

Lot Snijders Blok

List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/7097292/publications.pdf>

Version: 2024-02-01

14
papers

722
citations

759233

12
h-index

940533

16
g-index

17
all docs

17
docs citations

17
times ranked

1612
citing authors

#	ARTICLE	IF	CITATIONS
1	Mutations in DDX3X Are a Common Cause of Unexplained Intellectual Disability with Gender-Specific Effects on Wnt Signaling. <i>American Journal of Human Genetics</i> , 2015, 97, 343-352.	6.2	230
2	Pathogenic DDX3X Mutations Impair RNA Metabolism and Neurogenesis during Fetal Cortical Development. <i>Neuron</i> , 2020, 106, 404-420.e8.	8.1	121
3	CHD3 helicase domain mutations cause a neurodevelopmental syndrome with macrocephaly and impaired speech and language. <i>Nature Communications</i> , 2018, 9, 4619.	12.8	70
4	De novo mutations in MED13, a component of the Mediator complex, are associated with a novel neurodevelopmental disorder. <i>Human Genetics</i> , 2018, 137, 375-388.	3.8	46
5	<i>NBEA</i>: Developmental disease gene with early generalized epilepsy phenotypes. <i>Annals of Neurology</i> , 2018, 84, 788-795.	5.3	44
6	Mutation update for the <i>SATB2</i> gene. <i>Human Mutation</i> , 2019, 40, 1013-1029.	2.5	38
7	De Novo Variants Disturbing the Transactivation Capacity of POU3F3 Cause a Characteristic Neurodevelopmental Disorder. <i>American Journal of Human Genetics</i> , 2019, 105, 403-412.	6.2	35
8	Mutation-specific pathophysiological mechanisms define different neurodevelopmental disorders associated with SATB1 dysfunction. <i>American Journal of Human Genetics</i> , 2021, 108, 346-356.	6.2	30
9	Mutations of the Transcriptional Corepressor ZMYM2 Cause Syndromic Urinary Tract Malformations. <i>American Journal of Human Genetics</i> , 2020, 107, 727-742.	6.2	25
10	A second cohort of CHD3 patients expands the molecular mechanisms known to cause Snijders Blok-Campeau syndrome. <i>European Journal of Human Genetics</i> , 2020, 28, 1422-1431.	2.8	25
11	Loss-of-function and missense variants in NSD2 cause decreased methylation activity and are associated with a distinct developmental phenotype. <i>Genetics in Medicine</i> , 2021, 23, 1474-1483.	2.4	24
12	Heterozygous variants that disturb the transcriptional repressor activity of FOXP4 cause a developmental disorder with speech/language delays and multiple congenital abnormalities. <i>Genetics in Medicine</i> , 2021, 23, 534-542.	2.4	17
13	Inherited variants in CHD3 show variable expressivity in Snijders Blok-Campeau syndrome. <i>Genetics in Medicine</i> , 2022, 24, 1283-1296.	2.4	9
14	Speech&language profiles in the context of cognitive and adaptive functioning in SATB2 &associated syndrome. <i>Genes, Brain and Behavior</i> , 2021, 20, e12761.	2.2	4