Hildegard Kehrer-Sawatzki

List of Publications by Year
in descending order

86 papers	5,635 citations	117625 34 h-index	82547 72 g-index
94 all docs	94 docs citations	94 times ranked	8800 citing authors

#	Article	IF	Citations
1	Evolutionary and Biomedical Insights from the Rhesus Macaque Genome. Science, 2007, 316, 222-234.	12.6	1,283
2	Where genotype is not predictive of phenotype: towards an understanding of the molecular basis of reduced penetrance in human inherited disease. Human Genetics, 2013, 132, 1077-1130.	3.8	528
3	PRC2 loss amplifies Ras-driven transcription and confers sensitivity to BRD4-based therapies. Nature, 2014, 514, 247-251.	27.8	386
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	Emerging genotypeâ€“phenotype relationships in patients with large NF1 deletions. Human Genetics, 2017, 136, 349-376.	3.8	163
6	Genes, mutations, and human inherited disease at the dawn of the age of personalized genomics. Human Mutation, 2010, 31, 631-655.	2.5	161
7	Screening 500 unselected neurofibromatosis 1 patients for deletions of the NF1 gene. Human Mutation, 2004, 23, 111-116.	2.5	159
8	Genomic rearrangements in inherited disease and cancer. Seminars in Cancer Biology, 2010, 20, 222-233.	9.6	140
9	Molecular Characterization and Gene Content of Breakpoint Boundaries in Patients with Neurofibromatosis Type 1 with 17q11.2 Microdeletions. American Journal of Human Genetics, 2001, 69, 516-527.	6.2	122
10	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
11	The molecular pathogenesis of schwannomatosis, a paradigm for the co-involvement of multiple tumour suppressor genes in tumorigenesis. Human Genetics, 2017, 136, 129-148.	3.8	106
12	On the sequence-directed nature of human gene mutation: The role of genomic architecture and the local DNA sequence environment in mediating gene mutations underlying human inherited disease. Human Mutation, 2011, 32, 1075-1099.	2.5	99
13	Understanding the recent evolution of the human genome: insights from human-chimpanzee genome comparisons. Human Mutation, 2007, 28, 99-130.	2.5	98
14	Conservation of hotspots for recombination in low-copy repeats associated with the NF1 microdeletion. Nature Genetics, 2006, 38, 1419-1423.	21.4	76
15	Molecular Characterization of the Pericentric Inversion That Causes Differences Between Chimpanzee Chromosome 19 and Human Chromosome 17. American Journal of Human Genetics, 2002, 71, 375-388.	6.2	73
16	Complex patterns of copy number variation at sites of segmental duplications: an important category of structural variation in the human genome. Human Genetics, 2006, 120, 270-284.	3.8	68
17	Molecular mechanisms of chromosomal rearrangement during primate evolution. Chromosome Research, 2008, 16, 41-56.	2.2	68
18	Dissecting loss of heterozygosity (LOH) in neurofibromatosis type 1-associated neurofibromas: Importance of copy neutral LOH. Human Mutation, 2011, 32, 78-90.	2.5	66

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19	Mutation and expression analysis of the KRIT1 gene associated with cerebral cavernous malformations (CCM1). Acta Neuropathologica, 2002, 104, 231-240.	7.7	65
20	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
21	Type 2 NF1 Deletions Are Highly Unusual by Virtue of the Absence of Nonallelic Homologous Recombination Hotspots and an Apparent Preference for Female Mitotic Recombination. American Journal of Human Genetics, 2007, 81, 1201-1220.	6.2	60
22	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
23	Gene synteny comparisons between different vertebrates provide new insights into breakage and fusion events during mammalian karyotype evolution. BMC Evolutionary Biology, 2009, 9, 84.	3.2	54
24	Complete physical map and gene content of the human NF1 tumor suppressor region in human and mouse. Genes Chromosomes and Cancer, 2003, 37, 111-120.	2.8	49
25	Structural divergence between the human and chimpanzee genomes. Human Genetics, 2007, 120, 759-778.	3.8	49
26	Segmental duplication associated with the human-specific inversion of chromosomeï¿½18: a further example of the impact of segmental duplications on karyotype and genome evolution in primates. Human Genetics, 2004, 115, 116-22.	3.8	47
27	Breakpoint analysis of the pericentric inversion distinguishing human chromosome 4 from the homologous chromosome in the chimpanzee (Pan troglodytes). Human Mutation, 2005, 25, 45-55.	2.5	47
28	Childhood overgrowth in patients with common NF1 microdeletions. European Journal of Human Genetics, 2005, 13, 883-888.	2.8	46
29	Independent intrachromosomal recombination events underlie the pericentric inversions of chimpanzee and gorilla chromosomes homologous to human chromosome 16. Genome Research, 2005, 15, 1232-1242.	5.5	42
30	Mechanisms of Loss of Heterozygosity in Neurofibromatosis Type 1-Associated Plexiform Neurofibromas. Journal of Investigative Dermatology, 2009, 129, 615-621.	0.7	42
31	A novel third type of recurrent NF1 microdeletion mediated by nonallelic homologous recombination between LRRC37B-containing low-copy repeats in 17q11.2. Human Mutation, 2010, 31, 742-751.	2.5	42
32	Extensively high load of internal tumors determined by whole body MRI scanning in a patient with neurofibromatosis type 1 and a non-LCR-mediated 2-Mb deletion in 17q11.2. Human Genetics, 2005, 116, 466-475.	3.8	37
33	Intrachromosomal mitotic nonallelic homologous recombination is the major molecular mechanism underlying type-2 NF1 deletions. Human Mutation, 2010, 31, 1163-1173.	2.5	36
34	Whole exome sequencing reveals that the majority of schwannomatosis cases remain unexplained after excluding SMARCB1 and LZTR1 germline variants. Acta Neuropathologica, 2014, 128, 449-452.	7.7	36
35	Identification of large-scale human-specific copy number differences by inter-species array comparative genomic hybridization. Human Genetics, 2006, 119, 185-198.	3.8	35
36	Monozygotic twins discordant for neurofibromatosis type 1 due to a postzygotic NF1 gene mutation. Human Mutation, 2011, 32, E2134-E2147.	2.5	34

