# Shahriar Nafissi

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/9461722/publications.pdf

Version: 2024-02-01

| | | | |
|---|---|---|---|
| 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 | Disease severity and response to treatment in Iranian patients with myasthenia gravis. Neurological Sciences, 2022, 43, 1233-1237. | 1.9 | 3 |
| 2 | 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 |
| 3 | Clinical and genetic spectrum of a large cohort of patients with Î´-sarcoglycan muscular dystrophy. Brain, 2022, 145, 596-606. | 7.6 | 11 |
| 4 | 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 |
| 5 | Iranian Consensus Recommendations for Treatment of Myasthenia Gravis. Archives of Iranian Medicine, 2022, 25, 37-49. | 0.6 | 1 |
| 6 | 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 |
| 7 | Sporadic late-onset nemaline myopathy with monoclonal gammopathy of undetermined significance: Report of four patients. Neuromuscular Disorders, 2021, 31, 29-34. | 0.6 | 6 |
| 8 | 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 |
| 9 | Persian adaptation of Edinburgh Cognitive and Behavioural Screen (ECAS). Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2021, 22, 426-433. | 1.7 | 8 |
| 10 | Investigating the possible association between <scp>NLRP3</scp> gene polymorphisms and myasthenia gravis. Muscle and Nerve, 2021, 63, 730-736. | 2.2 | 7 |
| 11 | 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 |
| 12 | 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 |
| 13 | 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 |
| 14 | A case of adult onset Sandhoff disease that mimics Brown-Vialetto-Van Laere syndrome. Neuromuscular Disorders, 2021, 31, 528-531. | 0.6 | 1 |
| 15 | Thigh and Leg Muscle MRI Findings in GNE Myopathy. Journal of Neuromuscular Diseases, 2021, 8, 735-742. | 2.6 | 5 |
| 16 | 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 |
| 17 | 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 |
| 18 | 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 |

| # | Article | IF | Citations |
|---|---|---|---|
| 19 | 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 |
| 20 | Myasthenia gravis associated with novel coronavirus 2019 infection: A report of three cases. Clinical Neurology and Neurosurgery, 2021, 208, 106834. | 1.4 | 21 |
| 21 | Recommendations for Infantile-Onset and Late-Onset Pompe Disease: An Iranian Consensus. Frontiers in Neurology, 2021, 12, 739931. | 2.4 | 10 |
| 22 | Guillain-Barre Syndrome and COVID-19 Vaccine: A Report of Nine Patients. Basic and Clinical Neuroscience, 2021, 12, 703-710. | 0.6 | 24 |
| 23 | 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 |
| 24 | A novel heteroplasmic mutation in mitochondrial tRNAArg gene associated with non-dystrophic myotonias. Acta Neurologica Belgica, 2020, 120, 573-580. | 1.1 | 3 |
| 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 | 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 |
| 27 | Description of combined ARHSP/JALS phenotype in some patients with SPG11 mutations. Molecular Genetics & Genomic Medicine, 2020, 8, e1240. | 1.2 | 7 |
| 28 | 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 |
| 29 | Sequential targeted exome sequencing of 1001 patients affected by unexplained limb-girdle weakness. Genetics in Medicine, 2020, 22, 1478-1488. | 2.4 | 62 |
| 30 | 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 |
| 31 | 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 |
| 32 | 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 |
| 33 | POPDC3 Gene Variants Associate with a New Form of Limb Girdle Muscular Dystrophy. Annals of Neurology, 2019, 86, 832-843. | 5.3 | 27 |
| 34 | 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 |
| 35 | Mutation screening of SLC52A3, C19orf12, and TARDBP in Iranian ALS patients. Neurobiology of Aging, 2019, 75, 225.e9-225.e14. | 3.1 | 7 |
| 36 | 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 |

