Mohammad Keramatipour

List of Publications by Year
in descending order

41 papers	292 citations	1040056 9 h-index	996975 15 g-index
41 all docs	41 docs citations	41 times ranked	737 citing authors

#	Article	IF	Citations
1	Next-generation sequencing identified novel truncating mutations in BBS9 causing Bardet Biedl syndrome in two Iranian consanguineous families.. Iranian Journal of Child Neurology, 2022, 16, 123-133.	0.3	0
2	Identifying Novel Mutations in Iranian Patients with LPS-responsive Beige-like Anchor Protein (LRBA) Deficiency. Immunological Investigations, 2021, 50, 399-405.	2.0	1
3	SPOAN syndrome: a novel mutation and new ocular findings; a case report. BMC Neurology, 2021, 21, 24.	1.8	3
4	Four mutations in MITF, SOX10 and PAX3 genes were identified as genetic causes of waardenburg syndrome in four unrelated Iranian patients: case report. BMC Pediatrics, 2021, 21, 70.	1.7	4
5	The importance of CDC27 in cancer: molecular pathology and clinical aspects. Cancer Cell International, 2021, 21, 160.	4.1	24
6	Identification of a novel truncating variant in AHI1 gene and a brief review on mutations spectrum. Molecular Biology Reports, 2021, 48, 5339-5345.	2.3	3
7	Exome sequencing reveals novel rare variants in Iranian familial multiple sclerosis: The importance of POLD2 in the disease pathogenesis. Genomics, 2021, 113, 2645-2655.	2.9	5
8	Whole-Exome Sequencing Identified a Novel Variant (C.405_422+39del) in DSP Gene in an Iranian Pedigree with Familial Dilated Cardiomyopathy. Reports of Biochemistry and Molecular Biology, 2021, 10, 280-287.	1.4	2
9	Waardenburg syndrome type 2A in a large Iranian family with a novel MITF gene mutation. BMC Medical Genomics, 2021, 14, 230.	1.5	0
10	X-linked SCID with a rare mutation. Allergy, Asthma and Clinical Immunology, 2021, 17, 107.	2.0	3
11	Three Novel Variants identified in FBN1 and TGFBR2 in seven Iranian families with suspected Marfan syndrome. Molecular Genetics & Genomic Medicine, 2020, 8, e1274.	1.2	4
12	Dysfunction of the ciliary ARMC9/TOGARAM1 protein module causes Joubert syndrome. Journal of Clinical Investigation, 2020, 130, 4423-4439.	8.2	43
13	Whole Exome Sequencing Reveals a XPNPEP3 Novel Mutation Causing Nephronophthisis in a Pediatric Patient. Iranian Biomedical Journal, 2020, 24, 400-403.	0.7	4
14	Once in a Blue Moon, a Very Rare Coexistence of Glutaric Acidemia Type I and Mucopolysaccharidosis Type IIIB in a Patient. Iranian Biomedical Journal, 2020, 24, 201-205.	0.7	2
15	Succinate Dehydrogenase Deficiency: A Treatable Neurometabolic Disorder. Iranian Journal of Child Neurology, 2020, 14, 111-116.	0.3	1
16	Genetic mutations and immunological features of severe combined immunodeficiency patients in Iran. Immunology Letters, 2019, 216, 70-78.	2.5	14
17	Multi affected pedigree with congenital microcephaly: WES revealed PNKP gene mutation. Brain and Development, 2019, 41, 182-186.	1.1	11
18	CRB1-Related Leber Congenital Amaurosis: Reporting Novel Pathogenic Variants and a Brief Review on Mutations Spectrum. Iranian Biomedical Journal, 2019, 23, 362-8.	0.7	1

