Ignacio Blanco

List of Publications by Citations

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	Autoantibodies against type I IFNs in patients with life-threatening COVID-19. *Science*, **2020**, 370,	33.3	1090
206	Inborn errors of type I IFN immunity in patients with life-threatening COVID-19. *Science*, **2020**, 370,	33.3	994
205	Exemestane for breast-cancer prevention in postmenopausal women. *New England Journal of Medicine*, **2011**, 364, 2381-91	59.2	705
204	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
203	Peutz-Jeghers syndrome: a systematic review and recommendations for management. *Gut*, **2010**, 59, 975-86	19.2	482
202	Guidelines for the clinical management of familial adenomatous polyposis (FAP). *Gut*, **2008**, 57, 704-13	19.2	477
201	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
200	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
199	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
198	Guidelines for the clinical management of Lynch syndrome (hereditary non-polyposis cancer). *Journal of Medical Genetics*, **2007**, 44, 353-62	5.8	392
197	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
196	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
195	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
194	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
193	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
192	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
191	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

190	Polymorphisms in genes of nucleotide and base excision repair: risk and prognosis of colorectal cancer. *Clinical Cancer Research*, **2006**, 12, 2101-8	12.9	209
189	Posttraumatic growth in cancer: reality or illusion?. *Clinical Psychology Review*, **2009**, 29, 24-33	10.8	178
188	Identification of six new susceptibility loci for invasive epithelial ovarian cancer. *Nature Genetics*, **2015**, 47, 164-71	36.3	177
187	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
186	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
185	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
184	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
183	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
182	New insights into POLE and POLD1 germline mutations in familial colorectal cancer and polyposis. *Human Molecular Genetics*, **2014**, 23, 3506-12	5.6	110
181	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
180	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
179	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
178	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
177	Common variants in LSP1, 2q35 and 8q24 and breast cancer risk for BRCA1 and BRCA2 mutation carriers. *Human Molecular Genetics*, **2009**, 18, 4442-56	5.6	91
176	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
175	Identification of a BRCA2-specific modifier locus at 6p24 related to breast cancer risk. *PLoS Genetics*, **2013**, 9, e1003173	6	90
174	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
173	Identification and molecular characterization of a new ovarian cancer susceptibility locus at 17q21.31. *Nature Communications*, **2013**, 4, 1627	17.4	85

172	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
171	Recommendations to improve identification of hereditary and familial colorectal cancer in Europe. *Familial Cancer*, **2010**, 9, 109-15	3	84
170	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
169	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, 2861-9	12.9	77
168	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
167	Germline Mutations in FAN1 Cause Hereditary Colorectal Cancer by Impairing DNA Repair. *Gastroenterology*, **2015**, 149, 563-6	13.3	75
166	Analysis of FANCB and FANCN/PALB2 fanconi anemia genes in BRCA1/2-negative Spanish breast cancer families. *Breast Cancer Research and Treatment*, **2009**, 113, 545-51	4.4	75
165	Common genetic variants and modification of penetrance of BRCA2-associated breast cancer. *PLoS Genetics*, **2010**, 6, e1001183	6	74
164	Interplay between BRCA1 and RHAMM regulates epithelial apicobasal polarization and may influence risk of breast cancer. *PLoS Biology*, **2011**, 9, e1001199	9.7	73
163	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
162	Diagn□stico y tratamiento del d□ficit de alfa-1-antitripsina. *Archivos De Bronconeumologia*, **2006**, 42, 645-659	3.7	71
161	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
160	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
159	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
158	Low levels of microsatellite instability characterize MLH1 and MSH2 HNPCC carriers before tumor diagnosis. *Human Molecular Genetics*, **2005**, 14, 235-9	5.6	65
157	Abnormal overexpression of mastocytes in skin biopsies of fibromyalgia patients. *Clinical Rheumatology*, **2010**, 29, 1403-12	3.9	63
156	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
155	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

