Nejat Mahdieh

List of Publications by Year in Descending Order

75 papers	1,465 citations	18 h-index	37 g-index
87 ext. papers	1,714 ext. citations	2.4 avg, IF	4.66 L-index

#	Paper	IF	Citations
75	Genetics of Cardiovascular Disease and Applications of Genetic Testing **2022**, 665-674		
74	Epilepsia Partialis Continua a Clinical Feature of a Missense Variant in the ADCK3 Gene and Poor Response to Therapy.. *Journal of Molecular Neuroscience*, **2022**, 1	3.3	1
73	mutations and HbA2 level: Escaping the carrier screening programs. *Clinical Case Reports (discontinued)*, **2021**, 9, 973-977	0.7	0
72	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	1.6	0
71	A novel homozygous missense variant in the NAXE gene in an Iranian family with progressive encephalopathy with brain edema and leukoencephalopathy. *Acta Neurologica Belgica*, **2021**, 1	1.5	2
70	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	4.4	4
69	Identification of a novel missense c.386G > A variant in a boy with the POMGNT1-related muscular dystrophy-dystroglycanopathy. *Acta Neurologica Belgica*, **2021**, 121, 143-151	1.5	5
68	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	2	2
67	A case of autosomal recessive hypercholesterolemia with a novel mutation in the gene. *Clinical Pediatric Endocrinology*, **2021**, 30, 201-204	1.4	0
66	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	4.9	2
65	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	2	0
64	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	2.5	1
63	Nonsyndromic Early-Onset Epileptic Encephalopathies: Two Novel KCTD7 Pathogenic Variants and a Literature Review. *Developmental Neuroscience*, **2021**, 43, 348-357	2.2	
62	An Iranian Congenital Adrenal Hypoplasia Patient with Elevated Testosterone in Infancy due to a Novel Pathogenic Frameshift Variant in .. *International Journal of Endocrinology*, **2021**, 2021, 4367028	2.7	
61	Genetic homozygosity in a diverse population: An experience of long QT syndrome. *International Journal of Cardiology*, **2020**, 316, 117-124	3.2	1
60	A novel pathogenic variant of in an Iranian psuedohermaphrodite male. *Clinical Case Reports (discontinued)*, **2020**, 8, 1947-1951	0.7	
59	p.Gln318X and p.Val281Leu as the Major Variants of Gene in Children with Idiopathic Premature Pubarche. *International Journal of Endocrinology*, **2020**, 2020, 4329791	2.7	

58	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	2.6	4
57	Brown-Vialetto-Van Laere syndrome and Fazio-Londe syndrome: A novel mutation and in silico analyses. *Journal of Clinical Neuroscience*, **2020**, 72, 342-349	2.2	3
56	A systematic review of LDLR, PCSK9, and APOB variants in Asia. *Atherosclerosis*, **2020**, 305, 50-57	3.1	3
55	A novel de novo dominant mutation of NOTCH1 gene in an Iranian family with non-syndromic congenital heart disease. *Journal of Clinical Laboratory Analysis*, **2020**, 34, e23147	3	7
54	Carney Complex syndrome. *Cardiovascular Pathology*, **2020**, 49, 107231	3.8	0
53	The Genetic Perspective of Familial Glucocorticoid Deficiency: Analysis of Two Novel Variants. *International Journal of Endocrinology*, **2020**, 2020, 2190508	2.7	2
52	GATA4 screening in Iranian patients of various ethnicities affected with congenital heart disease: Co-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	3	5
51	Hypomyelinating Leukodystrophy with Spinal Cord Involvement Caused by a Novel Variant in RARS: Report of Two Unrelated Patients. *Neuropediatrics*, **2019**, 50, 130-134	1.6	6
50	A comprehensive in silico analysis, distribution and frequency of human mutations; A critical gene in congenital heart disease. *Journal of Cardiovascular and Thoracic Research*, **2019**, 11, 287-299	1.3	5
49	Investigation of Chromosomal Abnormalities and Microdeletion/ Microduplication(s) in Fifty Iranian Patients with Multiple Congenital Anomalies. *Cell Journal*, **2019**, 21, 337-349	2.4	1
48	Mosaic trisomy 22 in a 4-year-old boy with congenital heart disease and general hypotrophy: A case report. *Journal of Clinical Laboratory Analysis*, **2019**, 33, e22663	3	3
47	Genotypic effect of a mutation of the MYBPC3 gene and two phenotypes with different patterns of inheritance. *Journal of Clinical Laboratory Analysis*, **2018**, 32, e22419	3	4
46	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	2	4
45	Cardiovascular Genetics **2018**, 525-533		
44	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	2	1
43	Next generation sequencing applications for cardiovascular disease. *Annals of Medicine*, **2018**, 50, 91-109	1.5	16
42	A novel mutation and intrafamilial phenotypic variability in ARVC/D. *Medical Journal of the Islamic Republic of Iran*, **2018**, 32, 5	1.1	4
41	Seizure as the Early and Main Manifestation of Infantile Vanishing White Matter Disease: A Case Report. *Iranian Journal of Pediatrics*, **2018**, 28,	1	1

