

Sarah L Sawyer

List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/7239024/publications.pdf>

Version: 2024-02-01

19
papers

987
citations

758635

12
h-index

794141

19
g-index

20
all docs

20
docs citations

20
times ranked

2839
citing authors

#	ARTICLE	IF	CITATIONS
1	Alternative genomic diagnoses for individuals with a clinical diagnosis of Dubowitz syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 119-133.	0.7	17
2	SPEN haploinsufficiency causes a neurodevelopmental disorder overlapping proximal 1p36 deletion syndrome with an epismutation of X chromosomes in females. American Journal of Human Genetics, 2021, 108, 502-516.	2.6	48
3	Homozygous <i>WNT9B</i> variants in two families with bilateral renal agenesis/hypoplasia/dysplasia. American Journal of Medical Genetics, Part A, 2021, 185, 3005-3011.	0.7	5
4	Further delineation of the clinical spectrum of <i>KAT6B</i> disorders and allelic series of pathogenic variants. Genetics in Medicine, 2020, 22, 1338-1347.	1.1	25
5	Phenotate: crowdsourcing phenotype annotations as exercises in undergraduate classes. Genetics in Medicine, 2020, 22, 1391-1400.	1.1	2
6	De Novo Variants in <i>SPOP</i> Cause Two Clinically Distinct Neurodevelopmental Disorders. American Journal of Human Genetics, 2020, 106, 405-411.	2.6	8
7	Infantile Myofibromatosis With Intracranial Extradural Involvement and <i>PDGFRB</i> Mutation: A Case Report and Review of the Literature. Pediatric and Developmental Pathology, 2019, 22, 258-264.	0.5	11
8	Broad spectrum of neuropsychiatric phenotypes associated with white matter disease in <i>PTEN</i> hamartoma tumor syndrome. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2018, 177, 101-109.	1.1	34
9	Benchmarking outcomes in the Neonatal Intensive Care Unit: Cytogenetic and molecular diagnostic rates in a retrospective cohort. American Journal of Medical Genetics, Part A, 2017, 173, 1839-1847.	0.7	25
10	<i>MCM3AP</i> in recessive Charcot-Marie-Tooth neuropathy and mild intellectual disability. Brain, 2017, 140, 2093-2103.	3.7	31
11	H3.1 K36M mutation in a congenital-onset soft tissue neoplasm. Pediatric Blood and Cancer, 2017, 64, e26633.	0.8	7
12	Late diagnosis of cerebral folate deficiency: Fewer seizures with folinic acid in adult siblings. Neurology: Genetics, 2016, 2, e38.	0.9	11
13	Histone H3K36 mutations promote sarcomagenesis through altered histone methylation landscape. Science, 2016, 352, 844-849.	6.0	327
14	Biallelic Mutations in <i>BRCA1</i> Cause a New Fanconi Anemia Subtype. Cancer Discovery, 2015, 5, 135-142.	7.7	251
15	Homozygous mutations in <i>MFN2</i> cause multiple symmetric lipomatosis associated with neuropathy. Human Molecular Genetics, 2015, 24, 5109-5114.	1.4	61
16	Brain malformations in a patient with deletion 2p16.1: A refinement of the phenotype to <i>BCL11A</i> . European Journal of Medical Genetics, 2015, 58, 351-354.	0.7	24
17	Receptor tyrosine kinase mutations in developmental syndromes and cancer: two sides of the same coin. Human Molecular Genetics, 2015, 24, R60-R66.	1.4	70
18	Atypical fibrodysplasia ossificans progressiva diagnosed by whole-exome sequencing. American Journal of Medical Genetics, Part A, 2015, 167, 1337-1341.	0.7	11

#	ARTICLE	IF	CITATIONS
19	Longer Term Survival of a Child With Autosomal Recessive Cutis Laxa Due to a Mutation in <i>FBLN4</i> . American Journal of Medical Genetics, Part A, 2013, 161, 1148-1153.	0.7	19