List of Publications by Year in descending order

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#	Article	IF	CITATIONS
1	Identification of three clinical neurofibromatosis 1 subtypes: Latent class analysis of a series of 1351 patients. Journal of the European Academy of Dermatology and Venereology, 2022, 36, 739-743.	2.4	0
2	Severe Phenotype in Patients with Large Deletions of NF1. Cancers, 2021, 13, 2963.	3.7	15
3	NF1-like optic pathway gliomas in children: clinical and molecular characterization of this specific presentation. Neuro-Oncology Advances, 2020, 2, i98-i106.	0.7	4
4	One NF1 Mutation may Conceal Another. Genes, 2019, 10, 633.	2.4	5
5	<i>NF1</i> mutations identify molecular and clinical subtypes of lung adenocarcinomas. Cancer Medicine, 2019, 8, 4330-4337.	2.8	14
6	Non-invasive prenatal diagnosis of paternally inherited disorders from maternal plasma: detection of <i>NF1</i> and <i>CFTR</i> mutations using droplet digital PCR. Clinical Chemistry and Laboratory Medicine, 2018, 56, 728-738.	2.3	26
7	Targeted next-generation sequencing for differential diagnosis of neurofibromatosis type 2, schwannomatosis, and meningiomatosis. Neuro-Oncology, 2018, 20, 917-929.	1.2	48
8	Confirmation of mutation landscape of NF1â€associated malignant peripheral nerve sheath tumors. Genes Chromosomes and Cancer, 2017, 56, 421-426.	2.8	54
9	Moyamoya syndrome in children with neurofibromatosis type 1: Italian–French experience. American Journal of Medical Genetics, Part A, 2017, 173, 1521-1530.	1.2	36
10	Droplet digital PCR, a new approach to analyze fetal DNA from maternal blood: application to the determination of fetal RHD genotype. Annales De Biologie Clinique, 2016, 74, 269-277.	0.1	5
11	<i>SETD2</i> and <i>DNMT3A</i> screen in the Sotos-like syndrome French cohort. Journal of Medical Genetics, 2016, 53, 743-751.	3.2	54
12	Neurofibromatosis Type 1 Molecular Diagnosis: The RNA Point of View. EBioMedicine, 2016, 7, 21-22.	6.1	3
13	Copy number variants and rasopathies: germline KRAS duplication in a patient with syndrome including pigmentation abnormalities. Orphanet Journal of Rare Diseases, 2016, 11, 101.	2.7	7
14	NF1 single and multi-exons copy number variations in neurofibromatosis type 1. Journal of Human Genetics, 2015, 60, 221-224.	2.3	15
15	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
16	Juvenile myelomonocytic leukemia displays mutations in components of the RAS pathway and the PRC2 network. Nature Genetics, 2015, 47, 1334-1340.	21.4	152
17	Neurofibromatosis type 1 molecular diagnosis: what can NGS do for you when you have a large gene with loss of function mutations?. European Journal of Human Genetics, 2015, 23, 596-601.	2.8	97
18	RAS MAPK inhibitors deregulation in leukemia. Oncoscience, 2015, 2, 930-931.	2.2	4

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19	The Activation of the WNT Signaling Pathway Is a Hallmark in Neurofibromatosis Type 1 Tumorigenesis. Clinical Cancer Research, 2014, 20, 358-371.	7.0	44
20	MicroRNAome profiling in benign and malignant neurofibromatosis type 1-associated nerve sheath tumors: evidences of PTEN pathway alterations in early NF1 tumorigenesis. BMC Genomics, 2013, 14, 473.	2.8	46
21	Relevance of MPNST cell lines as models for NF1 associated-tumors. Journal of Neuro-Oncology, 2013, 114, 353-355.	2.9	5
22	<i>NF1</i> Molecular Characterization and Neurofibromatosis Type I Genotype-Phenotype Correlation: The French Experience. Human Mutation, 2013, 34, 1510-1518.	2.5	140
