Shahriar Nafissi

List of Publications by Year
in descending order

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119 papers	2,196 citations	218677 26 h-index	289244 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. American Journal of Human Genetics, 2011, 89, 219-230.	6.2	172
2	Hexosamine Biosynthetic Pathway Mutations Cause Neuromuscular Transmission Defect. American Journal of Human Genetics, 2011, 88, 162-172.	6.2	153
3	Safety and Efficacy of Nanocurcumin as Add-On Therapy to Riluzole in Patients With Amyotrophic Lateral Sclerosis: A Pilot Randomized Clinical Trial. Neurotherapeutics, 2018, 15, 430-438.	4.4	90
4	International clinimetric evaluation of the MGâ€QOL15, resulting in slight revision and subsequent validation of the MGâ€QOL15r. Muscle and Nerve, 2016, 54, 1015-1022.	2.2	85
5	Congenital myasthenic syndrome with tubular aggregates caused by GFPT1 mutations. Journal of Neurology, 2012, 259, 838-850.	3.6	72
6	Long-term follow-up of patients with congenital myasthenic syndrome caused by COLQ mutations. Neuromuscular Disorders, 2012, 22, 318-324.	0.6	64
7	Sequential targeted exome sequencing of 1001 patients affected by unexplained limb-girdle weakness. Genetics in Medicine, 2020, 22, 1478-1488.	2.4	62
8	Mitochondrial complex I deficiency and ATP/ADP ratio in lymphocytes of amyotrophic lateral sclerosis patients. Neurological Research, 2012, 34, 297-303.	1.3	54
9	Wilsonâ€™s Disease: A Great Masquerader. European Neurology, 2007, 57, 80-85.	1.4	50
10	Identification of mutation in GTPBP2 in patients of a family with neurodegeneration accompanied by iron deposition in the brain. Neurobiology of Aging, 2016, 38, 216.e11-216.e18.	3.1	43
11	Improved diagnostic yield of neuromuscular disorders applying clinical exome sequencing in patients arising from a consanguineous population. Clinical Genetics, 2017, 91, 386-402.	2.0	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. Skeletal Muscle, 2018, 8, 23.	4.2	40
13	Complex I and ATP Content Deficiency in Lymphocytes from Friedreich's Ataxia. Canadian Journal of Neurological Sciences, 2009, 36, 26-31.	0.5	39
14	Islamic fasting and multiple sclerosis. BMC Neurology, 2014, 14, 56.	1.8	38
15	Mutation inADORA1identified as likely cause of early-onset parkinsonism and cognitive dysfunction. Movement Disorders, 2016, 31, 1004-1011.	3.9	38
16	Impact of depression and disability on quality of life in Iranian patients with multiple sclerosis. Multiple Sclerosis Journal, 2007, 13, 275-277.	3.0	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. Anti-Cancer Drugs, 2010, 21, 252-260.	1.4	35
18	Phenotypic stratification and genotypeâ€“phenotype correlation in a heterogeneous, international cohort of GNE myopathy patients: First report from the GNE myopathy Disease Monitoring Program, registry portion. Neuromuscular Disorders, 2018, 28, 158-168.	0.6	35

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19	Genetic analysis and SOD1 mutation screening in Iranian amyotrophic lateral sclerosis patients. Neurobiology of Aging, 2013, 34, 1516.e1-1516.e8.	3.1	34
20	Gene Expression Profiling of Mitochondrial Oxidative Phosphorylation (OXPHOS) Complex I in Friedreich Ataxia (FRDA) Patients. PLoS ONE, 2014, 9, e94069.	2.5	32
21	Repeat expansion in C9ORF72 is not a major cause of amyotrophic lateral sclerosis among Iranian patients. Neurobiology of Aging, 2014, 35, 267.e1-267.e7.	3.1	32
22	Ryanodine receptor type 3 (<scp>RYR</scp>3) as a novel gene associated with a myopathy with nemaline bodies. European Journal of Neurology, 2018, 25, 841-847.	3.3	31
