Conxi Lazaro Garcia

List of Publications by Year
in descending order

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244 papers	14,528 citations	18482 62 h-index	24982 109 g-index
261 all docs	261 docs citations	261 times ranked	20279 citing authors

#	Article	IF	Citations
1	Solving the enigma of POLD1 p.V295M as a potential cause of increased cancer risk. European Journal of Human Genetics, 2022, 30, 485-489.	2.8	2
2	Breast and Prostate Cancer Risks for MaleBRCA1andBRCA2Pathogenic Variant Carriers Using Polygenic Risk Scores. Journal of the National Cancer Institute, 2022, 114, 109-122.	6.3	19
3	Revisiting the UK Genetic Severity Score for NF2: a proposal for the addition of a functional genetic component. Journal of Medical Genetics, 2022, 59, 678-686.	3.2	7
4	DGCR8 and the six hit, three-step model of schwannomatosis. Acta Neuropathologica, 2022, 143, 115-117.	7.7	10
5	CDK5RAP3, a New BRCA2 Partner That Regulates DNA Repair, Is Associated with Breast Cancer Survival. Cancers, 2022, 14, 353.	3.7	0
6	Polygenic risk modeling for prediction of epithelial ovarian cancer risk. European Journal of Human Genetics, 2022, 30, 349-362.	2.8	23
7	Mosaicism in PTENâ€”new case and comment on the literature. European Journal of Human Genetics, 2022, 30, 641-644.	2.8	6
8	Neurofibromatosis type 1 families with first-degree relatives harbouring distinct NF1 pathogenic variants. Genetic counselling and familial diagnosis: what should be offered?. Journal of Medical Genetics, 2022, 59, 1017-1023.	3.2	6
9	Modeling iPSC-derived human neurofibroma-like tumors in mice uncovers the heterogeneity of Schwann cells within plexiform neurofibromas. Cell Reports, 2022, 38, 110385.	6.4	19
10	Modification of BRCA1-associated breast cancer risk by HMMR overexpression. Nature Communications, 2022, 13, 1895.	12.8	19
11	A decade of RAD51C and RAD51D germline variants in cancer. Human Mutation, 2022, 43, 285-298.	2.5	6
12	A High-Throughput Screening Platform Identifies Novel Combination Treatments for Malignant Peripheral Nerve Sheath Tumors. Molecular Cancer Therapeutics, 2022, 21, 1246-1258.	4.1	2
13	Population-based targeted sequencing of 54 candidate genes identifiesPALB2as a susceptibility gene for high-grade serous ovarian cancer. Journal of Medical Genetics, 2021, 58, 305-313.	3.2	26
14	Exome and genome sequencing in adults with undiagnosed disease: a prospective cohort study. Journal of Medical Genetics, 2021, 58, 275-283.	3.2	14
15	TP53, a gene for colorectal cancer predisposition in the absence of Li-Fraumeni-associated phenotypes. Gut, 2021, 70, 1139-1146.	12.1	10
16	A Collaborative Effort to Define Classification Criteria forATMVariants in Hereditary Cancer Patients. Clinical Chemistry, 2021, 67, 518-533.	3.2	14
17	Characteristics of Adrenocortical Carcinoma Associated With Lynch Syndrome. Journal of Clinical Endocrinology and Metabolism, 2021, 106, 318-325.	3.6	20
18	Multigene panel testing for hereditary breast and ovarian cancer in the province of Ontario. Journal of Cancer Research and Clinical Oncology, 2021, 147, 871-879.	2.5	7

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19	BARD1 Pathogenic Variants Are Associated with Triple-Negative Breast Cancer in a Spanish Hereditary Breast and Ovarian Cancer Cohort. Genes, 2021, 12, 150.	2.4	11
20	Using antisense oligonucleotides for the physiological modulation of the alternative splicing of NF1 exon 23a during PC12 neuronal differentiation. Scientific Reports, 2021, 11, 3661.	3.3	4
