Maria Isabel Achatz

List of Publications by Year in descending order

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137 papers

4,323 citations

30 h-index 62 g-index

142 all docs

 $\begin{array}{c} 142 \\ \\ \text{docs citations} \end{array}$

times ranked

142

6646 citing authors

#	Article	IF	CITATIONS
1	Utility of interim blood tests for cancer screening in Li-Fraumeni syndrome. Familial Cancer, 2022, 21, 333-336.	0.9	1
2	Breast Cancer Phenotype Associated With Li-Fraumeni Syndrome: A Brazilian Cohort Enriched by TP53 p.R337H Carriers. Frontiers in Oncology, 2022, 12, 836937.	1.3	8
3	Cancer surveillance for patients with Li-Fraumeni Syndrome in Brazil: A cost-effectiveness analysis. The Lancet Regional Health Americas, 2022, 12, 100265.	1.5	1
4	Complete Clinical Response in Stage IVB Endometrioid Endometrial Carcinoma after First-Line Pembrolizumab Therapy: Report of a Case with Isolated Loss of PMS2 Protein. Case Reports in Oncology, 2021, 13, 1067-1074.	0.3	5
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.	1.1	2
6	Germline molecular data in hereditary breast cancer in Brazil: Lessons from a large single-center analysis. PLoS ONE, 2021, 16, e0247363.	1.1	14
7	Identification of the <i>TP53</i> p.R337H Variant in Tumor Genomic Profiling Should Prompt Consideration of Germline Testing for Li-Fraumeni Syndrome. JCO Global Oncology, 2021, 7, 1141-1150.	0.8	7
8	Expanding the phenotype of E318K (c.952G > A) MITF germline mutation carriers: case series and review of the literature. Hereditary Cancer in Clinical Practice, 2021, 19, 32.	0.6	1
9	Inherited Pediatric Cancer in Low- and Intermediate-Resource Countries. , 2021, , 361-386.		0
10	Novel Insights From the Germline Landscape of Breast Cancer in Brazil. Frontiers in Oncology, 2021, 11, 743231.	1.3	1
11	Family Health Leaders: Lessons on Living with Liâ€Fraumeni Syndrome across Generations. Family Process, 2020, 59, 1648-1663.	1.4	10
12	MIR605 rs2043556 is associated with the occurrence of multiple primary tumors in TP53 p.(Arg337His) mutation carriers. Cancer Genetics, 2020, 240, 54-58.	0.2	5
13	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.	2.2	9
14	CN4 Duty to recontact patients previously tested with negative results in a hereditary cancer syndrome center. Annals of Oncology, 2020, 31, S1123-S1124.	0.6	0
15	Suggested application of HER2+ breast tumor phenotype for germline <i>TP53</i> variant classification within ACMG/AMP guidelines. Human Mutation, 2020, 41, 1555-1562.	1.1	16
16	Li-Fraumeni Exploration Consortium Data Coordinating Center: Building an Interactive Web-Based Resource for Collaborative International Cancer Epidemiology Research for a Rare Condition. Cancer Epidemiology Biomarkers and Prevention, 2020, 29, 927-935.	1.1	7
17	Recommendations for Advancing the Diagnosis and Management of Hereditary Breast and Ovarian Cancer in Brazil. JCO Global Oncology, 2020, 6, 439-452.	0.8	25
18	XAF1 as a modifier of p53 function and cancer susceptibility. Science Advances, 2020, 6, eaba3231.	4.7	37