| # | Article | IF | Citations |
|---|---|---|---|
| 37 | 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 |
| 38 | An overview of motor unit number index reproducibility in amyotrophic lateral sclerosis. Iranian Journal of Neurology, 2019, 18, 119-126. | 0.5 | 2 |
| 39 | Delayedâ€onset inflammatory polyneuropathy without graft versus host disease after bone marrow transplantation. Muscle and Nerve, 2018, 58, E11-E13. | 2.2 | 2 |
| 40 | 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 |
| 41 | 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 |
| 42 | Genotype and phenotype analysis of 43 Iranian facioscapulohumeral muscular dystrophy patients; Evidence for anticipation. Neuromuscular Disorders, 2018, 28, 303-314. | 0.6 | 6 |
| 43 | 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 |
| 44 | Limb girdle muscular dystrophy due to mutations in POMT2. Journal of Neurology, Neurosurgery and Psychiatry, 2018, 89, 506-512. | 1.9 | 21 |
| 45 | 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 |
| 46 | 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 |
| 47 | 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 |
| 48 | 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 |
| 49 | LGMD2E is the most common type of sarcoglycanopathies in the Iranian population. Journal of Neurogenetics, 2017, 31, 161-169. | 1.4 | 18 |
| 50 | Lateâ€onset pompe disease in Iran: A clinical and genetic report. Muscle and Nerve, 2017, 55, 835-840. | 2.2 | 6 |
| 51 | 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 |
| 52 | Mutation inADORA1identified as likely cause of early-onset parkinsonism and cognitive dysfunction. Movement Disorders, 2016, 31, 1004-1011. | 3.9 | 38 |
| 53 | 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 |
| 54 | 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 |

| # | Article | IF | Citations |
|---|---|---|---|
| 55 | Iranian consensus on use of vitamin D in patients with multiple sclerosis. BMC Neurology, 2016, 16, 76. | 1.8 | 7 |
| 56 | 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 |
| 57 | Validation of the 15â€item myasthenia gravis quality of life questionnaire (MGâ€QOL15) Persian version. Muscle and Nerve, 2016, 54, 65-70. | 2.2 | 12 |
| 58 | Trends of quality of life changes in amyotrophic lateral sclerosis patients. Journal of the Neurological Sciences, 2016, 368, 35-40. | 0.6 | 17 |
| 59 | Genetics of GNE myopathy in the non-Jewish Persian population. European Journal of Human Genetics, 2016, 24, 243-251. | 2.8 | 18 |
| 60 | 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 |
| 61 | 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 |
| 62 | Personality characteristics in MS patients: The role of avoidant personality. Clinical Neurology and Neurosurgery, 2016, 144, 23-27. | 1.4 | 11 |
| 63 | 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 |
| 64 | Mitochondrial Copy Number and D-Loop Variants in Pompe Patients. Cell Journal, 2016, 18, 405-15. | 0.2 | 3 |
| 65 | 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 |
| 66 | Blink Reflex May Help Discriminate Alzheimer Disease From Vascular Dementia. Journal of Clinical Neurophysiology, 2015, 32, 505-511. | 1.7 | 4 |
| 67 | 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 |
| 68 | 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 |
| 69 | Granulocyte Colony-Stimulating Factor for Amyotrophic Lateral Sclerosis: A Randomized, | | |

