Magnus NordenskjÃ¶ld

List of Publications by Year
in descending order

83 papers	6,383 citations	136950 32 h-index	69250 77 g-index
85 all docs	85 docs citations	85 times ranked	8451 citing authors

#	Article	IF	Citations
1	Multiple endocrine neoplasia type 1 gene maps to chromosome 11 and is lost in insulinoma. Nature, 1988, 332, 85-87.	27.8	1,007
2	Cytogenetic analysis by chromosome painting using dopâ€pcr amplified flowâ€sorted chromosomes. Genes Chromosomes and Cancer, 1992, 4, 257-263.	2.8	555
3	Evidence for the complete inactivation of the NF2 gene in the majority of sporadic meningiomas. Nature Genetics, 1994, 6, 180-184.	21.4	514
4	Targeted sequencing identifies 91 neurodevelopmental-disorder risk genes with autism and developmental-disability biases. Nature Genetics, 2017, 49, 515-526.	21.4	443
5	Genetic mapping of a second locus predisposing to hereditary nonâ€“polyposis colon cancer. Nature Genetics, 1993, 5, 279-282.	21.4	408
6	Clonal culturing of human embryonic stem cells on laminin-521/E-cadherin matrix in defined and xeno-free environment. Nature Communications, 2014, 5, 3195.	12.8	248
7	Birt-Hogg-DubÃ© syndrome: mapping of a novel hereditary neoplasia gene to chromosome 17p12-q11.2. Oncogene, 2001, 20, 5239-5242.	5.9	199
8	Increased frequency of sister chromatid exchanges in cigarette smokers. Hereditas, 1978, 88, 147-149.	1.4	169
9	Genetic basis of PD-L1 overexpression in diffuse large B-cell lymphomas. Blood, 2016, 127, 3026-3034.	1.4	168
10	Hotspots of missense mutation identify neurodevelopmental disorder genes and functional domains. Nature Neuroscience, 2017, 20, 1043-1051.	14.8	152
11	Disruption of POGZ Is Associated with Intellectual Disability and Autism Spectrum Disorders. American Journal of Human Genetics, 2016, 98, 541-552.	6.2	132
12	Integration of whole genome sequencing into a healthcare setting: high diagnostic rates across multiple clinical entities in 3219 rare disease patients. Genome Medicine, 2021, 13, 40.	8.2	116
13	Loss of heterozygosity in malignant gliomas involves at least three distinct regions on chromosome 10. Human Genetics, 1993, 92, 169-74.	3.8	115
14	Deletions on chromosome 22 in sporadic meningioma. Genes Chromosomes and Cancer, 1994, 10, 122-130.	2.8	115
15	Fifty probands with extra structurally abnormal chromosomes characterized by fluorescence in situ hybridization. American Journal of Medical Genetics Part A, 1995, 55, 85-94.	2.4	114
16	Neu-Laxova Syndrome Is a Heterogeneous Metabolic Disorder Caused by Defects in Enzymes of the L-Serine Biosynthesis Pathway. American Journal of Human Genetics, 2014, 95, 285-293.	6.2	110
17	Large-scale targeted sequencing identifies risk genes for neurodevelopmental disorders. Nature Communications, 2020, 11, 4932.	12.8	105
18	From cytogenetics to cytogenomics: whole-genome sequencing as a first-line test comprehensively captures the diverse spectrum of disease-causing genetic variation underlying intellectual disability. Genome Medicine, 2019, 11, 68.	8.2	88