21	Chromosomal translocations inactivating CDKN2A support a single path for malignant peripheral nerve sheath tumor initiation. Human Genetics, 2021, 140, 1241-1252.	3.8	12
22	Revised diagnostic criteria for neurofibromatosis type 1 and Legius syndrome: an international consensus recommendation. Genetics in Medicine, 2021, 23, 1506-1513.	2.4	290
23	CNVfilteR: an R/Bioconductor package to identify false positives produced by germline NGS CNV detection tools. Bioinformatics, 2021, 37, 4227-4229.	4.1	1
24	Response to letter entitled: Re: ERCC3 a new ovarian cancer susceptibility gene?. European Journal of Cancer, 2021, 150, 281-282.	2.8	0
25	RAD51D Aberrant Splicing in Breast Cancer: Identification of Splicing Regulatory Elements and Minigene-Based Evaluation of 53 DNA Variants. Cancers, 2021, 13, 2845.	3.7	10
26	Genetic Screening for TLR7 Variants in Young and Previously Healthy Men With Severe COVID-19. Frontiers in Immunology, 2021, 12, 719115.	4.8	76
27	Altered regulation of BRCA1 exon 11 splicing is associated with breast cancer risk in carriers of BRCA1 pathogenic variants. Human Mutation, 2021, 42, 1488-1502.	2.5	7
28	Paired Somatic-Germline Testing of 15 Polyposis and Colorectal Cancerâ€“Predisposing Genes Highlights the Role of APC Mosaicism in de Novo Familial Adenomatous Polyposis. Journal of Molecular Diagnostics, 2021, 23, 1452-1459.	2.8	10
29	RNA assay identifies a previous misclassification of BARD1 c.1977A>G variant. Scientific Reports, 2021, 11, 22948.	3.3	0
30	Association of Genomic Domains in BRCA1 and BRCA2 with Prostate Cancer Risk and Aggressiveness. Cancer Research, 2020, 80, 624-638.	0.9	39
31	Mutational spectrum by phenotype: panelâ€based NGS testing of patients with clinical suspicion of RASopathy and children with multiple cafÃ©â€auâ€lait macules. Clinical Genetics, 2020, 97, 264-275.	2.0	13
32	Assessment of ovarian reserve and reproductive outcomes in BRCA1 or BRCA2 mutation carriers. International Journal of Gynecological Cancer, 2020, 30, 83-88.	2.5	12
33	Validation of an inÂVitro Mismatch Repair Assay Used in the Functional Characterization of Mismatch Repair Variants. Journal of Molecular Diagnostics, 2020, 22, 376-385.	2.8	5
34	Retesting of women who are negative for a BRCA1 and BRCA2 mutation using a 20-gene panel. Journal of Medical Genetics, 2020, 57, 380-384.	3.2	10
35	Haplotype analysis of the internationally distributed BRCA1 c.3331_3334delCAAG founder mutation reveals a common ancestral origin in Iberia. Breast Cancer Research, 2020, 22, 108.	5.0	9
36	Polygenic risk scores and breast and epithelial ovarian cancer risks for carriers of BRCA1 and BRCA2 pathogenic variants. Genetics in Medicine, 2020, 22, 1653-1666.	2.4	82

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37	Comprehensive Constitutional Genetic and Epigenetic Characterization of Lynch-Like Individuals. Cancers, 2020, 12, 1799.	3.7	15
38	Screening of CNVs using NGS data improves mutation detection yield and decreases costs in genetic testing for hereditary cancer. Journal of Medical Genetics, 2020, , jmedgenet-2020-107366.	3.2	3
39	Assessing Effectiveness of Colonic and Gynecological Risk Reducing Surgery in Lynch Syndrome Individuals. Cancers, 2020, 12, 3419.	3.7	11
40	Tumor BRCA Testing in High Grade Serous Carcinoma: Mutation Rates and Optimal Tissue Requirements. Cancers, 2020, 12, 3468.	3.7	12
41	Use of patient derived orthotopic xenograft models for real-time therapy guidance in a pediatric sporadic malignant peripheral nerve sheath tumor. Therapeutic Advances in Medical Oncology, 2020, 12, 175883592092957.	3.2	5
