Dalila Pinto

List of Publications by Year in Descending Order

92 papers	17,468 citations	48 h-index	97 g-index
97 ext. papers	21,582 ext. citations	13.7 avg, IF	5.06 L-index

#	Paper	IF	Citations
92	Neuronal and glial 3D chromatin architecture informs the cellular etiology of brain disorders. *Nature Communications*, **2021**, 12, 3968	17.4	2
91	Shared genetic risk between eating disorder- and substance-use-related phenotypes: Evidence from genome-wide association studies. *Addiction Biology*, **2021**, 26, e12880	4.6	12
90	Direct reprogramming induces vascular regeneration post muscle ischemic injury. *Molecular Therapy*, **2021**, 29, 3042-3058	11.7	6
89	Common Genetic Variation And Age at Onset Of Anorexia Nervosa. *Biological Psychiatry Global Open Science*, **2021**,		3
88	Dissecting the Shared Genetic Architecture of Suicide Attempt, Psychiatric Disorders, and Known Risk Factors. *Biological Psychiatry*, **2021**,	7.9	11
87	mTADA is a framework for identifying risk genes from de novo mutations in multiple traits. *Nature Communications*, **2020**, 11, 2929	17.4	2
86	Integrated Transcriptome and Network Analysis Reveals Spatiotemporal Dynamics of Calvarial Suturogenesis. *Cell Reports*, **2020**, 32, 107871	10.6	16
85	Associations Between Attention-Deficit/Hyperactivity Disorder and Various Eating Disorders: A Swedish Nationwide Population Study Using Multiple Genetically Informative Approaches. *Biological Psychiatry*, **2019**, 86, 577-586	7.9	24
84	Genome-wide association study identifies eight risk loci and implicates metabo-psychiatric origins for anorexia nervosa. *Nature Genetics*, **2019**, 51, 1207-1214	36.3	303
83	Genomic Relationships, Novel Loci, and Pleiotropic Mechanisms across Eight Psychiatric Disorders. *Cell*, **2019**, 179, 1469-1482.e11	56.2	402
82	Shared molecular neuropathology across major psychiatric disorders parallels polygenic overlap. *Science*, **2018**, 359, 693-697	33.3	547
81	Analysis of shared heritability in common disorders of the brain. *Science*, **2018**, 360,	33.3	666
80	Transcriptome and epigenome landscape of human cortical development modeled in organoids. *Science*, **2018**, 362,	33.3	142
79	Integrative functional genomic analysis of human brain development and neuropsychiatric risks. *Science*, **2018**, 362,	33.3	277
78	Transcriptome-wide isoform-level dysregulation in ASD, schizophrenia, and bipolar disorder. *Science*, **2018**, 362,	33.3	434
77	Comprehensive functional genomic resource and integrative model for the human brain. *Science*, **2018**, 362,	33.3	319
76	Influenza virus infection causes global RNAPII termination defects. *Nature Structural and Molecular Biology*, **2018**, 25, 885-893	17.6	24

