Alessia Fiorentino

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/3067347/publications.pdf

Version: 2024-02-01

25 papers	2,746 citations	430874 18 h-index	610901 24 g-index
27 all docs	27 docs citations	27 times ranked	3222 citing authors

#	Article	IF	Citations
1	Mapping genomic loci implicates genes and synaptic biology in schizophrenia. Nature, 2022, 604, 502-508.	27.8	929
2	Genome-wide association study of more than 40,000 bipolar disorder cases provides new insights into the underlying biology. Nature Genetics, 2021, 53, 817-829.	21.4	629
3	Rare coding variants in ten genes confer substantial risk for schizophrenia. Nature, 2022, 604, 509-516.	27.8	326
4	Mutations in REEP6 Cause Autosomal-Recessive Retinitis Pigmentosa. American Journal of Human Genetics, 2016, 99, 1305-1315.	6.2	121
5	Brain Derived Neurotrophic Factor (BDNF) Expression Is Regulated by MicroRNAs miR-26a and miR-26b Allele-Specific Binding. PLoS ONE, 2011, 6, e28656.	2.5	110
6	A Rare Functional Noncoding Variant at the GWAS-Implicated MIR137/MIR2682 Locus Might Confer Risk to Schizophrenia and Bipolar Disorder. American Journal of Human Genetics, 2014, 95, 744-753.	6.2	91
7	Structural Variants Create New Topological-Associated Domains and Ectopic Retinal Enhancer-Gene Contact in Dominant Retinitis Pigmentosa. American Journal of Human Genetics, 2020, 107, 802-814.	6.2	75
8	Mutations in the Spliceosome Component CWC27 Cause Retinal Degeneration with or without Additional Developmental Anomalies. American Journal of Human Genetics, 2017, 100, 592-604.	6.2	61
9	Association of rare variation in the glutamate receptor gene SLC1A2 with susceptibility to bipolar disorder and schizophrenia. European Journal of Human Genetics, 2015, 23, 1200-1206.	2.8	45
10	Analysis of ANK3 and CACNA1C variants identified in bipolar disorder whole genome sequence data. Bipolar Disorders, 2014, 16, 583-591.	1.9	44
11	Genetic Association, Mutation Screening, and Functional Analysis of a Kozak Sequence Variant in the Metabotropic Glutamate Receptor 3 Gene in Bipolar Disorder. JAMA Psychiatry, 2013, 70, 591.	11.0	43
12	Phenopolis: an open platform for harmonization and analysis of genetic and phenotypic data. Bioinformatics, 2017, 33, 2421-2423.	4.1	40
13	GUCY2D-Associated Leber Congenital Amaurosis: A Retrospective Natural History Study in Preparation for Trials of Novel Therapies. American Journal of Ophthalmology, 2020, 210, 59-70.	3.3	39
14	The functional GRM3 Kozak sequence variant rs148754219 affects the risk of schizophrenia and alcohol dependence as well as bipolar disorder. Psychiatric Genetics, 2014, 24, 277-278.	1.1	33
15	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.	6.2	26
16	Recessive Retinopathy Consequent on Mutant G-Protein Î² Subunit 3 (GNB3). JAMA Ophthalmology, 2016, 134, 924.	2.5	25
17	Loss-of-Function Mutations in the CFH Gene Affecting Alternatively Encoded Factor H-like 1 Protein Cause Dominant Early-Onset Macular Drusen. Ophthalmology, 2019, 126, 1410-1421.	5.2	25
18	Missense variants in the X-linked genePRPS1cause retinal degeneration in females. Human Mutation, 2018, 39, 80-91.	2.5	23

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19	Characterisation of age and polarity at onset in bipolar disorder. British Journal of Psychiatry, 2021, 219, 659-669.	2.8	20
20	Exome sequence analysis and follow up genotyping implicates rare ULK1 variants to be involved in susceptibility to schizophrenia. Annals of Human Genetics, 2018, 82, 88-92.	0.8	16
21	Genetic variation in the miRâ€708 gene and its binding targets in bipolar disorder. Bipolar Disorders, 2016, 18, 650-656.	1.9	14
22	Novel homozygous splicing mutations in cause autosomal recessive retinitis pigmentosa. Molecular Vision, 2018, 24, 603-612.	1.1	6
23	Genetic variant analysis of the putative regulatory regions of the LRRC7 gene in bipolar disorder. Psychiatric Genetics, 2016, 26, 99-100.	1.1	2
24	Genetic association and functional characterization of MCPH1 gene variation in bipolar disorder and schizophrenia. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2019, 180, 258-265.	1.7	2
25	Association study of rare nonsynonymous variants of FTO in bipolar disorder. Psychiatric Genetics, 2016, 26, 140-141.	1.1	0