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	Network modeling links breast cancer susceptibility and centrosome dysfunction. Nature Genetics, 2007, 39, 1338-1349.	21.4	602
2	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
3	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
4	Deletion of the late cornified envelope LCE3B and LCE3C genes as a susceptibility factor for psoriasis. Nature Genetics, 2009, 41, 211-215.	21.4	482
5	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
6	Identification of 12 new susceptibility loci for different histotypes of epithelial ovarian cancer. Nature Genetics, 2017, 49, 680-691.	21.4	356
7	An Absence of Cutaneous Neurofibromas Associated with a 3-bp Inframe Deletion in Exon 17 of the NF1 Gene (c.2970-2972 delAAT): Evidence of a Clinically Significant NF1 Genotype-Phenotype Correlation. American Journal of Human Genetics, 2007, 80, 140-151.	6.2	335
8	Mutations affecting mRNA splicing are the most common molecular defects in patients with neurofibromatosis type 1. Human Molecular Genetics, 2000, 9, 237-247.	2.9	319
9	A locus on 19p13 modifies risk of breast cancer in BRCA1 mutation carriers and is associated with hormone receptorâ€“negative breast cancer in the general population. Nature Genetics, 2010, 42, 885-892.	21.4	309
10	Revised diagnostic criteria for neurofibromatosis type 1 and Legius syndrome: an international consensus recommendation. Genetics in Medicine, 2021, 23, 1506-1513.	2.4	290
11	Identification of ten variants associated with risk of estrogen-receptor-negative breast cancer. Nature Genetics, 2017, 49, 1767-1778.	21.4	289
12	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
13	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
14	Schwann cells harbor the somatic NF1 mutation in neurofibromas: evidence of two different Schwann cell subpopulations. Human Molecular Genetics, 2000, 9, 3055-3064.	2.9	232
15	Mutational spectrum in a worldwide study of 29,700 families withBRCA1orBRCA2mutations. Human Mutation, 2018, 39, 593-620.	2.5	224
16	Identification of six new susceptibility loci for invasive epithelial ovarian cancer. Nature Genetics, 2015, 47, 164-171.	21.4	221
17	Confirmation of a Double-Hit Model for the NF1Gene in Benign Neurofibromas. American Journal of Human Genetics, 1997, 61, 512-519.	6.2	217
18	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

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19	Common Breast Cancer Susceptibility Alleles and the Risk of Breast Cancer for BRCA1 and BRCA2 Mutation Carriers: Implications for Risk Prediction. Cancer Research, 2010, 70, 9742-9754.	0.9	169
20	Recombination hotspot in NF1 microdeletion patients. Human Molecular Genetics, 2001, 10, 1387-1392.	2.9	159
21	Recurrent mutations in the NF1 gene are common among neurofibromatosis type 1 patients. Journal of Medical Genetics, 2003, 40, 82e-82.	3.2	154
22	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
23	Neurofibromatosis Type 1 Due to Germ-Line Mosaicism in a Clinically Normal Father. New England Journal of Medicine, 1994, 331, 1403-1407.	27.0	146
24	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
25	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
26	New insights into POLE and POLD1 germline mutations in familial colorectal cancer and polyposis. Human Molecular Genetics, 2014, 23, 3506-3512.	2.9	135
27	Predominant occurrence of somatic mutations of theNF2 gene in meningiomas and schwannomas. Genes Chromosomes and Cancer, 1995, 13, 211-216.	2.8	132
28	Coding haplotype analysis supports HCR as the putative susceptibility gene for psoriasis at the MHC PSORS1 locus. Human Molecular Genetics, 2002, 11, 589-597.	2.9	131
29	RANKL/RANK control Brca1 mutation-driven mammary tumors. Cell Research, 2016, 26, 761-774.	12.0	128
30	Integrative genomic analyses of neurofibromatosis tumours identify SOX9 as a biomarker and survival gene. EMBO Molecular Medicine, 2009, 1, 236-248.	6.9	112
31	Unequal Meiotic Crossover: A Frequent Cause of NF1 Microdeletions. American Journal of Human Genetics, 2000, 66, 1969-1974.	6.2	107
32	Mosaic type-1 NF1 microdeletions as a cause of both generalized and segmental neurofibromatosis type-1 (NF1). Human Mutation, 2011, 32, 213-219.	2.5	106
33	Identification of a BRCA2-Specific Modifier Locus at 6p24 Related to Breast Cancer Risk. PLoS Genetics, 2013, 9, e1003173.	3.5	105
34	Nature and mRNA effect of 282 differentNF1point mutations: focus on splicing alterations. Human Mutation, 2008, 29, E173-E193.	2.5	102
35	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
36	The location of constitutional neurofibromatosis 2 (NF2) splice site mutations is associated with the severity of NF2. Journal of Medical Genetics, 2005, 42, 540-546.	3.2	98

