Ignacio Blanco

List of Publications by Year in Descending Order

207 papers	13,917 citations	54 h-index	114 g-index
239 ext. papers	17,260 ext. citations	7.1 avg, IF	5.33 L-index

#	Paper	IF	Citations
207	Term pregnancy of a patient with severe pulmonary emphysema associated with (PI*ZZ) alpha-1 antitrypsin.. *Archivos De Bronconeumologia*, **2022**,	0.7	0
206	Comparison between mid-nasal swabs and buccal swabs for SARS-CoV-2 detection in mild COVID-19 patients.. *Journal of Infection*, **2022**,	18.9	
205	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**,	35.1	6
204	Modeling iPSC-derived human neurofibroma-like tumors in mice uncovers the heterogeneity of Schwann cells within plexiform neurofibromas.. *Cell Reports*, **2022**, 38, 110385	10.6	0
203	Prospective individual patient data meta-analysis of two randomized trials on convalescent plasma for COVID-19 outpatients.. *Nature Communications*, **2022**, 13, 2583	17.4	0
202	[Translated article] Term Pregnancy in a Patient With Severe Pulmonary Emphysema Associated With Pi*ZZ Alpha-1 Antitrypsin Deficiency.. *Archivos De Bronconeumologia*, **2022**,	0.7	
201	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	11.5	3
200	Identification of Plitidepsin as Potent Inhibitor of SARS-CoV-2-Induced Cytopathic Effect After a Drug Repurposing Screen. *Frontiers in Pharmacology*, **2021**, 12, 646676	5.6	17
199	The Challenge of Diagnosing Constitutional Mismatch Repair Deficiency Syndrome in Brain Malignancies from Young Individuals. *International Journal of Molecular Sciences*, **2021**, 22,	6.3	1
198	Clinical characteristics, imaging findings, and genetic results of a patient with -related cone-rod dystrophy. *Ophthalmic Genetics*, **2021**, 42, 474-479	1.2	0
197	Chromosomal translocations inactivating CDKN2A support a single path for malignant peripheral nerve sheath tumor initiation. *Human Genetics*, **2021**, 140, 1241-1252	6.3	2
196	Revised diagnostic criteria for neurofibromatosis type 1 and Legius syndrome: an international consensus recommendation. *Genetics in Medicine*, **2021**, 23, 1506-1513	8.1	43
195	Analytical and clinical performance of the panbio COVID-19 antigen-detecting rapid diagnostic test. *Journal of Infection*, **2021**, 82, 186-230	18.9	43
194	Previous SARS-CoV-2 Infection Increases B.1.1.7 Cross-Neutralization by Vaccinated Individuals. *Viruses*, **2021**, 13,	6.2	6
193	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	18.9	18
192	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,	4.6	5
191	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	7.5	2

190	Hydroxychloroquine pre-exposure prophylaxis for COVID-19 in healthcare workers. *Journal of Antimicrobial Chemotherapy*, **2021**, 76, 827-829	5.1	6
189	Using antisense oligonucleotides for the physiological modulation of the alternative splicing of NF1 exon 23a during PC12 neuronal differentiation. *Scientific Reports*, **2021**, 11, 3661	4.9	2
188	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,	28	91
187	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**,	18.9	2
186	Estimated Prevalence and Number of PiMZ Genotypes of Alpha-1 Antitrypsin in Seventy-Four Countries Worldwide. *International Journal of COPD*, **2021**, 16, 2617-2630	3	1
185	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	25.5	24
184	New -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	5.7	4
183	KIF11 and KIF15 mitotic kinesins are potential therapeutic vulnerabilities for malignant peripheral nerve sheath tumors. *Neuro-Oncology Advances*, **2020**, 2, i62-i74	0.9	6
182	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	3.7	16
181	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, 1758835920929579	5.4	2
180	Prevalence of □antitrypsin PiZZ genotypes in patients with COPD in Europe: a systematic review. *European Respiratory Review*, **2020**, 29,	9.8	16
179	Inborn errors of type I IFN immunity in patients with life-threatening COVID-19. *Science*, **2020**, 370,	33.3	994
178	Autoantibodies against type I IFNs in patients with life-threatening COVID-19. *Science*, **2020**, 370,	33.3	1090
177	Detection of SARS-CoV-2 in a cat owned by a COVID-19-affected patient in Spain. *Proceedings of the National Academy of Sciences of the United States of America*, **2020**, 117, 24790-24793	11.5	94
176	Mutational spectrum by phenotype: panel-based NGS testing of patients with clinical suspicion of RASopathy and children with multiple caf□au-lait macules. *Clinical Genetics*, **2020**, 97, 264-275	4	9
175	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	8.1	164
174	Impact of Host Genetics and Biological Response Modifiers on Respiratory Tract Infections. *Frontiers in Immunology*, **2019**, 10, 1013	8.4	10
173	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	2.3	24