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19	Targeted next generation sequencing identified a novel mutation in MYO7A causing Usher syndrome type 1 in an Iranian consanguineous pedigree. International Journal of Pediatric Otorhinolaryngology, 2018, 104, 10-13.	1.0	7
20	The association between <scp>ST</scp>18 gene polymorphism and severe pemphigus disease among Iranian population. Experimental Dermatology, 2018, 27, 1395-1398.	2.9	15
21	First report of Klein-Waardenburg Syndrome in Iran and a novel pathogenic splice site variant in PAX3 gene. International Journal of Pediatric Otorhinolaryngology, 2018, 113, 229-233.	1.0	5
22	Investigation of the association of G-7A and T-138C single nucleotide polymorphisms on the promoter of MGP gene with renal stone. Journal of Nephropharmacology, 2018, 7, 145-148.	0.4	1
23	Effect of Whole Exome Sequencing in Diagnosis of Inborn Errors of Metabolism and Neurogenetic Disorders. Iranian Journal of Child Neurology, 2018, 12, 7-15.	0.3	6
24	Comparison of the effects of nobiletin and letrozole on the activity and expression of aromatase in the MCF-7 breast cancer cell line. Biochemistry and Cell Biology, 2017, 95, 468-473.	2.0	10
25	Genetic study of the PAH locus in the Iranian population: familial gene mutations and minihaplotypes. Metabolic Brain Disease, 2017, 32, 1685-1691.	2.9	13
26	Homozygosity Mapping and Targeted Sanger Sequencing Identifies Three Novel CRB1 (Cumbs) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50 467 294-302.	0.7	
27	Comparison of Two Different PCR-based Methods for Detection of GAA Expansions in Frataxin Gene. Iranian Journal of Public Health, 2017, 46, 222-228.	0.5	2
28	Identification of a Novel Mutation in the PAH Gene in an Iranian Phenylketonuria Family: A Case Report. Iranian Journal of Public Health, 2017, 46, 560-564.	0.5	4
29	A Novel Variant in the Gene Causing Phenylketonuria in an Iranian Pedigree. Avicenna Journal of Medical Biotechnology, 2017, 9, 146-149.	0.3	2
30	A Novel Mutation in Gene Causes Malignant Infantile Osteopetrosis. Avicenna Journal of Medical Biotechnology, 2017, 9, 205-208.	0.3	6
31	PKD2 mutation in an Iranian autosomal dominant polycystic kidney disease family with misleading linkage analysis data. Kidney Research and Clinical Practice, 2016, 35, 96-101.	2.2	1
32	APOA II genotypes frequency and their interaction with saturated fatty acids consumption on lipid profile of patients with type 2 diabetes. Clinical Nutrition, 2016, 35, 907-911.	5.0	15
33	Whole Exome Sequencing Reveals a BSCL2 Mutation Causing Progressive Encephalopathy with Lipodystrophy (PELD) in an Iranian Pediatric Patient. Iranian Biomedical Journal, 2016, 20, 295-301.	0.7	8
34	A Case Series: Congenital Hyperinsulinism. International Journal of Endocrinology and Metabolism, 2016, 14, e37311.	1.0	9
35	The Relationship Between Coronary Artery Disease and Genetic Polymorphisms of Melanoma Inhibitory Activity 3. Iranian Red Crescent Medical Journal, 2016, 18, e31146.	0.5	1
36	Setting up Multiplex Panels for Genetic Testing of Familial Hypertrophic Cardiomyopathy Based on Linkage Analysis. Iranian Journal of Public Health, 2016, 45, 329-39.	0.5	0

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37	An Efficient Trio-Based Mini-Haplotyping Method for Genetic Diagnosis of Phenylketonuria. Cell Journal, 2016, 18, 229-36.	0.2	2
38	Functional Analysis of A Novel Splicing Mutation in The Mutase Gene of Two Unrelated Pedigrees. Cell Journal, 2016, 18, 397-404.	0.2	1
39	Effect of DHA-rich fish oil on PPARÎ³ target genes related to lipid metabolism in type 2 diabetes: A randomized, double-blind, placebo-controlled clinical trial. Journal of Clinical Lipidology, 2015, 9, 770-777.	1.5	43
40	Investigating the Effect of rs3783605 Single-nucleotide Polymorphism on the Activity of VCAM-1 Promoter in Human Umbilical Vein Endohelial Cells. Iranian Journal of Allergy, Asthma and Immunology, 2015, 14, 179-87.	0.4	2
41	Single Nucleotide Polymorphism rs 2476601 of PTPN22 Gene and Susceptibility to Rheumatoid Arthritis in Iranian Population. Iranian Journal of Allergy, Asthma and Immunology, 2015, 14, 437-42.	0.4	7