Anna M Lindstrand

List of Publications by Year
in descending order

80 papers	3,742 citations	172207 29 h-index	149479 56 g-index
87 all docs	87 docs citations	87 times ranked	7415 citing authors

#	Article	IF	Citations
1	Trailblazing precision medicine in Europe: A joint view by Genomic Medicine Sweden and the Centers for Personalized Medicine, ZPM, in Germany. Seminars in Cancer Biology, 2022, 84, 242-254.	4.3	22
2	Detection of germline mosaicism in fathers of children with intellectual disability syndromes caused by de novo variants. Molecular Genetics & Genomic Medicine, 2022, 10, e1880.	0.6	10
3	Targeted Exome Sequencing of Genes Involved in Rare CNVs in Early-Onset Severe Obesity. Frontiers in Genetics, 2022, 13, 839349.	1.1	3
4	PatientMatcher: A customizable Pythonâ€based openâ€source tool for matching undiagnosed rare disease patients via the Matchmaker Exchange network. Human Mutation, 2022, , .	1.1	5
5	Expanding the phenotype of the recurrent truncating <scp>eIF2Î³</scp> pathogenic variant p.() Tj ETQq1 1 0.784314 rgBT /Overlock 10 Case Reports (discontinued), 2022, 10, .	0.2	1
6	Multiâ€omics analysis reveals multiple mechanisms causing Praderâ€“Willi like syndrome in a family with a X;15 translocation. Human Mutation, 2022, 43, 1567-1575.	1.1	3
7	Complex genomic rearrangements: an underestimated cause of rare diseases. Trends in Genetics, 2022, 38, 1134-1146.	2.9	19
8	Novel pathogenic genomic variants leading to autosomal dominant and recessive Robinow syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 3593-3600.	0.7	16
9	DLG4-related synaptopathy: a new rare brain disorder. Genetics in Medicine, 2021, 23, 888-899.	1.1	16
10	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.	3.6	116
11	Rare deleterious mutations of HNRNP genes result in shared neurodevelopmental disorders. Genome Medicine, 2021, 13, 63.	3.6	50
12	High diagnostic yield in skeletal ciliopathies using massively parallel genome sequencing, structural variant screening and RNA analyses. Journal of Human Genetics, 2021, 66, 995-1008.	1.1	19
13	Solving patients with rare diseases through programmatic reanalysis of genome-phenome data. European Journal of Human Genetics, 2021, 29, 1337-1347.	1.4	34
14	Chromoanagenesis Event Underlies a de novo Pericentric and Multiple Paracentric Inversions in a Single Chromosome Causing Coffinâ€“Siris Syndrome. Frontiers in Genetics, 2021, 12, 708348.	1.1	5
15	Hybrid sequencing resolves two germline ultra-complex chromosomal rearrangements consisting of 137 breakpoint junctions in a single carrier. Human Genetics, 2021, 140, 775-790.	1.8	9
16	A missense mutation converts the Na+,K+-ATPase into an ion channel and causes therapy-resistant epilepsy. Journal of Biological Chemistry, 2021, 297, 101355.	1.6	9
17	Partial Monosomy 21 Mirrors Gene Expression of Trisomy 21 in a Patient-Derived Neuroepithelial Stem Cell Model. Frontiers in Genetics, 2021, 12, 803683.	1.1	1
18	Discovery of Novel Sequences in 1,000 Swedish Genomes. Molecular Biology and Evolution, 2020, 37, 18-30.	3.5	25

