Nazneen Rahman

List of Publications by Year in Descending Order

177 papers	28,340 citations	76 h-index	168 g-index
185 ext. papers	32,110 ext. citations	15.6 avg, IF	6.24 L-index

#	Paper	IF	Citations
177	Chromosomal instability by mutations in the novel minor spliceosome component CENATAC. *EMBO Journal*, **2021**, 40, e106536	13	4
176	Absence of the TRIP13 c.1060C>T Mutation in Wilms Tumor Patients From Pakistan. *Journal of Pediatric Hematology/Oncology*, **2020**, 42, e128-e131	1.2	
175	Insights into BRCA Cancer Predisposition from Integrated Germline and Somatic Analyses in 7632 Cancers. *JNCI Cancer Spectrum*, **2019**, 3, pkz028	4.6	6
174	Resolving the full spectrum of human genome variation using Linked-Reads. *Genome Research*, **2019**, 29, 635-645	9.7	86
173	Identification of new Wilms tumour predisposition genes: an exome sequencing study. *The Lancet Child and Adolescent Health*, **2019**, 3, 322-331	14.5	46
172	Genomic variant sharing: a position statement. *Wellcome Open Research*, **2019**, 4, 22	4.8	16
171	Associations of obesity and circulating insulin and glucose with breast cancer risk: a Mendelian randomization analysis. *International Journal of Epidemiology*, **2019**, 48, 795-806	7.8	52
170	Carboplatin in BRCA1/2-mutated and triple-negative breast cancer BRCAness subgroups: the TNT Trial. *Nature Medicine*, **2018**, 24, 628-637	50.5	410
169	A transcriptome-wide association study of 229,000 women identifies new candidate susceptibility genes for breast cancer. *Nature Genetics*, **2018**, 50, 968-978	36.3	101
168	Mosaic-variegated aneuploidy syndrome mutation or haploinsufficiency in Cep57 impairs tumor suppression. *Journal of Clinical Investigation*, **2018**, 128, 3517-3534	15.9	10
167	CoverView: a sequence quality evaluation tool for next generation sequencing data. *Wellcome Open Research*, **2018**, 3, 36	4.8	5
166	The Quality Sequencing Minimum (QSM): providing comprehensive, consistent, transparent next generation sequencing data quality assurance. *Wellcome Open Research*, **2018**, 3, 37	4.8	5
165	The Tatton-Brown-Rahman Syndrome: A clinical study of 55 individuals with constitutive variants. *Wellcome Open Research*, **2018**, 3, 46	4.8	41
164	The ICR639 CPG NGS validation series: A resource to assess analytical sensitivity of cancer predisposition gene testing. *Wellcome Open Research*, **2018**, 3, 68	4.8	4
163	ICR142 Benchmarker: evaluating, optimising and benchmarking variant calling performance using the ICR142 NGS validation series. *Wellcome Open Research*, **2018**, 3, 108	4.8	
162	ICR142 Benchmarker: evaluating, optimising and benchmarking variant calling using the ICR142 NGS validation series. *Wellcome Open Research*, **2018**, 3, 108	4.8	
161	Biallelic TRIP13 mutations predispose to Wilms tumor and chromosome missegregation. *Nature Genetics*, **2017**, 49, 1148-1151	36.3	75

160	A Cost-Effectiveness Evaluation of Germline BRCA1 and BRCA2 Testing in UK Women with Ovarian Cancer. *Value in Health*, **2017**, 20, 567-576	3.3	39
159	Association analysis identifies 65 new breast cancer risk loci. *Nature*, **2017**, 551, 92-94	50.4	643
158	Identification of ten variants associated with risk of estrogen-receptor-negative breast cancer. *Nature Genetics*, **2017**, 49, 1767-1778	36.3	186
157	Body mass index and breast cancer survival: a Mendelian randomization analysis. *International Journal of Epidemiology*, **2017**, 46, 1814-1822	7.8	27
156	The ICR96 exon CNV validation series: a resource for orthogonal assessment of exon CNV calling in NGS data. *Wellcome Open Research*, **2017**, 2, 35	4.8	7
155	Common variants upstream of MLF1 at 3q25 and within CPZ at 4p16 associated with neuroblastoma. *PLoS Genetics*, **2017**, 13, e1006787	6	40
