Patricia Ashton-Prolla

List of Publications by Year
in descending order

180	3,362	159585 30	206112 48
papers	citations	h-index	g-index

188	188	188	5219
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	The TP53 mutation, R337H, is associated with Li-Fraumeni and Li-Fraumeni-like syndromes in Brazilian families. Cancer Letters, 2007, 245, 96-102.	7.2	170
2	Detailed haplotype analysis at theTP53locus in p.R337H mutation carriers in the population of Southern Brazil: evidence for a founder effect. Human Mutation, 2010, 31, 143-150.	2.5	116
3	TSC1 and TSC2 gene mutations and their implications for treatment in Tuberous Sclerosis Complex: a review. Genetics and Molecular Biology, 2017, 40, 69-79.	1.3	98
4	Detection of R337H, a germline TP53 mutation predisposing to multiple cancers, in asymptomatic women participating in a breast cancer screening program in Southern Brazil. Cancer Letters, 2008, 261, 21-25.	7.2	94
5	Thalidomide, a current teratogen in South America. Teratology, 1996, 54, 273-277.	1.6	93
6	Tumor protein 53 mutations and inherited cancer: beyond Li-Fraumeni syndrome. Current Opinion in Oncology, 2010, 22, 64-69.	2.4	91
7	Genomic rearrangements in BRCA1 and BRCA2: a literature review. Genetics and Molecular Biology, 2009, 32, 437-446.	1.3	90
8	An alternative protocol for DNA extraction from formalin fixed and paraffin wax embedded tissue. Journal of Clinical Pathology, 2005, 58, 894-895.	2.0	68
9	Recommended Guidelines for Validation, Quality Control, and Reporting of TP53 Variants in Clinical Practice. Cancer Research, 2017, 77, 1250-1260.	0.9	68
10	Highly prevalent TP53 mutation predisposing to many cancers in the Brazilian population: a case for newborn screening?. Lancet Oncology, The, 2009, 10, 920-925.	10.7	67
11	Fabry disease: twenty-two novel mutations in the alpha-galactosidase A gene and genotype/phenotype correlations in severely and mildly affected hemizygotes and heterozygotes. Journal of Investigative Medicine, 2000, 48, 227-35.	1.6	67
12	TP53 PIN3 and MDM2 SNP309 polymorphisms as genetic modifiers in the Li-Fraumeni syndrome: impact on age at first diagnosis. Journal of Medical Genetics, 2009, 46, 766-772.	3.2	64
13	Pregnancy outcome after exposure to misoprostol in Brazil: a prospective, controlled study. Reproductive Toxicology, 1999, 13, 147-151.	2.9	62
14	Impact of Î²1-Adrenergic Receptor Polymorphisms on Susceptibility to Heart Failure, Arrhythmogenesis, Prognosis, and Response to Beta-Blocker Therapy. American Journal of Cardiology, 2008, 102, 726-732.	1.6	62
15	Development and validation of a simple questionnaire for the identification of hereditary breast cancer in primary care. BMC Cancer, 2009, 9, 283.	2.6	61
16	The germline mutational landscape of BRCA1 and BRCA2 in Brazil. Scientific Reports, 2018, 8, 9188.	3.3	61
17	miRNA-21 and miRNA-34a Are Potential Minimally Invasive Biomarkers for the Diagnosis of Pancreatic Ductal Adenocarcinoma. Pancreas, 2016, 45, 84-92.	1.1	56
18	PRIMA-1, a mutant p53 reactivator, induces apoptosis and enhances chemotherapeutic cytotoxicity in pancreatic cancer cell lines. Investigational New Drugs, 2014, 32, 783-794.	2.6	55

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19	BDNF/TrkB Content and Interaction with Gastrin-Releasing Peptide Receptor Blockade in Colorectal Cancer. Oncology, 2010, 79, 430-439.	1.9	50
20	Prevalence of the TP53 p.R337H Mutation in Breast Cancer Patients in Brazil. PLoS ONE, 2014, 9, e99893.	2.5	49
