Ludwine M Messiaen

List of Publications by Year
in descending order

118 papers	6,738 citations	61984 43 h-index	69250 77 g-index
123 all docs	123 docs citations	123 times ranked	7418 citing authors

#	Article	IF	Citations
1	Exhaustive mutation analysis of theNF1 gene allows identification of 95% of mutations and reveals a high frequency of unusual splicing defects. Human Mutation, 2000, 15, 541-555.	2.5	477
2	Germline loss-of-function mutations in SPRED1 cause a neurofibromatosis 1â€“like phenotype. Nature Genetics, 2007, 39, 1120-1126.	21.4	410
3	Revised diagnostic criteria for neurofibromatosis type 1 and Legius syndrome: an international consensus recommendation. Genetics in Medicine, 2021, 23, 1506-1513.	2.4	290
4	Germline loss-of-function mutations in LZTR1 predispose to an inherited disorder of multiple schwannomas. Nature Genetics, 2014, 46, 182-187.	21.4	242
5	FOXL2 and BPES: Mutational Hotspots, Phenotypic Variability, and Revision of the Genotype-Phenotype Correlation. American Journal of Human Genetics, 2003, 72, 478-487.	6.2	219
6	Elucidating Distinct Roles for NF1 in Melanomagenesis. Cancer Discovery, 2013, 3, 338-349.	9.4	213
7	Mutations ofVMD2Splicing Regulators Cause Nanophthalmos and Autosomal Dominant Vitreoretinochoroidopathy (ADVIRC). , 2004, 45, 3683.		205
8	Molecular pathogenesis of multiple gastrointestinal stromal tumors in NF1 patients. Human Molecular Genetics, 2006, 15, 1015-1023.	2.9	195
9	NF1 Is a Tumor Suppressor in Neuroblastoma that Determines Retinoic Acid Response and Disease Outcome. Cell, 2010, 142, 218-229.	28.9	190
10	Clinical and Mutational Spectrum of Neurofibromatosis Type 1â€“like Syndrome. JAMA - Journal of the American Medical Association, 2009, 302, 2111.	7.4	160
11	Molecular Dissection of Isolated Disease Features in Mosaic Neurofibromatosis Type 1. American Journal of Human Genetics, 2007, 81, 243-251.	6.2	157
12	Double Inactivation of NF1 in Tibial Pseudarthrosis. American Journal of Human Genetics, 2006, 79, 143-148.	6.2	145
13	Genotype-Phenotype Correlation in NF1: Evidence for a More Severe Phenotype Associated with Missense Mutations Affecting NF1 Codons 844â€“848. American Journal of Human Genetics, 2018, 102, 69-87.	6.2	144
14	High Incidence of Noonan Syndrome Features Including Short Stature and Pulmonic Stenosis in Patients carrying NF1 Missense Mutations Affecting p.Arg1809: Genotypeâ€“Phenotype Correlation. Human Mutation, 2015, 36, 1052-1063.	2.5	143
15	Proteasomal and Genetic Inactivation of the NF1 Tumor Suppressor in Gliomagenesis. Cancer Cell, 2009, 16, 44-54.	16.8	132
16	Glomus Tumors in Neurofibromatosis Type 1: Genetic, Functional, and Clinical Evidence of a Novel Association. Cancer Research, 2009, 69, 7393-7401.	0.9	122
17	Mosaic type-1 NF1 microdeletions as a cause of both generalized and segmental neurofibromatosis type-1 (NF1). Human Mutation, 2011, 32, 213-219.	2.5	106
18	ComprehensiveNF1 screening on cultured Schwann cells from neurofibromas. Human Mutation, 2006, 27, 1030-1040.	2.5	105

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19	Somatic Mutation Analysis in NF1 CafÃ© au lait Spots Reveals Two NF1 Hits in the Melanocytes. Journal of Investigative Dermatology, 2008, 128, 1050-1053.	0.7	95
20	Disruption of exonic splicing enhancer elements is the principal cause of exon skipping associated with seven nonsense or missense alleles of NF1. Human Mutation, 2004, 24, 491-501.	2.5	94
