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	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
2	The role of genomics in global cancer prevention. Nature Reviews Clinical Oncology, 2021, 18, 116-128.	27.6	22
3	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
4	The paradox of autophagy in Tuberous Sclerosis Complex. Genetics and Molecular Biology, 2021, 44, e20200014.	1.3	7
5	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
6	Brazilian Group of Gastrointestinal Tumoursâ€™ consensus guidelines for the management of oesophageal cancer. Ecancermedicalscience, 2021, 15, 1195.	1.1	1
7	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
8	Genetic epidemiology of BRCA1- and BRCA2-associated cancer across Latin America. Npj Breast Cancer, 2021, 7, 107.	5.2	13
9	Clinical and molecular characterization of patients fulfilling Chompret criteria for Li-Fraumeni syndrome in Southern Brazil. PLoS ONE, 2021, 16, e0251639.	2.5	4
10	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
11	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
12	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
13	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
14	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
15	Systems Biology Approaches Reveal Potential Phenotype-Modifier Genes in Neurofibromatosis Type 1. Cancers, 2020, 12, 2416.	3.7	7
16	Synchronous Periampullary Tumors in a Patient With Pancreas Divisum and Neurofibromatosis Type 1. Frontiers in Genetics, 2020, 11, 395.	2.3	3
17	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
18	Calcium Signaling Alterations Caused by Epigenetic Mechanisms in Pancreatic Cancer: From Early Markers to Prognostic Impact. Cancers, 2020, 12, 1735.	3.7	14

#	Article	IF	Citations
19	XAF1 as a modifier of p53 function and cancer susceptibility. Science Advances, 2020, 6, eaba3231.	10.3	37
20	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
21	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
22	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
23	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
24	Brazilian Group of Gastrointestinal Tumours' consensus guidelines for the management of gastric cancer. Ecancermedicalscience, 2020, 14, 1126.	1.1	3
25	TP53 variants of uncertain significance: increasing challenges in variant interpretation and genetic counseling. Familial Cancer, 2019, 18, 451-456.	1.9	12
26	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
27	The Role of Co-Deleted Genes in Neurofibromatosis Type 1 Microdeletions: An Evolutive Approach. Genes, 2019, 10, 839.	2.4	3
28	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
29	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
30	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
31	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
32	TULP3: A potential biomarker in colorectal cancer?. PLoS ONE, 2019, 14, e0210762.	2.5	10
33	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
34	Molecular profiling as predictor of outcomes in a Brazilian cohort of stage IV lung cancer.. Journal of Clinical Oncology, 2019, 37, e20668-e20668.	1.6	0
35	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
36	Clinical and molecular characterization of neurofibromatosis in southern Brazil. Expert Review of Molecular Diagnostics, 2018, 18, 577-586.	3.1	3

#	Article	IF	Citations
37	The Challenge of Evaluating Adnexal Masses in Patients With Breast Cancer. Clinical Breast Cancer, 2018, 18, e587-e594.	2.4	4
38	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
39	p53 signaling pathway polymorphisms, cancer risk and tumor phenotype in TP53 R337H mutation carriers. Familial Cancer, 2018, 17, 269-274.	1.9	11
40	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
41	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
42	The germline mutational landscape of BRCA1 and BRCA2 in Brazil. Scientific Reports, 2018, 8, 9188.	3.3	61
43	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
44	Recommended Guidelines for Validation, Quality Control, and Reporting of TP53 Variants in Clinical Practice. Cancer Research, 2017, 77, 1250-1260.	0.9	68
45	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
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	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
48	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
49	Genetic counseling to outpatient cancer population.. Journal of Clinical Oncology, 2017, 35, e13009-e13009.	1.6	0
50	Abstract 4282: GermlineTP53p.R337H mutations and Li-Fraumeni syndrome: A new variant form of the disease. , 2017, , .		0
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	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
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	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

#	Article	IF	Citations
55	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
56	Brazilian health-care policy for targeted oncology therapies and companion diagnostic testing. Lancet Oncology, The, 2016, 17, e363-e370.	10.7	9
57	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
58	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
59	Retinoblastoma in a pediatric oncology reference center in Southern Brazil. BMC Pediatrics, 2016, 16, 48.	1.7	16
60	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
61	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
62	The Development of the Study of Hereditary Cancer in South America. Genetics and Molecular Biology, 2016, 39, 166-167.	1.3	4
63	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
64	Abstract P2-09-12: Prevalence of HispanicBRCA1andBRCA2mutations among HBOC patients from Southern Brazil reveal differences among Latin American populations. , 2016, , .		0
65	Abstract P1-07-19: Spatial analyses of breast cancer in women 15-49 years-old in Rio Grande do Sul, southern Brazil. , 2016, , .		0
66	Germline mutational spectrum of Brazilian HBOC patients tested with hereditary cancer multigene panels.. Journal of Clinical Oncology, 2016, 34, e13113-e13113.	1.6	0
67	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
68	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
69	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
70	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
71	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
72	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

