancer. *Breast Cancer Research and Treatment*, **2012**, 132, 1185-6	4.4	5
83	Polymorphisms of CYP19A1 and response to aromatase inhibitors in metastatic breast cancer patients. *Breast Cancer Research and Treatment*, **2012**, 133, 1191-8	4.4	32
82	Review of biomarkers in colorectal cancer. *Colorectal Disease*, **2012**, 14, 3-17	2.1	81
81	The clinical management of relatives of young sudden unexplained death victims; implantable defibrillators are rarely indicated. *Heart*, **2012**, 98, 631-6	5.1	24
80	Fine-mapping CASP8 risk variants in breast cancer. *Cancer Epidemiology Biomarkers and Prevention*, **2012**, 21, 176-81	4	19
79	EGFR and KRAS mutational analysis and their correlation to survival in pancreatic and periampullary cancer. *Pancreas*, **2012**, 41, 428-34	2.6	43
78	Valuing pharmacogenetic testing services: a comparison of patientsPand health care professionalsP preferences. *Value in Health*, **2011**, 14, 121-34	3.3	49
77	Natural history of azathioprine-associated lymphopenia in inflammatory bowel disease patients: a prospective observational study. *European Journal of Gastroenterology and Hepatology*, **2011**, 23, 153-8	2.2	12
76	Meta-analysis identifies 29 additional ulcerative colitis risk loci, increasing the number of confirmed associations to 47. *Nature Genetics*, **2011**, 43, 246-52	36.3	1028
75	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	5.3	40
74	Pernicious anemia - genetic insights. *Autoimmunity Reviews*, **2011**, 10, 455-9	13.6	29
73	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, 273-82	24.3	94
72	A pragmatic randomized controlled trial of thiopurine methyltransferase genotyping prior to azathioprine treatment: the TARGET study. *Pharmacogenomics*, **2011**, 12, 815-26	2.6	59
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