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37	Sex differences in mutational rate and mutational mechanism in the NF1 gene in neurofibromatosis type 1 patients. Human Genetics, 1996, 98, 696-699.	3.8	97
38	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
39	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
40	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
41	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
42	Germline Mutations in FAN1 Cause Hereditary Colorectal Cancer by Impairing DNA Repair. Gastroenterology, 2015, 149, 563-566.	1.3	94
43	Identification of four novel susceptibility loci for oestrogen receptor negative breast cancer. Nature Communications, 2016, 7, 11375.	12.8	93
44	Interplay between BRCA1 and RHAMM Regulates Epithelial Apicobasal Polarization and May Influence Risk of Breast Cancer. PLoS Biology, 2011, 9, e1001199.	5.6	91
45	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
46	The Average Cumulative Risks of Breast and Ovarian Cancer for Carriers of Mutations in BRCA1 and BRCA2 Attending Genetic Counseling Units in Spain. Clinical Cancer Research, 2008, 14, 2861-2869.	7.0	90
47	Genome-wide association and transcriptome studies identify target genes and risk loci for breast cancer. Nature Communications, 2019, 10, 1741.	12.8	90
48	A highly informative CA/GT repeat polymorphism in intron 38 of the human neurofibromatosis type 1 (NF1) gene. Human Genetics, 1993, 92, 429-430.	3.8	85
49	Common Genetic Variants and Modification of Penetrance of BRCA2-Associated Breast Cancer. PLoS Genetics, 2010, 6, e1001183.	3.5	85
50	Analysis of FANCB and FANCN/PALB2 Fanconi Anemia genes in BRCA1/2-negative Spanish breast cancer families. Breast Cancer Research and Treatment, 2009, 113, 545-551.	2.5	83
51	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
52	Two CA/GT repeat polymorphisms in intron 27 of the human neurofibromatosis type 1 (NF1) gene. Human Genetics, 1994, 93, 351-352.	3.8	81
53	Common variants at 12p11, 12q24, 9p21, 9q31.2 and in ZNF365 are associated with breast cancer risk for BRCA1 and/or BRCA2mutation carriers. Breast Cancer Research, 2012, 14, R33.	5.0	78
54	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

