

Detelina Grozeva

List of Publications by Citations

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

57 papers	9,296 citations	30 h-index	61 g-index
61 ext. papers	11,340 ext. citations	12.3 avg, IF	3.93 L-index

#	Paper	IF	Citations
57	Genetic relationship between five psychiatric disorders estimated from genome-wide SNPs. <i>Nature Genetics</i> , 2013 , 45, 984-94	36.3	1628
56	Association scan of 14,500 nonsynonymous SNPs in four diseases identifies autoimmunity variants. <i>Nature Genetics</i> , 2007 , 39, 1329-37	36.3	1130
55	Collaborative genome-wide association analysis supports a role for ANK3 and CACNA1C in bipolar disorder. <i>Nature Genetics</i> , 2008 , 40, 1056-8	36.3	949
54	Genetic meta-analysis of diagnosed Alzheimer's disease identifies new risk loci and implicates A β tau, immunity and lipid processing. <i>Nature Genetics</i> , 2019 , 51, 414-430	36.3	917
53	The UK10K project identifies rare variants in health and disease. <i>Nature</i> , 2015 , 526, 82-90	50.4	776
52	Genome-wide association study of CNVs in 16,000 cases of eight common diseases and 3,000 shared controls. <i>Nature</i> , 2010 , 464, 713-20	50.4	639
51	Microduplications of 16p11.2 are associated with schizophrenia. <i>Nature Genetics</i> , 2009 , 41, 1223-7	36.3	550
50	Support for the involvement of large copy number variants in the pathogenesis of schizophrenia. <i>Human Molecular Genetics</i> , 2009 , 18, 1497-503	5.6	346
49	Comprehensive Rare Variant Analysis via Whole-Genome Sequencing to Determine the Molecular Pathology of Inherited Retinal Disease. <i>American Journal of Human Genetics</i> , 2017 , 100, 75-90	11	235
48	Operation of the schizophrenia susceptibility gene, neuregulin 1, across traditional diagnostic boundaries to increase risk for bipolar disorder. <i>Archives of General Psychiatry</i> , 2005 , 62, 642-8		209
47	Joint analysis of psychiatric disorders increases accuracy of risk prediction for schizophrenia, bipolar disorder, and major depressive disorder. <i>American Journal of Human Genetics</i> , 2015 , 96, 283-94	11	161
46	Rare copy number variants: a point of rarity in genetic risk for bipolar disorder and schizophrenia. <i>Archives of General Psychiatry</i> , 2010 , 67, 318-27		154
45	Whole-genome sequencing of patients with rare diseases in a national health system. <i>Nature</i> , 2020 , 583, 96-102	50.4	139
44	Variation at the DAOA/G30 locus influences susceptibility to major mood episodes but not psychosis in schizophrenia and bipolar disorder. <i>Archives of General Psychiatry</i> , 2006 , 63, 366-73		124
43	Targeted Next-Generation Sequencing Analysis of 1,000 Individuals with Intellectual Disability. <i>Human Mutation</i> , 2015 , 36, 1197-204	4.7	122
42	Mutations in the histone methyltransferase gene KMT2B cause complex early-onset dystonia. <i>Nature Genetics</i> , 2017 , 49, 223-237	36.3	116
41	Support for neuregulin 1 as a susceptibility gene for bipolar disorder and schizophrenia. <i>Biological Psychiatry</i> , 2008 , 64, 419-27	7.9	95

40	Phenotypic insights into ADCY5-associated disease. <i>Movement Disorders</i> , 2016 , 31, 1033-40	7	78
39	De novo loss-of-function mutations in SETD5, encoding a methyltransferase in a 3p25 microdeletion syndrome critical region, cause intellectual disability. <i>American Journal of Human Genetics</i> , 2014 , 94, 618-24	11	74
38	Complex structural variants in Mendelian disorders: identification and breakpoint resolution using short- and long-read genome sequencing. <i>Genome Medicine</i> , 2018 , 10, 95	14.4	70
37	Whole-genome sequencing of a sporadic primary immunodeficiency cohort. <i>Nature</i> , 2020 , 583, 90-95	50.4	69
36	Independent estimation of the frequency of rare CNVs in the UK population confirms their role in schizophrenia. <i>Schizophrenia Research</i> , 2012 , 135, 1-7	3.6	62
35	Identification of a human synaptotagmin-1 mutation that perturbs synaptic vesicle cycling. <i>Journal of Clinical Investigation</i> , 2015 , 125, 1670-8	15.9	57
34	Reciprocal duplication of the Williams-Beuren syndrome deletion on chromosome 7q11.23 is associated with schizophrenia. <i>Biological Psychiatry</i> , 2014 , 75, 371-7	7.9	54
33	Polymorphism of the 5-HT transporter and response to antidepressants: randomised controlled trial. <i>British Journal of Psychiatry</i> , 2011 , 198, 464-71	5.4	51
32	P2RX7: A bipolar and unipolar disorder candidate susceptibility gene?. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2009 , 150B, 1063-9	3.5	47
31	Bipolar Disorder is associated with the rs6971 polymorphism in the gene encoding 18 kDa Translocator Protein (TSPO). <i>Psychoneuroendocrinology</i> , 2013 , 38, 2826-9	5	37
30	Affective temperaments across the bipolar-unipolar spectrum: examination of the TEMPS-A in 927 patients and controls. <i>Journal of Affective Disorders</i> , 2010 , 123, 42-51	6.6	33
29	An inherited duplication at the gene p21 Protein-Activated Kinase 7 (PAK7) is a risk factor for psychosis. <i>Human Molecular Genetics</i> , 2014 , 23, 3316-26	5.6	32
28	Clinical and molecular consequences of disease-associated de novo mutations in SATB2. <i>Genetics in Medicine</i> , 2017 , 19, 900-908	8.1	30
27	Comprehensive Cancer-Predisposition Gene Testing in an Adult Multiple Primary Tumor Series Shows a Broad Range of Deleterious Variants and Atypical Tumor Phenotypes. <i>American Journal of Human Genetics</i> , 2018 , 103, 3-18	11	27
26	Identification of high risk DISC1 protein structural variants in patients with bipolar spectrum disorder. <i>Neuroscience Letters</i> , 2010 , 486, 136-40	3.3	27
25	Reduced burden of very large and rare CNVs in bipolar affective disorder. <i>Bipolar Disorders</i> , 2013 , 15, 893-8	3.8	22
24	Identification of a CACNA2D4 deletion in late onset bipolar disorder patients and implications for the involvement of voltage-dependent calcium channels in psychiatric disorders. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2012 , 159B, 465-75	3.5	19
23	MECP2 missense mutations outside the canonical MBD and TRD domains in males with intellectual disability. <i>Journal of Human Genetics</i> , 2016 , 61, 95-101	4.3	18

