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	Inherited variants in CHD3 show variable expressivity in Snijders Blok-Campeau syndrome. Genetics in Medicine, 2022, 24, 1283-1296.	1.1	9
2	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
3	Mutation-specific pathophysiological mechanisms define different neurodevelopmental disorders associated with SATB1 dysfunction. American Journal of Human Genetics, 2021, 108, 346-356.	2.6	30
4	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
5	<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
6	A call for global action for rare diseases in Africa. Nature Genetics, 2020, 52, 21-26.	9.4	31
7	The case for open science: rare diseases. JAMIA Open, 2020, 3, 472-486.	1.0	33
8	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
9	Title is missing!. , 2020, 16, e1008044.		0
10	Title is missing!. , 2020, 16, e1008044.		0
11	Title is missing!. , 2020, 16, e1008044.		0
12	Title is missing!. , 2020, 16, e1008044.		0
13	Title is missing!. , 2020, 16, e1008044.		0
14	Title is missing!. , 2020, 16, e1008044.		0
15	Big data phenotyping in rare diseases: some ethical issues. Genetics in Medicine, 2019, 21, 272-274.	1.1	22
16	Enabling Global Clinical Collaborations on Identifiable Patient Data: The Minerva Initiative. Frontiers in Genetics, 2019, 10, 611.	1.1	14
17	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
18	Next-generation phenotyping using computer vision algorithms in rare genomic neurodevelopmental disorders. Genetics in Medicine, 2019, 21, 1719-1725.	1.1	34

#	Article	IF	Citations
19	From Same Photo: Cheating on Visual Kinship Challenges. Lecture Notes in Computer Science, 2019, , 654-668.	1.0	8
20	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
21	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
22	Clinical and molecular consequences of disease-associated de novo mutations in SATB2. Genetics in Medicine, 2017, 19, 900-908.	1.1	46
23	Diagnostically relevant facial gestalt information from ordinary photos. ELife, 2014, 3, e02020.	2.8	129
24	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
25	Transcriptional Derepression of the ERVWE1 Locus following Influenza A Virus Infection. Journal of Virology, 2014, 88, 4328-4337.	1.5	56
26	The RNA-Editing Enzyme ADAR1 Controls Innate Immune Responses to RNA. Cell Reports, 2014, 9, 1482-1494.	2.9	508
27	The genomic landscape shaped by selection on transposable elements across 18 mouse strains. Genome Biology, 2012, 13, R45.	13.9	170
28	Rapid Turnover of Functional Sequence in Human and Other Genomes. Annual Review of Genomics and Human Genetics, 2011, 12, 275-299.	2.5	21
29	Mouse genomic variation and its effect on phenotypes and gene regulation. Nature, 2011, 477, 289-294.	13.7	1,461
30	Sequence-based characterization of structural variation in the mouse genome. Nature, 2011, 477, 326-329.	13.7	299
31	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
32	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
33	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