#	Article	IF	Citations
73	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
74	Genetic information and biobanking: a Brazilian perspective on biological and biographical issues. Journal of Community Genetics, 2015, 6, 295-299.	1.2	1
75	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
76	Brain Imaging and Genetic Risk in the Pediatric Population, Part 2. Neuroimaging Clinics of North America, 2015, 25, 53-67.	1.0	3
77	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
78	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
79	Prevalence of BRCA1 and BRCA2 mutations in patients fulfilling HBOC criteria in South Brazil.. Journal of Clinical Oncology, 2015, 33, e12529-e12529.	1.6	0
80	Abstract 804: Energetic metabolism and DNA damage response in fibroblasts from Li-Fraumeni syndrome patients: new insights into the molecular mechanisms of the disease. , 2015, , .		0
81	Genomic Alterations in Patients Showing Multiple Primary Tumors and Family History of Cancer. Annals of Oncology, 2014, 25, iv165.	1.2	2
82	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
83	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
84	p53 signaling pathway polymorphisms associated to recurrent pregnancy loss. Molecular Biology Reports, 2014, 41, 1871-1877.	2.3	33
85	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
86	The Brazilian Founder MutationTP53p.R337H is Uncommon in Portuguese Women Diagnosed with Breast Cancer. Breast Journal, 2014, 20, 534-536.	1.0	6
87	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
88	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
89	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
90	Prevalence of the TP53 p.R337H Mutation in Breast Cancer Patients in Brazil. PLoS ONE, 2014, 9, e99893.	2.5	49

#	Article	IF	Citations
91	Prevalence and impact of founder mutations in hereditary breast cancer in Latin America. Genetics and Molecular Biology, 2014, 37, 234-240.	1.3	34
92	Evaluation of the knowledge about breast cancer in Brazilian women seeking assistance in basic health care units.. Journal of Clinical Oncology, 2014, 32, e20573-e20573.	1.6	0
93	Identification of a rare germ-line variant in the TP53 3â€™UTR in individuals with the Li-Fraumeni-like phenotype: A new mechanism of cancer predisposition?. Journal of Clinical Oncology, 2014, 32, 11106-11106.	1.6	0
94	microRNA expression in circulating samples of pancreatic ductal adenocarcinoma patients: A search for circulating biomarkers.. Journal of Clinical Oncology, 2014, 32, e22111-e22111.	1.6	0
95	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
96	TP53p.R337H is a conditional cancer-predisposing mutation: further evidence from a homozygous patient. BMC Cancer, 2013, 13, 187.	2.6	15
97	Hereditary cancer syndromes: opportunities and challenges. BMC Proceedings, 2013, 7, K14.	1.6	3
98	Liâ€Fraumeni and Liâ€Fraumeniâ€”like syndrome among children diagnosed with pediatric cancer in Southern Brazil. Cancer, 2013, 119, 4341-4349.	4.1	39
99	Genetic Counseling for TP53 Germline Mutations. , 2013, , 327-343.		0
100	ESR1rs9340799 Is Associated with Endometriosis-Related Infertility andIn VitroFertilization Failure. Disease Markers, 2013, 35, 907-913.	1.3	25
101	Mutation spectrum in South American Lynch syndrome families. Hereditary Cancer in Clinical Practice, 2013, 11, 18.	1.5	26
102	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
103	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
104	Abstract P2-13-01: The Brazilian founder TP53 p.R337H mutation is uncommon in Portuguese women diagnosed with breast cancer. , 2013, , .		0
105	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
106	High twinning rate in Candido Godoi: a new role for p53 in human fertility. Human Reproduction, 2012, 27, 2866-2871.	0.9	19
107	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
108	Li-Fraumeni-like syndrome associated with a large BRCA1 intragenic deletion. BMC Cancer, 2012, 12, 237.	2.6	25