22	Novel ATP2A2 mutations in a large sample of individuals with Darier disease. <i>Journal of Dermatology</i> , 2013 , 40, 259-66	1.6	18
21	De Novo Truncating Mutations in WASF1 Cause Intellectual Disability with Seizures. <i>American Journal of Human Genetics</i> , 2018 , 103, 144-153	11	18
20	Bi-allelic Loss-of-Function CACNA1B Mutations in Progressive Epilepsy-Dyskinesia. <i>American Journal of Human Genetics</i> , 2019 , 104, 948-956	11	17
19	Biallelic Mutation of ARHGEF18, Involved in the Determination of Epithelial Apicobasal Polarity, Causes Adult-Onset Retinal Degeneration. <i>American Journal of Human Genetics</i> , 2017 , 100, 334-342	11	14
18	Variation at the GABAA receptor gene, Rho 1 (GABRR1) associated with susceptibility to bipolar schizoaffective disorder. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2010 , 153B, 1347-9	3.5	14
17	Genetic heterogeneity according to age at onset in bipolar disorder: a combined positional cloning and candidate gene approach. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2012 , 159B, 653-9	3.5	13
16	Gene-based analysis in HRC imputed genome wide association data identifies three novel genes for Alzheimer's disease. <i>PLoS ONE</i> , 2019 , 14, e0218111	3.7	12
15	Re-annotation of 191 developmental and epileptic encephalopathy-associated genes unmasks de novo variants in. <i>Npj Genomic Medicine</i> , 2019 , 4, 31	6.2	12
14	Genotype-phenotype correlations in Darier disease: A focus on the neuropsychiatric phenotype. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2018 , 177, 717-726	3.5	12
13	Functional Analyses of a Novel Splice Variant in the Gene, Found by Next Generation Sequencing, Confirm Its Pathogenicity in a Spanish Patient and Diagnose Him with CHARGE Syndrome. <i>Frontiers in Genetics</i> , 2018 , 9, 7	4.5	11
12	DNAJC6 Mutations Disrupt Dopamine Homeostasis in Juvenile Parkinsonism-Dystonia. <i>Movement Disorders</i> , 2020 , 35, 1357-1368	7	10
11	Complex Structural Variants Resolved by Short-Read and Long-Read Whole Genome Sequencing in Mendelian Disorders		4
10	Benefits and Challenges of Rare Genetic Variation in Alzheimer's Disease. <i>Current Genetic Medicine Reports</i> , 2019 , 7, 53-62	2.2	4
9	Rare Genetic Variation in 135 Families With Family History Suggestive of X-Linked Intellectual Disability. <i>Frontiers in Genetics</i> , 2019 , 10, 578	4.5	3
8	The Impact of COVID-19 on Cancer Symptom Experience and Help-Seeking Behaviour in the United Kingdom: A Cross-Sectional Population Survey. <i>SSRN Electronic Journal</i> ,	1	3
7	A systematic association mapping on chromosome 6q in bipolar affective disorder--evidence for the melanin-concentrating-hormone-receptor-2 gene as a risk factor for bipolar affective disorder. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2010 , 153B, 878-84	3.5	2
6	Impact of introducing procalcitonin testing on antibiotic usage in acute NHS hospitals during the first wave of COVID-19 in the UK: a controlled interrupted time series analysis of organization-level data.. <i>Journal of Antimicrobial Chemotherapy</i> , 2022 ,	5.1	2
5	Novel KAT6B proximal familial variant expands genotypic and phenotypic spectrum. <i>Clinical Genetics</i> , 2019 , 95, 334-335	4	2

4 Gene-Based Analysis in HRC Imputed Genome Wide Association Data Identifies Three Novel Genes For Alzheimer's Disease 1

3 Genome-wide significant locus for Research Diagnostic Criteria Schizoaffective Disorder Bipolar type. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, **2017**, 174, 767-771 3.5

2 P2-112: NEXT GENERATION EXOME SEQUENCING IN A LARGE SAMPLE OF ALZHEIMER'S PATIENTS **2018**, 14, P712-P712

1 Identification and functional modelling of plausibly causative cis-regulatory variants in a highly-selected cohort with X-linked intellectual disability. *PLoS ONE*, **2021**, 16, e0256181 3.7