

Giuseppe Gallone

List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/864376/publications.pdf>

Version: 2024-02-01

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