Seyedeh Sedigheh Abedini

List of Publications by Year
in descending order

29 papers	1,558 citations	840585 11 h-index	501076 28 g-index
30 all docs	30 docs citations	30 times ranked	3596 citing authors

#	Article	IF	Citations
1	Deep sequencing reveals 50 novel genes for recessive cognitive disorders. Nature, 2011, 478, 57-63.	13.7	805
2	A Defect in the Ionotropic Glutamate Receptor 6 Gene (GRIK2) Is Associated with Autosomal Recessive Mental Retardation. American Journal of Human Genetics, 2007, 81, 792-798.	2.6	137
3	Genetics of intellectual disability in consanguineous families. Molecular Psychiatry, 2019, 24, 1027-1039.	4.1	131
4	Homozygosity mapping in consanguineous families reveals extreme heterogeneity of non-syndromic autosomal recessive mental retardation and identifies 8 novel gene loci. Human Genetics, 2007, 121, 43-48.	1.8	92
5	Mutation of the conserved polyadenosine RNA binding protein, ZC3H14/dNab2, impairs neural function in Drosophila and humans. Proceedings of the National Academy of Sciences of the United States of America, 2011, 108, 12390-12395.	3.3	77
6	SNP array-based homozygosity mapping reveals MCPH1 deletion in family with autosomal recessive mental retardation and mild microcephaly. Human Genetics, 2006, 118, 708-715.	1.8	67
7	Next generation sequencing in a family with autosomal recessive Kahrizi syndrome (OMIM 612713) reveals a homozygous frameshift mutation in SRD5A3. European Journal of Human Genetics, 2011, 19, 115-117.	1.4	52
8	Autosomal recessive mental retardation: homozygosity mapping identifies 27 single linkage intervals, at least 14 novel loci and several mutation hotspots. Human Genetics, 2011, 129, 141-148.	1.8	45
9	Association Study of the TREM2 Gene and Identification of a Novel Variant in Exon 2 in Iranian Patients with Late-Onset Alzheimer's Disease. Medical Principles and Practice, 2015, 24, 351-354.	1.1	19
10	Fragile X syndrome screening of families with consanguineous and non-consanguineous parents in the Iranian population. European Journal of Medical Genetics, 2009, 52, 170-173.	0.7	13
11	Novel phenotype and genotype spectrum of NARS2 and literature review of previous mutations. Irish Journal of Medical Science, 2022, 191, 1877-1890.	0.8	13
12	GPR126: A novel candidate gene implicated in autosomal recessive intellectual disability. American Journal of Medical Genetics, Part A, 2019, 179, 13-19.	0.7	12
13	Chromosome abnormality rate among Iranian patients with idiopathic mental retardation from consanguineous marriages. Archives of Medical Science, 2011, 2, 321-325.	0.4	11
14	The influence of the BCL11A polymorphism on the phenotype of patients with beta thalassemia could be affected by the beta globin locus control region and/or the Xmn1-HBG2 genotypic background. Blood Cells, Molecules, and Diseases, 2013, 51, 80-84.	0.6	11
15	The modifying effect of Xmn1-HBG2 on thalassemic phenotype is associated with its linked elements in the beta globin locus control region, including the palindromic site at 5â€²HS4. Blood Cells, Molecules, and Diseases, 2012, 48, 1-5.	0.6	10
16	Two Iranian families with a novel mutation in GJB2 causing autosomal dominant nonsyndromic hearing loss. American Journal of Medical Genetics, Part A, 2011, 155, 1202-1211.	0.7	9
17	Subcellular relocalization and nuclear redistribution of the RNA methyltransferases TRMT1 and TRMT1L upon neuronal activation. RNA Biology, 2021, 18, 1905-1919.	1.5	9
18	Report of a patient with limb-girdle muscular dystrophy, ptosis and ophthalmoparesis caused by plectinopathy. Archives of Iranian Medicine, 2015, 18, 60-4.	0.2	8

#	Article	IF	Citations
19	Frequency of Î±-Globin Gene Triplications and Coinheritance with Î²-Globin Gene Mutations in the Iranian Population. Hemoglobin, 2018, 42, 252-256.	0.4	7
20	New evidence for the role of calpain 10 in autosomal recessive intellectual disability: identification of two novel nonsense variants by exome sequencing in Iranian families. Archives of Iranian Medicine, 2015, 18, 179-84.	0.2	6
21	Identification of diseaseâ€causing variants in the EXOSC gene family underlying autosomal recessive intellectual disability in Iranian families. Clinical Genetics, 2019, 95, 718-725.	1.0	5
22	tRNA Methyltransferase Defects and Intellectual Disability. Archives of Iranian Medicine, 2018, 21, 478-485.	0.2	5
23	Exome Sequencing and Linkage Analysis Identified Novel Candidate Genes in Recessive Intellectual Disability Associated with Ataxia. Archives of Iranian Medicine, 2015, 18, 670-82.	0.2	4
24	Analyzing 5â€²HS3 and 5â€²HS4 LCR core regions and NF-E2 in Iranian thalassemia intermedia patients with normal or carrier status for beta-globin mutations. Blood Cells, Molecules, and Diseases, 2011, 46, 201-205.	0.6	3
25	Comprehensive <scp>genotypeâ€phenotype</scp> correlation in <scp>AP</scp>â€4 deficiency syndrome; Adding data from a large cohort of Iranian patients. Clinical Genetics, 2021, 99, 187-192.	1.0	2
26	Carrier Testing in Known Autosomal Recessive Intellectual Disability Genes in an Iranian Healthy Individual Using Exome Sequencing. Archives of Iranian Medicine, 2015, 18, 643-69.	0.2	2
27	Whole genome sequencing identifies a duplicated region encompassing Xq13.2q13.3 in a large Iranian family with intellectual disability. Molecular Genetics & Genomic Medicine, 2020, 8, e1418.	0.6	1
28	Screening , and Mutations in 4 Iranian Families with Non-Syndromic Tooth Agenesis. Avicenna Journal of Medical Biotechnology, 2020, 12, 236-240.	0.2	1
29	Contribution of Iran in Elucidating the Genetic Causes of Autosomal Recessive Intellectual Disability. Archives of Iranian Medicine, 2019, 22, 461-471.	0.2	0