Suzanne Me Lewis

List of Publications by Year
in descending order

24 papers	1,752 citations	643344 15 h-index	721071 23 g-index
24 all docs	24 docs citations	24 times ranked	4594 citing authors

#	Article	IF	Citations
1	Novel diagnostic DNA methylation episignatures expand and refine the epigenetic landscapes of Mendelian disorders. Human Genetics and Genomics Advances, 2022, 3, 100075.	1.0	42
2	Genome-wide sequencing and the clinical diagnosis of genetic disease: The CAUSES study. Human Genetics and Genomics Advances, 2022, 3, 100108.	1.0	7
3	Contribution of Multiple Inherited Variants to Autism Spectrum Disorder (ASD) in a Family with 3 Affected Siblings. Genes, 2021, 12, 1053.	1.0	12
4	Exome sequencing identified a de novo mutation of PURA gene in a patient with familial Xp22.31 microduplication. European Journal of Medical Genetics, 2019, 62, 103-108.	0.7	17
5	RAPIDOMICS: rapid genome-wide sequencing in a neonatal intensive care unitâ€"successes and challenges. European Journal of Pediatrics, 2019, 178, 1207-1218.	1.3	59
6	Whole genome sequencing and variant discovery in the ASPIRE autism spectrum disorder cohort. Clinical Genetics, 2019, 96, 199-206.	1.0	18
7	Blood Mitochondrial DNA Content in HIV-Exposed Uninfected Children with Autism Spectrum Disorder. Viruses, 2018, 10, 77.	1.5	15
8	The phenotypic spectrum of Schaaf-Yang syndrome: 18 new affected individuals from 14 families. Genetics in Medicine, 2017, 19, 45-52.	1.1	94
9	Exome Sequencing and the Management of Neurometabolic Disorders. New England Journal of Medicine, 2016, 374, 2246-2255.	13.9	254
10	Exome sequencing identifies pathogenic variants of VPS13B in a patient with familial 16p11.2 duplication. BMC Medical Genetics, 2016, 17, 78.	2.1	13
11	Loss-of-function mutations inSCN4Acause severe foetal hypokinesia or â€˜classicalâ€™ congenital myopathy. Brain, 2016, 139, 674-691.	3.7	100
12	Identifying candidate genes for 2p15p16.1 microdeletion syndrome using clinical, genomic, and functional analysis. JCI Insight, 2016, 1, e85461.	2.3	22
13	MG-118â€...Towards understanding phenotypic variability using exome sequencing. Journal of Medical Genetics, 2015, 52, A4.3-A5.	1.5	0
14	Functional consequences of copy number variants in miscarriage. Molecular Cytogenetics, 2015, 8, 6.	0.4	17
15	16p11.2 600 kb Duplications confer risk for typical and atypical Rolandic epilepsy. Human Molecular Genetics, 2014, 23, 6069-6080.	1.4	61
16	Copy number variants (CNVs) analysis in a deeply phenotyped cohort of individuals with intellectual disability (ID). BMC Medical Genetics, 2014, 15, 82.	2.1	12
17	Opposite effects on facial morphology due to gene dosage sensitivity. Human Genetics, 2014, 133, 1117-1125.	1.8	16
18	Phenotypic and functional consequences of haploinsufficiency of genes from exocyst and retinoic acid pathway due to a recurrent microdeletion of 2p13.2. Orphanet Journal of Rare Diseases, 2013, 8, 100.	1.2	24

#	Article	IF	Citations
19	miRNA and miRNA target genes in copy number variations occurring in individuals with intellectual disability. BMC Genomics, 2013, 14, 544.	1.2	18
20	DRD2 and PPP1R1B (DARPP-32) polymorphisms independently confer increased risk for autism spectrum disorders and additively predict affected status in male-only affected sib-pair families. Behavioral and Brain Functions, 2012, 8, 19.	1.4	53
21	Mirror extreme BMI phenotypes associated with gene dosage at the chromosome 16p11.2 locus. Nature, 2011, 478, 97-102.	13.7	394
22	Understanding the impact of 1q21.1 copy number variant. Orphanet Journal of Rare Diseases, 2011, 6, 54.	1.2	48
23	Complete Rescue of Lipoprotein Lipaseâ€"Deficient Mice by Somatic Gene Transfer of the Naturally Occurring LPL S447X Beneficial Mutation. Arteriosclerosis, Thrombosis, and Vascular Biology, 2005, 25, 2143-2150.	1.1	60
24	Mutations in the human ortholog of Aristaless cause X-linked mental retardation and epilepsy. Nature Genetics, 2002, 30, 441-445.	9.4	396