Anna M Lindstrand

List of Publications by Year
in descending order

80	3,742	172457 29	149698 56
papers	citations	h-index	g-index

87	87	87	7415
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Targeted sequencing identifies 91 neurodevelopmental-disorder risk genes with autism and developmental-disability biases. Nature Genetics, 2017, 49, 515-526.	21.4	443
2	Exome Capture Reveals ZNF423 and CEP164 Mutations, Linking Renal Ciliopathies to DNA Damage Response Signaling. Cell, 2012, 150, 533-548.	28.9	347
3	Hotspots of missense mutation identify neurodevelopmental disorder genes and functional domains. Nature Neuroscience, 2017, 20, 1043-1051.	14.8	152
4	ARMC4 Mutations Cause Primary Ciliary Dyskinesia with Randomization of Left/Right Body Asymmetry. American Journal of Human Genetics, 2013, 93, 357-367.	6.2	150
5	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.	3.2	137
6	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
7	Copy-Number Variation Contributes to the Mutational Load of Bardet-Biedl Syndrome. American Journal of Human Genetics, 2016, 99, 318-336.	6.2	112
8	A novel intellectual disability syndrome caused by GPI anchor deficiency due to homozygous mutations inPIGT. Journal of Medical Genetics, 2013, 50, 521-528.	3.2	108
9	Clinical Presentation of a Complex Neurodevelopmental Disorder Caused by Mutations in ADNP. Biological Psychiatry, 2019, 85, 287-297.	1.3	108
10	Dominant Mutations in KAT6A Cause Intellectual Disability with Recognizable Syndromic Features. American Journal of Human Genetics, 2015, 96, 507-513.	6.2	107
11	Large-scale targeted sequencing identifies risk genes for neurodevelopmental disorders. Nature Communications, 2020, 11, 4932.	12.8	105
12	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.	6.2	100
13	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
14	Gain-of-function mutation of microRNA-140 in human skeletal dysplasia. Nature Medicine, 2019, 25, 583-590.	30.7	86
15	Recurrent CNVs and SNVs at the NPHP1 Locus Contribute Pathogenic Alleles to Bardet-Biedl Syndrome. American Journal of Human Genetics, 2014, 94, 745-754.	6.2	80
16	TIDDIT, an efficient and comprehensive structural variant caller for massive parallel sequencing data. F1000Research, 2017, 6, 664.	1.6	76
17	Zebrafish Models of Neurodevelopmental Disorders: Limitations and Benefits of Current Tools and Techniques. International Journal of Molecular Sciences, 2019, 20, 1296.	4.1	72
18	Low Copy Number of the AMY1 Locus Is Associated with Early-Onset Female Obesity in Finland. PLoS ONE, 2015, 10, e0131883.	2.5	70

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19	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
20	Detailed molecular and clinical characterization of three patients with 21q deletions. Clinical Genetics, 2010, 77, 145-154.	2.0	57
21	Different mutations inPDE4Dassociated with developmental disorders with mirror phenotypes. Journal of Medical Genetics, 2014, 51, 45-54.	3.2	57
22	Molecular and clinical characterization of patients with overlapping 10p deletions. American Journal of Medical Genetics, Part A, 2010, 152A, 1233-1243.	1.2	55
23	TIDDIT, an efficient and comprehensive structural variant caller for massive parallel sequencing data. F1000Research, 2017, 6, 664.	1.6	51
24	Rare deleterious mutations of HNRNP genes result in shared neurodevelopmental disorders. Genome Medicine, 2021, 13, 63.	8.2	50
25	De novo mutations in FLNC leading to early-onset restrictive cardiomyopathy and congenital myopathy. Human Mutation, 2018, 39, 1161-1172.	2.5	49
26	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.	3.8	43
27	PLS3 Deletions Lead to Severe Spinal Osteoporosis and Disturbed Bone Matrix Mineralization. Journal of Bone and Mineral Research, 2017, 32, 2394-2404.	2.8	41
28	Copy Number Variants Are Enriched in Individuals With Early-Onset Obesity and Highlight Novel Pathogenic Pathways. Journal of Clinical Endocrinology and Metabolism, 2017, 102, 3029-3039.	3.6	39
29	Comprehensive structural variation genome map of individuals carrying complex chromosomal rearrangements. PLoS Genetics, 2019, 15, e1007858.	3.5	36
30	CTNND2â€"a candidate gene for reading problems and mild intellectual disability. Journal of Medical Genetics, 2015, 52, 111-122.	3.2	35
31	Disruptive variants of CSDE1 associate with autism and interfere with neuronal development and synaptic transmission. Science Advances, 2019, 5, eaax2166.	10.3	35
32	Solving patients with rare diseases through programmatic reanalysis of genome-phenome data. European Journal of Human Genetics, 2021, 29, 1337-1347.	2.8	34
33	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
34	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.	2.8	29
35	Further evidence for specific IFIH1 mutation as a cause of Singletonâ€"Merten syndrome with phenotypic heterogeneity. American Journal of Medical Genetics, Part A, 2017, 173, 1396-1399.	1.2	28
36	Replicative and non-replicative mechanisms in the formation of clustered CNVs are indicated by whole genome characterization. PLoS Genetics, 2018, 14, e1007780.	3.5	28

