Nazneen Rahman

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/9276797/publications.pdf

Version: 2024-02-01

		5558	4323
173	34,575	82	173
papers	citations	h-index	g-index

185	185	185	37552
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	A census of human cancer genes. Nature Reviews Cancer, 2004, 4, 177-183.	12.8	2,868
2	Genome-wide association study identifies novel breast cancer susceptibility loci. Nature, 2007, 447, 1087-1093.	13.7	2,165
3	Association scan of 14,500 nonsynonymous SNPs in four diseases identifies autoimmunity variants. Nature Genetics, 2007, 39, 1329-1337.	9.4	1,298
4	Association analysis identifies 65 new breast cancer risk loci. Nature, 2017, 551, 92-94.	13.7	1,099
5	Low-penetrance susceptibility to breast cancer due to CHEK2*1100delC in noncarriers of BRCA1 or BRCA2 mutations. Nature Genetics, 2002, 31, 55-59.	9.4	1,001
6	Large-scale genotyping identifies 41 new loci associated with breast cancer risk. Nature Genetics, 2013, 45, 353-361.	9.4	960
7	PALB2, which encodes a BRCA2-interacting protein, is a breast cancer susceptibility gene. Nature Genetics, 2007, 39, 165-167.	9.4	858
8	Gene-Panel Sequencing and the Prediction of Breast-Cancer Risk. New England Journal of Medicine, 2015, 372, 2243-2257.	13.9	764
9	Prevalence of BRCA1 and BRCA2 Gene Mutations in Patients With Early-Onset Breast Cancer. Journal of the National Cancer Institute, 1999, 91, 943-949.	3.0	748
10	Breast-Cancer Risk in Families with Mutations in PALB2. New England Journal of Medicine, 2014, 371, 497-506.	13.9	745
11	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
12	Genome-wide association study identifies five new breast cancer susceptibility loci. Nature Genetics, 2010, 42, 504-507.	9.4	653
13	Carboplatin in BRCA1/2-mutated and triple-negative breast cancer BRCAness subgroups: the TNT Trial. Nature Medicine, 2018, 24, 628-637.	15.2	649
14	ATM mutations that cause ataxia-telangiectasia are breast cancer susceptibility alleles. Nature Genetics, 2006, 38, 873-875.	9.4	641
15	Truncating mutations in the Fanconi anemia J gene BRIP1 are low-penetrance breast cancer susceptibility alleles. Nature Genetics, 2006, 38, 1239-1241.	9.4	636
16	A common coding variant in CASP8 is associated with breast cancer risk. Nature Genetics, 2007, 39, 352-358.	9.4	591
17	Biallelic mutations in PALB2 cause Fanconi anemia subtype FA-N and predispose to childhood cancer. Nature Genetics, 2007, 39, 162-164.	9.4	556
18	Constitutional aneuploidy and cancer predisposition caused by biallelic mutations in BUB1B. Nature Genetics, 2004, 36, 1159-1161.	9.4	541

