Magnus Nordenskjld

List of Publications by Year in Descending Order

83 papers	4,938 citations	28 h-index	70 g-index
85 ext. papers	5,702 ext. citations	8.8 avg, IF	4.46 L-index

#	Paper	IF	Citations
83	A retrospective two centre study of Birt-Hogg-Dubé syndrome reveals a pathogenic founder mutation in FLCN in the Swedish population.. *PLoS ONE*, **2022**, 17, e0264056	3.7	0
82	An integrative proteomics method identifies a regulator of translation during stem cell maintenance and differentiation. *Nature Communications*, **2021**, 12, 6558	17.4	3
81	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	14.4	22
80	Rare deleterious mutations of HNRNP genes result in shared neurodevelopmental disorders. *Genome Medicine*, **2021**, 13, 63	14.4	9
79	Kostmann disease and other forms of severe congenital neutropenia. *Acta Paediatrica, International Journal of Paediatrics*, **2021**, 110, 2912-2920	3.1	1
78	Chromatin interactions in differentiating keratinocytes reveal novel atopic dermatitis- and psoriasis-associated genes. *Journal of Allergy and Clinical Immunology*, **2021**, 147, 1742-1752	11.5	1
77	Severe congenital neutropenia-associated JAGN1 mutations unleash a calpain-dependent cell death programme in myeloid cells. *British Journal of Haematology*, **2021**, 192, 200-211	4.5	3
76	Cell-free tumour DNA analysis detects copy number alterations in gastro-oesophageal cancer patients. *PLoS ONE*, **2021**, 16, e0245488	3.7	3
75	Diagnostic challenges for a novel SH2D1A mutation associated with X-linked lymphoproliferative disease. *Pediatric Blood and Cancer*, **2020**, 67, e28184	3	2
74	Large-scale targeted sequencing identifies risk genes for neurodevelopmental disorders. *Nature Communications*, **2020**, 11, 4932	17.4	25
73	Overexpression of chromatin remodeling and tyrosine kinase genes in iAMP21-positive acute lymphoblastic leukemia. *Leukemia and Lymphoma*, **2020**, 61, 604-613	1.9	2
72	Disruptive variants of associate with autism and interfere with neuronal development and synaptic transmission. *Science Advances*, **2019**, 5, eaax2166	14.3	16
71	Haploinsufficiency of UNC13D increases the risk of lymphoma. *Cancer*, **2019**, 125, 1848-1854	6.4	4
70	Gain-of-function mutation of microRNA-140 in human skeletal dysplasia. *Nature Medicine*, **2019**, 25, 583-590	30.5	38
69	Ataxia in Patients With Bi-Allelic Mutations and Absence of Full-Length NF186. *Frontiers in Genetics*, **2019**, 10, 896	4.5	4
68	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	14.4	44
67	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	11	17

66	Identification of putative pathogenic single nucleotide variants (SNVs) in genes associated with heart disease in 290 cases of stillbirth. *PLoS ONE*, **2019**, 14, e0210017	3.7	16
65	A RAB27A 5Tuntranslated region structural variant associated with late-onset hemophagocytic lymphohistiocytosis and normal pigmentation. *Journal of Allergy and Clinical Immunology*, **2018**, 142, 317-321.e8	11.5	16
64	Novel Features and Abnormal Pattern of Cerebral Glucose Metabolism in Spinocerebellar Ataxia 19. *Cerebellum*, **2018**, 17, 465-476	4.3	7
63	High-resolution detection of chromosomal rearrangements in leukemias through mate pair whole genome sequencing. *PLoS ONE*, **2018**, 13, e0193928	3.7	9
62	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	4	14
61	Targeted sequencing identifies 91 neurodevelopmental-disorder risk genes with autism and developmental-disability biases. *Nature Genetics*, **2017**, 49, 515-526	36.3	283
60	Hotspots of missense mutation identify neurodevelopmental disorder genes and functional domains. *Nature Neuroscience*, **2017**, 20, 1043-1051	25.5	94
59	Intragenic Copy Number Variation in the Filaggrin Gene in Ethiopian Patients with Atopic Dermatitis. *Pediatric Dermatology*, **2017**, 34, e140-e141	1.9	8
58	A KCNC3 mutation causes a neurodevelopmental, non-progressive SCA13 subtype associated with dominant negative effects and aberrant EGFR trafficking. *PLoS ONE*, **2017**, 12, e0173565	3.7	18
57	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	4.7	40
56	Knowledge and Attitudes Regarding Non-Invasive Prenatal Testing (NIPT) and Preferences for Risk Information among High School Students in Sweden. *Journal of Genetic Counseling*, **2017**, 26, 447-454	2.5	4
55	Differences in Granule Morphology yet Equally Impaired Exocytosis among Cytotoxic T Cells and NK Cells from Chediak-Higashi Syndrome Patients. *Frontiers in Immunology*, **2017**, 8, 426	8.4	19
54	Disruption of POGZ Is Associated with Intellectual Disability and Autism Spectrum Disorders. *American Journal of Human Genetics*, **2016**, 98, 541-552	11	89
53	Positive Attitudes towards Non-Invasive Prenatal Testing (NIPT) in a Swedish Cohort of 1,003 Pregnant Women. *PLoS ONE*, **2016**, 11, e0156088	3.7	19
52	Detailed gene dose analysis reveals recurrent focal gene deletions in pediatric B-cell precursor acute lymphoblastic leukemia. *Leukemia and Lymphoma*, **2016**, 57, 2161-70	1.9	9
51	Genetic basis of PD-L1 overexpression in diffuse large B-cell lymphomas. *Blood*, **2016**, 127, 3026-34	2.2	126
50	Pathogenenic variant in the COL2A1 gene is associated with Spondyloepiphyseal dysplasia type Stanescu. *American Journal of Medical Genetics, Part A*, **2016**, 170A, 266-9	2.5	2
49	Whole-exome sequencing of Ethiopian patients with ichthyosis vulgaris and atopic dermatitis. *Journal of Allergy and Clinical Immunology*, **2015**, 136, 507-9.e19	11.5	23

