Bahareh Rabbani

List of Publications by Year
in descending order

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44	1,203	687220 13	377752 34
papers	citations	h-index	g-index

49	49	49	2454
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Genetics of Cardiovascular Disease and Applications of Genetic Testing. , 2022, , 665-674.		0
2	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
3	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
4	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
5	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
6	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
7	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
8	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
9	Genetic homozygosity in a diverse population: An experience of long QT syndrome. International Journal of Cardiology, 2020, 316, 117-124.	0.8	2
10	A novel pathogenic variant of SRD5A2 in an Iranian psuedohermaphrodite male. Clinical Case Reports (discontinued), 2020, 8, 1947-1951.	0.2	1
11	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
12	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
13	A systematic review of LDLR, PCSK9, and APOB variants in Asia. Atherosclerosis, 2020, 305, 50-57.	0.4	6
14	Megalencephalic Leukoencephalopathy with Subcortical Cysts: Presentation of an Asymptomatic Patient and Review of Literature. Iranian Journal of Pediatrics, 2019, In Press, .	0.1	0
15	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
16	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
17	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
18	Clopidogrel Pharmacogenetics in Iranian Patients Undergoing Percutaneous Coronary Intervention. Cardiovascular Toxicology, 2018, 18, 482-491.	1.1	7

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19	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
20	The Frequency of HBB Mutations Among Î²-Thalassemia Patients in Hamadan Province, Iran. Hemoglobin, 2017, 41, 61-64.	0.4	8
21	Autosomal Recessive Nonsyndromic Arrhythmogenic Right Ventricular Cardiomyopathy without Cutaneous Involvements: A Novel Mutation. Annals of Human Genetics, 2017, 81, 135-140.	0.3	4
22	Next generation sequencing: implications in personalized medicine and pharmacogenomics. Molecular BioSystems, 2016, 12, 1818-1830.	2.9	82
23	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
24	Mutation detection of CYP21A2 gene in nonclassical congenital adrenal hyperplasia patients with premature pubarche. Advanced Biomedical Research, 2016, 5, 33.	0.2	4
25	The promise of whole-exome sequencing in medical genetics. Journal of Human Genetics, 2014, 59, 5-15.	1.1	404
26	HLA-DRB, -DQA, and DQB alleles andÂhaplotypes in Iranian patients with diabetes mellitus type I. Pediatric Diabetes, 2013, 14, 366-371.	1.2	13
27	PCR-ELISA: A diagnostic assay for identifying Iranian HIV seropositives. Molecular Genetics, Microbiology and Virology, 2013, 28, 127-131.	0.0	3
28	An overview of mutation detection methods in genetic disorders. Iranian Journal of Pediatrics, 2013, 23, 375-88.	0.1	41
29	PCR-ELISA: a diagnostic assay for identifying Iranian HIV seropositives. Molekuliarnaia Genetika, Mikrobiologiia I Virusologiia, 2013, , 36-9.	0.1	0
30	Mutation Analysis of theCYP21A2Gene in the Iranian Population. Genetic Testing and Molecular Biomarkers, 2012, 16, 82-90.	0.3	25
31	Screening of OTOF mutations in Iran: A novel mutation and review. International Journal of Pediatric Otorhinolaryngology, 2012, 76, 1610-1615.	0.4	37
32	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
33	Next-generation sequencing: impact of exome sequencing in characterizing Mendelian disorders. Journal of Human Genetics, 2012, 57, 621-632.	1.1	177
34	Genetics of Hearing Loss. , 2012, , .		1
35	21-hydroxylase deficiency: newborn screening in iran?. Iranian Journal of Pediatrics, 2012, 22, 279-80.	0.1	1
36	A girl with 45,X/46,XX Turner syndrome and salt wasting form of congenital adrenal hyperplasia due to regulatory changes. Clinical Laboratory, 2012, 58, 1063-6.	0.2	3

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37	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
38	Molecular Diagnosis of Congenital Adrenal Hyperplasia in Iran: Focusing on CYP21A2 Gene. Iranian Journal of Pediatrics, 2011, 21, 139-50.	0.1	10
39	Genetic causes of nonsyndromic hearing loss in Iran in comparison with other populations. Journal of Human Genetics, 2010, 55, 639-648.	1.1	71
40	Vitamin D Insufficiency among Children and Adolescents Living in Tehran, Iran. Journal of Tropical Pediatrics, 2009, 55, 189-191.	0.7	60
41	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
42	Characterization of minor bands of STR amplification reaction of FVIII gene by PCR cloning. Clinica Chimica Acta, 2008, 394, 114-115.	0.5	5
43	Analysing two dinucleotide repeats of FVIII gene in Iranian population. Haemophilia, 2007, 13, 740-744.	1.0	5
44	A novel single step double positive double negative selection strategy for Î²-globin gene replacement. Biochemical and Biophysical Research Communications, 2006, 345, 14-20.	1.0	5