Detelina Grozeva

List of Publications by Year in Descending Order

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Version: 2024-04-10

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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	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.. *Journal of Antimicrobial Chemotherapy*, **2022**,	5.1	2
56	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	
55	Whole-genome sequencing of a sporadic primary immunodeficiency cohort. *Nature*, **2020**, 583, 90-95	50.4	69
54	DNAJC6 Mutations Disrupt Dopamine Homeostasis in Juvenile Parkinsonism-Dystonia. *Movement Disorders*, **2020**, 35, 1357-1368	7	10
53	Whole-genome sequencing of patients with rare diseases in a national health system. *Nature*, **2020**, 583, 96-102	50.4	139
52	Bi-allelic Loss-of-Function CACNA1B Mutations in Progressive Epilepsy-Dyskinesia. *American Journal of Human Genetics*, **2019**, 104, 948-956	11	17
51	Gene-based analysis in HRC imputed genome wide association data identifies three novel genes for Alzheimer's disease. *PLoS ONE*, **2019**, 14, e0218111	3.7	12
50	Rare Genetic Variation in 135 Families With Family History Suggestive of X-Linked Intellectual Disability. *Frontiers in Genetics*, **2019**, 10, 578	4.5	3
49	Genetic meta-analysis of diagnosed Alzheimer's disease identifies new risk loci and implicates A tau, immunity and lipid processing. *Nature Genetics*, **2019**, 51, 414-430	36.3	917
48	Benefits and Challenges of Rare Genetic Variation in Alzheimer Disease. *Current Genetic Medicine Reports*, **2019**, 7, 53-62	2.2	4
47	Re-annotation of 191 developmental and epileptic encephalopathy-associated genes unmasks de novo variants in. *Npj Genomic Medicine*, **2019**, 4, 31	6.2	12
46	Novel KAT6B proximal familial variant expands genotypic and phenotypic spectrum. *Clinical Genetics*, **2019**, 95, 334-335	4	2
45	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. *Frontiers in Genetics*, **2018**, 9, 7	4.5	11
44	Comprehensive Cancer-Predisposition Gene Testing in an Adult Multiple Primary Tumor Series Shows a Broad Range of Deleterious Variants and Atypical Tumor Phenotypes. *American Journal of Human Genetics*, **2018**, 103, 3-18	11	27
43	P2-112: NEXT GENERATION EXOME SEQUENCING IN A LARGE SAMPLE OF ALZHEIMER'S PATIENTS **2018**, 14, P712-P712		
42	Complex structural variants in Mendelian disorders: identification and breakpoint resolution using short- and long-read genome sequencing. *Genome Medicine*, **2018**, 10, 95	14.4	70
41	Genotype-phenotype correlations in Darier disease: A focus on the neuropsychiatric phenotype. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, **2018**, 177, 717-726	3.5	12

40	De Novo Truncating Mutations in WASF1 Cause Intellectual Disability with Seizures. *American Journal of Human Genetics*, **2018**, 103, 144-153	11	18
39	Biallelic Mutation of ARHGEF18, Involved in the Determination of Epithelial Apicobasal Polarity, Causes Adult-Onset Retinal Degeneration. *American Journal of Human Genetics*, **2017**, 100, 334-342	11	14
38	Clinical and molecular consequences of disease-associated de novo mutations in SATB2. *Genetics in Medicine*, **2017**, 19, 900-908	8.1	30
37	Comprehensive Rare Variant Analysis via Whole-Genome Sequencing to Determine the Molecular Pathology of Inherited Retinal Disease. *American Journal of Human Genetics*, **2017**, 100, 75-90	11	235
36	Mutations in the histone methyltransferase gene KMT2B cause complex early-onset dystonia. *Nature Genetics*, **2017**, 49, 223-237	36.3	116
35	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	
34	MECP2 missense mutations outside the canonical MBD and TRD domains in males with intellectual disability. *Journal of Human Genetics*, **2016**, 61, 95-101	4.3	18
33	Phenotypic insights into ADCY5-associated disease. *Movement Disorders*, **2016**, 31, 1033-40	7	78
32	The UK10K project identifies rare variants in health and disease. *Nature*, **2015**, 526, 82-90	50.4	776
31	Targeted Next-Generation Sequencing Analysis of 1,000 Individuals with Intellectual Disability. *Human Mutation*, **2015**, 36, 1197-204	4.7	122
30	Joint analysis of psychiatric disorders increases accuracy of risk prediction for schizophrenia, bipolar disorder, and major depressive disorder. *American Journal of Human Genetics*, **2015**, 96, 283-94	11	161
29	Identification of a human synaptotagmin-1 mutation that perturbs synaptic vesicle cycling. *Journal of Clinical Investigation*, **2015**, 125, 1670-8	15.9	57
28	An inherited duplication at the gene p21 Protein-Activated Kinase 7 (PAK7) is a risk factor for psychosis. *Human Molecular Genetics*, **2014**, 23, 3316-26	5.6	32
27	De novo loss-of-function mutations in SETD5, encoding a methyltransferase in a 3p25 microdeletion syndrome critical region, cause intellectual disability. *American Journal of Human Genetics*, **2014**, 94, 618-24	11	74
26	Reciprocal duplication of the Williams-Beuren syndrome deletion on chromosome 7q11.23 is associated with schizophrenia. *Biological Psychiatry*, **2014**, 75, 371-7	7.9	54
25	Genetic relationship between five psychiatric disorders estimated from genome-wide SNPs. *Nature Genetics*, **2013**, 45, 984-94	36.3	1628
24	Bipolar Disorder is associated with the rs6971 polymorphism in the gene encoding 18 kDa Translocator Protein (TSPO). *Psychoneuroendocrinology*, **2013**, 38, 2826-9	5	37
23	Reduced burden of very large and rare CNVs in bipolar affective disorder. *Bipolar Disorders*, **2013**, 15, 893-8	3.8	22