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19	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.	1.3	17
20	Abstract P6-08-18: Breast cancer in Li-Fraumeni syndrome and risk-reduction mastectomy inTP53p.R337H carriers. , 2020, , .		1
21	Germline Mutation in MUS81 Resulting in Impaired Protein Stability is Associated with Familial Breast and Thyroid Cancer. Cancers, 2020, 12, 1289.	1.7	3
22	Pheochromocytoma and paraganglioma: implications of germline mutation investigation for treatment, screening, and surveillance. Archives of Endocrinology and Metabolism, 2019, 63, 369-375.	0.3	8
23	Couples coping with screening burden and diagnostic uncertainty in Li-Fraumeni syndrome: Connection versus independence. Journal of Psychosocial Oncology, 2019, 37, 178-193.	0.6	21
24	Reproductive factors associated with breast cancer risk in Li–Fraumeni syndrome. European Journal of Cancer, 2019, 116, 199-206.	1.3	10
25	Early-Onset Colorectal Cancer in Patients with Li Fraumeni Syndrome: Is It Really Enough to Justify Early Colon Cancer Screening?. Gastroenterology, 2019, 157, 264.	0.6	2
26	Genomic profiling in ovarian cancer retreated with platinum based chemotherapy presented homologous recombination deficiency and copy number imbalances of CCNE1 and RB1 genes. BMC Cancer, 2019, 19, 422.	1.1	22
27	Response to: Concern regarding classification of germline TP53 variants as likely pathogenic. Human Mutation, 2019, 40, 832-833.	1.1	1
28	Variable population prevalence estimates of germline <i>TP53</i> variants: A gnomAD-based analysis. Human Mutation, 2019, 40, 97-105.	1.1	66
29	Abstract 3316: Screening blood tests and cancer detection in Li-Fraumeni syndrome. , 2019, , .		O
30	Testing Positive on a Multigene Panel Does Not Suffice to Determine Disease Risks. Journal of the National Cancer Institute, 2018, 110, 797-798.	3.0	4
31	Germline <i><scp>MLH</scp>1, <scp>MSH</scp>2</i> and <i><scp>MSH</scp>6</i> variants in Brazilian patients with colorectal cancer and clinical features suggestive of Lynch Syndrome. Cancer Medicine, 2018, 7, 2078-2088.	1.3	23
32	Revisiting tumor patterns and penetrance in germline TP53 mutation carriers: temporal phases of Liâ€"Fraumeni syndrome. Current Opinion in Oncology, 2018, 30, 23-29.	1.1	129
33	Screening for germline mutations in mismatch repair genes in patients with Lynch syndrome by next generation sequencing. Familial Cancer, 2018, 17, 387-394.	0.9	15
34	p53 signaling pathway polymorphisms, cancer risk and tumor phenotype in TP53 R337H mutation carriers. Familial Cancer, 2018, 17, 269-274.	0.9	11
35	Prevalence of pathogenic/likely pathogenic variants in the 24 cancer genes of the ACMG Secondary Findings v2.0 list in a large cancer cohort and ethnicity-matched controls. Genome Medicine, 2018, 10, 99.	3.6	15
36	Whole-body magnetic resonance imaging of Li-Fraumeni syndrome patients: observations from a two rounds screening of Brazilian patients. Cancer Imaging, 2018, 18, 27.	1.2	19