172	Reprogramming Captures the Genetic and Tumorigenic Properties of Neurofibromatosis Type 1 Plexiform Neurofibromas. *Stem Cell Reports*, **2019**, 12, 411-426	8	13
171	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	8.1	23
170	Cutaneous neurofibromas: patientsN medical burden, current management and therapeutic expectations: results from an online European patient community survey. *Orphanet Journal of Rare Diseases*, **2019**, 14, 286	4.2	6
169	Boosting care and knowledge about hereditary cancer: European Reference Network on Genetic Tumour Risk Syndromes. *Familial Cancer*, **2019**, 18, 281-284	3	6
168	Geographical distribution of COPD prevalence in Europe, estimated by an inverse distance weighting interpolation technique. *International Journal of COPD*, **2018**, 13, 57-67	3	40
167	Consensus document on the implementation of next generation sequencing in the genetic diagnosis of hereditary cancer. *Medicina Cl□nica*, **2018**, 151, 80.e1-80.e10	1	5
166	Early Genetic Diagnosis of Neurofibromatosis Type 2 From Skin Plaque Plexiform Schwannomas in Childhood. *JAMA Dermatology*, **2018**, 154, 341-346	5.1	8
165	Cancer risk and survival in carriers by gene and gender up to 75 years of age: a report from the Prospective Lynch Syndrome Database. *Gut*, **2018**, 67, 1306-1316	19.2	259
164	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	13.3	25
163	Long-term evolution of lung function in individuals with alpha-1 antitrypsin deficiency from the Spanish registry (REDAAT). *International Journal of COPD*, **2018**, 13, 1001-1007	3	9
162	Consensus document on the implementation of next generation sequencing in the genetic diagnosis of hereditary cancer. *Medicina Cl□nica (English Edition)*, **2018**, 151, 80.e1-80.e10	0.3	0
161	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	5.7	16
160	Adaptaci□ espa□ola de la Escala de Control Personal Percibido ("Perceived Personal Control") en Consejo Gen□tico. *Psicooncologia*, **2018**, 15, 23-36	0.4	
159	Conviviendo con la Neurofibromatosis tipo 1: Revisi□ de la literatura. *Psicooncologia*, **2018**, 15, 37-48	0.4	
158	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	4.7	21
157	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	19.2	291
156	Spanish Registry of Patients With Alpha-1 Antitrypsin Deficiency: Database Evaluation and Population Analysis. *Archivos De Bronconeumologia*, **2017**, 53, 13-18	0.7	20
155	Spanish Registry of Patients With Alpha-1 Antitrypsin Deficiency: Database Evaluation and Population Analysis. *Archivos De Bronconeumologia*, **2017**, 53, 13-18	0.7	2