154	Mutations in Epigenetic Regulation Genes Are a Major Cause of Overgrowth with Intellectual Disability. *American Journal of Human Genetics*, **2017**, 100, 725-736	11	103
153	The integration of BRCA testing into oncology clinics. *British Journal of Nursing*, **2016**, 25, 690-4	0.7	37
152	Genome-Wide Meta-Analyses of Breast, Ovarian, and Prostate Cancer Association Studies Identify Multiple New Susceptibility Loci Shared by at Least Two Cancer Types. *Cancer Discovery*, **2016**, 6, 1052-67	24.4	104
151	Identification of four novel susceptibility loci for oestrogen receptor negative breast cancer. *Nature Communications*, **2016**, 7, 11375	17.4	64
150	OpEx - a validated, automated pipeline optimised for clinical exome sequence analysis. *Scientific Reports*, **2016**, 6, 31029	4.9	10
149	A cost analysis of a cancer genetic service model in the UK. *Journal of Community Genetics*, **2016**, 7, 185-94	2.5	24
148	The ICR142 NGS validation series: a resource for orthogonal assessment of NGS analysis. *F1000Research*, **2016**, 5, 386	3.6	5
147	The ICR142 NGS validation series: a resource for orthogonal assessment of NGS analysis. *F1000Research*, **2016**, 5, 386	3.6	5
146	Accurate clinical detection of exon copy number variants in a targeted NGS panel using DECoN. *Wellcome Open Research*, **2016**, 1, 20	4.8	40
145	Implementing rapid, robust, cost-effective, patient-centred, routine genetic testing in ovarian cancer patients. *Scientific Reports*, **2016**, 6, 29506	4.9	114
144	Identification of independent association signals and putative functional variants for breast cancer risk through fine-scale mapping of the 12p11 locus. *Breast Cancer Research*, **2016**, 18, 64	8.3	25
143	Cross-Cancer Genome-Wide Analysis of Lung, Ovary, Breast, Prostate, and Colorectal Cancer Reveals Novel Pleiotropic Associations. *Cancer Research*, **2016**, 76, 5103-14	10.1	66

142	Genome-wide association analysis of more than 120,000 individuals identifies 15 new susceptibility loci for breast cancer. *Nature Genetics*, **2015**, 47, 373-80	36.3	406
141	Mutations in the PP2A regulatory subunit B family genes PPP2R5B, PPP2R5C and PPP2R5D cause human overgrowth. *Human Molecular Genetics*, **2015**, 24, 4775-9	5.6	48
140	Identification of novel genetic markers of breast cancer survival. *Journal of the National Cancer Institute*, **2015**, 107,	9.7	38
139	Development of cancer genetic services in the UK: A national consultation. *Genome Medicine*, **2015**, 7, 18	14.4	29
138	Multi-stage genome-wide association study identifies new susceptibility locus for testicular germ cell tumour on chromosome 3q25. *Human Molecular Genetics*, **2015**, 24, 1169-76	5.6	26
137	Mutations in the transcriptional repressor REST predispose to Wilms tumor. *Nature Genetics*, **2015**, 47, 1471-4	36.3	36
136	Genetic predisposition to neuroblastoma mediated by a LMO1 super-enhancer polymorphism. *Nature*, **2015**, 528, 418-21	50.4	201
135	Identification and characterization of novel associations in the CASP8/ALS2CR12 region on chromosome 2 with breast cancer risk. *Human Molecular Genetics*, **2015**, 24, 285-98	5.6	35
134	Common germline polymorphisms associated with breast cancer-specific survival. *Breast Cancer Research*, **2015**, 17, 58	8.3	24
133	CSN and CAVA: variant annotation tools for rapid, robust next-generation sequencing analysis in the clinical setting. *Genome Medicine*, **2015**, 7, 76	14.4	41
132	Prediction of breast cancer risk based on profiling with common genetic variants. *Journal of the National Cancer Institute*, **2015**, 107,	9.7	324
131	Gene-panel sequencing and the prediction of breast-cancer risk. *New England Journal of Medicine*, **2015**, 372, 2243-57	59.2	587
130	Abstract S3-01: The TNT trial: A randomized phase III trial of carboplatin (C) compared with docetaxel (D) for patients with metastatic or recurrent locally advanced triple negative orBRCA1/2breast cancer (CRUK/07/012) **2015**,		52