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37	Mouse Homolog of the Human <i>TP53</i> R337H Mutation Reveals Its Role in Tumorigenesis. Cancer Research, 2018, 78, 5375-5383.	0.4	24
38	Complex Landscape of Germline Variants in Brazilian Patients With Hereditary and Early Onset Breast Cancer. Frontiers in Genetics, 2018, 9, 161.	1.1	21
39	The germline mutational landscape of BRCA1 and BRCA2 in Brazil. Scientific Reports, 2018, 8, 9188.	1.6	61
40	Abstract A20: Identification of new promising germline variants in melanoma-prone patients. , 2018, , .		0
41	Abstract A37: Complex landscape of germline variants in hereditary and early-onset breast cancer ascertained through whole exome sequencing. , 2018, , .		2
42	Effect of breastfeeding on the risk of breast cancer in Li-Fraumeni syndrome Journal of Clinical Oncology, 2018, 36, 1530-1530.	0.8	1
43	Abstract 1247: Hereditary paraganglioma-pheochromocytoma syndrome: Patterns of presentation in a Brazilian oncogenetics clinic. , 2018, , .		0
44	Frequency of Thyroid Carcinoma in Brazilian <i>TP53 </i> p.R337H Carriers With Li Fraumeni Syndrome. JAMA Oncology, 2017, 3, 1400.	3.4	21
45	Rare germline alterations in cancer-related genes associated with the risk of multiple primary tumor development. Journal of Molecular Medicine, 2017, 95, 523-533.	1.7	8
46	Germline large genomic alterations on 7q in patients with multiple primary cancers. Scientific Reports, 2017, 7, 41677.	1.6	2
47	<i>PTEN, DICER1, FH</i> , and Their Associated Tumor Susceptibility Syndromes: Clinical Features, Genetics, and Surveillance Recommendations in Childhood. Clinical Cancer Research, 2017, 23, e76-e82.	3.2	128
48	Cancer Screening Recommendations for Individuals with Li-Fraumeni Syndrome. Clinical Cancer Research, 2017, 23, e38-e45.	3.2	358
49	Clinical Management and Tumor Surveillance Recommendations of Inherited Mismatch Repair Deficiency in Childhood. Clinical Cancer Research, 2017, 23, e32-e37.	3.2	157
50	Higher-than-expected population prevalence of potentially pathogenic germline <i>TP53</i> variants in individuals unselected for cancer history. Human Mutation, 2017, 38, 1723-1730.	1.1	40
51	Baseline Surveillance in Li-Fraumeni Syndrome Using Whole-Body Magnetic Resonance Imaging. JAMA Oncology, 2017, 3, 1634.	3.4	148
52	Cancer Screening Recommendations and Clinical Management of Inherited Gastrointestinal Cancer Syndromes in Childhood. Clinical Cancer Research, 2017, 23, e107-e114.	3.2	91
53	Molecular analysis of TSC1 and TSC2 genes and phenotypic correlations in Brazilian families with tuberous sclerosis. PLoS ONE, 2017, 12, e0185713.	1.1	24
54	<i>HABP2</i> p.G534E variant in patients with family history of thyroid and breast cancer. Oncotarget, 2017, 8, 40896-40905.	0.8	7

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55	Homologous recombination deficiency and platinum rechallenge in platinum-resistant ovarian cancer patients Journal of Clinical Oncology, 2017, 35, 5576-5576.	0.8	1
56	Prevalence of germline <i>TP53</i> p.R337H mutation in Brazilian young breast cancer patients Journal of Clinical Oncology, 2017, 35, e13101-e13101.	0.8	0
57	Abstract 4287:HABP2p.G534E variant in patients with family history of thyroid and breast cancer. , 2017,		0
58	Abstract 4282: GermlineTP53p.R337H mutations and Li-Fraumeni syndrome: A new variant form of the disease. , 2017, , .		0
59	Abstract NG05: TP53-mediated human cancer susceptibility is defined by epigenetic dysregulation of microRNA-34A. , 2017, , .		0
60	Oral and maxillofacial considerations in Gardner's syndrome: a report of two cases. Ecancermedicalscience, 2016, 10, 623.	0.6	14
61	Genomic profile of Li-Fraumeni syndrome patients with adrenocortical carcinoma in childhood. Annals of Oncology, 2016, 27, vi11.	0.6	0
62	Early-onset breast cancer patients in the South and Southeast of Brazil should be tested for the TP53 p.R337H mutation. Genetics and Molecular Biology, 2016, 39, 199-202.	0.6	15
63	Role of rare germline copy number variation in melanoma-prone patients. Future Oncology, 2016, 12, 1345-1357.	1.1	8
64	Commentary regarding Schayek et al., entitled "The rate of recurrent BRCA1, BRCA2, and TP53 mutations in the general population, and unselected ovarian cancer cases, in Belo Horizonte, Brazil― Cancer Genetics, 2016, 209, 282-283.	0.2	0
65	The Inherited p53 Mutation in the Brazilian Population. Cold Spring Harbor Perspectives in Medicine, 2016, 6, a026195.	2.9	60
66	Contribution of rare germline copy number variations and common susceptibility loci in Lynch syndrome patients negative for mutations in the mismatch repair genes. International Journal of Cancer, 2016, 138, 1928-1935.	2.3	21
67	Genome-Wide DNA Methylation Analysis Reveals Epigenetic Dysregulation of MicroRNA-34A in <i>TP53</i> -Associated Cancer Susceptibility. Journal of Clinical Oncology, 2016, 34, 3697-3704.	0.8	33
68	Brazilian health-care policy for targeted oncology therapies and companion diagnostic testing. Lancet Oncology, The, 2016, 17, e363-e370.	5.1	9
69	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.	0.2	19
70	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.	1.1	31
71	ROBO1 deletion as a novel germline alteration in breast and colorectal cancer patients. Tumor Biology, 2016, 37, 3145-3153.	0.8	9
72	A Set of miRNAs, Their Gene and Protein Targets and Stromal Genes Distinguish Early from Late Onset ER Positive Breast Cancer. PLoS ONE, 2016, 11, e0154325.	1.1	7