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37	Marker chromosome genomic structure and temporal origin implicate a chromoanasynthesis event in a family with pleiotropic psychiatric phenotypes. Human Mutation, 2018, 39, 939-946.	2.5	26
38	Discovery of Novel Sequences in 1,000 Swedish Genomes. Molecular Biology and Evolution, 2020, 37, 18-30.	8.9	25
39	WNT3 involvement in human bladder exstrophy and cloaca development in zebrafish. Human Molecular Genetics, 2015, 24, 5069-5078.	2.9	23
40	A Large Inversion Involving GNAS Exon A/B and All Exons Encoding GsÎ± Is Associated With Autosomal Dominant Pseudohypoparathyroidism Type Ib (PHP1B). Journal of Bone and Mineral Research, 2017, 32, 776-783.	2.8	22
41	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.	9.6	22
42	Novel KIAA0753 mutations extend the phenotype of skeletal ciliopathies. Scientific Reports, 2017, 7, 15585.	3.3	21
43	Rare Copy Number Variants in Array-Based Comparative Genomic Hybridization in Early-Onset Skeletal Fragility. Frontiers in Endocrinology, 2018, 9, 380.	3.5	20
44	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.	2.3	19
45	AMYCNE: Confident copy number assessment using whole genome sequencing data. PLoS ONE, 2018, 13, e0189710.	2.5	19
46	Complex genomic rearrangements: an underestimated cause of rare diseases. Trends in Genetics, 2022, 38, 1134-1146.	6.7	19
47	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.	1.2	17
48	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.	2.5	17
49	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.	1.2	17
50	Alu-Alu mediated intragenic duplications in IFT81 and MATN3 are associated with skeletal dysplasias. Human Mutation, 2018, 39, 1456-1467.	2.5	16
51	Novel pathogenic genomic variants leading to autosomal dominant and recessive Robinow syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 3593-3600.	1.2	16
52	DLG4-related synaptopathy: a new rare brain disorder. Genetics in Medicine, 2021, 23, 888-899.	2.4	16
53	Meckel syndrome: Clinical and mutation profile in six fetuses. Clinical Genetics, 2019, 96, 560-565.	2.0	15
54	Early activating somatic PIK3CA mutations promote ectopic muscle development and upper limb overgrowth. Clinical Genetics, 2019, 96, 118-125.	2.0	14

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55	Rare copy number variants contribute pathogenic alleles in patients with intestinal malrotation. Molecular Genetics & Genomic Medicine, 2019, 7, e549.	1.2	12
56	Cytogenetically visible inversions are formed by multiple molecular mechanisms. Human Mutation, 2020, 41, 1979-1998.	2.5	12
57	Improved structural characterization of chromosomal breakpoints using high resolution custom arrayâ€CGH. Clinical Genetics, 2010, 77, 552-562.	2.0	11
58	High-resolution detection of chromosomal rearrangements in leukemias through mate pair whole genome sequencing. PLoS ONE, 2018, 13, e0193928.	2.5	11
59	Truncating Variant in Myof Gene Is Associated With Limb-Girdle Type Muscular Dystrophy and Cardiomyopathy. Frontiers in Genetics, 2019, 10, 608.	2.3	10
60	Detection of germline mosaicism in fathers of children with intellectual disability syndromes caused by de novo variants. Molecular Genetics & Genomic Medicine, 2022, 10, e1880.	1.2	10
61	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.	1.2	9
62	Autosomal recessive mutations in the <scp>COL2A1</scp> gene cause severe spondyloepiphyseal dysplasia. Clinical Genetics, 2015, 87, 496-498.	2.0	9
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73	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.	2.3	5
74	PatientMatcher: A customizable Pythonâ€based openâ€source tool for matching undiagnosed rare disease patients via the Matchmaker Exchange network. Human Mutation, 2022, , .	2.5	5
75	Targeted Exome Sequencing of Genes Involved in Rare CNVs in Early-Onset Severe Obesity. Frontiers in Genetics, 2022, 13, 839349.	2.3	3
76	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.	2.5	3
77	Partial Monosomy 21 Mirrors Gene Expression of Trisomy 21 in a Patient-Derived Neuroepithelial Stem Cell Model. Frontiers in Genetics, 2021, 12, 803683.	2.3	1
78	Expanding the phenotype of the recurrent truncating <scp>eIF2Î³</scp> pathogenic variant p.() Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50 54 Case Reports (discontinued), 2022, 10, .	0.5	1
79	Female representation on Swedish corporate boards. , 2018, , 169-182.		0
80	Defective membrane insertion of mutant Na,Kâ€ATPase, a cause of fatal epilepsy. FASEB Journal, 2020, 34, 1-1.	0.5	0