Thierry FrÃ©bourg

List of Publications by Year
in descending order

106 papers	10,274 citations	57758 44 h-index	33894 99 g-index
114 all docs	114 docs citations	114 times ranked	14626 citing authors

#	Article	IF	Citations
1	APP locus duplication causes autosomal dominant early-onset Alzheimer disease with cerebral amyloid angiopathy. Nature Genetics, 2006, 38, 24-26.	21.4	1,087
2	Cancer Risks Associated With Germline Mutations in <emph type="ital">MLH1</emph>, <emph type="ital">MSH2</emph>, and <emph type="ital">MSH6</emph> Genes in Lynch Syndrome. JAMA - Journal of the American Medical Association, 2011, 305, 2304.	7.4	878
3	Somatic and germline activating mutations of the ALK kinase receptor in neuroblastoma. Nature, 2008, 455, 967-970.	27.8	787
4	Early-Onset Autosomal Dominant Alzheimer Disease: Prevalence, Genetic Heterogeneity, and Mutation Spectrum. American Journal of Human Genetics, 1999, 65, 664-670.	6.2	696
5	Revisiting Li-Fraumeni Syndrome From TP53 Mutation Carriers. Journal of Clinical Oncology, 2015, 33, 2345-2352.	1.6	525
6	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
7	Recurrent Rearrangements in Synaptic and Neurodevelopmental Genes and Shared Biologic Pathways in Schizophrenia, Autism, and Mental Retardation. Archives of General Psychiatry, 2009, 66, 947.	12.3	374
8	Cancer Screening Recommendations for Individuals with Li-Fraumeni Syndrome. Clinical Cancer Research, 2017, 23, e38-e45.	7.0	358
9	2009 Version of the Chompret Criteria for Li Fraumeni Syndrome. Journal of Clinical Oncology, 2009, 27, e108-e109.	1.6	291
10	Germline Mutations of the Pairedâ€“Like Homeobox 2B (PHOX2B) Gene in Neuroblastoma. American Journal of Human Genetics, 2004, 74, 761-764.	6.2	288
11	Segregation of a Missense Mutation in the Microtubule-Associated Protein Tau Gene with Familial Frontotemporal Dementia and Parkinsonism. Human Molecular Genetics, 1998, 7, 1825-1829.	2.9	258
12	Screening patients for heterozygous p53 mutations using a functional assay in yeast. Nature Genetics, 1993, 5, 124-129.	21.4	243
13	Next-generation sequencing for the diagnosis of hereditary breast and ovarian cancer using genomic capture targeting multiple candidate genes. European Journal of Human Genetics, 2014, 22, 1305-1313.	2.8	217
14	Radio-induced malignancies after breast cancer postoperative radiotherapy in patients with Li-Fraumeni syndrome. Radiation Oncology, 2010, 5, 104.	2.7	168
15	Guidelines for the Liâ€“Fraumeni and heritable TP53-related cancer syndromes. European Journal of Human Genetics, 2020, 28, 1379-1386.	2.8	167
16	Rapid detection of novelBRCA1 rearrangements in high-risk breast-ovarian cancer families using multiplex PCR of short fluorescent fragments. Human Mutation, 2002, 20, 218-226.	2.5	161
17	Two metachronous tumors in the radiotherapy fields of a patient with Li-Fraumeni syndrome. International Journal of Cancer, 2001, 96, 238-242.	5.1	138
18	Exonic Splicing Mutations Are More Prevalent than Currently Estimated and Can Be Predicted by Using In Silico Tools. PLoS Genetics, 2016, 12, e1005756.	3.5	133