21	Nonâ€“pseudogene-derived complex acid Î²-glucosidase mutations causing mild type 1 and severe type 2 Gaucher disease. Journal of Clinical Investigation, 1999, 103, 817-823.	8.2	46
22	A survey of the clinicopathological and molecular characteristics of patients with suspected Lynch syndrome in Latin America. BMC Cancer, 2017, 17, 623.	2.6	40
23	Liâ€Fraumeni and Liâ€Fraumeniâ€”like syndrome among children diagnosed with pediatric cancer in Southern Brazil. Cancer, 2013, 119, 4341-4349.	4.1	39
24	TP53 PIN3 and PEX4 polymorphisms and infertility associated with endometriosis or with post-in vitro fertilization implantation failure. Cell Death and Disease, 2012, 3, e392-e392.	6.3	37
25	XAF1 as a modifier of p53 function and cancer susceptibility. Science Advances, 2020, 6, eaba3231.	10.3	37
26	Influence of CYP19A1 polymorphisms on the treatment of breast cancer with aromatase inhibitors: a systematic review and meta-analysis. BMC Medicine, 2015, 13, 139.	5.5	36
27	Reviewing the History of HIV-1: Spread of Subtype B in the Americas. PLoS ONE, 2011, 6, e27489.	2.5	34
28	Prevalence of the BRCA1 founder mutation c.5266dupin Brazilian individuals at-risk for the hereditary breast and ovarian cancer syndrome. Hereditary Cancer in Clinical Practice, 2011, 9, 12.	1.5	34
29	Prevalence and impact of founder mutations in hereditary breast cancer in Latin America. Genetics and Molecular Biology, 2014, 37, 234-240.	1.3	34
30	p53 signaling pathway polymorphisms associated to recurrent pregnancy loss. Molecular Biology Reports, 2014, 41, 1871-1877.	2.3	33
31	Prevalence of Hispanic BRCA1 and BRCA2 mutations among hereditary breast and ovarian cancer patients from Brazil reveals differences among Latin American populations. Cancer Genetics, 2016, 209, 417-422.	0.4	33
32	Germline variants in DNA repair genes associated with hereditary breast and ovarian cancer syndrome: analysis of a 21 gene panel in the Brazilian population. BMC Medical Genomics, 2020, 13, 21.	1.5	32
33	Association between myeloperoxidase polymorphisms and its plasma levels with severity of coronary artery disease. Clinical Biochemistry, 2010, 43, 57-62.	1.9	31
34	Characterization of individuals at high risk of developing melanoma in Latin America: bases for genetic counseling in melanoma. Genetics in Medicine, 2016, 18, 727-736.	2.4	31
35	Age at cancer onset in germline TP53 mutation carriers: association with polymorphisms in predicted G-quadruplex structures. Carcinogenesis, 2014, 35, 807-815.	2.8	29
36	BRCA1 and BRCA2 mutational profile and prevalence in hereditary breast and ovarian cancer (HBOC) probands from Southern Brazil: Are international testing criteria appropriate for this specific population?. PLoS ONE, 2017, 12, e0187630.	2.5	29

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37	Nerve conduction studies, electromyography and sympathetic skin response in Fabry's disease. Journal of the Neurological Sciences, 2003, 214, 21-25.	0.6	28
38	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
39	Additive effect of RET polymorphisms on sporadic medullary thyroid carcinoma susceptibility and tumor aggressiveness. European Journal of Endocrinology, 2012, 166, 847-854.	3.7	27
40	Mutation spectrum in South American Lynch syndrome families. Hereditary Cancer in Clinical Practice, 2013, 11, 18.	1.5	26
41	Fabry disease: Comparison of enzymatic, linkage, and mutation analysis for carrier detection in a family with a novel mutation (30delG). , 1999, 84, 420-424.		25
42	Li-Fraumeni-like syndrome associated with a large BRCA1 intragenic deletion. BMC Cancer, 2012, 12, 237.	2.6	25