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19	Cytogenetically visible inversions are formed by multiple molecular mechanisms. Human Mutation, 2020, 41, 1979-1998.	1.1	12
20	Rare variants in dynein heavy chain genes in two individuals with situs inversus and developmental dyslexia: a case report. BMC Medical Genetics, 2020, 21, 87.	2.1	5
21	Wholeâ€genome sequencing reveals complex chromosome rearrangement disrupting <scp>NIPBL</scp> in infant with Cornelia de Lange syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 1143-1151.	0.7	17
22	Loqusdb: added value of an observations database of local genomic variation. BMC Bioinformatics, 2020, 21, 273.	1.2	5
23	Whole genome sequencing unveils genetic heterogeneity in optic nerve hypoplasia. PLoS ONE, 2020, 15, e0228622.	1.1	6
24	Large-scale targeted sequencing identifies risk genes for neurodevelopmental disorders. Nature Communications, 2020, 11, 4932.	5.8	105
25	Defective membrane insertion of mutant Na,Kâ€ATPase, a cause of fatal epilepsy. FASEB Journal, 2020, 34, 1-1.	0.2	0
26	Meckel syndrome: Clinical and mutation profile in six fetuses. Clinical Genetics, 2019, 96, 560-565.	1.0	15
27	Truncating Variant in Myof Gene Is Associated With Limb-Girdle Type Muscular Dystrophy and Cardiomyopathy. Frontiers in Genetics, 2019, 10, 608.	1.1	10
28	Ataxia in Patients With Bi-Allelic NFASC Mutations and Absence of Full-Length NF186. Frontiers in Genetics, 2019, 10, 896.	1.1	7
29	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.	3.6	88
30	Disruptive variants of CSDE1 associate with autism and interfere with neuronal development and synaptic transmission. Science Advances, 2019, 5, eaax2166.	4.7	35
31	Further support linking the 22q11.2 microduplication to an increased risk of bladder exstrophy and highlighting LZTR1 as a candidate gene. Molecular Genetics & Genomic Medicine, 2019, 7, e666.	0.6	9
32	Zebrafish Models of Neurodevelopmental Disorders: Limitations and Benefits of Current Tools and Techniques. International Journal of Molecular Sciences, 2019, 20, 1296.	1.8	72
33	Early activating somatic PIK3CA mutations promote ectopic muscle development and upper limb overgrowth. Clinical Genetics, 2019, 96, 118-125.	1.0	14
34	Comprehensive structural variation genome map of individuals carrying complex chromosomal rearrangements. PLoS Genetics, 2019, 15, e1007858.	1.5	36
35	Gain-of-function mutation of microRNA-140 in human skeletal dysplasia. Nature Medicine, 2019, 25, 583-590.	15.2	86
36	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.	2.6	30