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19	Serum hyaluronate in liver diseases: Study by enzymoimmunological assay. Hepatology, 1986, 6, 392-395.	7.3	132
20	Genetic Association Studies of Suicidal Behavior: A Review of the Past 10â€‰Years, Progress, Limitations, and Future Directions. Frontiers in Psychiatry, 2016, 7, 158.	2.6	122
21	A rapid PCR fidelity assay. Nucleic Acids Research, 1994, 22, 3259-3260.	14.5	111
22	Cytoskeleton proteins are modulators of mutant tau-induced neurodegeneration in Drosophila. Human Molecular Genetics, 2007, 16, 555-566.	2.9	107
23	Significant Contribution of Germline BRCA2 Rearrangements in Male Breast Cancer Families. Cancer Research, 2004, 64, 8143-8147.	0.9	94
24	Early-Onset Brain Tumor and Lymphoma in MSH2-Deficient Children. American Journal of Human Genetics, 2003, 72, 213-216.	6.2	88
25	MSH2 in contrast to MLH1 and MSH6 is frequently inactivated by exonic and promoter rearrangements in hereditary nonpolyposis colorectal cancer. Cancer Research, 2002, 62, 848-53.	0.9	86
26	Drosophila models of human tauopathies indicate that Tau protein toxicity in vivo is mediated by soluble cytosolic phosphorylated forms of the protein. Journal of Neurochemistry, 2010, 113, 895-903.	3.9	83
27	TP63 gene mutation in ADULT syndrome. European Journal of Human Genetics, 2001, 9, 642-645.	2.8	82
28	Contribution of de novo and mosaic TP53 mutations to Li-Fraumeni syndrome. Journal of Medical Genetics, 2018, 55, 173-180.	3.2	78
29	Diagnosis of Constitutional Mismatch Repair-Deficiency Syndrome Based on Microsatellite Instability and Lymphocyte Tolerance to Methylating Agents. Gastroenterology, 2015, 149, 1017-1029.e3.	1.3	76
30	Detecting splicing patterns in genes involved in hereditary breast and ovarian cancer. European Journal of Human Genetics, 2017, 25, 1147-1154.	2.8	76
31	Identification of human p53 mutations with differential effects on the bax and p21 promoters using functional assays in yeast. Oncogene, 1998, 16, 1369-1372.	5.9	75
32	Screening for TP53 rearrangements in families with the Liâ€“Fraumeni syndrome reveals a complete deletion of the TP53 gene. Oncogene, 2003, 22, 840-846.	5.9	72
33	Polymorphisms of insulin degrading enzyme gene are not associated with Alzheimer's disease. Neuroscience Letters, 2002, 329, 121-123.	2.1	68
34	Deletion of the progranulin gene in patients with frontotemporal lobar degeneration or Parkinson disease. Neurobiology of Disease, 2008, 31, 41-45.	4.4	66
35	Plasmid vectors with multiple cloning sites and cat-reporter gene for promoter cloning and analysis in animal cells. Gene, 1988, 65, 315-318.	2.2	64
36	ABCA7 rare variants and Alzheimer disease risk. Neurology, 2016, 86, 2134-2137.	1.1	63

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37	Heterogeneity ofNSD1alterations in 116 patients with Sotos syndrome. Human Mutation, 2007, 28, 1098-1107.	2.5	62
38	Serum hyaluronate in malignant pleural mesothelioma. Cancer, 1987, 59, 2104-2107.	4.1	61
39	Clinical value of chip-based digital-PCR platform for the detection of circulating DNA in metastatic colorectal cancer. Digestive and Liver Disease, 2015, 47, 884-890.	0.9	59
40	High frequency in esophageal cancers of p53 alterations inactivating the regulation of genes involved in cell cycle and apoptosis. Carcinogenesis, 2000, 21, 563-565.	2.8	55
41	Accumulation of insoluble forms of FUS protein correlates with toxicity in Drosophila. Neurobiology of Aging, 2012, 33, 1008.e1-1008.e15.	3.1	52
42	The Rappâ€“Hodgkin syndrome results from mutations of the TP63 gene. European Journal of Human Genetics, 2003, 11, 700-704.	2.8	48
43	Complete germline deletion of the STK11 gene in a family with Peutzâ€“Jeghers syndrome. European Journal of Human Genetics, 2004, 12, 415-418.	2.8	48
44	Structure and expression of the Xenopus retinoblastoma gene. Developmental Biology, 1992, 153, 141-149.	2.0	47
45	The Challenge for the Next Generation of Medical Geneticists. Human Mutation, 2014, 35, 909-911.	2.5	47
46	Frontotemporal Dementia Phenotype Associated with MAPT Gene Duplication. Journal of Alzheimer's Disease, 2010, 21, 897-902.	2.6	46
47	Drastic Effect of GermlineTP53Missense Mutations in Li-Fraumeni Patients. Human Mutation, 2013, 34, 453-461.	2.5	46
48	Severe myoclonus-dystonia syndrome associated with a novel epsilon-sarcoglycan gene truncating mutation. , 2003, 119B, 114-117.		45
49	Tau is not normally degraded by the proteasome. Journal of Neuroscience Research, 2005, 80, 400-405.	2.9	43
50	Partial deletion of theMAPTgene: A novel mechanism of FTDP-17. Human Mutation, 2009, 30, E591-E602.	2.5	37
51	No evidence for involvement of KCNN3 (hSKCa3) potassium channel gene in familial and isolated cases of schizophrenia. European Journal of Human Genetics, 1999, 7, 247-250.	2.8	32
52	A sensitive assay for measuring SMN mRNA levels in peripheral blood and in muscle samples of patients affected with spinal muscular atrophy. European Journal of Human Genetics, 2007, 15, 1054-1062.	2.8	32
53	Contribution of genotoxic anticancer treatments to the development of multiple primary tumours in the context of germline TP53 mutations. European Journal of Cancer, 2018, 101, 254-262.	2.8	32
54	The 5â€² region of theMSH2gene involved in hereditary non-polyposis colorectal cancer contains a high density of recombinogenic sequences. Human Mutation, 2005, 26, 255-261.	2.5	31

