William G Newman

List of Publications by Citations

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	Host-microbe interactions have shaped the genetic architecture of inflammatory bowel disease. *Nature*, **2012**, 491, 119-24	50.4	3239
245	Genome-wide meta-analysis increases to 71 the number of confirmed Crohn's disease susceptibility loci. *Nature Genetics*, **2010**, 42, 1118-25	36.3	1946
244	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
243	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
242	Genome-wide association study of CNVs in 16,000 cases of eight common diseases and 3,000 shared controls. *Nature*, **2010**, 464, 713-20	50.4	639
241	Genome-wide association study implicates immune activation of multiple integrin genes in inflammatory bowel disease. *Nature Genetics*, **2017**, 49, 256-261	36.3	462
240	Genome-wide association study of ulcerative colitis identifies three new susceptibility loci, including the HNF4A region. *Nature Genetics*, **2009**, 41, 1330-4	36.3	411
239	Polygenic Risk Scores for Prediction of Breast Cancer and Breast Cancer Subtypes. *American Journal of Human Genetics*, **2019**, 104, 21-34	11	363
238	Heterozygous mutations in TREX1 cause familial chilblain lupus and dominant Aicardi-Goutieres syndrome. *American Journal of Human Genetics*, **2007**, 80, 811-5	11	286
237	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
236	Loss-of-function mutations in SMARCE1 cause an inherited disorder of multiple spinal meningiomas. *Nature Genetics*, **2013**, 45, 295-8	36.3	162
235	Dominant Mutations in the Autoimmune Regulator AIRE Are Associated with Common Organ-Specific Autoimmune Diseases. *Immunity*, **2015**, 42, 1185-96	32.3	156
234	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
233	Iron overload in the Asian community. *Blood*, **2009**, 114, 20-5	2.2	152
232	Breast Cancer Risk Genes - Association Analysis in More than 113,000 Women. *New England Journal of Medicine*, **2021**, 384, 428-439	59.2	143
231	Molecular characterisation of SMARCB1 and NF2 in familial and sporadic schwannomatosis. *Journal of Medical Genetics*, **2008**, 45, 332-9	5.8	141
230	CYP2D6 genotype and adjuvant tamoxifen: meta-analysis of heterogeneous study populations. *Clinical Pharmacology and Therapeutics*, **2014**, 95, 216-27	6.1	131

229	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
228	Gerodermia osteodysplastica is caused by mutations in SCYL1BP1, a Rab-6 interacting golgin. *Nature Genetics*, **2008**, 40, 1410-2	36.3	121
227	Histone Lysine Methylases and Demethylases in the Landscape of Human Developmental Disorders. *American Journal of Human Genetics*, **2018**, 102, 175-187	11	108
226	Germline selection shapes human mitochondrial DNA diversity. *Science*, **2019**, 364,	33.3	105
225	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
224	Impaired tamoxifen metabolism reduces survival in familial breast cancer patients. *Clinical Cancer Research*, **2008**, 14, 5913-8	12.9	99
223	Current use of pharmacogenetic testing: a national survey of thiopurine methyltransferase testing prior to azathioprine prescription. *Journal of Clinical Pharmacy and Therapeutics*, **2007**, 32, 187-95	2.2	97
222	The genetic basis of DOORS syndrome: an exome-sequencing study. *Lancet Neurology, The*, **2014**, 13, 44-58	24.1	96
221	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
220	PatientsPand healthcare professionalsPviews on pharmacogenetic testing and its future delivery in the NHS. *Pharmacogenomics*, **2007**, 8, 1511-9	2.6	85
219	Whole Genome Sequencing Increases Molecular Diagnostic Yield Compared with Current Diagnostic Testing for Inherited Retinal Disease. *Ophthalmology*, **2016**, 123, 1143-50	7.3	82
218	Germline SMARCE1 mutations predispose to both spinal and cranial clear cell meningiomas. *Journal of Pathology*, **2014**, 234, 436-40	9.4	81
217	Review of biomarkers in colorectal cancer. *Colorectal Disease*, **2012**, 14, 3-17	2.1	81
216	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
215	Rates of loss of heterozygosity and mitotic recombination in NF2 schwannomas, sporadic vestibular schwannomas and schwannomatosis schwannomas. *Oncogene*, **2010**, 29, 6216-21	9.2	77
214	Epidermal growth factor receptor in pancreatic cancer. *Cancers*, **2011**, 3, 1513-26	6.6	77
213	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
212	Comprehensive CYP2D6 genotype and adherence affect outcome in breast cancer patients treated with tamoxifen monotherapy. *Breast Cancer Research and Treatment*, **2011**, 125, 279-87	4.4	76