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37	What a difference copy number variation makes. BioEssays, 2007, 29, 311-313.	2.5	33
38	Somatic mosaicism of a greater than 1.7-Mb deletion of genomic DNA involving the entireNF1 gene as verified by FISH: Further evidence for a contiguous gene syndrome in 17q11.2. American Journal of Medical Genetics Part A, 1999, 87, 12-16.	2.4	31
39	Cruciform-forming inverted repeats appear to have mediated many of the microinversions that distinguish the human and chimpanzee genomes. Chromosome Research, 2009, 17, 469-483.	2.2	31
40	Exploring the potential relevance of human-specific genes to complex disease. Human Genomics, 2011, 5, 99.	2.9	30
41	Internal tumor burden in neurofibromatosis Type I patients with large NF1 deletions. Genes Chromosomes and Cancer, 2012, 51, 447-451.	2.8	30
42	No correlation between NF1 mutation position and risk of optic pathway glioma in 77 unrelated NF1 patients. Human Genetics, 2016, 135, 469-475.	3.8	29
43	Challenges in the diagnosis of neurofibromatosis type 1 (NF1) in young children facilitated by means of revised diagnostic criteria including genetic testing for pathogenic NF1 gene variants. Human Genetics, 2022, 141, 177-191.	3.8	29
44	Molecular characterization of the pericentric inversion of chimpanzee chromosome 11 homologous to human chromosome 9. Genomics, 2005, 85, 542-550.	2.9	28
45	Characterization of the nonallelic homologous recombination hotspot PRS3 associated with type-3NF1deletions. Human Mutation, 2012, 33, 372-383.	2.5	28
46	Molecular characterisation of the pericentric inversion that distinguishes human chromosome 5 from the homologous chimpanzee chromosome. Human Genetics, 2005, 117, 168-176.	3.8	27
47	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
48	Multifocal nerve lesions and LZTR1 germline mutations in segmental schwannomatosis. Annals of Neurology, 2016, 80, 625-628.	5.3	25
49	Phenotypic and genotypic overlap between mosaic NF2 and schwannomatosis in patients with multiple non-intradermal schwannomas. Human Genetics, 2018, 137, 543-552.	3.8	25
50	Tissue-specific differences in the proportion of mosaic large NF1 deletions are suggestive of a selective growth advantage of hematopoietic del(+/â˜) stem cells. Human Mutation, 2012, 33, 541-550.	2.5	23
51	Copy number variations in the NF1 gene region are infrequent and do not predispose to recurrent type-1 deletions. European Journal of Human Genetics, 2008, 16, 572-580.	2.8	22
52	Absence of cutaneous neurofibromas in an NF1 patient with an atypical deletion partially overlapping the common 1.4 Mb microdeleted region. American Journal of Medical Genetics, Part A, 2008, 146A, 691-699.	1.2	21
53	The chimpanzee-specific pericentric inversions that distinguish humans and chimpanzees have identical breakpoints in Pan troglodytes and Pan paniscus. Genomics, 2006, 87, 39-45.	2.9	20
54	Dissecting the clinical phenotype associated with mosaic type-2 NF1 microdeletions. Neurogenetics, 2012, 13, 229-236.	1.4	20