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73	The Value of Secondary Cytoreductive Surgery in Recurrent Ovarian Cancer and Application of a Prognostic Score. International Journal of Gynecological Cancer, 2016, 26, 449-55.	1.2	12
74	3311 Germline mutations and rare copy number variations in melanoma-prone patients. European Journal of Cancer, 2015, 51, S668.	1.3	0
75	DNA methylation patterns of candidate genes regulated by thymine DNA glycosylase in patients with TP53 germline mutations. Brazilian Journal of Medical and Biological Research, 2015, 48, 610-615.	0.7	6
76	DNA Methylation Levels of Melanoma Risk Genes Are Associated with Clinical Characteristics of Melanoma Patients. BioMed Research International, 2015, 2015, 1-8.	0.9	17
77	Neoadjuvant Chemotherapy Followed by Interval Debulking Surgery and the Risk of Platinum Resistance in Epithelial Ovarian Cancer. Annals of Surgical Oncology, 2015, 22, 971-978.	0.7	38
78	Li-Fraumeni Ontology: A Case Study of an Ontology for Knowledge Discovery in a Cancer Domain. , 2015, , .		1
79	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.	0.9	8
80	Molecular Characterization of Choroid Plexus Tumors Reveals Novel Clinically Relevant Subgroups. Clinical Cancer Research, 2015, 21, 184-192.	3.2	84
81	18F-FDG PET-CT for Surveillance of Brazilian Patients with Li-Fraumeni Syndrome. Frontiers in Oncology, 2015, 5, 38.	1.3	13
82	Xeroderma Pigmentosum: Low Prevalence of Germline XPA Mutations in a Brazilian XP Population. International Journal of Molecular Sciences, 2015, 16, 8988-8996.	1.8	9
83	Genomic profile of a Li-Fraumeni-like syndrome patient with a 45,X/46,XX karyotype, presenting neither mutations in TP53 nor clinical stigmata of Turner syndrome. Cancer Genetics, 2015, 208, 341-344.	0.2	5
84	LINE-1 hypermethylation in peripheral blood of cutaneous melanoma patients is associated with metastasis. Melanoma Research, 2015, 25, 173-177.	0.6	20
85	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.	1.1	8
86	Evaluation of rapid whole-body magnetic resonance as screening strategy for early cancer detection in 57 Brazilian Li-Fraumeni syndrome patients Journal of Clinical Oncology, 2015, 33, 1534-1534.	0.8	2
87	Molecular and clinical profile of Li-Fraumeni Syndrome in a Brazilian cohort Journal of Clinical Oncology, 2015, 33, e12533-e12533.	0.8	0
88	Abstract 2751: Identification of predisposition genes involved in thyroid and breast carcinomas in patients with family history of these tumors by whole exome sequencing. , 2015, , .		0
89	Abstract 2753: The germline TP53 p.R337H mutation: a putative selective advantage. , 2015, , .		0
90	Genomic Alterations in Patients Showing Multiple Primary Tumors and Family History of Cancer. Annals of Oncology, 2014, 25, iv165.	0.6	2

