John G Pappas

List of Publications by Year
in descending order

49 papers	1,586 citations	430874 18 h-index	345221 36 g-index
50 all docs	50 docs citations	50 times ranked	3401 citing authors

#	Article	IF	Citations
1	Heterozygous variants in CTR9, which encodes a major component of the PAF1 complex, are associated with a neurodevelopmental disorder. Genetics in Medicine, 2022, , .	2.4	1
2	A synonymous variant in MYO15A enriched in the Ashkenazi Jewish population causes autosomal recessive hearing loss due to abnormal splicing. European Journal of Human Genetics, 2021, 29, 988-997.	2.8	8
3	Severe epileptic encephalopathy associated with compound heterozygosity of <scp>THG1L</scp> variants in the Ashkenazi Jewish population. American Journal of Medical Genetics, Part A, 2021, 185, 1589-1597.	1.2	7
4	EIF3F-related neurodevelopmental disorder: refining the phenotypic and expanding the molecular spectrum. Orphanet Journal of Rare Diseases, 2021, 16, 136.	2.7	5
5	Rare deleterious mutations of HNRNP genes result in shared neurodevelopmental disorders. Genome Medicine, 2021, 13, 63.	8.2	50
6	<scp>PPP3CA</scp> truncating variants clustered in the regulatory domain cause earlyâ€onset refractory epilepsy. Clinical Genetics, 2021, 100, 227-233.	2.0	7
7	Expanding the phenotype of <scp>ASXL3</scp>â€related syndrome: A comprehensive description of 45 unpublished individuals with inherited and de novo pathogenic variants in <scp>ASXL3</scp>. American Journal of Medical Genetics, Part A, 2021, 185, 3446-3458.	1.2	12
8	CDK19-related disorder results from both loss-of-function and gain-of-function de novo missense variants. Genetics in Medicine, 2021, 23, 1050-1057.	2.4	7
9	De novo mutations in childhood cases of sudden unexplained death that disrupt intracellular Ca ²⁺ regulation. Proceedings of the National Academy of Sciences of the United States of America, 2021, 118, .	7.1	21
10	Clinical spectrum of individuals with pathogenic N F1 missense variants affecting p.Met1149, p.Arg1276, and p.Lys1423: genotypeâ€“phenotype study in neurofibromatosis type 1. Human Mutation, 2020, 41, 299-315.	2.5	80
11	Transcriptome sequencing identifies a noncoding, deep intronic variant in CLCN7 causing autosomal recessive osteopetrosis. Molecular Genetics & Genomic Medicine, 2020, 8, e1405.	1.2	7
12	De Novo Variants in the ATPase Module of MORC2 Cause a Neurodevelopmental Disorder with Growth Retardation and Variable Craniofacial Dysmorphism. American Journal of Human Genetics, 2020, 107, 352-363.	6.2	64
13	Germline AGO2 mutations impair RNA interference and human neurological development. Nature Communications, 2020, 11, 5797.	12.8	43
14	Genotypeâ€“phenotype correlation at codon 1740 of <scp>SETD2</scp>. American Journal of Medical Genetics, Part A, 2020, 182, 2037-2048.	1.2	14
15	HIST1H1E heterozygous proteinâ€truncating variants cause a recognizable syndrome with intellectual disability and distinctive facial gestalt: A study to clarify the HIST1H1E syndrome phenotype in 30 individuals. American Journal of Medical Genetics, Part A, 2019, 179, 2049-2055.	1.2	16
16	Hyponatremic Seizures and Adrenal Hypoplasia Congenita in a Neonate with Congenital Diaphragmatic Hernia. Case Reports in Pediatrics, 2019, 2019, 1-4.	0.4	1
17	Study of carrier frequency of Warsaw breakage syndrome in the Ashkenazi Jewish population and presentation of two cases. American Journal of Medical Genetics, Part A, 2019, 179, 2144-2151.	1.2	10
18	Recessive Mutations in AP1B1 Cause Ichthyosis, Deafness, and Photophobia. American Journal of Human Genetics, 2019, 105, 1023-1029.	6.2	21

