Alejandro Sifrim

List of Publications by Year
in descending order

44 papers	4,795 citations	218677 26 h-index	254184 43 g-index
55 all docs	55 docs citations	55 times ranked	10826 citing authors

#	Article	IF	Citations
1	Identification of the tumour transition states occurring during EMT. Nature, 2018, 556, 463-468.	27.8	1,083
2	Genetic diagnosis of developmental disorders in the DDD study: a scalable analysis of genome-wide research data. Lancet, The, 2015, 385, 1305-1314.	13.7	651
3	Distinct genetic architectures for syndromic and nonsyndromic congenital heart defects identified by exome sequencing. Nature Genetics, 2016, 48, 1060-1065.	21.4	351
4	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
5	Common genetic variants contribute to risk of rare severe neurodevelopmental disorders. Nature, 2018, 562, 268-271.	27.8	246
6	De novo mutations in regulatory elements in neurodevelopmental disorders. Nature, 2018, 555, 611-616.	27.8	232
7	Heterozygous missense mutations in SMARCA2 cause Nicolaides-Baraitser syndrome. Nature Genetics, 2012, 44, 445-449.	21.4	207
8	Histone Lysine Methylases and Demethylases in the Landscape of Human Developmental Disorders. American Journal of Human Genetics, 2018, 102, 175-187.	6.2	204
9	eXtasy: variant prioritization by genomic data fusion. Nature Methods, 2013, 10, 1083-1084.	19.0	153
10	Discovery of four recessive developmental disorders using probabilistic genotype and phenotype matching among 4,125 families. Nature Genetics, 2015, 47, 1363-1369.	21.4	133
11	Early lineage segregation of multipotent embryonic mammary gland progenitors. Nature Cell Biology, 2018, 20, 666-676.	10.3	124
12	BCL11A Haploinsufficiency Causes an Intellectual Disability Syndrome and Dysregulates Transcription. American Journal of Human Genetics, 2016, 99, 253-274.	6.2	118
13	Mechanisms of stretch-mediated skin expansion at single-cell resolution. Nature, 2020, 584, 268-273.	27.8	113
14	Heterotypic cell–cell communication regulates glandular stem cell multipotency. Nature, 2020, 584, 608-613.	27.8	82
15	Biallelic Variants in UBA5 Link Dysfunctional UFM1 Ubiquitin-like Modifier Pathway to Severe Infantile-Onset Encephalopathy. American Journal of Human Genetics, 2016, 99, 683-694.	6.2	72
16	Defining the Design Principles of Skin Epidermis Postnatal Growth. Cell, 2020, 181, 604-620.e22.	28.9	65
17	Interstitial Cell Remodeling Promotes Aberrant Adipogenesis in Dystrophic Muscles. Cell Reports, 2020, 31, 107597.	6.4	64
18	Mutations in Either TUBB or MAPRE2 Cause Circumferential Skin Creases Kunze Type. American Journal of Human Genetics, 2015, 97, 790-800.	6.2	63

#	Article	IF	Citations
19	Heterozygous Variants in KMT2E Cause a Spectrum of Neurodevelopmental Disorders and Epilepsy. American Journal of Human Genetics, 2019, 104, 1210-1222.	6.2	56
20	Medusa: A tool for exploring and clustering biological networks. BMC Research Notes, 2011, 4, 384.	1.4	49
21	Clinical and molecular consequences of disease-associated de novo mutations in SATB2. Genetics in Medicine, 2017, 19, 900-908.	2.4	46
22	Bi-allelic Loss-of-Function CACNA1B Mutations in Progressive Epilepsy-Dyskinesia. American Journal of Human Genetics, 2019, 104, 948-956.	6.2	45
23	Detection of structural mosaicism from targeted and whole-genome sequencing data. Genome Research, 2017, 27, 1704-1714.	5.5	44
24	Unraveling genomic variation from next generation sequencing data. BioData Mining, 2013, 6, 13.	4.0	43
25	Contribution of retrotransposition to developmental disorders. Nature Communications, 2019, 10, 4630.	12.8	43
26	Genetic and chemotherapeutic influences on germline hypermutation. Nature, 2022, 605, 503-508.	27.8	43
27	The contribution of X-linked coding variation to severe developmental disorders. Nature Communications, 2021, 12, 627.	12.8	33
28	<i>Beegle:</i> from literature mining to disease-gene discovery. Nucleic Acids Research, 2016, 44, e18-e18.	14.5	30
29	Annotate-it: a Swiss-knife approach to annotation, analysis and interpretation of single nucleotide variation in human disease. Genome Medicine, 2012, 4, 73.	8.2	28
30	Integrative analysis of genomic variants reveals new associations of candidate haploinsufficient genes with congenital heart disease. PLoS Genetics, 2021, 17, e1009679.	3.5	17
31	Systems genetics analysis identifies calcium-signaling defects as novel cause of congenital heart disease. Genome Medicine, 2020, 12, 76.	8.2	15
32	Detecting cryptic clinically relevant structural variation in exome-sequencing data increases diagnostic yield for developmental disorders. American Journal of Human Genetics, 2021, 108, 2186-2194.	6.2	12
33	Finding Diagnostically Useful Patterns in Quantitative Phenotypic Data. American Journal of Human Genetics, 2019, 105, 933-946.	6.2	8
34	Recent advances in congenital heart disease genomics. F1000Research, 2017, 6, 869.	1.6	7
35	Alternative Experimental Design with an Applied Normalization Scheme Can Improve Statistical Power in 2D-DIGE Experiments. Journal of Proteome Research, 2010, 9, 4919-4926.	3.7	6
36	Meander: visually exploring the structural variome using space-filling curves. Nucleic Acids Research, 2013, 41, e118-e118.	14.5	6

#	Article	IF	Citations
37	A Genetic Algorithm for Pancreatic Cancer Diagnosis. Communications in Computer and Information Science, 2013, , 222-230.	0.5	5
38	Problems with the nested granularity of feature domains in bioinformatics: the eXtasy case. BMC Bioinformatics, 2015, 16, S2.	2.6	5
39	TrioVis: a visualization approach for filtering genomic variants of parentâ€"child trios. Bioinformatics, 2013, 29, 1801-1802.	4.1	4
40	A Simple Genetic Algorithm for Biomarker Mining. Lecture Notes in Computer Science, 2012, , 222-232.	1.3	4
41	A Self-Tuning Genetic Algorithm with Applications in Biomarker Discovery. , 2014, , .		3
42	A Hybrid Approach to Feature Ranking for Microarray Data Classification. Communications in Computer and Information Science, 2013, , 241-248.	0.5	2
43	eXtasy simplified-towards opening the black box. , 2013, , .		0
44	A Note on the Evaluation of Mutation Prioritization Algorithms. , 2015, , .		0