Maryam Abiri

List of Publications by Year
in descending order

26 papers	341 citations	1039406 9 h-index	887659 17 g-index
26 all docs	26 docs citations	26 times ranked	495 citing authors

#	Article	IF	Citations
1	Comprehensive Mutation Analysis and Report of 12 Novel Mutations in a Cohort of Patients with Spinal Muscular Atrophy in Iran. Journal of Molecular Neuroscience, 2021, 71, 2281-2298.	1.1	9
2	Hints From the Cellular Functions to the Practical Outlook of Circular RNAs. Frontiers in Genetics, 2021, 12, 679446.	1.1	9
3	Comprehensive transcriptome mining identified the gene expression signature and differentially regulated pathways of the late-onset preeclampsia. Pregnancy Hypertension, 2021, 25, 91-102.	0.6	6
4	An Infertile Azoospermic Male with 45, X T(Yp;15) Karyotype. Journal of Family & Reproductive Health, 2021, 15, 271-274.	0.4	0
5	Genotype-phenotype correlation and description of two novel mutations in Iranian patients with glycogen storage disease 1b (GSD1b). Orphanet Journal of Rare Diseases, 2020, 15, 35.	1.2	4
6	Whole-Exome Sequencing Uncovers Novel Causative Variants and Additional Findings in Three Patients Affected by Glycogen Storage Disease Type VI and Fanconiâ˜Bickel Syndrome. Frontiers in Genetics, 2020, 11, 601566.	1.1	7
7	Maple syrup urine disease mutation spectrum in a cohort of 40 consanguineous patients and insilico analysis of novel mutations. Metabolic Brain Disease, 2019, 34, 1145-1156.	1.4	11
8	Genetic testing of Mucopolysaccharidoses disease using multiplex PCR- based panels of STR markers: in silico analysis of novel mutations. Metabolic Brain Disease, 2019, 34, 1447-1455.	1.4	5
9	Molecular genetic diagnosis of Glanzmann syndrome in Iranian population; reporting novel and recurrent mutations. Orphanet Journal of Rare Diseases, 2019, 14, 87.	1.2	10
10	Development and validation of a novel panel of 16 STR markers for simultaneous diagnosis of Î²-thalassemia, aneuploidy screening, maternal cell contamination detection and fetal sample authenticity in PND and PGD/PGS cases. Scientific Reports, 2019, 9, 7452.	1.6	5
11	Genomeâ€wide single nucleotide polymorphismâ€based autozygosity mapping facilitates identification of mutations in consanguineous families with epidermolysis bullosa. Experimental Dermatology, 2019, 28, 1118-1121.	1.4	19
12	Development and implementation of a novel panel consisting 20 markers for the detection of genetic causes of male infertility. Andrologia, 2018, 50, e12946.	1.0	1
13	Recessive mutation in tetraspanin CD151 causes Kindler syndrome-like epidermolysis bullosa with multi-systemic manifestations including nephropathy. Matrix Biology, 2018, 66, 22-33.	1.5	49
14	Molecular genetics of a cohort of 635 cases of phenylketonuria in a consanguineous population. Journal of Inherited Metabolic Disease, 2018, 41, 1159-1167.	1.7	14
15	Autozygosity mapping of methylmalonic acidemia associated genes by short tandem repeat markers facilitates the identification of five novel mutations in an Iranian patient cohort. Metabolic Brain Disease, 2018, 33, 1689-1697.	1.4	5
16	Multigene Next-Generation Sequencing Panel Identifies Pathogenic Variants in Patients with Unknown Subtype of Epidermolysis Bullosa: Subclassification with Prognostic Implications. Journal of Investigative Dermatology, 2017, 137, 2649-2652.	0.3	31
17	Dystrophic Epidermolysis Bullosa: COL7A1 Mutation Landscape in a Multi-Ethnic Cohort of 152 Extended Families with High Degree of Customary Consanguineous Marriages. Journal of Investigative Dermatology, 2017, 137, 660-669.	0.3	44
18	In silico analysis of novel mutations in maple syrup urine disease patients from Iran. Metabolic Brain Disease, 2017, 32, 105-113.	1.4	16

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19	Gene-Targeted Next Generation Sequencing Identifies PNPLA1 Mutations in Patients with a Phenotypic Spectrum of Autosomal Recessive Congenital Ichthyosis: The Impact of Consanguinity. Journal of Investigative Dermatology, 2017, 137, 678-685.	0.3	28
20	Hyaline Fibromatosis Syndrome: A Novel Mutation and Recurrent Founder Mutation in the CMG2/ANTXR2 Gene. Acta Dermato-Venereologica, 2017, 97, 108-109.	0.6	9
21	Co-existence of phenylketonuria either with maple syrup urine disease or Sandhoff disease in two patients from Iran: emphasizing the role of consanguinity. Journal of Pediatric Endocrinology and Metabolism, 2016, 29, 1215-1219.	0.4	7
22	Linkage Study Revealed Complex Haplotypes in a Multifamily due to Different Mutations in CAPN3 Gene in an Iranian Ethnic Group. Journal of Molecular Neuroscience, 2016, 59, 392-396.	1.1	5
23	Identification of six novel mutations in Iranian patients with maple syrup urine disease and their in silico analysisâ€‹. Mutation Research - Fundamental and Molecular Mechanisms of Mutagenesis, 2016, 786, 34-40.	0.4	12
24	Molecular Characterization of QDPR Gene in Iranian Families with BH4 Deficiency: Reporting Novel and Recurrent Mutations. JIMD Reports, 2015, 21, 123-128.	0.7	8
25	Replication of TCF7L2 rs7903146 association with type 2 diabetes in an Iranian population. Genetics and Molecular Biology, 2010, 33, 449-451.	0.6	27
26	Producing a Mammalian GFP Expression Vector Containing Neomycin Resistance Gene. Avicenna Journal of Medical Biotechnology, 2009, 1, 33-6.	0.2	0