154	Prevalence of germline MUTYH mutations among Lynch-like syndrome patients. *European Journal of Cancer*, **2014**, 50, 2241-50	7.5	54
153	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
152	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
151	Novel methylation panel for the early detection of colorectal tumors in stool DNA. *Clinical Colorectal Cancer*, **2010**, 9, 168-76	3.8	52
150	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
149	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
148	Alpha-1 antitrypsin Pi*SZ genotype: estimated prevalence and number of SZ subjects worldwide. *International Journal of COPD*, **2017**, 12, 1683-1694	3	46
147	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
146	Efficacy of alpha1-antitrypsin augmentation therapy in conditions other than pulmonary emphysema. *Orphanet Journal of Rare Diseases*, **2011**, 6, 14	4.2	44
145	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
144	Conventional renal cancer in a patient with fumarate hydratase mutation. *Human Pathology*, **2007**, 38, 793-6	3.7	43
143	Revised diagnostic criteria for neurofibromatosis type 1 and Legius syndrome: an international consensus recommendation. *Genetics in Medicine*, **2021**, 23, 1506-1513	8.1	43
142	Analytical and clinical performance of the panbio COVID-19 antigen-detecting rapid diagnostic test. *Journal of Infection*, **2021**, 82, 186-230	18.9	43
141	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
140	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
139	Screening for large rearrangements of the BRCA2 gene in Spanish families with breast/ovarian cancer. *Breast Cancer Research and Treatment*, **2007**, 103, 103-7	4.4	38
138	Antisense therapeutics for neurofibromatosis type 1 caused by deep intronic mutations. *Human Mutation*, **2009**, 30, 454-62	4.7	37
137	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

136	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
135	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
134	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
133	Allele-specific expression of APC in adenomatous polyposis families. *Gastroenterology*, **2010**, 139, 439-47, 447.e1	13.3	32
132	Ovarian cancer susceptibility alleles and risk of ovarian cancer in BRCA1 and BRCA2 mutation carriers. *Human Mutation*, **2012**, 33, 690-702	4.7	31
131	Severe alpha-1 antitrypsin deficiency in composite heterozygotes inheriting a new splicing mutation QOMadrid. *Respiratory Research*, **2014**, 15, 125	7.3	31
130	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
129	Identification and comprehensive characterization of large genomic rearrangements in the BRCA1 and BRCA2 genes. *Breast Cancer Research and Treatment*, **2010**, 122, 733-43	4.4	29
128	Detection of genetic alterations in hereditary colorectal cancer screening. *Mutation Research - Fundamental and Molecular Mechanisms of Mutagenesis*, **2010**, 693, 19-31	3.3	29
127	MLH1 founder mutations with moderate penetrance in Spanish Lynch syndrome families. *Cancer Research*, **2010**, 70, 7379-91	10.1	28
126	Stem cell-like transcriptional reprogramming mediates metastatic resistance to mTOR inhibition. *Oncogene*, **2017**, 36, 2737-2749	9.2	27
125	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
124	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
123	MLH1 methylation screening is effective in identifying epimutation carriers. *European Journal of Human Genetics*, **2012**, 20, 1256-64	5.3	26
122	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
121	Comprehensive functional assessment of MLH1 variants of unknown significance. *Human Mutation*, **2012**, 33, 1576-88	4.7	25
120	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
119	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

118	Genome-wide linkage scan reveals three putative breast-cancer-susceptibility loci. *American Journal of Human Genetics*, **2009**, 84, 115-22	11	24
117	No evidence for a genetic modifier for renal cell cancer risk in HLRCC syndrome. *Familial Cancer*, **2010**, 9, 245-51	3	24
116	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
115	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
114	Perception of breast cancer risk and surveillance behaviours of women with family history of breast cancer: a brief report on a Spanish cohort. *Psycho-Oncology*, **2003**, 12, 821-7	3.9	23
113	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
112	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
111	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
110	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
109	Uveal melanoma and BRCA1/BRCA2 genes: a relationship that needs further investigation. *Journal of Clinical Oncology*, **2011**, 29, e827-9	2.2	21
108	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
107	Detection of APC gene deletions using quantitative multiplex PCR of short fluorescent fragments. *Clinical Chemistry*, **2008**, 54, 1132-40	5.5	21
106	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
105	Spanish Registry of Patients With Alpha-1 Antitrypsin Deficiency: Database Evaluation and Population Analysis. *Archivos De Bronconeumologia*, **2017**, 53, 13-18	0.7	20
104	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
103	GALNT12 is not a major contributor of familial colorectal cancer type X. *Human Mutation*, **2014**, 35, 50-2	4.7	20
102	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
101	Telomere length and genetic anticipation in Lynch syndrome. *PLoS ONE*, **2013**, 8, e61286	3.7	20