48	Maternal filaggrin mutations increase the risk of atopic dermatitis in children: an effect independent of mutation inheritance. *PLoS Genetics*, **2015**, 11, e1005076	6	25
47	Disruption of tubular Flcn expression as a mouse model for renal tumor induction. *Kidney International*, **2015**, 88, 1057-69	9.9	20
46	CTNND2-a candidate gene for reading problems and mild intellectual disability. *Journal of Medical Genetics*, **2015**, 52, 111-22	5.8	24
45	Targeted high-throughput sequencing for genetic diagnostics of hemophagocytic lymphohistiocytosis. *Genome Medicine*, **2015**, 7, 130	14.4	28
44	Spectrum of Atypical Clinical Presentations in Patients with Biallelic PRF1 Missense Mutations. *Pediatric Blood and Cancer*, **2015**, 62, 2094-100	3	25
43	Hemophagocytic lymphohistiocytosis in 2 patients with underlying IFN- receptor deficiency. *Journal of Allergy and Clinical Immunology*, **2015**, 135, 1638-41	11.5	64
42	Amplification-free sequencing of cell-free DNA for prenatal non-invasive diagnosis of chromosomal aberrations. *Genomics*, **2015**, 105, 150-8	4.3	22
41	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, 285-93	11	82
40	Clonal culturing of human embryonic stem cells on laminin-521/E-cadherin matrix in defined and xeno-free environment. *Nature Communications*, **2014**, 5, 3195	17.4	183
39	Combined newborn screening for familial hemophagocytic lymphohistiocytosis and severe T- and B-cell immunodeficiencies. *Journal of Allergy and Clinical Immunology*, **2014**, 134, 226-8	11.5	14
38	Characterization of stem-like cells in mucoepidermoid tracheal paediatric tumor. *PLoS ONE*, **2014**, 9, e107712	3.7	2
37	An N-Terminal Missense Mutation in STX11 Causative of FHL4 Abrogates Syntaxin-11 Binding to Munc18-2. *Frontiers in Immunology*, **2014**, 4, 515	8.4	18
36	Analysis of short stature homeobox-containing gene (SHOX) and auxological phenotype in dyschondrosteosis and isolated Madelung deformity. *Human Genetics*, **2001**, 109, 551-8	6.3	51
35	Linkage and association to candidate regions in Swedish atopic dermatitis families. *Human Genetics*, **2001**, 109, 129-35	6.3	31
34	Clinical outcome of treatment cycles using preimplantation genetic diagnosis for structural chromosomal abnormalities. *Prenatal Diagnosis*, **2001**, 21, 781-7	3.2	27
33	Birt-Hogg-Dub syndrome: mapping of a novel hereditary neoplasia gene to chromosome 17p12-q11.2. *Oncogene*, **2001**, 20, 5239-42	9.2	162
32	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	3.8	13
31	Early prenatal diagnosis of the ICF syndrome. *Prenatal Diagnosis*, **2000**, 20, 828-31	3.2	18

