Christoffer NellÃ¥ker

List of Publications by Year
in descending order

33 papers	3,315 citations	430442 18 h-index	525886 27 g-index
35 all docs	35 docs citations	35 times ranked	6819 citing authors

#	Article	IF	Citations
1	Mouse genomic variation and its effect on phenotypes and gene regulation. Nature, 2011, 477, 289-294.	13.7	1,461
2	The RNA-Editing Enzyme ADAR1 Controls Innate Immune Responses to RNA. Cell Reports, 2014, 9, 1482-1494.	2.9	508
3	Sequence-based characterization of structural variation in the mouse genome. Nature, 2011, 477, 326-329.	13.7	299
4	The genomic landscape shaped by selection on transposable elements across 18 mouse strains. Genome Biology, 2012, 13, R45.	13.9	170
5	Genetic heterogeneity in Cornelia de Lange syndrome (CdLS) and CdLS-like phenotypes with observed and predicted levels of mosaicism. Journal of Medical Genetics, 2014, 51, 659-668.	1.5	141
6	Diagnostically relevant facial gestalt information from ordinary photos. ELife, 2014, 3, e02020.	2.8	129
7	Evaluation of minor groove binding probe and Taqman probe PCR assays: Influence of mismatches and template complexity on quantification. Molecular and Cellular Probes, 2006, 20, 311-316.	0.9	99
8	PURA syndrome: clinical delineation and genotype-phenotype study in 32 individuals with review of published literature. Journal of Medical Genetics, 2018, 55, 104-113.	1.5	59
9	Transcriptional Derepression of the ERVWE1 Locus following Influenza A Virus Infection. Journal of Virology, 2014, 88, 4328-4337.	1.5	56
10	Clinical and molecular consequences of disease-associated de novo mutations in SATB2. Genetics in Medicine, 2017, 19, 900-908.	1.1	46
11	De Novo and Inherited Loss-of-Function Variants in TLK2: Clinical and Genotype-Phenotype Evaluation of a Distinct Neurodevelopmental Disorder. American Journal of Human Genetics, 2018, 102, 1195-1203.	2.6	37
12	Next-generation phenotyping using computer vision algorithms in rare genomic neurodevelopmental disorders. Genetics in Medicine, 2019, 21, 1719-1725.	1.1	34
13	The case for open science: rare diseases. JAMIA Open, 2020, 3, 472-486.	1.0	33
14	A call for global action for rare diseases in Africa. Nature Genetics, 2020, 52, 21-26.	9.4	31
15	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
16	Mutation-specific pathophysiological mechanisms define different neurodevelopmental disorders associated with SATB1 dysfunction. American Journal of Human Genetics, 2021, 108, 346-356.	2.6	30
17	Big data phenotyping in rare diseases: some ethical issues. Genetics in Medicine, 2019, 21, 272-274.	1.1	22
18	Rapid Turnover of Functional Sequence in Human and Other Genomes. Annual Review of Genomics and Human Genetics, 2011, 12, 275-299.	2.5	21

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19	Molecular Beaconâ€“Based Temperature Control and Automated Analyses for Improved Resolution of Melting Temperature Analysis Using SYBR I Green Chemistry. Clinical Chemistry, 2007, 53, 98-103.	1.5	20
20	Machine Learning based histology phenotyping to investigate the epidemiologic and genetic basis of adipocyte morphology and cardiometabolic traits. PLoS Computational Biology, 2020, 16, e1008044.	1.5	16
21	Transcriptome and fatty-acid signatures of adipocyte hypertrophy and its non-invasive MR-based characterization in human adipose tissue. EBioMedicine, 2022, 79, 104020.	2.7	16
22	Enabling Global Clinical Collaborations on Identifiable Patient Data: The Minerva Initiative. Frontiers in Genetics, 2019, 10, 611.	1.1	14
23	Influenza A virus transactivates the mouse envelope gene encoding syncytin B and its regulator, glial cells missing 1. Journal of NeuroVirology, 2007, 13, 29-37.	1.0	13
24	Inherited variants in CHD3 show variable expressivity in Snijders Blok-Campeau syndrome. Genetics in Medicine, 2022, 24, 1283-1296.	1.1	9
25	From Same Photo: Cheating on Visual Kinship Challenges. Lecture Notes in Computer Science, 2019, , 654-668.	1.0	8
26	A DNA repair disorder caused by de novo monoallelic DDB1 variants is associated with a neurodevelopmental syndrome. American Journal of Human Genetics, 2021, 108, 749-756.	2.6	6
27	<scp>CHEDDA</scp> syndrome is an underrecognized neurodevelopmental disorder with a highly restricted <scp>ATN1</scp> mutation spectrum. Clinical Genetics, 2021, 100, 468-477.	1.0	4
28	Title is missing!. , 2020, 16, e1008044.		0
29	Title is missing!. , 2020, 16, e1008044.		0
30	Title is missing!. , 2020, 16, e1008044.		0
31	Title is missing!. , 2020, 16, e1008044.		0
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33	Title is missing!. , 2020, 16, e1008044.		0