21	The NF1 Gene Contains Hotspots for L1 Endonuclease-Dependent De Novo Insertion. PLoS Genetics, 2011, 7, e1002371.	3.5	93
22	Review and update ofSPRED1mutations causing legius syndrome. Human Mutation, 2012, 33, 1538-1546.	2.5	81
23	Clinical spectrum of individuals with pathogenic N F1 missense variants affecting p.Met1149, p.Arg1276, and p.Lys1423: genotypeâ€“phenotype study in neurofibromatosis type 1. Human Mutation, 2020, 41, 299-315.	2.5	80
24	Differentiating pathogenic mutations from polymorphic alterations in the splice sites ofBRCA1 andBRCA2. Genes Chromosomes and Cancer, 2003, 37, 314-320.	2.8	78
25	Conservation of hotspots for recombination in low-copy repeats associated with the NF1 microdeletion. Nature Genetics, 2006, 38, 1419-1423.	21.4	76
26	Neurofibromatosis-Noonan syndrome: Molecular evidence of the concurrence of both disorders in a patient. American Journal of Medical Genetics, Part A, 2005, 136A, 242-245.	1.2	74
27	panelcn.MOPS: Copy-number detection in targeted NGS panel data for clinical diagnostics. Human Mutation, 2017, 38, 889-897.	2.5	72
28	Elucidating the impact of neurofibromatosis-1 germline mutations on neurofibromin function and dopamine-based learning. Human Molecular Genetics, 2015, 24, 3518-3528.	2.9	70
29	The Development of Cutaneous Neurofibromas. American Journal of Pathology, 2011, 178, 500-505.	3.8	63
30	SVA retrotransposon insertion-associated deletion represents a novel mutational mechanism underlying large genomic copy number changes with non-recurrent breakpoints. Genome Biology, 2014, 15, R80.	9.6	63
31	Expanding the clinical phenotype of individuals with a 3-bp in-frame deletion of the NF1 gene (c.2970_2972del): an update of genotypeâ€“phenotype correlation. Genetics in Medicine, 2019, 21, 867-876.	2.4	62
32	Updated diagnostic criteria and nomenclature for neurofibromatosis type 2 and schwannomatosis: An international consensus recommendation. Genetics in Medicine, 2022, 24, 1967-1977.	2.4	60
33	Mitotic recombination as evidence of alternative pathogenesis of gastrointestinal stromal tumours in neurofibromatosis type 1. Journal of Medical Genetics, 2006, 44, e61-e61.	3.2	59
34	Pigment cellâ€related manifestations in neurofibromatosis type 1: an overview. Pigment Cell & Melanoma Research, 2005, 18, 13-24.	3.6	58
35	Somatic loss of wild typeNF1 allele in neurofibromas: Comparison ofNF1 microdeletion and non-microdeletion patients. Genes Chromosomes and Cancer, 2006, 45, 893-904.	2.8	56
36	Is Pulmonary Arterial Hypertension in Neurofibromatosis Type 1 Secondary to a Plexogenic Arteriopathy?. Chest, 2007, 132, 798-808.	0.8	56

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37	The humanFOXL2 mutation database. Human Mutation, 2004, 24, 189-193.	2.5	55
38	CafÃ©-au-lait spots in neurofibromatosis type 1 and in healthy control individuals: hyperpigmentation of a different kind?. Archives of Dermatological Research, 2006, 297, 439-449.	1.9	54
39	Genetic and clinical mosaicism in a patient with neurofibromatosis typeÂ1. Human Genetics, 2004, 114, 284-290.	3.8	53
40	Congenital pseudarthrosis of neurofibromatosis type 1: Impaired osteoblast differentiation and function and altered NF1 gene expression. Bone, 2009, 44, 243-250.	2.9	49
