Reza Maroofian

List of Publications by Year
in descending order

121 papers	2,825 citations	257450 24 h-index	276875 41 g-index
129 all docs	129 docs citations	129 times ranked	5581 citing authors

#	Article	IF	Citations
1	Biallelic variants in SLC38A3 encoding a glutamine transporter cause epileptic encephalopathy. Brain, 2022, 145, 909-924.	7.6	17
2	Unraveling the genetic complexities of combined retinal dystrophy and hearing impairment. Human Genetics, 2022, 141, 785-803.	3.8	6
3	High diagnostic rate of trio exome sequencing in consanguineous families with neurogenetic diseases. Brain, 2022, 145, 1507-1518.	7.6	14
4	Variable skeletal phenotypes associated with biallelic variants in PRKG2. Journal of Medical Genetics, 2022, 59, 947-950.	3.2	6
5	Heterozygous <scp>EIF2AK2</scp> Variant Causes Adolescenceâ€Onset Generalized Dystonia Partially Responsive to <scp>DBS</scp>. Movement Disorders Clinical Practice, 2022, 9, 268-271.	1.5	7
6	Biallelic Lossâ€ofâ€Function NDUFA12 Variants Cause a Wide Phenotypic Spectrum from Leigh/Leighâ€Like Syndrome to Isolated Optic Atrophy. Movement Disorders Clinical Practice, 2022, 9, 218-228.	1.5	5
7	Clinicoâ€radiological features, molecular spectrum, and identification of prognostic factors in developmental and epileptic encephalopathy due to inosine triphosphate pyrophosphatase (ITPase) deficiency. Human Mutation, 2022, 43, 403-419.	2.5	9
8	Bi-allelic variants inCHKAcause a neurodevelopmental disorder with epilepsy and microcephaly. Brain, 2022, 145, 1916-1923.	7.6	3
9	<scp>Elâ€Hattabâ€Alkuraya</scp> syndrome caused by biallelic <scp>WDR45B</scp> pathogenic variants: Further delineation of the phenotype and genotype. Clinical Genetics, 2022, 101, 530-540.	2.0	7
10	Biallelic ADAM22 pathogenic variants cause progressive encephalopathy and infantile-onset refractory epilepsy. Brain, 2022, 145, 2301-2312.	7.6	8
11	Response to: Phenotypic heterogeneity of Leigh syndrome due to NDUFA12 variants is multicausal. Human Mutation, 2022, 43, 99-100.	2.5	0
12	TMEM63C mutations cause mitochondrial morphology defects and underlie hereditary spastic paraplegia. Brain, 2022, 145, 3095-3107.	7.6	17
13	Biallelic Variants in the Ectonucleotidase <scp>ENTPD1</scp> Cause a Complex Neurodevelopmental Disorder with Intellectual Disability, Distinct White Matter Abnormalities, and Spastic Paraplegia. Annals of Neurology, 2022, 92, 304-321.	5.3	2
14	Biallelic <scp> KITLG </scp> variants lead to a distinct spectrum of hypomelanosis and sensorineural hearing loss. Journal of the European Academy of Dermatology and Venereology, 2022, , .	2.4	1
15	Biallelic variants in <scp>ZNF142</scp> lead to a syndromic neurodevelopmental disorder. Clinical Genetics, 2022, 102, 98-109.	2.0	6
16	Inhibition of G-protein signalling in cardiac dysfunction of intellectual developmental disorder with cardiac arrhythmia (IDDCA) syndrome. Journal of Medical Genetics, 2021, 58, 815-831.	3.2	3
17	Biallelic variants in ADARB1, encoding a dsRNA-specific adenosine deaminase, cause a severe developmental and epileptic encephalopathy. Journal of Medical Genetics, 2021, 58, 495-504.	3.2	14
18	PIGH deficiency can be associated with severe neurodevelopmental and skeletal manifestations. Clinical Genetics, 2021, 99, 313-317.	2.0	7

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19	Biallelic loss-of-function variants in NEMF cause central nervous system impairment and axonal polyneuropathy. Human Genetics, 2021, 140, 579-592.	3.8	14
