VÃ©ronique Geoffroy

List of Publications by Year
in descending order

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	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
2	AnnotSV and knotAnnotSV: a web server for human structural variations annotations, ranking and analysis. Nucleic Acids Research, 2021, 49, W21-W28.	14.5	38
3	NovelIQCEvariations confirm its role in postaxial polydactyly and cause ciliary defect phenotype in zebrafish. Human Mutation, 2020, 41, 240-254.	2.5	5
4	Proteasome subunit PSMC3 variants cause neurosensory syndrome combining deafness and cataract due to proteotoxic stress. EMBO Molecular Medicine, 2020, 12, e11861.	6.9	43
5	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
6	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
7	Mutations inKARScause a severe neurological and neurosensory disease with optic neuropathy. Human Mutation, 2019, 40, 1826-1840.	2.5	15
8	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
9	AnnotSV: an integrated tool for structural variations annotation. Bioinformatics, 2018, 34, 3572-3574.	4.1	231
10	Whole-genome sequencing in patients with ciliopathies uncovers a novel recurrent tandem duplication in IFT140. Human Mutation, 2018, 39, 983-992.	2.5	21
11	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
12	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
13	Detection of a Novel DSPP Mutation by NGS in a Population Isolate in Madagascar. Frontiers in Physiology, 2016, 7, 70.	2.8	10
14	A mutation in VPS15 (PIK3R4) causes a ciliopathy and affects IFT20 release from the cis-Golgi. Nature Communications, 2016, 7, 13586.	12.8	46
15	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
16	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
17	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
18	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

#	ARTICLE	IF	CITATIONS
19	VaRank: a simple and powerful tool for ranking genetic variants. PeerJ, 2015, 3, e796.	2.0	80
20	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
21	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
22	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
23	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