

Shahriar Nafissi

List of Publications by Year in descending order

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Version: 2024-02-01

119
papers

2,196
citations

218381

26
h-index

288905

40
g-index

120
all docs

120
docs citations

120
times ranked

4030
citing authors

#	ARTICLE	IF	CITATIONS
1	KIF1A, an Axonal Transporter of Synaptic Vesicles, Is Mutated in Hereditary Sensory and Autonomic Neuropathy Type 2. <i>American Journal of Human Genetics</i> , 2011, 89, 219-230.	2.6	172
2	Hexosamine Biosynthetic Pathway Mutations Cause Neuromuscular Transmission Defect. <i>American Journal of Human Genetics</i> , 2011, 88, 162-172.	2.6	153
3	Safety and Efficacy of Nanocurcumin as Add-On Therapy to Riluzole in Patients With Amyotrophic Lateral Sclerosis: A Pilot Randomized Clinical Trial. <i>Neurotherapeutics</i> , 2018, 15, 430-438.	2.1	90
4	International clinimetric evaluation of the MG-QOL15, resulting in slight revision and subsequent validation of the MG-QOL15r. <i>Muscle and Nerve</i> , 2016, 54, 1015-1022.	1.0	85
5	Congenital myasthenic syndrome with tubular aggregates caused by GFPT1 mutations. <i>Journal of Neurology</i> , 2012, 259, 838-850.	1.8	72
6	Long-term follow-up of patients with congenital myasthenic syndrome caused by COLQ mutations. <i>Neuromuscular Disorders</i> , 2012, 22, 318-324.	0.3	64
7	Sequential targeted exome sequencing of 1001 patients affected by unexplained limb-girdle weakness. <i>Genetics in Medicine</i> , 2020, 22, 1478-1488.	1.1	62
8	Mitochondrial complex I deficiency and ATP/ADP ratio in lymphocytes of amyotrophic lateral sclerosis patients. <i>Neurological Research</i> , 2012, 34, 297-303.	0.6	54
9	Wilson's Disease: A Great Masquerader. <i>European Neurology</i> , 2007, 57, 80-85.	0.6	50
10	Identification of mutation in GTPBP2 in patients of a family with neurodegeneration accompanied by iron deposition in the brain. <i>Neurobiology of Aging</i> , 2016, 38, 216.e11-216.e18.	1.5	43
11	Improved diagnostic yield of neuromuscular disorders applying clinical exome sequencing in patients arising from a consanguineous population. <i>Clinical Genetics</i> , 2017, 91, 386-402.	1.0	41
12	Detection of variants in dystroglycanopathy-associated genes through the application of targeted whole-exome sequencing analysis to a large cohort of patients with unexplained limb-girdle muscle weakness. <i>Skeletal Muscle</i> , 2018, 8, 23.	1.9	40
13	Complex I and ATP Content Deficiency in Lymphocytes from Friedreich's Ataxia. <i>Canadian Journal of Neurological Sciences</i> , 2009, 36, 26-31.	0.3	39
14	Islamic fasting and multiple sclerosis. <i>BMC Neurology</i> , 2014, 14, 56.	0.8	38
15	Mutation in <i>ADORA1</i> identified as likely cause of early-onset parkinsonism and cognitive dysfunction. <i>Movement Disorders</i> , 2016, 31, 1004-1011.	2.2	38
16	Impact of depression and disability on quality of life in Iranian patients with multiple sclerosis. <i>Multiple Sclerosis Journal</i> , 2007, 13, 275-277.	1.4	35
17	Silibinin inhibits invasive properties of human glioblastoma U87MG cells through suppression of cathepsin B and nuclear factor kappa B-mediated induction of matrix metalloproteinase 9. <i>Anti-Cancer Drugs</i> , 2010, 21, 252-260.	0.7	35
18	Phenotypic stratification and genotype-phenotype correlation in a heterogeneous, international cohort of GNE myopathy patients: First report from the GNE myopathy Disease Monitoring Program, registry portion. <i>Neuromuscular Disorders</i> , 2018, 28, 158-168.	0.3	35