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55	Germline TP53 mutations result into a constitutive defect of p53 DNA binding and transcriptional response to DNA damage. Human Molecular Genetics, 2017, 26, 2591-2602.	2.9	29
56	Largeâ€scale comparative evaluation of userâ€friendly tools for predicting variantâ€induced alterations of splicing regulatory elements. Human Mutation, 2020, 41, 1811-1829.	2.5	29
57	High Prevalence of Somatic Oncogenic Driver Alterations in Patients With NSCLC and Li-Fraumeni Syndrome. Journal of Thoracic Oncology, 2020, 15, 1232-1239.	1.1	29
58	Detection of a large genomic deletion in the pancreatic secretory trypsin inhibitor (SPINK1) gene. European Journal of Human Genetics, 2006, 14, 1204-1208.	2.8	28
59	Filamin-A and Myosin VI colocalize with fibrillary Tau protein in Alzheimer's disease and FTDP-17 brains. Brain Research, 2010, 1345, 182-189.	2.2	28
60	Evaluation of Lynch syndrome modifier genes in 748 MMR mutation carriers. European Journal of Human Genetics, 2011, 19, 887-892.	2.8	27
61	Detection of APC Gene Deletions Using Quantitative Multiplex PCR of Short Fluorescent Fragments. Clinical Chemistry, 2008, 54, 1132-1140.	3.2	26
62	Fraser syndrome: features suggestive of prenatal diagnosis in a review of 38 cases. Prenatal Diagnosis, 2016, 36, 1270-1275.	2.3	25
63	Diversity of genetic events associated with MLH1 promoter methylation in Lynch syndrome families with heritable constitutional epimutation. Genetics in Medicine, 2018, 20, 1589-1599.	2.4	25
64	Variable expressivity of the clinical and biochemical phenotype associated with the apolipoprotein E p.Leu149del mutation. European Journal of Human Genetics, 2005, 13, 1186-1191.	2.8	24
65	Lung Adenocarcinoma as Part of the Li-Fraumeni Syndrome Spectrum. JAMA Oncology, 2017, 3, 1736.	7.1	24
66	Inhibition of proteasome and Shaggy/Glycogen synthase kinase-3Î² kinase prevents clearance of phosphorylated tau inDrosophila. Journal of Neuroscience Research, 2006, 84, 1107-1115.	2.9	23
67	Gradual reduction of BUBR1 protein levels results in premature sister-chromatid separation then in aneuploidy. Human Genetics, 2008, 124, 473-478.	3.8	23
68	Detection of copy-number variations from NGS data using read depth information: a diagnostic performance evaluation. European Journal of Human Genetics, 2021, 29, 99-109.	2.8	23
69	The MDM2 285Gâ€“309G haplotype is associated with an earlier age of tumour onset in patients with Li-Fraumeni syndrome. Familial Cancer, 2014, 13, 127-130.	1.9	22
70	GermlineCDKN2A/P16INK4A mutations contribute to genetic determinism of sarcoma. Journal of Medical Genetics, 2017, 54, 607-612.	3.2	19
71	An Information-Theoretic Analysis of Genetics, Gender and Age in Cancer Patients. PLoS ONE, 2008, 3, e1951.	2.5	18
72	Diversity of the clinical presentation of the MMR gene biallelic mutations. Familial Cancer, 2014, 13, 131-135.	1.9	18

