Kimia Kahrizi

List of Publications by Year
in descending order

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87 papers	2,929 citations	186265 28 h-index	182427 51 g-index
91 all docs	91 docs citations	91 times ranked	5956 citing authors

#	Article	IF	Citations
1	SARSâ€CoVâ€2 outbreak in Iran: The dynamics of the epidemic and evidence on two independent introductions. Transboundary and Emerging Diseases, 2022, 69, 1375-1386.	3.0	19
2	Genetic etiology of hearing loss in Iran. Human Genetics, 2022, 141, 623-631.	3.8	6
3	ZBTB11 dysfunction: spectrum of brain abnormalities, biochemical signature and cellular consequences. Brain, 2022, 145, 2602-2616.	7.6	5
4	Phenotype and genotype spectrum of variants in guanine nucleotide exchange factor genes in a broad cohort of Iranian patients. Molecular Genetics & Genomic Medicine, 2022, 10, e1894.	1.2	5
5	Intellectual disability associated with craniofacial dysmorphism due to POLR3B mutation and defect in spliceosomal machinery. BMC Medical Genomics, 2022, 15, 89.	1.5	1
6	Comprehensive <scp>genotypeâ€phenotype</scp> correlation in <scp>AP</scp>â€4 deficiency syndrome; Adding data from a large cohort of Iranian patients. Clinical Genetics, 2021, 99, 187-192.	2.0	2
7	POLRMT mutations impair mitochondrial transcription causing neurological disease. Nature Communications, 2021, 12, 1135.	12.8	21
8	Exome sequencing utility in defining the genetic landscape of hearing loss and novelâ€gene discovery in Iran. Clinical Genetics, 2021, 100, 59-78.	2.0	4
9	CEP104 and CEP290; Genes with Ciliary Functions Cause Intellectual Disability in Multiple Families. Archives of Iranian Medicine, 2021, 24, 364-373.	0.6	3
10	The recurrent missense mutation p.(Arg367Trp) in YARS1 causes a distinct neurodevelopmental phenotype. Journal of Molecular Medicine, 2021, 99, 1755-1768.	3.9	3
11	The PTRHD1 Mutation in Intellectual Disability. Archives of Iranian Medicine, 2021, 24, 747-751.	0.6	3
12	A novel variant of C12orf4 linked to autosomal recessive intellectual disability type 66 with phenotype expansion. Journal of Gene Medicine, 2021, , e3406.	2.8	0
13	YIF1B mutations cause a post-natal neurodevelopmental syndrome associated with Golgi and primary cilium alterations. Brain, 2020, 143, 2911-2928.	7.6	13
14	Whole genome sequencing identifies a duplicated region encompassing Xq13.2q13.3 in a large Iranian family with intellectual disability. Molecular Genetics & Genomic Medicine, 2020, 8, e1418.	1.2	1
15	When transcripts matter: delineating between non-syndromic hearing loss DFNB32 and hearing impairment infertile male syndrome (HIIMS). Journal of Human Genetics, 2020, 65, 609-617.	2.3	2
16	A novel PTC mutation in the BTB domain of KLHL7 gene in two patients with Bohring-Opitz syndrome-like features. European Journal of Medical Genetics, 2020, 63, 103849.	1.3	3
17	Limbic System Associated Membrane Protein Mutation in an Iranian Family Diagnosed with MÃ©niÃ¨reâ€™s Disease. Archives of Iranian Medicine, 2020, 23, 319-325.	0.6	6
18	Novel Mutation in LARP7 in Two Iranian Consanguineous Families with Syndromic Intellectual Disability and Facial Dysmorphism. Archives of Iranian Medicine, 2020, 23, 842-847.	0.6	2