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19	Genetic analysis and SOD1 mutation screening in Iranian amyotrophic lateral sclerosis patients. <i>Neurobiology of Aging</i> , 2013, 34, 1516.e1-1516.e8.	1.5	34
20	Gene Expression Profiling of Mitochondrial Oxidative Phosphorylation (OXPHOS) Complex I in Friedreich Ataxia (FRDA) Patients. <i>PLoS ONE</i> , 2014, 9, e94069.	1.1	32
21	Repeat expansion in C9ORF72 is not a major cause of amyotrophic lateral sclerosis among Iranian patients. <i>Neurobiology of Aging</i> , 2014, 35, 267.e1-267.e7.	1.5	32
22	Ryanodine receptor type 3 (<i>RYR3</i>) as a novel gene associated with a myopathy with nemaline bodies. <i>European Journal of Neurology</i> , 2018, 25, 841-847.	1.7	31
23	Effect of Vitamin A Supplementation on fatigue and depression in Multiple Sclerosis patients: A Double-Blind Placebo-Controlled Clinical Trial. <i>Iranian Journal of Allergy, Asthma and Immunology</i> , 2016, 15, 13-9.	0.3	31
24	Abnormal viscoelastic behaviour of passive ankle joint movement in diabetic patients: an early or a late complication?. <i>Diabetologia</i> , 2005, 48, 1225-1228.	2.9	29
25	POGLUT1 biallelic mutations cause myopathy with reduced satellite cells, $\hat{\pm}$ -dystroglycan hypoglycosylation and a distinctive radiological pattern. <i>Acta Neuropathologica</i> , 2020, 139, 565-582.	3.9	29
26	Diagnosis and treatment of late-onset Pompe disease in the Middle East and North Africa region: consensus recommendations from an expert group. <i>BMC Neurology</i> , 2015, 15, 205.	0.8	28
27	<i>POPDC3</i> Gene Variants Associate with a New Form of Limb Girdle Muscular Dystrophy. <i>Annals of Neurology</i> , 2019, 86, 832-843.	2.8	27
28	Four novel C20orf54 mutations identified in Brown-Vialetto-Van Laere syndrome patients. <i>Journal of Human Genetics</i> , 2012, 57, 613-617.	1.1	26
29	Effect of skin thickness on sensory nerve action potential amplitude. <i>Clinical Neurophysiology</i> , 2008, 119, 1824-1828.	0.7	25
30	Impact of Vitamin A Supplementation on Disease Progression in Patients with Multiple Sclerosis. <i>Archives of Iranian Medicine</i> , 2015, 18, 435-40.	0.2	25
31	Guillain-Barre Syndrome and COVID-19 Vaccine: A Report of Nine Patients. <i>Basic and Clinical Neuroscience</i> , 2021, 12, 703-710.	0.3	24
32	Dietary intake of nutrients and its correlation with fatigue in multiple sclerosis patients. <i>Iranian Journal of Neurology</i> , 2014, 13, 28-32.	0.5	22
33	Intraspinal delivery of bone marrow stromal cell-derived neural stem cells in patients with amyotrophic lateral sclerosis: A safety and feasibility study. <i>Journal of the Neurological Sciences</i> , 2016, 362, 174-181.	0.3	21
34	Limb girdle muscular dystrophy due to mutations in <i>POMT2</i> . <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2018, 89, 506-512.	0.9	21
35	Myasthenia gravis associated with novel coronavirus 2019 infection: A report of three cases. <i>Clinical Neurology and Neurosurgery</i> , 2021, 208, 106834.	0.6	21
36	Identification and sizing of GAA trinucleotide repeat expansion, investigation for D-loop variations and mitochondrial deletions in Iranian patients with Friedreich's ataxia. <i>Mitochondrion</i> , 2006, 6, 87-93.	1.6	20

