Michele Iacomino

List of Publications by Year
in descending order

52	2,220	471509 17	265206 42
papers	citations	h-index	g-index

55	55	55	4886
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Expanding Phenotype of Poirierâ€“Bienvenu Syndrome: New Evidence from an Italian Multicentrical Cohort of Patients. Genes, 2022, 13, 276.	2.4	10
2	Loss of Neuron Navigator 2 Impairs Brain and Cerebellar Development. Cerebellum, 2022, , 1.	2.5	5
3	De novo GRIN2A variants associated with epilepsy and autism and literature review. Epilepsy and Behavior, 2022, 129, 108604.	1.7	9
4	A Phenotypic-Driven Approach for the Diagnosis of WOREE Syndrome. Frontiers in Pediatrics, 2022, 10, 847549.	1.9	3
5	Epilepsy Course and Developmental Trajectories in STXBP1 -DEE. Neurology: Genetics, 2022, 8, .	1.9	24
6	De novo truncating NOVA2 variants affect alternative splicing and lead to heterogeneous neurodevelopmental phenotypes. Human Mutation, 2022, 43, 1299-1313.	2.5	6
7	Complex Neurological Phenotype Associated with a De Novo DHDDS Mutation in a Boy with Intellectual Disability, Refractory Epilepsy, and Movement Disorder. Journal of Pediatric Genetics, 2021, 10, 236-238.	0.7	10
8	Musculoskeletal Features without Ataxia Associated with a Novel de novo Mutation in KCNA1 Impairing the Voltage Sensitivity of Kv1.1 Channel. Biomedicines, 2021, 9, 75.	3.2	5
9	Atypical Presentation of Aromatic L-Amino Acid Decarboxylase Deficiency with Developmental Epileptic Encephalopathy. Journal of Pediatric Epilepsy, 2021, 10, 124-127.	0.2	4
10	Biallelic Variants in KIF17 Associated with Microphthalmia and Coloboma Spectrum. International Journal of Molecular Sciences, 2021, 22, 4471.	4.1	8
11	Genotype-Phenotype Correlations in Neurofibromatosis Type 1: A Single-Center Cohort Study. Cancers, 2021, 13, 1879.	3.7	21
12	Temporalâ€parietalâ€occipital epilepsy in GEFS+ associated with SCN1A mutation. Epileptic Disorders, 2021, 23, 397-401.	1.3	2
13	Biallelic variants in HPDL cause pure and complicated hereditary spastic paraplegia. Brain, 2021, 144, 1422-1434.	7.6	22
14	Shared genetic basis between genetic generalized epilepsy and background electroencephalographic oscillations. Epilepsia, 2021, 62, 1518-1527.	5.1	5
15	Sub-genic intolerance, ClinVar, and the epilepsies: A whole-exome sequencing study of 29,165 individuals. American Journal of Human Genetics, 2021, 108, 965-982.	6.2	35
16	Clinical and Genetic Features in Patients With Reflex Bathing Epilepsy. Neurology, 2021, 97, e577-e586.	1.1	11
17	Neuromuscular and Neuroendocrinological Features Associated With ZC4H2-Related Arthrogryposis Multiplex Congenita in a Sicilian Family: A Case Report. Frontiers in Neurology, 2021, 12, 704747.	2.4	6
18	Mapping the human genetic architecture of COVID-19. Nature, 2021, 600, 472-477.	27.8	640