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19	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
20	GPR126: A novel candidate gene implicated in autosomal recessive intellectual disability. American Journal of Medical Genetics, Part A, 2019, 179, 13-19.	1.2	12
21	G130V de novo mutation in an Iranian pedigree with nonsyndromic hearing loss without palmoplantar keratoderma. International Journal of Pediatric Otorhinolaryngology, 2019, 126, 109607.	1.0	1
22	Iranome: A catalog of genomic variations in the Iranian population. Human Mutation, 2019, 40, 1968-1984.	2.5	116
23	Bi-allelic Pathogenic Variants in TUBGCP2 Cause Microcephaly and Lissencephaly Spectrum Disorders. American Journal of Human Genetics, 2019, 105, 1005-1015.	6.2	24
24	Distinct genetic variation and heterogeneity of the Iranian population. PLoS Genetics, 2019, 15, e1008385.	3.5	34
25	Homozygous variants in the gene SCAPER cause syndromic intellectual disability. American Journal of Medical Genetics, Part A, 2019, 179, 1214-1225.	1.2	10
26	Identification of diseaseâ€causing variants in the EXOSC gene family underlying autosomal recessive intellectual disability in Iranian families. Clinical Genetics, 2019, 95, 718-725.	2.0	5
27	Whole-Transcriptome Analysis Reveals Dysregulation of Actin-Cytoskeleton Pathway in Intellectual Disability Patients. Neuroscience, 2019, 404, 423-444.	2.3	4
28	A splice-altering variant in LARP7 gene leads to exon exclusion. Gene Reports, 2019, 15, 100375.	0.8	1
29	Effect of inbreeding on intellectual disability revisited by trio sequencing. Clinical Genetics, 2019, 95, 151-159.	2.0	49
30	Novel mutations in MYTH4-FERM domains of myosin 15 are associated with autosomal recessive nonsyndromic hearing loss. International Journal of Pediatric Otorhinolaryngology, 2019, 117, 115-126.	1.0	9
31	Genetics of intellectual disability in consanguineous families. Molecular Psychiatry, 2019, 24, 1027-1039.	7.9	131
32	Novel Mutations in KCNQ4, LHFPL5 and COCH Genes in Iranian Families with Hearing Impairment. Archives of Iranian Medicine, 2019, 22, 189-197.	0.6	9
33	Contribution of Iran in Elucidating the Genetic Causes of Autosomal Recessive Intellectual Disability. Archives of Iranian Medicine, 2019, 22, 461-471.	0.6	0
34	Exonic mutations and exon skipping: Lessons learned from DFNA5. Human Mutation, 2018, 39, 433-440.	2.5	44
35	Genotype and phenotype analysis of 43 Iranian facioscapulohumeral muscular dystrophy patients; Evidence for anticipation. Neuromuscular Disorders, 2018, 28, 303-314.	0.6	6
36	SLC52A2 mutations cause SCABD2 phenotype: A second report. International Journal of Pediatric Otorhinolaryngology, 2018, 104, 195-199.	1.0	9

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37	CDC14A phosphatase is essential for hearing and male fertility in mouse and human. Human Molecular Genetics, 2018, 27, 780-798.	2.9	49
38	Old gene, new phenotype: splice-altering variants in CEACAM16 cause recessive non-syndromic hearing impairment. Journal of Medical Genetics, 2018, 55, 555-560.	3.2	48
39	De novo and inherited mutations in the X-linked gene CLCN4 are associated with syndromic intellectual disability and behavior and seizure disorders in males and females. Molecular Psychiatry, 2018, 23, 222-230.	7.9	45
40	Variants in CIB2 cause DFNB48 and not USH1J. Clinical Genetics, 2018, 93, 812-821.	2.0	46
41	CNKSR1 gene defect can cause syndromic autosomal recessive intellectual disability. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2018, 177, 691-699.	1.7	4
42	The power of the Mediator complex-Expanding the genetic architecture and phenotypic spectrum of MED12 -related disorders. Clinical Genetics, 2018, 94, 450-456.	2.0	24
43	Biallelic missense variants in ZBTB11 can cause intellectual disability in humans. Human Molecular Genetics, 2018, 27, 3177-3188.	2.9	19
44	Intellectual Disability and Ataxia: Genetic Collisions. Archives of Iranian Medicine, 2018, 21, 29-40.	0.6	3
45	Calpains: Diverse Functions but Enigmatic. Archives of Iranian Medicine, 2018, 21, 170-179.	0.6	4
46	tRNA Methyltransferase Defects and Intellectual Disability. Archives of Iranian Medicine, 2018, 21, 478-485.	0.6	5
47	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
48	Mutations of the aminoacyl-tRNA-synthetases SARS and WARS2 are implicated in the etiology of autosomal recessive intellectual disability. Human Mutation, 2017, 38, 621-636.	2.5	54
49	Homozygous Truncating Variants in TBC1D23 Cause Pontocerebellar Hypoplasia and Alter Cortical Development. American Journal of Human Genetics, 2017, 101, 428-440.	6.2	39
50	CIB2, defective in isolated deafness, is key for auditory hair cell mechanotransduction and survival. EMBO Molecular Medicine, 2017, 9, 1711-1731.	6.9	66
51	LGMD2E is the most common type of sarcoglycanopathies in the Iranian population. Journal of Neurogenetics, 2017, 31, 161-169.	1.4	18
52	Adenosine kinase deficiency: expanding the clinical spectrum and evaluating therapeutic options. Journal of Inherited Metabolic Disease, 2016, 39, 273-283.	3.6	55
53	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
54	Missense variants in AIMP1 gene are implicated in autosomal recessive intellectual disability without neurodegeneration. European Journal of Human Genetics, 2016, 24, 392-399.	2.8	17