| # | Article | IF | Citations |
|---|---|---|---|
| 73 | Dysferlinopathy in Iran: Clinical and genetic report. Journal of the Neurological Sciences, 2015, 359, 256-259. | 0.6 | 12 |
| 74 | 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 |
| 75 | 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 |
| 76 | 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 |
| 77 | Impact of Vitamin A Supplementation on Disease Progression in Patients with Multiple Sclerosis. Archives of Iranian Medicine, 2015, 18, 435-40. | 0.6 | 25 |
| 78 | Gene Expression Profiling of Mitochondrial Oxidative Phosphorylation (OXPHOS) Complex I in Friedreich Ataxia (FRDA) Patients. PLoS ONE, 2014, 9, e94069. | 2.5 | 32 |
| 79 | 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 |
| 80 | Clinical and electrophysiological features of Guillainâ€“BarrÃ© syndrome in Iran. Journal of Clinical Neuroscience, 2014, 21, 1554-1557. | 1.5 | 4 |
| 81 | 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 |
| 82 | Muscle-specific kinase antibody associated myasthenia gravis after bone marrow transplantation. Neuromuscular Disorders, 2014, 24, 148-150. | 0.6 | 8 |
| 83 | 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 |
| 84 | Islamic fasting and multiple sclerosis. BMC Neurology, 2014, 14, 56. | 1.8 | 38 |
| 85 | Dietary intake of nutrients and its correlation with fatigue in multiple sclerosis patients. Iranian Journal of Neurology, 2014, 13, 28-32. | 0.5 | 22 |
| 86 | Comparison of electrophysiological findings in axonal and demyelinating Guillain-Barre syndrome. Iranian Journal of Neurology, 2014, 13, 138-43. | 0.5 | 15 |
| 87 | 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 |
| 88 | Impact of Vitamin A Supplementation on RAR Gene Expression in Multiple Sclerosis Patients. Journal of Molecular Neuroscience, 2013, 51, 478-484. | 2.3 | 12 |
| 89 | 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 |
| 90 | Genetic analysis and SOD1 mutation screening in Iranian amyotrophic lateral sclerosis patients. Neurobiology of Aging, 2013, 34, 1516.e1-1516.e8. | 3.1 | 34 |

| # | Article | IF | Citations |
|---|---|---|---|
| 91 | 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 |
| 92 | The role of cytomegalovirus, Haemophilus influenzae and Epstein Barr virus in Guillain Barre syndrome. Acta Medica Iranica, 2013, 51, 372-6. | 0.8 | 8 |
| 93 | Mitochondrial complex I deficiency and ATP/ADP ratio in lymphocytes of amyotrophic lateral sclerosis patients. Neurological Research, 2012, 34, 297-303. | 1.3 | 54 |
| 94 | Long-term follow-up of patients with congenital myasthenic syndrome caused by COLQ mutations. Neuromuscular Disorders, 2012, 22, 318-324. | 0.6 | 64 |
| 95 | 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 |
| 96 | Four novel C20orf54 mutations identified in Brownâ€“Vialettoâ€“Van Laere syndrome patients. Journal of Human Genetics, 2012, 57, 613-617. | 2.3 | 26 |
| 97 | 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 |
| 98 | Congenital myasthenic syndrome with tubular aggregates caused by GFPT1 mutations. Journal of Neurology, 2012, 259, 838-850. | 3.6 | 72 |
| 99 | 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 |
| 100 | Electrophysiological evaluation in lumbosacral radiculopathy. Iranian Journal of Neurology, 2012, 11, 83-6. | 0.5 | 8 |
| 101 | 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 |
| 102 | Hexosamine Biosynthetic Pathway Mutations Cause Neuromuscular Transmission Defect. American Journal of Human Genetics, 2011, 88, 162-172. | 6.2 | 153 |
| 103 | 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 |
| 104 | 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 |
| 105 | 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 |
| 106 | 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 |
| 107 | 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 |
| 108 | A Novel Mitochondrial Heteroplasmic C13806A Point Mutation Associated with Iranian Friedreichâ€™s Ataxia. Cellular and Molecular Neurobiology, 2009, 29, 225-233. | 3.3 | 13 |

| # | Article | IF | Citations |
|---|---|---|---|
| 109 | Complex I and ATP Content Deficiency in Lymphocytes from Friedreich's Ataxia. Canadian Journal of Neurological Sciences, 2009, 36, 26-31. | 0.5 | 39 |
| 110 | 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 |
| 111 | Effect of skin thickness on sensory nerve action potential amplitude. Clinical Neurophysiology, 2008, 119, 1824-1828. | 1.5 | 25 |
| 112 | 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 |
| 113 | Wilsonâ€™s Disease: A Great Masquerader. European Neurology, 2007, 57, 80-85. | 1.4 | 50 |
| 114 | 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 |
| 115 | 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 |
| 116 | Current Concepts in Botulism. Journal of Clinical Neuromuscular Disease, 2003, 4, 139-149. | 0.7 | 1 |
| 117 | An overview of motor unit number index reproducibility in amyotrophic lateral sclerosis. Current Journal of Neurology, 0, , . | 0.0 | 1 |
| 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 |