20	Autosomal recessive cardiomyopathy and sudden cardiac death associated with variants in MYL3. Genetics in Medicine, 2021, 23, 787-792.	2.4	16
21	Biallelic variants in HPDL, encoding 4-hydroxyphenylpyruvate dioxygenase-like protein, lead to an infantile neurodegenerative condition. Genetics in Medicine, 2021, 23, 524-533.	2.4	17
22	Novel variants broaden the phenotypic spectrum of PLEKHG5 â€associated neuropathies. European Journal of Neurology, 2021, 28, 1344-1355.	3.3	4
23	Pontocerebellar hypoplasia due to bi-allelic variants in MINPP1. European Journal of Human Genetics, 2021, 29, 411-421.	2.8	13
24	A relatively common homozygous TRAPPC4 splicing variant is associated with an early-infantile neurodegenerative syndrome. European Journal of Human Genetics, 2021, 29, 271-279.	2.8	8
25	Homozygous missense WIPI2 variants cause a congenital disorder of autophagy with neurodevelopmental impairments of variable clinical severity and disease course. Brain Communications, 2021, 3, fcab183.	3.3	10
26	Defective phosphatidylethanolamine biosynthesis leads to a broad ataxia-spasticity spectrum. Brain, 2021, 144, e30-e30.	7.6	12
27	Expanding the phenotype of PIGSâ€associated early onset epileptic developmental encephalopathy. Epilepsia, 2021, 62, e35-e41.	5.1	11
28	An ancestral 10-bp repeat expansion in VWA1 causes recessive hereditary motor neuropathy. Brain, 2021, 144, 584-600.	7.6	20
29	SCUBE3 loss-of-function causes a recognizable recessive developmental disorder due to defective bone morphogenetic protein signaling. American Journal of Human Genetics, 2021, 108, 115-133.	6.2	37
30	<scp>MED27</scp> Variants Cause Developmental Delay, Dystonia, and Cerebellar Hypoplasia. Annals of Neurology, 2021, 89, 828-833.	5.3	14
31	Bi-allelic variants in HOPS complex subunit VPS41 cause cerebellar ataxia and abnormal membrane trafficking. Brain, 2021, 144, 769-780.	7.6	33
32	Novel NDUFA12 variants are associated with isolated complex I defect and variable clinical manifestation. Human Mutation, 2021, 42, 699-710.	2.5	12
33	Homozygous SCN1B variants causing early infantile epileptic encephalopathy 52 affect voltageâ€gated sodium channel function. Epilepsia, 2021, 62, e82-e87.	5.1	9
34	Mitochondrial <scp>D</scp><scp>NA</scp> Analysis from Exome Sequencing Data Improves Diagnostic Yield in Neurological Diseases. Annals of Neurology, 2021, 89, 1240-1247.	5.3	12
35	Two novel biâ€allelic <scp>KDELR2</scp> missense variants cause osteogenesis imperfecta with neurodevelopmental features. American Journal of Medical Genetics, Part A, 2021, 185, 2241-2249.	1.2	7
36	Clinical, neuroimaging, and molecular spectrum of TECPR2 â€associated hereditary sensory and autonomic neuropathy with intellectual disability. Human Mutation, 2021, 42, 762-776.	2.5	18

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37	A form of muscular dystrophy associated with pathogenic variants in JAG2. American Journal of Human Genetics, 2021, 108, 840-856.	6.2	15
38	Biallelic and monoallelic variants in PLXNA1 are implicated in a novel neurodevelopmental disorder with variable cerebral and eye anomalies. Genetics in Medicine, 2021, 23, 1715-1725.	2.4	22
39	Biallelic variants in HPDL cause pure and complicated hereditary spastic paraplegia. Brain, 2021, 144, 1422-1434.	7.6	22
40	Loss of function mutations in GEMIN5 cause a neurodevelopmental disorder. Nature Communications, 2021, 12, 2558.	12.8	28