100	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
99	Usefulness of epithelial cell adhesion molecule expression in the algorithmic approach to Lynch syndrome identification. *Human Pathology*, **2013**, 44, 412-6	3.7	19
98	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
97	The impact of genetic counseling on knowledge and emotional responses in Spanish population with family history of breast cancer. *Patient Education and Counseling*, **2010**, 78, 382-8	3.1	19
96	Hereditary familial polyposis and Gardner's syndrome: contribution of the odonto-stomatology examination in its diagnosis and a case description. *Medicina Oral, Patologia Oral Y Cirugia Bucal*, **2005**, 10, 402-9	2.6	19
95	Role of Engrailed-2 (EN2) as a prostate cancer detection biomarker in genetically high risk men. *Scientific Reports*, **2013**, 3, 2059	4.9	18
94	Functional characterization of the novel APC N1026S variant associated with attenuated familial adenomatous polyposis. *Gastroenterology*, **2008**, 134, 56-64	13.3	18
93	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
92	Alpha1-antitrypsin replacement therapy controls fibromyalgia symptoms in 2 patients with PI ZZ alpha1-antitrypsin deficiency. *Journal of Rheumatology*, **2004**, 31, 2082-5	4.1	18
91	Genetic variant in the telomerase gene modifies cancer risk in Lynch syndrome. *European Journal of Human Genetics*, **2013**, 21, 511-6	5.3	17
90	The TP53 Arg72Pro and MDM2 309G>T polymorphisms are not associated with breast cancer risk in BRCA1 and BRCA2 mutation carriers. *British Journal of Cancer*, **2009**, 101, 1456-60	8.7	17
89	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
88	Non-Hodgkin lymphoma related to hereditary nonpolyposis colorectal cancer in a patient with a novel heterozygous complex deletion in the MSH2 gene. *Genes Chromosomes and Cancer*, **2008**, 47, 326-32	5	17
87	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
86	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
85	Exploring the link between MORF4L1 and risk of breast cancer. *Breast Cancer Research*, **2011**, 13, R40	8.3	16
84	Modulation of aberrant NF1 pre-mRNA splicing by kinetin treatment. *European Journal of Human Genetics*, **2010**, 18, 614-7	5.3	16
83	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

82	Prevalence of □antitrypsin PiZZ genotypes in patients with COPD in Europe: a systematic review. *European Respiratory Review*, **2020**, 29,	9.8	16
81	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
80	Haplotype structure in Ashkenazi Jewish BRCA1 and BRCA2 mutation carriers. *Human Genetics*, **2011**, 130, 685-99	6.3	15
79	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
78	Antiidiotypic response against murine monoclonal antibodies reactive with tumor-associated antigen TAG-72. *Journal of Clinical Immunology*, **1997**, 17, 96-106	5.7	15
77	Colorectal cancer risk and the APC D1822V variant. *International Journal of Cancer*, **2004**, 112, 161-3	7.5	15
76	Exome sequencing identifies MUTYH mutations in a family with colorectal cancer and an atypical phenotype. *Gut*, **2015**, 64, 355-6	19.2	14
75	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
74	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
73	Functional and structural analysis of C-terminal BRCA1 missense variants. *PLoS ONE*, **2013**, 8, e61302	3.7	14
72	Reprogramming Captures the Genetic and Tumorigenic Properties of Neurofibromatosis Type 1 Plexiform Neurofibromas. *Stem Cell Reports*, **2019**, 12, 411-426	8	13
71	Analysis of SLX4/FANCP in non-BRCA1/2-mutated breast cancer families. *BMC Cancer*, **2012**, 12, 84	4.8	13
70	Gene expression profiling integrated into network modelling reveals heterogeneity in the mechanisms of BRCA1 tumorigenesis. *British Journal of Cancer*, **2009**, 101, 1469-80	8.7	13
69	Germline hypermethylation of the APC promoter is not a frequent cause of familial adenomatous polyposis in APC/MUTYH mutation negative families. *International Journal of Cancer*, **2008**, 122, 1422-5	7.5	13
68	Founder mutation in familial adenomatous polyposis (FAP) in the Balearic Islands. *Cancer Genetics and Cytogenetics*, **2005**, 158, 70-4		13
67	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
66	Identification of a founder EPCAM deletion in Spanish Lynch syndrome families. *Clinical Genetics*, **2014**, 85, 260-6	4	11
65	Evidence for a link between TNFRSF11A and risk of breast cancer. *Breast Cancer Research and Treatment*, **2011**, 129, 947-54	4.4	11

