

# Dalila Pinto

## List of Publications by Citations

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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	Functional impact of global rare copy number variation in autism spectrum disorders. <i>Nature</i> , <b>2010</b> , 466, 368-72	50.4	1499
91	Origins and functional impact of copy number variation in the human genome. <i>Nature</i> , <b>2010</b> , 464, 704-12	50.4	1467
90	Structural variation of chromosomes in autism spectrum disorder. <i>American Journal of Human Genetics</i> , <b>2008</b> , 82, 477-88	11	1413
89	Analysis of shared heritability in common disorders of the brain. <i>Science</i> , <b>2018</b> , 360,	33.3	666
88	Genome-wide association study of CNVs in 16,000 cases of eight common diseases and 3,000 shared controls. <i>Nature</i> , <b>2010</b> , 464, 713-20	50.4	639
87	Convergence of genes and cellular pathways dysregulated in autism spectrum disorders. <i>American Journal of Human Genetics</i> , <b>2014</b> , 94, 677-94	11	635
86	Gene expression elucidates functional impact of polygenic risk for schizophrenia. <i>Nature Neuroscience</i> , <b>2016</b> , 19, 1442-1453	25.5	622
85	Shared molecular neuropathology across major psychiatric disorders parallels polygenic overlap. <i>Science</i> , <b>2018</b> , 359, 693-697	33.3	547
84	Contribution of copy number variants to schizophrenia from a genome-wide study of 41,321 subjects. <i>Nature Genetics</i> , <b>2017</b> , 49, 27-35	36.3	530
83	Contribution of SHANK3 mutations to autism spectrum disorder. <i>American Journal of Human Genetics</i> , <b>2007</b> , 81, 1289-97	11	492
82	A genome-wide linkage and association scan reveals novel loci for autism. <i>Nature</i> , <b>2009</b> , 461, 802-8	50.4	474
81	A genome-wide scan for common alleles affecting risk for autism. <i>Human Molecular Genetics</i> , <b>2010</b> , 19, 4072-82	5.6	443
80	Transcriptome-wide isoform-level dysregulation in ASD, schizophrenia, and bipolar disorder. <i>Science</i> , <b>2018</b> , 362,	33.3	434
79	Mutations in the SHANK2 synaptic scaffolding gene in autism spectrum disorder and mental retardation. <i>Nature Genetics</i> , <b>2010</b> , 42, 489-91	36.3	402
78	Genomic Relationships, Novel Loci, and Pleiotropic Mechanisms across Eight Psychiatric Disorders. <i>Cell</i> , <b>2019</b> , 179, 1469-1482.e11	56.2	402
77	Identifying signatures of natural selection in Tibetan and Andean populations using dense genome scan data. <i>PLoS Genetics</i> , <b>2010</b> , 6, e1001116	6	395
76	Meta-analysis of SHANK Mutations in Autism Spectrum Disorders: a gradient of severity in cognitive impairments. <i>PLoS Genetics</i> , <b>2014</b> , 10, e1004580	6	340

