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	Autoantibodies against type I IFNs in patients with life-threatening COVID-19. Science, 2020, 370, .	12.6	1,983
2	Inborn errors of type I IFN immunity in patients with life-threatening COVID-19. Science, 2020, 370, .	12.6	1,749
3	Exemestane for Breast-Cancer Prevention in Postmenopausal Women. New England Journal of Medicine, 2011, 364, 2381-2391.	27.0	847
4	Peutz-Jeghers syndrome: a systematic review and recommendations for management. Gut, 2010, 59, 975-986.	12.1	635
5	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
6	Guidelines for the clinical management of familial adenomatous polyposis (FAP). Gut, 2008, 57, 704-713.	12.1	591
7	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
8	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
9	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
10	Guidelines for the clinical management of Lynch syndrome (hereditary non-polyposis cancer). Journal of Medical Genetics, 2007, 44, 353-362.	3.2	461
11	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
12	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
13	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
14	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
15	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
16	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
17	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
18	Revised diagnostic criteria for neurofibromatosis type 1 and Legius syndrome: an international consensus recommendation. Genetics in Medicine, 2021, 23, 1506-1513.	2.4	290

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19	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
20	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
21	Polymorphisms in Genes of Nucleotide and Base Excision Repair: Risk and Prognosis of Colorectal Cancer. Clinical Cancer Research, 2006, 12, 2101-2108.	7.0	227
22	Posttraumatic growth in cancer: Reality or illusion?. Clinical Psychology Review, 2009, 29, 24-33.	11.4	222
23	Identification of six new susceptibility loci for invasive epithelial ovarian cancer. Nature Genetics, 2015, 47, 164-171.	21.4	221
24	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
25	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
26	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
27	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
28	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
29	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
30	New insights into POLE and POLD1 germline mutations in familial colorectal cancer and polyposis. Human Molecular Genetics, 2014, 23, 3506-3512.	2.9	135
31	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
32	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
33	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
34	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
35	A large-scale meta-analysis to refine colorectal cancer risk estimates associated with MUTYH variants. British Journal of Cancer, 2010, 103, 1875-1884.	6.4	107
36	Identification of a BRCA2-Specific Modifier Locus at 6p24 Related to Breast Cancer Risk. PLoS Genetics, 2013, 9, e1003173.	3.5	105

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37	Recommendations to improve identification of hereditary and familial colorectal cancer in Europe. Familial Cancer, 2010, 9, 109-115.	1.9	103
38	Common variants in LSP1, 2q35 and 8q24 and breast cancer risk for BRCA1 and BRCA2 mutation carriers. Human Molecular Genetics, 2009, 18, 4442-4456.	2.9	99
39	Identification and molecular characterization of a new ovarian cancer susceptibility locus at 17q21.31. Nature Communications, 2013, 4, 1627.	12.8	98
40	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
41	Germline Mutations in FAN1 Cause Hereditary Colorectal Cancer by Impairing DNA Repair. Gastroenterology, 2015, 149, 563-566.	1.3	94
42	DiagnÃ³stico y tratamiento del dÃ©ficit de alfa-1-antitripsina. Archivos De Bronconeumologia, 2006, 42, 645-659.	0.8	93
43	Interplay between BRCA1 and RHAMM Regulates Epithelial Apicobasal Polarization and May Influence Risk of Breast Cancer. PLoS Biology, 2011, 9, e1001199.	5.6	91
44	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
45	Common Genetic Variants and Modification of Penetrance of BRCA2-Associated Breast Cancer. PLoS Genetics, 2010, 6, e1001183.	3.5	85
46	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
47	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
48	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
49	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
50	Abnormal overexpression of mastocytes in skin biopsies of fibromyalgia patients. Clinical Rheumatology, 2010, 29, 1403-1412.	2.2	75
51	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.	2.5	73
52	Analytical and clinical performance of the panbio COVID-19 antigen-detecting rapid diagnostic test. Journal of Infection, 2021, 82, 186-230.	3.3	73
53	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
54	Low levels of microsatellite instability characterize MLH1 and MSH2 HNPCC carriers before tumor diagnosis. Human Molecular Genetics, 2005, 14, 235-239.	2.9	72

