

Kimia Kahrizi

List of Publications by Year in descending order

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Version: 2024-02-01

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	Deep sequencing reveals 50 novel genes for recessive cognitive disorders. <i>Nature</i> , 2011, 478, 57-63.	27.8	805
2	Utilizing Ethnic-Specific Differences in Minor Allele Frequency to Recategorize Reported Pathogenic Deafness Variants. <i>American Journal of Human Genetics</i> , 2014, 95, 445-453.	6.2	137
3	Genetics of intellectual disability in consanguineous families. <i>Molecular Psychiatry</i> , 2019, 24, 1027-1039.	7.9	131
4	Iranome: A catalog of genomic variations in the Iranian population. <i>Human Mutation</i> , 2019, 40, 1968-1984.	2.5	116
5	Characterising the spectrum of autosomal recessive hereditary hearing loss in Iran. <i>Journal of Medical Genetics</i> , 2015, 52, 823-829.	3.2	87
6	GJB2 mutations: Passage through Iran. <i>American Journal of Medical Genetics, Part A</i> , 2005, 133A, 132-137.	1.2	77
7	CIB2, defective in isolated deafness, is key for auditory hair cell mechanotransduction and survival. <i>EMBO Molecular Medicine</i> , 2017, 9, 1711-1731.	6.9	66
8	The Role of a Novel TRMT1 Gene Mutation and Rare GRM1 Gene Defect in Intellectual Disability in Two Azeri Families. <i>PLoS ONE</i> , 2015, 10, e0129631.	2.5	56
9	A comprehensive study to determine heterogeneity of autosomal recessive nonsyndromic hearing loss in Iran. <i>American Journal of Medical Genetics, Part A</i> , 2012, 158A, 2485-2492.	1.2	55
10	Adenosine kinase deficiency: expanding the clinical spectrum and evaluating therapeutic options. <i>Journal of Inherited Metabolic Disease</i> , 2016, 39, 273-283.	3.6	55
11	Screening for <i>MYO15A</i> gene mutations in autosomal recessive nonsyndromic, <i>GJB2</i> negative Iranian deaf population. <i>American Journal of Medical Genetics, Part A</i> , 2012, 158A, 1857-1864.	1.2	54
12	<i>PDZD7</i> and hearing loss: More than just a modifier. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 2957-2965.	1.2	54
13	Mutations of the aminoacyl-tRNA-synthetases SARS and WARS2 are implicated in the etiology of autosomal recessive intellectual disability. <i>Human Mutation</i> , 2017, 38, 621-636.	2.5	54
14	Redefining the MED13L syndrome. <i>European Journal of Human Genetics</i> , 2015, 23, 1308-1317.	2.8	53
15	Next generation sequencing in a family with autosomal recessive Kahrizi syndrome (OMIM 612713) reveals a homozygous frameshift mutation in SRD5A3. <i>European Journal of Human Genetics</i> , 2011, 19, 115-117.	2.8	52
16	CDC14A phosphatase is essential for hearing and male fertility in mouse and human. <i>Human Molecular Genetics</i> , 2018, 27, 780-798.	2.9	49
17	Effect of inbreeding on intellectual disability revisited by trio sequencing. <i>Clinical Genetics</i> , 2019, 95, 151-159.	2.0	49
18	Mutations in the first MyTH4 domain of <i>MYO15A</i> are a common cause of DFNB3 hearing loss. <i>Laryngoscope</i> , 2009, 119, 727-733.	2.0	48