129	The ICR1000 UK exome series: a resource of gene variation in an outbred population. *F1000Research*, **2015**, 4, 883	3.6	20
128	A genome-wide association study of early-onset breast cancer identifies PFKM as a novel breast cancer gene and supports a common genetic spectrum for breast cancer at any age. *Cancer Epidemiology Biomarkers and Prevention*, **2014**, 23, 658-69	4	63
127	Mutations in the DNA methyltransferase gene DNMT3A cause an overgrowth syndrome with intellectual disability. *Nature Genetics*, **2014**, 46, 385-8	36.3	196
126	Realizing the promise of cancer predisposition genes. *Nature*, **2014**, 505, 302-8	50.4	340
125	Characterizing genetic variants for clinical action. *American Journal of Medical Genetics, Part C: Seminars in Medical Genetics*, **2014**, 166C, 93-104	3.1	41

124	Germline mutations in the PAF1 complex gene CTR9 predispose to Wilms tumour. *Nature Communications*, **2014**, 5, 4398	17.4	53
123	Breast-cancer risk in families with mutations in PALB2. *New England Journal of Medicine*, **2014**, 371, 497-506	59.2	576
122	Pathway-based analysis of GWAs data identifies association of sex determination genes with susceptibility to testicular germ cell tumors. *Human Molecular Genetics*, **2014**, 23, 6061-8	5.6	25
121	MicroRNA related polymorphisms and breast cancer risk. *PLoS ONE*, **2014**, 9, e109973	3.7	37
120	Mainstreaming genetic testing of cancer predisposition genes. *Clinical Medicine*, **2014**, 14, 436-9	1.9	42
119	Genetic Predisposition to Wilms Tumour. *Pediatric Oncology*, **2014**, 19-38	0.5	1
118	Testicular germ cell tumor susceptibility associated with the UCK2 locus on chromosome 1q23. *Human Molecular Genetics*, **2013**, 22, 2748-53	5.6	53
117	Mosaic PPM1D mutations are associated with predisposition to breast and ovarian cancer. *Nature*, **2013**, 493, 406-10	50.4	191
116	Simple detection of germline microsatellite instability for diagnosis of constitutional mismatch repair cancer syndrome. *Human Mutation*, **2013**, 34, 847-52	4.7	35
115	Genome-wide association studies identify four ER negative-specific breast cancer risk loci. *Nature Genetics*, **2013**, 45, 392-8, 398e1-2	36.3	327
114	Large-scale genotyping identifies 41 new loci associated with breast cancer risk. *Nature Genetics*, **2013**, 45, 353-61, 361e1-2	36.3	813
113	Meta-analysis identifies four new loci associated with testicular germ cell tumor. *Nature Genetics*, **2013**, 45, 680-5	36.3	132
112	Identification of nine new susceptibility loci for testicular cancer, including variants near DAZL and PRDM14. *Nature Genetics*, **2013**, 45, 686-9	36.3	128
111	Weaver syndrome and EZH2 mutations: Clarifying the clinical phenotype. *American Journal of Medical Genetics, Part A*, **2013**, 161A, 2972-80	2.5	80
110	The NSD1 and EZH2 overgrowth genes, similarities and differences. *American Journal of Medical Genetics, Part C: Seminars in Medical Genetics*, **2013**, 163C, 86-91	3.1	56
109	Genome-wide association analysis identifies three new breast cancer susceptibility loci. *Nature Genetics*, **2012**, 44, 312-8	36.3	237
108	A meta-analysis of genome-wide association studies of breast cancer identifies two novel susceptibility loci at 6q14 and 20q11. *Human Molecular Genetics*, **2012**, 21, 5373-84	5.6	143
107	Breast cancer risk and 6q22.33: combined results from Breast Cancer Association Consortium and Consortium of Investigators on Modifiers of BRCA1/2. *PLoS ONE*, **2012**, 7, e35706	3.7	10

106	Stratification of Wilms tumor by genetic and epigenetic analysis. *Oncotarget*, **2012**, 3, 327-35	3.3	78
105	Predisposition gene identification in common cancers by exome sequencing: insights from familial breast cancer. *Breast Cancer Research and Treatment*, **2012**, 134, 429-33	4.4	46