41	Bi-allelic variants in IPO8 cause a connective tissue disorder associated with cardiovascular defects, skeletal abnormalities, and immune dysregulation. American Journal of Human Genetics, 2021, 108, 1126-1137.	6.2	14
42	A human importin-Î²-related disorder: Syndromic thoracic aortic aneurysm caused by bi-allelic loss-of-function variants in IPO8. American Journal of Human Genetics, 2021, 108, 1115-1125.	6.2	10
43	Pathogenic variants in PIDD1 lead to an autosomal recessive neurodevelopmental disorder with pachygyria and psychiatric features. European Journal of Human Genetics, 2021, 29, 1226-1234.	2.8	8
44	Bi-allelic loss-of-function variants in BCAS3 cause a syndromic neurodevelopmental disorder. American Journal of Human Genetics, 2021, 108, 1069-1082.	6.2	8
45	Bi-allelic premature truncating variants in LTBP1 cause cutis laxa syndrome. American Journal of Human Genetics, 2021, 108, 1095-1114.	6.2	19
46	Biallelic variants in KARS1 are associated with neurodevelopmental disorders and hearing loss recapitulated by the knockout zebrafish. Genetics in Medicine, 2021, 23, 1933-1943.	2.4	11
47	A Novel Homozygous <scp>ADCY5</scp> Variant is Associated with a Neurodevelopmental Disorder and Movement Abnormalities. Movement Disorders Clinical Practice, 2021, 8, 1140-1143.	1.5	3
48	Expanding the mutational landscape and clinical phenotype of the YIF1B related brain disorder. Brain, 2021, 144, e85-e85.	7.6	2
49	Delineating the molecular and phenotypic spectrum of the SETD1B-related syndrome. Genetics in Medicine, 2021, 23, 2122-2137.	2.4	16
50	A biallelic variant in CLRN2 causes non-syndromic hearing loss in humans. Human Genetics, 2021, 140, 915-931.	3.8	16
51	Bi-allelic variants in SPATA5L1 lead to intellectual disability, spastic-dystonic cerebral palsy, epilepsy, and hearing loss. American Journal of Human Genetics, 2021, 108, 2006-2016.	6.2	11
52	Early-onset phenotype of bi-allelic GRN mutations. Brain, 2021, 144, e22-e22.	7.6	5
53	Bi-allelic variants in OGDHL cause a neurodevelopmental spectrum disease featuring epilepsy, hearing loss, visual impairment, and ataxia. American Journal of Human Genetics, 2021, 108, 2368-2384.	6.2	12
54	Genetic and phenotypic characterization of NKX6â€2 â€related spastic ataxia and hypomyelination. European Journal of Neurology, 2020, 27, 334-342.	3.3	16

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55	Loss of UGP2 in brain leads to a severe epileptic encephalopathy, emphasizing that bi-allelic isoform-specific start-loss mutations of essential genes can cause genetic diseases. Acta Neuropathologica, 2020, 139, 415-442.	7.7	38
56	SVEP1 as a Genetic Modifier of TEK-Related Primary Congenital Glaucoma. , 2020, 61, 6.		25
57	Pathogenic Variants in the Myosin Chaperone UNC-45B Cause Progressive Myopathy with Eccentric Cores. American Journal of Human Genetics, 2020, 107, 1078-1095.	6.2	24
58	Early-infantile onset epilepsy and developmental delay caused by bi-allelic GAD1 variants. Brain, 2020, 143, 2388-2397.	7.6	28
59	De Novo and Bi-allelic Pathogenic Variants in NARS1 Cause Neurodevelopmental Delay Due to Toxic Gain-of-Function and Partial Loss-of-Function Effects. American Journal of Human Genetics, 2020, 107, 311-324.	6.2	32
60	Biallelic MADD variants cause a phenotypic spectrum ranging from developmental delay to a multisystem disorder. Brain, 2020, 143, 2437-2453.	7.6	21