154	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	19.2	87
153	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	4.9	32
152	Stem cell-like transcriptional reprogramming mediates metastatic resistance to mTOR inhibition. *Oncogene*, **2017**, 36, 2737-2749	9.2	27
151	Alpha-1 antitrypsin Pi*Z gene frequency and Pi*ZZ genotype numbers worldwide: an update. *International Journal of COPD*, **2017**, 12, 561-569	3	72
150	Colorectal cancer incidence in carriers subjected to different follow-up protocols: a Prospective Lynch Syndrome Database report. *Hereditary Cancer in Clinical Practice*, **2017**, 15, 18	2.3	27
149	Alpha-1 antitrypsin Pi*SZ genotype: estimated prevalence and number of SZ subjects worldwide. *International Journal of COPD*, **2017**, 12, 1683-1694	3	46
148	An□lisis de la comunicaci□ intra-familiar de los resultados gen□ticos diagn□sticos en c□ncer hereditario. *Psicooncologia*, **2017**, 14, 41-52	0.4	
147	Other Diseases Associated With Alpha-1 Antitrypsin Deficiency **2017**, 159-174		
146	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, 325-32	8.1	153
145	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 Cl□nica (English Edition)*, **2016**, 146, 148-154	0.3	
144	Von Hippel-Lindau Disease: Genetics and Role of Genetic Counseling in a Multiple Neoplasia Syndrome. *Journal of Clinical Oncology*, **2016**, 34, 2172-81	2.2	95
143	Identification of six new susceptibility loci for invasive epithelial ovarian cancer. *Nature Genetics*, **2015**, 47, 164-71	36.3	177
142	Population-based multicase-control study in common tumors in Spain (MCC-Spain): rationale and study design. *Gaceta Sanitaria*, **2015**, 29, 308-15	2.2	120
141	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	3.7	21
140	Germline Mutations in FAN1 Cause Hereditary Colorectal Cancer by Impairing DNA Repair. *Gastroenterology*, **2015**, 149, 563-6	13.3	75
139	Association of type and location of BRCA1 and BRCA2 mutations with risk of breast and ovarian cancer. *JAMA - Journal of the American Medical Association*, **2015**, 313, 1347-61	27.4	286
138	Exome sequencing identifies MUTYH mutations in a family with colorectal cancer and an atypical phenotype. *Gut*, **2015**, 64, 355-6	19.2	14
137	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, 185-92	0.7	48

136	Candidate genetic modifiers for breast and ovarian cancer risk in BRCA1 and BRCA2 mutation carriers. *Cancer Epidemiology Biomarkers and Prevention*, **2015**, 24, 308-16	4	20
135	Comprehensive establishment and characterization of orthoxenograft mouse models of malignant peripheral nerve sheath tumors for personalized medicine. *EMBO Molecular Medicine*, **2015**, 7, 608-27	12	29
134	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	8.3	16
133	Assessing associations between the AURKA-HMMR-TPX2-TUBG1 functional module and breast cancer risk in BRCA1/2 mutation carriers. *PLoS ONE*, **2015**, 10, e0120020	3.7	26
132	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	0.7	3
131	Germline mutation in BRCA1 or BRCA2 and ten-year survival for women diagnosed with epithelial ovarian cancer. *Clinical Cancer Research*, **2015**, 21, 652-7	12.9	107
130	Response to ℵRemarkable prevalence of celiac disease in patients with irritable bowel syndrome plus fibromyalgia in comparison with those with isolated irritable bowel syndrome: a case-finding studyℵ authorsℵreply. *Arthritis Research and Therapy*, **2014**, 16, 403	5.7	
129	Targeted prostate cancer screening in BRCA1 and BRCA2 mutation carriers: results from the initial screening round of the IMPACT study. *European Urology*, **2014**, 66, 489-99	10.2	156
128	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	36.3	332
127	GALNT12 is not a major contributor of familial colorectal cancer type X. *Human Mutation*, **2014**, 35, 50-2	4.7	20
126	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, 415-21	4.4	14
125	New insights into POLE and POLD1 germline mutations in familial colorectal cancer and polyposis. *Human Molecular Genetics*, **2014**, 23, 3506-12	5.6	110
124	Prevalence of germline MUTYH mutations among Lynch-like syndrome patients. *European Journal of Cancer*, **2014**, 50, 2241-50	7.5	54
123	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	4.8	1
122	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, 337-47	2.5	20
121	Comprehensive molecular characterisation of hereditary non-polyposis colorectal tumours with mismatch repair proficiency. *European Journal of Cancer*, **2014**, 50, 1964-72	7.5	7
120	Identification of a founder EPCAM deletion in Spanish Lynch syndrome families. *Clinical Genetics*, **2014**, 85, 260-6	4	11
119	DNA glycosylases involved in base excision repair may be associated with cancer risk in BRCA1 and BRCA2 mutation carriers. *PLoS Genetics*, **2014**, 10, e1004256	6	33