23	Sporadic NF1 mutation associated with a de-novo 20q11.3 deletion explains the association of unusual facies, Moyamoya vasculopathy, and developmental delay, reported by Bertoli et al. in 2009. Clinical Dysmorphology, 2013, 22, 42-43.	0.3	8
24	Somatic NF1 inactivation is a frequent event in sporadic pheochromocytoma. Human Molecular Genetics, 2012, 21, 5397-5405.	2.9	126
25	First description of ABCB4 gene deletions in familial low phospholipid-associated cholelithiasis and oral contraceptives-induced cholestasis. European Journal of Human Genetics, 2012, 20, 277-282.	2.8	42
26	Neurofibromatosis type 1: from genotype to phenotype. Journal of Medical Genetics, 2012, 49, 483-489.	3.2	133
27	IL28B polymorphism is associated with treatment response in patients with genotype 4 chronic hepatitis C. Journal of Hepatology, 2012, 56, 527-532.	3.7	165
28	Two independent de novo mutations as a cause for neurofibromatosis type 1 and Noonan syndrome in a single family. American Journal of Medical Genetics, Part A, 2012, 158A, 2290-2291.	1.2	4
29	Modifier Genes in NF1. , 2012, , 269-285.		1
30	SOX9 expression increases with malignant potential in tumors from patients with neurofibromatosis 1 and is not correlated to desert hedgehog. Human Pathology, 2011, 42, 434-443.	2.0	10
31	Identification of Genes Potentially Involved in the Increased Risk of Malignancy in NF1-Microdeleted Patients. Molecular Medicine, 2011, 17, 79-87.	4.4	46
32	Role of Noncoding RNA ANRIL in Genesis of Plexiform Neurofibromas in Neurofibromatosis Type 1. Journal of the National Cancer Institute, 2011, 103, 1713-1722.	6.3	106
33	NF1 microdeletions in neurofibromatosis type 1: from genotype to phenotype. Human Mutation, 2010, 31, E1506-E1518.	2.5	208
34	Differential Expression of <i>CCN1</i> / <i>CYR61</i> , <i>CCN3/NOV</i> , <i>CCN4/WISP1</i> , and <i>CCN5/WISP2</i> in Neurofibromatosis Type 1 Tumorigenesis. Journal of Neuropathology and Experimental Neurology, 2010, 69, 60-69.	1.7	16
35	Unravelling the genetic basis of variable clinical expression in neurofibromatosis 1. Human Molecular Genetics, 2009, 18, 2768-2778.	2.9	129
36	Detection and Characterization of NF1 Microdeletions by Custom High Resolution Array CGH. Journal of Molecular Diagnostics, 2009, 11, 524-529.	2.8	31

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37	SPRED1 disorder and predisposition to leukemia in children. Blood, 2009, 114, 1131-1131.	1.4	40
38	Characterization of a 7.6-Mb germline deletion encompassing the NF1 locus and about a hundred genes in an NF1 contiguous gene syndrome patient. European Journal of Human Genetics, 2008, 16, 1459-1466.	2.8	30
39	Liver Iron, HFE Gene Mutations, and Hepatocellular Carcinoma Occurrence in Patients With Cirrhosis. Gastroenterology, 2008, 134, 102-110.	1.3	115
40	Significant gene expression differences in histologically "Normal―liver biopsies: Implications for control tissue. Hepatology, 2008, 48, 953-962.	7.3	25
41	Microarray-Based Identification of Tenascin C and Tenascin XB, Genes Possibly Involved in Tumorigenesis Associated with Neurofibromatosis Type 1. Clinical Cancer Research, 2007, 13, 398-407.	7.0	48
42	Characterization of a Germ-Line Deletion, Including the Entire <i>INK4/ARF</i> Locus, in a Melanoma-Neural System Tumor Family: Identification of <i>ANRIL</i> , an Antisense Noncoding RNA Whose Expression Coclusters with <i>ARF</i> . Cancer Research, 2007, 67, 3963-3969.	0.9	582
43	Conservation of hotspots for recombination in low-copy repeats associated with the NF1 microdeletion. Nature Genetics, 2006, 38, 1419-1423.	21.4	76
44	Genetic Polymorphisms in Antioxidant Enzymes Modulate Hepatic Iron Accumulation and Hepatocellular Carcinoma Development in Patients with Alcohol-Induced Cirrhosis. Cancer Research, 2006, 66, 2844-2852.	0.9	70