61	Clinical and molecular description of 19 patients with GATAD2B-Associated Neurodevelopmental Disorder (GAND). European Journal of Medical Genetics, 2020, 63, 104004.	1.3	7
62	NEMF mutations that impair ribosome-associated quality control are associated with neuromuscular disease. Nature Communications, 2020, 11, 4625.	12.8	47
63	Expanding the clinical and genetic spectrum of PCYT2-related disorders. Brain, 2020, 143, e76-e76.	7.6	14
64	A homozygous GDAP2 loss-of-function variant in a patient with adult-onset cerebellar ataxia. Brain, 2020, 143, e49-e49.	7.6	5
65	Hereditary polyneuropathy with optic atrophy due to PDXK variant leading to impaired Vitamin B6 metabolism. Neuromuscular Disorders, 2020, 30, 583-589.	0.6	7
66	NR1H4â€related Progressive Familial Intrahepatic Cholestasis 5. Journal of Pediatric Gastroenterology and Nutrition, 2020, 70, e111-e113.	1.8	11
67	Biallelic MFSD2A variants associated with congenital microcephaly, developmental delay, and recognizable neuroimaging features. European Journal of Human Genetics, 2020, 28, 1509-1519.	2.8	21
68	Bi-allelic JAM2 Variants Lead to Early-Onset Recessive Primary Familial Brain Calcification. American Journal of Human Genetics, 2020, 106, 412-421.	6.2	47
69	Cell-based analysis of CAD variants identifies individuals likely to benefit from uridine therapy. Genetics in Medicine, 2020, 22, 1598-1605.	2.4	18
70	Loss of TNR causes a nonprogressive neurodevelopmental disorder with spasticity and transient opisthotonus. Genetics in Medicine, 2020, 22, 1061-1068.	2.4	14
71	Bi-allelic ADARB1 Variants Associated with Microcephaly, Intellectual Disability, and Seizures. American Journal of Human Genetics, 2020, 106, 467-483.	6.2	31
72	Bi-allelic Variants in the GPI Transamidase Subunit PIGK Cause a Neurodevelopmental Syndrome with Hypotonia, Cerebellar Atrophy, and Epilepsy. American Journal of Human Genetics, 2020, 106, 484-495.	6.2	22

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73	RSRC1 loss-of-function variants cause mild to moderate autosomal recessive intellectual disability. Brain, 2020, 143, e31-e31.	7.6	6
74	Bi-allelic GAD1 variants cause a neonatal onset syndromic developmental and epileptic encephalopathy. Brain, 2020, 143, 1447-1461.	7.6	18
75	KDM5A mutations identified in autism spectrum disorder using forward genetics. ELife, 2020, 9, .	6.0	27
76	Biallelic loss of function variants in PPP1R21 cause a neurodevelopmental syndrome with impaired endocytic function. Human Mutation, 2019, 40, 267-280.	2.5	15
77	AMPA receptor GluA2 subunit defects are a cause of neurodevelopmental disorders. Nature Communications, 2019, 10, 3094.	12.8	150
78	Homozygous Missense Variants in NTNG2, Encoding a Presynaptic Netrin-G2 Adhesion Protein, Lead to a Distinct Neurodevelopmental Disorder. American Journal of Human Genetics, 2019, 105, 1048-1056.	6.2	30
79	Bi-allelic variants in RNF170 are associated with hereditary spastic paraplegia. Nature Communications, 2019, 10, 4790.	12.8	39
80	Biallelic mutations in neurofascin cause neurodevelopmental impairment and peripheral demyelination. Brain, 2019, 142, 2948-2964.	7.6	43
81	Bi-allelic Loss of Human APC2, Encoding Adenomatous Polyposis Coli Protein 2, Leads to Lissencephaly, Subcortical Heterotopia, and Global Developmental Delay. American Journal of Human Genetics, 2019, 105, 844-853.	6.2	17
