Giuseppe Gallone

List of Publications by Year
in descending order

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15 papers	1,696 citations	759233 12 h-index	996975 15 g-index
21 all docs	21 docs citations	21 times ranked	4780 citing authors

#	Article	IF	Citations
1	TADAâ€”a machine learning tool for functional annotation-based prioritisation of pathogenic CNVs. Genome Biology, 2022, 23, 67.	8.8	4
2	Non-coding region variants upstream of MEF2C cause severe developmental disorder through three distinct loss-of-function mechanisms. American Journal of Human Genetics, 2021, 108, 1083-1094.	6.2	42
3	The contribution of X-linked coding variation to severe developmental disorders. Nature Communications, 2021, 12, 627.	12.8	33
4	Optical Interrogation of Sympathetic Neuronal Effects on Macroscopic Cardiomyocyte Network Dynamics. IScience, 2020, 23, 101334.	4.1	13
5	Evidence for 28 genetic disorders discovered by combining healthcare and research data. Nature, 2020, 586, 757-762.	27.8	343
6	Ranbow: A fast and accurate method for polyploid haplotype reconstruction. PLoS Computational Biology, 2020, 16, e1007843.	3.2	23
7	Targeted RNA sequencing enhances gene expression profiling of ultra-low input samples. RNA Biology, 2020, 17, 1741-1753.	3.1	10
8	Contribution of retrotransposition to developmental disorders. Nature Communications, 2019, 10, 4630.	12.8	43
9	Exome-wide assessment of the functional impact and pathogenicity of multinucleotide mutations. Genome Research, 2019, 29, 1047-1056.	5.5	34
10	Pathogenicity and selective constraint on variation near splice sites. Genome Research, 2019, 29, 159-170.	5.5	70
11	Making new genetic diagnoses with old data: iterative reanalysis and reporting from genome-wide data in 1,133 families with developmental disorders. Genetics in Medicine, 2018, 20, 1216-1223.	2.4	255
12	De novo mutations in regulatory elements in neurodevelopmental disorders. Nature, 2018, 555, 611-616.	27.8	232
13	Quantifying the contribution of recessive coding variation to developmental disorders. Science, 2018, 362, 1161-1164.	12.6	158
14	Common genetic variants contribute to risk of rare severe neurodevelopmental disorders. Nature, 2018, 562, 268-271.	27.8	246
15	Foxn1 regulates key target genes essential for T cell development in postnatal thymic epithelial cells. Nature Immunology, 2016, 17, 1206-1215.	14.5	142