43	ESR1rs9340799 Is Associated with Endometriosis-Related Infertility andIn VitroFertilization Failure. Disease Markers, 2013, 35, 907-913.	1.3	25
44	Recommendations for Advancing the Diagnosis and Management of Hereditary Breast and Ovarian Cancer in Brazil. JCO Global Oncology, 2020, 6, 439-452.	1.8	25
45	Adherence to a Breast Cancer Screening Program and Its Predictors in Underserved Women in Southern Brazil. Cancer Epidemiology Biomarkers and Prevention, 2010, 19, 2673-2679.	2.5	24
46	Molecular analysis of TSC1 and TSC2 genes and phenotypic correlations in Brazilian families with tuberous sclerosis. PLoS ONE, 2017, 12, e0185713.	2.5	24
47	Germline <scp>MLH</scp>1, <scp>MSH</scp>2 and <scp>MSH</scp>6 variants in Brazilian patients with colorectal cancer and clinical features suggestive of Lynch Syndrome. Cancer Medicine, 2018, 7, 2078-2088.	2.8	23
48	Clinical Characterization and Risk Profile of Individuals Seeking Genetic Counseling for Hereditary Breast Cancer in Brazil. Journal of Genetic Counseling, 2007, 16, 363-371.	1.6	22
49	A TP53 founder mutation, p.R337H, is associated with phyllodes breast tumors in Brazil. Virchows Archiv Fur Pathologische Anatomie Und Physiologie Und Fur Klinische Medizin, 2013, 463, 17-22.	2.8	22
50	Pediatric cancer and Li-Fraumeni/Li-Fraumeni-like syndromes: a review for the pediatrician. Revista Da AssociaÃ§Ã£o MÃ©dica Brasileira, 2015, 61, 282-289.	0.7	22
51	BRCA1 and BRCA2 rearrangements in Brazilian individuals with Hereditary Breast and Ovarian Cancer Syndrome. Genetics and Molecular Biology, 2016, 39, 223-231.	1.3	22
52	The role of genomics in global cancer prevention. Nature Reviews Clinical Oncology, 2021, 18, 116-128.	27.6	22
53	Screening for germline BRCA1, BRCA2, TP53 and CHEK2 mutations in families at-risk for hereditary breast cancer identified in a population-based study from Southern Brazil. Genetics and Molecular Biology, 2016, 39, 210-222.	1.3	21
54	Increased Oxidative Damage in Carriers of the Germline TP53 p.R337H Mutation. PLoS ONE, 2012, 7, e47010.	2.5	21

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55	A model to optimize public health care and downstage breast cancer in limited-resource populations in southern Brazil. (Porto Alegre Breast Health Intervention Cohort). BMC Public Health, 2009, 9, 83.	2.9	20
56	GSTM1, GSTT1, and GSTP1 polymorphisms, breast cancer risk factors and mammographic density in women submitted to breast cancer screening. Revista Brasileira De Epidemiologia, 2012, 15, 246-255.	0.8	20
57	Identification of patients at-risk for Lynch syndrome in a hospital-based colorectal surgery clinic. World Journal of Gastroenterology, 2011, 17, 766.	3.3	20
58	Founder effect of the BRCA1 5382insC mutation in Brazilian patients with hereditary breast ovary cancer syndrome. Cancer Genetics and Cytogenetics, 2008, 184, 62-66.	1.0	19
59	Consistency of self-reported first-degree family history of cancer in a population-based study. Familial Cancer, 2009, 8, 195-202.	1.9	19
60	The CDKN2A p.A148T variant is associated with cutaneous melanoma in Southern Brazil. Experimental Dermatology, 2011, 20, 890-893.	2.9	19
61	High twinning rate in Candido Godoi: a new role for p53 in human fertility. Human Reproduction, 2012, 27, 2866-2871.	0.9	19
62	Rare germline variant (rs78378222) in the TP53 3' UTR: Evidence for a new mechanism of cancer predisposition in Li-Fraumeni syndrome. Cancer Genetics, 2016, 209, 97-106.	0.4	19
63	Genetic Variations in the TP53 Pathway in Native Americans Strongly Suggest Adaptation to the High Altitudes of the Andes. PLoS ONE, 2015, 10, e0137823.	2.5	18