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55	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
56	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
57	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
58	Prevalence of germline MUTYH mutations among Lynch-like syndrome patients. European Journal of Cancer, 2014, 50, 2241-2250.	2.8	66
59	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
60	Survival in women with MMR mutations and ovarian cancer: a multicentre study in Lynch syndrome kindreds. Journal of Medical Genetics, 2010, 47, 99-102.	3.2	61
61	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
62	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
63	Novel Methylation Panel for the Early Detection of Colorectal Tumors in Stool DNA. Clinical Colorectal Cancer, 2010, 9, 168-176.	2.3	59
64	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
65	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
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	Efficacy of alpha1-antitrypsin augmentation therapy in conditions other than pulmonary emphysema. Orphanet Journal of Rare Diseases, 2011, 6, 14.	2.7	54
68	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
69	Conventional renal cancer in a patient with fumarate hydratase mutation. Human Pathology, 2007, 38, 793-796.	2.0	47
70	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
71	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
72	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

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73	Antisense therapeutics for neurofibromatosis type 1 caused by deep intronic mutations. Human Mutation, 2009, 30, 454-462.	2.5	46
74	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
75	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
76	Screening for large rearrangements of the BRCA2 gene in Spanish families with breast/ovarian cancer. Breast Cancer Research and Treatment, 2007, 103, 103-107.	2.5	43
77	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
78	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
79	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
80	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
81	Clinical and genetic characterization of classical forms of familial adenomatous polyposis: a Spanish population study. Annals of Oncology, 2011, 22, 903-909.	1.2	39
82	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
83	Severe alpha-1 antitrypsin deficiency in composite heterozygotes inheriting a new splicing mutation QOMadrid. Respiratory Research, 2014, 15, 125.	3.6	38
84	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
85	MLH1 methylation screening is effective in identifying epimutation carriers. European Journal of Human Genetics, 2012, 20, 1256-1264.	2.8	36
86	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
87	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
88	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
89	Allele-Specific Expression of APC in Adenomatous Polyposis Families. Gastroenterology, 2010, 139, 439-447.e1.	1.3	34
90	Ovarian cancer susceptibility alleles and risk of ovarian cancer inBRCA1andBRCA2mutation carriers. Human Mutation, 2012, 33, 690-702.	2.5	34

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91	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
92	Stem cell-like transcriptional reprogramming mediates metastatic resistance to mTOR inhibition. Oncogene, 2017, 36, 2737-2749.	5.9	34
93	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
94	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
95	Common variants of the BRCA1 wild-type allele modify the risk of breast cancer in BRCA1 mutation carriers. Human Molecular Genetics, 2011, 20, 4732-4747.	2.9	32
96	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
97	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
98	Genome-wide Linkage Scan Reveals Three Putative Breast-Cancer-Susceptibility Loci. American Journal of Human Genetics, 2009, 84, 115-122.	6.2	30
99	Comprehensive functional assessment ofMLH1variants of unknown significance. Human Mutation, 2012, 33, 1576-1588.	2.5	30
100	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
101	Detection of genetic alterations in hereditary colorectal cancer screeningâ˜†. Mutation Research - Fundamental and Molecular Mechanisms of Mutagenesis, 2010, 693, 19-31.	1.0	29
102	MLH1 Founder Mutations with Moderate Penetrance in Spanish Lynch Syndrome Families. Cancer Research, 2010, 70, 7379-7391.	0.9	29
103	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
104	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, 821-827.	2.3	28
105	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, 382-388.	2.2	28
106	Reprogramming Captures the Genetic and Tumorigenic Properties of Neurofibromatosis Type 1 Plexiform Neurofibromas. Stem Cell Reports, 2019, 12, 411-426.	4.8	28
107	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
108	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

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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.	3.5	27
110	Detection of APC Gene Deletions Using Quantitative Multiplex PCR of Short Fluorescent Fragments. Clinical Chemistry, 2008, 54, 1132-1140.	3.2	26
111	No evidence for a genetic modifier for renal cell cancer risk in HLRCC syndrome. Familial Cancer, 2010, 9, 245-251.	1.9	26
112	Role of Engrailed-2 (EN2) as a prostate cancer detection biomarker in genetically high risk men. Scientific Reports, 2013, 3, 2059.	3.3	26
113	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
114	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
115	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
116	Low plasma levels of monocyte chemoattractant protein-1 (MCP-1), tumor necrosis factor-alpha (TNFÎ±), and vascular endothelial growth factor (VEGF) in patients with alpha1-antitrypsin deficiency-related fibromyalgia. Clinical Rheumatology, 2010, 29, 189-197.	2.2	24
117	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
118	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
119	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.	1.7	24
120	Exploring the link between MORF4L1 and risk of breast cancer. Breast Cancer Research, 2011, 13, R40.	5.0	23
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	Uveal Melanoma and BRCA1/BRCA2 Genes: A Relationship That Needs Further Investigation. Journal of Clinical Oncology, 2011, 29, e827-e829.	1.6	22
123	GALNT12is Not a Major Contributor of Familial Colorectal Cancer Type X. Human Mutation, 2014, 35, 50-52.	2.5	22
124	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
125	Telomere Length and Genetic Anticipation in Lynch Syndrome. PLoS ONE, 2013, 8, e61286.	2.5	21
126	Antiidiotypic response against murine monoclonal antibodies reactive with tumor-associated antigen TAG-72. Journal of Clinical Immunology, 1997, 17, 96-106.	3.8	20

