Meena Upadhyaya

List of Publications by Year
in descending order

89 papers	6,058 citations	94415 37 h-index	74160 75 g-index
94 all docs	94 docs citations	94 times ranked	5703 citing authors

#	Article	IF	Citations
1	High-resolution breakpoint junction mapping of proximally extended D4Z4 deletions in FSHD1 reveals evidence for a founder effect. Human Molecular Genetics, 2022, 31, 748-760.	2.9	8
2	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
3	Management of neurofibromatosis type 1-associated plexiform neurofibromas. Neuro-Oncology, 2022, 24, 1827-1844.	1.2	29
4	Revised diagnostic criteria for neurofibromatosis type 1 and Legius syndrome: an international consensus recommendation. Genetics in Medicine, 2021, 23, 1506-1513.	2.4	290
5	Breast cancer risk in neurofibromatosis type 1 is a function of the type of NF1 gene mutation: a new genotype-phenotype correlation. Journal of Medical Genetics, 2019, 56, 209-219.	3.2	26
6	Emerging therapeutic targets for neurofibromatosis type 1. Expert Opinion on Therapeutic Targets, 2018, 22, 419-437.	3.4	53
7	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
8	Extreme clustering of type-1 NF1 deletion breakpoints co-locating with G-quadruplex forming sequences. Human Genetics, 2018, 137, 511-520.	3.8	13
9	Confirmation of mutation landscape of NF1â€associated malignant peripheral nerve sheath tumors. Genes Chromosomes and Cancer, 2017, 56, 421-426.	2.8	54
10	The NF1 somatic mutational landscape in sporadic human cancers. Human Genomics, 2017, 11, 13.	2.9	203
11	Telomere erosion in NF1 tumorigenesis. Oncotarget, 2017, 8, 40132-40139.	1.8	8
12	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
13	Remotely acting SMCHD1 gene regulatory elements: in silico prediction and identification of potential regulatory variants in patients with FSHD. Human Genomics, 2015, 9, 25.	2.9	0
14	Evaluation of copy number variation and gene expression in neurofibromatosis type-1-associated malignant peripheral nerve sheath tumours. Human Genomics, 2015, 9, 3.	2.9	17
15	STAT3 and HIF1Î± Signaling Drives Oncogenic Cellular Phenotypes in Malignant Peripheral Nerve Sheath Tumors. Molecular Cancer Research, 2015, 13, 1149-1160.	3.4	25
16	Identification of two novel SMCHD1 sequence variants in families with FSHD-like muscular dystrophy. European Journal of Human Genetics, 2015, 23, 67-71.	2.8	17
17	Inter-individual differences in CpG methylation at D4Z4 correlate with clinical variability in FSHD1 and FSHD2. Human Molecular Genetics, 2015, 24, 659-669.	2.9	130
18	The Molecular Biology of Neurofibromatosis Type 1. Colloquium Series on Genomic and Molecular Medicine, 2014, 3, 1-79.	0.2	0