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37	Association of HLA class II (DRB1, DQA1, DQB1) alleles and haplotypes with myasthenia gravis and its subgroups in the Iranian population. <i>Journal of the Neurological Sciences</i> , 2015, 359, 335-342.	0.3	20
38	Carrier frequency of SMA by quantitative analysis of the SMN1 deletion in the Iranian population. <i>European Journal of Neurology</i> , 2010, 17, 160-162.	1.7	18
39	Identification of mutation in NPC2 by exome sequencing results in diagnosis of Niemann-Pick disease type C. <i>Molecular Genetics and Metabolism</i> , 2013, 110, 139-144.	0.5	18
40	Genetics of GNE myopathy in the non-Jewish Persian population. <i>European Journal of Human Genetics</i> , 2016, 24, 243-251.	1.4	18
41	LGMD2E is the most common type of sarcoglycanopathies in the Iranian population. <i>Journal of Neurogenetics</i> , 2017, 31, 161-169.	0.6	18
42	Trends of quality of life changes in amyotrophic lateral sclerosis patients. <i>Journal of the Neurological Sciences</i> , 2016, 368, 35-40.	0.3	17
43	Therapeutic Efficacy of Silibinin on Human Neuroblastoma Cells: Akt and NF- κ B Expressions May Play an Important Role in Silibinin-Induced Response. <i>Neurochemical Research</i> , 2012, 37, 2053-2063.	1.6	16
44	Comparison of electrophysiological findings in axonal and demyelinating Guillain-Barre syndrome. <i>Iranian Journal of Neurology</i> , 2014, 13, 138-43.	0.5	15
45	Comparing efficacy and side effects of a weekly intramuscular biogeneric/biosimilar interferon beta-1a with Avonex in relapsing remitting multiple sclerosis: A double blind randomized clinical trial. <i>Clinical Neurology and Neurosurgery</i> , 2012, 114, 986-989.	0.6	14
46	HMSN-P caused by p.Pro285Leu mutation in TFG is not confined to patients with Far East ancestry. <i>Neurobiology of Aging</i> , 2015, 36, 1606.e1-1606.e7.	1.5	14
47	Multiple acyl-coenzyme A dehydrogenase deficiency shows a possible founder effect and is the most frequent cause of lipid storage myopathy in Iran. <i>Journal of the Neurological Sciences</i> , 2020, 411, 116707.	0.3	14
48	A Novel Mitochondrial Heteroplasmic C13806A Point Mutation Associated with Iranian Friedreich's Ataxia. <i>Cellular and Molecular Neurobiology</i> , 2009, 29, 225-233.	1.7	13
49	Impact of Vitamin A Supplementation on RAR Gene Expression in Multiple Sclerosis Patients. <i>Journal of Molecular Neuroscience</i> , 2013, 51, 478-484.	1.1	12
50	Granulocyte Colony-Stimulating Factor for Amyotrophic Lateral Sclerosis: A Randomized,		

