Pelagia Deriziotis

List of Publications by Year in descending order

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23 papers	2,418 citations	361413 20 h-index	552781 26 g-index
26 all docs	26 docs citations	26 times ranked	4913 citing authors

#	Article	IF	Citations
1	Exome sequencing in sporadic autism spectrum disorders identifies severe de novo mutations. Nature Genetics, 2011, 43, 585-589.	21.4	1,080
2	Disease-Associated Prion Protein Oligomers Inhibit the 26S Proteasome. Molecular Cell, 2007, 26, 175-188.	9.7	237
3	Genetic risk factors for variant Creutzfeldtâ€“Jakob disease: a genome-wide association study. Lancet Neurology, The, 2009, 8, 57-66.	10.2	131
4	BCL11A Haploinsufficiency Causes an Intellectual Disability Syndrome and Dysregulates Transcription. American Journal of Human Genetics, 2016, 99, 253-274.	6.2	118
5	De novo TBR1 mutations in sporadic autism disrupt protein functions. Nature Communications, 2014, 5, 4954.	12.8	109
6	Misfolded PrP impairs the UPS by interaction with the 20S proteasome and inhibition of substrate entry. EMBO Journal, 2011, 30, 3065-3077.	7.8	104
7	CHD3 helicase domain mutations cause a neurodevelopmental syndrome with macrocephaly and impaired speech and language. Nature Communications, 2018, 9, 4619.	12.8	70
8	Identification and functional characterization ofde novo FOXP1variants provides novel insights into the etiology of neurodevelopmental disorder. Human Molecular Genetics, 2016, 25, 546-557.	2.9	69
9	Speech and Language: Translating the Genome. Trends in Genetics, 2017, 33, 642-656.	6.7	57
10	Proteomic analysis of FOXP proteins reveals interactions between cortical transcription factors associated with neurodevelopmental disorders. Human Molecular Genetics, 2018, 27, 1212-1227.	2.9	53
11	A de novo FOXP1 variant in a patient with autism, intellectual disability and severe speech and language impairment. European Journal of Human Genetics, 2015, 23, 1702-1707.	2.8	45
12	The DISC1 promoter: characterization and regulation by FOXP2. Human Molecular Genetics, 2012, 21, 2862-2872.	2.9	39
13	The language-related transcription factor FOXP2 is post-translationally modified with small ubiquitin-like modifiers. Scientific Reports, 2016, 6, 20911.	3.3	38
14	Overlapping SETBP1 gain-of-function mutations in Schinzel-Giedion syndrome and hematologic malignancies. PLoS Genetics, 2017, 13, e1006683.	3.5	35
15	De Novo Variants Disturbing the Transactivation Capacity of POU3F3 Cause a Characteristic Neurodevelopmental Disorder. American Journal of Human Genetics, 2019, 105, 403-412.	6.2	35
16	Insights into the Genetic Foundations of Human Communication. Neuropsychology Review, 2015, 25, 3-26.	4.9	33
17	Neurogenomics of speech and language disorders: the road ahead. Genome Biology, 2013, 14, 204.	9.6	28
18	Equivalent missense variant in the FOXP2 and FOXP1 transcription factors causes distinct neurodevelopmental disorders. Human Mutation, 2017, 38, 1542-1554.	2.5	28

#	Article	IF	Citations
19	Functional characterization of rare FOXP2 variants in neurodevelopmental disorder. Journal of Neurodevelopmental Disorders, 2016, 8, 44.	3.1	26
20	Functional characterization of TBR1 variants in neurodevelopmental disorder. Scientific Reports, 2018, 8, 14279.	3.3	26
21	Prions and the proteasome. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2008, 1782, 713-722.	3.8	25
22	Investigating Protein-protein Interactions in Live Cells Using Bioluminescence Resonance Energy Transfer. Journal of Visualized Experiments, 2014, , .	0.3	17
23	Proteomic analysis of FOXP proteins reveals interactions between cortical transcription factors associated with neurodevelopmental disorders. Human Molecular Genetics, 2018, , .	2.9	2