#	Article	IF	Citations
19	Genome-wide association analysis of more than 120,000 individuals identifies 15 new susceptibility loci for breast cancer. Nature Genetics, 2015, 47, 373-380.	9.4	513
20	Realizing the promise of cancer predisposition genes. Nature, 2014, 505, 302-308.	13.7	483
21	Bayesian refinement of association signals for 14 loci in 3 common diseases. Nature Genetics, 2012, 44, 1294-1301.	9.4	469
22	Germline mutations in RAD51D confer susceptibility to ovarian cancer. Nature Genetics, 2011, 43, 879-882.	9.4	460
23	Newly discovered breast cancer susceptibility loci on 3p24 and 17q23.2. Nature Genetics, 2009, 41, 585-590.	9.4	434
24	Prediction of Breast Cancer Risk Based on Profiling With Common Genetic Variants. Journal of the National Cancer Institute, 2015, 107, .	3.0	428
25	The emerging landscape of breast cancer susceptibility. Nature Genetics, 2008, 40, 17-22.	9.4	418
26	Genome-wide association studies identify four ER negativeâ€“specific breast cancer risk loci. Nature Genetics, 2013, 45, 392-398.	9.4	374
27	Mutation of the RAD51C gene in a Fanconi anemiaâ€“like disorder. Nature Genetics, 2010, 42, 406-409.	9.4	360
28	Variants near DMRT1, TERT and ATF7IP are associated with testicular germ cell cancer. Nature Genetics, 2010, 42, 604-607.	9.4	320
29	A genome-wide association study of testicular germ cell tumor. Nature Genetics, 2009, 41, 807-810.	9.4	317
30	DICER1 syndrome: clarifying the diagnosis, clinical features and management implications of a pleiotropic tumour predisposition syndrome. Journal of Medical Genetics, 2011, 48, 273-278.	1.5	312
31	Genotype-Phenotype Associations in Sotos Syndrome: An Analysis of 266 Individuals with NSD1 Aberrations. American Journal of Human Genetics, 2005, 77, 193-204.	2.6	298
32	Identification of ten variants associated with risk of estrogen-receptor-negative breast cancer. Nature Genetics, 2017, 49, 1767-1778.	9.4	289
33	Mutations in the DNA methyltransferase gene DNMT3A cause an overgrowth syndrome with intellectual disability. Nature Genetics, 2014, 46, 385-388.	9.4	280
34	Integrative genomics identifies LMO1 as a neuroblastoma oncogene. Nature, 2011, 469, 216-220.	13.7	276
35	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, 132-143.	2.6	273
36	Chromosome 6p22 Locus Associated with Clinically Aggressive Neuroblastoma. New England Journal of Medicine, 2008, 358, 2585-2593.	13.9	271

#	Article	IF	Citations
37	Common variations in BARD1 influence susceptibility to high-risk neuroblastoma. Nature Genetics, 2009, 41, 718-723.	9.4	266
38	Syndromes and constitutional chromosomal abnormalities associated with Wilms tumour. Journal of Medical Genetics, 2006, 43, 705-715.	1.5	264
39	Genetic predisposition to neuroblastoma mediated by a LMO1 super-enhancer polymorphism. Nature, 2015, 528, 418-421.	13.7	263
40	Genome-wide association analysis identifies three new breast cancer susceptibility loci. Nature Genetics, 2012, 44, 312-318.	9.4	256
41	THE GENETICS OF BREAST CANCER SUSCEPTIBILITY. Annual Review of Genetics, 1998, 32, 95-121.	3.2	245
42	Genetic Predisposition to Breast Cancer: Past, Present, and Future. Annual Review of Genomics and Human Genetics, 2008, 9, 321-345.	2.5	233
43	Germline RAD51C mutations confer susceptibility to ovarian cancer. Nature Genetics, 2012, 44, 475-476.	9.4	219
44	Mosaic PPM1D mutations are associated with predisposition to breast and ovarian cancer. Nature, 2013, 493, 406-410.	13.7	218
45	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.	2.6	209
46	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, 21830-21835.	3.3	190
47	ATM and breast cancer susceptibility. Oncogene, 2006, 25, 5906-5911.	2.6	189
48	Human Chromosome 7: DNA Sequence and Biology. Science, 2003, 300, 767-772.	6.0	185
49	A transcriptome-wide association study of 229,000 women identifies new candidate susceptibility genes for breast cancer. Nature Genetics, 2018, 50, 968-978.	9.4	184
50	Resolving the full spectrum of human genome variation using Linked-Reads. Genome Research, 2019, 29, 635-645.	2.4	182
51	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, 5373-5384.	1.4	168
52	Mutations in Epigenetic Regulation Genes Are a Major Cause of Overgrowth with Intellectual Disability. American Journal of Human Genetics, 2017, 100, 725-736.	2.6	168
53	Evidence for a familial Wilms' tumour gene (FWT1) on chromosome 17q12â€“q21. Nature Genetics, 1996, 13, 461-463.	9.4	166
54	Implementing rapid, robust, cost-effective, patient-centred, routine genetic testing in ovarian cancer patients. Scientific Reports, 2016, 6, 29506.	1.6	165

