Ignacio Blanco

List of Publications by Year in descending order

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214 papers	19,632 citations	23567 58 h-index	12597 132 g-index
239 all docs	239 docs citations	239 times ranked	28698 citing authors

#	Article	IF	Citations
1	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
2	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
3	Embarazo a tÃ©rmino de una paciente con enfisema pulmonar grave asociado a dÃ©ficit grave (PI*ZZ) de alfa-1 antitripsina. Archivos De Bronconeumologia, 2022, 58, 427-428.	0.8	1
4	Comparison between mid-nasal swabs and buccal swabs for SARS-CoV-2 detection in mild COVID-19 patients. Journal of Infection, 2022, 84, e78-e79.	3.3	1
5	High-titre methylene blue-treated convalescent plasma as an early treatment for outpatients with COVID-19: a randomised, placebo-controlled trial. Lancet Respiratory Medicine,the, 2022, 10, 278-288.	10.7	61
6	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
7	ERN GENTURIS clinical practice guidelines for the diagnosis, treatment, management and surveillance of people with schwannomatosis. European Journal of Human Genetics, 2022, 30, 812-817.	2.8	11
8	Prospective individual patient data meta-analysis of two randomized trials on convalescent plasma for COVID-19 outpatients. Nature Communications, 2022, 13, 2583.	12.8	25
9	[Translated article] Term Pregnancy in a Patient With Severe Pulmonary Emphysema Associated With Pi*ZZ Alpha-1 Antitrypsin Deficiency. Archivos De Bronconeumologia, 2022, , .	0.8	0
10	Heterogeneous Infectivity and Pathogenesis of SARS-CoV-2 Variants Beta, Delta and Omicron in Transgenic K18-hACE2 and Wildtype Mice. Frontiers in Microbiology, 2022, 13, .	3.5	39
11	Performance of SARS-CoV-2 Antigen-Detecting Rapid Diagnostic Tests for Omicron and Other Variants of Concern. Frontiers in Microbiology, 2022, 13, .	3.5	15
12	The risk of COVID-19 death is much greater and age dependent with type I IFN autoantibodies. Proceedings of the National Academy of Sciences of the United States of America, 2022, 119, e2200413119.	7.1	110
13	Follow up of the Humoral Response in Healthcare Workers after the Administration of Two Dose of the Anti SARS-CoV-2 Vaccinesâ€"Effectiveness in Delta Variant Breakthrough Infections. Viruses, 2022, 14, 1385.	3.3	1
14	Analysis in the Prospective Lynch Syndrome Database identifies sarcoma as part of the Lynch syndrome tumor spectrum. International Journal of Cancer, 2021, 148, 512-513.	5.1	9
15	Hydroxychloroquine pre-exposure prophylaxis for COVID-19 in healthcare workers. Journal of Antimicrobial Chemotherapy, 2021, 76, 827-829.	3.0	7
16	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
17	Identification of Plitidepsin as Potent Inhibitor of SARS-CoV-2-Induced Cytopathic Effect After a Drug Repurposing Screen. Frontiers in Pharmacology, 2021, 12, 646676.	3.5	40
18	The Challenge of Diagnosing Constitutional Mismatch Repair Deficiency Syndrome in Brain Malignancies from Young Individuals. International Journal of Molecular Sciences, 2021, 22, 4629.	4.1	9

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19	Clinical characteristics, imaging findings, and genetic results of a patient with CEP290-related cone-rod dystrophy. Ophthalmic Genetics, 2021, 42, 474-479.	1.2	1
20	Chromosomal translocations inactivating CDKN2A support a single path for malignant peripheral nerve sheath tumor initiation. Human Genetics, 2021, 140, 1241-1252.	3.8	12
21	Revised diagnostic criteria for neurofibromatosis type 1 and Legius syndrome: an international consensus recommendation. Genetics in Medicine, 2021, 23, 1506-1513.	2.4	290
22	Analytical and clinical performance of the panbio COVID-19 antigen-detecting rapid diagnostic test. Journal of Infection, 2021, 82, 186-230.	3.3	73