64	Population prevalence of hereditary breast cancer phenotypes and implementation of a genetic cancer risk assessment program in southern Brazil. Genetics and Molecular Biology, 2009, 32, 447-455.	1.3	17
65	Comprehensive germline mutation analysis and clinical profile in a large cohort of Brazilian xeroderma pigmentosum patients. Journal of the European Academy of Dermatology and Venereology, 2020, 34, 2392-2401.	2.4	17
66	Clinical and Molecular Characterization of Patients at Risk for Hereditary Melanoma in Southern Brazil. Journal of Investigative Dermatology, 2008, 128, 421-425.	0.7	16
67	Apolipoprotein E genetic polymorphism, serum lipoprotein levels and breast cancer risk: A case-control study. Molecular and Clinical Oncology, 2014, 2, 1009-1015.	1.0	16
68	Genomic analysis in the clinic: benefits and challenges for health care professionals and patients in Brazil. Journal of Community Genetics, 2015, 6, 275-283.	1.2	16
69	Retinoblastoma in a pediatric oncology reference center in Southern Brazil. BMC Pediatrics, 2016, 16, 48.	1.7	16
70	Frequency of the common germline MUTYH mutations p.G396D and p.Y179C in patients diagnosed with colorectal cancer in Southern Brazil. International Journal of Colorectal Disease, 2011, 26, 841-846.	2.2	15
71	TP53p.R337H is a conditional cancer-predisposing mutation: further evidence from a homozygous patient. BMC Cancer, 2013, 13, 187.	2.6	15
72	Knowledge about breast cancer and hereditary breast cancer among nurses in a public hospital. Revista Latino-Americana De Enfermagem, 2015, 23, 90-97.	1.0	15

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73	Screening for germline mutations in mismatch repair genes in patients with Lynch syndrome by next generation sequencing. Familial Cancer, 2018, 17, 387-394.	1.9	15
74	Association of adipokines and adhesion molecules with indicators of obesity in women undergoing mammography screening. Nutrition and Metabolism, 2012, 9, 97.	3.0	14
75	Costs of genetic testing: Supporting Brazilian Public Policies for the incorporating of molecular diagnostic technologies. Genetics and Molecular Biology, 2015, 38, 332-337.	1.3	14
76	Reviewing the characteristics of BRCA and PALB2-related cancers in the precision medicine era. Genetics and Molecular Biology, 2019, 42, 215-231.	1.3	14
77	From colorectal cancer pattern to the characterization of individuals at risk: Picture for genetic research in Latin America. International Journal of Cancer, 2019, 145, 318-326.	5.1	14
78	Calcium Signaling Alterations Caused by Epigenetic Mechanisms in Pancreatic Cancer: From Early Markers to Prognostic Impact. Cancers, 2020, 12, 1735.	3.7	14
79	The TP53 fertility network. Genetics and Molecular Biology, 2012, 35, 939-946.	1.3	14
80	Cancer Genetic Counseling in Public Health Care Hospitals: The Experience of Three Brazilian Services. Public Health Genomics, 2007, 10, 110-119.	1.0	13
81	Pooling of samples to optimize SARS-CoV-2 diagnosis by RT-qPCR: comparative analysis of two protocols. European Journal of Clinical Microbiology and Infectious Diseases, 2021, 40, 889-892.	2.9	13
82	Genetic epidemiology of BRCA1- and BRCA2-associated cancer across Latin America. Npj Breast Cancer, 2021, 7, 107.	5.2	13
83	HFE gene mutations prevent sustained virological response to interferon plus ribavirin in chronic hepatitis C patients with serum markers of iron overload. American Journal of Gastroenterology, 2002, 97, 1570-1572.	0.4	12
84	CHEK2 1100DELC germline mutation: a frequency study in hereditary breast and colon cancer Brazilian families. Arquivos De Gastroenterologia, 2012, 49, 273-278.	0.8	12