118	Severe alpha-1 antitrypsin deficiency in composite heterozygotes inheriting a new splicing mutation QOMadrid. *Respiratory Research*, **2014**, 15, 125	7.3	31
117	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	5.7	12
116	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	8.3	46
115	Longer telomeres are associated with cancer risk in MMR-proficient hereditary non-polyposis colorectal cancer. *PLoS ONE*, **2014**, 9, e86063	3.7	11
114	Mammographic density and breast cancer in women from high-risk families.. *Journal of Clinical Oncology*, **2014**, 32, 1525-1525	2.2	
113	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	2.2	
112	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, 1389-95	5.3	5
111	Multiple independent variants at the TERT locus are associated with telomere length and risks of breast and ovarian cancer. *Nature Genetics*, **2013**, 45, 371-84, 384e1-2	36.3	422
110	Genetic variant in the telomerase gene modifies cancer risk in Lynch syndrome. *European Journal of Human Genetics*, **2013**, 21, 511-6	5.3	17
109	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	5.7	21
108	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	3	22
107	Refining the role of PMS2 in Lynch syndrome: germline mutational analysis improved by comprehensive assessment of variants. *Journal of Medical Genetics*, **2013**, 50, 552-63	5.8	40
106	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, 864-70	5.3	84
105	Revised guidelines for the clinical management of Lynch syndrome (HNPCC): recommendations by a group of European experts. *Gut*, **2013**, 62, 812-23	19.2	500
104	Usefulness of epithelial cell adhesion molecule expression in the algorithmic approach to Lynch syndrome identification. *Human Pathology*, **2013**, 44, 412-6	3.7	19
103	In vitro antisense therapeutics for a deep intronic mutation causing Neurofibromatosis type 2. *European Journal of Human Genetics*, **2013**, 21, 769-73	5.3	14
102	Identification of a BRCA2-specific modifier locus at 6p24 related to breast cancer risk. *PLoS Genetics*, **2013**, 9, e1003173	6	90
101	Genome-wide association study in BRCA1 mutation carriers identifies novel loci associated with breast and ovarian cancer risk. *PLoS Genetics*, **2013**, 9, e1003212	6	209