23	Previous SARS-CoV-2 Infection Increases B.1.1.7 Cross-Neutralization by Vaccinated Individuals. Viruses, 2021, 13, 1135.	3.3	17
24	Performance characteristics of five antigen-detecting rapid diagnostic test (Ag-RDT) for SARS-CoV-2 asymptomatic infection: a head-to-head benchmark comparison. Journal of Infection, 2021, 82, 269-275.	3.3	42
25	A Cost-Benefit Analysis of the COVID-19 Asymptomatic Mass Testing Strategy in the North Metropolitan Area of Barcelona. International Journal of Environmental Research and Public Health, 2021, 18, 7028.	2.6	17
26	Autoantibodies neutralizing type I IFNs are present in ~4% of uninfected individuals over 70 years old and account for ~20% of COVID-19 deaths. Science Immunology, 2021, 6, .	11.9	357
27	Self-collected mid-nasal swabs and saliva specimens, compared with nasopharyngeal swabs, for SARS-CoV-2 detection in mild COVID-19 patients. Journal of Infection, 2021, 83, 709-737.	3.3	10
28	Estimated Prevalence and Number of PiMZ Genotypes of Alpha-1 Antitrypsin in Seventy-Four Countries Worldwide. International Journal of COPD, 2021, Volume 16, 2617-2630.	2.3	9
29	Same-day SARS-CoV-2 antigen test screening in an indoor mass-gathering live music event: a randomised controlled trial. Lancet Infectious Diseases, The, 2021, 21, 1365-1372.	9.1	73
30	Mutational spectrum by phenotype: panelâ€based NGS testing of patients with clinical suspicion of RASopathy and children with multiple cafÃ©â€uâ€lait macules. Clinical Genetics, 2020, 97, 264-275.	2.0	13
31	Cancer risks by gene, age, and gender in 6350 carriers of pathogenic mismatch repair variants: findings from the Prospective Lynch Syndrome Database. Genetics in Medicine, 2020, 22, 15-25.	2.4	365
32	Î±₁-antitrypsin PI*SZ genotype: a SERPINA1 deficiency haplotype with uncertain clinical and therapeutic implications. European Respiratory Journal, 2020, 55, 2000713.	6.7	1
33	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
34	Prevalence of Î±₁-antitrypsin PiZZ genotypes in patients with COPD in Europe: a systematic review. European Respiratory Review, 2020, 29, 200014.	7.1	31
35	Inborn errors of type I IFN immunity in patients with life-threatening COVID-19. Science, 2020, 370, .	12.6	1,749
36	Autoantibodies against type I IFNs in patients with life-threatening COVID-19. Science, 2020, 370, .	12.6	1,983

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37	Detection of SARS-CoV-2 in a cat owned by a COVID-19â˜affected patient in Spain. Proceedings of the National Academy of Sciences of the United States of America, 2020, 117, 24790-24793.	7.1	154
38	New cis-Acting Variants in PI*S Background Produce Null Phenotypes Causing Alpha-1 Antitrypsin Deficiency. American Journal of Respiratory Cell and Molecular Biology, 2020, 63, 444-451.	2.9	5
39	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
40	Seroprevalence of SARS-CoV-2 IgG specific antibodies among healthcare workers in the Northern Metropolitan Area of Barcelona, Spain, after the first pandemic wave. PLoS ONE, 2020, 15, e0244348.	2.5	28
41	From exome analysis in idiopathic azoospermia to the identification of a high-risk subgroup for occult Fanconi anemia. Genetics in Medicine, 2019, 21, 189-194.	2.4	38
42	Impact of Host Genetics and Biological Response Modifiers on Respiratory Tract Infections. Frontiers in Immunology, 2019, 10, 1013.	4.8	16
43	Lack of association between screening interval and cancer stage in Lynch syndrome may be accounted for by over-diagnosis; a prospective Lynch syndrome database report. Hereditary Cancer in Clinical Practice, 2019, 17, 8.	1.5	42
44	Reprogramming Captures the Genetic and Tumorigenic Properties of Neurofibromatosis Type 1 Plexiform Neurofibromas. Stem Cell Reports, 2019, 12, 411-426.	4.8	28