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19	Gain-of-function mutation of microRNA-140 in human skeletal dysplasia. Nature Medicine, 2019, 25, 583-590.	30.7	86
20	A high degree of aneuploidy in frozen-thawed human preimplantation embryos. Human Genetics, 1999, 104, 376.	3.8	78
21	Tumorigenesis in colorectal tumors from patients with hereditary non-polyposis colorectal cancer. Human Genetics, 1997, 101, 51-55.	3.8	75
22	Highly abnormal cleavage divisions in preimplantation embryos from translocation carriers. Prenatal Diagnosis, 2000, 20, 1038-1047.	2.3	74
23	Hemophagocytic lymphohistiocytosis in 2 patients with underlying IFN-Î³ receptor deficiency. Journal of Allergy and Clinical Immunology, 2015, 135, 1638-1641.e5.	2.9	69
24	Genomic alterations in human breast carcinomas. Genes Chromosomes and Cancer, 1990, 2, 191-197.	2.8	64
25	Analysis of short stature homeobox-containing gene (SHOX) and auxological phenotype in dyschondrosteosis and isolated Madelung deformity. Human Genetics, 2001, 109, 551-558.	3.8	60
26	Whole-Genome Sequencing of Cytogenetically Balanced Chromosome Translocations Identifies Potentially Pathological Gene Disruptions and Highlights the Importance of Microhomology in the Mechanism of Formation. Human Mutation, 2017, 38, 180-192.	2.5	58
27	Rare deleterious mutations of HNRNP genes result in shared neurodevelopmental disorders. Genome Medicine, 2021, 13, 63.	8.2	50
28	Distinct deleted regions on chromosome segment 16q23-24 associated with metastases in prostate cancer. Genes Chromosomes and Cancer, 1999, 24, 175-182.	2.8	49
29	Spectrum of Atypical Clinical Presentations in Patients with Biallelic PRF1 Missense Mutations. Pediatric Blood and Cancer, 2015, 62, 2094-2100.	1.5	38
30	Targeted high-throughput sequencing for genetic diagnostics of hemophagocytic lymphohistiocytosis. Genome Medicine, 2015, 7, 130.	8.2	37
31	CTNND2â€”a candidate gene for reading problems and mild intellectual disability. Journal of Medical Genetics, 2015, 52, 111-122.	3.2	35
32	Disruptive variants of CSDE1 associate with autism and interfere with neuronal development and synaptic transmission. Science Advances, 2019, 5, eaax2166.	10.3	35
33	Sporadic cardiac myxomas and tumors from patients with Carney complex are not associated with activating mutations of the GsÎ± gene. Human Genetics, 1996, 98, 185-188.	3.8	33
34	Maternal Filaggrin Mutations Increase the Risk of Atopic Dermatitis in Children: An Effect Independent of Mutation Inheritance. PLoS Genetics, 2015, 11, e1005076.	3.5	33
35	Linkage and association to candidate regions in Swedish atopic dermatitis families. Human Genetics, 2001, 109, 129-135.	3.8	32
36	Clinical outcome of treatment cycles using preimplantation genetic diagnosis for structural chromosomal abnormalities. Prenatal Diagnosis, 2001, 21, 781-787.	2.3	31