211	Are patients with intermediate TPMT activity at increased risk of myelosuppression when taking thiopurine medications?. *Pharmacogenomics*, **2010**, 11, 177-88	2.6	76
210	The mutational spectrum of brachydactyly type C. *American Journal of Medical Genetics Part A*, **2002**, 112, 291-6		76
209	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
208	Muscarinic Acetylcholine Receptor M3 Mutation Causes Urinary Bladder Disease and a Prune-Belly-like Syndrome. *American Journal of Human Genetics*, **2011**, 89, 668-74	11	74
207	Mutations in HPSE2 cause urofacial syndrome. *American Journal of Human Genetics*, **2010**, 86, 963-9	11	73
206	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
205	Identification and characterization of an inborn error of metabolism caused by dihydrofolate reductase deficiency. *American Journal of Human Genetics*, **2011**, 88, 216-25	11	68
204	Whole Exome Sequencing Reveals the Major Genetic Contributors to Nonsyndromic Tetralogy of Fallot. *Circulation Research*, **2019**, 124, 553-563	15.7	62
203	CTLA4/ICOS gene variants and haplotypes are associated with rheumatoid arthritis and primary biliary cirrhosis in the Canadian population. *Arthritis and Rheumatism*, **2009**, 60, 931-7		61
202	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
201	A pragmatic randomized controlled trial of thiopurine methyltransferase genotyping prior to azathioprine treatment: the TARGET study. *Pharmacogenomics*, **2011**, 12, 815-26	2.6	59
200	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
199	LRIG2 mutations cause urofacial syndrome. *American Journal of Human Genetics*, **2013**, 92, 259-64	11	55
198	Expanding the genotypic spectrum of Perrault syndrome. *Clinical Genetics*, **2017**, 91, 302-312	4	54
197	ORAI1 Mutations with Distinct Channel Gating Defects in Tubular Aggregate Myopathy. *Human Mutation*, **2017**, 38, 426-438	4.7	54
196	Validation of copy number variation analysis for next-generation sequencing diagnostics. *European Journal of Human Genetics*, **2017**, 25, 719-724	5.3	53
195	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
194	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

193	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
192	Valuing pharmacogenetic testing services: a comparison of patientsPand health care professionalsP preferences. *Value in Health*, **2011**, 14, 121-34	3.3	49
191	Rheumatoid arthritis association with the FCRL3 -169C polymorphism is restricted to PTPN22 1858T-homozygous individuals in a Canadian population. *Arthritis and Rheumatism*, **2006**, 54, 3820-7		49
190	Genetics of mitochondrial dysfunction and infertility. *Clinical Genetics*, **2017**, 91, 199-207	4	48
189	Irbesartan in Marfan syndrome (AIMS): a double-blind, placebo-controlled randomised trial. *Lancet, The*, **2019**, 394, 2263-2270	40	46
188	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
187	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
186	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
185	EGFR and KRAS mutational analysis and their correlation to survival in pancreatic and periampullary cancer. *Pancreas*, **2012**, 41, 428-34	2.6	43
184	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
183	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, 547-55	5.3	42
182	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
181	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
180	Follicle-stimulating hormone receptor gene polymorphisms are not associated with ovarian reserve markers. *Fertility and Sterility*, **2012**, 97, 677-81	4.8	38
179	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
178	Gender-stratified analysis of DLG5 R30Q in 4707 patients with Crohn disease and 4973 controls from 12 Caucasian cohorts. *Journal of Medical Genetics*, **2008**, 45, 36-42	5.8	37
177	Improving pharmacovigilance in Europe: TPMT genotyping and phenotyping in the UK and Spain. *European Journal of Human Genetics*, **2009**, 17, 991-8	5.3	36
176	Pharmacogenetics education in British medical schools. *Genomic Medicine*, **2008**, 2, 101-5		35