104	Gene-gene interactions in breast cancer susceptibility. *Human Molecular Genetics*, **2012**, 21, 958-62	5.6	33
103	Mammographic breast density and breast cancer: evidence of a shared genetic basis. *Cancer Research*, **2012**, 72, 1478-84	10.1	50
102	BRCA1 testing should be offered to individuals with triple-negative breast cancer diagnosed below 50 years. *British Journal of Cancer*, **2012**, 106, 1234-8	8.7	76
101	A genome-wide association study identifies susceptibility loci for Wilms tumor. *Nature Genetics*, **2012**, 44, 681-4	36.3	58
100	Bayesian refinement of association signals for 14 loci in 3 common diseases. *Nature Genetics*, **2012**, 44, 1294-301	36.3	347
99	Germline RAD51C mutations confer susceptibility to ovarian cancer. *Nature Genetics*, **2012**, 44, 475-6; author reply 476	36.3	190
98	Comparison of 6q25 breast cancer hits from Asian and European Genome Wide Association Studies in the Breast Cancer Association Consortium (BCAC). *PLoS ONE*, **2012**, 7, e42380	3.7	49
97	Exploring the link between MORF4L1 and risk of breast cancer. *Breast Cancer Research*, **2011**, 13, R40	8.3	16
96	Germline mutations in the oncogene EZH2 cause Weaver syndrome and increased human height. *Oncotarget*, **2011**, 2, 1127-33	3.3	110
95	Mutations in CEP57 cause mosaic variegated aneuploidy syndrome. *Nature Genetics*, **2011**, 43, 527-9	36.3	98
94	Integrative genomics identifies LMO1 as a neuroblastoma oncogene. *Nature*, **2011**, 469, 216-20	50.4	231
93	Heterogeneity of familial medulloblastoma and contribution of germline PTCH1 and SUFU mutations to sporadic medulloblastoma. *Familial Cancer*, **2011**, 10, 337-42	3	51
92	Confirmation of 5p12 as a susceptibility locus for progesterone-receptor-positive, lower grade breast cancer. *Cancer Epidemiology Biomarkers and Prevention*, **2011**, 20, 2222-31	4	27
91	Facial dysmorphism and digit anomalies in three siblings with severe developmental delay. *Clinical Dysmorphology*, **2011**, 20, 92-94	0.9	
90	Associations of common variants at 1p11.2 and 14q24.1 (RAD51L1) with breast cancer risk and heterogeneity by tumor subtype: findings from the Breast Cancer Association Consortium. *Human Molecular Genetics*, **2011**, 20, 4693-706	5.6	66
89	Common variants of the BRCA1 wild-type allele modify the risk of breast cancer in BRCA1 mutation carriers. *Human Molecular Genetics*, **2011**, 20, 4732-47	5.6	21

88	Germline mutations in RAD51D confer susceptibility to ovarian cancer. *Nature Genetics*, **2011**, 43, 879-882	36.3	379
87	DICER1 syndrome: clarifying the diagnosis, clinical features and management implications of a pleiotropic tumour predisposition syndrome. *Journal of Medical Genetics*, **2011**, 48, 273-8	5.8	278
86	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
85	Mutation of the RAD51C gene in a Fanconi anemia-like disorder. *Nature Genetics*, **2010**, 42, 406-9	36.3	311
84	Genome-wide association study identifies five new breast cancer susceptibility loci. *Nature Genetics* , **2010**, 42, 504-7	36.3	582
83	Variants near DMRT1, TERT and ATF7IP are associated with testicular germ cell cancer. *Nature Genetics*, **2010**, 42, 604-7	36.3	289
82	Molecular causes for BUBR1 dysfunction in the human cancer predisposition syndrome mosaic variegated aneuploidy. *Cancer Research*, **2010**, 70, 4891-900	10.1	85
81	A novel HER2-positive breast cancer phenotype arising from germline TP53 mutations. *Journal of Medical Genetics*, **2010**, 47, 771-4	5.8	85
80	Generation of trisomies in cancer cells by multipolar mitosis and incomplete cytokinesis. *Proceedings of the National Academy of Sciences of the United States of America*, **2010**, 107, 20489-93	11.5	57
79	Mutation and association analysis of GEN1 in breast cancer susceptibility. *Breast Cancer Research and Treatment*, **2010**, 124, 283-8	4.4	7