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91	Genome-wide DNA methylation profile of leukocytes from melanoma patients with and without CDKN2A mutations. Experimental and Molecular Pathology, 2014, 97, 425-432.	0.9	5
92	Germline CDKN2A mutations in Brazilian patients of hereditary cutaneous melanoma. Familial Cancer, 2014, 13, 645-649.	0.9	18
93	Association of <i>TP53</i> polymorphisms on the risk of Wilms tumor. Pediatric Blood and Cancer, 2014, 61, 436-441.	0.8	23
94	Whole-genome sequencing analysis of phenotypic heterogeneity and anticipation in Li–Fraumeni cancer predisposition syndrome. Proceedings of the National Academy of Sciences of the United States of America, 2014, 111, 15497-15501.	3.3	29
95	Does germ-line deletion of the PIP gene constitute a widespread risk for cancer?. European Journal of Human Genetics, 2014, 22, 307-309.	1.4	4
96	Hereditary breast and ovarian cancer: assessment of point mutations and copy number variations in Brazilian patients. BMC Medical Genetics, 2014, 15, 55.	2.1	57
97	The profile and contribution of rare germline copy number variants to cancer risk in Li-Fraumeni patients negative for TP53 mutations. Orphanet Journal of Rare Diseases, 2014, 9, 63.	1.2	12
98	Age at cancer onset in germline TP53 mutation carriers: association with polymorphisms in predicted G-quadruplex structures. Carcinogenesis, 2014, 35, 807-815.	1.3	29
99	Primary versus interval debulking surgery and the risk to induce platinum resitance Journal of Clinical Oncology, 2014, 32, 5588-5588.	0.8	3
100	Prevalence of the TP53 p.R337H Mutation in Breast Cancer Patients in Brazil. PLoS ONE, 2014, 9, e99893.	1.1	49
101	Identification of a rare germ-line variant in the <i>TP53</i> 3'UTR in individuals with the Li-Fraumeni-like phenotype: A new mechanism of cancer predisposition?. Journal of Clinical Oncology, 2014, 32, 11106-11106.	0.8	0
102	Abstract 3418: Rare germline copy number variations in hereditary cutaneous melanoma. , 2014, , .		0
103	Mutational spectrum of the APC and MUTYH genes and genotype–phenotype correlations in Brazilian FAP, AFAP, and MAP patients. Orphanet Journal of Rare Diseases, 2013, 8, 54.	1.2	35
104	Integration of Genomics in Cancer Care. Journal of Nursing Scholarship, 2013, 45, 43-51.	1.1	20
105	Germline BAX Deletion in a Patient With Melanoma and Gastrointestinal Stromal Tumor. American Journal of Gastroenterology, 2013, 108, 1372-1375.	0.2	3
106	Comprehensive Analysis of BRCA1, BRCA2 and TP53 Germline Mutation and Tumor Characterization: A Portrait of Early-Onset Breast Cancer in Brazil. PLoS ONE, 2013, 8, e57581.	1.1	70
107	Molecular Analyses of Early-Onset Gastric Cancer in Brazilian Patients: <i>TP</i> 53 Mutations, Cadherin-Catenin and Mucins Proteins Expression. Journal of Cancer Therapy, 2013, 04, 33-42.	0.1	1
108	Keratocystic odontogenic tumor related to nevoid basal cell carcinoma syndrome: clinicopathological study. Brazilian Journal of Oral Sciences, 2013, 12, 23-29.	0.1	2

