Sonia Mayo

List of Publications by Year
in descending order

23 papers	789 citations	840776 11 h-index	610901 24 g-index
24 all docs	24 docs citations	24 times ranked	1868 citing authors

#	Article	IF	Citations
1	The Arabidopsis heavy metal P-type ATPase HMA5 interacts with metallochaperones and functions in copper detoxification of roots. Plant Journal, 2006, 45, 225-236.	5.7	290
2	High diagnostic yield of syndromic intellectual disability by targeted next-generation sequencing. Journal of Medical Genetics, 2017, 54, 87-92.	3.2	93
3	Identification of Intellectual Disability Genes in Female Patients with a Skewed X-Inactivation Pattern. Human Mutation, 2016, 37, 804-811.	2.5	92
4	Copy-Number Gains of HUWE1 Due to Replication- and Recombination-Based Rearrangements. American Journal of Human Genetics, 2012, 91, 252-264.	6.2	71
5	Detection of Hypomethylation Syndrome among Patients with Epigenetic Alterations at theGNASLocus. Journal of Clinical Endocrinology and Metabolism, 2012, 97, E1060-E1067.	3.6	37
6	Novel mutations of NFIX gene causing Marshall-Smith syndrome or Sotos-like syndrome: one gene, two phenotypes. Pediatric Research, 2015, 78, 533-539.	2.3	35
7	De novo mutations in genes of mediator complex causing syndromic intellectual disability: mediatorpathy or transcriptomopathy?. Pediatric Research, 2016, 80, 809-815.	2.3	27
8	Enrichment of ultraconserved elements among genomic imbalances causing mental delay and congenital anomalies. BMC Medical Genomics, 2010, 3, 54.	1.5	18
9	Phenotype profiling of patients with intellectual disability and copy number variations. European Journal of Paediatric Neurology, 2014, 18, 558-566.	1.6	18
10	Intragenic CNVs for epigenetic regulatory genes in intellectual disability: Survey identifies pathogenic and benign single exon changes. American Journal of Medical Genetics, Part A, 2016, 170, 2916-2926.	1.2	14
11	Candidate Genes for Eyelid Myoclonia with Absences, Review of the Literature. International Journal of Molecular Sciences, 2021, 22, 5609.	4.1	13
12	Pure duplication of 19p13.3 in three members of a family with intellectual disability and literature review. Definition of a new microduplication syndrome. American Journal of Medical Genetics, Part A, 2015, 167, 1614-1620.	1.2	12
13	Duplication at Xq13.3â€“q21.1 with syndromic intellectual disability, a probable role for the ATRX gene. American Journal of Medical Genetics, Part A, 2014, 164, 918-923.	1.2	10
14	Haploinsufficiency of the MYT1L gene causes intellectual disability frequently associated with behavioral disorder. Genetics in Medicine, 2015, 17, 683-684.	2.4	10
15	Chimeric Genes in Deletions and Duplications Associated with Intellectual Disability. International Journal of Genomics, 2017, 2017, 1-11.	1.6	10
16	Prenatal Diagnosis of a Female Fetus with Ring Chromosome 9, 46,XX,r(9)(p24q34), and a de novo Interstitial 9p Deletion. Cytogenetic and Genome Research, 2014, 144, 275-279.	1.1	8
17	Large deletion in the Factor <scp>VIII</scp> gene (<scp>F</scp>8) involving segmental duplications in int22h shows no haematological phenotype in female carriers, but may be embryonic lethal in males. British Journal of Haematology, 2012, 158, 138-140.	2.5	7
18	A novel missense mutation in the NSDHL gene identified in a Lithuanian family by targeted nextâ€generation sequencing causes CK syndrome. American Journal of Medical Genetics, Part A, 2015, 167, 1342-1348.	1.2	6

#	Article	IF	Citations
19	Hypomethylation of the KCNQ1OT1 imprinting center of chromosome 11 associated to Sotos-like features. Journal of Human Genetics, 2012, 57, 153-156.	2.3	4
20	Noninvasive prenatal testing: How far can we reach detecting fetal copy number variations. European Journal of Obstetrics, Gynecology and Reproductive Biology, 2022, 272, 150-155.	1.1	4
21	In Pursuit of New Imprinting Syndromes by Epimutation Screening in Idiopathic Neurodevelopmental Disorder Patients. BioMed Research International, 2015, 2015, 1-8.	1.9	3
22	CfDNA Measurement as a Diagnostic Tool for the Detection of Brain Somatic Mutations in Refractory Epilepsy. International Journal of Molecular Sciences, 2022, 23, 4879.	4.1	2
23	Noninvasive Prenatal Testing: Comparison of Two Mappers and Influence in the Diagnostic Yield. BioMed Research International, 2018, 2018, 1-6.	1.9	1