Hossein Darvish

List of Publications by Year
in descending order

75 papers	1,290 citations	430874 18 h-index	414414 32 g-index
75 all docs	75 docs citations	75 times ranked	2307 citing authors

#	Article	IF	Citations
1	Unraveling the genetic complexities of combined retinal dystrophy and hearing impairment. Human Genetics, 2022, 141, 785-803.	3.8	6
2	The phenotypic spectrum of PCDH12 associated disorders - Five new cases and review of the literature. European Journal of Paediatric Neurology, 2022, 36, 7-13.	1.6	4
3	Leu226Trp CACNA1A variant associated with juvenile myoclonic epilepsy with and without intellectual disability. Clinical Neurology and Neurosurgery, 2022, 213, 107108.	1.4	2
4	Bi-allelic variants inCHKAcause a neurodevelopmental disorder with epilepsy and microcephaly. Brain, 2022, 145, 1916-1923.	7.6	3
5	A novel splice site mutation in the SDCCAG8 gene in an Iranian family with Bardetâ€“Biedl syndrome. International Ophthalmology, 2021, 41, 389-397.	1.4	4
6	Genotypeâ€“phenotype associations in hereditary spastic paraplegia: a systematic review and meta-analysis on 13,570 patients. Journal of Neurology, 2021, 268, 2065-2082.	3.6	60
7	Recessive COL4A2 Mutation Leads to Intellectual Disability, Epilepsy, and Spastic Cerebral Palsy. Neurology: Genetics, 2021, 7, e583.	1.9	3
8	Three functional variants in the NLRP3 gene are associated with susceptibility and clinical characteristics of systemic lupus erythematosus. Lupus, 2021, 30, 1273-1282.	1.6	16
9	Mutational analysis of CYP1B1 gene in Iranian pedigrees with glaucoma reveals known and novel mutations. International Ophthalmology, 2021, 41, 3269-3276.	1.4	5
10	Biallelic variants in HPDL cause pure and complicated hereditary spastic paraplegia. Brain, 2021, 144, 1422-1434.	7.6	22
11	<scp>ANXA1</scp> with Antiâ€Inflammatory Properties Might Contribute to Parkinsonism. Annals of Neurology, 2021, 90, 319-323.	5.3	7
12	Biallelic loss-of-function variants in the splicing regulator NSRP1 cause a severe neurodevelopmental disorder with spastic cerebral palsy and epilepsy. Genetics in Medicine, 2021, 23, 2455-2460.	2.4	9
13	PRDM12 Is Transcriptionally Active and Required for Nociceptor Function Throughout Life. Frontiers in Molecular Neuroscience, 2021, 14, 720973.	2.9	7
14	Clinical and molecular spectrum of P/Q type calcium channel Cav2.1 in epileptic patients. Orphanet Journal of Rare Diseases, 2021, 16, 461.	2.7	7
15	The Phenotypic Spectrum of PCDH12-Associated Disorders: Five New Cases and Review of the Literature. , 2021, 52, .		0
16	Biallelic truncation variants in ATP9A are associated with a novel autosomal recessive neurodevelopmental disorder. Npj Genomic Medicine, 2021, 6, 94.	3.8	10
17	Defining the clinical, molecular and imaging spectrum of adaptor protein complex 4-associated hereditary spastic paraplegia. Brain, 2020, 143, 2929-2944.	7.6	29
18	Bi-allelic HPDL Variants Cause a Neurodegenerative Disease Ranging from Neonatal Encephalopathy to Adolescent-Onset Spastic Paraplegia. American Journal of Human Genetics, 2020, 107, 364-373.	6.2	30