75	Comprehensive assessment of array-based platforms and calling algorithms for detection of copy number variants. <i>Nature Biotechnology</i> , <b>2011</b> , 29, 512-20	44.5	333
74	Comprehensive functional genomic resource and integrative model for the human brain. <i>Science</i> , <b>2018</b> , 362,	33.3	319
73	Genome-wide association study identifies eight risk loci and implicates metabo-psychiatric origins for anorexia nervosa. <i>Nature Genetics</i> , <b>2019</b> , 51, 1207-1214	36.3	303
72	Genetic and functional analyses of SHANK2 mutations suggest a multiple hit model of autism spectrum disorders. <i>PLoS Genetics</i> , <b>2012</b> , 8, e1002521	6	297
71	Individual common variants exert weak effects on the risk for autism spectrum disorders. <i>Human Molecular Genetics</i> , <b>2012</b> , 21, 4781-92	5.6	279
70	Integrative functional genomic analysis of human brain development and neuropsychiatric risks. <i>Science</i> , <b>2018</b> , 362,	33.3	277
69	Significant Locus and Metabolic Genetic Correlations Revealed in Genome-Wide Association Study of Anorexia Nervosa. <i>American Journal of Psychiatry</i> , <b>2017</b> , 174, 850-858	11.9	276
68	SHANK1 Deletions in Males with Autism Spectrum Disorder. <i>American Journal of Human Genetics</i> , <b>2012</b> , 90, 879-87	11	233
67	The PsychENCODE project. <i>Nature Neuroscience</i> , <b>2015</b> , 18, 1707-12	25.5	226
66	A genome-wide association study of anorexia nervosa. <i>Molecular Psychiatry</i> , <b>2014</b> , 19, 1085-94	15.1	224
65	Genome-wide linkage scan of epilepsy-related photoparoxysmal electroencephalographic response: evidence for linkage on chromosomes 7q32 and 16p13. <i>Human Molecular Genetics</i> , <b>2005</b> , 14, 171-8	5.6	210
64	Towards a comprehensive structural variation map of an individual human genome. <i>Genome Biology</i> , <b>2010</b> , 11, R52	18.3	204
63	Copy-number variation in control population cohorts. <i>Human Molecular Genetics</i> , <b>2007</b> , 16 Spec No. 2, R168-73	5.6	186
62	Rare deletions at the neurexin 3 locus in autism spectrum disorder. <i>American Journal of Human Genetics</i> , <b>2012</b> , 90, 133-41	11	155
61	A novel approach of homozygous haplotype sharing identifies candidate genes in autism spectrum disorder. <i>Human Genetics</i> , <b>2012</b> , 131, 565-79	6.3	150
60	Transcriptome and epigenome landscape of human cortical development modeled in organoids. <i>Science</i> , <b>2018</b> , 362,	33.3	142
59	Disruption at the PTCHD1 Locus on Xp22.11 in Autism spectrum disorder and intellectual disability. <i>Science Translational Medicine</i> , <b>2010</b> , 2, 49ra68	17.5	140
58	Gene and miRNA expression profiles in autism spectrum disorders. <i>Brain Research</i> , <b>2011</b> , 1380, 85-97	3.7	138

57	The autism sequencing consortium: large-scale, high-throughput sequencing in autism spectrum disorders. <i>Neuron</i> , <b>2012</b> , 76, 1052-6	13.9	124
56	Brain-expressed exons under purifying selection are enriched for de novo mutations in autism spectrum disorder. <i>Nature Genetics</i> , <b>2014</b> , 46, 742-7	36.3	121
55	Effective detection of human leukocyte antigen risk alleles in celiac disease using tag single nucleotide polymorphisms. <i>PLoS ONE</i> , <b>2008</b> , 3, e2270	3.7	113
54	Identification of novel genetic causes of Rett syndrome-like phenotypes. <i>Journal of Medical Genetics</i> , <b>2016</b> , 53, 190-9	5.8	108
53	HLA-DRB1*11 and variants of the MHC class II locus are strong risk factors for systemic juvenile idiopathic arthritis. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , <b>2015</b> , 112, 15970-5	11.5	103
52	Genetic architecture distinguishes systemic juvenile idiopathic arthritis from other forms of juvenile idiopathic arthritis: clinical and therapeutic implications. <i>Annals of the Rheumatic Diseases</i> , <b>2017</b> , 76, 906-913	2.4	89
51	A novel approach identifies new differentially methylated regions (DMRs) associated with imprinted genes. <i>Genome Research</i> , <b>2011</b> , 21, 465-76	9.7	87
50	Rare familial 16q21 microdeletions under a linkage peak implicate cadherin 8 (CDH8) in susceptibility to autism and learning disability. <i>Journal of Medical Genetics</i> , <b>2011</b> , 48, 48-54	5.8	81
49	Candidate genes and biological processes in de novo CNVs from autistic individuals <b>2010</b> , 11, O9		78
48	Landscape of Conditional eQTL in Dorsolateral Prefrontal Cortex and Co-localization with Schizophrenia GWAS. <i>American Journal of Human Genetics</i> , <b>2018</b> , 102, 1169-1184	11	73
47	Genetics of photosensitivity (photoparoxysmal response): a review. <i>Epilepsia</i> , <b>2004</b> , 45 Suppl 1, 19-23	6.4	61
46	Bone marrow failure and developmental delay caused by mutations in poly(A)-specific ribonuclease (PARN). <i>Journal of Medical Genetics</i> , <b>2015</b> , 52, 738-48	5.8	56
45	Hemizygous deletions on chromosome 1p21.3 involving the DPYD gene in individuals with autism spectrum disorder. <i>Clinical Genetics</i> , <b>2011</b> , 80, 435-43	4	50
44	Integrated Bayesian analysis of rare exonic variants to identify risk genes for schizophrenia and neurodevelopmental disorders. <i>Genome Medicine</i> , <b>2017</b> , 9, 114	14.4	48
43	Network topologies and convergent aetiologies arising from deletions and duplications observed in individuals with autism. <i>PLoS Genetics</i> , <b>2013</b> , 9, e1003523	6	47
42	The impact of the metabotropic glutamate receptor and other gene family interaction networks on autism. <i>Nature Communications</i> , <b>2014</b> , 5, 4074	17.4	45
41	Genome-wide survey of large rare copy number variants in Alzheimer's disease among Caribbean hispanics. <i>G3: Genes, Genomes, Genetics</i> , <b>2012</b> , 2, 71-8	3.2	43
40	A CTNNA3 compound heterozygous deletion implicates a role for $\beta$ -catenin in susceptibility to autism spectrum disorder. <i>Journal of Neurodevelopmental Disorders</i> , <b>2014</b> , 6, 17	4.6	32

