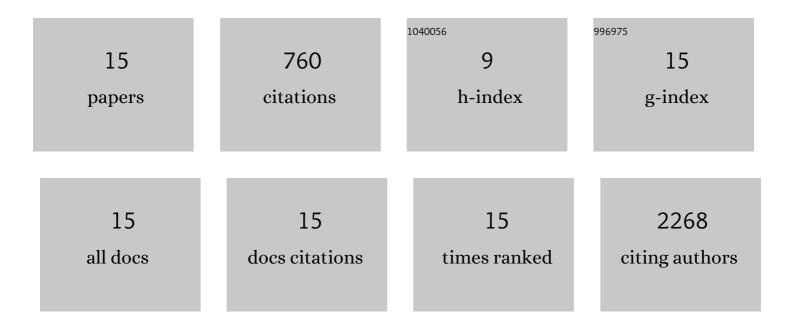
## Maxime Cadieux-Dion

List of Publications by Year in descending order

Source: https://exaly.com/author-pdf/3912572/publications.pdf Version: 2024-02-01



| #  | Article  | IF   | CITATIONS |
|----|--|------|-----------|
| 1  | Phenotypic expansion and variable expressivity in individuals with <i>JARID2</i> â€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<br>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<br>missed by Sanger and whole-exome sequencing. European Journal of Human Genetics, 2020, 28, 783-789.         | 2.8  | 10        |
| 5  | Variants in <i>CHRNB2</i> and <i>CHRNA4</i> Identified in Patients with Insular Epilepsy. Canadian<br>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<br>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<br>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<br>Disability with Either Autism or Cerebellar Ataxia. American Journal of Human Genetics, 2018, 102,<br>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.<br>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<br>Journal of Human Genetics, 2017, 101, 664-685.   | 6.2  | 337       |
| 14 | Diagnosis and misdiagnosis of adult neuronal ceroid lipofuscinosis (Kufs disease). Neurology, 2016,<br>87, 579-584.  | 1.1  | 28        |
| 15 | Expanding the Clinical Phenotype Associated With <i>ELOVL4</i> Mutation. JAMA Neurology, 2014, 71, 470.  | 9.0  | 110       |