#	Article	IF	Citations
55	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, 1052-1067.	7.7	157
56	Constitutional 11p15 abnormalities, including heritable imprinting center mutations, cause nonsyndromic Wilms tumor. Nature Genetics, 2008, 40, 1329-1334.	9.4	154
57	Meta-analysis identifies four new loci associated with testicular germ cell tumor. Nature Genetics, 2013, 45, 680-685.	9.4	154
58	Identification of nine new susceptibility loci for testicular cancer, including variants near DAZL and PRDM14. Nature Genetics, 2013, 45, 686-689.	9.4	149
59	Distinct Genomic Profiles in Hereditary Breast Tumors Identified by Array-Based Comparative Genomic Hybridization. Cancer Research, 2005, 65, 7612-7621.	0.4	147
60	Sotos syndrome. European Journal of Human Genetics, 2007, 15, 264-271.	1.4	146
61	The Gene for Cherubism Maps to Chromosome 4p16.3. American Journal of Human Genetics, 1999, 65, 151-157.	2.6	145
62	Germline mutations in the oncogene EZH2 cause Weaver syndrome and increased human height. Oncotarget, 2011, 2, 1127-1133.	0.8	145
63	Surveillance for Wilms tumour in at-risk children: pragmatic recommendations for best practice. Archives of Disease in Childhood, 2006, 91, 995-999.	1.0	136
64	Variants in CHEK2 Other than 1100delC Do Not Make a Major Contribution to Breast Cancer Susceptibility. American Journal of Human Genetics, 2003, 72, 1023-1028.	2.6	119
65	Weaver syndrome and EZH2 mutations: Clarifying the clinical phenotype. American Journal of Medical Genetics, Part A, 2013, 161, 2972-2980.	0.7	119
66	Mutations in CEP57 cause mosaic variegated aneuploidy syndrome. Nature Genetics, 2011, 43, 527-529.	9.4	117
67	A genome wide linkage search for breast cancer susceptibility genes. Genes Chromosomes and Cancer, 2006, 45, 646-655.	1.5	111
68	Biallelic TRIP13 mutations predispose to Wilms tumor and chromosome missegregation. Nature Genetics, 2017, 49, 1148-1151.	9.4	111
69	A Gene for Lymphedema-Distichiasis Maps to 16q24.3. American Journal of Human Genetics, 1999, 65, 427-432.	2.6	106
70	Molecular Causes for BUBR1 Dysfunction in the Human Cancer Predisposition Syndrome Mosaic Variegated Aneuploidy. Cancer Research, 2010, 70, 4891-4900.	0.4	105
71	Mutations in RNF135, a gene within the NF1 microdeletion region, cause phenotypic abnormalities including overgrowth. Nature Genetics, 2007, 39, 963-965.	9.4	103
72	A novel HER2-positive breast cancer phenotype arising from germline TP53 mutations. Journal of Medical Genetics, 2010, 47, 771-774.	1.5	102