42	Immune Cell Associations with Cancer Risk. IScience, 2020, 23, 101296.	4.1	6
43	Role of POLE and POLD1 in familial cancer. Genetics in Medicine, 2020, 22, 2089-2100.	2.4	76
44	ERCC3, a new ovarian cancer susceptibility gene?. European Journal of Cancer, 2020, 141, 1-8.	2.8	8
45	Association of germline variation with the survival of women with BRCA1/2 pathogenic variants and breast cancer. Npj Breast Cancer, 2020, 6, 44.	5.2	5
46	Comprehensive analysis and ACMGâ€based classification of CHEK2 variants in hereditary cancer patients. Human Mutation, 2020, 41, 2128-2142.	2.5	10
47	Editorial: Hereditary Breast and Ovarian Cancer: Current Concepts of Prevention and Treatment. Frontiers in Oncology, 2020, 10, 618369.	2.8	2
48	Genome-wide association study identifies 32 novel breast cancer susceptibility loci from overall and subtype-specific analyses. Nature Genetics, 2020, 52, 572-581.	21.4	265
49	High-sensitivity microsatellite instability assessment for the detection of mismatch repair defects in normal tissue of biallelic germline mismatch repair mutation carriers. Journal of Medical Genetics, 2020, 57, 269-273.	3.2	20
50	Evaluation of CNV detection tools for NGS panel data in genetic diagnostics. European Journal of Human Genetics, 2020, 28, 1645-1655.	2.8	67
51	Tumors defective in homologous recombination rely on oxidative metabolism: relevance to treatments with <scp>PARP</scp> inhibitors. EMBO Molecular Medicine, 2020, 12, e11217.	6.9	37
52	High Prevalence of Somatic Oncogenic Driver Alterations in Patients With NSCLC and Li-Fraumeni Syndrome. Journal of Thoracic Oncology, 2020, 15, 1232-1239.	1.1	29
53	Transcriptomeâ€wide association study of breast cancer risk by estrogenâ€receptor status. Genetic Epidemiology, 2020, 44, 442-468.	1.3	32
54	The Spectrum of FANCM Protein Truncating Variants in European Breast Cancer Cases. Cancers, 2020, 12, 292.	3.7	11

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55	KIF11 and KIF15 mitotic kinesins are potential therapeutic vulnerabilities for malignant peripheral nerve sheath tumors. Neuro-Oncology Advances, 2020, 2, i62-i74.	0.7	12
56	Exploring the Role of Mutations in Fanconi Anemia Genes in Hereditary Cancer Patients. Cancers, 2020, 12, 829.	3.7	41
57	HuR/ELAVL1 drives malignant peripheral nerve sheath tumor growth and metastasis. Journal of Clinical Investigation, 2020, 130, 3848-3864.	8.2	38
58	Genomics of Peripheral Nerve Sheath Tumors Associated with Neurofibromatosis Type 1. , 2020, , 117-147.		1
59	Improving Genetic Testing in Hereditary Cancer by RNA Analysis. Journal of Molecular Diagnostics, 2020, 22, 1453-1468.	2.8	9
60	The FANCM:p.Arg658* truncating variant is associated with risk of triple-negative breast cancer. Npj Breast Cancer, 2019, 5, 38.	5.2	28
61	POT1 and Damage Response Malfunction Trigger Acquisition of Somatic Activating Mutations in the VEGF Pathway in Cardiac Angiosarcomas. Journal of the American Heart Association, 2019, 8, e012875.	3.7	8
62	GFP-Fragment Reassembly Screens for the Functional Characterization of Variants of Uncertain Significance in Protein Interaction Domains of the BRCA1 and BRCA2 Genes. Cancers, 2019, 11, 151.	3.7	4
63	Large scale multifactorial likelihood quantitative analysis of BRCA1 and BRCA2 variants: An ENIGMA resource to support clinical variant classification. Human Mutation, 2019, 40, 1557-1578.	2.5	102