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55	Conservation of hotspots for recombination in low-copy repeats associated with the NF1 microdeletion. Nature Genetics, 2006, 38, 1419-1423.	21.4	76
56	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
57	Role of POLE and POLD1 in familial cancer. Genetics in Medicine, 2020, 22, 2089-2100.	2.4	76
58	Genetic Screening for TLR7 Variants in Young and Previously Healthy Men With Severe COVID-19. Frontiers in Immunology, 2021, 12, 719115.	4.8	76
59	Mitotic recombination effects homozygosity for NF1 germline mutations in neurofibromas. Nature Genetics, 2001, 28, 294-296.	21.4	72
60	Common breast cancer susceptibility alleles are associated with tumour subtypes in BRCA1 and BRCA2 mutation carriers: results from the Consortium of Investigators of Modifiers of BRCA1/2. Breast Cancer Research, 2011, 13, R110.	5.0	71
61	Naturally occurringBRCA2alternative mRNA splicing events in clinically relevant samples. Journal of Medical Genetics, 2016, 53, 548-558.	3.2	69
62	Common alleles at 6q25.1 and 1p11.2 are associated with breast cancer risk for BRCA1 and BRCA2 mutation carriers. Human Molecular Genetics, 2011, 20, 3304-3321.	2.9	68
63	Evaluation of CNV detection tools for NGS panel data in genetic diagnostics. European Journal of Human Genetics, 2020, 28, 1645-1655.	2.8	67
64	Dissecting loss of heterozygosity (LOH) in neurofibromatosis type 1-associated neurofibromas: Importance of copy neutral LOH. Human Mutation, 2011, 32, 78-90.	2.5	66
65	Prevalence of germline MUTYH mutations among Lynch-like syndrome patients. European Journal of Cancer, 2014, 50, 2241-2250.	2.8	66
66	The wide spectrum of POT1 gene variants correlates with multiple cancer types. European Journal of Human Genetics, 2017, 25, 1278-1281.	2.8	66
67	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
68	PheoSeq. Journal of Molecular Diagnostics, 2017, 19, 575-588.	2.8	63
69	Molecular studies in 20 submicroscopic neurofibromatosis type 1 gene deletions. Human Mutation, 1999, 14, 387-393.	2.5	62
70	A Clinical Variant of Neurofibromatosis Type 1: Familial Spinal Neurofibromatosis with a Frameshift Mutation in the NF1 Gene. American Journal of Human Genetics, 1998, 62, 834-841.	6.2	61
71	Type 2 NF1 Deletions Are Highly Unusual by Virtue of the Absence of Nonallelic Homologous Recombination Hotspots and an Apparent Preference for Female Mitotic Recombination. American Journal of Human Genetics, 2007, 81, 1201-1220.	6.2	60
72	Biological reprogramming in acquired resistance to endocrine therapy of breast cancer. Oncogene, 2010, 29, 6071-6083.	5.9	59

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73	Therapeutic potential and mechanism of kinetin as a treatment for the human splicing disease familial dysautonomia. Journal of Molecular Medicine, 2007, 85, 149-161.	3.9	58
74	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
75	Parity and the risk of breast and ovarian cancer in BRCA1 and BRCA2 mutation carriers. Breast Cancer Research and Treatment, 2010, 119, 221-232.	2.5	56
76	Characterization of BRCA1 and BRCA2 splicing variants: a collaborative report by ENIGMA consortium members. Breast Cancer Research and Treatment, 2012, 132, 1009-1023.	2.5	56
77	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
78	Missense mutations in the cystic fibrosis gene in adult patients with asthma. Human Mutation, 1999, 14, 510-519.	2.5	51
79	Cold shock induces the insertion of a cryptic exon in the neurofibromatosis type 1 (NF1) mRNA. Nucleic Acids Research, 2000, 28, 1307-1312.	14.5	48
80	Novel alleles, hemizygosity and deletions at an Alu-repeat within the neurofibromatosis type 1 (NF1) gene. Human Molecular Genetics, 1993, 2, 725-730.	2.9	47
81	The Role of KRAS rs61764370 in Invasive Epithelial Ovarian Cancer: Implications for Clinical Testing. Clinical Cancer Research, 2011, 17, 3742-3750.	7.0	47
82	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
83	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
84	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
85	Antisense therapeutics for neurofibromatosis type 1 caused by deep intronic mutations. Human Mutation, 2009, 30, 454-462.	2.5	46
86	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
87	New alleles at microsatellite loci in CEPH families mainly arise from somatic mutations in the lymphoblastoid cell lines. Human Mutation, 1994, 3, 365-372.	2.5	44
88	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
89	Exploring the Role of Mutations in Fanconi Anemia Genes in Hereditary Cancer Patients. Cancers, 2020, 12, 829.	3.7	41
90	Association Study of the Chromosomal Region Containing the FCER2 Gene Suggests It Has a Regulatory Role in Atopic Disorders. American Journal of Respiratory and Critical Care Medicine, 2000, 161, 700-706.	5.6	40