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37	Whole-exome sequencing of Ethiopian patients with ichthyosis vulgaris and atopic dermatitis. Journal of Allergy and Clinical Immunology, 2015, 136, 507-509.e19.	2.9	30
38	Missense Variants in the Histone Acetyltransferase Complex Component Gene TRRAP Cause Autism and Syndromic Intellectual Disability. American Journal of Human Genetics, 2019, 104, 530-541.	6.2	30
39	1p and 3p deletions in meningiomas without detectable aberrations of chromosome 22 identified by comparative genomic hybridization. , 1997, 20, 419-424.		29
40	Genomic screening in rare disorders: New mutations and phenotypes, highlighting ALG14 as a novel cause of severe intellectual disability. Clinical Genetics, 2018, 94, 528-537.	2.0	29
41	Positive Attitudes towards Non-Invasive Prenatal Testing (NIPT) in a Swedish Cohort of 1,003 Pregnant Women. PLoS ONE, 2016, 11, e0156088.	2.5	28
42	Disruption of tubular Flcn expression as a mouse model for renal tumor induction. Kidney International, 2015, 88, 1057-1069.	5.2	27
43	Amplification-free sequencing of cell-free DNA for prenatal non-invasive diagnosis of chromosomal aberrations. Genomics, 2015, 105, 150-158.	2.9	26
44	Differences in Granule Morphology yet Equally Impaired Exocytosis among Cytotoxic T Cells and NK Cells from Chediakâ€“Higashi Syndrome Patients. Frontiers in Immunology, 2017, 8, 426.	4.8	26
45	Four separate regions on chromosome 17 show loss of heterozygosity in familial breast carcinomas. Human Genetics, 1993, 91, 6-12.	3.8	25
46	Bilateral multiple renal oncocytomas and cysts associated with a constitutional translocation (8;9)(q24.1;q34.3) and a rare constitutional VHL missense substitution. , 1998, 21, 260-264.		24
47	Identification of putative pathogenic single nucleotide variants (SNVs) in genes associated with heart disease in 290 cases of stillbirth. PLoS ONE, 2019, 14, e0210017.	2.5	24
48	A KCNC3 mutation causes a neurodevelopmental, non-progressive SCA13 subtype associated with dominant negative effects and aberrant EGFR trafficking. PLoS ONE, 2017, 12, e0173565.	2.5	22
49	A RAB27A 5â€² untranslated region structural variant associated with late-onset hemophagocytic lymphohistiocytosis and normal pigmentation. Journal of Allergy and Clinical Immunology, 2018, 142, 317-321.e8.	2.9	22
50	Early prenatal diagnosis of the ICF syndrome. Prenatal Diagnosis, 2000, 20, 828-831.	2.3	20
51	An N-Terminal Missense Mutation in STX11 Causative of FHL4 Abrogates Syntaxin-11 Binding to Munc18-2. Frontiers in Immunology, 2014, 4, 515.	4.8	20
52	Combined newborn screening for familial hemophagocytic lymphohistiocytosis and severe T- and B-cell immunodeficiencies. Journal of Allergy and Clinical Immunology, 2014, 134, 226-228.e7.	2.9	20
53	Chromatin interactions in differentiating keratinocytes reveal novel atopic dermatitisâ€“ and psoriasis-associated genes. Journal of Allergy and Clinical Immunology, 2021, 147, 1742-1752.	2.9	18
54	An integrative proteomics method identifies a regulator of translation during stem cell maintenance and differentiation. Nature Communications, 2021, 12, 6558.	12.8	16