64	Mendelian randomisation study of height and body mass index as modifiers of ovarian cancer risk in 22,588 BRCA1 and BRCA2 mutation carriers. British Journal of Cancer, 2019, 121, 180-192.	6.4	19
65	NTHL1 biallelic mutations seldom cause colorectal cancer, serrated polyposis or a multi-tumor phenotype, in absence of colorectal adenomas. Scientific Reports, 2019, 9, 9020.	3.3	23
66	Genome-wide association and transcriptome studies identify target genes and risk loci for breast cancer. Nature Communications, 2019, 10, 1741.	12.8	90
67	Opportunistic testing of BRCA1, BRCA2 and mismatch repair genes improves the yield of phenotype driven hereditary cancer gene panels. International Journal of Cancer, 2019, 145, 2682-2691.	5.1	30
68	Reprogramming Captures the Genetic and Tumorigenic Properties of Neurofibromatosis Type 1 Plexiform Neurofibromas. Stem Cell Reports, 2019, 12, 411-426.	4.8	28
69	AhR controls redox homeostasis and shapes the tumor microenvironment in BRCA1-associated breast cancer. Proceedings of the National Academy of Sciences of the United States of America, 2019, 116, 3604-3613.	7.1	96
70	Highly sensitive MLH1 methylation analysis in blood identifies a cancer patient with low-level mosaic MLH1 epimutation. Clinical Epigenetics, 2019, 11, 171.	4.1	7
71	Does multilocus inherited neoplasia alleles syndrome have severe clinical expression?. Journal of Medical Genetics, 2019, 56, 521-525.	3.2	11
72	Breast cancer risk in neurofibromatosis type 1 is a function of the type of NF1 gene mutation: a new genotype-phenotype correlation. Journal of Medical Genetics, 2019, 56, 209-219.	3.2	26

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73	Prospective study of germline and somatic alterations for early onset lung cancer patients (EOLUNG) Tj ETQq1 1 0.784314 rgBT /Ove	1.6	1
74	Decapping protein EDC4 regulates DNA repair and phenocopies BRCA1. Nature Communications, 2018, 9, 967.	12.8	33
75	Tumor xenograft modeling identifies TCF4/ITF2 loss associated with breast cancer chemoresistance. DMM Disease Models and Mechanisms, 2018, 11, .	2.4	15
76	Mutational spectrum in a worldwide study of 29,700 families withBRCA1orBRCA2mutations. Human Mutation, 2018, 39, 593-620.	2.5	224
77	Documento de consenso sobre la implementaciÃ³n de la secuenciaciÃ³n masiva de nueva generaciÃ³n en el diagnÃ³stico genÃ©tico de la predisposiciÃ³n hereditaria al cÃ¡ncer. Medicina ClÃnica, 2018, 151, 80.e1-80.e10.	0.6	7
78	Early Genetic Diagnosis of Neurofibromatosis Type 2 From Skin Plaque Plexiform Schwannomas in Childhood. JAMA Dermatology, 2018, 154, 341.	4.1	16
79	Association Between Germline Mutations in BRF1, a Subunit of the RNA Polymerase III Transcription Complex, and Hereditary Colorectal Cancer. Gastroenterology, 2018, 154, 181-194.e20.	1.3	32
80	Genetic Testing and Clinical Management Practices for Variants in Non-BRCA1/2 Breast (and) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50 472 for the Interpretation of Germline Mutant Alleles (ENIGMA) Clinical Working Group. JCO Precision Oncology, 2018, 2, 1-42.	3.0	19
81	Primary constitutional MLH1 epimutations: a focal epigenetic event. British Journal of Cancer, 2018, 119, 978-987.	6.4	22
82	Computational Tools for Splicing Defect Prediction in Breast/Ovarian Cancer Genes: How Efficient Are They at Predicting RNA Alterations?. Frontiers in Genetics, 2018, 9, 366.	2.3	53
83	Analysis of intratumor heterogeneity in Neurofibromatosis type 1 plexiform neurofibromas and neurofibromas with atypical features: Correlating histological and genomic findings. Human Mutation, 2018, 39, 1112-1125.	2.5	34
