

John G Pappas

List of Publications by Year in descending order

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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	Genotype-Phenotype Correlation in NF1: Evidence for a More Severe Phenotype Associated with Missense Mutations Affecting NF1 Codons 844-848. <i>American Journal of Human Genetics</i> , 2018, 102, 69-87.	6.2	144
2	Loss-of-function HDAC8 mutations cause a phenotypic spectrum of Cornelia de Lange syndrome-like features, ocular hypertelorism, large fontanelle and X-linked inheritance. <i>Human Molecular Genetics</i> , 2014, 23, 2888-2900.	2.9	120
3	UPD detection using homozygosity profiling with a SNP genotyping microarray. , 2011, 155, 757-768.		112
4	A molecular and clinical study of Larsen syndrome caused by mutations in FLNB. <i>Journal of Medical Genetics</i> , 2006, 44, 89-98.	3.2	102
5	De novo mutations in KIF1A cause progressive encephalopathy and brain atrophy. <i>Annals of Clinical and Translational Neurology</i> , 2015, 2, 623-635.	3.7	96
6	A Syndromic Neurodevelopmental Disorder Caused by De Novo Variants in EBF3. <i>American Journal of Human Genetics</i> , 2017, 100, 128-137.	6.2	96
7	DAX1 Mutations Map to Putative Structural Domains in a Deduced Three-Dimensional Model. <i>American Journal of Human Genetics</i> , 1998, 62, 855-864.	6.2	91
8	Clinical spectrum of individuals with pathogenic <i>NF1</i> missense variants affecting p.Met1149, p.Arg1276, and p.Lys1423: genotype-phenotype study in neurofibromatosis type 1. <i>Human Mutation</i> , 2020, 41, 299-315.	2.5	80
9	Mutations in <i>SLC1A4</i> , encoding the brain serine transporter, are associated with developmental delay, microcephaly and hypomyelination. <i>Journal of Medical Genetics</i> , 2015, 52, 541-547.	3.2	68
10	Variants of α -1-Antitrypsin in Puerto Rican Children With Asthma. <i>Chest</i> , 1993, 103, 812-815.	0.8	66
11	De Novo Variants in the ATPase Module of MORC2 Cause a Neurodevelopmental Disorder with Growth Retardation and Variable Craniofacial Dysmorphism. <i>American Journal of Human Genetics</i> , 2020, 107, 352-363.	6.2	64
12	Rare deleterious mutations of HNRNP genes result in shared neurodevelopmental disorders. <i>Genome Medicine</i> , 2021, 13, 63.	8.2	50
13	Well-differentiated Pancreatic Neuroendocrine Carcinoma in Tuberous Sclerosis-Case Report and Review of the Literature. <i>American Journal of Surgical Pathology</i> , 2012, 36, 149-153.	3.7	49
14	Germline AGO2 mutations impair RNA interference and human neurological development. <i>Nature Communications</i> , 2020, 11, 5797.	12.8	43
15	Expansion of phenotype and genotypic data in CRB2-related syndrome. <i>European Journal of Human Genetics</i> , 2016, 24, 1436-1444.	2.8	36
16	Genotype-phenotype correlations in individuals with pathogenic <i>RERE</i> variants. <i>Human Mutation</i> , 2018, 39, 666-675.	2.5	34
17	In vivo epigenetic editing of Sema6a promoter reverses transcallosal dysconnectivity caused by C11orf46/Arl14ep risk gene. <i>Nature Communications</i> , 2019, 10, 4112.	12.8	34
18	Variants in TCF20 in neurodevelopmental disability: description of 27 new patients and review of literature. <i>Genetics in Medicine</i> , 2019, 21, 2036-2042.	2.4	23