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55	BOD1 Is Required for Cognitive Function in Humans and Drosophila. PLoS Genetics, 2016, 12, e1006022.	3.5	18
56	Heterogeneity of Hereditary Hearing Loss in Iran: a Comprehensive Review. Archives of Iranian Medicine, 2016, 19, 720-728.	0.6	18
57	Genetic Studies in Intellectual Disability and Behavioral Impairment. Archives of Iranian Medicine, 2016, 19, 363-75.	0.6	10
58	PDZD7 and hearing loss: More than just a modifier. American Journal of Medical Genetics, Part A, 2015, 167, 2957-2965.	1.2	54
59	Finding mutation within non-coding region of GJB2 reveals its importance in genetic testing of Hearing Loss in Iranian population. International Journal of Pediatric Otorhinolaryngology, 2015, 79, 136-138.	1.0	4
60	Redefining the MED13L syndrome. European Journal of Human Genetics, 2015, 23, 1308-1317.	2.8	53
61	Characterising the spectrum of autosomal recessive hereditary hearing loss in Iran. Journal of Medical Genetics, 2015, 52, 823-829.	3.2	87
62	A defect in the CLIP1 gene (CLIP-170) can cause autosomal recessive intellectual disability. European Journal of Human Genetics, 2015, 23, 331-336.	2.8	22
63	The Role of a Novel TRMT1 Gene Mutation and Rare GRM1 Gene Defect in Intellectual Disability in Two Azeri Families. PLoS ONE, 2015, 10, e0129631.	2.5	56
64	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
65	New evidence for the role of calpain 10 in autosomal recessive intellectual disability: identification of two novel nonsense variants by exome sequencing in Iranian families. Archives of Iranian Medicine, 2015, 18, 179-84.	0.6	6
66	Carrier Testing in Known Autosomal Recessive Intellectual Disability Genes in an Iranian Healthy Individual Using Exome Sequencing. Archives of Iranian Medicine, 2015, 18, 643-69.	0.6	2
67	Exome Sequencing and Linkage Analysis Identified Novel Candidate Genes in Recessive Intellectual Disability Associated with Ataxia. Archives of Iranian Medicine, 2015, 18, 670-82.	0.6	4
68	Genetic Investigation of an Iranian Supercentenarian by Whole Exome Sequencing. Archives of Iranian Medicine, 2015, 18, 688-97.	0.6	6
69	Impact of whole exome sequencing among Iranian patients with autosomal recessive retinitis pigmentosa. Archives of Iranian Medicine, 2015, 18, 776-85.	0.6	15
70	Integrated Sequence Analysis Pipeline Provides One-Stop Solution for Identifying Disease-Causing Mutations. Human Mutation, 2014, 35, 1427-1435.	2.5	31
71	A novel mutation of the USH2C (GPR98) gene in an Iranian family with Usher syndrome type II. Journal of Genetics, 2014, 93, 837-841.	0.7	14
72	Investigation of ATP6V1B1 and ATP6V0A4 genes causing hereditary hearing loss associated with distal renal tubular acidosis in Iranian families. Journal of Laryngology and Otology, 2014, 128, 1056-1059.	0.8	5

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73	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
74	Genetics of non-syndromic hearing loss in the Middle East. International Journal of Pediatric Otorhinolaryngology, 2014, 78, 2026-2036.	1.0	45
75	Mutation profile of BBS genes in Iranian patients with Bardetâ€“Biedl syndrome: genetic characterization and report of nine novel mutations in five BBS genes. Journal of Human Genetics, 2014, 59, 368-375.	2.3	33
76	Utilizing Ethnic-Specific Differences in Minor Allele Frequency to Recategorize Reported Pathogenic Deafness Variants. American Journal of Human Genetics, 2014, 95, 445-453.	6.2	137
77	Association of polymorphisms at LDLR locus with coronary artery disease independently from lipid profile. Acta Medica Iranica, 2014, 52, 352-9.	0.8	11
78	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
79	A comprehensive study to determine heterogeneity of autosomal recessive nonsyndromic hearing loss in Iran. American Journal of Medical Genetics, Part A, 2012, 158A, 2485-2492.	1.2	55
80	Screening for MYO15A gene mutations in autosomal recessive nonsyndromic, GJB2 negative Iranian deaf population. American Journal of Medical Genetics, Part A, 2012, 158A, 1857-1864.	1.2	54
81	Deep sequencing reveals 50 novel genes for recessive cognitive disorders. Nature, 2011, 478, 57-63.	27.8	805
82	Next generation sequencing in a family with autosomal recessive Kahrizi syndrome (OMIM 612713) reveals a homozygous frameshift mutation in SRD5A3. European Journal of Human Genetics, 2011, 19, 115-117.	2.8	52
83	Mutations in the first MyTH4 domain of MYO15A are a common cause of DFNB3 hearing loss. Laryngoscope, 2009, 119, 727-733.	2.0	48
84	Identification of SLC26A4 gene mutations in Iranian families with hereditary hearing impairment. European Journal of Pediatrics, 2009, 168, 651-653.	2.7	30
85	An autosomal recessive syndrome of severe mental retardation, cataract, coloboma and kyphosis maps to the pericentromeric region of chromosome 4. European Journal of Human Genetics, 2009, 17, 125-128.	2.8	17
86	A novel DFNA5 mutation does not cause hearing loss in an Iranian family. Journal of Human Genetics, 2007, 52, 549-552.	2.3	26
87	GJB2 mutations: Passage through Iran. American Journal of Medical Genetics, Part A, 2005, 133A, 132-137.	1.2	77