Mahshid S Azamian

List of Publications by Year
in descending order

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70 papers	2,300 citations	304743 22 h-index	254184 43 g-index
76 all docs	76 docs citations	76 times ranked	5256 citing authors

#	Article	IF	Citations
1	Clinical application of a scale to assess genomic healthcare empowerment (GEmS): Process and illustrative case examples. Journal of Genetic Counseling, 2022, 31, 59-70.	1.6	3
2	Genetic counselor roles in the undiagnosed diseases network research study: Clinical care, collaboration, and curation. Journal of Genetic Counseling, 2022, 31, 326-337.	1.6	1
3	Delineation of a novel neurodevelopmental syndrome associated with PAX5 haploinsufficiency. Human Mutation, 2022, 43, 461-470.	2.5	5
4	Loss of IRF2BPL impairs neuronal maintenance through excess Wnt signaling. Science Advances, 2022, 8, eabl5613.	10.3	12
5	<scp>LMOD2</scp>â€related dilated cardiomyopathy presenting in late infancy. American Journal of Medical Genetics, Part A, 2022, 188, 1858-1862.	1.2	5
6	The microRNA processorDROSHAis a candidate gene for a severe progressive neurological disorder. Human Molecular Genetics, 2022, 31, 2934-2950.	2.9	6
7	Cardiac Crises: Cardiac Arrhythmias and Cardiomyopathy during TANGO2-deficiency related Metabolic Crises. Heart Rhythm, 2022, , .	0.7	13
8	An autosomal dominant neurological disorder caused by de novo variants in FAR1 resulting in uncontrolled synthesis of ether lipids. Genetics in Medicine, 2021, 23, 740-750.	2.4	25
9	Family genetic result communication in rare and undiagnosed disease communities: Understanding the practice. Journal of Genetic Counseling, 2021, 30, 439-447.	1.6	4
10	Clinical sites of the Undiagnosed Diseases Network: unique contributions to genomic medicine and science. Genetics in Medicine, 2021, 23, 259-271.	2.4	18
11	Commonalities across computational workflows for uncovering explanatory variants in undiagnosed cases. Genetics in Medicine, 2021, 23, 1075-1085.	2.4	16
12	<scp>Healthâ€related</scp> quality of life in adults with osteogenesis imperfecta. Clinical Genetics, 2021, 99, 772-779.	2.0	4
13	OTUD5 Variants Associated With X-Linked Intellectual Disability and Congenital Malformation. Frontiers in Cell and Developmental Biology, 2021, 9, 631428.	3.7	4
14	Variants in PRKAR1B cause a neurodevelopmental disorder with autism spectrum disorder, apraxia, and insensitivity to pain. Genetics in Medicine, 2021, 23, 1465-1473.	2.4	10
15	Missense and truncating variants in CHD5 in a dominant neurodevelopmental disorder with intellectual disability, behavioral disturbances, and epilepsy. Human Genetics, 2021, 140, 1109-1120.	3.8	18
16	Progressive cerebellar atrophy in a patient with complex II and III deficiency and a novel deleterious variant in SDHA: A Counseling Conundrum. Molecular Genetics & Genomic Medicine, 2021, 9, e1692.	1.2	1
17	Detection of a mosaic CDKL5 deletion and inversion by optical genome mapping ends an exhaustive diagnostic odyssey. Molecular Genetics & Genomic Medicine, 2021, 9, e1665.	1.2	11
18	A rare description of pure partial trisomy of 16q12.2q24.3 and review of the literature. American Journal of Medical Genetics, Part A, 2021, 185, 2903-2912.	1.2	2