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19	The association of CNTNAP2 rs2710102 and ENGRAILED-2 rs1861972 genes polymorphism and autism in Iranian population. Meta Gene, 2020, 24, 100664.	0.6	5
20	Phenotypic and genotypic characterization of families with complex intellectual disability identified pathogenic genetic variations in known and novel disease genes. Scientific Reports, 2020, 10, 968.	3.3	8
21	Incomplete penetrance of MITF gene c.943C>T mutation in an extended family with Waardenburg syndrome type II. International Journal of Pediatric Otorhinolaryngology, 2020, 135, 110014.	1.0	6
22	Whole-exome sequencing identified a novel mutation of MLH1 in an extended family with lynch syndrome. Genes and Diseases, 2020, 7, 614-619.	3.4	2
23	Novel ABCD1 gene mutations in Iranian pedigrees with X-linked adrenoleukodystrophy. Journal of Pediatric Endocrinology and Metabolism, 2019, 32, 1207-1215.	0.9	0
24	Bi-allelic variants in RNF170 are associated with hereditary spastic paraplegia. Nature Communications, 2019, 10, 4790.	12.8	39
25	Genetic analysis of rs11038167, rs11038172 and rs835784 polymorphisms of the TSPAN18 gene in Iranian schizophrenia patients. Meta Gene, 2019, 22, 100609.	0.6	2
26	Incomplete penetrance of CRX gene for autosomal dominant form of cone-rod dystrophy. Ophthalmic Genetics, 2019, 40, 259-266.	1.2	11
27	Expression analysis and genotyping of DGKZ: a GWAS-derived risk gene for schizophrenia. Molecular Biology Reports, 2019, 46, 4105-4111.	2.3	7
28	A novel PUS7 mutation causes intellectual disability with autistic and aggressive behaviors. Neurology: Genetics, 2019, 5, e356.	1.9	18
29	Homozygous Mutation in TWNK Cases Ataxia, Sensorineural Hearing Loss and Optic Nerve Atrophy. Archives of Iranian Medicine, 2019, 22, 728-730.	0.6	3
30	hOGG1 gene polymorphism and breast cancer risk: A systematic review and meta-analysis study. Breast Journal, 2018, 24, 70-73.	1.0	3
31	A Clinical and Molecular Genetic Study of 50 Families with Autosomal Recessive Parkinsonism Revealed Known and Novel Gene Mutations. Molecular Neurobiology, 2018, 55, 3477-3489.	4.0	67
32	Genetic screening in two Iranian families with early-onset Alzheimer's disease identified a novel PSEN1 mutation. Neurobiology of Aging, 2018, 62, 244.e15-244.e17.	3.1	9
33	A novel c.240_241insGG mutation in NDP gene in a family with Norrie disease. Australasian journal of optometry, The, 2018, 101, 255-259.	1.3	4
34	Identification of a large homozygous VPS13C deletion in a patient with earlyâ€onset Parkinsonism. Movement Disorders, 2018, 33, 1968-1970.	3.9	38
35	Molecular characterization of PRKN structural variations identified through wholeâ€genome sequencing. Molecular Genetics & Genomic Medicine, 2018, 6, 1243-1248.	1.2	6
36	RIT2: responsible and susceptible gene for neurological and psychiatric disorders. Molecular Genetics and Genomics, 2018, 293, 785-792.	2.1	12

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37	COL18A1 is a candidate eye iridocorneal angle-closure gene in humans. Human Molecular Genetics, 2018, 27, 3772-3786.	2.9	30
38	RIT2 Polymorphisms: Is There a Differential Association?. Molecular Neurobiology, 2017, 54, 2234-2240.	4.0	31
39	The human RIT2 core promoter short tandem repeat predominant allele is species-specific in length: a selective advantage for human evolution?. Molecular Genetics and Genomics, 2017, 292, 611-617.	2.1	18
40	Association of Î²-Secretase Functional Polymorphism with Risk of Schizophrenia. Genetic Testing and Molecular Biomarkers, 2017, 21, 248-251.	0.7	3
41	RAB7L1 promoter polymorphism and risk of Parkinsonâ€™s disease; a case-control study. Neurological Research, 2017, 39, 468-471.	1.3	12
42	Genetic analysis of SNCA gene polymorphisms in Parkinsonâ€™s disease in an Iranian population. Basal Ganglia, 2017, 10, 4-7.	0.3	2
43	A novel mutation in SMOC1 and variable phenotypic expression in two patients with Waardenburg anophthalmia syndrome. European Journal of Medical Genetics, 2017, 60, 578-582.	1.3	8
44	Whole genome sequencing identifies a novel homozygous exon deletion in the NT5C2 gene in a family with intellectual disability and spastic paraplegia. Npj Genomic Medicine, 2017, 2, .	3.8	10
45	PTRHD1(C2orf79) mutations lead to autosomal-recessive intellectual disability and parkinsonism. Movement Disorders, 2017, 32, 287-291.	3.9	38
46	A genetic variant in miRNA binding site of glutamate receptor 4, metabotropic (GRM4) is associated with increased risk of major depressive disorder. Journal of Affective Disorders, 2017, 208, 218-222.	4.1	25
47	Copy Number Variants in Patients with Autism and Additional Clinical Features: Report of VIPR2 Duplication and a Novel Microduplication Syndrome. Molecular Neurobiology, 2017, 54, 7019-7027.	4.0	20
48	Bioinformatic tools to determine the pathogenicity of a missense mutation in PKHD1 in autosomal recessive polycystic kidney disease. Nephrology, 2017, 22, 330-331.	1.6	0
49	Screening for intermediate CGG alleles of FMR1 gene in male Iranian patients with Parkinsonism. Neurological Sciences, 2017, 38, 123-128.	1.9	4
50	Novel Mutations in Gene in Families with Gelatinous Drop-like Corneal Dystrophy (GDLD). International Journal of Molecular and Cellular Medicine, 2017, 6, 204-211.	1.1	1
51	Glutamate receptor, metabotropic 7 (GRM7) gene variations and susceptibility to autism: A caseâ€“control study. Autism Research, 2016, 9, 1161-1168.	3.8	57
52	Vitamin D receptor gene rs4334089 polymorphism and Parkinsonâ€™s disease in Iranian population. Basal Ganglia, 2016, 6, 157-160.	0.3	2
53	Genetic Analysis of the ZNF512B, SLC41A1, and ALDH2 Polymorphisms in Parkinson's Disease in the Iranian Population. Genetic Testing and Molecular Biomarkers, 2016, 20, 629-632.	0.7	18
54	SIPA1L2 , MIR4697 , GCH1 and VPS13C loci and risk of Parkinson's diseases in Iranian population: A case-control study. Journal of the Neurological Sciences, 2016, 369, 1-4.	0.6	15