85	A DNA repair variant in POLQ (c.-1060A > G) is associated to hereditary breast cancer patients: a caseâ€“control study. BMC Cancer, 2014, 14, 850.	2.6	12
86	TP53 variants of uncertain significance: increasing challenges in variant interpretation and genetic counseling. Familial Cancer, 2019, 18, 451-456.	1.9	12
87	Performance of the Gail and Tyrer-Cuzick breast cancer risk assessment models in women screened in a primary care setting with the FHS-7 questionnaire. Genetics and Molecular Biology, 2019, 42, 232-237.	1.3	12
88	The Brazilian Hereditary Cancer Network: historical aspects and challenges for clinical cancer genetics in the public health care system in Brazil. Genetics and Molecular Biology, 2016, 39, 163-165.	1.3	12
89	Interaction between TP63 and MDM2 genes and the risk of recurrent pregnancy loss. European Journal of Obstetrics, Gynecology and Reproductive Biology, 2014, 182, 7-10.	1.1	11
90	p53 signaling pathway polymorphisms, cancer risk and tumor phenotype in TP53 R337H mutation carriers. Familial Cancer, 2018, 17, 269-274.	1.9	11

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91	Maternal hyperphenylalaninaemia as a cause of microcephaly and mental retardation. Acta Paediatrica, International Journal of Paediatrics, 1996, 85, 943-946.	1.5	10
92	Clinical and histomolecular endometrial tumor characterization of patients at-risk for Lynch syndrome in South of Brazil. Familial Cancer, 2010, 9, 131-139.	1.9	10
93	TP53 p.Arg337His germline mutation prevalence in Southern Brazil: Further evidence for mutation testing in young breast cancer patients. PLoS ONE, 2018, 13, e0209934.	2.5	10
94	TULP3: A potential biomarker in colorectal cancer?. PLoS ONE, 2019, 14, e0210762.	2.5	10
95	Brazilian health-care policy for targeted oncology therapies and companion diagnostic testing. Lancet Oncology, The, 2016, 17, e363-e370.	10.7	9
96	Information and Diagnosis Networks â€“ tools to improve diagnosis and treatment for patients with rare genetic diseases. Genetics and Molecular Biology, 2019, 42, 155-164.	1.3	9
97	Haplotype analysis of the internationally distributed BRCA1 c.3331_3334delCAAG founder mutation reveals a common ancestral origin in Iberia. Breast Cancer Research, 2020, 22, 108.	5.0	9
98	Skin pigmentation polymorphisms associated with increased risk of melanoma in a case-control sample from southern Brazil. BMC Cancer, 2020, 20, 1069.	2.6	9
99	Haplotypic characterization of BRCA1 c.5266dupC, the prevailing mutation in Brazilian hereditary breast/ovarian cancer. Genetics and Molecular Biology, 2020, 43, e20190072.	1.3	9
100	Effects of imilglucerase withdrawal on an adult with gaucher disease. British Journal of Haematology, 2001, 113, 1088-1089.	2.5	8
101	miRNAs as Diagnostic and Prognostic Biomarkers in Pancreatic Ductal Adenocarcinoma and Its Precursor Lesions: A Review. Biomarker Insights, 2015, 10, BMI.S27679.	2.5	8
102	The breast cancer immunophenotype of TP53-p.R337H carriers is different from that observed among other pathogenic TP53 mutation carriers. Familial Cancer, 2015, 14, 333-336.	1.9	8
103	Comparison of multiple genotyping methods for the identification of the cancer predisposing founder mutation p.R337H in TP53. Genetics and Molecular Biology, 2016, 39, 203-209.	1.3	8
104	Rare germline alterations in cancer-related genes associated with the risk of multiple primary tumor development. Journal of Molecular Medicine, 2017, 95, 523-533.	3.9	8
