Maxime Cadieux-Dion

List of Publications by Year
in descending order

15 papers	760 citations	1040056 9 h-index	996975 15 g-index
15 all docs	15 docs citations	15 times ranked	2268 citing authors

#	Article	IF	Citations
1	Phenotypic expansion and variable expressivity in individuals with JARID2â€related intellectual disability: A case series. Clinical Genetics, 2022, 102, 136-141.	2.0	3
2	Delayed diagnosis of holocarboxylase synthetase deficiency in three patients with prominent skin findings. Pediatric Dermatology, 2021, 38, 655-658.	0.9	4
3	Factors Affecting Migration to GRCh38 in Laboratories Performing Clinical Next-Generation Sequencing. Journal of Molecular Diagnostics, 2021, 23, 651-657.	2.8	13
4	Autosomal-dominant adult neuronal ceroid lipofuscinosis caused by duplication in DNAJC5 initially missed by Sanger and whole-exome sequencing. European Journal of Human Genetics, 2020, 28, 783-789.	2.8	10
5	Variants in CHRNB2 and CHRNA4 Identified in Patients with Insular Epilepsy. Canadian Journal of Neurological Sciences, 2020, 47, 800-809.	0.5	7
6	Deficient histone H3 propionylation by BRPF1-KAT6 complexes in neurodevelopmental disorders and cancer. Science Advances, 2020, 6, eaax0021.	10.3	56
7	Haploinsufficiency of the Notch Ligand DLL1 Causes Variable Neurodevelopmental Disorders. American Journal of Human Genetics, 2019, 105, 631-639.	6.2	42
8	De Novo and Inherited Pathogenic Variants in KDM3B Cause Intellectual Disability, Short Stature, and Facial Dysmorphism. American Journal of Human Genetics, 2019, 104, 758-766.	6.2	34
9	Dual Molecular Effects of Dominant RORA Mutations Cause Two Variants of Syndromic Intellectual Disability with Either Autism or Cerebellar Ataxia. American Journal of Human Genetics, 2018, 102, 744-759.	6.2	51
10	On the verge of diagnosis: Detection, reporting, and investigation of de novo variants in novel genes identified by clinical sequencing. Human Mutation, 2018, 39, 1505-1516.	2.5	9
11	Global characterization of copy number variants in epilepsy patients from whole genome sequencing. PLoS Genetics, 2018, 14, e1007285.	3.5	50
12	Novel heterozygous pathogenic variants in CHUK in a patient with AEC-like phenotype, immune deficiencies and 1q21.1 microdeletion syndrome: a case report. BMC Medical Genetics, 2018, 19, 41.	2.1	6
13	High Rate of Recurrent De Novo Mutations in Developmental and Epileptic Encephalopathies. American Journal of Human Genetics, 2017, 101, 664-685.	6.2	337
14	Diagnosis and misdiagnosis of adult neuronal ceroid lipofuscinosis (Kufs disease). Neurology, 2016, 87, 579-584.	1.1	28
15	Expanding the Clinical Phenotype Associated WithELOVL4Mutation. JAMA Neurology, 2014, 71, 470.	9.0	110