

Mahshid S Azamian

List of Publications by Year in descending order

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Version: 2024-02-01

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	Use of Exome Sequencing for Infants in Intensive Care Units. <i>JAMA Pediatrics</i> , 2017, 171, e173438.	6.2	348
2	Identification of rare-disease genes using blood transcriptome sequencing and large control cohorts. <i>Nature Medicine</i> , 2019, 25, 911-919.	30.7	221
3	MARRVEL: Integration of Human and Model Organism Genetic Resources to Facilitate Functional Annotation of the Human Genome. <i>American Journal of Human Genetics</i> , 2017, 100, 843-853.	6.2	181
4	Recurrent Muscle Weakness with Rhabdomyolysis, Metabolic Crises, and Cardiac Arrhythmia Due to Bi-allelic TANGO2 Mutations. <i>American Journal of Human Genetics</i> , 2016, 98, 347-357.	6.2	98
5	Mutations in PURA Cause Profound Neonatal Hypotonia, Seizures, and Encephalopathy in 5q31.3 Microdeletion Syndrome. <i>American Journal of Human Genetics</i> , 2014, 95, 579-583.	6.2	92
6	IRF2BPL Is Associated with Neurological Phenotypes. <i>American Journal of Human Genetics</i> , 2018, 103, 245-260.	6.2	69
7	De Novo GMNN Mutations Cause Autosomal-Dominant Primordial Dwarfism Associated with Meier-Gorlin Syndrome. <i>American Journal of Human Genetics</i> , 2015, 97, 904-913.	6.2	65
8	A comprehensive iterative approach is highly effective in diagnosing individuals who are exome negative. <i>Genetics in Medicine</i> , 2019, 21, 161-172.	2.4	60
9	Biallelic Mutations in ATP5F1D, which Encodes a Subunit of ATP Synthase, Cause a Metabolic Disorder. <i>American Journal of Human Genetics</i> , 2018, 102, 494-504.	6.2	59
10	Lysosomal Storage and Albinism Due to Effects of a De Novo CLCN7 Variant on Lysosomal Acidification. <i>American Journal of Human Genetics</i> , 2019, 104, 1127-1138.	6.2	59
11	Expanding the Spectrum of BAF-Related Disorders: De Novo Variants in SMARCC2 Cause a Syndrome with Intellectual Disability and Developmental Delay. <i>American Journal of Human Genetics</i> , 2019, 104, 164-178.	6.2	59
12	Bi-allelic Mutations in PKD1L1 Are Associated with Laterality Defects in Humans. <i>American Journal of Human Genetics</i> , 2016, 99, 886-893.	6.2	57
13	Genetic architecture of laterality defects revealed by whole exome sequencing. <i>European Journal of Human Genetics</i> , 2019, 27, 563-573.	2.8	44
14	TM4SF20 Ancestral Deletion and Susceptibility to a Pediatric Disorder of Early Language Delay and Cerebral White Matter Hyperintensities. <i>American Journal of Human Genetics</i> , 2013, 93, 197-210.	6.2	43
15	De Novo Variants in WDR37 Are Associated with Epilepsy, Colobomas, Dysmorphism, Developmental Delay, Intellectual Disability, and Cerebellar Hypoplasia. <i>American Journal of Human Genetics</i> , 2019, 105, 413-424.	6.2	43
16	Looking beyond the exome: a phenotype-first approach to molecular diagnostic resolution in rare and undiagnosed diseases. <i>Genetics in Medicine</i> , 2018, 20, 464-469.	2.4	42
17	Partial Loss of USP9X Function Leads to a Male Neurodevelopmental and Behavioral Disorder Converging on Transforming Growth Factor β Signaling. <i>Biological Psychiatry</i> , 2020, 87, 100-112.	1.3	42
18	Whole exome sequencing in 342 congenital cardiac left sided lesion cases reveals extensive genetic heterogeneity and complex inheritance patterns. <i>Genome Medicine</i> , 2017, 9, 95.	8.2	37