64	Transcriptional characteristics of familial non-BRCA1/BRCA2 breast tumors. *International Journal of Cancer*, **2011**, 128, 2635-44	7.5	11
63	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
62	Alpha1-Antitrypsin Polymorphism in Fibromyalgia Syndrome Patients from the Asturias Province in Northern Spain: A Significantly Higher Prevalence of the PI*Z Deficiency Allele in Patients Than in the General Population. *Journal of Musculoskeletal Pain*, **2006**, 14, 5-12		11
61	Longer telomeres are associated with cancer risk in MMR-proficient hereditary non-polyposis colorectal cancer. *PLoS ONE*, **2014**, 9, e86063	3.7	11
60	Analytical and Clinical Performance of the Panbio COVID-19 Antigen-Detecting Rapid Diagnostic Test		11
59	Impact of Host Genetics and Biological Response Modifiers on Respiratory Tract Infections. *Frontiers in Immunology*, **2019**, 10, 1013	8.4	10
58	Founder effect of a pathogenic MSH2 mutation identified in Spanish families with Lynch syndrome. *Clinical Genetics*, **2010**, 78, 186-90	4	10
57	Crecimiento Post-traumático en supervivientes de cáncer y sus otros significativos: ¿Crecimiento vicario o secundario?. *Terapia Psicologica*, **2013**, 31, 81-92	1.9	10
56	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
55	High risk of endometrial cancer in colorectal cancer kindred is pathognomonic for MMR-mutation carriers. *Familial Cancer*, **2009**, 8, 145-51	3	9
54	Mutational spectrum by phenotype: panel-based NGS testing of patients with clinical suspicion of RASopathy and children with multiple café-au-lait macules. *Clinical Genetics*, **2020**, 97, 264-275	4	9
53	Early Genetic Diagnosis of Neurofibromatosis Type 2 From Skin Plaque Plexiform Schwannomas in Childhood. *JAMA Dermatology*, **2018**, 154, 341-346	5.1	8
52	Alpha-1-antitrypsin phenotypes and HLA-B27 typing in uveitis patients in southeast Iran. *Clinical Biochemistry*, **2005**, 38, 425-32	3.5	8
51	Comprehensive molecular characterisation of hereditary non-polyposis colorectal tumours with mismatch repair proficiency. *European Journal of Cancer*, **2014**, 50, 1964-72	7.5	7
50	Identification of Plitidepsin as Potent Inhibitor of SARS-CoV-2-Induced Cytopathic Effect after a Drug Repurposing Screen		7
49	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
48	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
47	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

46	Previous SARS-CoV-2 Infection Increases B.1.1.7 Cross-Neutralization by Vaccinated Individuals. *Viruses*, **2021**, 13,	6.2	6
45	Cutaneous neurofibromas: patientsNmedical 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
44	Boosting care and knowledge about hereditary cancer: European Reference Network on Genetic Tumour Risk Syndromes. *Familial Cancer*, **2019**, 18, 281-284	3	6
43	Hydroxychloroquine pre-exposure prophylaxis for COVID-19 in healthcare workers. *Journal of Antimicrobial Chemotherapy*, **2021**, 76, 827-829	5.1	6
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