78	A new familial cancer syndrome including predisposition to Wilms tumor and neuroblastoma. *Familial Cancer*, **2010**, 9, 425-30	3	8
77	Epigenetic inactivation of the Sotos overgrowth syndrome gene histone methyltransferase NSD1 in human neuroblastoma and glioma. *Proceedings of the National Academy of Sciences of the United States of America*, **2009**, 106, 21830-5	11.5	162
76	Risk of estrogen receptor-positive and -negative breast cancer and single-nucleotide polymorphism 2q35-rs13387042. *Journal of the National Cancer Institute*, **2009**, 101, 1012-8	9.7	90
75	15q overgrowth syndrome: a newly recognized phenotype associated with overgrowth, learning difficulties, characteristic facial appearance, renal anomalies and increased dosage of distal chromosome 15q. *American Journal of Medical Genetics, Part A*, **2009**, 149A, 147-54	2.5	41
74	Newly discovered breast cancer susceptibility loci on 3p24 and 17q23.2. *Nature Genetics*, **2009**, 41, 585-90	36.3	393
73	Common variations in BARD1 influence susceptibility to high-risk neuroblastoma. *Nature Genetics*, **2009**, 41, 718-23	36.3	226
72	A genome-wide association study of testicular germ cell tumor. *Nature Genetics*, **2009**, 41, 807-10	36.3	282
71	The emerging landscape of breast cancer susceptibility. *Nature Genetics*, **2008**, 40, 17-22	36.3	365

70	Constitutional 11p15 abnormalities, including heritable imprinting center mutations, cause nonsyndromic Wilms tumor. *Nature Genetics*, **2008**, 40, 1329-34	36.3	134
69	Genetic predisposition to breast cancer: past, present, and future. *Annual Review of Genomics and Human Genetics*, **2008**, 9, 321-45	9.7	202
68	Chromosome 6p22 locus associated with clinically aggressive neuroblastoma. *New England Journal of Medicine*, **2008**, 358, 2585-93	59.2	224
67	Association study of prostate cancer susceptibility variants with risks of invasive ovarian, breast, and colorectal cancer. *Cancer Research*, **2008**, 68, 8837-42	10.1	10
66	Mosaic variegated aneuploidy without microcephaly: implications for cytogenetic diagnosis. *American Journal of Medical Genetics, Part A*, **2007**, 143A, 1890-3	2.5	13
65	Association scan of 14,500 nonsynonymous SNPs in four diseases identifies autoimmunity variants. *Nature Genetics*, **2007**, 39, 1329-37	36.3	1130
64	Biallelic mutations in PALB2 cause Fanconi anemia subtype FA-N and predispose to childhood cancer. *Nature Genetics*, **2007**, 39, 162-4	36.3	485
63	PALB2, which encodes a BRCA2-interacting protein, is a breast cancer susceptibility gene. *Nature Genetics*, **2007**, 39, 165-7	36.3	719
62	A common coding variant in CASP8 is associated with breast cancer risk. *Nature Genetics*, **2007**, 39, 352-8	36.3	557
61	Mutations in RNF135, a gene within the NF1 microdeletion region, cause phenotypic abnormalities including overgrowth. *Nature Genetics*, **2007**, 39, 963-5	36.3	90
60	Sotos syndrome. *European Journal of Human Genetics*, **2007**, 15, 264-71	5.3	113
59	Genome-wide association study identifies novel breast cancer susceptibility loci. *Nature*, **2007**, 447, 1087-93	36.3	1957
58	Medulloblastoma, acute myelocytic leukemia and colonic carcinomas in a child with biallelic MSH6 mutations. *Nature Clinical Practice Oncology*, **2007**, 4, 130-4		73
57	Familial T-cell non-Hodgkin lymphoma caused by biallelic MSH2 mutations. *Journal of Medical Genetics*, **2007**, 44, e83	5.8	35
56	Estrogen receptor status could modulate the genomic pattern in familial and sporadic breast cancer. *Clinical Cancer Research*, **2007**, 13, 7305-13	12.9	28
55	Cancer genes associated with phenotypes in monoallelic and biallelic mutation carriers: new lessons from old players. *Human Molecular Genetics*, **2007**, 16 Spec No 1, R60-6	5.6	68
54	A genome wide linkage search for breast cancer susceptibility genes. *Genes Chromosomes and Cancer*, **2006**, 45, 646-55	5	100