30	Highly abnormal cleavage divisions in preimplantation embryos from translocation carriers. *Prenatal Diagnosis*, **2000**, 20, 1038-1047	3.2	67
29	Hereditary cancer. *Acta Oncologica*, **1999**, 38, 439-47	3.2	10
28	A high degree of aneuploidy in frozen-thawed human preimplantation embryos. *Human Genetics*, **1999**, 104, 376-82	6.3	62
27	Distinct deleted regions on chromosome segment 16q23.4 associated with metastases in prostate cancer. *Genes Chromosomes and Cancer*, **1999**, 24, 175-182	5	44
26	Distinct deleted regions on chromosome segment 16q23.4 associated with metastases in prostate cancer **1999**, 24, 175		2
25	Germline mutations detected in the von Hippel-Lindau disease tumor suppressor gene by Southern blot and direct genomic DNA sequencing. *Human Mutation*, **1998**, Suppl 1, S31-3	4.7	2
24	Bilateral multiple renal oncocytomas and cysts associated with a constitutional translocation (8;9)(q24.1;q34.3) and a rare constitutional VHL missense substitution. *Genes Chromosomes and Cancer*, **1998**, 21, 260-4	5	18
23	Tumorigenesis in colorectal tumors from patients with hereditary non-polyposis colorectal cancer. *Human Genetics*, **1997**, 101, 51-5	6.3	67
22	1p and 3p deletions in meningiomas without detectable aberrations of chromosome 22 identified by comparative genomic hybridization **1997**, 20, 419-424		22
21	Exclusion of the phosphoinositide-specific phospholipase C beta 3 (PLCB3) gene as a candidate for multiple endocrine neoplasia type 1. *Human Genetics*, **1997**, 99, 130-2	6.3	5
20	Sporadic cardiac myxomas and tumors from patients with Carney complex are not associated with activating mutations of the Gs alpha gene. *Human Genetics*, **1996**, 98, 185-8	6.3	30
19	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 3p22-p21 tumour-suppressor region by two-colour fluorescence in situ hybridization. *Chromosome Research*, **1996**, 4, 310-3	4.4	10
18	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	6.3	11
17	Finnish mutations in Swedish HNPCC families. *Nature Medicine*, **1995**, 1, 1104	50.5	11
16	Fifty probands with extra structurally abnormal chromosomes characterized by fluorescence in situ hybridization. *American Journal of Medical Genetics Part A*, **1995**, 55, 85-94		109
15	Characterization of add(1)(p36) in non-Hodgkin lymphomas by fluorescence in situ hybridization. *Genes Chromosomes and Cancer*, **1995**, 13, 34-9	5	9
14	A constitutional mutation within the retinoblastoma gene detected by PFGE. *Clinical Genetics*, **1994**, 45, 5-10	4	5
13	Family screening in multiple endocrine neoplasia type 1 (MEN 1). *Annals of Medicine*, **1994**, 26, 191-8	1.5	8

12	Sublocalization of a locus at 3p21.3-23 predisposing to hereditary nonpolyposis colon cancer. *Human Genetics*, **1994**, 94, 210-4	6.3	8
11	Deletions on chromosome 22 in sporadic meningioma. *Genes Chromosomes and Cancer*, **1994**, 10, 122-30	5	99
10	Evidence for the complete inactivation of the NF2 gene in the majority of sporadic meningiomas. *Nature Genetics*, **1994**, 6, 180-4	36.3	450
9	Loss of heterozygosity in malignant gliomas involves at least three distinct regions on chromosome 10. *Human Genetics*, **1993**, 92, 169-74	6.3	107
8	Four separate regions on chromosome 17 show loss of heterozygosity in familial breast carcinomas. *Human Genetics*, **1993**, 91, 6-12	6.3	23
7	Tight linkage between the Beckwith-Wiedemann syndrome and a microsatellite marker for the TH locus. *Human Genetics*, **1993**, 92, 296-8	6.3	8
6	Genetic mapping of a second locus predisposing to hereditary non-polyposis colon cancer. *Nature Genetics*, **1993**, 5, 279-82	36.3	375
5	Hereditary breast cancer in Sweden: a predominance of maternally inherited cases. *Breast Cancer Research and Treatment*, **1992**, 24, 159-65	4.4	11
4	Cytogenetic analysis by chromosome painting using DOP-PCR amplified flow-sorted chromosomes. *Genes Chromosomes and Cancer*, **1992**, 4, 257-63	5	474
3	Genomic alterations in human breast carcinomas. *Genes Chromosomes and Cancer*, **1990**, 2, 191-7	5	61
2	Multiple endocrine neoplasia type 1 gene maps to chromosome 11 and is lost in insulinoma. *Nature*, **1988**, 332, 85-7	50.4	875
1	Increased frequency of sister chromatid exchanges in cigarette smokers. *Hereditas*, **1978**, 88, 147-9	2.4	156