Nejat Mahdieh

List of Publications by Year
in descending order

81	1,822	430754	289141
papers	citations	18 h-index	40 g-index

87	87	87	3174
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	The promise of whole-exome sequencing in medical genetics. Journal of Human Genetics, 2014, 59, 5-15.	1.1	404
2	Next-generation sequencing: impact of exome sequencing in characterizing Mendelian disorders. Journal of Human Genetics, 2012, 57, 621-632.	1.1	177
3	Whole-Exome Sequencing Efficiently Detects Rare Mutations in Autosomal Recessive Nonsyndromic Hearing Loss. PLoS ONE, 2012, 7, e50628.	1.1	143
4	Comprehensive analysis via exome sequencing uncovers genetic etiology in autosomal recessive nonsyndromic deafness in a large multiethnic cohort. Genetics in Medicine, 2016, 18, 364-371.	1.1	124
5	Next generation sequencing: implications in personalized medicine and pharmacogenomics. Molecular BioSystems, 2016, 12, 1818-1830.	2.9	82
6	GJB2 mutations: Passage through Iran. American Journal of Medical Genetics, Part A, 2005, 133A, 132-137.	0.7	77
7	Genetic causes of nonsyndromic hearing loss in Iran in comparison with other populations. Journal of Human Genetics, 2010, 55, 639-648.	1.1	71
8	Statistical study of 35delG mutation of GJB2 gene: A meta-analysis of carrier frequency. International Journal of Audiology, 2009, 48, 363-370.	0.9	60
9	Variations in Multiple Syndromic Deafness Genes Mimic Non-syndromic Hearing Loss. Scientific Reports, 2016, 6, 31622.	1.6	44
10	An overview of mutation detection methods in genetic disorders. Iranian Journal of Pediatrics, 2013, 23, 375-88.	0.1	41
11	Screening of OTOF mutations in Iran: A novel mutation and review. International Journal of Pediatric Otorhinolaryngology, 2012, 76, 1610-1615.	0.4	37
12	The frequency of GJB2 mutations and the Î” (GJB6-D13S1830) deletion as a cause of autosomal recessive non-syndromic deafness in the Kurdish population. Clinical Genetics, 2004, 65, 506-508.	1.0	34
13	Impact of Consanguineous Marriages in GJB2-Related Hearing Loss in the Iranian Population: A Report of a Novel Variant. Genetic Testing and Molecular Biomarkers, 2011, 15, 489-493.	0.3	31
14	Beta thalassemia in 31,734 cases with HBB gene mutations: Pathogenic and structural analysis of the common mutations; Iran as the crossroads of the Middle East. Blood Reviews, 2016, 30, 493-508.	2.8	31
15	Next generation sequencing applications for cardiovascular disease. Annals of Medicine, 2018, 50, 91-109.	1.5	29
16	In silico structural, functional and pathogenicity evaluation of a novel mutation: An overview of HSD3B2 gene mutations. Gene, 2012, 503, 215-221.	1.0	27
17	Mutation Analysis of theCYP21A2Gene in the Iranian Population. Genetic Testing and Molecular Biomarkers, 2012, 16, 82-90.	0.3	25
18	High level of intrafamilial phenotypic variability of non-syndromic hearing loss in a Lur family due to delE120 mutation in GJB2 gene. International Journal of Pediatric Otorhinolaryngology, 2010, 74, 1089-1091.	0.4	23