#	Article	IF	Citations
73	Biallelic BRCA2 mutations are associated with multiple malignancies in childhood including familial Wilms tumour. Journal of Medical Genetics, 2005, 42, 147-151.	1.5	101
74	Stratification of Wilms tumor by genetic and epigenetic analysis. Oncotarget, 2012, 3, 327-335.	0.8	101
75	Cross-Cancer Genome-Wide Analysis of Lung, Ovary, Breast, Prostate, and Colorectal Cancer Reveals Novel Pleiotropic Associations. Cancer Research, 2016, 76, 5103-5114.	0.4	100
76	Risk of Estrogen Receptorâ€“Positive and â€“Negative Breast Cancer and Singleâ€“Nucleotide Polymorphism 2q35-rs13387042. Journal of the National Cancer Institute, 2009, 101, 1012-1018.	3.0	99
77	Clinical features of NSD1-positive Sotos syndrome. Clinical Dysmorphology, 2004, 13, 199-204.	0.1	94
78	Frequency and Heritability of WT1 Mutations in Nonsyndromic Wilms' Tumor Patients: A UK Childrenâ€™s Cancer Study Group Study. Journal of Clinical Oncology, 2004, 22, 4140-4146.	0.8	93
79	Identification of four novel susceptibility loci for oestrogen receptor negative breast cancer. Nature Communications, 2016, 7, 11375.	5.8	93
80	Mutations of the Uromodulin gene in MCKD type 2 patients cluster in exon 4, which encodes three EGF-like domains. Kidney International, 2003, 64, 1580-1587.	2.6	87
81	Medulloblastoma, acute myelocytic leukemia and colonic carcinomas in a child with biallelic MSH6 mutations. Nature Clinical Practice Oncology, 2007, 4, 130-134.	4.3	86
82	BRCA1 testing should be offered to individuals with triple-negative breast cancer diagnosed below 50 years. British Journal of Cancer, 2012, 106, 1234-1238.	2.9	85
83	Germline mutations in the PAF1 complex gene CTR9 predispose to Wilms tumour. Nature Communications, 2014, 5, 4398.	5.8	85
84	Mutations in the PP2A regulatory subunit B family genesPPP2R5B,PPP2R5CandPPP2R5Dcause human overgrowth. Human Molecular Genetics, 2015, 24, 4775-4779.	1.4	85
85	Identification of new Wilms tumour predisposition genes: an exome sequencing study. The Lancet Child and Adolescent Health, 2019, 3, 322-331.	2.7	82
86	Associations of obesity and circulating insulin and glucose with breast cancer risk: a Mendelian randomization analysis. International Journal of Epidemiology, 2019, 48, 795-806.	0.9	81
87	Accurate clinical detection of exon copy number variants in a targeted NGS panel using DECoN. Wellcome Open Research, 2016, 1, 20.	0.9	79
88	Cancer genes associated with phenotypes in monoallelic and biallelic mutation carriers: new lessons from old players. Human Molecular Genetics, 2007, 16, R60-R66.	1.4	78
89	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 or BRCA1/2 breast cancer (CRUK/07/012). Cancer Research, 2015, 75, S3-01-S3-01.	0.4	78
90	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, 658-669.	1.1	77