84	Germline mutations in the spindle assembly checkpoint genes BUB1 and BUB3 are infrequent in familial colorectal cancer and polyposis. Molecular Cancer, 2018, 17, 23.	19.2	19
85	Genetic Testing in Hereditary Colorectal Cancer. , 2018, , 209-232.		0
86	Substantial evidence for the clinical significance of missense variant BRCA1 c.5309G>T p.(Gly1770Val). Breast Cancer Research and Treatment, 2018, 172, 497-503.	2.5	7
87	Germline variation in the oxidative DNA repair genes NUDT1 and OGG1 is not associated with hereditary colorectal cancer or polyposis. Human Mutation, 2018, 39, 1214-1225.	2.5	10
88	Consensus document on the implementation of next generation sequencing in the genetic diagnosis of hereditary cancer. Medicina ClÃnica (English Edition), 2018, 151, 80.e1-80.e10.	0.2	3
89	Evaluation of copy-number variants as modifiers of breast and ovarian cancer risk for BRCA1 pathogenic variant carriers. European Journal of Human Genetics, 2017, 25, 432-438.	2.8	26
90	Elucidating the clinical significance of two PMS2 missense variants coexisting in a family fulfilling hereditary cancer criteria. Familial Cancer, 2017, 16, 501-507.	1.9	3

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91	Elucidating the molecular basis of MSH2â€deficient tumors by combined germline and somatic analysis. International Journal of Cancer, 2017, 141, 1365-1380.	5.1	26
92	Identification of 12 new susceptibility loci for different histotypes of epithelial ovarian cancer. Nature Genetics, 2017, 49, 680-691.	21.4	356
93	Benchmarking of Whole Exome Sequencing and Ad Hoc Designed Panels for Genetic Testing of Hereditary Cancer. Scientific Reports, 2017, 7, 37984.	3.3	35
94	A comprehensive custom panel design for routine hereditary cancer testing: preserving control, improving diagnostics and revealing a complex variation landscape. Scientific Reports, 2017, 7, 39348.	3.3	45
95	Stem cell-like transcriptional reprogramming mediates metastatic resistance to mTOR inhibition. Oncogene, 2017, 36, 2737-2749.	5.9	34
96	Identification of ten variants associated with risk of estrogen-receptor-negative breast cancer. Nature Genetics, 2017, 49, 1767-1778.	21.4	289
97	The wide spectrum of POT1 gene variants correlates with multiple cancer types. European Journal of Human Genetics, 2017, 25, 1278-1281.	2.8	66
98	Mutational Heterogeneity in APC and KRAS Arises at the Crypt Level and Leads to Polyclonality in Early Colorectal Tumorigenesis. Clinical Cancer Research, 2017, 23, 5936-5947.	7.0	25
99	PheoSeq. Journal of Molecular Diagnostics, 2017, 19, 575-588.	2.8	63
100	Association of breast cancer risk in BRCA1 and BRCA2 mutation carriers with genetic variants showing differential allelic expression: identification of a modifier of breast cancer risk at locus 11q22.3. Breast Cancer Research and Treatment, 2017, 161, 117-134.	2.5	18
101	Prediction of Breast and Prostate Cancer Risks in Male BRCA1 and BRCA2 Mutation Carriers Using Polygenic Risk Scores. Journal of Clinical Oncology, 2017, 35, 2240-2250.	1.6	152
102	Identification of a founder <scp>BRCA1</scp> mutation in the Moroccan population. Clinical Genetics, 2016, 90, 361-365.	2.0	13
103	Naturally occurringBRCA2alternative mRNA splicing events in clinically relevant samples. Journal of Medical Genetics, 2016, 53, 548-558.	3.2	69
104	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.	5.0	31