39	A genome-wide copy number association study of osteoporotic fractures points to the 6p25.1 locus. <i>Journal of Medical Genetics</i> , <b>2014</b> , 51, 122-31	5.8	32
38	A novel splicing mutation in KCNQ2 in a multigenerational family with BFNC followed for 25 years. <i>Epilepsia</i> , <b>2006</b> , 47, 851-9	6.4	32
37	Evidence for three genetic loci involved in both anorexia nervosa risk and variation of body mass index. <i>Molecular Psychiatry</i> , <b>2017</b> , 22, 192-201	15.1	31
36	Microdeletions of ELP4 Are Associated with Language Impairment, Autism Spectrum Disorder, and Mental Retardation. <i>Human Mutation</i> , <b>2015</b> , 36, 842-50	4.7	31
35	Using ancestry-informative markers to identify fine structure across 15 populations of European origin. <i>European Journal of Human Genetics</i> , <b>2014</b> , 22, 1190-200	5.3	30
34	Gene-ontology enrichment analysis in two independent family-based samples highlights biologically plausible processes for autism spectrum disorders. <i>European Journal of Human Genetics</i> , <b>2011</b> , 19, 1082-9	5.3	30
33	A deletion involving CD38 and BST1 results in a fusion transcript in a patient with autism and asthma. <i>Autism Research</i> , <b>2014</b> , 7, 254-63	5.1	29
32	Evidence for linkage between juvenile myoclonic epilepsy-related idiopathic generalized epilepsy and 6p11-12 in Dutch families. <i>Epilepsia</i> , <b>2004</b> , 45, 211-7	6.4	29
31	A family with autism and rare copy number variants disrupting the Duchenne/Becker muscular dystrophy gene DMD and TRPM3. <i>Journal of Neurodevelopmental Disorders</i> , <b>2011</b> , 3, 124-31	4.6	28
30	The phenotypic manifestations of rare genic CNVs in autism spectrum disorder. <i>Molecular Psychiatry</i> , <b>2015</b> , 20, 1366-72	15.1	27
29	The Autism Simplex Collection: an international, expertly phenotyped autism sample for genetic and phenotypic analyses. <i>Molecular Autism</i> , <b>2014</b> , 5, 34	6.5	25
28	Explorative two-locus linkage analysis suggests a multiplicative interaction between the 7q32 and 16p13 myoclonic seizures-related photosensitivity loci. <i>Genetic Epidemiology</i> , <b>2007</b> , 31, 42-50	2.6	25
27	Associations Between Attention-Deficit/Hyperactivity Disorder and Various Eating Disorders: A Swedish Nationwide Population Study Using Multiple Genetically Informative Approaches. <i>Biological Psychiatry</i> , <b>2019</b> , 86, 577-586	7.9	24
26	Influenza virus infection causes global RNAPII termination defects. <i>Nature Structural and Molecular Biology</i> , <b>2018</b> , 25, 885-893	17.6	24
25	A genotype resource for postmortem brain samples from the Autism Tissue Program. <i>Autism Research</i> , <b>2011</b> , 4, 89-97	5.1	20
24	Novel method for combined linkage and genome-wide association analysis finds evidence of distinct genetic architecture for two subtypes of autism. <i>Journal of Neurodevelopmental Disorders</i> , <b>2011</b> , 3, 113-23	4.6	20
23	Molecular characteristics of a pancreatic adenocarcinoma associated with Shwachman-Diamond syndrome. <i>Pediatric Blood and Cancer</i> , <b>2013</b> , 60, 754-60	3	19
22	A genome-wide association study of anorexia nervosa suggests a risk locus implicated in dysregulated leptin signaling. <i>Scientific Reports</i> , <b>2017</b> , 7, 3847	4.9	16