#	Article	IF	Citations
91	The NSD1 and EZH2 Overgrowth Genes, Similarities and Differences. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2013, 163, 86-91.	0.7	75
92	The Tatton-Brown-Rahman Syndrome: A clinical study of 55 individuals with de novo constitutive DNMT3A variants. Wellcome Open Research, 2018, 3, 46.	0.9	75
93	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, 827-831.	3.3	73
94	Aneuploidy-Cancer Predisposition Syndromes: A New Link between the Mitotic Spindle Checkpoint and Cancer. Cell Cycle, 2005, 4, 228-230.	1.3	72
95	A genome-wide association study identifies susceptibility loci for Wilms tumor. Nature Genetics, 2012, 44, 681-684.	9.4	72
96	The Gene for Juvenile Hyaline Fibromatosis Maps to Chromosome 4q21. American Journal of Human Genetics, 2002, 71, 975-980.	2.6	71
97	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 Consortiumâ€. Human Molecular Genetics, 2011, 20, 4693-4706.	1.4	71
98	Localization of the Gene for Distal Hereditary Motor Neuronopathy VII (dHMN-VII) to Chromosome 2q14. American Journal of Human Genetics, 2001, 68, 1270-1276.	2.6	68
99	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, 20489-20493.	3.3	67
100	Common variants upstream of MLF1 at 3q25 and within CPZ at 4p16 associated with neuroblastoma. PLoS Genetics, 2017, 13, e1006787.	1.5	62
101	Mainstreaming genetic testing of cancer predisposition genes. Clinical Medicine, 2014, 14, 436-439.	0.8	61
102	Testicular germ cell tumor susceptibility associated with the UCK2 locus on chromosome 1q23. Human Molecular Genetics, 2013, 22, 2748-2753.	1.4	59
103	Identification of Novel Genetic Markers of Breast Cancer Survival. Journal of the National Cancer Institute, 2015, 107, .	3.0	56
104	Heterogeneity of familial medulloblastoma and contribution of germline PTCH1 and SUFU mutations to sporadic medulloblastoma. Familial Cancer, 2011, 10, 337-342.	0.9	55
105	Mammographic Breast Density and Breast Cancer: Evidence of a Shared Genetic Basis. Cancer Research, 2012, 72, 1478-1484.	0.4	54
106	CSN and CAVA: variant annotation tools for rapid, robust next-generation sequencing analysis in the clinical setting. Genome Medicine, 2015, 7, 76.	3.6	54
107	Mutations in the transcriptional repressor REST predispose to Wilms tumor. Nature Genetics, 2015, 47, 1471-1474.	9.4	54
108	A Cost-Effectiveness Evaluation of Germline BRCA1 and BRCA2 Testing in UK Women with Ovarian Cancer. Value in Health, 2017, 20, 567-576.	0.1	54

#	Article	IF	Citations
109	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.	2.6	51
110	Evaluation ofRAD50 in familial breast cancer predisposition. International Journal of Cancer, 2006, 118, 2911-2916.	2.3	51
111	A Multicenter Study of Cancer Incidence in CHEK2 1100delC Mutation Carriers: Table 1.. Cancer Epidemiology Biomarkers and Prevention, 2006, 15, 2542-2545.	1.1	51
112	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, 147-154.	0.7	51
113	Predisposition gene identification in common cancers by exome sequencing: insights from familial breast cancer. Breast Cancer Research and Treatment, 2012, 134, 429-433.	1.1	51
114	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.	1.1	51
115	Characterizing genetic variants for clinical action. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2014, 166, 93-104.	0.7	50
116	MicroRNA Related Polymorphisms and Breast Cancer Risk. PLoS ONE, 2014, 9, e109973.	1.1	49
117	The integration of BRCA testing into oncology clinics. British Journal of Nursing, 2016, 25, 690-694.	0.3	48
118	Evaluation of Fanconi Anemia genes in familial breast cancer predisposition. Cancer Research, 2003, 63, 8596-9.	0.4	48
119	Mechanisms predisposing to childhood overgrowth and cancer. Current Opinion in Genetics and Development, 2005, 15, 227-233.	1.5	46
120	Body mass index and breast cancer survival: a Mendelian randomization analysis. International Journal of Epidemiology, 2017, 46, 1814-1822.	0.9	45
121	PHOX2B analysis in non-syndromic neuroblastoma cases shows novel mutations and genotypeâ€“phenotype associations. American Journal of Medical Genetics, Part A, 2006, 140A, 1297-1301.	0.7	44
122	Geneâ€“gene interactions in breast cancer susceptibility. Human Molecular Genetics, 2012, 21, 958-962.	1.4	41
123	Simple Detection of Germline Microsatellite Instability for Diagnosis of Constitutional Mismatch Repair Cancer Syndrome. Human Mutation, 2013, 34, 847-852.	1.1	40
124	Comparative genomic hybridization and BUB1B mutation analyses in childhood cancers associated with mosaic variegated aneuploidy syndrome. Cancer Letters, 2006, 239, 234-238.	3.2	39
125	Familial T-cell non-Hodgkin lymphoma caused by biallelic MSH2 mutations. Journal of Medical Genetics, 2007, 44, e83-e83.	1.5	39
126	Identification and characterization of novel associations in the CASP8/ALS2CR12 region on chromosome 2 with breast cancer risk. Human Molecular Genetics, 2015, 24, 285-298.	1.4	38