105	Cancer network activity associated with therapeutic response and synergism. Genome Medicine, 2016, 8, 88.	8.2	7
106	Inheritance of deleterious mutations at both BRCA1 and BRCA2 in an international sample of 32,295 women. Breast Cancer Research, 2016, 18, 112.	5.0	42
107	Identification of four novel susceptibility loci for oestrogen receptor negative breast cancer. Nature Communications, 2016, 7, 11375.	12.8	93
108	Scarce evidence of the causal role of germline mutations in UNC5C in hereditary colorectal cancer and polyposis. Scientific Reports, 2016, 6, 20697.	3.3	9

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109	RANKL/RANK control Brca1 mutation-driven mammary tumors. Cell Research, 2016, 26, 761-774.	12.0	128
110	Investigating the effect of 28 BRCA1 and BRCA2 mutations on their related transcribed mRNA. Breast Cancer Research and Treatment, 2016, 155, 253-260.	2.5	6
111	BRCA2 Polymorphic Stop Codon K3326X and the Risk of Breast, Prostate, and Ovarian Cancers. Journal of the National Cancer Institute, 2016, 108, djv315.	6.3	77
112	POLE and POLD1 mutations in 529 kindred with familial colorectal cancer and/or polyposis: review of reported cases and recommendations for genetic testing and surveillance. Genetics in Medicine, 2016, 18, 325-332.	2.4	209
113	Mutations in JMJD1C are involved in Rett syndrome and intellectual disability. Genetics in Medicine, 2016, 18, 378-385.	2.4	40
114	No clinical utility of KRAS variant rs61764370 for ovarian or breast cancer. Gynecologic Oncology, 2016, 141, 386-401.	1.4	18
115	Treatment focused genetic testing (TFGT) for ovarian cancer (OC) patients: The Catalan Institute of Oncology (ICO) network experience.. Journal of Clinical Oncology, 2016, 34, e17071-e17071.	1.6	0
116	High Incidence of Noonan Syndrome Features Including Short Stature and Pulmonic Stenosis in Patients carrying NF1 Missense Mutations Affecting p.Arg1809: Genotypeâ€“Phenotype Correlation. Human Mutation, 2015, 36, 1052-1063.	2.5	143
117	Comprehensive establishment and characterization of orthoxenograft mouse models of malignant peripheral nerve sheath tumors for personalized medicine. EMBO Molecular Medicine, 2015, 7, 608-627.	6.9	36
118	An original phylogenetic approach identified mitochondrial haplogroup T1a1 as inversely associated with breast cancer risk in BRCA2 mutation carriers. Breast Cancer Research, 2015, 17, 61.	5.0	26
119	Assessing Associations between the AURKA-HMMR-TPX2-TUBG1 Functional Module and Breast Cancer Risk in BRCA1/2 Mutation Carriers. PLoS ONE, 2015, 10, e0120020.	2.5	34
120	Germline Mutation in BRCA1 or BRCA2 and Ten-Year Survival for Women Diagnosed with Epithelial Ovarian Cancer. Clinical Cancer Research, 2015, 21, 652-657.	7.0	138
121	Identification of six new susceptibility loci for invasive epithelial ovarian cancer. Nature Genetics, 2015, 47, 164-171.	21.4	221
122	Segmental neurofibromatosis type 2: discriminating two hit from four hit in a patient presenting multiple schwannomas confined to one limb. BMC Medical Genomics, 2015, 8, 2.	1.5	24
123	FANCMc.5791C>T nonsense mutation (rs144567652) induces exon skipping, affects DNA repair activity and is a familial breast cancer risk factor. Human Molecular Genetics, 2015, 24, 5345-5355.	2.9	91
124	Germline Mutations in FAN1 Cause Hereditary Colorectal Cancer by Impairing DNA Repair. Gastroenterology, 2015, 149, 563-566.	1.3	94
125	Association of Type and Location ofBRCA1andBRCA2Mutations With Risk of Breast and Ovarian Cancer. JAMA - Journal of the American Medical Association, 2015, 313, 1347.	7.4	390