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55	Exclusion of the 13-kDa rapamycin binding protein gene (FKBP2) as a candidate gene for multiple endocrine neoplasia type 1. Human Genetics, 1995, 95, 455-8.	3.8	14
56	Identification of numerical and structural chromosome aberrations in 15 high hyperdiploid childhood acute lymphoblastic leukemias using spectral karyotyping. European Journal of Haematology, 2001, 66, 297-304.	2.2	13
57	Novel Features and Abnormal Pattern of Cerebral Glucose Metabolism in Spinocerebellar Ataxia 19. Cerebellum, 2018, 17, 465-476.	2.5	13
58	Cell-free tumour DNA analysis detects copy number alterations in gastro-oesophageal cancer patients. PLoS ONE, 2021, 16, e0245488.	2.5	13
59	Hereditary breast cancer in sweden: a predominance of maternally inherited cases. Breast Cancer Research and Treatment, 1992, 24, 159-165.	2.5	12
60	Finnish mutations in Swedish HNPCC families. Nature Medicine, 1995, 1, 1104-1104.	30.7	12
61	Mapping of a new MAP kinase activated protein kinase gene (3PK) to human chromosome band 3p21.2 and ordering of 3PK and two cosmid markers in the 3p22â€“p21 tumour-suppressor region by two-colour fluorescencein situ hybridization. Chromosome Research, 1996, 4, 310-313.	2.2	11
62	Hereditary Cancer. Acta OncolÃ³gica, 1999, 38, 439-447.	1.8	11
63	High-resolution detection of chromosomal rearrangements in leukemias through mate pair whole genome sequencing. PLoS ONE, 2018, 13, e0193928.	2.5	11
64	Sublocalization of a locus at 3p21.3?23 predisposing to hereditary nonpolyposis colon cancer. Human Genetics, 1994, 94, 210-4.	3.8	10
65	Characterization of add(I)(p36) in non-hodgkin lymphomas by fluorescence in situ hybridization. Genes Chromosomes and Cancer, 1995, 13, 34-39.	2.8	10
66	Detailed gene dose analysis reveals recurrent focal gene deletions in pediatric B-cell precursor acute lymphoblastic leukemia. Leukemia and Lymphoma, 2016, 57, 2161-2170.	1.3	10
67	Intragenic Copy Number Variation in the Filaggrin Gene in Ethiopian Patients with Atopic Dermatitis. Pediatric Dermatology, 2017, 34, e140-e141.	0.9	10
68	Knowledge and Attitudes Regarding Nonâ€Invasive Prenatal Testing (NIPT) and Preferences for Risk Information among High School Students in Sweden. Journal of Genetic Counseling, 2017, 26, 447-454.	1.6	10
69	Tight linkage between the Beckwith-Wiedemann syndrome and a microsatellite marker for the TH locus. Human Genetics, 1993, 92, 296-8.	3.8	9
70	Family Screening in Multiple Endocrine Type 1 (MEN 1) Neoplasia. Annals of Medicine, 1994, 26, 191-198.	3.8	9
71	Exclusion of the phosphoinositide-specific phospholipase C?3 (PLCB3) gene as a candidate for multiple endocrine neoplasia type 1. Human Genetics, 1996, 99, 130-132.	3.8	8
72	Haploinsufficiency of UNC13D increases the risk of lymphoma. Cancer, 2019, 125, 1848-1854.	4.1	8

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73	Ataxia in Patients With Bi-Allelic NFASC Mutations and Absence of Full-Length NF186. Frontiers in Genetics, 2019, 10, 896.	2.3	7
74	Overexpression of chromatin remodeling and tyrosine kinase genes in iAMP21-positive acute lymphoblastic leukemia. Leukemia and Lymphoma, 2020, 61, 604-613.	1.3	7
75	Severe congenital neutropeniaâ€associated JAGN1 mutations unleash a calpainâ€dependent cell death programme in myeloid cells. British Journal of Haematology, 2021, 192, 200-211.	2.5	7
76	Kostmann disease and other forms of severe congenital neutropenia. Acta Paediatrica, International Journal of Paediatrics, 2021, 110, 2912-2920.	1.5	7
77	A constitutional mutation within the retinoblastoma gene detected by PFGE. Clinical Genetics, 1994, 45, 5-10.	2.0	6
78	A retrospective two centre study of Birt-Hogg-DubÃ© syndrome reveals a pathogenic founder mutation in FLCN in the Swedish population. PLoS ONE, 2022, 17, e0264056.	2.5	5
79	Diagnostic challenges for a novel SH2D1A mutation associated with Xâ€linked lymphoproliferative disease. Pediatric Blood and Cancer, 2020, 67, e28184.	1.5	4
80	Germline mutations detected in the von Hippel-Lindau disease tumor suppressor gene by southern blot and direct genomic DNA sequencing. Human Mutation, 1998, 11, S31-S33.	2.5	3
81	Characterization of Stem-Like Cells in Mucoepidermoid Tracheal Paediatric Tumor. PLoS ONE, 2014, 9, e107712.	2.5	2
82	Pathogenenic variant in the COL2A1 gene is associated with Spondyloepiphyseal dysplasia type Stanescu. American Journal of Medical Genetics, Part A, 2016, 170, 266-269.	1.2	2
83	Distinct deleted regions on chromosome segment 16q23â€“24 associated with metastases in prostate cancer. Genes Chromosomes and Cancer, 1999, 24, 175-182.	2.8	2