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127	Functional Characterization of the Novel APC N1026S Variant Associated With Attenuated Familial Adenomatous Polyposis. Gastroenterology, 2008, 134, 56-64.	1.3	20
128	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
129	Genetic variant in the telomerase gene modifies cancer risk in Lynch syndrome. European Journal of Human Genetics, 2013, 21, 511-516.	2.8	20
130	Usefulness of epithelial cell adhesion molecule expression in the algorithmic approach to Lynch syndrome identification. Human Pathology, 2013, 44, 412-416.	2.0	20
131	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
132	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, 1456-1460.	6.4	19
133	Alpha1-antitrypsin replacement therapy controls fibromyalgia symptoms in 2 patients with PI ZZ alpha1-antitrypsin deficiency. Journal of Rheumatology, 2004, 31, 2082-5.	2.0	19
134	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
135	Nonâ€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, 326-332.	2.8	18
136	Haplotype structure in Ashkenazi Jewish BRCA1 and BRCA2 mutation carriers. Human Genetics, 2011, 130, 685-699.	3.8	18
137	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
138	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.	0.3	17
139	Boosting care and knowledge about hereditary cancer: European Reference Network on Genetic Tumour Risk Syndromes. Familial Cancer, 2019, 18, 281-284.	1.9	17
140	Previous SARS-CoV-2 Infection Increases B.1.1.7 Cross-Neutralization by Vaccinated Individuals. Viruses, 2021, 13, 1135.	3.3	17
141	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
142	Modulation of aberrant NF1 pre-mRNA splicing by kinetin treatment. European Journal of Human Genetics, 2010, 18, 614-617.	2.8	16
143	Functional and Structural Analysis of C-Terminal BRCA1 Missense Variants. PLoS ONE, 2013, 8, e61302.	2.5	16
144	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

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145	Early Genetic Diagnosis of Neurofibromatosis Type 2 From Skin Plaque Plexiform Schwannomas in Childhood. JAMA Dermatology, 2018, 154, 341.	4.1	16
146	Impact of Host Genetics and Biological Response Modifiers on Respiratory Tract Infections. Frontiers in Immunology, 2019, 10, 1013.	4.8	16
147	Colorectal cancer risk and theAPC D1822V variant. International Journal of Cancer, 2004, 112, 161-163.	5.1	15
148	Founder mutation in familial adenomatous polyposis (FAP) in the Balearic Islands. Cancer Genetics and Cytogenetics, 2005, 158, 70-74.	1.0	15
149	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, 1422-1425.	5.1	15
150	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
151	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
152	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
153	Analysis of SLX4/FANCP in non-BRCA1/2-mutated breast cancer families. BMC Cancer, 2012, 12, 84.	2.6	14
154	Exome sequencing identifiesMUTYHmutations in a family with colorectal cancer and an atypical phenotype. Gut, 2015, 64, 355-356.	12.1	14
155	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
156	Gene expression profiling integrated into network modelling reveals heterogeneity in the mechanisms of BRCA1 tumorigenesis. British Journal of Cancer, 2009, 101, 1469-1480.	6.4	13
157	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
158	Longer Telomeres Are Associated with Cancer Risk in MMR-Proficient Hereditary Non-Polyposis Colorectal Cancer. PLoS ONE, 2014, 9, e86063.	2.5	13
159	Evidence for a link between TNFRSF11A and risk of breast cancer. Breast Cancer Research and Treatment, 2011, 129, 947-954.	2.5	12
160	Identification of a founder EPCAM deletion in Spanish Lynch syndrome families. Clinical Genetics, 2014, 85, 260-266.	2.0	12
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