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19	Cortical and Subcortical Network Dysfunction in a Female Patient With NEXMIF Encephalopathy. Frontiers in Neurology, 2021, 12, 722664.	2.4	3
20	Using common genetic variants to find drugs for common epilepsies. Brain Communications, 2021, 3, fcab287.	3.3	9
21	New Trends and Most Promising Therapeutic Strategies for Epilepsy Treatment. Frontiers in Neurology, 2021, 12, 753753.	2.4	23
22	Homozygous STXBP1 variant causes encephalopathy and gain-of-function in synaptic transmission. Brain, 2020, 143, 441-451.	7.6	46
23	Brain Organoids as Model Systems for Genetic Neurodevelopmental Disorders. Frontiers in Cell and Developmental Biology, 2020, 8, 590119.	3.7	31
24	Epilepsy subtype-specific copy number burden observed in a genome-wide study of 17â€‰458 subjects. Brain, 2020, 143, 2106-2118.	7.6	47
25	Targeted re-sequencing in malformations of cortical development: genotype-phenotype correlations. Seizure: the Journal of the British Epilepsy Association, 2020, 80, 145-152.	2.0	13
26	Emerging treatments for progressive myoclonus epilepsies. Expert Review of Neurotherapeutics, 2020, 20, 341-350.	2.8	11
27	Loss of Wwox Perturbs Neuronal Migration and Impairs Early Cortical Development. Frontiers in Neuroscience, 2020, 14, 644.	2.8	22
28	Clinical spectrum and genotype-phenotype correlations in PRRT2 Italian patients. European Journal of Paediatric Neurology, 2020, 28, 193-197.	1.6	14
29	Distal motor neuropathy associated with novel EMILIN1 mutation. Neurobiology of Disease, 2020, 137, 104757.	4.4	6
30	Chiari malformation type I: what information from the genetics?. Child's Nervous System, 2019, 35, 1665-1671.	1.1	12
31	Ultra-Rare Genetic Variation in the Epilepsies: A Whole-Exome Sequencing Study of 17,606 Individuals. American Journal of Human Genetics, 2019, 105, 267-282.	6.2	237
32	AMPA receptor GluA2 subunit defects are a cause of neurodevelopmental disorders. Nature Communications, 2019, 10, 3094.	12.8	150
33	Polygenic burden in focal and generalized epilepsies. Brain, 2019, 142, 3473-3481.	7.6	90
34	Assessing the genetic association between vitamin B6 metabolism and genetic generalized epilepsy. Molecular Genetics and Metabolism Reports, 2019, 21, 100518.	1.1	2
35	Intronic ATTTC repeat expansions in STARD7 in familial adult myoclonic epilepsy linked to chromosome 2. Nature Communications, 2019, 10, 4920.	12.8	99
36	No evidence for a BRD 2 promoter hypermethylation inÂblood leukocytes of Europeans with juvenile myoclonicÂepilepsy. Epilepsia, 2019, 60, e31-e36.	5.1	4

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37	Novel mutation in sarcotubular myopathy. Acta Myologica, 2019, 38, 8-12.	1.5	13
38	Spinal motor neuron involvement in a patient with homozygous PRUNE mutation. European Journal of Paediatric Neurology, 2018, 22, 541-543.	1.6	10
39	Clinical and molecular consequences of exon 78 deletion in DMD gene. Journal of Human Genetics, 2018, 63, 761-764.	2.3	7
40	Inflammatory myopathy in a patient with collagen VI mutations. Scandinavian Journal of Rheumatology, 2018, 47, 166-167.	1.1	0
41	Genome-wide mega-analysis identifies 16 loci and highlights diverse biological mechanisms in the common epilepsies. Nature Communications, 2018, 9, 5269.	12.8	331
42	Rare coding variants in genes encoding GABAA receptors in genetic generalised epilepsies: an exome-based case-control study. Lancet Neurology, The, 2018, 17, 699-708.	10.2	67
43	Novel AMPD2 mutation in pontocerebellar hypoplasia, dysmorphisms, and teeth abnormalities. Neurology: Genetics, 2017, 3, e179.	1.9	22
44	Alterations in the Î±₂Î´ ligand, thrombospondinâ€1, in a rat model of spontaneous absence epilepsy and in patients with idiopathic/genetic generalized epilepsies. Epilepsia, 2017, 58, 1993-2001.	5.1	8
45	Rare GABRA3 variants are associated with epileptic seizures, encephalopathy and dysmorphic features. Brain, 2017, 140, 2879-2894.	7.6	33
46	White matter involvement in a family with a novel PDGFB mutation. Neurology: Genetics, 2016, 2, e77.	1.9	19
47	Exome Sequencing Fails to Identify the Genetic Cause of Aicardi Syndrome. Molecular Syndromology, 2016, 7, 234-238.	0.8	16
48	Novel GABRG2 mutations cause familial febrile seizures. Neurology: Genetics, 2015, 1, e35.	1.9	29
49	Use of a Lower Dosage Liver-Detargeted AAV Vector to Prevent Hamster Muscular Dystrophy. Human Gene Therapy, 2013, 24, 424-430.	2.7	7
50	O.17 Mutation spectrum of limb-girdle muscular dystrophies by New Generation Sequencing approaches. Neuromuscular Disorders, 2013, 23, 849-850.	0.6	0
51	D.O.3 Next generation sequencing applications are ready for genetic diagnosis of muscular dystrophies. Neuromuscular Disorders, 2012, 22, 806.	0.6	0
52	Enhancer Chip: Detecting Human Copy Number Variations in Regulatory Elements. PLoS ONE, 2012, 7, e52264.	2.5	8