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	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
2	Reâ€evaluation of missense variant classifications in NF2. Human Mutation, 2022, 43, 643-654.	2.5	5
3	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
4	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
5	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
6	Typical 22q11.2 deletion syndrome appears to confer a reduced risk of schwannoma. Genetics in Medicine, 2021, 23, 1779-1782.	2.4	3
7	Revised diagnostic criteria for neurofibromatosis type 1 and Legius syndrome: an international consensus recommendation. Genetics in Medicine, 2021, 23, 1506-1513.	2.4	290
8	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
9	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
10	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
11	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
12	Molecular Diagnosis for NF1. , 2020, , 15-34.		2
13	Child Neurology: Spastic paraparesis and dystonia with a novel ADCY5 mutation. Neurology, 2019, 93, 510-514.	1.1	7
14	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
15	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
16	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
17	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
18	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

#	Article	IF	Citations
19	Genetically engineered minipigs model the major clinical features of human neurofibromatosis type 1. Communications Biology, 2018, 1, 158.	4.4	49
20	Extreme clustering of type-1 NF1 deletion breakpoints co-locating with G-quadruplex forming sequences. Human Genetics, 2018, 137, 511-520.	3.8	13
21	Germline and Somatic NF1 Alterations Are Linked to Increased HER2 Expression in Breast Cancer. Cancer Prevention Research, 2018, 11, 655-664.	1.5	4
22	High-Throughput Tabular Data Processor â€“ Platform independent graphical tool for processing large data sets. PLoS ONE, 2018, 13, e0192858.	2.5	1
23	Alternative outcomes of pathogenic complex somatic structural variations in the genomes of NF1 and NF2 patients. Neurogenetics, 2017, 18, 169-174.	1.4	0
24	panelcn.MOPS: Copy-number detection in targeted NGS panel data for clinical diagnostics. Human Mutation, 2017, 38, 889-897.	2.5	72
25	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
26	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
27	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
28	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
29	Paraspinal neurofibromas and hypertrophic neuropathy in Noonan syndrome with multiple lentigines. Journal of Medical Genetics, 2016, 53, 123-126.	3.2	19
30	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
31	Fine mapping of meiotic NAHR-associated crossovers causing largeNF1deletions. Human Molecular Genetics, 2016, 25, 484-496.	2.9	15
32	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
33	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
34	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
35	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
36	Decoding NF1 Intragenic Copy-Number Variations. American Journal of Human Genetics, 2015, 97, 238-249.	6.2	24

#	Article	IF	Citations
37	Hypomagnesemia due to two novel TRPM6 mutations. Journal of Pediatric Endocrinology and Metabolism, 2015, 28, 1373-8.	0.9	10
38	Clinical Implications of Rabphillin-3A-Like Gene Alterations in Breast Cancer. PLoS ONE, 2015, 10, e0129216.	2.5	4
39	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
40	Palindrome-Mediated and Replication-Dependent Pathogenic Structural Rearrangements within theNF1Gene. Human Mutation, 2014, 35, 891-898.	2.5	13
41	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
42	Germline loss-of-function mutations in LZTR1 predispose to an inherited disorder of multiple schwannomas. Nature Genetics, 2014, 46, 182-187.	21.4	242
43	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
44	Identification of LargeNF1Duplications Reciprocal to NAHR-Mediated Type-1NF1Deletions. Human Mutation, 2014, 35, 1469-1475.	2.5	7
45	Mutation Spectrum of NF1 and Clinical Characteristics in 78 Korean Patients With Neurofibromatosis Type 1. Pediatric Neurology, 2013, 48, 447-453.	2.1	43
46	Elucidating Distinct Roles for NF1 in Melanomagenesis. Cancer Discovery, 2013, 3, 338-349.	9.4	213
47	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
48	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
49	Prognostic Significance and Gene Expression Profiles of p53 Mutations in Microsatellite-Stable Stage III Colorectal Adenocarcinomas. PLoS ONE, 2012, 7, e30020.	2.5	27
50	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
51	Review and update ofSPRED1mutations causing legius syndrome. Human Mutation, 2012, 33, 1538-1546.	2.5	81
52	Characterization of the nonallelic homologous recombination hotspot PRS3 associated with type-3NF1deletions. Human Mutation, 2012, 33, 372-383.	2.5	28
53	The NF1 Gene Contains Hotspots for L1 Endonuclease-Dependent De Novo Insertion. PLoS Genetics, 2011, 7, e1002371.	3.5	93
54	The Development of Cutaneous Neurofibromas. American Journal of Pathology, 2011, 178, 500-505.	3.8	63