41	Interaction between a Domain of the Negative Regulator of the Ras-ERK Pathway, SPRED1 Protein, and the GTPase-activating Protein-related Domain of Neurofibromin Is Implicated in Legius Syndrome and Neurofibromatosis Type 1. Journal of Biological Chemistry, 2016, 291, 3124-3134.	3.4	49
42	Genetically engineered minipigs model the major clinical features of human neurofibromatosis type 1. Communications Biology, 2018, 1, 158.	4.4	49
43	Neurofibromin binds to caveolin-1 and regulates ras, FAK, and Akt. Biochemical and Biophysical Research Communications, 2006, 340, 1200-1208.	2.1	44
44	Neurofibromatosis Type 1 Protein and Amyloid Precursor Protein Interact in Normal Human Melanocytes and Colocalize with Melanosomes. Journal of Investigative Dermatology, 2006, 126, 653-659.	0.7	44
45	Human Myosin V Gene Produces Different Transcripts in a Cell Type-Specific Manner. Biochemical and Biophysical Research Communications, 1998, 252, 329-333.	2.1	43
46	Mutation Spectrum of NF1 and Clinical Characteristics in 78 Korean Patients With Neurofibromatosis Type 1. Pediatric Neurology, 2013, 48, 447-453.	2.1	43
47	Radiation-induced rhabdomyosarcoma of the brainstem in a patient with neurofibromatosis Type 2. Journal of Neurosurgery, 2010, 112, 81-87.	1.6	42
48	Quantification ofNF1transcripts reveals novel highly expressed splice variants. FEBS Letters, 2002, 522, 71-76.	2.8	41
49	Constitutional mismatch repair deficiency as a differential diagnosis of neurofibromatosis type 1: consensus guidelines for testing a child without malignancy. Journal of Medical Genetics, 2019, 56, 53-62.	3.2	40
50	Bax expression is a candidate prognostic and predictive marker of colorectal cancer. Journal of Gastrointestinal Oncology, 2010, 1, 76-89.	1.4	40
51	Neurofibromin is actively transported to the nucleus. FEBS Letters, 2004, 560, 98-102.	2.8	39
52	Pathological splice mutations outside the invariant AG/GT splice sites of BRCA1 exon 5 increase alternative transcript levels in the 5â€² end of the BRCA1 gene. Oncogene, 2002, 21, 4171-4175.	5.9	35
53	Report of a patient with a constitutional missense mutation in SMARCB1, Coffinâ€“Siris phenotype, and schwannomatosis. American Journal of Medical Genetics, Part A, 2015, 167, 3186-3191.	1.2	35
54	Exon 10b of the NF1 gene represented a mutational hotspot and harbors a recurrent missense mutation y489c associated with aberrant splicing. Genetics in Medicine, 1999, 1, 248-253.	2.4	34

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55	Jaffeâ€“Campanacci syndrome, revisited: detailed clinical and molecular analyses determine whether patients have neurofibromatosis type 1, coincidental manifestations, or a distinct disorder. Genetics in Medicine, 2014, 16, 448-459.	2.4	33
56	Closing in on the BPES Gene on 3q23: Mapping of ade NovoReciprocal Translocation t(3;4)(q23;p15.2) Breakpoint within a 45-kb Cosmid and Mapping of Three Candidate Genes, RBP1, RBP2, and Iº²â€²-COP, Distal to the Breakpoint. Genomics, 1999, 57, 70-78.	2.9	32
57	Diverse prevalence of large deletions within the OA1 gene in ocular albinism type 1 patients from Europe and North America. Human Genetics, 2001, 108, 51-54.	3.8	31
58	Aberrant splicing in several human tumors in the tumor suppressor genes neurofibromatosis type 1, neurofibromatosis type 2, and tuberous sclerosis 2. Cancer Research, 2002, 62, 1503-9.	0.9	29
59	Combined Retinal Hamartomas Leading to the Diagnosis of Neurofibromatosis Type 2. Ophthalmic Genetics, 2008, 29, 133-138.	1.2	28