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19	Successful pregnancy outcome in Ehlers-Danlos syndrome, vascular type. <i>Journal of Maternal-Fetal and Neonatal Medicine</i> , 2009, 22, 924-927.	1.5	21
20	Recessive Mutations in AP1B1 Cause Ichthyosis, Deafness, and Photophobia. <i>American Journal of Human Genetics</i> , 2019, 105, 1023-1029.	6.2	21
21	De novo mutations in childhood cases of sudden unexplained death that disrupt intracellular Ca ²⁺ regulation. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2021, 118, .	7.1	21
22	Three cases of isolated terminal deletion of chromosome 8p without heart defects presenting with a mild phenotype. <i>American Journal of Medical Genetics, Part A</i> , 2013, 161, 822-828.	1.2	20
23	The Clinical Course of an Overgrowth Syndrome, From Diagnosis in Infancy Through Adulthood: The Case of Beckwith-Wiedemann Syndrome. <i>Current Problems in Pediatric and Adolescent Health Care</i> , 2015, 45, 112-117.	1.7	18
24	Early presentation of bilateral gonadoblastomas in a Denys-Drash syndrome patient: a cautionary tale for prophylactic gonadectomy. <i>Journal of Pediatric Endocrinology and Metabolism</i> , 2013, 26, 971-4.	0.9	16
25	Loss-of-function variants in <i>NFIA</i> provide further support that <i>NFIA</i> is a critical gene in 1p32-p31 deletion syndrome: A four patient series. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 3158-3164.	1.2	16
26	<i>HIST1H1E</i> heterozygous protein-truncating variants cause a recognizable syndrome with intellectual disability and distinctive facial gestalt: A study to clarify the <i>HIST1H1E</i> syndrome phenotype in 30 individuals. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 2049-2055.	1.2	16
27	Genotype-phenotype correlation at codon 1740 of <i>SETD2</i> . <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 2037-2048.	1.2	14
28	Expanding the phenotype of <i>ASXL3</i> -related syndrome: A comprehensive description of 45 unpublished individuals with inherited and de novo pathogenic variants in <i>ASXL3</i> . <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 3446-3458.	1.2	12
29	Study of carrier frequency of Warsaw breakage syndrome in the Ashkenazi Jewish population and presentation of two cases. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 2144-2151.	1.2	10
30	Complex Chromosome Rearrangement of 6p25.3->p23 and 12q24.32->qter in a Child With Moyamoya. <i>Pediatrics</i> , 2013, 131, e1996-e2001.	2.1	9
31	Intellectual disability due to monoallelic variant in <i>GATAD2B</i> and mosaicism in unaffected parent. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 2907-2910.	1.2	8
32	A synonymous variant in MYO15A enriched in the Ashkenazi Jewish population causes autosomal recessive hearing loss due to abnormal splicing. <i>European Journal of Human Genetics</i> , 2021, 29, 988-997.	2.8	8
33	Multifocal renal oncocytoma in a patient with Von Hippel-Lindau mutation. <i>Urology</i> , 2005, 66, 1320.e11-1320.e12.	1.0	7
34	Transcriptome sequencing identifies a noncoding, deep intronic variant in <i>CLCN7</i> causing autosomal recessive osteopetrosis. <i>Molecular Genetics & Genomic Medicine</i> , 2020, 8, e1405.	1.2	7
35	Severe epileptic encephalopathy associated with compound heterozygosity of <i>THG1L</i> variants in the Ashkenazi Jewish population. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 1589-1597.	1.2	7
36	<i>PPP3CA</i> truncating variants clustered in the regulatory domain cause early-onset refractory epilepsy. <i>Clinical Genetics</i> , 2021, 100, 227-233.	2.0	7

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37	CDK19-related disorder results from both loss-of-function and gain-of-function de novo missense variants. <i>Genetics in Medicine</i> , 2021, 23, 1050-1057.	2.4	7
38	Potocki-Lupski syndrome in conjunction with bilateral clubfoot. <i>Journal of Pediatric Orthopaedics Part B</i> , 2015, 24, 373-376.	0.6	6
39	Effect of Growth Hormone Therapy on Severe Short Stature and Skeletal Deformities in a Patient with Combined Turner Syndrome and Langer Mesomelic Dysplasia. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2009, 94, 5028-5033.	3.6	5
40	Unusual phenotype of congenital adrenal hyperplasia (CAH) with a novel mutation of the CYP21A2 gene. <i>Journal of Pediatric Endocrinology and Metabolism</i> , 2016, 29, 867-71.	0.9	5
41	EIF3F-related neurodevelopmental disorder: refining the phenotypic and expanding the molecular spectrum. <i>Orphanet Journal of Rare Diseases</i> , 2021, 16, 136.	2.7	5
42	Greig Cephalopolysyndactyly Syndrome. <i>Journal of Ultrasound in Medicine</i> , 2009, 28, 1735-1742.	1.7	4
43	Clinical Phenotype in a Toddler with a Novel Heterozygous Mutation of the Vitamin D Receptor. <i>Case Reports in Endocrinology</i> , 2017, 2017, 1-4.	0.4	3
44	Kufor-Rakeb Syndrome Due to a Novel <i>ATP13A2</i> Mutation in 2 Chinese-American Brothers. <i>Movement Disorders Clinical Practice</i> , 2018, 5, 92-95.	1.5	3
45	Hyponatremic Seizures and Adrenal Hypoplasia Congenita in a Neonate with Congenital Diaphragmatic Hernia. <i>Case Reports in Pediatrics</i> , 2019, 2019, 1-4.	0.4	1
46	Heterozygous variants in <i>CTR9</i> , which encodes a major component of the PAF1 complex, are associated with a neurodevelopmental disorder. <i>Genetics in Medicine</i> , 2022, , .	2.4	1
47	Congenital absence of the superior oblique tendon in Noonan-neurofibromatosis syndrome. <i>Journal of AAPOS</i> , 2011, 15, 593-594.	0.3	0
48	Hereditary Predisposition to Primary CNS Tumors. <i>Molecular Pathology Library</i> , 2015, , 1-22.	0.1	0
49	Early Presentation of Bilateral Gonadoblastoma in Denys-Drash Syndrome: A Cautionary Tale for Prophylactic Gonadectomy. , 2011, , P3-64-P3-64.		0