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19	PRC2 loss amplifies Ras-driven transcription and confers sensitivity to BRD4-based therapies. Nature, 2014, 514, 247-251.	27.8	386
20	Screening in silico predicted remotely acting NF1gene regulatory elements for mutations in patients with neurofibromatosis type 1. Human Genomics, 2013, 7, 18.	2.9	4
21	Can the diagnosis of NF1 be excluded clinically? A lack of pigmentary findings in families with spinal neurofibromatosis demonstrates a limitation of clinical diagnosis. Journal of Medical Genetics, 2013, 50, 606-613.	3.2	28
22	Increased rate of missense/in-frame mutations in individuals with NF1-related pulmonary stenosis: a novel genotypeâ€“phenotype correlation. European Journal of Human Genetics, 2013, 21, 535-539.	2.8	27
23	Exploring the somatic NF1 mutational spectrum associated with NF1 cutaneous neurofibromas. European Journal of Human Genetics, 2012, 20, 411-419.	2.8	25
24	Abnormal Achromatic and Chromatic Contrast Sensitivity in Neurofibromatosis Type 1. , 2012, 53, 287.		19
25	Assessment of the potential pathogenicity of missense mutations identified in the GTPase-activating protein (GAP)-related domain of the neurofibromatosis type-1 (NF1) gene. Human Mutation, 2012, 33, 1687-1696.	2.5	21
26	Genotype-phenotype associations in neurofibromatosis type 1 (NF1): an increased risk of tumor complications in patients with NF1splice-site mutations?. Human Genomics, 2012, 6, 12.	2.9	50
27	Molecular heterogeneity in malignant peripheral nerve sheath tumors associated with neurofibromatosis type 1. Human Genomics, 2012, 6, 18.	2.9	21
28	An emerging role for microRNAs in NF1 tumorigenesis. Human Genomics, 2012, 6, 23.	2.9	16
29	The Germline Mutational Spectrum in Neurofibromatosis Type 1 and Genotypeâ€“Phenotype Correlations. , 2012, , 115-134.		6
30	Microarray-based copy number analysis of neurofibromatosis type-1 (NF1)-associated malignant peripheral nerve sheath tumors reveals a role for Rho-GTPase pathway genes in NF1 tumorigenesis. Human Mutation, 2012, 33, 763-776.	2.5	44
31	Facioscapulohumeral muscular dystrophy (FSHD): an enigma unravelled?. Human Genetics, 2012, 131, 325-340.	3.8	128
32	The Somatic Mutational Spectrum of the NF1 Gene. , 2012, , 211-233.		3
33	Somatic Copy Number Alterations: Gene and Protein Expression Correlates in NF1-Associated Malignant Peripheral Nerve Sheath Tumors. , 2012, , 405-428.		0
34	Genetic basis of tumorigenesis in NF1 malignant peripheral nerve sheath tumors. Frontiers in Bioscience - Landmark, 2011, 16, 937.	3.0	51
35	Neurofibromatosis type 1-associated tumours: Their somatic mutational spectrum and pathogenesis. Human Genomics, 2011, 5, 623.	2.9	113
36	Different sized somatic NF1 locus rearrangements in neurofibromatosisÂ1-associated malignant peripheral nerve sheath tumors. Journal of Neuro-Oncology, 2011, 102, 341-346.	2.9	9

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37	A molecular analysis of individuals with neurofibromatosis type 1 (NF1) and optic pathway gliomas (OPGs), and an assessment of genotype-phenotype correlations. Journal of Medical Genetics, 2011, 48, 256-260.	3.2	90
38	Analysis of NF1 somatic mutations in cutaneous neurofibromas from patients with high tumor burden. Neurogenetics, 2010, 11, 391-400.	1.4	25
39	Molecular evolution of a neurofibroma to malignant peripheral nerve sheath tumor (MPNST) in an NF1 patient: correlation between histopathological, clinical and molecular findings. Journal of Cancer Research and Clinical Oncology, 2010, 136, 1869-1880.	2.5	53
40	NF1 microdeletions in neurofibromatosis type 1: from genotype to phenotype. Human Mutation, 2010, 31, E1506-E1518.	2.5	208
41	Neurofibromatosis type 1: diagnosis and recent advances. Expert Opinion on Medical Diagnostics, 2010, 4, 307-322.	1.6	38
42	Confirmation that the specific SSLP microsatellite allele 4qA161 segregates with fascioscapulohumeral muscular dystrophy (FSHD) in a cohort of multiplex and simplex FSHD families. Muscle and Nerve, 2010, 42, 820-821.	2.2	17
43	Glomus Tumors in Neurofibromatosis Type 1: Genetic, Functional, and Clinical Evidence of a Novel Association. Cancer Research, 2009, 69, 7393-7401.	0.9	122
44	Integrative genomic analyses of neurofibromatosis tumours identify SOX9 as a biomarker and survival gene. EMBO Molecular Medicine, 2009, 1, 236-248.	6.9	112
45	Genomeâ€wide highâ€resolution analysis of DNA copy number alterations in NF1â€associated malignant peripheral nerve sheath tumors using 32K BAC array. Genes Chromosomes and Cancer, 2009, 48, 897-907.	2.8	50
46	The spectrum of somatic and germline NF1 mutations in NF1 patients with spinal neurofibromas. Neurogenetics, 2009, 10, 251-263.	1.4	61
47	A novel mutation in theNF1gene in two siblings with neurofibromatosis type 1 and bilateral optic pathway glioma. Pediatric Blood and Cancer, 2008, 50, 713-715.	1.5	8
48	Germline and somaticNF1 gene mutation spectrum in NF1-associated malignant peripheral nerve sheath tumors (MPNSTs). Human Mutation, 2008, 29, 74-82.	2.5	106
49	Profiling of copy number variations (CNVs) in healthy individuals from three ethnic groups using a human genome 32â€‰K BAC-clone-based array. Human Mutation, 2008, 29, 398-408.	2.5	46
50	Germline and somaticNF1gene mutations in plexiform neurofibromas. Human Mutation, 2008, 29, E103-E111.	2.5	51
51	Telomerase activity is a biomarker for high grade malignant peripheral nerve sheath tumors in neurofibromatosis type 1 individuals. Genes Chromosomes and Cancer, 2008, 47, 238-246.	2.8	23
52	Neurofibromatosis type 1 & Related Disorders. , 2008, , 51-151.		12
53	High-Resolution DNA Copy Number Profiling of Malignant Peripheral Nerve Sheath Tumors Using Targeted Microarray-Based Comparative Genomic Hybridization. Clinical Cancer Research, 2008, 14, 1015-1024.	7.0	119
54	An Absence of Cutaneous Neurofibromas Associated with a 3-bp Inframe Deletion in Exon 17 of the NF1 Gene (c.2970-2972 delAAT): Evidence of a Clinically Significant NF1 Genotype-Phenotype Correlation. American Journal of Human Genetics, 2007, 80, 140-151.	6.2	335

