Alejandro Sifrim

List of Publications by Year in Descending Order

46 papers	2,924 citations	25 h-index	54 g-index
55 ext. papers	4,122 ext. citations	19 avg, IF	4.35 L-index

#	Paper	IF	Citations
46	Genetic and chemotherapeutic influences on germline hypermutation.. *Nature*, **2022**, 605, 503-508	50.4	0
45	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	11	0
44	Integrative analysis of genomic variants reveals new associations of candidate haploinsufficient genes with congenital heart disease. *PLoS Genetics*, **2021**, 17, e1009679	6	1
43	The contribution of X-linked coding variation to severe developmental disorders. *Nature Communications*, **2021**, 12, 627	17.4	5
42	Defining the Design Principles of Skin Epidermis Postnatal Growth. *Cell*, **2020**, 181, 604-620.e22	56.2	26
41	Interstitial Cell Remodeling Promotes Aberrant Adipogenesis in Dystrophic Muscles. *Cell Reports*, **2020**, 31, 107597	10.6	28
40	Mechanisms of stretch-mediated skin expansion at single-cell resolution. *Nature*, **2020**, 584, 268-273	50.4	35
39	Systems genetics analysis identifies calcium-signaling defects as novel cause of congenital heart disease. *Genome Medicine*, **2020**, 12, 76	14.4	5
38	Heterotypic cell-cell communication regulates glandular stem cell multipotency. *Nature*, **2020**, 584, 608-613	50.4	27
37	Heterozygous Variants in KMT2E Cause a Spectrum of Neurodevelopmental Disorders and Epilepsy. *American Journal of Human Genetics*, **2019**, 104, 1210-1222	11	31
36	Bi-allelic Loss-of-Function CACNA1B Mutations in Progressive Epilepsy-Dyskinesia. *American Journal of Human Genetics*, **2019**, 104, 948-956	11	17
35	Finding Diagnostically Useful Patterns in Quantitative Phenotypic Data. *American Journal of Human Genetics*, **2019**, 105, 933-946	11	4
34	Contribution of retrotransposition to developmental disorders. *Nature Communications*, **2019**, 10, 4630	17.4	25
33	Identification of the tumour transition states occurring during EMT. *Nature*, **2018**, 556, 463-468	50.4	652
32	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	8.1	161
31	Histone Lysine Methylases and Demethylases in the Landscape of Human Developmental Disorders. *American Journal of Human Genetics*, **2018**, 102, 175-187	11	108
30	De novo mutations in regulatory elements in neurodevelopmental disorders. *Nature*, **2018**, 555, 611-616	50.4	146

29	Common genetic variants contribute to risk of rare severe neurodevelopmental disorders. *Nature*, **2018**, 562, 268-271	50.4	149
28	Early lineage segregation of multipotent embryonic mammary gland progenitors. *Nature Cell Biology*, **2018**, 20, 666-676	23.4	84
27	Clinical and molecular consequences of disease-associated de novo mutations in SATB2. *Genetics in Medicine*, **2017**, 19, 900-908	8.1	30
26	Detection of structural mosaicism from targeted and whole-genome sequencing data. *Genome Research*, **2017**, 27, 1704-1714	9.7	26
25	Recent advances in congenital heart disease genomics. *F1000Research*, **2017**, 6, 869	3.6	4
24	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	11	43
23	BCL11A Haploinsufficiency Causes an Intellectual Disability Syndrome and Dysregulates Transcription. *American Journal of Human Genetics*, **2016**, 99, 253-74	11	84
22	Beegle: from literature mining to disease-gene discovery. *Nucleic Acids Research*, **2016**, 44, e18	20.1	18
21	Distinct genetic architectures for syndromic and nonsyndromic congenital heart defects identified by exome sequencing. *Nature Genetics*, **2016**, 48, 1060-5	36.3	200
20	Discovery of four recessive developmental disorders using probabilistic genotype and phenotype matching among 4,125 families. *Nature Genetics*, **2015**, 47, 1363-9	36.3	91
19	Problems with the nested granularity of feature domains in bioinformatics: the eXtasy case. *BMC Bioinformatics*, **2015**, 16 Suppl 4, S2	3.6	4
18	Genetic diagnosis of developmental disorders in the DDD study: a scalable analysis of genome-wide research data. *Lancet, The*, **2015**, 385, 1305-14	40	451
17	Mutations in Either TUBB or MAPRE2 Cause Circumferential Skin Creases Kunze Type. *American Journal of Human Genetics*, **2015**, 97, 790-800	11	41
16	A Self-Tuning Genetic Algorithm with Applications in Biomarker Discovery **2014**,		3
15	Unraveling genomic variation from next generation sequencing data. *BioData Mining*, **2013**, 6, 13	4.3	33
14	eXtasy: variant prioritization by genomic data fusion. *Nature Methods*, **2013**, 10, 1083-4	21.6	119
13	TrioVis: a visualization approach for filtering genomic variants of parent-child trios. *Bioinformatics*, **2013**, 29, 1801-2	7.2	4
12	A Hybrid Approach to Feature Ranking for Microarray Data Classification. *Communications in Computer and Information Science*, **2013**, 241-248	0.3	1

11	A Genetic Algorithm for Pancreatic Cancer Diagnosis. *Communications in Computer and Information Science*, **2013**, 222-230	0.3	2
10	Meander: visually exploring the structural variome using space-filling curves. *Nucleic Acids Research*, **2013**, 41, e118	20.1	6
9	Heterozygous missense mutations in SMARCA2 cause Nicolaides-Baraitser syndrome. *Nature Genetics*, **2012**, 44, 445-9, S1	36.3	170
8	Annotate-it: a Swiss-knife approach to annotation, analysis and interpretation of single nucleotide variation in human disease. *Genome Medicine*, **2012**, 4, 73	14.4	27
7	A Simple Genetic Algorithm for Biomarker Mining. *Lecture Notes in Computer Science*, **2012**, 222-232	0.9	3
6	Medusa: A tool for exploring and clustering biological networks. *BMC Research Notes*, **2011**, 4, 384	2.3	44
5	Alternative experimental design with an applied normalization scheme can improve statistical power in 2D-DIGE experiments. *Journal of Proteome Research*, **2010**, 9, 4919-26	5.6	5
4	Common genetic variants contribute to risk of rare severe neurodevelopmental disorders		1
3	De novo mutations in regulatory elements cause neurodevelopmental disorders		2
2	Contribution of Retrotransposition to Developmental Disorders		2
1	Genetic and pharmacological causes of germline hypermutation		3