45	Cutaneous neurofibromas: patientsâ€™ medical burden, current management and therapeutic expectations: results from an online European patient community survey. Orphanet Journal of Rare Diseases, 2019, 14, 286.	2.7	25
46	Boosting care and knowledge about hereditary cancer: European Reference Network on Genetic Tumour Risk Syndromes. Familial Cancer, 2019, 18, 281-284.	1.9	17
47	Geographical distribution of COPD prevalence in Europe, estimated by an inverse distance weighting interpolation technique. International Journal of COPD, 2018, Volume 13, 57-67.	2.3	60
48	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
49	Early Genetic Diagnosis of Neurofibromatosis Type 2 From Skin Plaque Plexiform Schwannomas in Childhood. JAMA Dermatology, 2018, 154, 341.	4.1	16
50	Cancer risk and survival in path_MMR carriers by gene and gender up to 75 years of age: a report from the Prospective Lynch Syndrome Database. Gut, 2018, 67, 1306-1316.	12.1	410
51	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
52	Characterization of Novel Missense Variants of SERPINA1 Gene Causing Alpha-1 Antitrypsin Deficiency. American Journal of Respiratory Cell and Molecular Biology, 2018, 58, 706-716.	2.9	24
53	AdaptaciÃ³n espaÃ±ola de la Escala de Control Personal Percibido ("Perceived Personal Control") en Consejo GenÃ©tico. Psicooncologia, 2018, 15, 23-36.	0.3	0
54	Conviviendo con la Neurofibromatosis tipo 1: RevisiÃ³n de la literatura. Psicooncologia, 2018, 15, 37-48.	0.3	0

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55	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
56	Long-term evolution of lung function in individuals with alpha-1 antitrypsin deficiency from the Spanish registry (REDAAT). International Journal of COPD, 2018, Volume 13, 1001-1007.	2.3	15
57	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
58	Cancer incidence and survival in Lynch syndrome patients receiving colonoscopic and gynaecological surveillance: first report from the prospective Lynch syndrome database. Gut, 2017, 66, 464-472.	12.1	411
59	Registro espaÃ±ol de pacientes con dÃ©ficit de alfa-1 antitripsina: evaluaciÃ³n de la base de datos y anÃ¡lisis de la poblaciÃ³n incluida. Archivos De Bronconeumologia, 2017, 53, 13-18.	0.8	29
60	Spanish Registry of Patients With Alpha-1 Antitrypsin Deficiency: Database Evaluation and Population Analysis. Archivos De Bronconeumologia, 2017, 53, 13-18.	0.8	12
61	Incidence of and survival after subsequent cancers in carriers of pathogenic MMR variants with previous cancer: a report from the prospective Lynch syndrome database. Gut, 2017, 66, 1657-1664.	12.1	127
62	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
63	Stem cell-like transcriptional reprogramming mediates metastatic resistance to mTOR inhibition. Oncogene, 2017, 36, 2737-2749.	5.9	34
64	Alpha-1 antitrypsin Pi*Z gene frequency and Pi*ZZ genotype numbers worldwide: an update. International Journal of COPD, 2017, Volume 12, 561-569.	2.3	117
65	Colorectal cancer incidence in path_MLH1 carriers subjected to different follow-up protocols: a Prospective Lynch Syndrome Database report. Hereditary Cancer in Clinical Practice, 2017, 15, 18.	1.5	49
66	Alpha-1 antitrypsin Pi*SZ genotype: estimated prevalence and number of SZ subjects worldwide. International Journal of COPD, 2017, Volume 12, 1683-1694.	2.3	56
67	AnÃ¡lisis de la comunicaciÃ³n intra-familiar de los resultados genÃ©ticos diagnÃ³sticos en cÃ¡ncer hereditario. Psicooncologia, 2017, 14, 41-52.	0.3	0
68	Other Diseases Associated With Alpha-1 Antitrypsin Deficiency. , 2017, , 159-174.		0
69	Cultural scale adaptation and validation of the Spanish version of the BRCA Self-Concept Scale in women carriers at high risk for hereditary breast and ovarian cancer. Medicina ClÃnica (English) Tj ETQq1 1 0.784314 rgBT /Overlock	1.4	0