100	Identification and molecular characterization of a new ovarian cancer susceptibility locus at 17q21.31. *Nature Communications*, **2013**, 4, 1627	17.4	85
99	Role of Engrailed-2 (EN2) as a prostate cancer detection biomarker in genetically high risk men. *Scientific Reports*, **2013**, 3, 2059	4.9	18
98	¿Por qu las mujeres con cncer de mama deben estar guapas y los hombres con cncer de prstata pueden ir sin afeitar? oncologa, disidencia y cultura hegemnica. *Psicooncologia*, **2013**, 10,	0.4	2
97	Telomere length and genetic anticipation in Lynch syndrome. *PLoS ONE*, **2013**, 8, e61286	3.7	20
96	Functional and structural analysis of C-terminal BRCA1 missense variants. *PLoS ONE*, **2013**, 8, e61302	3.7	14
95	Crecimiento Post-traumtico en supervivientes de cncer y sus otros significativos: ¿Crecimiento vicario o secundario?. *Terapia Psicologica*, **2013**, 31, 81-92	1.9	10
94	Analysis of SLX4/FANCP in non-BRCA1/2-mutated breast cancer families. *BMC Cancer*, **2012**, 12, 84	4.8	13
93	Common variants at 12p11, 12q24, 9p21, 9q31.2 and in ZNF365 are associated with breast cancer risk for BRCA1 and/or BRCA2 mutation carriers. *Breast Cancer Research*, **2012**, 14, R33	8.3	70
92	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	4.7	211
91	Ovarian cancer susceptibility alleles and risk of ovarian cancer in BRCA1 and BRCA2 mutation carriers. *Human Mutation*, **2012**, 33, 690-702	4.7	31
90	Association between BRCA1 and BRCA2 mutations and survival in women with invasive epithelial ovarian cancer. *JAMA - Journal of the American Medical Association*, **2012**, 307, 382-90	27.4	427
89	MLH1 methylation screening is effective in identifying epimutation carriers. *European Journal of Human Genetics*, **2012**, 20, 1256-64	5.3	26
88	Comprehensive functional assessment of MLH1 variants of unknown significance. *Human Mutation*, **2012**, 33, 1576-88	4.7	25
87	Assessing the RNA effect of 26 DNA variants in the BRCA1 and BRCA2 genes. *Breast Cancer Research and Treatment*, **2012**, 132, 979-92	4.4	19
86	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, 1362-70	4	20
85	Evidence of linkage to chromosomes 10p15.3-p15.1, 14q24.3-q31.1 and 9q33.3-q34.3 in non-syndromic colorectal cancer families. *European Journal of Human Genetics*, **2012**, 20, 91-6	5.3	11
84	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, 645-57	4	44
83	MLH1 promoter hypermethylation in the analytical algorithm of Lynch syndrome: a cost-effectiveness study. *European Journal of Human Genetics*, **2012**, 20, 762-8	5.3	67

82	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, 134-47	4	411
81	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	4.9	35
80	Efectos de la primera visita de consejo gen□ico sobre la percepci□ de riesgo y el malestar emocional. *Psicooncologia*, **2012**, 8,	0.4	2
79	Exemestane for breast-cancer prevention in postmenopausal women. *New England Journal of Medicine*, **2011**, 364, 2381-91	59.2	705
78	Exploring the link between MORF4L1 and risk of breast cancer. *Breast Cancer Research*, **2011**, 13, R40	8.3	16
77	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	8.3	62
76	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	5.6	76
75	International distribution and age estimation of the Portuguese BRCA2 c.156_157insAlu founder mutation. *Breast Cancer Research and Treatment*, **2011**, 127, 671-9	4.4	21
74	Germline ATM mutational analysis in BRCA1/BRCA2 negative hereditary breast cancer families by MALDI-TOF mass spectrometry. *Breast Cancer Research and Treatment*, **2011**, 128, 573-9	4.4	6
73	Evidence for a link between TNFRSF11A and risk of breast cancer. *Breast Cancer Research and Treatment*, **2011**, 129, 947-54	4.4	11
72	Identification of a new complex rearrangement affecting exon 20 of BRCA1. *Breast Cancer Research and Treatment*, **2011**, 130, 341-4	4.4	2
71	Haplotype structure in Ashkenazi Jewish BRCA1 and BRCA2 mutation carriers. *Human Genetics*, **2011**, 130, 685-99	6.3	15
70	Efficacy of alpha1-antitrypsin augmentation therapy in conditions other than pulmonary emphysema. *Orphanet Journal of Rare Diseases*, **2011**, 6, 14	4.2	44
69	Estimating probability of germline mismatch repair mutations in colorectal cancer patients with microsatellite stable tumors. *Hereditary Cancer in Clinical Practice*, **2011**, 9, P12	2.3	78
68	Dissecting loss of heterozygosity (LOH) in neurofibromatosis type 1-associated neurofibromas: Importance of copy neutral LOH. *Human Mutation*, **2011**, 32, 78-90	4.7	52
67	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, 705-9	4.7	15
66	Transcriptional characteristics of familial non-BRCA1/BRCA2 breast tumors. *International Journal of Cancer*, **2011**, 128, 2635-44	7.5	11
65	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, 3304-21	5.6	62