#	Article	IF	Citations
55	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
56	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
57	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
58	Radiation-induced rhabdomyosarcoma of the brainstem in a patient with neurofibromatosis Type 2. Journal of Neurosurgery, 2010, 112, 81-87.	1.6	42
59	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
60	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
61	NF1 Is a Tumor Suppressor in Neuroblastoma that Determines Retinoic Acid Response and Disease Outcome. Cell, 2010, 142, 218-229.	28.9	190
62	Bax expression is a candidate prognostic and predictive marker of colorectal cancer. Journal of Gastrointestinal Oncology, 2010, 1, 76-89.	1.4	40
63	Glomus Tumors in Neurofibromatosis Type 1: Genetic, Functional, and Clinical Evidence of a Novel Association. Cancer Research, 2009, 69, 7393-7401.	0.9	122
64	Clinical and Mutational Spectrum of Neurofibromatosis Type 1â€“like Syndrome. JAMA - Journal of the American Medical Association, 2009, 302, 2111.	7.4	160
65	Proteasomal and Genetic Inactivation of the NF1 Tumor Suppressor in Gliomagenesis. Cancer Cell, 2009, 16, 44-54.	16.8	132
66	Congenital pseudarthrosis of neurofibromatosis type 1: Impaired osteoblast differentiation and function and altered NF1 gene expression. Bone, 2009, 44, 243-250.	2.9	49
67	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
68	Combined Retinal Hamartomas Leading to the Diagnosis of Neurofibromatosis Type 2. Ophthalmic Genetics, 2008, 29, 133-138.	1.2	28
69	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
70	Is Pulmonary Arterial Hypertension in Neurofibromatosis Type 1 Secondary to a Plexogenic Arteriopathy?. Chest, 2007, 132, 798-808.	0.8	56
71	Molecular Dissection of Isolated Disease Features in Mosaic Neurofibromatosis Type 1. American Journal of Human Genetics, 2007, 81, 243-251.	6.2	157
72	Germline loss-of-function mutations in SPRED1 cause a neurofibromatosis 1â€“like phenotype. Nature Genetics, 2007, 39, 1120-1126.	21.4	410

#	Article	IF	Citations
73	Double Inactivation of NF1 in Tibial Pseudarthrosis. American Journal of Human Genetics, 2006, 79, 143-148.	6.2	145
74	Neurofibromin binds to caveolin-1 and regulates ras, FAK, and Akt. Biochemical and Biophysical Research Communications, 2006, 340, 1200-1208.	2.1	44
75	Conservation of hotspots for recombination in low-copy repeats associated with the NF1 microdeletion. Nature Genetics, 2006, 38, 1419-1423.	21.4	76
76	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
77	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
78	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
79	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
80	ComprehensiveNF1 screening on cultured Schwann cells from neurofibromas. Human Mutation, 2006, 27, 1030-1040.	2.5	105
81	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
82	Molecular pathogenesis of multiple gastrointestinal stromal tumors in NF1 patients. Human Molecular Genetics, 2006, 15, 1015-1023.	2.9	195
83	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
84	Pigment cellâ€related manifestations in neurofibromatosis type 1: an overview. Pigment Cell & Melanoma Research, 2005, 18, 13-24.	3.6	58
85	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
86	Mutations ofVMD2Splicing Regulators Cause Nanophthalmos and Autosomal Dominant Vitreoretinochoroidopathy (ADVIRC). , 2004, 45, 3683.		205
87	Multiple Myofibromas and an Epidermal Verrucous Nevus in a Child with Neurofibromatosis Type 1. Dermatology, 2004, 209, 223-227.	2.1	3
88	Genetic and clinical mosaicism in a patient with neurofibromatosis typeÂ1. Human Genetics, 2004, 114, 284-290.	3.8	53
89	The humanFOXL2 mutation database. Human Mutation, 2004, 24, 189-193.	2.5	55
90	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

#	Article	IF	Citations
91	Neurofibromin is actively transported to the nucleus. FEBS Letters, 2004, 560, 98-102.	2.8	39
92	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
93	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
94	Differentiating pathogenic mutations from polymorphic alterations in the splice sites ofBRCA1 andBRCA2. Genes Chromosomes and Cancer, 2003, 37, 314-320.	2.8	78
95	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
96	Quantification ofNF1transcripts reveals novel highly expressed splice variants. FEBS Letters, 2002, 522, 71-76.	2.8	41
97	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
98	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
99	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
100	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
101	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
102	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
103	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
104	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
105	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
106	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
107	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
108	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

#	Article	IF	Citations
109	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
110	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 Î²â€²-COP, Distal to the Breakpoint. Genomics, 1999, 57, 70-78.	2.9	32
111	Improved conditions for PTT analysis of the BRCA1, BRCA2, NF1 and APC genes. Technical Tips Online, 1998, 3, 90-93.	0.2	2
112	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
113	Chondrodysplasia punctata with multiple congenital anomalies: a new syndrome?. Pediatric Radiology, 1998, 28, 790-793.	2.0	16
114	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
115	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
116	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
117	Refined Genetic and Physical Mapping of BPES Type II. European Journal of Human Genetics, 1996, 4, 34-38.	2.8	20
118	Lack of independence between five DNA polymorphisms in the NF1 gene. Human Molecular Genetics, 1993, 2, 485-485.	2.9	15