# Sophie Ehresmann

List of Publications by Year
in descending order



| 10 | 378 | 933447 10 | 1199594 12 |
|---|---|---|---|
| papers | citations | h-index | g-index |

| 12 | 12 | 12 | 917 |
|---|---|---|---|
| all docs | docs citations | times ranked | citing authors |

| # | Article | IF | Citations |
|---|---|---|---|
| 1 | CHD3 helicase domain mutations cause a neurodevelopmental syndrome with macrocephaly and impaired speech and language. Nature Communications, 2018, 9, 4619. | 12.8 | 70 |
| 2 | Mutations in ACTL6B Cause Neurodevelopmental Deficits and Epilepsy and Lead to Loss of Dendrites in Human Neurons. American Journal of Human Genetics, 2019, 104, 815-834. | 6.2 | 59 |
| 3 | Expanding the Spectrum of BAF-Related Disorders: De Novo Variants in SMARCC2 Cause a Syndrome with Intellectual Disability and Developmental Delay. American Journal of Human Genetics, 2019, 104, 164-178. | 6.2 | 59 |
| 4 | Mutations in GPAA1 , Encoding a GPI Transamidase Complex Protein, Cause Developmental Delay, Epilepsy, Cerebellar Atrophy, and Osteopenia. American Journal of Human Genetics, 2017, 101, 856-865. | 6.2 | 49 |
| 5 | Loss of Oxidation Resistance 1, OXR1, Is Associated with an Autosomal-Recessive Neurological Disease with Cerebellar Atrophy and Lysosomal Dysfunction. American Journal of Human Genetics, 2019, 105, 1237-1253. | 6.2 | 34 |
| 6 | A Syndromic Neurodevelopmental Disorder Caused by Mutations in SMARCD1, a Core SWI/SNF Subunit Needed for Context-Dependent Neuronal Gene Regulation in Flies. American Journal of Human Genetics, 2019, 104, 596-610. | 6.2 | 32 |
| 7 | Missense Variants in the Histone Acetyltransferase Complex Component Gene TRRAP Cause Autism and Syndromic Intellectual Disability. American Journal of Human Genetics, 2019, 104, 530-541. | 6.2 | 30 |
| 8 | A variant of neonatal progeroid syndrome, or Wiedemannâ€“Rautenstrauch syndrome, is associated with a nonsense variant in POLR3GL. European Journal of Human Genetics, 2020, 28, 461-468. | 2.8 | 16 |
| 9 | De Novo KAT5 Variants Cause a Syndrome with Recognizable Facial Dysmorphisms, Cerebellar Atrophy, Sleep Disturbance, and Epilepsy. American Journal of Human Genetics, 2020, 107, 564-574. | 6.2 | 14 |
| 10 | Lowry-Wood syndrome: further evidence of association with RNU4ATAC, and correlation between genotype and phenotype. Human Genetics, 2018, 137, 905-909. | 3.8 | 11 |