105	Ancestry of the Brazilian TP53 c.1010G>A (p.Arg337His, R337H) Founder Mutation: Clues from Haplotyping of Short Tandem Repeats on Chromosome 17p. PLoS ONE, 2015, 10, e0143262.	2.5	8
106	Melanoma hereditÃ¡rio: prevalÃªncia de fatores de risco em um grupo de pacientes no Sul do Brasil. Anais Brasileiros De Dermatologia, 2004, 79, 53-60.	1.1	8
107	Say syndrome: A new case with cystic renal dysplasia in discordant monozygotic twins. , 1997, 70, 353-356.		7
108	Vitamin D Status and VDR Genotype in NF1 Patients: A Case-Control Study from Southern Brazil. International Journal of Endocrinology, 2015, 2015, 1-9.	1.5	7

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109	Polymorphisms in CYP19A1 and NFKB1 genes are associated with cutaneous melanoma risk in southern Brazilian patients. Melanoma Research, 2016, 26, 348-353.	1.2	7
110	Systems Biology Approaches Reveal Potential Phenotype-Modifier Genes in Neurofibromatosis Type 1. Cancers, 2020, 12, 2416.	3.7	7
111	The paradox of autophagy in Tuberous Sclerosis Complex. Genetics and Molecular Biology, 2021, 44, e20200014.	1.3	7
112	Prevalence of the STK15 F31I polymorphism and its relationship with mammographic density. Brazilian Journal of Medical and Biological Research, 2011, 44, 291-296.	1.5	6
113	The Brazilian Founder MutationTP53p.R337H is Uncommon in Portuguese Women Diagnosed with Breast Cancer. Breast Journal, 2014, 20, 534-536.	1.0	6
114	Screening and characterization of BRCA2 c.156_157insAlu in Brazil: Results from 1380 individuals from the South and Southeast. Cancer Genetics, 2018, 228-229, 93-97.	0.4	6
115	Cancer-related worry and risk perception in Brazilian individuals seeking genetic counseling for hereditary breast cancer. Genetics and Molecular Biology, 2020, 43, e20190097.	1.3	6
116	Optic nerve enlargement and leukodystrophy: an unusual finding of the infantile form of Krabbe disease. Arquivos De Neuro-Psiquiatria, 2010, 68, 816-818.	0.8	6
117	Prevalence of ERÎ±-397 PvuII C/T, ERÎ±-351 XbaI A/G and PGR PROGINS polymorphisms in Brazilian breast cancer-unaffected women. Brazilian Journal of Medical and Biological Research, 2012, 45, 891-897.	1.5	5
118	MIR605 rs2043556 is associated with the occurrence of multiple primary tumors in TP53 p.(Arg337His) mutation carriers. Cancer Genetics, 2020, 240, 54-58.	0.4	5
119	Prenatal diagnosis of a familial interchromosomal insertion of Y chromosome heterochromatin. , 1997, 73, 470-473.		4
120	The Challenge of Evaluating Adnexal Masses in Patients With Breast Cancer. Clinical Breast Cancer, 2018, 18, e587-e594.	2.4	4
121	Clinical and molecular characterization of patients fulfilling Chompret criteria for Li-Fraumeni syndrome in Southern Brazil. PLoS ONE, 2021, 16, e0251639.	2.5	4
122	The Development of the Study of Hereditary Cancer in South America. Genetics and Molecular Biology, 2016, 39, 166-167.	1.3	4
123	Hereditary cancer syndromes: opportunities and challenges. BMC Proceedings, 2013, 7, K14.	1.6	3
124	Fabry disease: Evidence for a regional founder effect of the GLA gene mutation 30delG in Brazilian patients. Molecular Genetics and Metabolism Reports, 2014, 1, 414-421.	1.1	3
125	Brain Imaging and Genetic Risk in the Pediatric Population, Part 2. Neuroimaging Clinics of North America, 2015, 25, 53-67.	1.0	3
126	Clinical and molecular characterization of neurofibromatosis in southern Brazil. Expert Review of Molecular Diagnostics, 2018, 18, 577-586.	3.1	3

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127	The Role of Co-Deleted Genes in Neurofibromatosis Type 1 Microdeletions: An Evolutive Approach. Genes, 2019, 10, 839.	2.4	3