75	Landscape of Conditional eQTL in Dorsolateral Prefrontal Cortex and Co-localization with Schizophrenia GWAS. *American Journal of Human Genetics*, **2018**, 102, 1169-1184	11	73
74	Evidence for three genetic loci involved in both anorexia nervosa risk and variation of body mass index. *Molecular Psychiatry*, **2017**, 22, 192-201	15.1	31
73	Parallel changes in serum proteins and diffusion tensor imaging in methamphetamine-associated psychosis. *Scientific Reports*, **2017**, 7, 43777	4.9	7
72	De novo unbalanced translocation (4p duplication/8p deletion) in a patient with autism, OCD, and overgrowth syndrome. *American Journal of Medical Genetics, Part A*, **2017**, 173, 1656-1662	2.5	7
71	Significant Locus and Metabolic Genetic Correlations Revealed in Genome-Wide Association Study of Anorexia Nervosa. *American Journal of Psychiatry*, **2017**, 174, 850-858	11.9	276
70	A genome-wide association study of anorexia nervosa suggests a risk locus implicated in dysregulated leptin signaling. *Scientific Reports*, **2017**, 7, 3847	4.9	16
69	Genetic architecture distinguishes systemic juvenile idiopathic arthritis from other forms of juvenile idiopathic arthritis: clinical and therapeutic implications. *Annals of the Rheumatic Diseases*, **2017**, 76, 906-913	2.4	89
68	Integrated Bayesian analysis of rare exonic variants to identify risk genes for schizophrenia and neurodevelopmental disorders. *Genome Medicine*, **2017**, 9, 114	14.4	48
67	Contribution of copy number variants to schizophrenia from a genome-wide study of 41,321 subjects. *Nature Genetics*, **2017**, 49, 27-35	36.3	530
66	Characterization of Large Copy Number Variation in Mexican Type 2 Diabetes subjects. *Scientific Reports*, **2017**, 7, 17105	4.9	9
65	Phenotypic Association Analyses With Copy Number Variation in Recurrent Depressive Disorder. *Biological Psychiatry*, **2016**, 79, 329-36	7.9	15
64	Identification of novel genetic causes of Rett syndrome-like phenotypes. *Journal of Medical Genetics*, **2016**, 53, 190-9	5.8	108
63	Gene expression elucidates functional impact of polygenic risk for schizophrenia. *Nature Neuroscience*, **2016**, 19, 1442-1453	25.5	622
62	Bone marrow failure and developmental delay caused by mutations in poly(A)-specific ribonuclease (PARN). *Journal of Medical Genetics*, **2015**, 52, 738-48	5.8	56
61	The PsychENCODE project. *Nature Neuroscience*, **2015**, 18, 1707-12	25.5	226
60	The phenotypic manifestations of rare genic CNVs in autism spectrum disorder. *Molecular Psychiatry*, **2015**, 20, 1366-72	15.1	27
59	Microdeletions of ELP4 Are Associated with Language Impairment, Autism Spectrum Disorder, and Mental Retardation. *Human Mutation*, **2015**, 36, 842-50	4.7	31
58	ISDN2014_0322: REMOVED: Identification of novel genetic causes of Rett syndrome-like phenotypes by whole exome sequencing. *International Journal of Developmental Neuroscience*, **2015**, 47, 99-99	2.7	

57	HLA-DRB1*11 and variants of the MHC class II locus are strong risk factors for systemic juvenile idiopathic arthritis. *Proceedings of the National Academy of Sciences of the United States of America*, **2015**, 112, 15970-5	11.5	103
56	A deletion involving CD38 and BST1 results in a fusion transcript in a patient with autism and asthma. *Autism Research*, **2014**, 7, 254-63	5.1	29
55	Convergence of genes and cellular pathways dysregulated in autism spectrum disorders. *American Journal of Human Genetics*, **2014**, 94, 677-94	11	635
54	Brain-expressed exons under purifying selection are enriched for de novo mutations in autism spectrum disorder. *Nature Genetics*, **2014**, 46, 742-7	36.3	121
53	A CTNNA3 compound heterozygous deletion implicates a role for □-catenin in susceptibility to autism spectrum disorder. *Journal of Neurodevelopmental Disorders*, **2014**, 6, 17	4.6	32
52	Recurrent duplications of the annexin A1 gene (ANXA1) in autism spectrum disorders. *Molecular Autism*, **2014**, 5, 28	6.5	9
51	A genome-wide association study of anorexia nervosa. *Molecular Psychiatry*, **2014**, 19, 1085-94	15.1	224
50	The impact of the metabotropic glutamate receptor and other gene family interaction networks on autism. *Nature Communications*, **2014**, 5, 4074	17.4	45
49	Using ancestry-informative markers to identify fine structure across 15 populations of European origin. *European Journal of Human Genetics*, **2014**, 22, 1190-200	5.3	30
48	Meta-analysis of SHANK Mutations in Autism Spectrum Disorders: a gradient of severity in cognitive impairments. *PLoS Genetics*, **2014**, 10, e1004580	6	340
47	A genome-wide copy number association study of osteoporotic fractures points to the 6p25.1 locus. *Journal of Medical Genetics*, **2014**, 51, 122-31	5.8	32
46	The Autism Simplex Collection: an international, expertly phenotyped autism sample for genetic and phenotypic analyses. *Molecular Autism*, **2014**, 5, 34	6.5	25
45	Network topologies and convergent aetiologies arising from deletions and duplications observed in individuals with autism. *PLoS Genetics*, **2013**, 9, e1003523	6	47
44	Molecular characteristics of a pancreatic adenocarcinoma associated with Shwachman-Diamond syndrome. *Pediatric Blood and Cancer*, **2013**, 60, 754-60	3	19
43	The autism sequencing consortium: large-scale, high-throughput sequencing in autism spectrum disorders. *Neuron*, **2012**, 76, 1052-6	13.9	124
42	A novel approach of homozygous haplotype sharing identifies candidate genes in autism spectrum disorder. *Human Genetics*, **2012**, 131, 565-79	6.3	150
41	Rare deletions at the neurexin 3 locus in autism spectrum disorder. *American Journal of Human Genetics*, **2012**, 90, 133-41	11	155
40	SHANK1 Deletions in Males with Autism Spectrum Disorder. *American Journal of Human Genetics*, **2012**, 90, 879-87	11	233

