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	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
2	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
3	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
4	SLC12A2 mutations cause NKCC1 deficiency with encephalopathy and impaired secretory epithelia. Neurology: Genetics, 2020, 6, e478.	0.9	20
5	Loqusdb: added value of an observations database of local genomic variation. BMC Bioinformatics, 2020, 21, 273.	1.2	5
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	MultiQC: summarize analysis results for multiple tools and samples in a single report. Bioinformatics, 2016, 32, 3047-3048.	1.8	4,633
8	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
9	Rapid pulsed whole genome sequencing for comprehensive acute diagnostics of inborn errors of metabolism. BMC Genomics, 2014, 15, 1090.	1.2	54
10	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
11	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
12	The Relationship between Caloric Response, Oculomotor Dysfunction and Size of Cerebello-Pontine Angle Tumours. Acta Oto-Laryngologica, 1988, 106, 361-367.	0.3	19
13	Chanjo: Clincal grade sequence coverage analysis. F1000Research, 0, 9, 615.	0.8	1