128	Synchronous Periampullary Tumors in a Patient With Pancreas Divisum and Neurofibromatosis Type 1. Frontiers in Genetics, 2020, 11, 395.	2.3	3
129	Breast cancer screening in 10.000 women of an underserved population in South Brazil: The NMAMAPOA cohort. Journal of Clinical Oncology, 2005, 23, 1020-1020.	1.6	3
130	Effect of HFE gene polymorphism on sustained virological response in patients with chronic hepatitis C and elevated serum ferritin. Arquivos De Gastroenterologia, 2012, 49, 9-13.	0.8	3
131	Brazilian Group of Gastrointestinal Tumours' consensus guidelines for the management of gastric cancer. Ecancermedicalscience, 2020, 14, 1126.	1.1	3
132	Polymorphic variation of mononucleotide microsatellites in healthy humans and its implication for microsatellite instability screening. Arquivos De Gastroenterologia, 2007, 44, 64-67.	0.8	2
133	Genomic Alterations in Patients Showing Multiple Primary Tumors and Family History of Cancer. Annals of Oncology, 2014, 25, iv165.	1.2	2
134	A comprehensive analysis of core polyadenylation sequences and regulation by microRNAs in a set of cancer predisposition genes. Gene, 2019, 712, 143943.	2.2	2
135	Early-Onset Colorectal Cancer in Patients with Li Fraumeni Syndrome: Is It Really Enough to Justify Early Colon Cancer Screening?. Gastroenterology, 2019, 157, 264.	1.3	2
136	Tuberous Sclerosis Complex with rare associated findings in the gastrointestinal system: a case report and review of the literature. BMC Gastroenterology, 2020, 20, 394.	2.0	2
137	Prevalence of the Brazilian TP53 Founder c.1010G>A (p.Arg337His) in Lung Adenocarcinoma: Is Genotyping Warranted in All Brazilian Patients?. Frontiers in Genetics, 2021, 12, 606537.	2.3	2
138	Prevalence of BRCA1 and BRCA2 founder mutations in Brazilian hereditary breast and ovarian cancer families. Journal of Clinical Oncology, 2008, 26, 22108-22108.	1.6	2
139	Hereditary non-polipomatous colorectal cancer: hereditary predisposition, diagnosis and prevention. Arquivos De Gastroenterologia, 2005, 42, 99-106.	0.8	2
140	Pineal region hemangioblastoma in a patient with Von Hippel-Lindau disease. Arquivos De Neuro-Psiquiatria, 2011, 69, 988-988.	0.8	2
141	Response to â€œGermline TP53 R337H mutation is not sufficient to establish Li-Fraumeni or Li-Fraumeni-like syndromeâ€, by Ribeiro et al.. Cancer Letters, 2007, 247, 356-358.	7.2	1
142	Genetic information and biobanking: a Brazilian perspective on biological and biographical issues. Journal of Community Genetics, 2015, 6, 295-299.	1.2	1
143	Brazilian Group of Gastrointestinal Tumoursâ€™ consensus guidelines for the management of oesophageal cancer. Ecancermedicalscience, 2021, 15, 1195.	1.1	1
144	Functional Polymorphisms in the p53 Pathway Genes on the Genetic Susceptibility to Zika Virus Teratogenesis. Frontiers in Cellular and Infection Microbiology, 2021, 11, 641413.	3.9	1

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145	Genetic counseling and cancer risk perception in Brazilian patients at-risk for hereditary breast and ovarian cancer. Journal of Clinical Oncology, 2004, 22, 9698-9698.	1.6	1
146	Reviewing the occurrence of large genomic rearrangements in patients with inherited cancer predisposing syndromes: importance of a comprehensive molecular diagnosis. Expert Review of Molecular Diagnostics, 2022, 22, 319-346.	3.1	1
147	High prevalence of gastrointestinal symptoms in Fabry's disease. Gastroenterology, 1998, 114, A369.	1.3	0
148	H63D and C282Y HFE mutations prevent sustained virologic response to interferon plus ribavirin in chronic hepatitis C patients with serum markers of iron overload. Journal of Hepatology, 2002, 36, 232.	3.7	0
