# VÃ©ronique Geoffroy

List of Publications by Year
in descending order



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|---|---|---|---|
| 23 papers | 1,472 citations | 471509 17 h-index | 610901 24 g-index |
| 26 all docs | 26 docs citations | 26 times ranked | 3237 citing authors |

| # | ARTICLE | IF | CITATIONS |
|---|---|---|---|
| 1 | AnnotSV: an integrated tool for structural variations annotation. Bioinformatics, 2018, 34, 3572-3574. | 4.1 | 231 |
| 2 | Efficient strategy for the molecular diagnosis of intellectual disability using targeted high-throughput sequencing. Journal of Medical Genetics, 2014, 51, 724-736. | 3.2 | 229 |
| 3 | Exome sequencing of Bardetâ€“Biedl syndrome patient identifies a null mutation in the BBSome subunitBBIP1(BBS18). Journal of Medical Genetics, 2014, 51, 132-136. | 3.2 | 124 |
| 4 | Targeted high-throughput sequencing for diagnosis of genetically heterogeneous diseases: efficient mutation detection in Bardet-Biedl and AlstrÃ¶m Syndromes. Journal of Medical Genetics, 2012, 49, 502-512. | 3.2 | 104 |
| 5 | A targeted next-generation sequencing assay for the molecular diagnosis of genetic disorders with orodental involvement. Journal of Medical Genetics, 2016, 53, 98-110. | 3.2 | 100 |
| 6 | Homozygosity Mapping and Candidate Prioritization Identify Mutations, Missed by Whole-Exome Sequencing, in SMOC2, Causing Major Dental Developmental Defects. American Journal of Human Genetics, 2011, 89, 773-781. | 6.2 | 88 |
| 7 | VaRank: a simple and powerful tool for ranking genetic variants. PeerJ, 2015, 3, e796. | 2.0 | 80 |
| 8 | Identification of a novel mutation confirms the implication of IFT172 (BBS20) in Bardetâ€“Biedl syndrome. Journal of Human Genetics, 2016, 61, 447-450. | 2.3 | 64 |
| 9 | Mutations in TUBGCP4 Alter Microtubule Organization via the Î³-Tubulin Ring Complex in Autosomal-Recessive Microcephaly with Chorioretinopathy. American Journal of Human Genetics, 2015, 96, 666-674. | 6.2 | 60 |
| 10 | Rare De Novo Missense Variants in RNA Helicase DDX6 Cause Intellectual Disability and Dysmorphic Features and Lead to P-Body Defects and RNA Dysregulation. American Journal of Human Genetics, 2019, 105, 509-525. | 6.2 | 50 |
| 11 | Intragenic FMR1 disease-causing variants: a significant mutational mechanism leading to Fragile-X syndrome. European Journal of Human Genetics, 2017, 25, 423-431. | 2.8 | 48 |
| 12 | A mutation in VPS15 (PIK3R4) causes a ciliopathy and affects IFT20 release from the cis-Golgi. Nature Communications, 2016, 7, 13586. | 12.8 | 46 |
| 13 | Proteasome subunit PSMC3 variants cause neurosensory syndrome combining deafness and cataract due to proteotoxic stress. EMBO Molecular Medicine, 2020, 12, e11861. | 6.9 | 43 |
| 14 | Mutations in the latent TGF-beta binding protein 3 (LTBP3) gene cause brachyolmia with amelogenesis imperfecta. Human Molecular Genetics, 2015, 24, 3038-3049. | 2.9 | 40 |
| 15 | AnnotSV and knotAnnotSV: a web server for human structural variations annotations, ranking and analysis. Nucleic Acids Research, 2021, 49, W21-W28. | 14.5 | 38 |
| 16 | Identification and Characterization of Known Biallelic Mutations in the IFT27 (BBS19) Gene in a Novel Family With Bardet-Biedl Syndrome. Frontiers in Genetics, 2019, 10, 21. | 2.3 | 30 |
| 17 | Whole-genome sequencing in patients with ciliopathies uncovers a novel recurrent tandem duplication in IFT140. Human Mutation, 2018, 39, 983-992. | 2.5 | 21 |
| 18 | A <scp>BBS1 SVA</scp> F retrotransposon insertion is a frequent cause of <scp>Bardetâ€Biedl</scp> syndrome. Clinical Genetics, 2021, 99, 318-324. | 2.0 | 21 |

| # | ARTICLE | IF | CITATIONS |
|---|---|---|---|
| 19 | Mutations inKARScause a severe neurological and neurosensory disease with optic neuropathy. Human Mutation, 2019, 40, 1826-1840. | 2.5 | 15 |
| 20 | A New SLC10A7 Homozygous Missense Mutation Responsible for a Milder Phenotype of Skeletal Dysplasia With Amelogenesis Imperfecta. Frontiers in Genetics, 2019, 10, 504. | 2.3 | 11 |
| 21 | Detection of a Novel DSPP Mutation by NGS in a Population Isolate in Madagascar. Frontiers in Physiology, 2016, 7, 70. | 2.8 | 10 |
| 22 | Genetic Evidence Supporting the Role of the Calcium Channel, CACNA1S, in Tooth Cusp and Root Patterning. Frontiers in Physiology, 2018, 9, 1329. | 2.8 | 10 |
| 23 | NovelIQCEvariations confirm its role in postaxial polydactyly and cause ciliary defect phenotype in zebrafish. Human Mutation, 2020, 41, 240-254. | 2.5 | 5 |