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55	Molecular diagnosis of neurofibromatosis type 1: 2Âyears experience. Familial Cancer, 2007, 6, 21-34.	1.9	74
56	Somatic alterations of the NF1 gene in an NF1 individual with multiple benign tumours (internal and) Tj ETQq0 0 0 rgBT /Overlock 10 Tf	1.9	28
57	Guidelines for the diagnosis and management of individuals with neurofibromatosis 1. Journal of Medical Genetics, 2006, 44, 81-88.	3.2	778
58	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
59	The heterogeneous nature of germline mutations in NF1 patients with malignant peripheral serve sheath tumours (MPNSTs). Human Mutation, 2006, 27, 716-716.	2.5	46
60	Large-Scale Molecular Comparison of Human Schwann Cells to Malignant Peripheral Nerve Sheath Tumor Cell Lines and Tissues. Cancer Research, 2006, 66, 2584-2591.	0.9	191
61	Gonosomal Mosaicism for a Nonsense Mutation (R1947X) in the NF1 Gene in Segmental Neurofibromatosis Type 1. Journal of Investigative Dermatology, 2005, 125, 463-466.	0.7	68
62	Neurofibromatosis Type 1: A Common Familial Cancer Syndrome. , 2004, , 285-310.		2
63	Functional analysis of polymorphic variation within the promoter and 5? untranslated region of the neurofibromatosis type 1 (NF1) gene. American Journal of Medical Genetics Part A, 2004, 131A, 227-231.	2.4	9
64	Characterization of the somatic mutational spectrum of the neurofibromatosis type 1 (NF1) gene in neurofibromatosis patients with benign and malignant tumors. Human Mutation, 2004, 23, 134-146.	2.5	97
65	Neurofibromatosis type 1: a common familial cancer syndrome. Methods in Molecular Medicine, 2004, 92, 285-310.	0.8	1
66	Detection of NF1 Mutations Utilizing the Protein Truncation Test (PTT). , 2003, 217, 315-328.		3
67	Three different pathological lesions in the NF1 gene originating de novo in a family with neurofibromatosis type 1. Human Genetics, 2003, 112, 12-17.	3.8	24
68	Neurofibromatosis typeÂ1 gene as a mutational target in a mismatch repair-deficient cell type. Human Genetics, 2003, 112, 117-123.	3.8	92
69	Molecular diagnosis of facioscapulohumeral muscular dystrophy. Expert Review of Molecular Diagnostics, 2002, 2, 160-171.	3.1	28
70	Evaluation of denaturing high performance liquid chromatography (DHPLC) for the mutational analysis of the neurofibromatosis type 1 (NF1) gene. Human Genetics, 2001, 109, 487-497.	3.8	71
71	Molecular analysis of the 5â€²-flanking region of the neurofibromatosis type 1 (NF1) gene: identification of five sequence variants. Clinical Genetics, 2001, 57, 221-224.	2.0	12
72	Multiple coronary artery aneurysms in a child with neurofibromatosis type 1. European Journal of Pediatrics, 2000, 159, 477-480.	2.7	30

