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	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
2	AMPA receptor GluA2 subunit defects are a cause of neurodevelopmental disorders. Nature Communications, 2019, 10, 3094.	12.8	150
3	Mutations in B4GALNT1 (GM2 synthase) underlie a new disorder of ganglioside biosynthesis. Brain, 2013, 136, 3618-3624.	7.6	115
4	Spectrum of DNA variants for non-syndromic deafness in a large cohort from multiple continents. Human Genetics, 2016, 135, 953-961.	3.8	102
5	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
6	Loss of the sphingolipid desaturase DEGS1 causes hypomyelinating leukodystrophy. Journal of Clinical Investigation, 2019, 129, 1240-1256.	8.2	68
7	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
8	Genetic, Phenotypic, and Interferon Biomarker Status in ADAR1-Related Neurological Disease. Neuropediatrics, 2017, 48, 166-184.	0.6	62
9	PRUNE is crucial for normal brain development and mutated in microcephaly with neurodevelopmental impairment. Brain, 2017, 140, 940-952.	7.6	62
10	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
11	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
12	Biallelic variants in KIF14 cause intellectual disability with microcephaly. European Journal of Human Genetics, 2018, 26, 330-339.	2.8	52
13	NEMF mutations that impair ribosome-associated quality control are associated with neuromuscular disease. Nature Communications, 2020, 11, 4625.	12.8	47
14	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
15	Biallelic mutations in neurofascin cause neurodevelopmental impairment and peripheral demyelination. Brain, 2019, 142, 2948-2964.	7.6	43
16	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
17	SLC10A7 mutations cause a skeletal dysplasia with amelogenesis imperfecta mediated by GAG biosynthesis defects. Nature Communications, 2018, 9, 3087.	12.8	39
18	Bi-allelic variants in RNF170 are associated with hereditary spastic paraplegia. Nature Communications, 2019, 10, 4790.	12.8	39

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19	Growth disrupting mutations in epigenetic regulatory molecules are associated with abnormalities of epigenetic aging. Genome Research, 2019, 29, 1057-1066.	5.5	38
20	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
21	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
22	Mutations in KPTN Cause Macrocephaly, Neurodevelopmental Delay, and Seizures. American Journal of Human Genetics, 2014, 94, 87-94.	6.2	35
23	Cardiomyopathy with lethal arrhythmias associated with inactivation of KLHL24. Human Molecular Genetics, 2019, 28, 1919-1929.	2.9	35
24	Bi-allelic variants in HOPS complex subunit VPS41 cause cerebellar ataxia and abnormal membrane trafficking. Brain, 2021, 144, 769-780.	7.6	33
25	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
26	Bi-allelic ADARB1 Variants Associated with Microcephaly, Intellectual Disability, and Seizures. American Journal of Human Genetics, 2020, 106, 467-483.	6.2	31
27	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
28	Early-infantile onset epilepsy and developmental delay caused by bi-allelic GAD1 variants. Brain, 2020, 143, 2388-2397.	7.6	28
29	Loss of function mutations in GEMIN5 cause a neurodevelopmental disorder. Nature Communications, 2021, 12, 2558.	12.8	28
30	KDM5A mutations identified in autism spectrum disorder using forward genetics. ELife, 2020, 9, .	6.0	27
31	Neurologic Phenotypes Associated With Mutations in RTN4IP1 (OPA10) in Children and Young Adults. JAMA Neurology, 2018, 75, 105.	9.0	26
32	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
33	MAB21L1 loss of function causes a syndromic neurodevelopmental disorder with distinctivecerebellar,ocular, craniofacial andgenital features (COFG) Tj ETQq1 1 0.784314 rgBT /Overlock 10	11.4	25
34	SVEP1 as a Genetic Modifier of TEK-Related Primary Congenital Glaucoma. , 2020, 61, 6.		25
35	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
36	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