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37	Rare copy number variants contribute pathogenic alleles in patients with intestinal malrotation. Molecular Genetics & Genomic Medicine, 2019, 7, e549.	0.6	12
38	Clinical Presentation of a Complex Neurodevelopmental Disorder Caused by Mutations in ADNP. Biological Psychiatry, 2019, 85, 287-297.	0.7	108
39	Targeted copy number screening highlights an intragenic deletion of WDR63 as the likely cause of human occipital encephalocele and abnormal CNS development in zebrafish. Human Mutation, 2018, 39, 495-505.	1.1	17
40	Marker chromosome genomic structure and temporal origin implicate a chromoanasynthesis event in a family with pleiotropic psychiatric phenotypes. Human Mutation, 2018, 39, 939-946.	1.1	26
41	Replicative and non-replicative mechanisms in the formation of clustered CNVs are indicated by whole genome characterization. PLoS Genetics, 2018, 14, e1007780.	1.5	28
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55	PLS3 Deletions Lead to Severe Spinal Osteoporosis and Disturbed Bone Matrix Mineralization. Journal of Bone and Mineral Research, 2017, 32, 2394-2404.	3.1	41
56	Novel KIAA0753 mutations extend the phenotype of skeletal ciliopathies. Scientific Reports, 2017, 7, 15585.	1.6	21
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.	1.1	58
58	Identification of new TRIP12 variants and detailed clinical evaluation of individuals with non-syndromic intellectual disability with or without autism. Human Genetics, 2017, 136, 179-192.	1.8	43
59	TIDDIT, an efficient and comprehensive structural variant caller for massive parallel sequencing data. F1000Research, 2017, 6, 664.	0.8	76
60	TIDDIT, an efficient and comprehensive structural variant caller for massive parallel sequencing data. F1000Research, 2017, 6, 664.	0.8	51
61	Periodontal Ehlers-Danlos Syndrome Is Caused by Mutations in C1R and C1S , which Encode Subcomponents C1r and C1s of Complement. American Journal of Human Genetics, 2016, 99, 1005-1014.	2.6	100
62	Copy-Number Variation Contributes to the Mutational Load of Bardet-Biedl Syndrome. American Journal of Human Genetics, 2016, 99, 318-336.	2.6	112
63	A novel phenotype in N-glycosylation disorders: Gillessen-Kaesbachâ€“Nishimura skeletal dysplasia due to pathogenic variants in ALG9. European Journal of Human Genetics, 2016, 24, 198-207.	1.4	29
64	Autosomal recessive mutations in the <scp>COL2A1</scp> gene cause severe spondyloepiphyseal dysplasia. Clinical Genetics, 2015, 87, 496-498.	1.0	9
65	Dominant Mutations in KAT6A Cause Intellectual Disability with Recognizable Syndromic Features. American Journal of Human Genetics, 2015, 96, 507-513.	2.6	107
66	WNT3 involvement in human bladder exstrophy and cloaca development in zebrafish. Human Molecular Genetics, 2015, 24, 5069-5078.	1.4	23
67	CTNND2â€”a candidate gene for reading problems and mild intellectual disability. Journal of Medical Genetics, 2015, 52, 111-122.	1.5	35
68	Low Copy Number of the AMY1 Locus Is Associated with Early-Onset Female Obesity in Finland. PLoS ONE, 2015, 10, e0131883.	1.1	70
69	Identification of three novelFGF16mutations in Xâ€linked recessive fusion of the fourth and fifth metacarpals and possible correlation with heart disease. Molecular Genetics & Genomic Medicine, 2014, 2, 402-411.	0.6	17
70	Different mutations inPDE4Dassociated with developmental disorders with mirror phenotypes. Journal of Medical Genetics, 2014, 51, 45-54.	1.5	57
71	Recurrent CNVs and SNVs at the NPHP1 Locus Contribute Pathogenic Alleles to Bardet-Biedl Syndrome. American Journal of Human Genetics, 2014, 94, 745-754.	2.6	80
72	ARMC4 Mutations Cause Primary Ciliary Dyskinesia with Randomization of Left/Right Body Asymmetry. American Journal of Human Genetics, 2013, 93, 357-367.	2.6	150

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73	A novel intellectual disability syndrome caused by GPI anchor deficiency due to homozygous mutations inPIGT. Journal of Medical Genetics, 2013, 50, 521-528.	1.5	108
74	Exome Capture Reveals ZNF423 and CEP164 Mutations, Linking Renal Ciliopathies to DNA Damage Response Signaling. Cell, 2012, 150, 533-548.	13.5	347
75	Inherited mosaicism for the supernumerary marker chromosome in cat eye syndrome: Interâ€and intraâ€individual variation and correlation to the phenotype. American Journal of Medical Genetics, Part A, 2012, 158A, 1111-1117.	0.7	9
76	Molecular and clinical characterization of patients with overlapping 10p deletions. American Journal of Medical Genetics, Part A, 2010, 152A, 1233-1243.	0.7	55
77	Detailed molecular and clinical characterization of three patients with 21q deletions. Clinical Genetics, 2010, 77, 145-154.	1.0	57
78	Improved structural characterization of chromosomal breakpoints using high resolution custom arrayâ€CGH. Clinical Genetics, 2010, 77, 552-562.	1.0	11
79	Further molecular and clinical delineation of co-locating 17p13.3 microdeletions and microduplications that show distinctive phenotypes. Journal of Medical Genetics, 2010, 47, 299-311.	1.5	137
80	Molecular cytogenetic characterization of a constitutional, highly complex intrachromosomal rearrangement of chromosome 1, with 14 breakpoints and a 0.5 Mb submicroscopic deletion. American Journal of Medical Genetics, Part A, 2008, 146A, 3217-3222.	0.7	8