45	Molecular profiling of early stage liver fibrosis in patients with chronic hepatitis C virus infection. Virology, 2005, 332, 130-144.	2.4	145
46	Liver Gene Expression Signature of Mild Fibrosis in Patients With Chronic Hepatitis C. Gastroenterology, 2005, 129, 2064-2075.	1.3	161
47	Molecular Profiles of Neurofibromatosis Type 1-Associated Plexiform Neurofibromas. Clinical Cancer Research, 2004, 10, 3763-3771.	7.0	34
48	Molecular profiling of malignant peripheral nerve sheath tumors associated with neurofibromatosis type 1, based on large-scale real-time RT-PCR. Molecular Cancer, 2004, 3, 20.	19.2	85
49	Quantitative RT-PCR reveals a ubiquitous but preferentially neural expression of the KIS gene in rat and human. Molecular Brain Research, 2003, 114, 55-64.	2.3	24
50	Analyses ofMYC,ERBB2, andCCND1Genes in Benign and Malignant Thyroid Follicular Cell Tumors by Real-Time Polymerase Chain Reaction. Thyroid, 2001, 11, 147-152.	4.5	60
51	A novel mutation in the neurofibromatosis type 1 (NF1) gene promotes skipping of two exons by preventing exon definition11Edited by M. Yaniv. Journal of Molecular Biology, 2001, 307, 1261-1270.	4.2	29
52	The CGA gene as new predictor of the response to endocrine therapy in ERα-positive postmenopausal breast cancer patients. Oncogene, 2001, 20, 6955-6959.	5.9	22
53	Prenatal Detection by Real-Time Quantitative PCR and Characterization of a New CFTR Deletion, 3600+15kbdel5.3kb (or CFTRdele19). Clinical Chemistry, 2000, 46, 1417-1420.	3.2	25
54	Real-Time Reverse Transcription-PCR Assay for Future Management of ERBB2-based Clinical Applications. Clinical Chemistry, 1999, 45, 1148-1156.	3.2	235

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55	Novel approach to quantitative polymerase chain reaction using real-time detection: Application to the detection of gene amplification in breast cancer. International Journal of Cancer, 1998, 78, 661-666.	5.1	191
56	Novel recurrent nonsense mutation causing neurofibromatosis type 1 (NF1) in a family segregating both NF1 and Noonan syndrome. , 1998, 75, 265-272.		31
57	Novel approach to quantitative polymerase chain reaction using realâ€time detection: Application to the detection of gene amplification in breast cancer. International Journal of Cancer, 1998, 78, 661-666.	5.1	5
58	Echinococcus multilocularis:Microsatellite Polymorphism in U1 snRNA Genes. Experimental Parasitology, 1996, 82, 324-328.	1.2	39
59	Sex differences in mutational rate and mutational mechanism in the NF1 gene in neurofibromatosis type 1 patients. Human Genetics, 1996, 98, 696-699.	3.8	97
60	Nucleotide substitutions at the -6 position in the promoter region of the factor IX gene result in different severity of hemophilia B Leyden: consequences for genetic counseling. Human Genetics, 1993, 91, 241-4.	3.8	10
61	Detection of a molecular defect in 40 of 44 patients with haemophilia B by PCR and denaturing gradient gel electrophoresis. British Journal of Haematology, 1993, 84, 662-669.	2.5	35
62	Haemophilia B Due to a De Novo Insertion of a Human-Specific Alu Subfamily Member within the Coding Region of the Factor IX Gene. European Journal of Human Genetics, 1993, 1, 30-36.	2.8	106
63	Structure of the Echinococcus multilocularis U1 snRNA gene repeat. Molecular and Biochemical Parasitology, 1991, 46, 285-292.	1.1	15
64	Mutations in the catalytic domain of human coagulation factor IX: Rapid characterization by direct genomic sequencing of DNA fragments displaying an altered melting behavior. Genomics, 1989, 4, 266-272.	2.9	110