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37	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
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	Biallelic MADD variants cause a phenotypic spectrum ranging from developmental delay to a multisystem disorder. Brain, 2020, 143, 2437-2453.	7.6	21
41	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
42	De novo and biallelic DEAF1 variants cause a phenotypic spectrum. Genetics in Medicine, 2019, 21, 2059-2069.	2.4	20
43	An ancestral 10-bp repeat expansion in VWA1 causes recessive hereditary motor neuropathy. Brain, 2021, 144, 584-600.	7.6	20
44	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
45	MPZL2 is a novel gene associated with autosomal recessive nonsyndromic moderate hearing loss. Human Genetics, 2018, 137, 479-486.	3.8	19
46	Bi-allelic premature truncating variants in LTBP1 cause cutis laxa syndrome. American Journal of Human Genetics, 2021, 108, 1095-1114.	6.2	19
47	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
48	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
49	Cell-based analysis of CAD variants identifies individuals likely to benefit from uridine therapy. Genetics in Medicine, 2020, 22, 1598-1605.	2.4	18
50	Bi-allelic GAD1 variants cause a neonatal onset syndromic developmental and epileptic encephalopathy. Brain, 2020, 143, 1447-1461.	7.6	18
51	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
52	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
53	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
54	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

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55	Biallelic variants in SLC38A3 encoding a glutamine transporter cause epileptic encephalopathy. Brain, 2022, 145, 909-924.	7.6	17
56	TMEM63C mutations cause mitochondrial morphology defects and underlie hereditary spastic paraplegia. Brain, 2022, 145, 3095-3107.	7.6	17
57	Novel EYA1 variants causing Branchio-oto-renal syndrome. International Journal of Pediatric Otorhinolaryngology, 2017, 98, 59-63.	1.0	16
58	Expanding the clinical phenotype of IARS2-related mitochondrial disease. BMC Medical Genetics, 2018, 19, 196.	2.1	16
59	Genetic and phenotypic characterization of NKX6â€2 â€related spastic ataxia and hypomyelination. European Journal of Neurology, 2020, 27, 334-342.	3.3	16
60	Autosomal recessive cardiomyopathy and sudden cardiac death associated with variants in MYL3. Genetics in Medicine, 2021, 23, 787-792.	2.4	16
61	Delineating the molecular and phenotypic spectrum of the SETD1B-related syndrome. Genetics in Medicine, 2021, 23, 2122-2137.	2.4	16
62	A biallelic variant in CLRN2 causes non-syndromic hearing loss in humans. Human Genetics, 2021, 140, 915-931.	3.8	16
63	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
64	Biallelic loss of function variants in PPP1R21 cause a neurodevelopmental syndrome with impaired endocytic function. Human Mutation, 2019, 40, 267-280.	2.5	15
65	A form of muscular dystrophy associated with pathogenic variants in JAG2. American Journal of Human Genetics, 2021, 108, 840-856.	6.2	15
66	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
67	Expanding the clinical and genetic spectrum of PCYT2-related disorders. Brain, 2020, 143, e76-e76.	7.6	14
68	Loss of TNR causes a nonprogressive neurodevelopmental disorder with spasticity and transient opisthotonus. Genetics in Medicine, 2020, 22, 1061-1068.	2.4	14
69	Biallelic loss-of-function variants in NEMF cause central nervous system impairment and axonal polyneuropathy. Human Genetics, 2021, 140, 579-592.	3.8	14
70	<scp>MED27</scp> Variants Cause Developmental Delay, Dystonia, and Cerebellar Hypoplasia. Annals of Neurology, 2021, 89, 828-833.	5.3	14
71	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
72	High diagnostic rate of trio exome sequencing in consanguineous families with neurogenetic diseases. Brain, 2022, 145, 1507-1518.	7.6	14