175	First evidence of genotype-phenotype correlations in Gorlin syndrome. *Journal of Medical Genetics*, **2017**, 54, 530-536	5.8	34
174	Spectrum of PEX1 and PEX6 variants in Heimler syndrome. *European Journal of Human Genetics*, **2016**, 24, 1565-1571	5.3	34
173	Breast cancer susceptibility variants alter risks in familial disease. *Journal of Medical Genetics*, **2010**, 47, 126-31	5.8	34
172	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
171	Expanding the clinical spectrum of SLC29A3 gene defects. *European Journal of Medical Genetics*, **2010**, 53, 309-13	2.6	33
170	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		33
169	Haplotype analysis of the 185delAG BRCA1 mutation in ethnically diverse populations. *European Journal of Human Genetics*, **2013**, 21, 212-6	5.3	32
168	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
167	UGT1A1*28 genotype predicts gastrointestinal toxicity in patients treated with intermediate-dose irinotecan. *Pharmacogenomics*, **2009**, 10, 733-9	2.6	31
166	Mutations in the G6PC3 gene cause Dursun syndrome. *American Journal of Medical Genetics, Part A*, **2010**, 152A, 2609-11	2.5	31
165	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
164	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
163	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
162	Pernicious anemia - genetic insights. *Autoimmunity Reviews*, **2011**, 10, 455-9	13.6	29
161	Genetic variants in IL-23R and ATG16L1 independently predispose to increased susceptibility to Crohnß disease in a Canadian population. *Journal of Clinical Gastroenterology*, **2009**, 43, 444-7	3	29
160	Urinary tract effects of HPSE2 mutations. *Journal of the American Society of Nephrology: JASN*, **2015**, 26, 797-804	12.7	25
159	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, 521-9	5.1	25
158	Chiari I malformation without hydrocephalus: acute intracranial hypertension managed with endoscopic third ventriculostomy (ETV). *Childjs Nervous System*, **2008**, 24, 1493-7	1.7	25

157	Heparanase 2, mutated in urofacial syndrome, mediates peripheral neural development in Xenopus. *Human Molecular Genetics*, **2014**, 23, 4302-14	5.6	24
156	Prevention of breast cancer in the context of a national breast screening programme. *Journal of Internal Medicine*, **2012**, 271, 321-30	10.8	24
155	Perrault syndrome: further evidence for genetic heterogeneity. *Journal of Neurology*, **2012**, 259, 974-6	5.5	24
154	The clinical management of relatives of young sudden unexplained death victims; implantable defibrillators are rarely indicated. *Heart*, **2012**, 98, 631-6	5.1	24
153	Non lethal Raine syndrome and differential diagnosis. *European Journal of Medical Genetics*, **2016**, 59, 577-583	2.6	24
152	SMARCE1 mutations in pediatric clear cell meningioma: case report. *Journal of Neurosurgery: Pediatrics*, **2015**, 16, 296-300	2.1	23
151	Intronic splicing mutations in PTCH1 cause Gorlin syndrome. *Familial Cancer*, **2014**, 13, 477-80	3	22
150	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
149	Replication and meta-analysis of 13,000 cases defines the risk for interleukin-23 receptor and autophagy-related 16-like 1 variants in Crohn□s disease. *Canadian Journal of Gastroenterology & Hepatology*, **2010**, 24, 297-302		22
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	DLG5 variants contribute to Crohn disease risk in a Canadian population. *Human Mutation*, **2006**, 27, 353-8	4.7	21
146	A network analysis to identify mediators of germline-driven differences in breast cancer prognosis. *Nature Communications*, **2020**, 11, 312	17.4	20
145	Urofacial syndrome: a genetic and congenital disease of aberrant urinary bladder innervation. *Pediatric Nephrology*, **2014**, 29, 513-8	3.2	20
144	Leri□ 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
143	Foramen ovale closure is a process of endothelial-to-mesenchymal transition leading to fibrosis. *PLoS ONE*, **2014**, 9, e107175	3.7	19
142	Fine-mapping CASP8 risk variants in breast cancer. *Cancer Epidemiology Biomarkers and Prevention*, **2012**, 21, 176-81	4	19
141	Removing barriers to a clinical pharmacogenetics service. *Personalized Medicine*, **2008**, 5, 471-480	2.2	19
140	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