126	Detailed characterization of <scp>MLH1</scp> p.<scp>D41H</scp> and p.<scp>N710D</scp> variants coexisting in a Lynch syndrome family with conserved <scp>MLH1</scp> expression tumors. Clinical Genetics, 2015, 87, 543-548.	2.0	6

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127	Exome sequencing identifiesMUTYHmutations in a family with colorectal cancer and an atypical phenotype. Gut, 2015, 64, 355-356.	12.1	14
128	Candidate Genetic Modifiers for Breast and Ovarian Cancer Risk in BRCA1 and BRCA2 Mutation Carriers. Cancer Epidemiology Biomarkers and Prevention, 2015, 24, 308-316.	2.5	22
129	Unenhanced Magnetic Resonance Imaging in the evaluation of High Risk Breast Cancer Individuals.. Journal of Clinical Oncology, 2015, 33, e12579-e12579.	1.6	0
130	Abstract 2739: Transcontinental characterization of the Hispanic BRCA1 3450del4 breast cancer founder mutation. , 2015, , .		0
131	DNA Glycosylases Involved in Base Excision Repair May Be Associated with Cancer Risk in BRCA1 and BRCA2 Mutation Carriers. PLoS Genetics, 2014, 10, e1004256.	3.5	47
132	Associations of common breast cancer susceptibility alleles with risk of breast cancer subtypes in BRCA1 and BRCA2 mutation carriers. Breast Cancer Research, 2014, 16, 3416.	5.0	57
133	ICO Amplicon NGS Data Analysis: A Web Tool for Variant Detection in Common High-Risk Hereditary Cancer Genes Analyzed by Amplicon GS Junior Next-Generation Sequencing. Human Mutation, 2014, 35, 271-277.	2.5	2
134	GALNT12is Not a Major Contributor of Familial Colorectal Cancer Type X. Human Mutation, 2014, 35, 50-52.	2.5	22
135	Tubers from patients with tuberous sclerosis complex are characterized by changes in microtubule biology through <scp>ROCK2</scp> signalling. Journal of Pathology, 2014, 233, 247-257.	4.5	7
136	New insights into POLE and POLD1 germline mutations in familial colorectal cancer and polyposis. Human Molecular Genetics, 2014, 23, 3506-3512.	2.9	135
137	Prevalence of germline MUTYH mutations among Lynch-like syndrome patients. European Journal of Cancer, 2014, 50, 2241-2250.	2.8	66
138	Comparison of mRNA Splicing Assay Protocols across Multiple Laboratories: Recommendations for Best Practice in Standardized Clinical Testing. Clinical Chemistry, 2014, 60, 341-352.	3.2	95
139	Comprehensive molecular characterisation of hereditary non-polyposis colorectal tumours with mismatch repair proficiency. European Journal of Cancer, 2014, 50, 1964-1972.	2.8	8
140	Identification of a founder EPCAM deletion in Spanish Lynch syndrome families. Clinical Genetics, 2014, 85, 260-266.	2.0	12
141	SVA retrotransposon insertion-associated deletion represents a novel mutational mechanism underlying large genomic copy number changes with non-recurrent breakpoints. Genome Biology, 2014, 15, R80.	9.6	63
142	Longer Telomeres Are Associated with Cancer Risk in MMR-Proficient Hereditary Non-Polyposis Colorectal Cancer. PLoS ONE, 2014, 9, e86063.	2.5	13
143	Genetic variant in the telomerase gene modifies cancer risk in Lynch syndrome. European Journal of Human Genetics, 2013, 21, 511-516.	2.8	20
144	Refining the role ofpms2in Lynch syndrome: germline mutational analysis improved by comprehensive assessment of variants. Journal of Medical Genetics, 2013, 50, 552-563.	3.2	47

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145	Next-generation sequencing meets genetic diagnostics: development of a comprehensive workflow for the analysis of BRCA1 and BRCA2 genes. European Journal of Human Genetics, 2013, 21, 864-870.	2.8	94