39	Genetic and functional analyses of SHANK2 mutations suggest a multiple hit model of autism spectrum disorders. *PLoS Genetics*, **2012**, 8, e1002521	6	297
38	Individual common variants exert weak effects on the risk for autism spectrum disorders. *Human Molecular Genetics*, **2012**, 21, 4781-92	5.6	279
37	Genome-wide survey of large rare copy number variants in Alzheimer's disease among Caribbean hispanics. *G3: Genes, Genomes, Genetics*, **2012**, 2, 71-8	3.2	43
36	Hemizygous deletions on chromosome 1p21.3 involving the DPYD gene in individuals with autism spectrum disorder. *Clinical Genetics*, **2011**, 80, 435-43	4	50
35	Gene-ontology enrichment analysis in two independent family-based samples highlights biologically plausible processes for autism spectrum disorders. *European Journal of Human Genetics* , **2011**, 19, 1082-9	5.3	30
34	Comprehensive assessment of array-based platforms and calling algorithms for detection of copy number variants. *Nature Biotechnology*, **2011**, 29, 512-20	44.5	333
33	Gene and miRNA expression profiles in autism spectrum disorders. *Brain Research*, **2011**, 1380, 85-97	3.7	138
32	A genotype resource for postmortem brain samples from the Autism Tissue Program. *Autism Research*, **2011**, 4, 89-97	5.1	20
31	Novel method for combined linkage and genome-wide association analysis finds evidence of distinct genetic architecture for two subtypes of autism. *Journal of Neurodevelopmental Disorders*, **2011**, 3, 113-23	4.6	20
30	A family with autism and rare copy number variants disrupting the Duchenne/Becker muscular dystrophy gene DMD and TRPM3. *Journal of Neurodevelopmental Disorders*, **2011**, 3, 124-31	4.6	28
29	A scan statistic to extract causal gene clusters from case-control genome-wide rare CNV data. *BMC Bioinformatics*, **2011**, 12, 205	3.6	4
28	A novel approach identifies new differentially methylated regions (DMRs) associated with imprinted genes. *Genome Research*, **2011**, 21, 465-76	9.7	87
27	Rare familial 16q21 microdeletions under a linkage peak implicate cadherin 8 (CDH8) in susceptibility to autism and learning disability. *Journal of Medical Genetics*, **2011**, 48, 48-54	5.8	81
26	Origins and functional impact of copy number variation in the human genome. *Nature*, **2010**, 464, 704-12	50.4	1467
25	Genome-wide association study of CNVs in 16,000 cases of eight common diseases and 3,000 shared controls. *Nature*, **2010**, 464, 713-20	50.4	639
24	Functional impact of global rare copy number variation in autism spectrum disorders. *Nature*, **2010**, 466, 368-72	50.4	1499
23	Mutations in the SHANK2 synaptic scaffolding gene in autism spectrum disorder and mental retardation. *Nature Genetics*, **2010**, 42, 489-91	36.3	402
22	A genome-wide scan for common alleles affecting risk for autism. *Human Molecular Genetics*, **2010**, 19, 4072-82	5.6	443