21	Integrated Transcriptome and Network Analysis Reveals Spatiotemporal Dynamics of Calvarial Suturogenesis. <i>Cell Reports</i> , <b>2020</b> , 32, 107871	10.6	16
20	Phenotypic Association Analyses With Copy Number Variation in Recurrent Depressive Disorder. <i>Biological Psychiatry</i> , <b>2016</b> , 79, 329-36	7.9	15
19	Heterogeneity at the JME 6p11-12 locus: absence of mutations in the EFHC1 gene in linked Dutch families. <i>Epilepsia</i> , <b>2006</b> , 47, 1743-6	6.4	14
18	Association analysis of BRD2 (RING3) and epilepsy in a Dutch population. <i>Epilepsia</i> , <b>2007</b> , 48, 2191-2	6.4	13
17	Shared genetic risk between eating disorder- and substance-use-related phenotypes: Evidence from genome-wide association studies. <i>Addiction Biology</i> , <b>2021</b> , 26, e12880	4.6	12
16	Dissecting the Shared Genetic Architecture of Suicide Attempt, Psychiatric Disorders, and Known Risk Factors. <i>Biological Psychiatry</i> , <b>2021</b> ,	7.9	11
15	Recurrent duplications of the annexin A1 gene (ANXA1) in autism spectrum disorders. <i>Molecular Autism</i> , <b>2014</b> , 5, 28	6.5	9
14	Characterization of Large Copy Number Variation in Mexican Type 2 Diabetes subjects. <i>Scientific Reports</i> , <b>2017</b> , 7, 17105	4.9	9
13	Parallel changes in serum proteins and diffusion tensor imaging in methamphetamine-associated psychosis. <i>Scientific Reports</i> , <b>2017</b> , 7, 43777	4.9	7
12	De novo unbalanced translocation (4p duplication/8p deletion) in a patient with autism, OCD, and overgrowth syndrome. <i>American Journal of Medical Genetics, Part A</i> , <b>2017</b> , 173, 1656-1662	2.5	7
11	Analysis of the allelic diversity of a (CA) <sub>n</sub> repeat polymorphism among alpha 1-antitrypsin gene products from northern Portugal. <i>Human Genetics</i> , <b>1997</b> , 99, 194-8	6.3	7
10	Co-localization of Conditional eQTL and GWAS Signatures in Schizophrenia		6
9	Direct reprogramming induces vascular regeneration post muscle ischemic injury. <i>Molecular Therapy</i> , <b>2021</b> , 29, 3042-3058	11.7	6
8	A scan statistic to extract causal gene clusters from case-control genome-wide rare CNV data. <i>BMC Bioinformatics</i> , <b>2011</b> , 12, 205	3.6	4
7	Common Genetic Variation And Age at Onset Of Anorexia Nervosa. <i>Biological Psychiatry Global Open Science</i> , <b>2021</b> ,		3
6	mTADA is a framework for identifying risk genes from de novo mutations in multiple traits. <i>Nature Communications</i> , <b>2020</b> , 11, 2929	17.4	2
5	Neuronal and glial 3D chromatin architecture informs the cellular etiology of brain disorders. <i>Nature Communications</i> , <b>2021</b> , 12, 3968	17.4	2
4	Cell type-specific isolation and transcriptomic profiling informs glial pathology in human temporal lobe epilepsy		1

- 3 Identifying tissues implicated in Anorexia Nervosa using Transcriptomic Imputation 1
- 2 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
- 1 Endophenotype Strategy in Epilepsy Genetics 2009, 135-149