53	Evaluation of RAD50 in familial breast cancer predisposition. *International Journal of Cancer*, **2006**, 118, 2911-6	7.5	47

52	PHOX2B analysis in non-syndromic neuroblastoma cases shows novel mutations and genotype-phenotype associations. *American Journal of Medical Genetics, Part A*, **2006**, 140, 1297-301	2.5	39
51	Surveillance for Wilms tumour in at-risk children: pragmatic recommendations for best practice. *Archives of Disease in Childhood*, **2006**, 91, 995-9	2.2	122
50	A multicenter study of cancer incidence in CHEK2 1100delC mutation carriers. *Cancer Epidemiology Biomarkers and Prevention*, **2006**, 15, 2542-5	4	44
49	Syndromes and constitutional chromosomal abnormalities associated with Wilms tumour. *Journal of Medical Genetics*, **2006**, 43, 705-15	5.8	217
48	Comparative genomic hybridization and BUB1B mutation analyses in childhood cancers associated with mosaic variegated aneuploidy syndrome. *Cancer Letters*, **2006**, 239, 234-8	9.9	31
47	ATM mutations that cause ataxia-telangiectasia are breast cancer susceptibility alleles. *Nature Genetics*, **2006**, 38, 873-5	36.3	553
46	Truncating mutations in the Fanconi anemia J gene BRIP1 are low-penetrance breast cancer susceptibility alleles. *Nature Genetics*, **2006**, 38, 1239-41	36.3	553
45	ATM and breast cancer susceptibility. *Oncogene*, **2006**, 25, 5906-11	9.2	167
44	Genotype-phenotype associations in Sotos syndrome: an analysis of 266 individuals with NSD1 aberrations. *American Journal of Human Genetics*, **2005**, 77, 193-204	11	218
43	Mechanisms predisposing to childhood overgrowth and cancer. *Current Opinion in Genetics and Development*, **2005**, 15, 227-33	4.9	38
42	Distinct genomic profiles in hereditary breast tumors identified by array-based comparative genomic hybridization. *Cancer Research*, **2005**, 65, 7612-21	10.1	141
41	Evaluation of NSD2 and NSD3 in overgrowth syndromes. *European Journal of Human Genetics*, **2005**, 13, 150-3	5.3	26
40	Periodontal treatment of two siblings with juvenile hyaline fibromatosis. *Journal of Clinical Periodontology*, **2005**, 32, 1016-21	7.7	16
39	Familial gigantism caused by an NSD1 mutation. *American Journal of Medical Genetics, Part A*, **2005**, 139, 40-4	2.5	12
38	Confirmation of a hereditary motor and sensory neuropathy IIC locus at chromosome 12q23-q24. *Annals of Neurology*, **2005**, 57, 293-7	9.4	25
37	A WT1 exon 1 mutation in a child diagnosed with Denys-Drash syndrome. *Pediatric Nephrology*, **2005**, 20, 81-5	3.2	23
36	Direct role of PDGF-BB in lymphangiogenesis and lymphatic metastasis. *Cell Cycle*, **2005**, 4, 228-30	4.7	65
35	Biallelic BRCA2 mutations are associated with multiple malignancies in childhood including familial Wilms tumour. *Journal of Medical Genetics*, **2005**, 42, 147-51	5.8	86

34	Frequency and heritability of WT1 mutations in nonsyndromic Wilms tumor patients: a UK Children's Cancer Study Group Study. *Journal of Clinical Oncology*, **2004**, 22, 4140-6	2.2	85
33	Constitutional aneuploidy and cancer predisposition caused by biallelic mutations in BUB1B. *Nature Genetics*, **2004**, 36, 1159-61	36.3	471
32	A census of human cancer genes. *Nature Reviews Cancer*, **2004**, 4, 177-83	31.3	2424
31	Clinical features of NSD1-positive Sotos syndrome. *Clinical Dysmorphology*, **2004**, 13, 199-204	0.9	72
30	Clinical features of NSD1-positive Sotos syndrome. *Clinical Dysmorphology*, **2004**, 13, 199-204	0.9	19
29	Skeletal muscle involvement in infantile systemic hyalinosis. *European Journal of Paediatric Neurology*, **2003**, 7, 401-6	3.8	5
28	Mutations of the Uromodulin gene in MCKD type 2 patients cluster in exon 4, which encodes three EGF-like domains. *Kidney International*, **2003**, 64, 1580-7	9.9	73