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19	De novo EIF2AK1 and EIF2AK2 Variants Are Associated with Developmental Delay, Leukoencephalopathy, and Neurologic Decompensation. <i>American Journal of Human Genetics</i> , 2020, 106, 570-583.	6.2	37
20	Disruptive variants of <i>CSDE1</i> associate with autism and interfere with neuronal development and synaptic transmission. <i>Science Advances</i> , 2019, 5, eaax2166.	10.3	35
21	A genome-wide association study of congenital cardiovascular left-sided lesions shows association with a locus on chromosome 20. <i>Human Molecular Genetics</i> , 2016, 25, 2331-2341.	2.9	31
22	De Novo Pathogenic Variants in N-cadherin Cause a Syndromic Neurodevelopmental Disorder with Corpus Callosum, Axon, Cardiac, Ocular, and Genital Defects. <i>American Journal of Human Genetics</i> , 2019, 105, 854-868.	6.2	29
23	Bi-allelic Variants in TONSL Cause SPONASTRIME Dysplasia and a Spectrum of Skeletal Dysplasia Phenotypes. <i>American Journal of Human Genetics</i> , 2019, 104, 422-438.	6.2	27
24	An autosomal dominant neurological disorder caused by de novo variants in FAR1 resulting in uncontrolled synthesis of ether lipids. <i>Genetics in Medicine</i> , 2021, 23, 740-750.	2.4	25
25	Congenital heart defects and left ventricular non-compaction in males with loss-of-function variants in <i>NONO</i> . <i>Journal of Medical Genetics</i> , 2017, 54, 47-53.	3.2	24
26	Biallelic MADD variants cause a phenotypic spectrum ranging from developmental delay to a multisystem disorder. <i>Brain</i> , 2020, 143, 2437-2453.	7.6	21
27	Heterozygous variants in <i>MYBPC1</i> are associated with an expanded neuromuscular phenotype beyond arthrogryposis. <i>Human Mutation</i> , 2019, 40, 1115-1126.	2.5	19
28	An exome sequencing study of Moebius syndrome including atypical cases reveals an individual with CFEOM3A and a <i>TUBB3</i> mutation. <i>Journal of Physical Education and Sports Management</i> , 2017, 3, a000984.	1.2	18
29	Whole genome sequencing reveals novel <i>IGHMBP2</i> variant leading to unique cryptic splice site and Charcot-Marie-Tooth phenotype with early onset symptoms. <i>Molecular Genetics & Genomic Medicine</i> , 2019, 7, e00676.	1.2	18
30	Clinical sites of the Undiagnosed Diseases Network: unique contributions to genomic medicine and science. <i>Genetics in Medicine</i> , 2021, 23, 259-271.	2.4	18
31	Missense and truncating variants in CHD5 in a dominant neurodevelopmental disorder with intellectual disability, behavioral disturbances, and epilepsy. <i>Human Genetics</i> , 2021, 140, 1109-1120.	3.8	18
32	Assessment of large copy number variants in patients with apparently isolated congenital left-sided cardiac lesions reveals clinically relevant genomic events. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 2176-2188.	1.2	17
33	Plasma Glutamine Is a Minor Precursor for the Synthesis of Citrulline: A Multispecies Study. <i>Journal of Nutrition</i> , 2017, 147, 549-555.	2.9	16
34	Commonalities across computational workflows for uncovering explanatory variants in undiagnosed cases. <i>Genetics in Medicine</i> , 2021, 23, 1075-1085.	2.4	16
35	Phenotypic expansion of CACNA1C-associated disorders to include isolated neurological manifestations. <i>Genetics in Medicine</i> , 2021, 23, 1922-1932.	2.4	16
36	Early infantile epileptic encephalopathy due to biallelic pathogenic variants in <i>PIGQ</i> : Report of seven new subjects and review of the literature. <i>Journal of Inherited Metabolic Disease</i> , 2020, 43, 1321-1332.	3.6	15