#	Article	IF	Citations
127	Development of cancer genetic services in the UK: A national consultation. Genome Medicine, 2015, 7, 18.	3.6	37
128	Absence of evidence for a familial breast cancer susceptibility gene at chromosome 8p12-p22. Oncogene, 2000, 19, 4170-4173.	2.6	35
129	Evaluation of NSD2 and NSD3 in overgrowth syndromes. European Journal of Human Genetics, 2005, 13, 150-153.	1.4	32
130	Common variants of the BRCA1 wild-type allele modify the risk of breast cancer in BRCA1 mutation carriers. Human Molecular Genetics, 2011, 20, 4732-4747.	1.4	32
131	Estrogen Receptor Status Could Modulate the Genomic Pattern in Familial and Sporadic Breast Cancer. Clinical Cancer Research, 2007, 13, 7305-7313.	3.2	31
132	Multi-stage genome-wide association study identifies new susceptibility locus for testicular germ cell tumour on chromosome 3q25. Human Molecular Genetics, 2015, 24, 1169-1176.	1.4	31
133	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.	2.2	31
134	Genomic variant sharing: a position statement. Wellcome Open Research, 2019, 4, 22.	0.9	31
135	Confirmation of a hereditary motor and sensory neuropathy IIC locus at chromosome 12q23-q24. Annals of Neurology, 2005, 57, 293-297.	2.8	30
136	A cost analysis of a cancer genetic service model in the UK. Journal of Community Genetics, 2016, 7, 185-194.	0.5	29
137	Pathway-based analysis of GWAs data identifies association of sex determination genes with susceptibility to testicular germ cell tumors. Human Molecular Genetics, 2014, 23, 6061-6068.	1.4	28
138	Confirmation of 5p12 As a Susceptibility Locus for Progesterone-Receptorâ€“Positive, Lower Grade Breast Cancer. Cancer Epidemiology Biomarkers and Prevention, 2011, 20, 2222-2231.	1.1	27
139	Clinical features of NSD1-positive Sotos syndrome. Clinical Dysmorphology, 2004, 13, 199-204.	0.1	27
140	Common germline polymorphisms associated with breast cancer-specific survival. Breast Cancer Research, 2015, 17, 58.	2.2	26
141	Chromosomal instability by mutations in the novel minor spliceosome component CENATAC. EMBO Journal, 2021, 40, e106536.	3.5	26
142	Confirmation of a gene locus for medullary cystic kidney disease (MCKD2) on chromosome 16p12. Kidney International, 2001, 60, 1233-1239.	2.6	25
143	A WT1 exon 1 mutation in a child diagnosed with Denys-Drash syndrome. Pediatric Nephrology, 2005, 20, 81-85.	0.9	24
144	Exploring the link between MORF4L1 and risk of breast cancer. Breast Cancer Research, 2011, 13, R40.	2.2	23