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91	Genetic Variation at 9p22.2 and Ovarian Cancer Risk for BRCA1 and BRCA2 Mutation Carriers. Journal of the National Cancer Institute, 2011, 103, 105-116.	6.3	40
92	Mutations in JMJD1C are involved in Rett syndrome and intellectual disability. Genetics in Medicine, 2016, 18, 378-385.	2.4	40
93	A new approach for identifying non-pathogenic mutations. An analysis of the cystic fibrosis transmembrane regulator gene in normal individuals. Human Genetics, 2000, 106, 172-178.	3.8	39
94	Association of Genomic Domains in BRCA1 and BRCA2 with Prostate Cancer Risk and Aggressiveness. Cancer Research, 2020, 80, 624-638.	0.9	39
95	HuR/ELAVL1 drives malignant peripheral nerve sheath tumor growth and metastasis. Journal of Clinical Investigation, 2020, 130, 3848-3864.	8.2	38
96	Mosaicism for the fragile X syndrome full mutation and deletions within the CGG repeat of the FMR1 gene.. Journal of Medical Genetics, 1996, 33, 338-340.	3.2	37
97	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
98	Gene Expression Analysis Identifies Potential Biomarkers of Neurofibromatosis Type 1 Including Adrenomedullin. Clinical Cancer Research, 2010, 16, 5048-5057.	7.0	36
99	MLH1 methylation screening is effective in identifying epimutation carriers. European Journal of Human Genetics, 2012, 20, 1256-1264.	2.8	36
100	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
101	Benchmarking of Whole Exome Sequencing and Ad Hoc Designed Panels for Genetic Testing of Hereditary Cancer. Scientific Reports, 2017, 7, 37984.	3.3	35
102	HLA Class II Genes in Soybean Epidemic Asthma Patients. American Journal of Respiratory and Critical Care Medicine, 1997, 156, 1394-1398.	5.6	34
103	Identification and comprehensive characterization of large genomic rearrangements in the BRCA1 and BRCA2 genes. Breast Cancer Research and Treatment, 2010, 122, 733-743.	2.5	34
104	Allele-Specific Expression of APC in Adenomatous Polyposis Families. Gastroenterology, 2010, 139, 439-447.e1.	1.3	34
105	Ovarian cancer susceptibility alleles and risk of ovarian cancer inBRCA1andBRCA2mutation carriers. Human Mutation, 2012, 33, 690-702.	2.5	34
106	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
107	Stem cell-like transcriptional reprogramming mediates metastatic resistance to mTOR inhibition. Oncogene, 2017, 36, 2737-2749.	5.9	34
108	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

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109	Exploring the Link between Germline and Somatic Genetic Alterations in Breast Carcinogenesis. PLoS ONE, 2010, 5, e14078.	2.5	33
110	Decapping protein EDC4 regulates DNA repair and phenocopies BRCA1. Nature Communications, 2018, 9, 967.	12.8	33
111	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
112	Transcriptomeâ€wide association study of breast cancer risk by estrogenâ€receptor status. Genetic Epidemiology, 2020, 44, 442-468.	1.3	32
113	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
114	Genome-wide Linkage Scan Reveals Three Putative Breast-Cancer-Susceptibility Loci. American Journal of Human Genetics, 2009, 84, 115-122.	6.2	30
115	Comprehensive functional assessment ofMLH1variants of unknown significance. Human Mutation, 2012, 33, 1576-1588.	2.5	30
116	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
117	Detection of genetic alterations in hereditary colorectal cancer screeningâ˜†. Mutation Research - Fundamental and Molecular Mechanisms of Mutagenesis, 2010, 693, 19-31.	1.0	29
118	MLH1 Founder Mutations with Moderate Penetrance in Spanish Lynch Syndrome Families. Cancer Research, 2010, 70, 7379-7391.	0.9	29
119	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
120	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
121	The FANCM:p.Arg658* truncating variant is associated with risk of triple-negative breast cancer. Npj Breast Cancer, 2019, 5, 38.	5.2	28
122	Reprogramming Captures the Genetic and Tumorigenic Properties of Neurofibromatosis Type 1 Plexiform Neurofibromas. Stem Cell Reports, 2019, 12, 411-426.	4.8	28
123	International distribution and age estimation of the Portuguese BRCA2 c.156_157insAlu founder mutation. Breast Cancer Research and Treatment, 2011, 127, 671-679.	2.5	27
124	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
125	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
126	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