60	Characterization of the nonallelic homologous recombination hotspot PRS3 associated with type-3NF1deletions. Human Mutation, 2012, 33, 372-383.	2.5	28
61	Predicting neurofibromatosis type 1 riskÂamong children with isolated cafÃ©-au-laitÂmacules. Journal of the American Academy of Dermatology, 2017, 76, 1077-1083.e3.	1.2	28
62	Mutation Analysis of the BRCA1 and BRCA2 Genes in the Belgian Patient Population and Identification of a Belgian Founder Mutation BRCA1 IVS5+3A>G. Disease Markers, 1999, 15, 69-73.	1.3	27
63	Prognostic Significance and Gene Expression Profiles of p53 Mutations in Microsatellite-Stable Stage III Colorectal Adenocarcinomas. PLoS ONE, 2012, 7, e30020.	2.5	27
64	Identification of recurrent type-2NF1microdeletions reveals a mitotic nonallelic homologous recombination hotspot underlying a human genomic disorder. Human Mutation, 2012, 33, 1599-1609.	2.5	26
65	A patient severely affected by spinal neurofibromas carries a recurrent splice site mutation in the NF1 gene. European Journal of Human Genetics, 2002, 10, 334-338.	2.8	25
66	Decoding NF1 Intragenic Copy-Number Variations. American Journal of Human Genetics, 2015, 97, 238-249.	6.2	24
67	AGâ€exclusion zone revisited: Lessons to learn from 91 intronic NF1 3â€² splice site mutations outside the canonical AGâ€dinucleotides. Human Mutation, 2020, 41, 1145-1156.	2.5	24
68	Noonan-like phenotype in monozygotic twins with a duplication-deficiency of the long arm of chromosome 18 resulting from a maternal paracentric inversion. Human Genetics, 1998, 103, 497-505.	3.8	23
69	Identification of BPESC1, a Novel Gene Disrupted by a Balanced Chromosomal Translocation, t(3;4)(q23;p15.2), in a Patient with BPES. Genomics, 2000, 68, 296-304.	2.9	23
70	Identification of PKHD1 Multiexon Deletions Using Multiplex Ligation-Dependent Probe Amplification and Quantitative Polymerase Chain Reaction. Genetic Testing and Molecular Biomarkers, 2010, 14, 505-510.	0.7	23
71	Mice with missense and nonsense NF1 mutations display divergent phenotypes compared to NF1 patients. DMM Disease Models and Mechanisms, 2016, 9, 759-67.	2.4	23
72	Breast cancer risk and germline genomic profiling of women with neurofibromatosis type 1 who developed breast cancer. Genes Chromosomes and Cancer, 2018, 57, 19-27.	2.8	22

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73	Novel Frameshift Mutation in a Heterozygous Woman with Fabry Disease and End-Stage Renal Failure. American Journal of Nephrology, 1996, 16, 352-357.	3.1	20
74	FunctionalPMS2hybrid alleles containing a pseudogene-specific missense variant trace back to a single ancient intrachromosomal recombination event. Human Mutation, 2010, 31, n/a-n/a.	2.5	20
75	Refined Genetic and Physical Mapping of BPES Type II. European Journal of Human Genetics, 1996, 4, 34-38.	2.8	20
76	Improved multiplex ligationâ€dependent probe amplification analysis identifies a deleterious PMS2 allele generated by recombination with crossover between PMS2 and PMS2CL. Genes Chromosomes and Cancer, 2012, 51, 819-831.	2.8	19
77	Paraspinal neurofibromas and hypertrophic neuropathy in Noonan syndrome with multiple lentigines. Journal of Medical Genetics, 2016, 53, 123-126.	3.2	19
78	Familial pericentric inversion of chromosome 18: behavioral abnormalities in patients heterozygous for either the dup(18p)/del(18q) or dup(18q)/del(18p) recombinant chromosome. European Journal of Human Genetics, 2005, 13, 52-58.	2.8	18