82	Growth disrupting mutations in epigenetic regulatory molecules are associated with abnormalities of epigenetic aging. Genome Research, 2019, 29, 1057-1066.	5.5	38
83	De novo and biallelic DEAF1 variants cause a phenotypic spectrum. Genetics in Medicine, 2019, 21, 2059-2069.	2.4	20
84	A combined immunodeficiency with severe infections, inflammation, and allergy caused by ARPC1B deficiency. Journal of Allergy and Clinical Immunology, 2019, 143, 2296-2299.	2.9	87
85	Homozygous Mutations in CSF1R Cause a Pediatric-Onset Leukoencephalopathy and Can Result in Congenital Absence of Microglia. American Journal of Human Genetics, 2019, 104, 936-947.	6.2	157
86	Cardiomyopathy with lethal arrhythmias associated with inactivation of KLHL24. Human Molecular Genetics, 2019, 28, 1919-1929.	2.9	35
87	Further supporting evidence for REEP1 phenotypic and allelic heterogeneity. Neurology: Genetics, 2019, 5, e379.	1.9	3
88	TMX2 Is a Crucial Regulator of Cellular Redox State, and Its Dysfunction Causes Severe Brain Developmental Abnormalities. American Journal of Human Genetics, 2019, 105, 1126-1147.	6.2	25
89	Homozygous Null TBX4 Mutations Lead to Posterior Amelia with Pelvic and Pulmonary Hypoplasia. American Journal of Human Genetics, 2019, 105, 1294-1301.	6.2	17
90	MAB21L1 loss of function causes a syndromic neurodevelopmental disorder with distinctivecerebellar,ocular, craniofacial andgenital features (COFG) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50 57	3.2	15

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91	Loss of the sphingolipid desaturase DEGS1 causes hypomyelinating leukodystrophy. Journal of Clinical Investigation, 2019, 129, 1240-1256.	8.2	68
92	Dual Diagnosis of Ellis-van Creveld Syndrome and Hearing Loss in a Consanguineous Family. Molecular Syndromology, 2018, 9, 5-14.	0.8	12
93	Biallelic variants in KIF14 cause intellectual disability with microcephaly. European Journal of Human Genetics, 2018, 26, 330-339.	2.8	52
94	Potential role of gender specific effect of leptin receptor deficiency in an extended consanguineous family with severe early-onset obesity. European Journal of Medical Genetics, 2018, 61, 465-467.	1.3	15
95	A Genotype-First Approach for Clinical and Genetic Evaluation of Wolcott-Rallison Syndrome in a Large Cohort of Iranian Children With Neonatal Diabetes. Canadian Journal of Diabetes, 2018, 42, 272-275.	0.8	19
96	Neurologic Phenotypes Associated With Mutations in RTN4IP1 (OPA10) in Children and Young Adults. JAMA Neurology, 2018, 75, 105.	9.0	26
97	Novel Homozygous Missense Mutation in RYR1 Leads to Severe Congenital Ptosis, Ophthalmoplegia, and Scoliosis in the Absence of Myopathy. Molecular Syndromology, 2018, 9, 25-29.	0.8	3
98	Expanding the clinical phenotype of IARS2-related mitochondrial disease. BMC Medical Genetics, 2018, 19, 196.	2.1	16
99	Parental Whole-Exome Sequencing Enables Sialidosis Type II Diagnosis due to an NEU1 Missense Mutation as an Underlying Cause of Nephrotic Syndrome in the Child. Kidney International Reports, 2018, 3, 1454-1463.	0.8	11
100	MPZL2 is a novel gene associated with autosomal recessive nonsyndromic moderate hearing loss. Human Genetics, 2018, 137, 479-486.	3.8	19
101	The conserved p.Arg108 residue in S1PR2 (DFNB68) is fundamental for proper hearing: evidence from a consanguineous Iranian family. BMC Medical Genetics, 2018, 19, 81.	2.1	10