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55	The effect of pregnancy on growth-dynamics of neurofibromas in Neurofibromatosis type 1. PLoS ONE, 2020, 15, e0232031.	2.5	18
56	Polymorphic micro-inversions contribute to the genomic variability of humans and chimpanzees. Human Genetics, 2006, 119, 103-112.	3.8	17
57	Characterization of the human lineage-specific pericentric inversion that distinguishes human chromosome 1 from the homologous chromosomes of the great apes. Human Genetics, 2006, 120, 126-138.	3.8	17
58	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
59	Interstitial deletion del(10)(q25.2q25.3 â^¼ 26.11)â€"case report and review of the literature. Prenatal Diagnosis, 2005, 25, 954-959.	2.3	16
60	Fine mapping of meiotic NAHR-associated crossovers causing largeNF1deletions. Human Molecular Genetics, 2016, 25, 484-496.	2.9	15
61	A gene conversion hotspot in the human growth hormone (GH1) gene promoter. Human Mutation, 2009, 30, 239-247.	2.5	13
62	Coâ€œoccurrence of schwannomatosis and rhabdoid tumor predisposition syndrome 1. Molecular Genetics & Genomic Medicine, 2018, 6, 627-637.	1.2	13
63	Extreme clustering of type-1 NF1 deletion breakpoints co-locating with G-quadruplex forming sequences. Human Genetics, 2018, 137, 511-520.	3.8	13
64	Triangulation of the human, chimpanzee, and Neanderthal genome sequences identifies potentially compensated mutations. Human Mutation, 2010, 31, 1286-1293.	2.5	12
65	Delineating the Hemostaseome as an aid to individualize the analysis of the hereditary basis of thrombotic and bleeding disorders. Human Genetics, 2011, 130, 149-166.	3.8	12
66	Delineation of the clinical phenotype associated with non-mosaic type-2 NF1 deletions: two case reports. Journal of Medical Case Reports, 2011, 5, 577.	0.8	12
67	Pronounced maternal parent-of-origin bias for type-1 NF1 microdeletions. Human Genetics, 2018, 137, 365-373.	3.8	12
68	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
69	Clinical characterization of children and adolescents with NF1 microdeletions. Child's Nervous System, 2020, 36, 2297-2310.	1.1	12
70	Classification of NF1 microdeletions and its importance for establishing genotype/phenotype correlations in patients with NF1 microdeletions. Human Genetics, 2021, 140, 1635-1649.	3.8	12
71	Non-coding RNA ANRIL and the number of plexiform neurofibromas in patients with NF1microdeletions. BMC Medical Genetics, 2012, 13, 98.	2.1	11
72	NF1 Microdeletions and Their Underlying Mutational Mechanisms. , 2012, , 187-209.		10

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73	Atypical NF1 Microdeletions: Challenges and Opportunities for Genotype/Phenotype Correlations in Patients with Large NF1 Deletions. Genes, 2021, 12, 1639.	2.4	10
74	Extended runs of homozygosity at 17q11.2: an association with type-2NF1deletions?. Human Mutation, 2010, 31, 325-334.	2.5	9
75	Femoral facial syndrome associated with a de novo complex chromosome 2q37 rearrangement. American Journal of Medical Genetics, Part A, 2016, 170, 1202-1207.	1.2	9
76	Consideration of the haplotype diversity at nonallelic homologous recombination hotspots improves the precision of rearrangement breakpoint identification. Human Mutation, 2017, 38, 1711-1722.	2.5	9
77	Determination of the mutant allele frequency in patients with neurofibromatosis type 2 and somatic mosaicism by means of deep sequencing. Genes Chromosomes and Cancer, 2015, 54, 482-488.	2.8	8
78	Identification of LargeNF1Duplications Reciprocal to NAHR-Mediated Type-1NF1Deletions. Human Mutation, 2014, 35, 1469-1475.	2.5	7
79	C-Fiber Loss as a Possible Cause of Neuropathic Pain in Schwannomatosis. International Journal of Molecular Sciences, 2020, 21, 3569.	4.1	5
80	Population-specific differences in gene conversion patterns between human SUZ12 and SUZ12P are indicative of the dynamic nature of interparalog gene conversion. Human Genetics, 2014, 133, 383-401.	3.8	3
81	SWI/SNF-Komplex-assoziierte Tumordispositions-Syndrome. Medizinische Genetik, 2017, 29, 296-305.	0.2	3
82	Null phenotype of neurofibromatosis type 1 in a carrier of a heterozygous atypical NF1 deletion due to mosaicism. Human Mutation, 2020, 41, 1226-1231.	2.5	3
83	Distinct sequence features underlie microdeletions and gross deletions in the human genome. Human Mutation, 2022, 43, 328-346.	2.5	3
84	The NF1 microdeletion syndrome: early genetic diagnosis facilitates the management of a clinically defined disease. JDDG - Journal of the German Society of Dermatology, 2022, 20, 273-277.	0.8	3
85	Neurofibromatosis Type 1 Without Neurofibromas: Genotype-Phenotype Correlations in NF1. Human Mutation, 2015, 36, v-v.	2.5	2
86	Das NF1 â€Mikrodeletionsâ€Syndrom: Die frÃ¼hzeitige genetische Diagnose erleichtert den Umgang mit einer klinisch definierten Erkrankung. JDDG - Journal of the German Society of Dermatology, 2022, 20, 273-278.	0.8	0