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19	In vivo epigenetic editing of Sema6a promoter reverses transcallosal dysconnectivity caused by C11orf46/Arl14ep risk gene. Nature Communications, 2019, 10, 4112.	12.8	34
20	Variants in TCF20 in neurodevelopmental disability: description of 27 new patients and review of literature. Genetics in Medicine, 2019, 21, 2036-2042.	2.4	23
21	Kuforâ€Rakeb Syndrome Due to a Novel <scp>ATP</scp>13A2 Mutation in 2 Chineseâ€American Brothers. Movement Disorders Clinical Practice, 2018, 5, 92-95.	1.5	3
22	Genotype-phenotype correlations in individuals with pathogenicREREvariants. Human Mutation, 2018, 39, 666-675.	2.5	34
23	Genotype-Phenotype Correlation in NF1: Evidence for a More Severe Phenotype Associated with Missense Mutations Affecting NF1 Codons 844â€“848. American Journal of Human Genetics, 2018, 102, 69-87.	6.2	144
24	Intellectual disability due to monoallelic variant in GATAD2B and mosaicism in unaffected parent. American Journal of Medical Genetics, Part A, 2018, 176, 2907-2910.	1.2	8
25	A Syndromic Neurodevelopmental Disorder Caused by De Novo Variants in EBF3. American Journal of Human Genetics, 2017, 100, 128-137.	6.2	96
26	Lossâ€ofâ€function variants in NFIA provide further support that NFIA is a critical gene in 1p32â€p31 deletion syndrome: A four patient series. American Journal of Medical Genetics, Part A, 2017, 173, 3158-3164.	1.2	16
27	Clinical Phenotype in a Toddler with a Novel Heterozygous Mutation of the Vitamin D Receptor. Case Reports in Endocrinology, 2017, 2017, 1-4.	0.4	3
28	Unusual phenotype of congenital adrenal hyperplasia (CAH) with a novel mutation of the CYP21A2 gene. Journal of Pediatric Endocrinology and Metabolism, 2016, 29, 867-71.	0.9	5
29	Expansion of phenotype and genotypic data in CRB2-related syndrome. European Journal of Human Genetics, 2016, 24, 1436-1444.	2.8	36
30	Potockiâ€“Lupski syndrome in conjunction with bilateral clubfoot. Journal of Pediatric Orthopaedics Part B, 2015, 24, 373-376.	0.6	6
31	Hereditary Predisposition to Primary CNS Tumors. Molecular Pathology Library, 2015, , 1-22.	0.1	0
32	The Clinical Course of an Overgrowth Syndrome, From Diagnosis in Infancy Through Adulthood: The Case of Beckwithâ€“Wiedemann Syndrome. Current Problems in Pediatric and Adolescent Health Care, 2015, 45, 112-117.	1.7	18
33	Mutations inSLC1A4, encoding the brain serine transporter, are associated with developmental delay, microcephaly and hypomyelination. Journal of Medical Genetics, 2015, 52, 541-547.	3.2	68
34	De novo mutations in KIF1A cause progressive encephalopathy and brain atrophy. Annals of Clinical and Translational Neurology, 2015, 2, 623-635.	3.7	96
35	Loss-of-function HDAC8 mutations cause a phenotypic spectrum of Cornelia de Lange syndrome-like features, ocular hypertelorism, large fontanelle and X-linked inheritance. Human Molecular Genetics, 2014, 23, 2888-2900.	2.9	120
36	Three cases of isolated terminal deletion of chromosome 8p without heart defects presenting with a mild phenotype. American Journal of Medical Genetics, Part A, 2013, 161, 822-828.	1.2	20

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37	Complex Chromosome Rearrangement of 6p25.3->p23 and 12q24.32->qter in a Child With Moyamoya. Pediatrics, 2013, 131, e1996-e2001.	2.1	9
38	Early presentation of bilateral gonadoblastomas in a Denys-Drash syndrome patient: a cautionary tale for prophylactic gonadectomy. Journal of Pediatric Endocrinology and Metabolism, 2013, 26, 971-4.	0.9	16
39	Well-differentiated Pancreatic Neuroendocrine Carcinoma in Tuberous Sclerosisâ€"Case Report and Review of the Literature. American Journal of Surgical Pathology, 2012, 36, 149-153.	3.7	49
40	Congenital absence of the superior oblique tendon in Noonan-neurofibromatosis syndrome. Journal of AAPOS, 2011, 15, 593-594.	0.3	0
41	UPD detection using homozygosity profiling with a SNP genotyping microarray. , 2011, 155, 757-768.		112
42	Early Presentation of Bilateral Gonadoblastoma in Denys-Drash Syndrome: A Cautionary Tale for Prophylactic Gonadectomy. , 2011, , P3-64-P3-64.		0
43	Successful pregnancy outcome in Ehlersâ€"Danlos syndrome, vascular type. Journal of Maternal-Fetal and Neonatal Medicine, 2009, 22, 924-927.	1.5	21
44	Effect of Growth Hormone Therapy on Severe Short Stature and Skeletal Deformities in a Patient with Combined Turner Syndrome and Langer Mesomelic Dysplasia. Journal of Clinical Endocrinology and Metabolism, 2009, 94, 5028-5033.	3.6	5
45	Greig Cephalopolysyndactyly Syndrome. Journal of Ultrasound in Medicine, 2009, 28, 1735-1742.	1.7	4
46	A molecular and clinical study of Larsen syndrome caused by mutations in FLNB. Journal of Medical Genetics, 2006, 44, 89-98.	3.2	102
47	Multifocal renal oncocytoma in a patient with Von Hippel-Lindau mutation. Urology, 2005, 66, 1320.e11-1320.e12.	1.0	7
48	DAX1 Mutations Map to Putative Structural Domains in a Deduced Three-Dimensional Model. American Journal of Human Genetics, 1998, 62, 855-864.	6.2	91
49	Variants of Î±1-Antitrypsin in Puerto Rican Children With Asthma. Chest, 1993, 103, 812-815.	0.8	66