146	Probe-Based Quantitative PCR Assay for Detecting Constitutional and Somatic Deletions in the NF1 Gene: Application to Genetic Testing and Tumor Analysis. Clinical Chemistry, 2013, 59, 928-937.	3.2	12
147	In vitro antisense therapeutics for a deep intronic mutation causing Neurofibromatosis type 2. European Journal of Human Genetics, 2013, 21, 769-773.	2.8	20
148	Identification of a BRCA2-Specific Modifier Locus at 6p24 Related to Breast Cancer Risk. PLoS Genetics, 2013, 9, e1003173.	3.5	105
149	Genome-Wide Association Study in BRCA1 Mutation Carriers Identifies Novel Loci Associated with Breast and Ovarian Cancer Risk. PLoS Genetics, 2013, 9, e1003212.	3.5	244
150	Evaluation of Rare Variants in the New Fanconi Anemia GeneERCC4(FANCQ) as Familial Breast/Ovarian Cancer Susceptibility Alleles. Human Mutation, 2013, 34, 1615-1618.	2.5	28
151	Whole Exome Sequencing Suggests Much of Non-BRCA1/BRCA2 Familial Breast Cancer Is Due to Moderate and Low Penetrance Susceptibility Alleles. PLoS ONE, 2013, 8, e55681.	2.5	95
152	Telomere Length and Genetic Anticipation in Lynch Syndrome. PLoS ONE, 2013, 8, e61286.	2.5	21
153	Functional and Structural Analysis of C-Terminal BRCA1 Missense Variants. PLoS ONE, 2013, 8, e61302.	2.5	16
154	Genomic imbalance ofHMMR/RHAMMregulates the sensitivity and response of malignant peripheral nerve sheath tumour cells to aurora kinase inhibition. Oncotarget, 2013, 4, 80-93.	1.8	27
155	Abstract LB-214: Common genomic alterations in malignant peripheral nerve sheath tumors augment Aurora A activity and sensitize tumors to aurora kinase inhibitors.. , 2013, , .		0
156	A Nonsynonymous Polymorphism in IRS1 Modifies Risk of Developing Breast and Ovarian Cancers in BRCA1 and Ovarian Cancer in BRCA2 Mutation Carriers. Cancer Epidemiology Biomarkers and Prevention, 2012, 21, 1362-1370.	2.5	23
157	Common Variants at the 19p13.1 and ZNF365 Loci Are Associated with ER Subtypes of Breast Cancer and Ovarian Cancer Risk in BRCA1 and BRCA2 Mutation Carriers. Cancer Epidemiology Biomarkers and Prevention, 2012, 21, 645-657.	2.5	47
158	MLH1 promoter hypermethylation in the analytical algorithm of Lynch syndrome: a cost-effectiveness study. European Journal of Human Genetics, 2012, 20, 762-768.	2.8	76
159	Pathology of Breast and Ovarian Cancers among BRCA1 and BRCA2 Mutation Carriers: Results from the Consortium of Investigators of Modifiers of BRCA1/2 (CIMBA). Cancer Epidemiology Biomarkers and Prevention, 2012, 21, 134-147.	2.5	513
160	Association Between <emph type="ital">BRCA1</emph> and <emph type="ital">BRCA2</emph> Mutations and Survival in Women With Invasive Epithelial Ovarian Cancer. JAMA - Journal of the American Medical Association, 2012, 307, 382.	7.4	546
161	MLH1 methylation screening is effective in identifying epimutation carriers. European Journal of Human Genetics, 2012, 20, 1256-1264.	2.8	36
162	Applying Microsatellite Multiplex PCR Analysis (MMPA) for Determining Allele Copy-Number Status and Percentage of Normal Cells within Tumors. PLoS ONE, 2012, 7, e42682.	2.5	3

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163	Comprehensive functional assessment ofMLH1variants of unknown significance. Human Mutation, 2012, 33, 1576-1588.	2.5	30
164	Assessing the RNA effect of 26 DNA variants in the BRCA1 and BRCA2 genes. Breast Cancer Research and Treatment, 2012, 132, 979-992.	2.5	20
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