

Sophie Ehresmann

List of Publications by Year in descending order

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Version: 2024-02-01

10
papers

378
citations

933447

10
h-index

1199594

12
g-index

12
all docs

12
docs citations

12
times ranked

917
citing authors

#	ARTICLE	IF	CITATIONS
1	A variant of neonatal progeroid syndrome, or Wiedemann-Rautenstrauch syndrome, is associated with a nonsense variant in POLR3GL. <i>European Journal of Human Genetics</i> , 2020, 28, 461-468.	2.8	16
2	De Novo KAT5 Variants Cause a Syndrome with Recognizable Facial Dysmorphisms, Cerebellar Atrophy, Sleep Disturbance, and Epilepsy. <i>American Journal of Human Genetics</i> , 2020, 107, 564-574.	6.2	14
3	Mutations in ACTL6B Cause Neurodevelopmental Deficits and Epilepsy and Lead to Loss of Dendrites in Human Neurons. <i>American Journal of Human Genetics</i> , 2019, 104, 815-834.	6.2	59
4	A Syndromic Neurodevelopmental Disorder Caused by Mutations in SMARCD1, a Core SWI/SNF Subunit Needed for Context-Dependent Neuronal Gene Regulation in Flies. <i>American Journal of Human Genetics</i> , 2019, 104, 596-610.	6.2	32
5	Missense Variants in the Histone Acetyltransferase Complex Component Gene TRRAP Cause Autism and Syndromic Intellectual Disability. <i>American Journal of Human Genetics</i> , 2019, 104, 530-541.	6.2	30
6	Loss of Oxidation Resistance 1, OXR1, Is Associated with an Autosomal-Recessive Neurological Disease with Cerebellar Atrophy and Lysosomal Dysfunction. <i>American Journal of Human Genetics</i> , 2019, 105, 1237-1253.	6.2	34
7	Expanding the Spectrum of BAF-Related Disorders: De Novo Variants in SMARCC2 Cause a Syndrome with Intellectual Disability and Developmental Delay. <i>American Journal of Human Genetics</i> , 2019, 104, 164-178.	6.2	59
8	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
9	Lowry-Wood syndrome: further evidence of association with RNU4ATAC, and correlation between genotype and phenotype. <i>Human Genetics</i> , 2018, 137, 905-909.	3.8	11
10	Mutations in GPAA1, Encoding a GPI Transamidase Complex Protein, Cause Developmental Delay, Epilepsy, Cerebellar Atrophy, and Osteopenia. <i>American Journal of Human Genetics</i> , 2017, 101, 856-865.	6.2	49