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127	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
128	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
129	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
130	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
131	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
132	NF1mutation rather than individual genetic variability is the main determinant of theNF1-transcriptional profile of mutations affecting splicing. Human Mutation, 2006, 27, 1104-1114.	2.5	23
133	Exploring the link between MORF4L1 and risk of breast cancer. Breast Cancer Research, 2011, 13, R40.	5.0	23
134	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
135	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
136	Polygenic risk modeling for prediction of epithelial ovarian cancer risk. European Journal of Human Genetics, 2022, 30, 349-362.	2.8	23
137	GALNT12is Not a Major Contributor of Familial Colorectal Cancer Type X. Human Mutation, 2014, 35, 50-52.	2.5	22
138	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
139	Primary constitutional MLH1 epimutations: a focal epigenetic event. British Journal of Cancer, 2018, 119, 978-987.	6.4	22
140	Telomere Length and Genetic Anticipation in Lynch Syndrome. PLoS ONE, 2013, 8, e61286.	2.5	21
141	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
142	Genetic variant in the telomerase gene modifies cancer risk in Lynch syndrome. European Journal of Human Genetics, 2013, 21, 511-516.	2.8	20
143	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
144	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

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145	Characteristics of Adrenocortical Carcinoma Associated With Lynch Syndrome. Journal of Clinical Endocrinology and Metabolism, 2021, 106, 318-325.	3.6	20
146	Genetic Testing and Clinical Management Practices for Variants in Non-BRCA1/2 Breast (and) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50 712 for the Interpretation of Germline Mutant Alleles (ENIGMA) Clinical Working Group. JCO Precision Oncology, 2018, 2, 1-42.	3.0	19
147	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
148	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
149	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
150	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
151	Modification of BRCA1-associated breast cancer risk by HMMR overexpression. Nature Communications, 2022, 13, 1895.	12.8	19
152	Haplotype structure in Ashkenazi Jewish BRCA1 and BRCA2 mutation carriers. Human Genetics, 2011, 130, 685-699.	3.8	18
153	A mild neurofibromatosis type 1 phenotype produced by the combination of the benign nature of a leaky NF1-splice mutation and the presence of a complex mosaicism. Human Mutation, 2011, 32, 705-709.	2.5	18
154	No clinical utility of KRAS variant rs61764370 for ovarian or breast cancer. Gynecologic Oncology, 2016, 141, 386-401.	1.4	18
155	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
156	Prenatal diagnosis of sporadic neurofibromatosis type 1 (NF1) by RNA and DNA analysis of a splicing mutation. , 1999, 19, 739-742.		17
157	Modulation of aberrant NF1 pre-mRNA splicing by kinetin treatment. European Journal of Human Genetics, 2010, 18, 614-617.	2.8	16
158	Functional and Structural Analysis of C-Terminal BRCA1 Missense Variants. PLoS ONE, 2013, 8, e61302.	2.5	16
159	Early Genetic Diagnosis of Neurofibromatosis Type 2 From Skin Plaque Plexiform Schwannomas in Childhood. JAMA Dermatology, 2018, 154, 341.	4.1	16
160	Tumor xenograft modeling identifies TCF4/ITF2 loss associated with breast cancer chemoresistance. DMM Disease Models and Mechanisms, 2018, 11, .	2.4	15
161	Comprehensive Constitutional Genetic and Epigenetic Characterization of Lynch-Like Individuals. Cancers, 2020, 12, 1799.	3.7	15
162	Analysis of SLX4/FANCP in non-BRCA1/2-mutated breast cancer families. BMC Cancer, 2012, 12, 84.	2.6	14