70	Von Hippel-Lindau Disease: Genetics and Role of Genetic Counseling in a Multiple Neoplasia Syndrome. Journal of Clinical Oncology, 2016, 34, 2172-2181.	1.6	132
71	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
72	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

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73	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
74	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
75	Indications for Active Case Searches and Intravenous Alpha-1 Antitrypsin Treatment for Patients With Alpha-1 Antitrypsin Deficiency Chronic Pulmonary Obstructive Disease: An Update. Archivos De Bronconeumologia, 2015, 51, 185-192.	0.8	30
76	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
77	Identification of six new susceptibility loci for invasive epithelial ovarian cancer. Nature Genetics, 2015, 47, 164-171.	21.4	221
78	Population-based multicase-control study in common tumors in Spain (MCC-Spain): rationale and study design. Gaceta Sanitaria, 2015, 29, 308-315.	1.5	158
79	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
80	Germline Mutations in FAN1 Cause Hereditary Colorectal Cancer by Impairing DNA Repair. Gastroenterology, 2015, 149, 563-566.	1.3	94
81	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
82	Exome sequencing identifiesMUTYHmutations in a family with colorectal cancer and an atypical phenotype. Gut, 2015, 64, 355-356.	12.1	14
83	ActualizaciÃ³n sobre indicaciones de bÃºsqueda activa de casos y tratamiento con alfa-1 antitripsina por vÃa intravenosa en pacientes con enfermedad pulmonar obstructiva crÃ³nica asociada a dÃ©ficit de alfa-1 antitripsina. Archivos De Bronconeumologia, 2015, 51, 185-192.	0.8	66
84	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
85	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
86	Severe alpha-1 antitrypsin deficiency in composite heterozygotes inheriting a new splicing mutation QOMadrid. Respiratory Research, 2014, 15, 125.	3.6	38
87	Effect of one year of a gluten-free diet on the clinical evolution of irritable bowel syndrome plus fibromyalgia in patients with associated lymphocytic enteritis: a case-control study. Arthritis Research and Therapy, 2014, 16, 421.	3.5	16
88	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
89	Response to â€˜Remarkable prevalence of celiac disease in patients with irritable bowel syndrome plus fibromyalgia in comparison with those with isolated irritable bowel syndrome: a case-finding studyâ€™ â€“ authorsâ€™ reply. Arthritis Research and Therapy, 2014, 16, 403.	3.5	0
90	Targeted Prostate Cancer Screening in BRCA1 and BRCA2 Mutation Carriers: Results from the Initial Screening Round of the IMPACT Study. European Urology, 2014, 66, 489-499.	1.9	195

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91	Application of a 5-tiered scheme for standardized classification of 2,360 unique mismatch repair gene variants in the InSiGHT locus-specific database. Nature Genetics, 2014, 46, 107-115.	21.4	410
92	GALNT12is Not a Major Contributor of Familial Colorectal Cancer Type X. Human Mutation, 2014, 35, 50-52.	2.5	22
93	Limited family structure and triple-negative breast cancer (TNBC) subtype as predictors of BRCA mutations in a genetic counseling cohort of early-onset sporadic breast cancers. Breast Cancer Research and Treatment, 2014, 148, 415-421.	2.5	15
94	New insights into POLE and POLD1 germline mutations in familial colorectal cancer and polyposis. Human Molecular Genetics, 2014, 23, 3506-3512.	2.9	135
95	Prevalence of germline MUTYH mutations among Lynch-like syndrome patients. European Journal of Cancer, 2014, 50, 2241-2250.	2.8	66
96	Little evidence for association between the TGFBR1*6A variant and colorectal cancer: a family-based association study on non-syndromic family members from Australia and Spain. BMC Cancer, 2014, 14, 475.	2.6	1