23	Effect of Vitamin A Supplementation on fatigue and depression in Multiple Sclerosis patients: A Double-Blind Placebo-Controlled Clinical Trial. Iranian Journal of Allergy, Asthma and Immunology, 2016, 15, 13-9.	0.4	31
24	Abnormal viscoelastic behaviour of passive ankle joint movement in diabetic patients: an early or a late complication?. Diabetologia, 2005, 48, 1225-1228.	6.3	29
25	POGLUT1 biallelic mutations cause myopathy with reduced satellite cells, Î±-dystroglycan hypoglycosylation and a distinctive radiological pattern. Acta Neuropathologica, 2020, 139, 565-582.	7.7	29
26	Diagnosis and treatment of late-onset Pompe disease in the Middle East and North Africa region: consensus recommendations from an expert group. BMC Neurology, 2015, 15, 205.	1.8	28
27	POPDC3 Gene Variants Associate with a New Form of Limb Girdle Muscular Dystrophy. Annals of Neurology, 2019, 86, 832-843.	5.3	27
28	Four novel C20orf54 mutations identified in Brownâ€“Vialettoâ€“Van Laere syndrome patients. Journal of Human Genetics, 2012, 57, 613-617.	2.3	26
29	Effect of skin thickness on sensory nerve action potential amplitude. Clinical Neurophysiology, 2008, 119, 1824-1828.	1.5	25
30	Impact of Vitamin A Supplementation on Disease Progression in Patients with Multiple Sclerosis. Archives of Iranian Medicine, 2015, 18, 435-40.	0.6	25
31	Guillain-Barre Syndrome and COVID-19 Vaccine: A Report of Nine Patients. Basic and Clinical Neuroscience, 2021, 12, 703-710.	0.6	24
32	Dietary intake of nutrients and its correlation with fatigue in multiple sclerosis patients. Iranian Journal of Neurology, 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. Journal of the Neurological Sciences, 2016, 362, 174-181.	0.6	21
34	Limb girdle muscular dystrophy due to mutations in POMT2. Journal of Neurology, Neurosurgery and Psychiatry, 2018, 89, 506-512.	1.9	21
35	Myasthenia gravis associated with novel coronavirus 2019 infection: A report of three cases. Clinical Neurology and Neurosurgery, 2021, 208, 106834.	1.4	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. Mitochondrion, 2006, 6, 87-93.	3.4	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. Journal of the Neurological Sciences, 2015, 359, 335-342.	0.6	20
38	Carrier frequency of SMA by quantitative analysis of the SMN1 deletion in the Iranian population. European Journal of Neurology, 2010, 17, 160-162.	3.3	18
39	Identification of mutation in NPC2 by exome sequencing results in diagnosis of Niemannâ€“Pick disease type C. Molecular Genetics and Metabolism, 2013, 110, 139-144.	1.1	18
40	Genetics of GNE myopathy in the non-Jewish Persian population. European Journal of Human Genetics, 2016, 24, 243-251.	2.8	18
41	LGMD2E is the most common type of sarcoglycanopathies in the Iranian population. Journal of Neurogenetics, 2017, 31, 161-169.	1.4	18
42	Trends of quality of life changes in amyotrophic lateral sclerosis patients. Journal of the Neurological Sciences, 2016, 368, 35-40.	0.6	17
43	Therapeutic Efficacy of Silibinin on Human Neuroblastoma Cells: Akt and NF-Î¹B Expressions May Play an Important Role in Silibinin-Induced Response. Neurochemical Research, 2012, 37, 2053-2063.	3.3	16
44	Comparison of electrophysiological findings in axonal and demyelinating Guillain-Barre syndrome. Iranian Journal of Neurology, 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. Clinical Neurology and Neurosurgery, 2012, 114, 986-989.	1.4	14
46	HMSN-P caused by p.Pro285Leu mutation in TFG is not confined to patients with Far East ancestry. Neurobiology of Aging, 2015, 36, 1606.e1-1606.e7.	3.1	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. Journal of the Neurological Sciences, 2020, 411, 116707.	0.6	14