#	Article	IF	Citations
19	A transversion mutation in non-coding exon 3 of the TMC1 gene in two ethnically related Iranian deaf families from different geographical regions; evidence for founder effect. International Journal of Pediatric Otorhinolaryngology, 2013, 77, 821-826.	0.4	20
20	Genetic testing of leukodystrophies unraveling extensive heterogeneity in a large cohort and report of five common diseases and 38 novel variants. Scientific Reports, 2021, 11, 3231.	1.6	18
21	The frequency of eight common point mutations in CYP21 gene in Iranian patients with congenital adrenal hyperplasia. Iranian Biomedical Journal, 2008, 12, 49-53.	0.4	18
22	MYO15A splicing mutations in hearing loss: A review literature and report of a novel mutation. International Journal of Pediatric Otorhinolaryngology, 2017, 96, 35-38.	0.4	17
23	Design, expression and evaluation of a novel humanized single chain antibody against epidermal growth factor receptor (EGFR). Protein Expression and Purification, 2016, 127, 8-15.	0.6	14
24	GJB2 mutations in deaf population of Ilam (Western Iran): a different pattern of mutation distribution. European Archives of Oto-Rhino-Laryngology, 2016, 273, 1161-1165.	0.8	14
25	A novel de novo dominant mutation of NOTCH1 gene in an Iranian family with nonâ€syndromic congenital heart disease. Journal of Clinical Laboratory Analysis, 2020, 34, e23147.	0.9	13
26	Novel pathogenic variants underlie SLC26A4 -related hearing loss in a multiethnic cohort. International Journal of Pediatric Otorhinolaryngology, 2017, 101, 167-171.	0.4	11
27	Identification of a novel missense c.386Gâ€‰>â€‰A variant in a boy with the POMGNT1-related muscular dystrophy-dystroglycanopathy. Acta Neurologica Belgica, 2021, 121, 143-151.	0.5	11
28	A novel homozygous missense variant in the NAXE gene in an Iranian family with progressive encephalopathy with brain edema and leukoencephalopathy. Acta Neurologica Belgica, 2022, 122, 1201-1210.	0.5	10
29	Novel cases of pediatric sudden cardiac death secondary to TRDN mutations presenting as long QT syndrome at rest and catecholaminergic polymorphic ventricular tachycardia during exercise: The TRDN arrhythmia syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 3433-3445.	0.7	10
30	A novel PKP2 mutation and intrafamilial phenotypic variability in ARVC/D. Medical Journal of the Islamic Republic of Iran, 2018, 32, 22-26.	0.9	10
31	Molecular Diagnosis of Congenital Adrenal Hyperplasia in Iran: Focusing on CYP21A2 Gene. Iranian Journal of Pediatrics, 2011, 21, 139-50.	0.1	10
32	Construction, expression, and activity of a novel immunotoxin comprising a humanized antiepidermal growth factor receptor scFv and modified Pseudomonas aeruginosa exotoxin A. Anti-Cancer Drugs, 2017, 28, 263-270.	0.7	9
33	Unexpected heterogeneity due to recessive and de novo dominant mutations of GJB2 in an Iranian family with nonsyndromic hearing loss: Implication for genetic counseling. Biochemical and Biophysical Research Communications, 2010, 402, 305-307.	1.0	8
34	Development of Doxorubicin-Loaded Nanostructured Lipid Carriers: Preparation, Characterization, and In Vitro Evaluation on MCF-7 Cell Line. BioNanoScience, 2017, 7, 32-39.	1.5	8
35	The Frequency of HBB Mutations Among Î²-Thalassemia Patients in Hamadan Province, Iran. Hemoglobin, 2017, 41, 61-64.	0.4	8
36	Hypomyelinating Leukodystrophy with Spinal Cord Involvement Caused by a Novel Variant in RARS: Report of Two Unrelated Patients. Neuropediatrics, 2019, 50, 130-134.	0.3	8