149	Role of Î²1-Adrenergic Receptor Polymorphism on Risk of Complex Ventricular Arrhythmias in Patients with Heart Failure: Potential Pharmacogenetic Interactions. Journal of Cardiac Failure, 2006, 12, S39.	1.7	0
150	Microsatellite Instability Testing in Genetically Heterogeneous Populations. Journal of Clinical Oncology, 2007, 25, 913-914.	1.6	0
151	Neurophysiological studies in Fabry disease. Acta Paediatrica, International Journal of Paediatrics, 2007, 91, 125-125.	1.5	0
152	Chitotriosidase: is it a useful biochemical marker for the assessment of Fabry patients treated by enzyme replacement therapy?. Acta Paediatrica, International Journal of Paediatrics, 2007, 91, 152-153.	1.5	0
153	577 Pharmacological Reactivation of Mutant p53 by PRIMA-1 Induces Apoptosis and Enhances Chemotherapeutic Cytotoxicity in Pancreatic Cancer Cells. European Journal of Cancer, 2012, 48, 177.	2.8	0
154	P-0231 Occurrence of 2152 C>T â€“MHS2 Mutation in Brazilian Lynch Syndrome Families. Annals of Oncology, 2012, 23, iv96.	1.2	0
155	Genetic Counseling for TP53 Germline Mutations. , 2013, , 327-343.		0
156	Commentary regarding Schayek et al., entitled â€œThe rate of recurrent BRCA1, BRCA2, and TP53 mutations in the general population, and unselected ovarian cancer cases, in Belo Horizonte, Brazilâ€• Cancer Genetics, 2016, 209, 282-283.	0.4	0
157	Primary cells derived from Tuberous Sclerosis Complex patients show autophagy alteration in the haploinsufficiency state. Genetics and Molecular Biology, 2021, 44, e20200475.	1.3	0
158	Genetic counseling and cancer risk perception in Brazilian patients at-risk for hereditary breast and ovarian cancer. Journal of Clinical Oncology, 2004, 22, 9698-9698.	1.6	0
159	Prevalence of breast cancer risk factors in two different cohorts in relation to health care access and preventive programs in southern Brazil. Journal of Clinical Oncology, 2007, 25, 1538-1538.	1.6	0
160	Detection of R337H, a germline mutation predisposing to multiple cancers, in asymptomatic women participating in a breast cancer screening programme in southern Brazil. Journal of Clinical Oncology, 2007, 25, 21029-21029.	1.6	0
161	Screening for germline mutations in families at-risk for hereditary breast cancer in Southern Brazil. Journal of Clinical Oncology, 2008, 26, 22128-22128.	1.6	0
162	Development and validation of a questionnaire for the identification of hereditary breast cancer in primary care. Journal of Clinical Oncology, 2008, 26, 22094-22094.	1.6	0

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163	Abstract P3-06-02: Prevalence ofTP53p.R337H Mutation in Cases of Breast Phyllodes Tumours in Southern Brazil. , 2010, , .		0
164	Abstract 1837: Clinical diversity and tumor spectrum in Xeroderma Pigmentosum Brazilian patients. , 2011, , .		0
165	P1-09-07: Contribution of TP53 p.R337H Mutation to Breast Cancer Incidence in Brazil.. , 2011, , .		0
166	Presence of familial colorectal cancer type X in families fulfilling Amsterdam criteria for Lynch syndrome in southern Brazil.. Journal of Clinical Oncology, 2013, 31, e12546-e12546.	1.6	0
167	Abstract P2-07-04: HER2 overexpressing breast cancers are more frequent in carriers of TP53 DNA-binding domain mutations than in carriers of oligomerization-site mutations. , 2013, , .		0
168	Abstract P2-13-01: The Brazilian founder TP53 p.R337H mutation is uncommon in Portuguese women diagnosed with breast cancer. , 2013, , .		0
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