48	A Novel Mitochondrial Heteroplasmic C13806A Point Mutation Associated with Iranian Friedreichâ€™s Ataxia. Cellular and Molecular Neurobiology, 2009, 29, 225-233.	3.3	13
49	Impact of Vitamin A Supplementation on RAR Gene Expression in Multiple Sclerosis Patients. Journal of Molecular Neuroscience, 2013, 51, 478-484.	2.3	12
50	Granulocyte Colony-Stimulating Factor for Amyotrophic Lateral Sclerosis: A Randomized,		

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55	Extending the clinical and mutational spectrum ofTRIM32-related myopathies in a non-Hutterite population. Journal of Neurology, Neurosurgery and Psychiatry, 2019, 90, 490-493.	1.9	11
56	Clinical and genetic spectrum of a large cohort of patients with Î´-sarcoglycan muscular dystrophy. Brain, 2022, 145, 596-606.	7.6	11
57	Mitochondrial <i>tRNALeu/Lys</i> and <i>ATPase 6/8</i> Gene Variations in Spinocerebellar Ataxias. Neurodegenerative Diseases, 2009, 6, 16-22.	1.4	10
58	Identification of novel TFG mutation in HMSN-P pedigree: Emphasis on variable clinical presentations. Journal of the Neurological Sciences, 2016, 369, 318-323.	0.6	10
59	Recommendations for Infantile-Onset and Late-Onset Pompe Disease: An Iranian Consensus. Frontiers in Neurology, 2021, 12, 739931.	2.4	10
60	Correlation between distribution of muscle weakness, electrophysiological findings and CTG expansion in myotonic dystrophy. Journal of Clinical Neuroscience, 2014, 21, 1123-1126.	1.5	9
61	Intellectual disability, muscle weakness and characteristic face in three siblings: A newly described recessive syndrome mapping to 3p24.3â€“p25.3. American Journal of Medical Genetics, Part A, 2015, 167, 2508-2515.	1.2	9
62	Association between trinucleotide CAG repeats of the DNA polymerase gene (POLG) with age of onset of Iranian Friedreichâ€™s ataxia patients. Neurological Sciences, 2008, 29, 489-493.	1.9	8
63	Muscle-specific kinase antibody associated myasthenia gravis after bone marrow transplantation. Neuromuscular Disorders, 2014, 24, 148-150.	0.6	8
64	High Frequency of Tc22 and Th22 Cells in Myasthenia Gravis Patients and Their Significant Reduction after Thymectomy. NeuroImmunoModulation, 2018, 25, 80-88.	1.8	8
65	Persian adaptation of Edinburgh Cognitive and Behavioural Screen (ECAS). Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2021, 22, 426-433.	1.7	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. Journal of Neurogenetics, 2021, 35, 84-94.	1.4	8
67	Electrophysiological evaluation in lumbosacral radiculopathy. Iranian Journal of Neurology, 2012, 11, 83-6.	0.5	8
68	The role of cytomegalovirus, Haemophilus influenzae and Epstein Barr virus in Guillain Barre syndrome. Acta Medica Iranica, 2013, 51, 372-6.	0.8	8
69	Report of a patient with limb-girdle muscular dystrophy, ptosis and ophthalmoparesis caused by plectinopathy. Archives of Iranian Medicine, 2015, 18, 60-4.	0.6	8
70	Dystrophin Gene Mutation Analysis in Iranian Males and Females Using Multiplex Polymerase Chain Reaction and Multiplex Ligation-Dependent Probe Amplification Methods. Genetic Testing and Molecular Biomarkers, 2011, 15, 893-899.	0.7	7
71	Iranian consensus on use of vitamin D in patients with multiple sclerosis. BMC Neurology, 2016, 16, 76.	1.8	7
72	Continuum of phenotypes in hereditary motor and sensory neuropathy with proximal predominance and Charcotâ€“Marieâ€“Tooth patients with TFG mutation. American Journal of Medical Genetics, Part A, 2019, 179, 1507-1515.	1.2	7

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73	Mutation screening of SLC52A3, C19orf12, and TARDBP in Iranian ALS patients. Neurobiology of Aging, 2019, 75, 225.e9-225.e14.	3.1	7