102	SLC10A7 mutations cause a skeletal dysplasia with amelogenesis imperfecta mediated by GAG biosynthesis defects. Nature Communications, 2018, 9, 3087.	12.8	39
103	Biallelic Mutations in ADPRHL2, Encoding ADP-Ribosylhydrolase 3, Lead to a Degenerative Pediatric Stress-Induced Epileptic Ataxia Syndrome. American Journal of Human Genetics, 2018, 103, 431-439.	6.2	62
104	Clinical presentation and natural history of infantile-onset ascending spastic paralysis from three families with an ALS2 founder variant. Neurological Sciences, 2018, 39, 1917-1925.	1.9	18
105	Targeted sequencing with expanded gene profile enables high diagnostic yield in non-5q-spinal muscular atrophies. Human Mutation, 2018, 39, 1284-1298.	2.5	42
106	Mutations in INPP5K Cause a Form of Congenital Muscular Dystrophy Overlapping Marinesco-SjÃ¶gren Syndrome and Dystroglycanopathy. American Journal of Human Genetics, 2017, 100, 537-545.	6.2	67
107	Genetic, Phenotypic, and Interferon Biomarker Status in ADAR1-Related Neurological Disease. Neuropediatrics, 2017, 48, 166-184.	0.6	62
108	Novel EYA1 variants causing Branchio-oto-renal syndrome. International Journal of Pediatric Otorhinolaryngology, 2017, 98, 59-63.	1.0	16

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109	Biallelic MCM3AP mutations cause Charcot-Marie-Tooth neuropathy with variable clinical presentation. Brain, 2017, 140, e65-e65.	7.6	13
110	A Novel Loss-of-Function Mutation in HOXB1 Associated with Autosomal Recessive Hereditary Congenital Facial Palsy in a Large Iranian Family. Molecular Syndromology, 2017, 8, 261-265.	0.8	5
111	A homozygous loss-of-function mutation in PTPN14 causes a syndrome of bilateral choanal atresia and early infantile-onset lymphedema. Meta Gene, 2017, 14, 53-58.	0.6	6
112	Digenic inheritance of mutations in the cardiac troponin (TNNT2) and cardiac beta myosin heavy chain (MYH7) as the cause of severe dilated cardiomyopathy. European Journal of Medical Genetics, 2017, 60, 485-488.	1.3	23
113	PRUNE is crucial for normal brain development and mutated in microcephaly with neurodevelopmental impairment. Brain, 2017, 140, 940-952.	7.6	62
114	B3GALNT2 mutations associated with non-syndromic autosomal recessive intellectual disability reveal a lack of genotypeâ€“phenotype associations in the muscular dystrophy-dystroglycanopathies. Genome Medicine, 2017, 9, 118.	8.2	13
115	Genetic screening of Congenital Short Bowel Syndrome patients confirms CLMP as the major gene involved in the recessive form of this disorder. European Journal of Human Genetics, 2016, 24, 1627-1629.	2.8	18
116	Spectrum of DNA variants for non-syndromic deafness in a large cohort from multiple continents. Human Genetics, 2016, 135, 953-961.	3.8	102
117	A Novel Mutation in the OFD1 Gene in a Family with Oral-Facial-Digital Syndrome Type 1: A Case Report. Iranian Journal of Public Health, 2016, 45, 1359-1366.	0.5	2
118	Recessive nephrocerebellar syndrome on the Galloway-Mowat syndrome spectrum is caused by homozygous protein-truncating mutations ofWDR73. Brain, 2015, 138, 2173-2190.	7.6	60
119	Mutations in KPTN Cause Macrocephaly, Neurodevelopmental Delay, and Seizures. American Journal of Human Genetics, 2014, 94, 87-94.	6.2	35
120	Mutations in B4GALNT1 (GM2 synthase) underlie a new disorder of ganglioside biosynthesis. Brain, 2013, 136, 3618-3624.	7.6	115
121	Biallelic loss of <scp> EMC10 </scp> leads to mild to severe intellectual disability. Annals of Clinical and Translational Neurology, 0, , .	3.7	1