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55	Extending the clinical and mutational spectrum of <i>TRIM32</i> -related myopathies in a non-Hutterite population. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2019, 90, 490-493.	0.9	11
56	Clinical and genetic spectrum of a large cohort of patients with β -sarcoglycan muscular dystrophy. <i>Brain</i> , 2022, 145, 596-606.	3.7	11
57	Mitochondrial <i>tRNA^{Leu/Lys}</i> and <i>ATPase 6/8</i> Gene Variations in Spinocerebellar Ataxias. <i>Neurodegenerative Diseases</i> , 2009, 6, 16-22.	0.8	10
58	Identification of novel TFG mutation in HMSN-P pedigree: Emphasis on variable clinical presentations. <i>Journal of the Neurological Sciences</i> , 2016, 369, 318-323.	0.3	10
59	Recommendations for Infantile-Onset and Late-Onset Pompe Disease: An Iranian Consensus. <i>Frontiers in Neurology</i> , 2021, 12, 739931.	1.1	10
60	Correlation between distribution of muscle weakness, electrophysiological findings and CTG expansion in myotonic dystrophy. <i>Journal of Clinical Neuroscience</i> , 2014, 21, 1123-1126.	0.8	9
61	Intellectual disability, muscle weakness and characteristic face in three siblings: A newly described recessive syndrome mapping to 3p24.3â€“p25.3. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 2508-2515.	0.7	9
62	Association between trinucleotide CAG repeats of the DNA polymerase gene (POLG) with age of onset of Iranian Friedreichâ€™s ataxia patients. <i>Neurological Sciences</i> , 2008, 29, 489-493.	0.9	8
63	Muscle-specific kinase antibody associated myasthenia gravis after bone marrow transplantation. <i>Neuromuscular Disorders</i> , 2014, 24, 148-150.	0.3	8
64	High Frequency of Tc22 and Th22 Cells in Myasthenia Gravis Patients and Their Significant Reduction after Thymectomy. <i>NeuroImmunoModulation</i> , 2018, 25, 80-88.	0.9	8
65	Persian adaptation of Edinburgh Cognitive and Behavioural Screen (ECAS). <i>Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration</i> , 2021, 22, 426-433.	1.1	8
66	Description of clinical features and genetic analysis of one ultra-rare (SPG64) and two common forms (SPG5A and SPG15) of hereditary spastic paraplegia families. <i>Journal of Neurogenetics</i> , 2021, 35, 84-94.	0.6	8
67	Electrophysiological evaluation in lumbosacral radiculopathy. <i>Iranian Journal of Neurology</i> , 2012, 11, 83-6.	0.5	8
68	The role of cytomegalovirus, Haemophilus influenzae and Epstein Barr virus in Guillain Barre syndrome. <i>Acta Medica Iranica</i> , 2013, 51, 372-6.	0.8	8
69	Report of a patient with limb-girdle muscular dystrophy, ptosis and ophthalmoparesis caused by plectinopathy. <i>Archives of Iranian Medicine</i> , 2015, 18, 60-4.	0.2	8
70	Dystrophin Gene Mutation Analysis in Iranian Males and Females Using Multiplex Polymerase Chain Reaction and Multiplex Ligation-Dependent Probe Amplification Methods. <i>Genetic Testing and Molecular Biomarkers</i> , 2011, 15, 893-899.	0.3	7
71	Iranian consensus on use of vitamin D in patients with multiple sclerosis. <i>BMC Neurology</i> , 2016, 16, 76.	0.8	7
72	Continuum of phenotypes in hereditary motor and sensory neuropathy with proximal predominance and Charcotâ€™s Marieâ€™Tooth patients with <i>TFG</i> mutation. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 1507-1515.	0.7	7