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19	â€œDoctors can read about it, they can know about it, but they've never lived with itâ€: How parents use social media throughout the diagnostic odyssey. Journal of Genetic Counseling, 2021, 30, 1707-1718.	1.6	10
20	Heterozygous loss-of-function variants significantly expand the phenotypes associated with loss of GDF11. Genetics in Medicine, 2021, 23, 1889-1900.	2.4	13
21	Phenotypic expansion of CACNA1C-associated disorders to include isolated neurological manifestations. Genetics in Medicine, 2021, 23, 1922-1932.	2.4	16
22	<scp>PPP3CA</scp> truncating variants clustered in the regulatory domain cause earlyâ€onset refractory epilepsy. Clinical Genetics, 2021, 100, 227-233.	2.0	7
23	One is the loneliest number: genotypic matchmaking using the electronic health record. Genetics in Medicine, 2021, 23, 1830-1832.	2.4	6
24	Vertical transmission of a large calvarial ossification defect due to heterozygous variants of ALX4 and TWIST1. American Journal of Medical Genetics, Part A, 2021, 185, 916-922.	1.2	1
25	Partial Loss of USP9X Function Leads to a Male Neurodevelopmental and Behavioral Disorder Converging on Transforming Growth Factor Î² Signaling. Biological Psychiatry, 2020, 87, 100-112.	1.3	42
26	A novel CACNA1A variant in a child with early stroke and intractable epilepsy. Molecular Genetics & Genomic Medicine, 2020, 8, e1383.	1.2	11
27	Variants in SCAF4 Cause a Neurodevelopmental Disorder and Are Associated with Impaired mRNA Processing. American Journal of Human Genetics, 2020, 107, 544-554.	6.2	13
28	Biallelic MADD variants cause a phenotypic spectrum ranging from developmental delay to a multisystem disorder. Brain, 2020, 143, 2437-2453.	7.6	21
29	DYRK1A pathogenic variants in two patients with syndromic intellectual disability and a review of the literature. Molecular Genetics & Genomic Medicine, 2020, 8, e1544.	1.2	8
30	Recessive ACO2 variants as a cause of isolated ophthalmologic phenotypes. American Journal of Medical Genetics, Part A, 2020, 182, 1960-1966.	1.2	8
31	De novo EIF2AK1 and EIF2AK2 Variants Are Associated with Developmental Delay, Leukoencephalopathy, and Neurologic Decompensation. American Journal of Human Genetics, 2020, 106, 570-583.	6.2	37
32	<scp>Wolffâ€“Parkinsonâ€“White</scp> syndrome: De novo variants and evidence for mutational burden in genes associated with atrial fibrillation. American Journal of Medical Genetics, Part A, 2020, 182, 1387-1399.	1.2	14
33	Early infantile epileptic encephalopathy due to biallelic pathogenic variants in <scp>PIGQ</scp>: Report of seven new subjects and review of the literature. Journal of Inherited Metabolic Disease, 2020, 43, 1321-1332.	3.6	15
34	A comprehensive iterative approach is highly effective in diagnosing individuals who are exome negative. Genetics in Medicine, 2019, 21, 161-172.	2.4	60
35	De Novo Variants in WDR37 Are Associated with Epilepsy, Colobomas, Dysmorphism, Developmental Delay, Intellectual Disability, and Cerebellar Hypoplasia. American Journal of Human Genetics, 2019, 105, 413-424.	6.2	43
36	Magnetic Resonance Imaging characteristics in case of TOR1AIP1 muscular dystrophy. Clinical Imaging, 2019, 58, 108-113.	1.5	6

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37	Loss of CLTRN function produces a neuropsychiatric disorder and a biochemical phenotype that mimics Hartnup disease. American Journal of Medical Genetics, Part A, 2019, 179, 2459-2468.	1.2	14
38	De Novo Pathogenic Variants in N-cadherin Cause a Syndromic Neurodevelopmental Disorder with Corpus Callosum, Axon, Cardiac, Ocular, and Genital Defects. American Journal of Human Genetics, 2019, 105, 854-868.	6.2	29
39	Disruptive variants of CSDE1 associate with autism and interfere with neuronal development and synaptic transmission. Science Advances, 2019, 5, eaax2166.	10.3	35
40	Lysosomal Storage and Albinism Due to Effects of a De Novo CLCN7 Variant on Lysosomal Acidification. American Journal of Human Genetics, 2019, 104, 1127-1138.	6.2	59
41	Identification of rare-disease genes using blood transcriptome sequencing and large control cohorts. Nature Medicine, 2019, 25, 911-919.	30.7	221
42	Heterozygous variants in MYBPC1 are associated with an expanded neuromuscular phenotype beyond arthrogryposis. Human Mutation, 2019, 40, 1115-1126.	2.5	19
43	Aberrant DNA methylation as a diagnostic biomarker of diabetic embryopathy. Genetics in Medicine, 2019, 21, 2453-2461.	2.4	8
44	IgG4â€related disease: Association with a rare gene variant expressed in cytotoxic T cells. Molecular Genetics & Genomic Medicine, 2019, 7, e686.	1.2	8
45	Whole genome sequencing reveals novel IGHMBP2 variant leading to unique cryptic spliceâ€site and Charcotâ€Marieâ€Tooth phenotype with early onset symptoms. Molecular Genetics & Genomic Medicine, 2019, 7, e00676.	1.2	18
46	Exome sequencing reveals novel variants and unique allelic spectrum for hearing impairment in Filipino cochlear implantees. Clinical Genetics, 2019, 95, 634-636.	2.0	9
47	Bi-allelic Variants in TONSL Cause SPONASTRIME Dysplasia and a Spectrum of Skeletal Dysplasia Phenotypes. American Journal of Human Genetics, 2019, 104, 422-438.	6.2	27
48	Expanding the Spectrum of BAF-Related Disorders: De Novo Variants in SMARCC2 Cause a Syndrome with Intellectual Disability and Developmental Delay. American Journal of Human Genetics, 2019, 104, 164-178.	6.2	59
49	Genetic architecture of laterality defects revealed by whole exome sequencing. European Journal of Human Genetics, 2019, 27, 563-573.	2.8	44
50	Biallelic Mutations in ATP5F1D, which Encodes a Subunit of ATP Synthase, Cause a Metabolic Disorder. American Journal of Human Genetics, 2018, 102, 494-504.	6.2	59
51	A randomized trial to study the comparative efficacy of phenylbutyrate and benzoate on nitrogen excretion and ureagenesis in healthy volunteers. Genetics in Medicine, 2018, 20, 708-716.	2.4	8
52	Looking beyond the exome: a phenotype-first approach to molecular diagnostic resolution in rare and undiagnosed diseases. Genetics in Medicine, 2018, 20, 464-469.	2.4	42
53	Copy Number Variants of Undetermined Significance Are Not Associated with Worse Clinical Outcomes in Hypoplastic Left Heart Syndrome. Journal of Pediatrics, 2018, 202, 206-211.e2.	1.8	3
54	IRF2BPL Is Associated with Neurological Phenotypes. American Journal of Human Genetics, 2018, 103, 245-260.	6.2	69