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163	Exome sequencing identifiesMUTYHmutations in a family with colorectal cancer and an atypical phenotype. Gut, 2015, 64, 355-356.	12.1	14
164	Exome and genome sequencing in adults with undiagnosed disease: a prospective cohort study. Journal of Medical Genetics, 2021, 58, 275-283.	3.2	14
165	A Collaborative Effort to Define Classification Criteria forATMVariants in Hereditary Cancer Patients. Clinical Chemistry, 2021, 67, 518-533.	3.2	14
166	Mutation analysis of genetic diseases by asymmetric-PCR SSCP and ethidium bromide staining: application to neurofibromatosis and cystic fibrosis. Molecular and Cellular Probes, 1992, 6, 357-359.	2.1	13
167	Identification of a founder <scp>BRCA1</scp> mutation in the Moroccan population. Clinical Genetics, 2016, 90, 361-365.	2.0	13
168	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
169	Longer Telomeres Are Associated with Cancer Risk in MMR-Proficient Hereditary Non-Polyposis Colorectal Cancer. PLoS ONE, 2014, 9, e86063.	2.5	13
170	Evidence for a link between TNFRSF11A and risk of breast cancer. Breast Cancer Research and Treatment, 2011, 129, 947-954.	2.5	12
171	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
172	Identification of a founder EPCAM deletion in Spanish Lynch syndrome families. Clinical Genetics, 2014, 85, 260-266.	2.0	12
173	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
174	Tumor BRCA Testing in High Grade Serous Carcinoma: Mutation Rates and Optimal Tissue Requirements. Cancers, 2020, 12, 3468.	3.7	12
175	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
176	Chromosomal translocations inactivating CDKN2A support a single path for malignant peripheral nerve sheath tumor initiation. Human Genetics, 2021, 140, 1241-1252.	3.8	12
177	Mutation and linkage disequilibrium analysis in genetic counselling of Spanish cystic fibrosis families.. Journal of Medical Genetics, 1991, 28, 771-776.	3.2	11
178	Does multilocus inherited neoplasia alleles syndrome have severe clinical expression?. Journal of Medical Genetics, 2019, 56, 521-525.	3.2	11
179	Assessing Effectiveness of Colonic and Gynecological Risk Reducing Surgery in Lynch Syndrome Individuals. Cancers, 2020, 12, 3419.	3.7	11
180	The Spectrum of FANCM Protein Truncating Variants in European Breast Cancer Cases. Cancers, 2020, 12, 292.	3.7	11

#	Article	IF	Citations
181	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
182	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
183	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
184	Comprehensive analysis and ACMGâ€based classification of CHEK2 variants in hereditary cancer patients. Human Mutation, 2020, 41, 2128-2142.	2.5	10
185	TP53, a gene for colorectal cancer predisposition in the absence of Li-Fraumeni-associated phenotypes. Gut, 2021, 70, 1139-1146.	12.1	10
186	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
187	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
188	DGCR8 and the six hit, three-step model of schwannomatosis. Acta Neuropathologica, 2022, 143, 115-117.	7.7	10
189	Prenatal diagnosis of sporadic neurofibromatosis 1. Lancet, The, 1992, 339, 119-120.	13.7	9
190	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
191	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
192	Improving Genetic Testing in Hereditary Cancer by RNA Analysis. Journal of Molecular Diagnostics, 2020, 22, 1453-1468.	2.8	9
193	Prenatal diagnosis of neurofibromatosis type 1: From flanking rflps to intragenic microsatellite markers. Prenatal Diagnosis, 1995, 15, 129-134.	2.3	8
194	Association study of proposed candidate genes/regions in a population of Spanish asthmatics. European Journal of Epidemiology, 2000, 16, 745-750.	5.7	8
195	Comprehensive molecular characterisation of hereditary non-polyposis colorectal tumours with mismatch repair proficiency. European Journal of Cancer, 2014, 50, 1964-1972.	2.8	8
196	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
197	ERCC3, a new ovarian cancer susceptibility gene?. European Journal of Cancer, 2020, 141, 1-8.	2.8	8
198	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