40	Clopidogrel Pharmacogenetics in Iranian Patients Undergoing Percutaneous Coronary Intervention. *Cardiovascular Toxicology*, **2018**, 18, 482-491	3.4	5
39	Development of Doxorubicin-Loaded Nanostructured Lipid Carriers: Preparation, Characterization, and In Vitro Evaluation on MCF-7 Cell Line. *BioNanoScience*, **2017**, 7, 32-39	3.4	4
38	The Frequency of HBB Mutations Among Thalassemia Patients in Hamadan Province, Iran. *Hemoglobin*, **2017**, 41, 61-64	0.6	5
37	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	2.4	7
36	Autosomal Recessive Nonsyndromic Arrhythmogenic Right Ventricular Cardiomyopathy without Cutaneous Involvements: A Novel Mutation. *Annals of Human Genetics*, **2017**, 81, 135-140	2.2	3
35	MYO15A splicing mutations in hearing loss: A review literature and report of a novel mutation. *International Journal of Pediatric Otorhinolaryngology*, **2017**, 96, 35-38	1.7	12
34	Novel pathogenic variants underlie SLC26A4-related hearing loss in a multiethnic cohort. *International Journal of Pediatric Otorhinolaryngology*, **2017**, 101, 167-171	1.7	6
33	Comprehensive analysis via exome sequencing uncovers genetic etiology in autosomal recessive nonsyndromic deafness in a large multiethnic cohort. *Genetics in Medicine*, **2016**, 18, 364-71	8.1	92
32	GJB2 mutations in deaf population of Ilam (Western Iran): a different pattern of mutation distribution. *European Archives of Oto-Rhino-Laryngology*, **2016**, 273, 1161-5	3.5	10
31	Variations in Multiple Syndromic Deafness Genes Mimic Non-syndromic Hearing Loss. *Scientific Reports*, **2016**, 6, 31622	4.9	28
30	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	2	13
29	Homozygosity mapping and CDH23 mutation analysis in Iranian deaf families. *Hearing, Balance and Communication*, **2016**, 14, 189-193	0.7	
28	Next generation sequencing: implications in personalized medicine and pharmacogenomics. *Molecular BioSystems*, **2016**, 12, 1818-30		63
27	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	11.1	18
26	The promise of whole-exome sequencing in medical genetics. *Journal of Human Genetics*, **2014**, 59, 5-15	4.3	310
25	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	2	5
24	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, 821-6	1.7	14
23	PCR-ELISA: A diagnostic assay for identifying Iranian HIV seropositives. *Molecular Genetics, Microbiology and Virology*, **2013**, 28, 127-131	0.4	2

22	An overview of mutation detection methods in genetic disorders. *Iranian Journal of Pediatrics*, **2013**, 23, 375-88	1	38
21	A novel PCBD gene mutation in an Iranian patient with hyperphenylalaninemia. *Clinical Laboratory*, **2013**, 59, 925-8	2	2
20	PCR-ELISA: a diagnostic assay for identifying Iranian HIV seropositives. *Molekuliarnaia Genetika, Mikrobiologiia I Virusologiia*, **2013**, 36-9	0.3	
19	Screening of OTOF mutations in Iran: a novel mutation and review. *International Journal of Pediatric Otorhinolaryngology*, **2012**, 76, 1610-5	1.7	32
18	In silico structural, functional and pathogenicity evaluation of a novel mutation: an overview of HSD3B2 gene mutations. *Gene*, **2012**, 503, 215-21	3.8	22
17	Next-generation sequencing: impact of exome sequencing in characterizing Mendelian disorders. *Journal of Human Genetics*, **2012**, 57, 621-32	4.3	155
16	Whole-exome sequencing efficiently detects rare mutations in autosomal recessive nonsyndromic hearing loss. *PLoS ONE*, **2012**, 7, e50628	3.7	120
15	Genetics of Hearing Loss **2012**,		1
14	Mutation analysis of the CYP21A2 gene in the Iranian population. *Genetic Testing and Molecular Biomarkers*, **2012**, 16, 82-90	1.6	19
13	21-hydroxylase deficiency: newborn screening in iran?. *Iranian Journal of Pediatrics*, **2012**, 22, 279-80	1	1
12	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, 489-93	1.6	26
11	Molecular Diagnosis of Congenital Adrenal Hyperplasia in Iran: Focusing on CYP21A2 Gene. *Iranian Journal of Pediatrics*, **2011**, 21, 139-50	1	9
10	Genetic causes of nonsyndromic hearing loss in Iran in comparison with other populations. *Journal of Human Genetics*, **2010**, 55, 639-48	4.3	63
9	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, 1089-91	1.7	22
8	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, 305-7	3.4	8
7	Investigation of GJB6 large deletions in Iranian patients using quantitative real-time PCR. *Clinical Laboratory*, **2010**, 56, 467-71	2	7
6	Statistical study of 35delG mutation of GJB2 gene: a meta-analysis of carrier frequency. *International Journal of Audiology*, **2009**, 48, 363-70	2.6	51
5	Characterization of minor bands of STR amplification reaction of FVIII gene by PCR cloning. *Clinica Chimica Acta*, **2008**, 394, 114-5	6.2	4

4	The frequency of eight common point mutations in CYP21 gene in Iranian patients with congenital adrenal hyperplasia. *Iranian Biomedical Journal*, **2008**, 12, 49-53	2	16
3	GJB2 mutations: passage through Iran. *American Journal of Medical Genetics, Part A*, **2005**, 133A, 132-7	2.5	72
2	Heterozygosity and allele frequencies of the two VNTRs (ApoB and D1S80) in Iranian population. *Indian Journal of Human Genetics*, **2005**, 11, 31		5
1	The frequency of GJB2 mutations and the Delta (GJB6-D13S1830) deletion as a cause of autosomal recessive non-syndromic deafness in the Kurdish population. *Clinical Genetics*, **2004**, 65, 506-8	4	29