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109	Abstract 3771: Establishment of primary cultures of cancer stem cells from patients with Li-Fraumeni Syndrome, carrier of TP53 p.R337H Brazilian founder mutation, 2013, , .		О
110	Abstract A025: Screening for genomic rearrangements and germline mutations in BRCA1 and BRCA2 genes in hereditary breast cancer unrelated Brazilian families. , 2013, , .		0
111	Li-Fraumeni-like syndrome associated with a large BRCA1 intragenic deletion. BMC Cancer, 2012, 12, 237.	1.1	25
112	Number of rare germline CNVs and TP53 mutation types. Orphanet Journal of Rare Diseases, 2012, 7, 101.	1.2	9
113	CHEK2 1100DELC germline mutation: a frequency study in hereditary breast and colon cancer Brazilian families. Arquivos De Gastroenterologia, 2012, 49, 273-278.	0.3	12
114	Germline DNA copy number variation in familial and early-onset breast cancer. Breast Cancer Research, 2012, 14, R24.	2.2	76
115	Abstract 5349: Cancer stem cells isolation and characterization from breast tumor of a germline carrier of TP53 p.R337H (Brazilian founder) mutation. , 2012, , .		0
116	Gastric cancer in individuals with Li-Fraumeni syndrome. Genetics in Medicine, 2011, 13, 651-657.	1.1	118
117	International distribution and age estimation of the Portuguese BRCA2 c.156_157insAlu founder mutation. Breast Cancer Research and Treatment, 2011, 127, 671-679.	1.1	27
118	TP53 mutation p.R337H in gastric cancer tissues of a 12-year-old male child - evidence for chimerism involving a common mutant founder haplotype: case report. BMC Cancer, 2011, 11, 449.	1.1	34
119	Abstract 1836: Distinct tumor spectrum and age of onset in a Brazilian cohort of p.R337HTP53mutation carriers. , 2011, , .		О
120	Abstract 5598: Germline TP53 mutation in very early onset breast cancer patients without BRCA1 and BRCA2 mutation in Brazilian population. , 2011, , .		0
121	Abstract 1837: Clinical diversity and tumor spectrum in Xeroderma Pigmentosum Brazilian patients. , 2011, , .		О
122	Abstract LB-329: Copy number variations associated with hereditary breast and colorectal carcinomas. , 2011, , .		0
123	Abstract B10: Germline copy number variation in Li-Fraumeni syndrome patients with TP53 mutations. , 2011, , .		O
124	Abstract A24: Germline deletion at $3p12.3$ in patients with hereditary breast and colorectal carcinoma. , $2011, , .$		0
125	P1-09-07: Contribution of TP53 p.R337H Mutation to Breast Cancer Incidence in Brazil, 2011, , .		0
126	Tumor protein 53 mutations and inherited cancer: beyond Li-Fraumeni syndrome. Current Opinion in Oncology, 2010, 22, 64-69.	1.1	91

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127	A Common Molecular Mechanism Underlies Two Phenotypically Distinct 17p13.1 Microdeletion Syndromes. American Journal of Human Genetics, 2010, 87, 631-642.	2.6	36
128	Detailed haplotype analysis at the <i>TP53</i> locus in p.R337H mutation carriers in the population of Southern Brazil: evidence for a founder effect. Human Mutation, 2010, 31, 143-150.	1.1	116
129	Abstract 3038: Determination of microRNA expression profile in tumors from young women with breast cancer. , 2010, , .		O
130	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.	1.5	64
131	Highly prevalent TP53 mutation predisposing to many cancers in the Brazilian population: a case for newborn screening?. Lancet Oncology, The, 2009, 10, 920-925.	5.1	67
132	Differential Gene Expression Profiles in Breast Cancer Tumors from Young and Post-Menopausal Patients, 2009,,.		0
133	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.	3.2	94
134	The TP53 mutation, R337H, is associated with Li-Fraumeni and Li-Fraumeni-like syndromes in Brazilian families. Cancer Letters, 2007, 245, 96-102.	3.2	170
135	Response to "Germline 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.	3.2	1
136	Brain tumors in individuals with familial adenomatous polyposis. Cancer, 2007, 109, 761-766.	2.0	88
137	TP53 mutations in human cancers: functional selection and impact on cancer prognosis and outcomes. Oncogene, 2007, 26, 2157-2165.	2.6	796