79	Analysis of Crossover Breakpoints Yields New Insights into the Nature of the Gene Conversion Events Associated with LargeNF1Deletions Mediated by Nonallelic Homologous Recombination. Human Mutation, 2014, 35, 215-226.	2.5	17
80	Chondrodysplasia punctata with multiple congenital anomalies: a new syndrome?. Pediatric Radiology, 1998, 28, 790-793.	2.0	16
81	Lack of independence between five DNA polymorphisms in the NF1 gene. Human Molecular Genetics, 1993, 2, 485-485.	2.9	15
82	Cloning and Characterization of HumanWDR10, a Novel Gene Located at 3q21 Encoding a WD-Repeat Protein That Is Highly Expressed in Pituitary and Testis. DNA and Cell Biology, 2001, 20, 41-52.	1.9	15
83	Phenotypic variability among cafÃ©-au-lait macules in neurofibromatosis type 1. Journal of the American Academy of Dermatology, 2010, 63, 440-447.	1.2	15
84	Fine mapping of meiotic NAHR-associated crossovers causing largeNF1deletions. Human Molecular Genetics, 2016, 25, 484-496.	2.9	15
85	Real-time quantitative allele discrimination assay using 3â€² locked nucleic acid primers for detection of low-percentage mosaic mutations. Analytical Biochemistry, 2006, 359, 144-146.	2.4	14
86	Constitutional mismatch repair deficiency is the diagnosis in 0.41% of pathogenic NF1/SPRED1 variant negative children suspected of sporadic neurofibromatosis type 1. Genetics in Medicine, 2020, 22, 2081-2088.	2.4	14
87	Palindrome-Mediated and Replication-Dependent Pathogenic Structural Rearrangements within theNF1Gene. Human Mutation, 2014, 35, 891-898.	2.5	13
88	Extreme clustering of type-1 NF1 deletion breakpoints co-locating with G-quadruplex forming sequences. Human Genetics, 2018, 137, 511-520.	3.8	13
89	Analysis of copy number variants in 11 pairs of monozygotic twins with neurofibromatosis type 1. American Journal of Medical Genetics, Part A, 2017, 173, 647-653.	1.2	12
90	Ultra-deep amplicon sequencing indicates absence of low-grade mosaicism with normal cells in patients with type-1 NF1 deletions. Human Genetics, 2019, 138, 73-81.	3.8	12

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91	Concurrent DNA Copy-Number Alterations and Mutations in Genes Related to Maintenance of Genome Stability in Uninvolved Mammary Glandular Tissue from Breast Cancer Patients. Human Mutation, 2015, 36, 1088-1099.	2.5	11
92	Argâ€“Cys Substitution at Codon 1246 of the Human Myosin Va Gene is not Associated with Griscelli Syndrome. Journal of Investigative Dermatology, 2000, 114, 731-733.	0.7	10
93	Hypomagnesemia due to two novel TRPM6 mutations. Journal of Pediatric Endocrinology and Metabolism, 2015, 28, 1373-8.	0.9	10
94	Association between Cys282Tyr missense mutation and haptoglobin phenotype polymorphism in patients with chronic hepatitis C. European Journal of Gastroenterology and Hepatology, 2001, 13, 1077-1081.	1.6	9
95	Multiple orbital neurofibromas, painful peripheral nerve tumors, distinctive face and marfanoid habitus: a new syndrome. European Journal of Human Genetics, 2012, 20, 618-625.	2.8	9
96	Mosaicism for a SPRED1 deletion revealed in a patient with clinically suspected mosaic neurofibromatosis. British Journal of Dermatology, 2017, 176, 1077-1078.	1.5	8