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73	Hypermethylation of the neurofibromatosis type 1 (NF1) gene promoter is not a common event in the inactivation of the NF1 gene in NF1-specific tumours. Human Genetics, 2000, 107, 33-39.	3.8	13
74	Hypermethylation of the neurofibromatosis type 1 (NF1) gene promoter is not a common event in the inactivation of the NF1 gene in NF1-specific tumours. Human Genetics, 2000, 107, 33-39.	3.8	30
75	Mutation analysis of iduronate-2-sulphatase gene in 24 patients with Hunter syndrome: Characterisation of 6 novel mutations. Human Mutation, 1999, 14, 87-87.	2.5	11
76	Genetic Variation in the 3â€² Untranslated Region of the Neurofibromatosis 1 Gene: Application to Unequal Allelic Expression. Somatic Cell and Molecular Genetics, 1998, 24, 107-119.	0.7	18
77	Blind Analysis of Denaturing High-Performance Liquid Chromatography as a Tool for Mutation Detection. Genomics, 1998, 52, 44-49.	2.9	334
78	High-Throughput Screening for the Detection of Unknown Mutations: Improved Productivity Using Heteroduplex Analysis. BioTechniques, 1998, 25, 648-651.	1.8	6
79	Sex ratio and absence of uniparental disomy in spontaneous abortions with a normal karyotype. Clinical Genetics, 1998, 53, 258-261.	2.0	12
80	Molecular genetics of facioscapulohumeral muscular dystrophy (FSHD). Neuromuscular Disorders, 1997, 7, 55-62.	0.6	35
81	Characterization of six mutations in exon 37 of neurofibromatosis type 1 gene. , 1996, 67, 421-423.		26
82	Germinal mosaicism in facioscapulohumeral muscular dystrophy (FSHD). Muscle and Nerve, 1995, 18, S45-S49.	2.2	33
83	Phenotypic-genotypic correlation will assist genetic counseling in 4q35-facioscapulohumeral muscular dystrophy. Muscle and Nerve, 1995, 18, S103-S109.	2.2	55
84	Correlation between fragment size at D4F104S1 and age at onset or at wheelchair use, with a possible generational effect, accounts for much phenotypic variation in 4q35-facioscapulohumeral muscular dystrophy (FSHD). Human Molecular Genetics, 1995, 4, 951-958.	2.9	191
85	Prenatal diagnosis of X-linked hypohidrotic ectodermal dysplasia by linkage analysis. American Journal of Medical Genetics Part A, 1990, 35, 132-135.	2.4	20
86	Chorionic villus sampling for prenatal diagnosis in wales using DNA probesâ€”5 years' experience. Prenatal Diagnosis, 1990, 10, 593-603.	2.3	0
87	Lack of sampling site variation in chorion villus biopsy as assessed by DNA, enzymatic, morphological and cytogenetical analyses. Prenatal Diagnosis, 1987, 7, 119-127.	2.3	5
88	Seminal Acid Phosphatase in Relation to Fertility. Acta Obstetricia Et Gynecologica Scandinavica, 1986, 65, 49-52.	2.8	6
89	Antisperm Antibodies and Male Infertility. British Journal of Urology, 1984, 56, 531-536.	0.1	22