97	Developing a framework for implementation of genetic services: learning from examples of testing for monogenic forms of common diseases. Journal of Community Genetics, 2014, 5, 337-347.	1.2	33
98	Comprehensive molecular characterisation of hereditary non-polyposis colorectal tumours with mismatch repair proficiency. European Journal of Cancer, 2014, 50, 1964-1972.	2.8	8
99	Identification of a founder EPCAM deletion in Spanish Lynch syndrome families. Clinical Genetics, 2014, 85, 260-266.	2.0	12
100	Longer Telomeres Are Associated with Cancer Risk in MMR-Proficient Hereditary Non-Polyposis Colorectal Cancer. PLoS ONE, 2014, 9, e86063.	2.5	13
101	Mammographic density and breast cancer in women from high-risk families.. Journal of Clinical Oncology, 2014, 32, 1525-1525.	1.6	0
102	Second primary malignances (SPMs) in patients with gastrointestinal stromal tumors (GIST): The potential influence of imatinib treatment.. Journal of Clinical Oncology, 2014, 32, 10552-10552.	1.6	0
103	An association between the PTGS2 rs5275 polymorphism and colorectal cancer risk in families with inherited non-syndromic predisposition. European Journal of Human Genetics, 2013, 21, 1389-1395.	2.8	6
104	Multiple independent variants at the TERT locus are associated with telomere length and risks of breast and ovarian cancer. Nature Genetics, 2013, 45, 371-384.	21.4	493
105	Genetic variant in the telomerase gene modifies cancer risk in Lynch syndrome. European Journal of Human Genetics, 2013, 21, 511-516.	2.8	20
106	Remarkable prevalence of coeliac disease in patients with irritable bowel syndrome plus fibromyalgia in comparison with those with isolated irritable bowel syndrome: a case-finding study. Arthritis Research and Therapy, 2013, 15, R201.	3.5	27
107	Clinical impact of a gluten-free diet on health-related quality of life in seven fibromyalgia syndrome patients with associated celiac disease. BMC Gastroenterology, 2013, 13, 157.	2.0	36
108	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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109	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
110	Revised guidelines for the clinical management of Lynch syndrome (HNPCC): recommendations by a group of European experts. Gut, 2013, 62, 812-823.	12.1	630
111	Usefulness of epithelial cell adhesion molecule expression in the algorithmic approach to Lynch syndrome identification. Human Pathology, 2013, 44, 412-416.	2.0	20
112	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
113	Identification of a BRCA2-Specific Modifier Locus at 6p24 Related to Breast Cancer Risk. PLoS Genetics, 2013, 9, e1003173.	3.5	105
114	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
115	Identification and molecular characterization of a new ovarian cancer susceptibility locus at 17q21.31. Nature Communications, 2013, 4, 1627.	12.8	98
116	Role of Engrailed-2 (EN2) as a prostate cancer detection biomarker in genetically high risk men. Scientific Reports, 2013, 3, 2059.	3.3	26
117	Â¿Por quÃ© las mujeres con cÃ¡ncer de mama deben estar guapas y los hombres con cÃ¡ncer de prÃ³stata pueden ir sin afeitar? oncologÃa, disidencia y cultura hegemÃ³nica. Psicooncologia, 2013, 10, .	0.3	4
118	Telomere Length and Genetic Anticipation in Lynch Syndrome. PLoS ONE, 2013, 8, e61286.	2.5	21
119	Functional and Structural Analysis of C-Terminal BRCA1 Missense Variants. PLoS ONE, 2013, 8, e61302.	2.5	16
120	Crecimiento Post-traumÃ¡tico en supervivientes de cÃ¡ncer y sus otros significativos: Â¿Crecimiento vicario o secundario?. Terapia Psicologica, 2013, 31, 81-92.	0.3	14
121	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
