Sarah L Sawyer

List of Publications by Year
in descending order

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	Histone H3K36 mutations promote sarcomagenesis through altered histone methylation landscape. Science, 2016, 352, 844-849.	6.0	327
2	Biallelic Mutations in BRCA1 Cause a New Fanconi Anemia Subtype. Cancer Discovery, 2015, 5, 135-142.	7.7	251
3	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
4	Homozygous mutations inMFN2cause multiple symmetric lipomatosis associated with neuropathy. Human Molecular Genetics, 2015, 24, 5109-5114.	1.4	61
5	SPEN haploinsufficiency causes a neurodevelopmental disorder overlapping proximal 1p36 deletion syndrome with an episignature of X chromosomes in females. American Journal of Human Genetics, 2021, 108, 502-516.	2.6	48
6	Broad spectrum of neuropsychiatric phenotypes associated with white matter disease in PTEN hamartoma tumor syndrome. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2018, 177, 101-109.	1.1	34
7	MCM3AP in recessive Charcot-Marie-Tooth neuropathy and mild intellectual disability. Brain, 2017, 140, 2093-2103.	3.7	31
8	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
9	Further delineation of the clinical spectrum of KAT6B disorders and allelic series of pathogenic variants. Genetics in Medicine, 2020, 22, 1338-1347.	1.1	25
10	Brain malformations in a patient with deletion 2p16.1: A refinement ofÂthe phenotype to BCL11A. European Journal of Medical Genetics, 2015, 58, 351-354.	0.7	24
11	Longer Term Survival of a Child With Autosomal Recessive Cutis Laxa Due to a Mutation in FBLN4. American Journal of Medical Genetics, Part A, 2013, 161, 1148-1153.	0.7	19
12	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
13	Atypical fibrodysplasia ossificans progressiva diagnosed by wholeâ€exome sequencing. American Journal of Medical Genetics, Part A, 2015, 167, 1337-1341.	0.7	11
14	Late diagnosis of cerebral folate deficiency: Fewer seizures with folinic acid in adult siblings. Neurology: Genetics, 2016, 2, e38.	0.9	11
15	Infantile Myofibromatosis With Intracranial Extradural Involvement and PDGFRB Mutation: A Case Report and Review of the Literature. Pediatric and Developmental Pathology, 2019, 22, 258-264.	0.5	11
16	De Novo Variants in SPOP Cause Two Clinically Distinct Neurodevelopmental Disorders. American Journal of Human Genetics, 2020, 106, 405-411.	2.6	8
17	H3.1 K36M mutation in a congenitalâ€onset soft tissue neoplasm. Pediatric Blood and Cancer, 2017, 64, e26633.	0.8	7
18	Homozygous <scp>WNT9B</scp> variants in two families with bilateral renal agenesis/hypoplasia/dysplasia. American Journal of Medical Genetics, Part A, 2021, 185, 3005-3011.	0.7	5

#	Article	IF	Citations
19	Phenotate: crowdsourcing phenotype annotations as exercises in undergraduate classes. Genetics in Medicine, 2020, 22, 1391-1400.	1.1	2