21	Disruption at the PTCHD1 Locus on Xp22.11 in Autism spectrum disorder and intellectual disability. *Science Translational Medicine*, **2010**, 2, 49ra68	17.5	140
20	Towards a comprehensive structural variation map of an individual human genome. *Genome Biology* , **2010**, 11, R52	18.3	204
19	Candidate genes and biological processes in de novo CNVs from autistic individuals **2010**, 11, O9		78
18	Identifying signatures of natural selection in Tibetan and Andean populations using dense genome scan data. *PLoS Genetics*, **2010**, 6, e1001116	6	395
17	A genome-wide linkage and association scan reveals novel loci for autism. *Nature*, **2009**, 461, 802-8	50.4	474
16	Endophenotype Strategy in Epilepsy Genetics **2009**, 135-149		
15	Effective detection of human leukocyte antigen risk alleles in celiac disease using tag single nucleotide polymorphisms. *PLoS ONE*, **2008**, 3, e2270	3.7	113
14	Structural variation of chromosomes in autism spectrum disorder. *American Journal of Human Genetics*, **2008**, 82, 477-88	11	1413
13	Explorative two-locus linkage analysis suggests a multiplicative interaction between the 7q32 and 16p13 myoclonic seizures-related photosensitivity loci. *Genetic Epidemiology*, **2007**, 31, 42-50	2.6	25
12	Association analysis of BRD2 (RING3) and epilepsy in a Dutch population. *Epilepsia*, **2007**, 48, 2191-2	6.4	13
11	Copy-number variation in control population cohorts. *Human Molecular Genetics*, **2007**, 16 Spec No. 2, R168-73	5.6	186
10	Contribution of SHANK3 mutations to autism spectrum disorder. *American Journal of Human Genetics*, **2007**, 81, 1289-97	11	492
9	A novel splicing mutation in KCNQ2 in a multigenerational family with BFNC followed for 25 years. *Epilepsia*, **2006**, 47, 851-9	6.4	32
8	Heterogeneity at the JME 6p11-12 locus: absence of mutations in the EFHC1 gene in linked Dutch families. *Epilepsia*, **2006**, 47, 1743-6	6.4	14
7	Genome-wide linkage scan of epilepsy-related photoparoxysmal electroencephalographic response: evidence for linkage on chromosomes 7q32 and 16p13. *Human Molecular Genetics*, **2005**, 14, 171-8	5.6	210
6	Evidence for linkage between juvenile myoclonic epilepsy-related idiopathic generalized epilepsy and 6p11-12 in Dutch families. *Epilepsia*, **2004**, 45, 211-7	6.4	29
5	Genetics of photosensitivity (photoparoxysmal response): a review. *Epilepsia*, **2004**, 45 Suppl 1, 19-23	6.4	61
4	Analysis of the allelic diversity of a (CA)n repeat polymorphism among alpha 1-antitrypsin gene products from northern Portugal. *Human Genetics*, **1997**, 99, 194-8	6.3	7

List of Publications

3	Cell type-specific isolation and transcriptomic profiling informs glial pathology in human temporal lobe epilepsy	1
2	Co-localization of Conditional eQTL and GWAS Signatures in Schizophrenia	6
1	Identifying tissues implicated in Anorexia Nervosa using Transcriptomic Imputation	1