139	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
138	TPMT testing in azathioprine: a R cost-effective use of healthcare resourcesR. *Personalized Medicine*, **2009**, 6, 103-113	2.2	18
137	Clinical Pharmacogenetics Implementation Consortium Guideline for the Use of Aminoglycosides Based on MT-RNR1 Genotype. *Clinical Pharmacology and Therapeutics*, **2021**,	6.1	18
136	Delivery of a clinical genomics service. *Genes*, **2014**, 5, 1001-17	4.2	17
135	Genetic analysis of thiopurine methyltransferase polymorphism in the Jordanian population. *European Journal of Clinical Pharmacology*, **2010**, 66, 999-1003	2.8	17
134	Array comparative genomic hybridization for diagnosis of developmental delay: an exploratory cost-consequences analysis. *Clinical Genetics*, **2007**, 71, 254-9	4	17
133	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
132	G6PC3 mutations cause non-syndromic severe congenital neutropenia. *Molecular Genetics and Metabolism*, **2013**, 108, 138-41	3.7	16
131	Variability of bone marrow morphology in G6PC3 mutations: is there a genotype-phenotype correlation or age-dependent relationship?. *American Journal of Hematology*, **2011**, 86, 235-7	7.1	16
130	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
129	Lrig2 and Hpse2, mutated in urofacial syndrome, pattern nerves in the urinary bladder. *Kidney International*, **2019**, 95, 1138-1152	9.9	15
128	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
127	Genetics of human congenital urinary bladder disease. *Pediatric Nephrology*, **2014**, 29, 353-60	3.2	15
126	Pharmacogenetic testing in the United Kingdom genetics and immunogenetics laboratories. *Genetic Testing and Molecular Biomarkers*, **2010**, 14, 121-5	1.6	15
125	Pharmacogenetics and pharmacogenomics: a clinical reality. *Annals of Clinical Biochemistry*, **2011**, 48, 410-7	2.2	15
124	RASSF1A polymorphism in familial breast cancer. *Familial Cancer*, **2010**, 9, 263-5	3	15
123	Genomic and healthcare dynamics of nosocomial SARS-CoV-2 transmission. *ELife*, **2021**, 10,	8.9	15
122	Delivering a pharmacogenetic service: is there a role for genetic counselors?. *Journal of Genetic Counseling*, **2012**, 21, 527-35	2.5	14

121	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
120	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
119	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
118	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
117	The Impact of CYP2D6 Genotyping on Tamoxifen Treatment. *Pharmaceuticals*, **2010**, 3, 1122-1138	5.2	13
116	Early diagnosis and treatment of cobalamin deficiency of infancy owing to occult maternal pernicious anemia. *Journal of Pediatric Hematology/Oncology*, **2010**, 32, 319-22	1.2	13
115	Refusal of viral testing during the SARS-CoV-2 pandemic. *Clinical Medicine*, **2020**, 20, e163-e164	1.9	13
114	Impaired eIF5A function causes a Mendelian disorder that is partially rescued in model systems by spermidine. *Nature Communications*, **2021**, 12, 833	17.4	13
113	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
112	Disease modeling of core pre-mRNA splicing factor haploinsufficiency. *Human Molecular Genetics*, **2019**, 28, 3704-3723	5.6	12
111	The :p.Arg658* truncating variant is associated with risk of triple-negative breast cancer. *Npj Breast Cancer*, **2019**, 5, 38	7.8	12
110	PCOS and peripheral AMH levels in relation to FSH receptor gene single nucleotide polymorphisms. *Gynecological Endocrinology*, **2012**, 28, 375-7	2.4	12
109	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
108	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
107	Avoidance of CYP2D6 inhibitors in patients receiving tamoxifen. *Journal of Clinical Oncology*, **2010**, 28, e584-5; author reply e586	2.2	12
106	Breast Cancer Polygenic Risk Score and Contralateral Breast Cancer Risk. *American Journal of Human Genetics*, **2020**, 107, 837-848	11	12
105	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
104	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