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73	Hyperprolinemia is not associated with childhood onset schizophrenia. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2006, 141B, 192-192.	1.7	17
74	Splicing factors act as genetic modulators of TDP-43 production in a new autoregulatory TDP-43 Drosophila model. Human Molecular Genetics, 2017, 26, 3396-3408.	2.9	17
75	Early-onset low-grade papillary carcinoma of the bladder associated with Apert syndrome and a germlineFGFR2 mutation (Pro253Arg). American Journal of Medical Genetics, Part A, 2006, 140A, 2245-2247.	1.2	16
76	Germline TP53 Testing in Breast Cancers: Why, When and How?. Cancers, 2020, 12, 3762.	3.7	16
77	A risk for early-onset Alzheimer's disease associated with the APBB1 gene (FE65) intron 13 polymorphism. Neuroscience Letters, 2003, 342, 5-8.	2.1	14
78	Partial duplications of the MSH2 and MLH1 genes in hereditary nonpolyposis colorectal cancer. European Journal of Human Genetics, 2007, 15, 383-386.	2.8	14
79	Pitfalls in the use of DGV for CNV interpretation. American Journal of Medical Genetics, Part A, 2011, 155, 2593-2596.	1.2	14
80	Neuron-to-Neuron Transfer of FUS in Drosophila Primary Neuronal Culture Is Enhanced by ALS-Associated Mutations. Journal of Molecular Neuroscience, 2017, 62, 114-122.	2.3	14
81	Optimization of the diagnosis of inherited colorectal cancer using NGS and capture of exonic and intronic sequences of panel genes. European Journal of Human Genetics, 2018, 26, 1597-1602.	2.8	12
82	Value of microsatellite instability typing in detecting hereditary non-polyposis colorectal cancer. Gastroenterologie Clinique Et Biologique, 2005, 29, 667-675.	0.9	11
83	Germline Mutations of Inhibins in Earlyâ€Onset Ovarian Epithelial Tumors. Human Mutation, 2014, 35, 294-297.	2.5	11
84	A Connected Network of Interacting Proteins Is Involved in Human-Tau Toxicity in Drosophila. Frontiers in Neuroscience, 2020, 14, 68.	2.8	11
85	No replication of the association between the Nicastrin gene and familial early-onset Alzheimer's disease. Neuroscience Letters, 2003, 353, 153-155.	2.1	10
86	Identification of TCERG1 as a new genetic modulator of TDP-43 production in Drosophila. Acta Neuropathologica Communications, 2018, 6, 138.	5.2	10
87	Prognostic value of TP53 transcriptional activity on p21 and bax in patients with esophageal squamous cell carcinomas treated by definitive chemoradiotherapy. International Journal of Radiation Oncology Biology Physics, 2002, 54, 379-385.	0.8	9
88	Duodenal adenocarcinoma and Mut Y human homologue-associated polyposis. European Journal of Gastroenterology and Hepatology, 2008, 20, 1024-1027.	1.6	9
89	Reply to Talseth-Palmer et al. European Journal of Human Genetics, 2012, 20, 488-488.	2.8	9
90	Familial solitary chondrosarcoma resulting from germline EXT2 mutation. Genes Chromosomes and Cancer, 2017, 56, 128-134.	2.8	7

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91	Blood functional assay for rapid clinical interpretation of germline TP53 variants. Journal of Medical Genetics, 2021, 58, 796-805.	3.2	7
92	Further delineation of the <scp>NTHL1</scp> associated syndrome: A report from the French Oncogenetic Consortium. Clinical Genetics, 2021, 99, 662-672.	2.0	7
93	Direct-to-consumer genetic testing services: what are the medical benefits?. European Journal of Human Genetics, 2012, 20, 483-483.	2.8	5
94	Gestational choriocarcinoma associated with a germline TP53 mutation. Familial Cancer, 2018, 17, 113-117.	1.9	5
95	A remarkable APC mosaicism with two mutant alleles in a family with familial adenomatous polyposis. American Journal of Medical Genetics, Part A, 2011, 155, 1500-1502.	1.2	4
96	Transmission of germlineTP53mutations from male carriers to female partners. Journal of Medical Genetics, 2015, 52, 145-146.	3.2	4
97	Mosaic PTEN alteration in the neural crest during embryogenesis results in multiple nervous system hamartomas. Acta Neuropathologica Communications, 2019, 7, 191.	5.2	4
98	Reply to Kratz et al.. European Journal of Human Genetics, 2020, 28, 1483-1485.	2.8	4
99	Molecular cloning of flounder Xp18, a newly identified highly conserved protein mainly expressed in the ovary. Gene, 2003, 307, 13-21.	2.2	3
100	Patients with 10q22.3q23.1 recurrent deletion syndrome are at risk for juvenile polyposis. European Journal of Medical Genetics, 2020, 63, 103773.	1.3	3
101	Cost-effectiveness evaluation of pre-counseling telephone interviews before face-to-face genetic counseling in cancer genetics. Familial Cancer, 2018, 17, 451-457.	1.9	1
102	Moderate Overexpression of Tau in Drosophila Exacerbates Amyloid-Î²-Induced Neuronal Phenotypes and Correlates with Tau Oligomerization. Journal of Alzheimer's Disease, 2020, 74, 637-647.	2.6	1
103	Li-Fraumeni Syndrome. , 2021, , 1-21.		0
104	Syndrome de Li-Fraumeni. , 2009, , 257-269.		0
105	Abstract C5: Worse prognosis ofKRASc.35 G > A mutant metastatic colorectal cancer (MCRC) patients treated with intensive triplet chemotherapy plus bevacizumab (FIr-B/FOx).. , 2011, , .		0
106	Generation of 17q21.31 duplication iPSC-derived neurons as a model for primary tauopathies. Stem Cell Research, 2022, 61, 102762.	0.7	0