22	Novel ATP2A2 mutations in a large sample of individuals with Darier disease. *Journal of Dermatology*, **2013**, 40, 259-66	1.6	18
21	Independent estimation of the frequency of rare CNVs in the UK population confirms their role in schizophrenia. *Schizophrenia Research*, **2012**, 135, 1-7	3.6	62
20	Identification of a CACNA2D4 deletion in late onset bipolar disorder patients and implications for the involvement of voltage-dependent calcium channels in psychiatric disorders. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, **2012**, 159B, 465-75	3.5	19
19	Genetic heterogeneity according to age at onset in bipolar disorder: a combined positional cloning and candidate gene approach. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, **2012**, 159B, 653-9	3.5	13
18	Polymorphism of the 5-HT transporter and response to antidepressants: randomised controlled trial. *British Journal of Psychiatry*, **2011**, 198, 464-71	5.4	51
17	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
16	Identification of high risk DISC1 protein structural variants in patients with bipolar spectrum disorder. *Neuroscience Letters*, **2010**, 486, 136-40	3.3	27
15	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. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, **2010**, 153B, 878-84	3.5	2
14	Rare copy number variants: a point of rarity in genetic risk for bipolar disorder and schizophrenia. *Archives of General Psychiatry*, **2010**, 67, 318-27		154
13	Affective temperaments across the bipolar-unipolar spectrum: examination of the TEMPS-A in 927 patients and controls. *Journal of Affective Disorders*, **2010**, 123, 42-51	6.6	33
12	Variation at the GABAA receptor gene, Rho 1 (GABRR1) associated with susceptibility to bipolar schizoaffective disorder. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, **2010**, 153B, 1347-9	3.5	14
11	Support for the involvement of large copy number variants in the pathogenesis of schizophrenia. *Human Molecular Genetics*, **2009**, 18, 1497-503	5.6	346
10	P2RX7: A bipolar and unipolar disorder candidate susceptibility gene?. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics*, **2009**, 150B, 1063-9	3.5	47
9	Microduplications of 16p11.2 are associated with schizophrenia. *Nature Genetics*, **2009**, 41, 1223-7	36.3	550
8	Collaborative genome-wide association analysis supports a role for ANK3 and CACNA1C in bipolar disorder. *Nature Genetics*, **2008**, 40, 1056-8	36.3	949
7	Support for neuregulin 1 as a susceptibility gene for bipolar disorder and schizophrenia. *Biological Psychiatry*, **2008**, 64, 419-27	7.9	95
6	Association scan of 14,500 nonsynonymous SNPs in four diseases identifies autoimmunity variants. *Nature Genetics*, **2007**, 39, 1329-37	36.3	1130
5	Variation at the DAOA/G30 locus influences susceptibility to major mood episodes but not psychosis in schizophrenia and bipolar disorder. *Archives of General Psychiatry*, **2006**, 63, 366-73		124

4	Operation of the schizophrenia susceptibility gene, neuregulin 1, across traditional diagnostic boundaries to increase risk for bipolar disorder. *Archives of General Psychiatry*, **2005**, 62, 642-8		209
3	Complex Structural Variants Resolved by Short-Read and Long-Read Whole Genome Sequencing in Mendelian Disorders		4
2	Gene-Based Analysis in HRC Imputed Genome Wide Association Data Identifies Three Novel Genes For Alzheimer□ Disease		1
1	The Impact of COVID-19 on Cancer Symptom Experience and Help-Seeking Behaviour in the United Kingdom: A Cross-Sectional Population Survey. *SSRN Electronic Journal*,	1	3