Pelagia Deriziotis

List of Publications by Year in descending order

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#	Article	IF	CITATIONS
1	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
2	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
3	Functional characterization of TBR1 variants in neurodevelopmental disorder. Scientific Reports, 2018, 8, 14279.	3.3	26
4	CHD3 helicase domain mutations cause a neurodevelopmental syndrome with macrocephaly and impaired speech and language. Nature Communications, 2018, 9, 4619.	12.8	70
5	Proteomic analysis of FOXP proteins reveals interactions between cortical transcription factors associated with neurodevelopmental disorders. Human Molecular Genetics, 2018, , .	2.9	2
6	Equivalent missense variant in the <i>FOXP2</i> and <i>FOXP1</i> transcription factors causes distinct neurodevelopmental disorders. Human Mutation, 2017, 38, 1542-1554.	2.5	28
7	Speech and Language: Translating the Genome. Trends in Genetics, 2017, 33, 642-656.	6.7	57
8	Overlapping SETBP1 gain-of-function mutations in Schinzel-Giedion syndrome and hematologic malignancies. PLoS Genetics, 2017, 13, e1006683.	3.5	35
9	The language-related transcription factor FOXP2 is post-translationally modified with small ubiquitin-like modifiers. Scientific Reports, 2016, 6, 20911.	3.3	38
10	BCL11A Haploinsufficiency Causes an Intellectual Disability Syndrome and Dysregulates Transcription. American Journal of Human Genetics, 2016, 99, 253-274.	6.2	118
11	Functional characterization of rare FOXP2 variants in neurodevelopmental disorder. Journal of Neurodevelopmental Disorders, 2016, 8, 44.	3.1	26
12	Identification and functional characterization of <i>de novo FOXP1</i> variants provides novel insights into the etiology of neurodevelopmental disorder. Human Molecular Genetics, 2016, 25, 546-557.	2.9	69
13	Insights into the Genetic Foundations of Human Communication. Neuropsychology Review, 2015, 25, 3-26.	4.9	33
14	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
15	De novo TBR1 mutations in sporadic autism disrupt protein functions. Nature Communications, 2014, 5, 4954.	12.8	109
16	Investigating Protein-protein Interactions in Live Cells Using Bioluminescence Resonance Energy Transfer. Journal of Visualized Experiments, 2014, , .	0.3	17
17	Neurogenomics of speech and language disorders: the road ahead. Genome Biology, 2013, 14, 204.	9.6	28
18	The DISC1 promoter: characterization and regulation by FOXP2. Human Molecular Genetics, 2012, 21, 2862-2872.	2.9	39

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#	Article	IF	CITATIONS
19	Exome sequencing in sporadic autism spectrum disorders identifies severe de novo mutations. Nature Genetics, 2011, 43, 585-589.	21.4	1,080
20	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
21	Genetic risk factors for variant Creutzfeldt–Jakob disease: a genome-wide association study. Lancet Neurology, The, 2009, 8, 57-66.	10.2	131
22	Prions and the proteasome. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2008, 1782, 713-722.	3.8	25
23	Disease-Associated Prion Protein Oligomers Inhibit the 26S Proteasome. Molecular Cell, 2007, 26, 175-188.	9.7	237