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19	Old gene, new phenotype: splice-altering variants in <i>CEACAM16</i> cause recessive non-syndromic hearing impairment. <i>Journal of Medical Genetics</i> , 2018, 55, 555-560.	3.2	48
20	Variants in <i>CIB2</i> cause DFNB48 and not USH1J. <i>Clinical Genetics</i> , 2018, 93, 812-821.	2.0	46
21	Genetics of non-syndromic hearing loss in the Middle East. <i>International Journal of Pediatric Otorhinolaryngology</i> , 2014, 78, 2026-2036.	1.0	45
22	De novo and inherited mutations in the X-linked gene <i>CLCN4</i> are associated with syndromic intellectual disability and behavior and seizure disorders in males and females. <i>Molecular Psychiatry</i> , 2018, 23, 222-230.	7.9	45
23	Exonic mutations and exon skipping: Lessons learned from <i>DFNA5</i> . <i>Human Mutation</i> , 2018, 39, 433-440.	2.5	44
24	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.	2.0	41
25	Homozygous Truncating Variants in <i>TBC1D23</i> Cause Pontocerebellar Hypoplasia and Alter Cortical Development. <i>American Journal of Human Genetics</i> , 2017, 101, 428-440.	6.2	39
26	Distinct genetic variation and heterogeneity of the Iranian population. <i>PLoS Genetics</i> , 2019, 15, e1008385.	3.5	34
27	Mutation profile of BBS genes in Iranian patients with Bardet-Biedl syndrome: genetic characterization and report of nine novel mutations in five BBS genes. <i>Journal of Human Genetics</i> , 2014, 59, 368-375.	2.3	33
28	Integrated Sequence Analysis Pipeline Provides One-Stop Solution for Identifying Disease-Causing Mutations. <i>Human Mutation</i> , 2014, 35, 1427-1435.	2.5	31
29	Identification of <i>SLC26A4</i> gene mutations in Iranian families with hereditary hearing impairment. <i>European Journal of Pediatrics</i> , 2009, 168, 651-653.	2.7	30
30	A novel <i>DFNA5</i> mutation does not cause hearing loss in an Iranian family. <i>Journal of Human Genetics</i> , 2007, 52, 549-552.	2.3	26
31	The power of the Mediator complex-Expanding the genetic architecture and phenotypic spectrum of <i>MED12</i> -related disorders. <i>Clinical Genetics</i> , 2018, 94, 450-456.	2.0	24
32	Bi-allelic Pathogenic Variants in <i>TUBGCP2</i> Cause Microcephaly and Lissencephaly Spectrum Disorders. <i>American Journal of Human Genetics</i> , 2019, 105, 1005-1015.	6.2	24
33	A defect in the <i>CLIP1</i> gene (<i>CLIP-170</i>) can cause autosomal recessive intellectual disability. <i>European Journal of Human Genetics</i> , 2015, 23, 331-336.	2.8	22
34	<i>POLRMT</i> mutations impair mitochondrial transcription causing neurological disease. <i>Nature Communications</i> , 2021, 12, 1135.	12.8	21
35	Biallelic missense variants in <i>ZBTB11</i> can cause intellectual disability in humans. <i>Human Molecular Genetics</i> , 2018, 27, 3177-3188.	2.9	19
36	SARS-CoV-2 outbreak in Iran: The dynamics of the epidemic and evidence on two independent introductions. <i>Transboundary and Emerging Diseases</i> , 2022, 69, 1375-1386.	3.0	19

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37	LGMD2E is the most common type of sarcoglycanopathies in the Iranian population. <i>Journal of Neurogenetics</i> , 2017, 31, 161-169.	1.4	18
38	BOD1 Is Required for Cognitive Function in Humans and Drosophila. <i>PLoS Genetics</i> , 2016, 12, e1006022.	3.5	18
39	Heterogeneity of Hereditary Hearing Loss in Iran: a Comprehensive Review. <i>Archives of Iranian Medicine</i> , 2016, 19, 720-728.	0.6	18
40	An autosomal recessive syndrome of severe mental retardation, cataract, coloboma and kyphosis maps to the pericentromeric region of chromosome 4. <i>European Journal of Human Genetics</i> , 2009, 17, 125-128.	2.8	17
41	Missense variants in AIMP1 gene are implicated in autosomal recessive intellectual disability without neurodegeneration. <i>European Journal of Human Genetics</i> , 2016, 24, 392-399.	2.8	17
42	Impact of whole exome sequencing among Iranian patients with autosomal recessive retinitis pigmentosa. <i>Archives of Iranian Medicine</i> , 2015, 18, 776-85.	0.6	15
43	A novel mutation of the USH2C (GPR98) gene in an Iranian family with Usher syndrome type II. <i>Journal of Genetics</i> , 2014, 93, 837-841.	0.7	14
44	YIF1B mutations cause a post-natal neurodevelopmental syndrome associated with Golgi and primary cilium alterations. <i>Brain</i> , 2020, 143, 2911-2928.	7.6	13
45	<i>GPR126</i> : A novel candidate gene implicated in autosomal recessive intellectual disability. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 13-19.	1.2	12
46	Association of polymorphisms at LDLR locus with coronary artery disease independently from lipid profile. <i>Acta Medica Iranica</i> , 2014, 52, 352-9.	0.8	11
47	Homozygous variants in the gene <i>SCAPER</i> cause syndromic intellectual disability. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 1214-1225.	1.2	10
48	Genetic Studies in Intellectual Disability and Behavioral Impairment. <i>Archives of Iranian Medicine</i> , 2016, 19, 363-75.	0.6	10
49	Correlation between distribution of muscle weakness, electrophysiological findings and CTG expansion in myotonic dystrophy. <i>Journal of Clinical Neuroscience</i> , 2014, 21, 1123-1126.	1.5	9
50	SLC52A2 mutations cause SCABD2 phenotype: A second report. <i>International Journal of Pediatric Otorhinolaryngology</i> , 2018, 104, 195-199.	1.0	9
51	Novel mutations in MYTH4-FERM domains of myosin 15 are associated with autosomal recessive nonsyndromic hearing loss. <i>International Journal of Pediatric Otorhinolaryngology</i> , 2019, 117, 115-126.	1.0	9
52	Novel Mutations in KCNQ4, LHFPL5 and COCH Genes in Iranian Families with Hearing Impairment. <i>Archives of Iranian Medicine</i> , 2019, 22, 189-197.	0.6	9
53	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.	0.6	8
54	Genotype and phenotype analysis of 43 Iranian facioscapulohumeral muscular dystrophy patients; Evidence for anticipation. <i>Neuromuscular Disorders</i> , 2018, 28, 303-314.	0.6	6