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55	Congenital heart defects and left ventricular non-compaction in males with loss-of-function variants inNONO. Journal of Medical Genetics, 2017, 54, 47-53.	3.2	24
56	MARRVEL: Integration of Human and Model Organism Genetic Resources to Facilitate Functional Annotation of the Human Genome. American Journal of Human Genetics, 2017, 100, 843-853.	6.2	181
57	An exome sequencing study of Moebius syndrome including atypical cases reveals an individual with CFEOM3A and a TUBB3 mutation. Journal of Physical Education and Sports Management, 2017, 3, a000984.	1.2	18
58	Plasma Glutamine Is a Minor Precursor for the Synthesis of Citrulline: A Multispecies Study. Journal of Nutrition, 2017, 147, 549-555.	2.9	16
59	Use of Exome Sequencing for Infants in Intensive Care Units. JAMA Pediatrics, 2017, 171, e173438.	6.2	348
60	Assessment of large copy number variants in patients with apparently isolated congenital leftâ€sided cardiac lesions reveals clinically relevant genomic events. American Journal of Medical Genetics, Part A, 2017, 173, 2176-2188.	1.2	17
61	Xp11.22 deletions encompassing CENPVL1, CENPVL2, MAGED1 and GSPT2 as a cause of syndromic X-linked intellectual disability. PLoS ONE, 2017, 12, e0175962.	2.5	14
62	Whole exome sequencing in 342 congenital cardiac left sided lesion cases reveals extensive genetic heterogeneity and complex inheritance patterns. Genome Medicine, 2017, 9, 95.	8.2	37
63	Bi-allelic Mutations in PKD1L1 Are Associated with Laterality Defects in Humans. American Journal of Human Genetics, 2016, 99, 886-893.	6.2	57
64	Recurrent Muscle Weakness with Rhabdomyolysis, Metabolic Crises, and Cardiac Arrhythmia Due to Bi-allelic TANGO2 Mutations. American Journal of Human Genetics, 2016, 98, 347-357.	6.2	98
65	A genome-wide association study of congenital cardiovascular left-sided lesions shows association with a locus on chromosome 20. Human Molecular Genetics, 2016, 25, 2331-2341.	2.9	31
66	Cytogenomic Aberrations in Congenital Cardiovascular Malformations. Molecular Syndromology, 2016, 7, 51-61.	0.8	7
67	De Novo GMNN Mutations Cause Autosomal-Dominant Primordial Dwarfism Associated with Meier-Gorlin Syndrome. American Journal of Human Genetics, 2015, 97, 904-913.	6.2	65
68	Mutations in PURA Cause Profound Neonatal Hypotonia, Seizures, and Encephalopathy in 5q31.3 Microdeletion Syndrome. American Journal of Human Genetics, 2014, 95, 579-583.	6.2	92
69	TM4SF20 Ancestral Deletion and Susceptibility to a Pediatric Disorder of Early Language Delay and Cerebral White Matter Hyperintensities. American Journal of Human Genetics, 2013, 93, 197-210.	6.2	43
70	Quality of life, illness perceptions, and parental lived experiences in TANGO2-related metabolic encephalopathy and arrhythmias. European Journal of Human Genetics, 0, , .	2.8	1