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73	Biallelic MCM3AP mutations cause Charcot-Marie-Tooth neuropathy with variable clinical presentation. Brain, 2017, 140, e65-e65.	7.6	13
74	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
75	Pontocerebellar hypoplasia due to bi-allelic variants in MINPP1. European Journal of Human Genetics, 2021, 29, 411-421.	2.8	13
76	Dual Diagnosis of Ellis-van Creveld Syndrome and Hearing Loss in a Consanguineous Family. Molecular Syndromology, 2018, 9, 5-14.	0.8	12
77	Defective phosphatidylethanolamine biosynthesis leads to a broad ataxia-spasticity spectrum. Brain, 2021, 144, e30-e30.	7.6	12
78	Novel NDUFA12 variants are associated with isolated complex I defect and variable clinical manifestation. Human Mutation, 2021, 42, 699-710.	2.5	12
79	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
80	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
81	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
82	NR1H4â€related Progressive Familial Intrahepatic Cholestasis 5. Journal of Pediatric Gastroenterology and Nutrition, 2020, 70, e111-e113.	1.8	11
83	Expanding the phenotype of PIGSâ€associated early onset epileptic developmental encephalopathy. Epilepsia, 2021, 62, e35-e41.	5.1	11
84	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
85	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
86	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
87	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
88	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
89	Homozygous SCN1B variants causing early infantile epileptic encephalopathy 52 affect voltageâ€gated sodium channel function. Epilepsia, 2021, 62, e82-e87.	5.1	9
90	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

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91	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
92	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
93	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
94	Biallelic ADAM22 pathogenic variants cause progressive encephalopathy and infantile-onset refractory epilepsy. Brain, 2022, 145, 2301-2312.	7.6	8
95	Clinical and molecular description of 19 patients with GATAD2B-Associated Neurodevelopmental Disorder (GAND). European Journal of Medical Genetics, 2020, 63, 104004.	1.3	7
96	Hereditary polyneuropathy with optic atrophy due to PDXK variant leading to impaired Vitamin B6 metabolism. Neuromuscular Disorders, 2020, 30, 583-589.	0.6	7
97	PIGH deficiency can be associated with severe neurodevelopmental and skeletal manifestations. Clinical Genetics, 2021, 99, 313-317.	2.0	7
98	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
99	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
100	<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
101	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
102	RSRC1 loss-of-function variants cause mild to moderate autosomal recessive intellectual disability. Brain, 2020, 143, e31-e31.	7.6	6
103	Unraveling the genetic complexities of combined retinal dystrophy and hearing impairment. Human Genetics, 2022, 141, 785-803.	3.8	6
104	Variable skeletal phenotypes associated with biallelic variants in PRKG2. Journal of Medical Genetics, 2022, 59, 947-950.	3.2	6
105	Biallelic variants in <scp>ZNF142</scp> lead to a syndromic neurodevelopmental disorder. Clinical Genetics, 2022, 102, 98-109.	2.0	6
106	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
107	A homozygous GDAP2 loss-of-function variant in a patient with adult-onset cerebellar ataxia. Brain, 2020, 143, e49-e49.	7.6	5
108	Early-onset phenotype of bi-allelic GRN mutations. Brain, 2021, 144, e22-e22.	7.6	5

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109	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
110	Novel variants broaden the phenotypic spectrum of PLEKHG5 â€associated neuropathies. European Journal of Neurology, 2021, 28, 1344-1355.	3.3	4
111	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
112	Further supporting evidence for REEP1 phenotypic and allelic heterogeneity. Neurology: Genetics, 2019, 5, e379.	1.9	3
113	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
114	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
115	Bi-allelic variants inCHKAcause a neurodevelopmental disorder with epilepsy and microcephaly. Brain, 2022, 145, 1916-1923.	7.6	3
116	Expanding the mutational landscape and clinical phenotype of the YIF1B related brain disorder. Brain, 2021, 144, e85-e85.	7.6	2
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	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
119	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
120	Biallelic loss of <scp> EMC10 </scp> leads to mild to severe intellectual disability. Annals of Clinical and Translational Neurology, 0, , .	3.7	1
121	Response to: Phenotypic heterogeneity of Leigh syndrome due to NDUFA12 variants is multicausal. Human Mutation, 2022, 43, 99-100.	2.5	0