122	Evidence of linkage to chromosomes 10p15.3â€“p15.1, 14q24.3â€“q31.1 and 9q33.3â€“q34.3 in non-syndromic colorectal cancer families. European Journal of Human Genetics, 2012, 20, 91-96.	2.8	11
123	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
124	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
125	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
126	Reduction of severe exacerbations and hospitalization-derived costs in alpha-1-antitrypsin-deficient patients treated with alpha-1-antitrypsin augmentation therapy. Therapeutic Advances in Respiratory Disease, 2012, 6, 67-78.	2.6	44

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127	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
128	MLH1 methylation screening is effective in identifying epimutation carriers. European Journal of Human Genetics, 2012, 20, 1256-1264.	2.8	36
129	Comprehensive functional assessment ofMLH1variants of unknown significance. Human Mutation, 2012, 33, 1576-1588.	2.5	30
130	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
131	Analysis of SLX4/FANCP in non-BRCA1/2-mutated breast cancer families. BMC Cancer, 2012, 12, 84.	2.6	14
132	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
133	ENIGMA-Evidence-based network for the interpretation of germline mutant alleles: An international initiative to evaluate risk and clinical significance associated with sequence variation in BRCA1 and BRCA2 genes. Human Mutation, 2012, 33, 2-7.	2.5	269
134	Ovarian cancer susceptibility alleles and risk of ovarian cancer inBRCA1andBRCA2mutation carriers. Human Mutation, 2012, 33, 690-702.	2.5	34
135	Efectos de la primera visita de consejo genÃ©tico sobre la percepciÃ³n de riesgo y el malestar emocional. Psicooncologia, 2012, 8, .	0.3	2
136	Abstract 90: An evaluation of the genes involved in the Base Excision Repair (BER) pathway as potential phenotypic modifiers of breast cancer risk inBRCA1andBRCA2mutation carriers.. , 2012, , .		1
137	Neurofibromatosis tipo 2: La historia de Lidia. Index De Enfermeria, 2012, 21, 77-81.	0.2	0
138	Exemestane for Breast-Cancer Prevention in Postmenopausal Women. New England Journal of Medicine, 2011, 364, 2381-2391.	27.0	847
139	Exploring the link between MORF4L1 and risk of breast cancer. Breast Cancer Research, 2011, 13, R40.	5.0	23
140	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
141	Targeted prostate cancer screening in men with mutations in BRCA1 and BRCA2 detects aggressive prostate cancer: preliminary analysis of the results of the IMPACT study. BJU International, 2011, 107, 28-39.	2.5	83
142	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
143	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
144	Evidence for a link between TNFRSF11A and risk of breast cancer. Breast Cancer Research and Treatment, 2011, 129, 947-954.	2.5	12

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145	Identification of a new complex rearrangement affecting exon 20 of BRCA1. Breast Cancer Research and Treatment, 2011, 130, 341-344.	2.5	3
146	Haplotype structure in Ashkenazi Jewish BRCA1 and BRCA2 mutation carriers. Human Genetics, 2011, 130, 685-699.	3.8	18
147	Efficacy of alpha1-antitrypsin augmentation therapy in conditions other than pulmonary emphysema. Orphanet Journal of Rare Diseases, 2011, 6, 14.	2.7	54
148	Estimating probability of germline mismatch repair mutations in colorectal cancer patients with microsatellite stable tumors. Hereditary Cancer in Clinical Practice, 2011, 9, P12.	1.5	0
149	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
150	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
151	Transcriptional characteristics of familial nonâ€BRCA1/BRCA2 breast tumors. International Journal of Cancer, 2011, 128, 2635-2644.	5.1	11
152	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
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