64	Uveal melanoma and BRCA1/BRCA2 genes: a relationship that needs further investigation. *Journal of Clinical Oncology*, **2011**, 29, e827-9	2.2	21
63	Common variants of the BRCA1 wild-type allele modify the risk of breast cancer in BRCA1 mutation carriers. *Human Molecular Genetics*, **2011**, 20, 4732-47	5.6	21
62	Genetic variation at 9p22.2 and ovarian cancer risk for BRCA1 and BRCA2 mutation carriers. *Journal of the National Cancer Institute*, **2011**, 103, 105-16	9.7	37
61	Interplay between BRCA1 and RHAMM regulates epithelial apicobasal polarization and may influence risk of breast cancer. *PLoS Biology*, **2011**, 9, e1001199	9.7	73
60	Clinical and genetic characterization of classical forms of familial adenomatous polyposis: a Spanish population study. *Annals of Oncology*, **2011**, 22, 903-909	10.3	25
59	A large-scale meta-analysis to refine colorectal cancer risk estimates associated with MUTYH variants. *British Journal of Cancer*, **2010**, 103, 1875-84	8.7	91
58	Modulation of aberrant NF1 pre-mRNA splicing by kinetin treatment. *European Journal of Human Genetics*, **2010**, 18, 614-7	5.3	16
57	A locus on 19p13 modifies risk of breast cancer in BRCA1 mutation carriers and is associated with hormone receptor-negative breast cancer in the general population. *Nature Genetics*, **2010**, 42, 885-92	36.3	276
56	Founder effect of a pathogenic MSH2 mutation identified in Spanish families with Lynch syndrome. *Clinical Genetics*, **2010**, 78, 186-90	4	10
55	The rs10993994 risk allele for prostate cancer results in clinically relevant changes in microseminoprotein-beta expression in tissue and urine. *PLoS ONE*, **2010**, 5, e13363	3.7	68
54	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, 9742-54	10.1	147
53	MLH1 founder mutations with moderate penetrance in Spanish Lynch syndrome families. *Cancer Research*, **2010**, 70, 7379-91	10.1	28
52	Survival in women with MMR mutations and ovarian cancer: a multicentre study in Lynch syndrome kindreds. *Journal of Medical Genetics*, **2010**, 47, 99-102	5.8	53
51	Common genetic variants and modification of penetrance of BRCA2-associated breast cancer. *PLoS Genetics*, **2010**, 6, e1001183	6	74
50	Novel methylation panel for the early detection of colorectal tumors in stool DNA. *Clinical Colorectal Cancer*, **2010**, 9, 168-76	3.8	52
49	Allele-specific expression of APC in adenomatous polyposis families. *Gastroenterology*, **2010**, 139, 439-47, 447.e1	13.3	32
48	Peutz-Jeghers syndrome: a systematic review and recommendations for management. *Gut*, **2010**, 59, 975-86	19.2	482
47	Low plasma levels of monocyte chemoattractant protein-1 (MCP-1), tumor necrosis factor-alpha (TNFalpha), and vascular endothelial growth factor (VEGF) in patients with alpha1-antitrypsin deficiency-related fibromyalgia. *Clinical Rheumatology*, **2010**, 29, 189-97	3.9	17

46	Abnormal overexpression of mastocytes in skin biopsies of fibromyalgia patients. *Clinical Rheumatology*, **2010**, 29, 1403-12	3.9	63
45	Parity and the risk of breast and ovarian cancer in BRCA1 and BRCA2 mutation carriers. *Breast Cancer Research and Treatment*, **2010**, 119, 221-32	4.4	48
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