103	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
102	Pharmacogenetics of aromatase inhibitors. *Pharmacogenomics*, **2012**, 13, 699-707	2.6	11
101	Advantages of a subcutaneous implantable cardioverter-defibrillator in LAMP2 hypertrophic cardiomyopathy. *Journal of Cardiovascular Electrophysiology*, **2013**, 24, 1051-3	2.7	11
100	Loss-of-function variants in myocardin cause congenital megabladder in humans and mice. *Journal of Clinical Investigation*, **2019**, 129, 5374-5380	15.9	11
99	Functional and in-silico interrogation of rare genomic variants impacting RNA splicing for the diagnosis of genomic disorders		10
98	Diagnosing and Preventing Hearing Loss in the Genomic Age. *Trends in Hearing*, **2019**, 23, 2331216519878983	3.2	9
97	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
96	Inflammatory bowel disease in the South Asian population of Northwest England. *American Journal of Gastroenterology*, **2008**, 103, 242-3; author reply 243-4	0.7	8
95	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
94	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
93	Accounting for Capacity Constraints in Economic Evaluations of Precision Medicine: A Systematic Review. *Pharmacoeconomics*, **2019**, 37, 1011-1027	4.4	7
92	Geroderma osteodysplastica maps to a 4 Mb locus on chromosome 1q24. *American Journal of Medical Genetics, Part A*, **2008**, 146A, 3034-7	2.5	7
91	EFTUD2 missense variants disrupt protein function and splicing in mandibulofacial dysostosis Guion-Almeida type. *Human Mutation*, **2020**, 41, 1372-1382	4.7	6
90	Exome Sequencing Identifies a Dominant TNNT3 Mutation in a Large Family with Distal Arthrogryposis. *Molecular Syndromology*, **2014**, 5, 218-28	1.5	6
89	Marfanoid habitus is a nonspecific feature of Perrault syndrome. *Clinical Dysmorphology*, **2017**, 26, 200-204	0.9	6
88	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
87	Breast cancer pharmacogenomics: where we are going. *Pharmacogenomics*, **2012**, 13, 629-31	2.6	6
86	Is Involved in Urinary Tract and Urorectal Development. *Frontiers in Cell and Developmental Biology*, **2020**, 8, 567	5.7	6

85	Modelling the developmental spliceosomal craniofacial disorder Burn-McKeown syndrome using induced pluripotent stem cells. *PLoS ONE*, **2020**, 15, e0233582	3.7	6
84	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
83	Clinical and genetic heterogeneity in Melkersson-Rosenthal Syndrome. *European Journal of Medical Genetics*, **2019**, 62, 103536	2.6	6
82	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
81	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
80	Rapid Point-of-Care Genotyping to Avoid Aminoglycoside-Induced Ototoxicity in Neonatal Intensive Care.. *JAMA Pediatrics*, **2022**,	8.3	6
79	A role for genetic counsellors and clinical geneticists in pharmacogenetics?. *Clinical Genetics*, **2012**, 82, 201-2; author reply 203	4	5
78	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
77	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
76	Identification of a novel familial FGF16 mutation in metacarpal 4-5 fusion. *Clinical Dysmorphology*, **2014**, 23, 95-97	0.9	5
75	Lack of caveolin-1 (P132L) somatic mutations in breast cancer. *Breast Cancer Research and Treatment*, **2012**, 132, 1185-6	4.4	5
74	Pharmacogenomics in the UK National Health Service: opportunities and challenges. *Pharmacogenomics*, **2020**, 21, 1237-1246	2.6	5
73	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
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