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55	Limbic System Associated Membrane Protein Mutation in an Iranian Family Diagnosed with MÃ©niÃ©reâ€™s Disease. <i>Archives of Iranian Medicine</i> , 2020, 23, 319-325.	0.6	6
56	Molecular Diagnosis of Hereditary Neuropathies by Whole Exome Sequencing and Expanding the Phenotype Spectrum. <i>Archives of Iranian Medicine</i> , 2020, 23, 426-433.	0.6	6
57	Genetic etiology of hearing loss in Iran. <i>Human Genetics</i> , 2022, 141, 623-631.	3.8	6
58	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. <i>Archives of Iranian Medicine</i> , 2015, 18, 179-84.	0.6	6
59	Genetic Investigation of an Iranian Supercentenarian by Whole Exome Sequencing. <i>Archives of Iranian Medicine</i> , 2015, 18, 688-97.	0.6	6
60	Investigation of ATP6V1B1 and ATP6VOA4 genes causing hereditary hearing loss associated with distal renal tubular acidosis in Iranian families. <i>Journal of Laryngology and Otology</i> , 2014, 128, 1056-1059.	0.8	5
61	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.	0.6	5
62	Identification of diseaseâ€™causing variants in the <i>EXOSC</i> gene family underlying autosomal recessive intellectual disability in Iranian families. <i>Clinical Genetics</i> , 2019, 95, 718-725.	2.0	5
63	ZBTB11 dysfunction: spectrum of brain abnormalities, biochemical signature and cellular consequences. <i>Brain</i> , 2022, 145, 2602-2616.	7.6	5
64	tRNA Methyltransferase Defects and Intellectual Disability. <i>Archives of Iranian Medicine</i> , 2018, 21, 478-485.	0.6	5
65	Phenotype and genotype spectrum of variants in guanine nucleotide exchange factor genes in a broad cohort of Iranian patients. <i>Molecular Genetics & Genomic Medicine</i> , 2022, 10, e1894.	1.2	5
66	Finding mutation within non-coding region of GJB2 reveals its importance in genetic testing of Hearing Loss in Iranian population. <i>International Journal of Pediatric Otorhinolaryngology</i> , 2015, 79, 136-138.	1.0	4
67	<i>CNKSRL</i> gene defect can cause syndromic autosomal recessive intellectual disability. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2018, 177, 691-699.	1.7	4
68	Whole-Transcriptome Analysis Reveals Dysregulation of Actin-Cytoskeleton Pathway in Intellectual Disability Patients. <i>Neuroscience</i> , 2019, 404, 423-444.	2.3	4
69	Exome sequencing utility in defining the genetic landscape of hearing loss and novelâ€™gene discovery in Iran. <i>Clinical Genetics</i> , 2021, 100, 59-78.	2.0	4
70	Calpains: Diverse Functions but Enigmatic. <i>Archives of Iranian Medicine</i> , 2018, 21, 170-179.	0.6	4
71	Exome Sequencing and Linkage Analysis Identified Novel Candidate Genes in Recessive Intellectual Disability Associated with Ataxia. <i>Archives of Iranian Medicine</i> , 2015, 18, 670-82.	0.6	4
72	A novel PTC mutation in the BTB domain of KLHL7 gene in two patients with Bohring-Opitz syndrome-like features. <i>European Journal of Medical Genetics</i> , 2020, 63, 103849.	1.3	3

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73	<i>CEP104</i> and <i>CEP290</i>; Genes with Ciliary Functions Cause Intellectual Disability in Multiple Families. Archives of Iranian Medicine, 2021, 24, 364-373.	0.6	3
74	The recurrent missense mutation p.(Arg367Trp) in YARS1 causes a distinct neurodevelopmental phenotype. Journal of Molecular Medicine, 2021, 99, 1755-1768.	3.9	3
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
76	The <i>PTRHD1</i> Mutation in Intellectual Disability. Archives of Iranian Medicine, 2021, 24, 747-751.	0.6	3
77	Intellectual Disability and Ataxia: Genetic Collisions. Archives of Iranian Medicine, 2018, 21, 29-40.	0.6	3
78	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
79	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
80	Novel Mutation in <i>LARP7</i> in Two Iranian Consanguineous Families with Syndromic Intellectual Disability and Facial Dysmorphism. Archives of Iranian Medicine, 2020, 23, 842-847.	0.6	2
81	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
82	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
83	A splice-altering variant in LARP7 gene leads to exon exclusion. Gene Reports, 2019, 15, 100375.	0.8	1
84	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
85	Intellectual disability associated with craniofacial dysmorphism due to POLR3B mutation and defect in spliceosomal machinery. BMC Medical Genomics, 2022, 15, 89.	1.5	1
86	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
87	Contribution of Iran in Elucidating the Genetic Causes of Autosomal Recessive Intellectual Disability. Archives of Iranian Medicine, 2019, 22, 461-471.	0.6	0