#	Article	IF	Citations
145	The ICR1000 UK exome series: a resource of gene variation in an outbred population. F1000Research, 2015, 4, 883.	0.8	22
146	Familial vocal fold paralysis. Journal of Laryngology and Otology, 2002, 116, 1047-1049.	0.4	20
147	Periodontal treatment of two siblings with juvenile hyaline fibromatosis. Journal of Clinical Periodontology, 2005, 32, 1016-1021.	2.3	19
148	The familial Wilms' tumour susceptibility gene, FWT1, may not be a tumour suppressor gene. Oncogene, 1997, 14, 3099-3102.	2.6	17
149	Mosaic-variegated aneuploidy syndrome mutation or haploinsufficiency in Cep57 impairs tumor suppression. Journal of Clinical Investigation, 2018, 128, 3517-3534.	3.9	17
150	Familial gigantism caused by anNSD1 mutation. American Journal of Medical Genetics, Part A, 2005, 139A, 40-44.	0.7	16
151	Mosaic variegated aneuploidy without microcephaly: Implications for cytogenetic diagnosis. American Journal of Medical Genetics, Part A, 2007, 143A, 1890-1893.	0.7	16
152	Association Study of Prostate Cancer Susceptibility Variants with Risks of Invasive Ovarian, Breast, and Colorectal Cancer. Cancer Research, 2008, 68, 8837-8842.	0.4	14
153	The ICR96 exon CNV validation series: a resource for orthogonal assessment of exon CNV calling in NGS data. Wellcome Open Research, 2017, 2, 35.	0.9	14
154	Case of interstitial 12q deletion in association with Wilms tumor. American Journal of Medical Genetics Part A, 2001, 104, 246-249.	2.4	13
155	Mutation and association analysis of GEN1 in breast cancer susceptibility. Breast Cancer Research and Treatment, 2010, 124, 283-288.	1.1	12
156	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.	1.1	11
157	OpEx - a validated, automated pipeline optimised for clinical exome sequence analysis. Scientific Reports, 2016, 6, 31029.	1.6	10
158	Insights into BRCA Cancer Predisposition from Integrated Germline and Somatic Analyses in 7632 Cancers. JNCI Cancer Spectrum, 2019, 3, pkz028.	1.4	10
159	A new familial cancer syndrome including predisposition to Wilms tumor and neuroblastoma. Familial Cancer, 2010, 9, 425-430.	0.9	9
160	Skeletal muscle involvement in infantile systemic hyalinosis. European Journal of Paediatric Neurology, 2003, 7, 401-406.	0.7	8
161	CoverView: a sequence quality evaluation tool for next generation sequencing data. Wellcome Open Research, 2018, 3, 36.	0.9	7
162	Genomic variant sharing: a position statement. Wellcome Open Research, 0, 4, 22.	0.9	7

#	Article	IF	Citations
163	The Quality Sequencing Minimum (QSM): providing comprehensive, consistent, transparent next generation sequencing Âdata quality assurance. Wellcome Open Research, 2018, 3, 37.	0.9	6
164	A new gene on the X involved in Fanconi anemia. Nature Genetics, 2004, 36, 1142-1143.	9.4	5
165	The ICR142 NGS validation series: a resource for orthogonal assessment of NGS analysis. F1000Research, 2016, 5, 386.	0.8	5
166	The ICR142 NGS validation series: a resource for orthogonal assessment of NGS analysis. F1000Research, 2016, 5, 386.	0.8	5
167	The ICR639 CPG NGS validation series: A resource to assess analytical sensitivity of cancer predisposition gene testing. Wellcome Open Research, 2018, 3, 68.	0.9	4
168	Genetic Predisposition to Wilms Tumour. Pediatric Oncology, 2014, , 19-38.	0.5	2
169	Facial dysmorphism and digit anomalies in three siblings with severe developmental delay. Clinical Dysmorphology, 2011, 20, 92-94.	0.1	0
170	ICR142 Benchmarker: evaluating, optimising and benchmarking variant calling using the ICR142 NGS validation series. Wellcome Open Research, 2018, 3, 108.	0.9	0
171	Absence of the TRIP13 c.1060C>T Mutation in Wilms Tumor Patients From Pakistan. Journal of Pediatric Hematology/Oncology, 2020, 42, e128-e131.	0.3	0
172	ICR142 Benchmarker: evaluating, optimising and benchmarking variant calling performance using the ICR142 NGS validation series. Wellcome Open Research, 2018, 3, 108.	0.9	0
173	Clinical Annotation Reference Templates: a resource for consistent variant annotation. Wellcome Open Research, 0, 3, 146.	0.9	0