Periklis Makrythanasis

List of Publications by Year
in descending order

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Version: 2024-02-01

30 papers	1,690 citations	394421 19 h-index	454955 30 g-index
30 all docs	30 docs citations	30 times ranked	5038 citing authors

#	Article	IF	Citations
1	Taurine treatment of retinal degeneration and cardiomyopathy in a consanguineous family with SLC6A6 taurine transporter deficiency. Human Molecular Genetics, 2020, 29, 618-623.	2.9	29
2	De Novo KAT5 Variants Cause a Syndrome with Recognizable Facial Dysmorphisms, Cerebellar Atrophy, Sleep Disturbance, and Epilepsy. American Journal of Human Genetics, 2020, 107, 564-574.	6.2	14
3	Bi-allelic Variants in IQSEC1 Cause Intellectual Disability, Developmental Delay, and Short Stature. American Journal of Human Genetics, 2019, 105, 907-920.	6.2	22
4	Bi-allelic Variants in DYNC1I2 Cause Syndromic Microcephaly with Intellectual Disability, Cerebral Malformations, and Dysmorphic Facial Features. American Journal of Human Genetics, 2019, 104, 1073-1087.	6.2	19
5	Biallelic variants in FBXL3 cause intellectual disability, delayed motor development and short stature. Human Molecular Genetics, 2019, 28, 972-979.	2.9	17
6	Biallelic variants in KIF14 cause intellectual disability with microcephaly. European Journal of Human Genetics, 2018, 26, 330-339.	2.8	52
7	Biallelic variants in LINGO1 are associated with autosomal recessive intellectual disability, microcephaly, speech and motor delay. Genetics in Medicine, 2018, 20, 778-784.	2.4	21
8	Bi-allelic Loss-of-Function Variants in DNMBP Cause Infantile Cataracts. American Journal of Human Genetics, 2018, 103, 568-578.	6.2	29
9	Visual impairment and progressive phthisis bulbi caused by recessive pathogenic variant in MARK3. Human Molecular Genetics, 2018, 27, 2703-2711.	2.9	21
10	Germline PMS2 and somatic POLE exonuclease mutations cause hypermutability of the leading DNA strand in biallelic mismatch repair deficiency syndrome brain tumours. Journal of Pathology, 2017, 243, 331-341.	4.5	12
11	Autosomal-Recessive Mutations in AP3B2, Adaptor-Related Protein Complex 3 Beta 2 Subunit, Cause an Early-Onset Epileptic Encephalopathy with Optic Atrophy. American Journal of Human Genetics, 2016, 99, 1368-1376.	6.2	46
12	Exome sequencing discloses KALRN homozygous variant as likely cause of intellectual disability and short stature in a consanguineous pedigree. Human Genomics, 2016, 10, 26.	2.9	13
13	Pathogenic Variants in PIGG Cause Intellectual Disability with Seizures and Hypotonia. American Journal of Human Genetics, 2016, 98, 615-626.	6.2	71
14	CATCHing putative causative variants in consanguineous families. BMC Bioinformatics, 2015, 16, 310.	2.6	12
15	Tissue-Specific Effects of Genetic and Epigenetic Variation on Gene Regulation and Splicing. PLoS Genetics, 2015, 11, e1004958.	3.5	185
16	Exome sequencing reveals pathogenic mutations in 91 strains of mice with Mendelian disorders. Genome Research, 2015, 25, 948-957.	5.5	54
17	DNA-Methylation Patterns in Trisomy 21 Using Cells from Monozygotic Twins. PLoS ONE, 2015, 10, e0135555.	2.5	47
18	BTNL2 gene SNPs as a contributing factor to sarcoidosis pathogenesis in a cohort of Greek patients. Meta Gene, 2014, 2, 619-630.	0.6	7

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19	Recessive thrombocytopenia likely due to a homozygous pathogenic variant in the FYBgene: case report. BMC Medical Genetics, 2014, 15, 135.	2.1	27
20	DEPDC5 mutations in families presenting as autosomal dominant nocturnal frontal lobe epilepsy. Neurology, 2014, 82, 2101-2106.	1.1	126
21	Loss of Function Mutation in the Palmitoyl-Transferase HHAT Leads to Syndromic 46,XY Disorder of Sex Development by Impeding Hedgehog Protein Palmitoylation and Signaling. PLoS Genetics, 2014, 10, e1004340.	3.5	63
22	Exome sequencing reveals a mutation in DMP1 in a family with familial sclerosing bone dysplasia. Bone, 2014, 68, 142-145.	2.9	15
23	Simultaneous identification and prioritization of variants in familial, de novo, and somatic genetic disorders with VariantMaster. Genome Research, 2014, 24, 349-355.	5.5	36
24	Diagnostic Exome Sequencing to Elucidate the Genetic Basis of Likely Recessive Disorders in Consanguineous Families. Human Mutation, 2014, 35, 1203-1210.	2.5	75
25	A Novel Homozygous Mutation in FGFR3 Causes Tall Stature, Severe Lateral Tibial Deviation, Scoliosis, Hearing Impairment, Camptodactyly, and Arachnodactyly. Human Mutation, 2014, 35, 959-963.	2.5	64
26	Mutations in ZMYND10, a Gene Essential for Proper Axonemal Assembly of Inner and Outer Dynein Arms in Humans and Flies, Cause Primary Ciliary Dyskinesia. American Journal of Human Genetics, 2013, 93, 346-356.	6.2	167
27	The complex SNP and CNV genetic architecture of the increased risk of congenital heart defects in Down syndrome. Genome Research, 2013, 23, 1410-1421.	5.5	65
28	Passive and active DNA methylation and the interplay with genetic variation in gene regulation. ELife, 2013, 2, e00523.	6.0	374
29	From sequence to functional understanding: the difficult road ahead. Genome Medicine, 2011, 3, 21.	8.2	1
30	Cystic Fibrosis Conductance Regulator, Tumor Necrosis Factor, Interferon Alpha-10, Interferon Alpha-17, and Interferon Gamma Genotyping as Potential Risk Markers in Pulmonary Sarcoidosis Pathogenesis in Greek Patients. Genetic Testing and Molecular Biomarkers, 2010, 14, 577-584.	0.7	6