#	Article	IF	Citations
109	Association of adipokines and adhesion molecules with indicators of obesity in women undergoing mammography screening. Nutrition and Metabolism, 2012, 9, 97.	3.0	14
110	P-0231 Occurrence of 2152 C>T â€“MHS2 Mutation in Brazilian Lynch Syndrome Families. Annals of Oncology, 2012, 23, iv96.	1.2	0
111	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
112	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
113	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
114	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
115	Increased Oxidative Damage in Carriers of the Germline TP53 p.R337H Mutation. PLoS ONE, 2012, 7, e47010.	2.5	21
116	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
117	The TP53 fertility network. Genetics and Molecular Biology, 2012, 35, 939-946.	1.3	14
118	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
119	Reviewing the History of HIV-1: Spread of Subtype B in the Americas. PLoS ONE, 2011, 6, e27489.	2.5	34
120	The CDKN2A p.A148T variant is associated with cutaneous melanoma in Southern Brazil. Experimental Dermatology, 2011, 20, 890-893.	2.9	19
121	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
122	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
123	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
124	Pineal region hemangioblastoma in a patient with Von Hippel-Lindau disease. Arquivos De Neuro-Psiquiatria, 2011, 69, 988-988.	0.8	2
125	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
126	Abstract 1837: Clinical diversity and tumor spectrum in Xeroderma Pigmentosum Brazilian patients. , 2011, , .		0

#	Article	IF	Citations
127	P1-09-07: Contribution of TP53 p.R337H Mutation to Breast Cancer Incidence in Brazil.. , 2011, , .		0
128	Tumor protein 53 mutations and inherited cancer: beyond Li-Fraumeni syndrome. Current Opinion in Oncology, 2010, 22, 64-69.	2.4	91
129	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
130	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
131	Association between myeloperoxidase polymorphisms and its plasma levels with severity of coronary artery disease. Clinical Biochemistry, 2010, 43, 57-62.	1.9	31
132	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
133	BDNF/TrkB Content and Interaction with Gastrin-Releasing Peptide Receptor Blockade in Colorectal Cancer. Oncology, 2010, 79, 430-439.	1.9	50
134	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
135	Abstract P3-06-02: Prevalence ofTP53p.R337H Mutation in Cases of Breast Phyllodes Tumours in Southern Brazil. , 2010, , .		0
136	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
137	Genomic rearrangements in BRCA1 and BRCA2: a literature review. Genetics and Molecular Biology, 2009, 32, 437-446.	1.3	90
138	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
139	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
140	Consistency of self-reported first-degree family history of cancer in a population-based study. Familial Cancer, 2009, 8, 195-202.	1.9	19
141	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
142	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
143	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
144	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

#	Article	IF	Citations
145	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
146	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
147	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
148	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
149	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
150	Microsatellite Instability Testing in Genetically Heterogeneous Populations. Journal of Clinical Oncology, 2007, 25, 913-914.	1.6	0
151	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
152	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
153	Cancer Genetic Counseling in Public Health Care Hospitals: The Experience of Three Brazilian Services. Public Health Genomics, 2007, 10, 110-119.	1.0	13
154	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
155	Neurophysiological studies in Fabry disease. Acta Paediatrica, International Journal of Paediatrics, 2007, 91, 125-125.	1.5	0
156	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
157	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
158	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
159	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
160	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
161	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
162	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

#	Article	IF	Citations
163	Hereditary non-polipomatous colorectal cancer: hereditary predisposition, diagnosis and prevention. Arquivos De Gastroenterologia, 2005, 42, 99-106.	0.8	2
164	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
165	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
166	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
167	Nerve conduction studies, electromyography and sympathetic skin response in Fabry's disease. Journal of the Neurological Sciences, 2003, 214, 21-25.	0.6	28
168	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
169	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
170	Effects of imilglucerase withdrawal on an adult with gaucher disease. British Journal of Haematology, 2001, 113, 1088-1089.	2.5	8
171	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
172	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
173	Pregnancy outcome after exposure to misoprostol in Brazil: a prospective, controlled study. Reproductive Toxicology, 1999, 13, 147-151.	2.9	62
174	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
175	High prevalence of gastrointestinal symptoms in Fabry's disease. Gastroenterology, 1998, 114, A369.	1.3	0
176	Say syndrome: A new case with cystic renal dysplasia in discordant monozygotic twins. , 1997, 70, 353-356.		7
177	Prenatal diagnosis of a familial interchromosomal insertion of Y chromosome heterochromatin. , 1997, 73, 470-473.		4
178	Say syndrome: a new case with cystic renal dysplasia in discordant monozygotic twins. American Journal of Medical Genetics Part A, 1997, 70, 353-6.	2.4	0
179	Maternal hyperphenylalaninaemia as a cause of microcephaly and mental retardation. Acta Paediatrica, International Journal of Paediatrics, 1996, 85, 943-946.	1.5	10
180	Thalidomide, a current teratogen in South America. Teratology, 1996, 54, 273-277.	1.6	93