#	Article	IF	Citations
37	A comprehensive in silico analysis, distribution and frequency of human Nkx2-5 mutations; A critical gene in congenital heart disease. Journal of Cardiovascular and Thoracic Research, 2019, 11, 287-299.	0.3	8
38	A Case-Control Study on the Association of Common Variants of CAPN10 Gene and the Risk of Type 2 Diabetes in an Iranian Population. Clinical Laboratory, 2014, 60, 663-70.	0.2	8
39	Clopidogrel Pharmacogenetics in Iranian Patients Undergoing Percutaneous Coronary Intervention. Cardiovascular Toxicology, 2018, 18, 482-491.	1.1	7
40	GATA4 screening in Iranian patients of various ethnicities affected with congenital heart disease: Coâ€•occurrence of a novel de novo translocation (5;7) and a likely pathogenic heterozygous GATA4 mutation in a family with autosomal dominant congenital heart disease. Journal of Clinical Laboratory Analysis, 2019, 33, e22923.	0.9	7
41	The Genetic Perspective of Familial Glucocorticoid Deficiency: In Silico Analysis of Two Novel Variants. International Journal of Endocrinology, 2020, 2020, 1-8.	0.6	7
42	Patient-Specific Induced Pluripotent Stem Cell-Derived Hepatocyte-Like Cells as a Model to Study Autosomal Recessive Hypercholesterolemia. Stem Cells and Development, 2021, 30, 714-724.	1.1	7
43	Investigation of GJB6 large deletions in Iranian patients using quantitative real-time PCR. Clinical Laboratory, 2010, 56, 467-71.	0.2	7
44	Genotypic effect of a mutation of the <scp>MYBPC</scp>3 gene and two phenotypes with different patterns of inheritance. Journal of Clinical Laboratory Analysis, 2018, 32, e22419.	0.9	6
45	Mosaic trisomy 22 in a 4â€•yearâ€•old boy with congenital heart disease and general hypotrophy: A case report. Journal of Clinical Laboratory Analysis, 2019, 33, e22663.	0.9	6
46	Novel disease-causing variants in a cohort of Iranian patients with metachromatic leukodystrophy and in silico analysis of their pathogenicity. Clinical Neurology and Neurosurgery, 2021, 201, 106448.	0.6	6
47	A systematic review of LDLR, PCSK9, and APOB variants in Asia. Atherosclerosis, 2020, 305, 50-57.	0.4	6
48	Epilepsia Partialis Continua a Clinical Feature of a Missense Variant in the ADCK3 Gene and Poor Response to Therapy. Journal of Molecular Neuroscience, 2022, 72, 1125-1132.	1.1	6
49	Characterization of minor bands of STR amplification reaction of FVIII gene by PCR cloning. Clinica Chimica Acta, 2008, 394, 114-115.	0.5	5
50	A novel missense variant in GPT2 causes non-syndromic autosomal recessive intellectual disability in a consanguineous Iranian family. European Journal of Medical Genetics, 2020, 63, 103853.	0.7	5
51	Brown-Vialetto-Van Laere syndrome and Fazio-Londe syndrome: A novel mutation and in silico analyses. Journal of Clinical Neuroscience, 2020, 72, 342-349.	0.8	5
52	Heterozygosity and allele frequencies of the two VNTRs (ApoB and D1S80) in Iranian population. Indian Journal of Human Genetics, 2005, 11, 31.	0.7	5
53	Autosomal Recessive Nonsyndromic Arrhythmogenic Right Ventricular Cardiomyopathy without Cutaneous Involvements: A Novel Mutation. Annals of Human Genetics, 2017, 81, 135-140.	0.3	4
54	Genotype, phenotype and in silico pathogenicity analysis of HEXB mutations: Panel based sequencing for differential diagnosis of gangliosidosis. Clinical Neurology and Neurosurgery, 2018, 167, 43-53.	0.6	4