74	Description of combined ARHSP/JALS phenotype in some patients with SPG11 mutations. Molecular Genetics & Genomic Medicine, 2020, 8, e1240.	1.2	7
75	Investigating the possible association between <scp>NLRP3</scp> gene polymorphisms and myasthenia gravis. Muscle and Nerve, 2021, 63, 730-736.	2.2	7
76	Association of amyotrophic lateral sclerosis and Behcetâ€™s disease: is there a relationship? A multi-national case series. Clinical Rheumatology, 2012, 31, 733-738.	2.2	6
77	Lateâ€onset pompe disease in Iran: A clinical and genetic report. Muscle and Nerve, 2017, 55, 835-840.	2.2	6
78	Genotype and phenotype analysis of 43 Iranian facioscapulohumeral muscular dystrophy patients; Evidence for anticipation. Neuromuscular Disorders, 2018, 28, 303-314.	0.6	6
79	Sporadic late-onset nemaline myopathy with monoclonal gammopathy of undetermined significance: Report of four patients. Neuromuscular Disorders, 2021, 31, 29-34.	0.6	6
80	Anticipation Can Be More Common in Hereditary Spastic Paraplegia with SPAST Mutations Than It Appears. Canadian Journal of Neurological Sciences, 2022, 49, 651-661.	0.5	6
81	Molecular Diagnosis of Hereditary Neuropathies by Whole Exome Sequencing and Expanding the Phenotype Spectrum. Archives of Iranian Medicine, 2020, 23, 426-433.	0.6	6
82	Validation of the Persian version of the 40-item amyotrophic lateral sclerosis assessment questionnaire. Iranian Journal of Neurology, 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. Journal of Clinical Neurophysiology, 2014, 31, 547-551.	1.7	5
84	Report of limb girdle muscular dystrophy type 2a in 6 Iranian patients, one with a novel deletion in CAPN3 gene. Neuromuscular Disorders, 2016, 26, 277-282.	0.6	5
85	Distinct Clinical and Genetic Findings in Iranian Patients With Glycogen Storage Disease Type 3. Journal of Clinical Neuromuscular Disease, 2018, 19, 203-210.	0.7	5
86	Thigh and Leg Muscle MRI Findings in GNE Myopathy. Journal of Neuromuscular Diseases, 2021, 8, 735-742.	2.6	5
87	Clinical and electrophysiological features of Guillainâ€“BarrÃ© syndrome in Iran. Journal of Clinical Neuroscience, 2014, 21, 1554-1557.	1.5	4
88	Blink Reflex May Help Discriminate Alzheimer Disease From Vascular Dementia. Journal of Clinical Neurophysiology, 2015, 32, 505-511.	1.7	4
89	Evaluation of point mutations in dystrophin gene in Iranian Duchenne and Becker muscular dystrophy patients: introducing three novel variants. Journal of Genetics, 2016, 95, 325-329.	0.7	4
90	Zytux in Refractory Myasthenia Gravis: A Multicenter, Open-Labeled, Clinical Trial Study of Effectiveness and Safety of a Rituximab Biosimilar. Frontiers in Neurology, 2021, 12, 682622.	2.4	4

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91	The value of <scp>MUNIX</scp> as an objective electrophysiological biomarker of disease progression in chronic inflammatory demyelinating polyneuropathy. Muscle and Nerve, 2022, 65, 433-439.	2.2	4
92	A novel heteroplasmic mutation in mitochondrial tRNAArg gene associated with non-dystrophic myotonias. Acta Neurologica Belgica, 2020, 120, 573-580.	1.1	3
93	Application of muscle ultrasound for the evaluation of patients with amyotrophic lateral sclerosis: An observational crossâ€sectional study. Muscle and Nerve, 2020, 62, 516-521.	2.2	3
94	BVVL/ FL: features caused by SLC52A3 mutations; WDFY4 and TNFSF13B may be novel causative genes. Neurobiology of Aging, 2021, 99, 102.e1-102.e10.	3.1	3
95	Application of CMAP scan for the evaluation of patients with chronic inflammatory demyelinating polyneuropathy: a prospective study. Neurophysiologie Clinique, 2021, 51, 175-181.	2.2	3
96	Disease severity and response to treatment in Iranian patients with myasthenia gravis. Neurological Sciences, 2022, 43, 1233-1237.	1.9	3
