MÃ¥ns Magnusson

List of Publications by Year
in descending order

13 papers	5,350 citations	933264 10 h-index	1199470 12 g-index
13 all docs	13 docs citations	13 times ranked	11217 citing authors

#	Article	IF	Citations
1	MultiQC: summarize analysis results for multiple tools and samples in a single report. Bioinformatics, 2016, 32, 3047-3048.	1.8	4,633
2	Dominant Mutations in GRHL3 Cause Van der Woude Syndrome and Disrupt Oral Periderm Development. American Journal of Human Genetics, 2014, 94, 23-32.	2.6	195
3	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
4	An international effort towards developing standards for best practices in analysis, interpretation and reporting of clinical genome sequencing results in the CLARITY Challenge. Genome Biology, 2014, 15, R53.	13.9	101
5	Rescue of primary ubiquinone deficiency due to a novelCOQ7defect using 2,4â€“dihydroxybensoic acid. Journal of Medical Genetics, 2015, 52, 779-783.	1.5	94
6	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
7	Rapid pulsed whole genome sequencing for comprehensive acute diagnostics of inborn errors of metabolism. BMC Genomics, 2014, 15, 1090.	1.2	54
8	SLC12A2 mutations cause NKCC1 deficiency with encephalopathy and impaired secretory epithelia. Neurology: Genetics, 2020, 6, e478.	0.9	20
9	The Relationship between Caloric Response, Oculomotor Dysfunction and Size of Cerebello-Pontine Angle Tumours. Acta Oto-Laryngologica, 1988, 106, 361-367.	0.3	19
10	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
11	Loqusdb: added value of an observations database of local genomic variation. BMC Bioinformatics, 2020, 21, 273.	1.2	5
12	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
13	Chanjo: Clincal grade sequence coverage analysis. F1000Research, 0, 9, 615.	0.8	1