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55	SNAP-25gene variations and attention-deficit hyperactivity disorder in Iranian population. Neurological Research, 2016, 38, 959-964.	1.3	1
56	Identification of a LargeDNAJB2Deletion in a Family with Spinal Muscular Atrophy and Parkinsonism. Human Mutation, 2016, 37, 1180-1189.	2.5	36
57	c.376G>A mutation in WFS1 gene causes Wolfram syndrome without deafness. European Journal of Medical Genetics, 2016, 59, 65-69.	1.3	7
58	The analysis of association between SNCA, HUSEYO and CSMD1 gene variants and Parkinsonâ€™s disease in Iranian population. Neurological Sciences, 2016, 37, 731-736.	1.9	20
59	A genetic variant in CAMKK2 gene is possibly associated with increased risk of bipolar disorder. Journal of Neural Transmission, 2016, 123, 323-328.	2.8	13
60	XRCC1 and OGG1 Gene Polymorphisms and Breast Cancer: A Systematic Review of Literature. Iranian Journal of Cancer Prevention, 2016, 9, e3467.	0.7	9
61	Analysis of CYP17, CYP19 and CYP1A1 Gene Polymorphisms in Iranian Women with Breast Cancer. Asian Pacific Journal of Cancer Prevention, 2016, 17, 23-26.	1.2	12
62	A Novel PKD1 Mutation in a Patient with Autosomal Dominant Polycystic Kidney Disease. International Journal of Molecular and Cellular Medicine, 2016, 5, 123-4.	1.1	3
63	Emery-Dreifuss Muscular Dystrophy: a Report of a Large Family with 11 Affected Individuals. International Journal of Molecular and Cellular Medicine, 2016, 5, 196-198.	1.1	3
64	Analysis of Copy Number Variations in Patients with Autism Using Cytogenetic and MLPA Techniques: Report of 16p13.1p13.3 and 10q26.3 Duplications. International Journal of Molecular and Cellular Medicine, 2016, 5, 236-245.	1.1	7
65	Association analysis of DISC1 gene polymorphisms with Attention-deficit/hyperactivity disorder in Iranian population. Pakistan Journal of Medical Sciences, 2015, 31, 1162-6.	0.6	2
66	Variation in the miRNA-433 binding site of FGF20 is a risk factor for Parkinson's disease in Iranian population. Journal of the Neurological Sciences, 2015, 355, 72-74.	0.6	25
67	Mutations in the histamineN-methyltransferase gene,HNMT, are associated with nonsyndromic autosomal recessive intellectual disability. Human Molecular Genetics, 2015, 24, 5697-5710.	2.9	27
68	Monoamine Oxidase AGene polymorphisms and Bipolar Disorder in Iranian Population. Iranian Red Crescent Medical Journal, 2015, 17, e23095.	0.5	5
69	No Evidence for Association Between Norepinephrine Transporter-3081 (A/T) Polymorphism and Attention Deficit Hyperactivity Disorder in Iranian Population. Iranian Red Crescent Medical Journal, 2015, 17, e22996.	0.5	1
70	RIT2 , a susceptibility gene for Parkinson's disease in Iranian population. Neurobiology of Aging, 2014, 35, e27-e28.	3.1	23
71	Variable phenotypic expression in families with early-onset Parkinsonism due to PRKN mutations. Journal of Neurology, 2014, 261, 1223-1226.	3.6	3
72	FABP9 Mutations Are Not Detected in Cases of Infertility due to Sperm Morphological Defects in Iranian Men. International Journal of Fertility & Sterility, 2014, 7, 275-80.	0.2	1

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73	The Sac1 Domain of <scp>SYNJ</scp> 1 Identified Mutated in a Family with Earlyâ€Onset Progressive <scp>P</scp> arkinsonism with Generalized Seizures. Human Mutation, 2013, 34, 1200-1207.	2.5	302
74	Detection of copy number changes in genes associated with Parkinson's disease in Iranian patients. Neuroscience Letters, 2013, 551, 75-78.	2.1	29
75	Identification of Chromosome Abnormalities in Subtelomeric Regions Using Multiplex Ligation Dependent Probe Amplification (MLPA) Technique in 100 Iranian Patients With Idiopathic Mental Retardation. Iranian Red Crescent Medical Journal, 2013, 15, e8221.	0.5	3