27	NSD1 mutations are the major cause of Sotos syndrome and occur in some cases of Weaver syndrome but are rare in other overgrowth phenotypes. *American Journal of Human Genetics*, **2003**, 72, 132-43	11	221
26	Variants in CHEK2 other than 1100delC do not make a major contribution to breast cancer susceptibility. *American Journal of Human Genetics*, **2003**, 72, 1023-8	11	104
25	Ehlers-Danlos syndrome with severe early-onset periodontal disease (EDS-VIII) is a distinct, heterogeneous disorder with one predisposition gene at chromosome 12p13. *American Journal of Human Genetics*, **2003**, 73, 198-204	11	40
24	Mutations in the gene encoding capillary morphogenesis protein 2 cause juvenile hyaline fibromatosis and infantile systemic hyalinosis. *American Journal of Human Genetics*, **2003**, 73, 791-800	11	180
23	Human chromosome 7: DNA sequence and biology. *Science*, **2003**, 300, 767-72	33.3	159
22	Evaluation of Fanconi Anemia genes in familial breast cancer predisposition. *Cancer Research*, **2003**, 63, 8596-9	10.1	41
21	Low-penetrance susceptibility to breast cancer due to CHEK2(*)1100delC in noncarriers of BRCA1 or BRCA2 mutations. *Nature Genetics*, **2002**, 31, 55-9	36.3	863
20	Evaluation of linkage of breast cancer to the putative BRCA3 locus on chromosome 13q21 in 128 multiple case families from the Breast Cancer Linkage Consortium. *Proceedings of the National Academy of Sciences of the United States of America*, **2002**, 99, 827-31	11.5	64
19	Familial vocal fold paralysis. *Journal of Laryngology and Otology*, **2002**, 116, 1047-9	1.8	19
18	The gene for juvenile hyaline fibromatosis maps to chromosome 4q21. *American Journal of Human Genetics*, **2002**, 71, 975-80	11	62
17	Confirmation of a gene locus for medullary cystic kidney disease (MCKD2) on chromosome 16p12. *Kidney International*, **2001**, 60, 1233-9	9.9	19

16	Case of interstitial 12q deletion in association with Wilms tumor. *American Journal of Medical Genetics Part A*, **2001**, 104, 246-249		13
15	Localization of the gene for distal hereditary motor neuronopathy VII (dHMN-VII) to chromosome 2q14. *American Journal of Human Genetics*, **2001**, 68, 1270-6	11	63
14	Absence of evidence for a familial breast cancer susceptibility gene at chromosome 8p12-p22. *Oncogene*, **2000**, 19, 4170-3	9.2	32
13	Prevalence of BRCA1 and BRCA2 gene mutations in patients with early-onset breast cancer. *Journal of the National Cancer Institute*, **1999**, 91, 943-9	9.7	654
12	The gene for cherubism maps to chromosome 4p16.3. *American Journal of Human Genetics*, **1999**, 65, 151-7	11	117
11	A gene for lymphedema-distichiasis maps to 16q24.3. *American Journal of Human Genetics*, **1999**, 65, 427-32	11	98
10	The genetics of breast cancer susceptibility. *Annual Review of Genetics*, **1998**, 32, 95-121	14.5	221
9	The familial WilmsQtumour susceptibility gene, FWT1, may not be a tumour suppressor gene. *Oncogene*, **1997**, 14, 3099-102	9.2	15
8	Evidence for a familial WilmsQtumour gene (FWT1) on chromosome 17q12-q21. *Nature Genetics*, **1996**, 13, 461-3	36.3	145
7	Genomic variant sharing: a position statement. *Wellcome Open Research*,4, 22	4.8	5
6	Clinical Annotation Reference Templates: a resource for consistent variant annotation. *Wellcome Open Research*,3, 146	4.8	
5	A discrete event simulation to evaluate the cost effectiveness of germline BRCA1 and BRCA2 testing in UK women with ovarian cancer		1
4	One in seven pathogenic variants can be challenging to detect by NGS: An analysis of 450,000 patients with implications for clinical sensitivity and genetic test implementation		3
3	Chromosomal instability by mutations in a novel specificity factor of the minor spliceosome		1
2	An interlaboratory study of complex variant detection		4
1	Implementing rapid, robust, cost-effective, patient-centred, routine genetic testing in ovarian cancer patients		1