97	Clinical significance of a novel single nucleotide polymorphism in the 5' untranslated region of the Rabphillin-3A-Like gene in colorectal adenocarcinoma. Frontiers in Bioscience - Landmark, 2008, 13, 1050.	3.0	8
98	Evidence for involvement of a tumor suppressor gene on 1p in malignant peripheral nerve sheath tumors. Cancer Genetics and Cytogenetics, 2003, 143, 120-124.	1.0	7
99	Identification of LargeNF1Duplications Reciprocal to NAHR-Mediated Type-1NF1Deletions. Human Mutation, 2014, 35, 1469-1475.	2.5	7
100	Child Neurology: Spastic paraparesis and dystonia with a novel ADCY5 mutation. Neurology, 2019, 93, 510-514.	1.1	7
101	Targeted exon skipping of NF1 exon 17 as a therapeutic for neurofibromatosis type I. Molecular Therapy - Nucleic Acids, 2022, 28, 261-278.	5.1	7
102	Exhaustive mutation analysis of the NF1 gene allows identification of 95% of mutations and reveals a high frequency of unusual splicing defects. Human Mutation, 2000, 15, 541.	2.5	6
103	Analysis of 22 cystic fibrosis mutations in 62 patients from the Flanders, Belgium, reveals a high prevalence of Nordic mutation 394delTT. Human Mutation, 1997, 10, 236-238.	2.5	5
104	Interphase FISH, the structure of reciprocal translocation chromosomes and physical mapping studies rule out the duplication of the NF1 gene at 17q11.2. A reply. Human Genetics, 2003, 113, 188-190.	3.8	5
105	Reâ€evaluation of missense variant classifications in NF2. Human Mutation, 2022, 43, 643-654.	2.5	5
106	Natural history of NF1 c.2970_2972del p.(Met992del): confirmation of a low risk of complications in a longitudinal study. European Journal of Human Genetics, 2022, 30, 291-297.	2.8	5
107	Germline and Somatic NF1 Alterations Are Linked to Increased HER2 Expression in Breast Cancer. Cancer Prevention Research, 2018, 11, 655-664.	1.5	4
108	Exhaustive mutation analysis of the NF1 gene allows identification of 95% of mutations and reveals a high frequency of unusual splicing defects. , 2000, 15, 541.		4

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109	Clinical Implications of Rabphillin-3A-Like Gene Alterations in Breast Cancer. PLoS ONE, 2015, 10, e0129216.	2.5	4
110	Multiple Myofibromas and an Epidermal Verrucous Nevus in a Child with Neurofibromatosis Type 1. Dermatology, 2004, 209, 223-227.	2.1	3
111	Typical 22q11.2 deletion syndrome appears to confer a reduced risk of schwannoma. Genetics in Medicine, 2021, 23, 1779-1782.	2.4	3
112	Targeted massively parallel sequencing of candidate regions on chromosome 22q predisposing to multiple schwannomas: an analysis of 51 individuals in a single center experience. Human Mutation, 2021, , .	2.5	3
113	Improved conditions for PTT analysis of the BRCA1, BRCA2, NF1 and APC genes. Technical Tips Online, 1998, 3, 90-93.	0.2	2
114	Molecular Diagnosis for NF1. , 2020, , 15-34.		2
115	Error in a Study of the Clinical and Mutational Spectrum of Neurofibromatosis Type 1â€“like Syndrome. JAMA - Journal of the American Medical Association, 2010, 303, 2476.	7.4	1
116	High-Throughput Tabular Data Processor â€“ Platform independent graphical tool for processing large data sets. PLoS ONE, 2018, 13, e0192858.	2.5	1
117	Alternative outcomes of pathogenic complex somatic structural variations in the genomes of NF1 and NF2 patients. Neurogenetics, 2017, 18, 169-174.	1.4	0
118	eP330: Mosaicism for SMARCB1 or LZTR1 variants in patients with schwannomatosis in the UAB cohort. Genetics in Medicine, 2022, 24, S206-S207.	2.4	0