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73	Mutation screening of SLC52A3, C19orf12, and TARDBP in Iranian ALS patients. <i>Neurobiology of Aging</i> , 2019, 75, 225.e9-225.e14.	1.5	7
74	Description of combined ARHSP/JALS phenotype in some patients with SPG11 mutations. <i>Molecular Genetics & Genomic Medicine</i> , 2020, 8, e1240.	0.6	7
75	Investigating the possible association between <scp>NLRP3</scp> gene polymorphisms and myasthenia gravis. <i>Muscle and Nerve</i> , 2021, 63, 730-736.	1.0	7
76	Association of amyotrophic lateral sclerosis and Behcetâ€™s disease: is there a relationship? A multi-national case series. <i>Clinical Rheumatology</i> , 2012, 31, 733-738.	1.0	6
77	Lateâ€œonset pompe disease in Iran: A clinical and genetic report. <i>Muscle and Nerve</i> , 2017, 55, 835-840.	1.0	6
78	Genotype and phenotype analysis of 43 Iranian facioscapulohumeral muscular dystrophy patients; Evidence for anticipation. <i>Neuromuscular Disorders</i> , 2018, 28, 303-314.	0.3	6
79	Sporadic late-onset nemaline myopathy with monoclonal gammopathy of undetermined significance: Report of four patients. <i>Neuromuscular Disorders</i> , 2021, 31, 29-34.	0.3	6
80	Anticipation Can Be More Common in Hereditary Spastic Paraplegia with <i>SPAST</i> Mutations Than It Appears. <i>Canadian Journal of Neurological Sciences</i> , 2022, 49, 651-661.	0.3	6
81	Molecular Diagnosis of Hereditary Neuropathies by Whole Exome Sequencing and Expanding the Phenotype Spectrum. <i>Archives of Iranian Medicine</i> , 2020, 23, 426-433.	0.2	6
82	Validation of the Persian version of the 40-item amyotrophic lateral sclerosis assessment questionnaire. <i>Iranian Journal of Neurology</i> , 2013, 12, 102-5.	0.5	6
83	Effect of Repetitive Transcranial Magnetic Stimulation on Reducing Spasticity in Patients Suffering From HTLV-1â€™Associated Myelopathy. <i>Journal of Clinical Neurophysiology</i> , 2014, 31, 547-551.	0.9	5
84	Report of limb girdle muscular dystrophy type 2a in 6 Iranian patients, one with a novel deletion in CAPN3 gene. <i>Neuromuscular Disorders</i> , 2016, 26, 277-282.	0.3	5
85	Distinct Clinical and Genetic Findings in Iranian Patients With Glycogen Storage Disease Type 3. <i>Journal of Clinical Neuromuscular Disease</i> , 2018, 19, 203-210.	0.3	5
86	Thigh and Leg Muscle MRI Findings in GNE Myopathy. <i>Journal of Neuromuscular Diseases</i> , 2021, 8, 735-742.	1.1	5
87	Clinical and electrophysiological features of Guillainâ€™BarrÃ© syndrome in Iran. <i>Journal of Clinical Neuroscience</i> , 2014, 21, 1554-1557.	0.8	4
88	Blink Reflex May Help Discriminate Alzheimer Disease From Vascular Dementia. <i>Journal of Clinical Neurophysiology</i> , 2015, 32, 505-511.	0.9	4
89	Evaluation of point mutations in dystrophin gene in Iranian Duchenne and Becker muscular dystrophy patients: introducing three novel variants. <i>Journal of Genetics</i> , 2016, 95, 325-329.	0.4	4
90	Zytux in Refractory Myasthenia Gravis: A Multicenter, Open-Labeled, Clinical Trial Study of Effectiveness and Safety of a Rituximab Biosimilar. <i>Frontiers in Neurology</i> , 2021, 12, 682622.	1.1	4

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91	The value of <sc>MUNIX</sc> as an objective electrophysiological biomarker of disease progression in chronic inflammatory demyelinating polyneuropathy. <i>Muscle and Nerve</i> , 2022, 65, 433-439.	1.0	4
92	A novel heteroplasmic mutation in mitochondrial tRNA ^{Arg} gene associated with non-dystrophic myotonias. <i>Acta Neurologica Belgica</i> , 2020, 120, 573-580.	0.5	3
93	Application of muscle ultrasound for the evaluation of patients with amyotrophic lateral sclerosis: An observational cross-sectional study. <i>Muscle and Nerve</i> , 2020, 62, 516-521.	1.0	3
94	BVVL/ FL: features caused by SLC52A3 mutations; WDFY4 and TNFSF13B may be novel causative genes. <i>Neurobiology of Aging</i> , 2021, 99, 102.e1-102.e10.	1.5	3
95	Application of CMAP scan for the evaluation of patients with chronic inflammatory demyelinating polyneuropathy: a prospective study. <i>Neurophysiologie Clinique</i> , 2021, 51, 175-181.	1.0	3
96	Disease severity and response to treatment in Iranian patients with myasthenia gravis. <i>Neurological Sciences</i> , 2022, 43, 1233-1237.	0.9	3
97	Retrospective analysis of response to rituximab in chronic inflammatory demyelinating polyneuropathy refractory to first-line therapy. <i>Journal of the Peripheral Nervous System</i> , 2021, 26, 469-474.	1.4	3
98	Observation of c.260A > G mutation in superoxide dismutase 1 that causes p.Asn86Ser in Iranian amyotrophic lateral sclerosis patient and absence of genotype/phenotype correlation. <i>Iranian Journal of Neurology</i> , 2015, 14, 152-7.	0.5	3
99	Mutation analysis in exons 22 and 24 of SCN4A gene in Iranian patients with non-dystrophic myotonia. <i>Iranian Journal of Neurology</i> , 2015, 14, 190-4.	0.5	3
100	Mitochondrial Copy Number and D-Loop Variants in Pompe Patients. <i>Cell Journal</i> , 2016, 18, 405-15.	0.2	3
101	Delayed-onset inflammatory polyneuropathy without graft versus host disease after bone marrow transplantation. <i>Muscle and Nerve</i> , 2018, 58, E11-E13.	1.0	2
102	Deep geno- and phenotyping in two consanguineous families with CMT2 reveals HADHA as an unusual disease-causing gene and an intronic variant in GDAP1 as an unusual mutation. <i>Journal of Neurology</i> , 2021, 268, 640-650.	1.8	2
103	Novel Mutations of the TYMP Gene in Mitochondrial Neurogastrointestinal Encephalomyopathy: Case Series and Literature Review. <i>Journal of Molecular Neuroscience</i> , 2021, 71, 2526-2533.	1.1	2
104	The correlation of the serum level of L-carnitine with disease severity in patients with Amyotrophic lateral sclerosis. <i>Journal of Clinical Neuroscience</i> , 2021, 89, 232-236.	0.8	2
105	An Iranian familial amyotrophic lateral sclerosis pedigree with p.Val48Phe causing mutation in SOD1: a genetic and clinical report. <i>Iranian Journal of Basic Medical Sciences</i> , 2014, 17, 735-9.	1.0	2
106	An overview of motor unit number index reproducibility in amyotrophic lateral sclerosis. <i>Iranian Journal of Neurology</i> , 2019, 18, 119-126.	0.5	2
107	Current Concepts in Botulism. <i>Journal of Clinical Neuromuscular Disease</i> , 2003, 4, 139-149.	0.3	1
108	A case of adult onset Sandhoff disease that mimics Brown-Vialetto-Van Laere syndrome. <i>Neuromuscular Disorders</i> , 2021, 31, 528-531.	0.3	1