#	Article	IF	Citations
199	Cancer network activity associated with therapeutic response and synergism. Genome Medicine, 2016, 8, 88.	8.2	7
200	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
201	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
202	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
203	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
204	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
205	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
206	Germline ATM mutational analysis in BRCA1/BRCA2 negative hereditary breast cancer families by MALDI-TOF mass spectrometry. Breast Cancer Research and Treatment, 2011, 128, 573-579.	2.5	6
207	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
208	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
209	Immune Cell Associations with Cancer Risk. IScience, 2020, 23, 101296.	4.1	6
210	Mosaicism in PTENâ€"new case and comment on the literature. European Journal of Human Genetics, 2022, 30, 641-644.	2.8	6
211	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
212	A decade of RAD51C and RAD51D germline variants in cancer. Human Mutation, 2022, 43, 285-298.	2.5	6
213	Conservation of a polymorphic microsatellite at orthologous positions in the human and mouse CD5 gene promoter. Immunogenetics, 1997, 45, 233-234.	2.4	5
214	Tumor LOH analysis provides reliable linkage information for prenatal genetic testing of sporadic NF1 patients. Genes Chromosomes and Cancer, 2007, 46, 820-827.	2.8	5
215	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
216	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

#	Article	IF	Citations
217	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
218	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
219	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
220	Identification of a new complex rearrangement affecting exon 20 of BRCA1. Breast Cancer Research and Treatment, 2011, 130, 341-344.	2.5	3
221	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
222	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
223	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
224	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
225	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
226	Editorial: Hereditary Breast and Ovarian Cancer: Current Concepts of Prevention and Treatment. Frontiers in Oncology, 2020, 10, 618369.	2.8	2
227	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
228	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
229	Mosaicismo clÃnico y genÃ©tico en la neurofibromatosis tipo 1. Piel, 2006, 21, 477-483.	0.0	1
230	Comments on: Sluiter MD and van Rensburg EJ, Large genomic rearrangements of the BRCA1 and BRCA2 genes: review of the literature and report of a novel BRCA1 mutation. Breast Cancer Research and Treatment, 2010, 124, 295-296.	2.5	1
231	CNVfilteR: an R/Bioconductor package to identify false positives produced by germline NGS CNV detection tools. Bioinformatics, 2021, 37, 4227-4229.	4.1	1
232	Deep Intronic NF1 Mutations and Possible Therapeutic Interventions. , 2012, , 173-186.		1
233	Prospective study of germline and somatic alterations for early onset lung cancer patients (EOLUNG) Tj ETQq1 1 0.784314 rgBT /Ove	1.6	1
234	Genomics of Peripheral Nerve Sheath Tumors Associated with Neurofibromatosis Type 1. , 2020, , 117-147.		1

#	Article	IF	Citations
235	Incidence of CDKN2A mutations in melanoma families: Inherited susceptibility to several cancers but not always association with dysplastic nevus syndrome. Journal of Dermatological Science, 1998, 16, S145.	1.9	0
236	Genetic Testing in Hereditary Colorectal Cancer. , 2018, , 209-232.		0
237	Response to letter entitled: Re: ERCC3 a new ovarian cancer susceptibility gene?. European Journal of Cancer, 2021, 150, 281-282.	2.8	0
238	Abstract 4445: Defining a pipeline to use next generation sequencing for genetic testing in hereditary cancer. , 2012, , .		0
239	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
240	Unenhanced Magnetic Resonance Imaging in the evaluation of High Risk Breast Cancer Individuals.. Journal of Clinical Oncology, 2015, 33, e12579-e12579.	1.6	0
241	Abstract 2739: Transcontinental characterization of the Hispanic BRCA1 3450del4 breast cancer founder mutation. , 2015, , .		0
242	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
243	RNA assay identifies a previous misclassification of BARD1 c.1977A>G variant. Scientific Reports, 2021, 11, 22948.	3.3	0
244	CDK5RAP3, a New BRCA2 Partner That Regulates DNA Repair, Is Associated with Breast Cancer Survival. Cancers, 2022, 14, 353.	3.7	0