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37	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
38	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
39	<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
40	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
41	Heterozygous loss-of-function variants significantly expand the phenotypes associated with loss of GDF11. Genetics in Medicine, 2021, 23, 1889-1900.	2.4	13
42	Cardiac Crises: Cardiac Arrhythmias and Cardiomyopathy during TANGO2-deficiency related Metabolic Crises. Heart Rhythm, 2022, , .	0.7	13
43	Loss of IRF2BPL impairs neuronal maintenance through excess Wnt signaling. Science Advances, 2022, 8, eabl5613.	10.3	12
44	A novel <i>CACNA1A</i> variant in a child with early stroke and intractable epilepsy. Molecular Genetics & Genomic Medicine, 2020, 8, e1383.	1.2	11
45	Detection of a mosaic <i>CDKL5</i> deletion and inversion by optical genome mapping ends an exhaustive diagnostic odyssey. Molecular Genetics & Genomic Medicine, 2021, 9, e1665.	1.2	11
46	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
47	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
48	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
49	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
50	Aberrant DNA methylation as a diagnostic biomarker of diabetic embryopathy. Genetics in Medicine, 2019, 21, 2453-2461.	2.4	8
51	IgG4â€“related disease: Association with a rare gene variant expressed in cytotoxic T cells. Molecular Genetics & Genomic Medicine, 2019, 7, e686.	1.2	8
52	<i>DYRK1A</i> 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
53	Recessive ACO2 variants as a cause of isolated ophthalmologic phenotypes. American Journal of Medical Genetics, Part A, 2020, 182, 1960-1966.	1.2	8
54	<scp><i>PPP3CA</i></scp> truncating variants clustered in the regulatory domain cause earlyâ€“onset refractory epilepsy. Clinical Genetics, 2021, 100, 227-233.	2.0	7

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55	Cytogenomic Aberrations in Congenital Cardiovascular Malformations. <i>Molecular Syndromology</i> , 2016, 7, 51-61.	0.8	7
56	Magnetic Resonance Imaging characteristics in case of TOR1AIP1 muscular dystrophy. <i>Clinical Imaging</i> , 2019, 58, 108-113.	1.5	6
57	One is the loneliest number: genotypic matchmaking using the electronic health record. <i>Genetics in Medicine</i> , 2021, 23, 1830-1832.	2.4	6
58	The microRNA processor <i>DROSHA</i> is a candidate gene for a severe progressive neurological disorder. <i>Human Molecular Genetics</i> , 2022, 31, 2934-2950.	2.9	6
59	Delineation of a novel neurodevelopmental syndrome associated with <i>PAX5</i> haploinsufficiency. <i>Human Mutation</i> , 2022, 43, 461-470.	2.5	5
60	<i>LMOD2</i> -related dilated cardiomyopathy presenting in late infancy. <i>American Journal of Medical Genetics, Part A</i> , 2022, 188, 1858-1862.	1.2	5
61	Family genetic result communication in rare and undiagnosed disease communities: Understanding the practice. <i>Journal of Genetic Counseling</i> , 2021, 30, 439-447.	1.6	4
62	Health-related quality of life in adults with osteogenesis imperfecta. <i>Clinical Genetics</i> , 2021, 99, 772-779.	2.0	4
63	<i>OTUD5</i> Variants Associated With X-Linked Intellectual Disability and Congenital Malformation. <i>Frontiers in Cell and Developmental Biology</i> , 2021, 9, 631428.	3.7	4
64	Copy Number Variants of Undetermined Significance Are Not Associated with Worse Clinical Outcomes in Hypoplastic Left Heart Syndrome. <i>Journal of Pediatrics</i> , 2018, 202, 206-211.e2.	1.8	3
65	Clinical application of a scale to assess genomic healthcare empowerment (GEmS): Process and illustrative case examples. <i>Journal of Genetic Counseling</i> , 2022, 31, 59-70.	1.6	3
66	A rare description of pure partial trisomy of 16q12.2q24.3 and review of the literature. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 2903-2912.	1.2	2
67	Progressive cerebellar atrophy in a patient with complex II and III deficiency and a novel deleterious variant in <i>SDHA</i> : A Counseling Conundrum. <i>Molecular Genetics & Genomic Medicine</i> , 2021, 9, e1692.	1.2	1
68	Genetic counselor roles in the undiagnosed diseases network research study: Clinical care, collaboration, and curation. <i>Journal of Genetic Counseling</i> , 2022, 31, 326-337.	1.6	1
69	Vertical transmission of a large calvarial ossification defect due to heterozygous variants of <i>ALX4</i> and <i>TWIST1</i> . <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 916-922.	1.2	1
70	Quality of life, illness perceptions, and parental lived experiences in <i>TANGO2</i> -related metabolic encephalopathy and arrhythmias. <i>European Journal of Human Genetics</i> , 0, , .	2.8	1