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109	Association of HLA Class II Alleles with Disease Severity and Treatment Response in Iranian Patients with Myasthenia Gravis. <i>Journal of Neuromuscular Diseases</i> , 2021, 8, 827-829.	1.1	1
110	Evaluation of Quality of Life in Patients With Chronic Inflammatory Demyelinating Polyneuropathy in Iran. <i>Journal of Clinical Neuromuscular Disease</i> , 2019, 21, 77-83.	0.3	1
111	An overview of motor unit number index reproducibility in amyotrophic lateral sclerosis. <i>Current Journal of Neurology</i> , 0, , .	0.0	1
112	Clinical and Pathological Features of Lipid Storage Myopathy; A Retrospective Study of a Large Group from Iran. <i>International Clinical Neuroscience Journal</i> , 2020, 8, 26-29.	0.1	1
113	Four cases of brown-vialetto-van laere syndrome from Iran: Clinical and electrophysiologic findings. <i>Iranian Journal of Neurology</i> , 2011, 10, 54-7.	0.5	1
114	Iranian Consensus Recommendations for Treatment of Myasthenia Gravis. <i>Archives of Iranian Medicine</i> , 2022, 25, 37-49.	0.2	1
115	Practical needs and considerations for refugees and other forcibly displaced persons with neurological disorders: Recommendations using a modified Delphi approach. <i>Gates Open Research</i> , 2021, 5, 178.	2.0	1
116	De novo Mutation in CACNA1S Gene in a 20-Year-Old Man Diagnosed with Metabolic Myopathy. <i>Archives of Iranian Medicine</i> , 2017, 20, 617-620.	0.2	1
117	The role of opening CSF pressure in response to treatment for idiopathic intracranial hypertension (IIH). <i>Journal of Clinical Neuroscience</i> , 2020, 76, 171-176.	0.8	0
118	Validation of Persian Individualized Neuromuscular Quality of Life in patients with muscular dystrophies. <i>Current Journal of Neurology</i> , 0, , .	0.0	0
119	RABENOSYN separation-of-function mutations uncouple endosomal recycling from lysosomal degradation, causing a distinct Mendelian Disorder. <i>Human Molecular Genetics</i> , 0, , .	1.4	0