#	Article	IF	Citations
55	Whole-Exome Sequencing Reveals a Novel Mutation of FLNA Gene in an Iranian Family with Nonsyndromic Tetralogy of Fallot. Laboratory Medicine, 2021, 52, 614-618.	0.8	4
56	PCR-ELISA: A diagnostic assay for identifying Iranian HIV seropositives. Molecular Genetics, Microbiology and Virology, 2013, 28, 127-131.	0.0	3
57	A case of autosomal recessive hypercholesterolemia with a novel mutation in the <i>LDLRAP1</i> gene. Clinical Pediatric Endocrinology, 2021, 30, 201-204.	0.4	3
58	GFAP variants leading to infantile Alexander disease: Phenotype and genotype analysis of 135 cases and report of a de novo variant. Clinical Neurology and Neurosurgery, 2021, 207, 106754.	0.6	3
59	Pathogenic significance of SCN1A splicing variants causing Dravet syndrome: Improving diagnosis with targeted sequencing for variants by in silico analysis. Clinical Neurology and Neurosurgery, 2018, 166, 80-90.	0.6	2
60	Carney Complex syndrome. Cardiovascular Pathology, 2020, 49, 107231.	0.7	2
61	Genetic homozygosity in a diverse population: An experience of long QT syndrome. International Journal of Cardiology, 2020, 316, 117-124.	0.8	2
62	Nonsyndromic Early-Onset Epileptic Encephalopathies: Two Novel <i>KCTD7</i> Pathogenic Variants and a Literature Review. Developmental Neuroscience, 2021, 43, 348-357.	1.0	2
63	Seizure as the Early and Main Manifestation of Infantile Vanishing White Matter Disease: A Case Report. Iranian Journal of Pediatrics, 2018, 28, .	0.1	2
64	A Novel PCBD Gene Mutation in an Iranian Patient with Hyperphenylalaninemia. Clinical Laboratory, 2013, 59, 925-8.	0.2	2
65	CASE REPORT: A Girl with 45,X/46,XX Turner Syndrome and Salt Wasting Form of Congenital Adrenal Hyperplasia Due to Regulatory Changes. Clinical Laboratory, 2013, 59, .	0.2	2
66	Genetics of Hearing Loss. , 2012, , .		1
67	A novel pathogenic variant of SRD5A2 in an Iranian psuedohermaphrodite male. Clinical Case Reports (discontinued), 2020, 8, 1947-1951.	0.2	1
68	p.Gln318X and p.Val281Leu as the Major Variants of CYP21A2 Gene in Children with Idiopathic Premature Pubarche. International Journal of Endocrinology, 2020, 2020, 1-9.	0.6	1
69	HBB mutations and HbA2 level: Escaping the carrier screening programs. Clinical Case Reports (discontinued), 2021, 9, 973-977.	0.2	1
70	21-hydroxylase deficiency: newborn screening in iran?. Iranian Journal of Pediatrics, 2012, 22, 279-80.	0.1	1
71	Investigation of Chromosomal Abnormalities and Microdeletion/ Microduplication(s) in Fifty Iranian Patients with Multiple Congenital Anomalies. Cell Journal, 2019, 21, 337-349.	0.2	1
72	Clinical and Molecular Findings of Autosomal Recessive Spastic Ataxia of Charlevoix Saguenay: an Iranian Case Series Expanding the Genetic and Neuroimaging Spectra. Cerebellum, 0, , .	1.4	1

#	Article	IF	Citations
73	Homozygosity mapping and CDH23 mutation analysis in Iranian deaf families. Hearing, Balance and Communication, 2016, 14, 189-193.	0.1	0
74	Cardiovascular Genetics. , 2018, , 525-533.		0
75	Genetics of Cardiovascular Disease and Applications of Genetic Testing. , 2022, , 665-674.		0
76	Amplicon Secondary Structure Formation and Elongation during the Process of Sequencing. Journal of Proteomics and Bioinformatics, 2012, 05, .	0.4	0
77	Design of a Biological Method for Rapid Elimination of PCR Inhibitors in Aged Bone DNA. Clinical Laboratory, 2013, 59, .	0.2	0
78	Megalencephalic Leukoencephalopathy with Subcortical Cysts: Presentation of an Asymptomatic Patient and Review of Literature. Iranian Journal of Pediatrics, 2019, In Press, .	0.1	0
79	An Asymptomatic Case of Megalencephalic Leukoencephalopathy with Subcortical Cysts. Iranian Journal of Pediatrics, 2019, 29, .	0.1	0
80	PCR-ELISA: a diagnostic assay for identifying Iranian HIV seropositives. Molekuliarnaia Genetika, Mikrobiologiia I Virusologiia, 2013, , 36-9.	0.1	0
81	An Iranian Congenital Adrenal Hypoplasia Patient with Elevated Testosterone in Infancy due to a Novel Pathogenic Frameshift Variant in NR0B1. International Journal of Endocrinology, 2021, 2021, 1-5.	0.6	0