97	Retrospective analysis of response to rituximab in chronic inflammatory demyelinating polyneuropathy refractory to firstâ€line therapy. Journal of the Peripheral Nervous System, 2021, 26, 469-474.	3.1	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. Iranian Journal of Neurology, 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. Iranian Journal of Neurology, 2015, 14, 190-4.	0.5	3
100	Mitochondrial Copy Number and D-Loop Variants in Pompe Patients. Cell Journal, 2016, 18, 405-15.	0.2	3
101	Delayedâ€onset inflammatory polyneuropathy without graft versus host disease after bone marrow transplantation. Muscle and Nerve, 2018, 58, E11-E13.	2.2	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. Journal of Neurology, 2021, 268, 640-650.	3.6	2
103	Novel Mutations of the TYMP Gene in Mitochondrial Neurogastrointestinal Encephalomyopathy: Case Series and Literature Review. Journal of Molecular Neuroscience, 2021, 71, 2526-2533.	2.3	2
104	The correlation of the serum level of L-carnitine with disease severity in patients with Amyotrophic lateral sclerosis. Journal of Clinical Neuroscience, 2021, 89, 232-236.	1.5	2
105	An Iranian familial amyotrophic lateral sclerosis pedigree with p.Val48Phe causing mutation in SOD1: a genetic and clinical report. Iranian Journal of Basic Medical Sciences, 2014, 17, 735-9.	1.0	2
106	An overview of motor unit number index reproducibility in amyotrophic lateral sclerosis. Iranian Journal of Neurology, 2019, 18, 119-126.	0.5	2
107	Current Concepts in Botulism. Journal of Clinical Neuromuscular Disease, 2003, 4, 139-149.	0.7	1
108	A case of adult onset Sandhoff disease that mimics Brown-Vialetto-Van Laere syndrome. Neuromuscular Disorders, 2021, 31, 528-531.	0.6	1

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109	Association of HLA Class II Alleles with Disease Severity and Treatment Response in Iranian Patients with Myasthenia Gravis. Journal of Neuromuscular Diseases, 2021, 8, 827-829.	2.6	1
110	Evaluation of Quality of Life in Patients With Chronic Inflammatory Demyelinating Polyneuropathy in Iran. Journal of Clinical Neuromuscular Disease, 2019, 21, 77-83.	0.7	1
111	An overview of motor unit number index reproducibility in amyotrophic lateral sclerosis. Current Journal of Neurology, 0, , .	0.0	1
112	Clinical and Pathological Features of Lipid Storage Myopathy; A Retrospective Study of a Large Group from Iran. International Clinical Neuroscience Journal, 2020, 8, 26-29.	0.1	1
113	Four cases of brown-vialetto-van laere syndrome from Iran: Clinical and electrophysiologic findings. Iranian Journal of Neurology, 2011, 10, 54-7.	0.5	1
114	Iranian Consensus Recommendations for Treatment of Myasthenia Gravis. Archives of Iranian Medicine, 2022, 25, 37-49.	0.6	1
115	Practical needs and considerations for refugees and other forcibly displaced persons with neurological disorders: Recommendations using a modified Delphi approach. Gates Open Research, 2021, 5, 178.	1.1	1
116	De novo Mutation in CACNA1S Gene in a 20-Year-Old Man Diagnosed with Metabolic Myopathy. Archives of Iranian Medicine, 2017, 20, 617-620.	0.6	1
117	The role of opening CSF pressure in response to treatment for idiopathic intracranial hypertension (IIH). Journal of Clinical Neuroscience, 2020, 76, 171-176.	1.5	0
118	Validation of Persian Individualized Neuromuscular Quality of Life in patients with muscular dystrophies. Current Journal of Neurology, 0, , .	0.0	0
119	RABENOSYN separation-of-function mutations uncouple endosomal recycling from lysosomal degradation, causing a distinct Mendelian Disorder. Human Molecular Genetics, 0, , .	2.9	0