Mohammadreza Dehghani

List of Publications by Year
in descending order

23 papers	280 citations	1040056 9 h-index	940533 16 g-index
23 all docs	23 docs citations	23 times ranked	808 citing authors

#	Article	IF	Citations
1	Testis development in the absence of SRY: chromosomal rearrangements at SOX9 and SOX3. European Journal of Human Genetics, 2015, 23, 1025-1032.	2.8	59
2	Loss of UGP2 in brain leads to a severe epileptic encephalopathy, emphasizing that bi-allelic isoform-specific start-loss mutations of essential genes can cause genetic diseases. Acta Neuropathologica, 2020, 139, 415-442.	7.7	38
3	Homozygous Missense Variants in NTNG2, Encoding a Presynaptic Netrin-G2 Adhesion Protein, Lead to a Distinct Neurodevelopmental Disorder. American Journal of Human Genetics, 2019, 105, 1048-1056.	6.2	30
4	Recessive mutations in ATP8A2 cause severe hypotonia, cognitive impairment, hyperkinetic movement disorders and progressive optic atrophy. Orphanet Journal of Rare Diseases, 2018, 13, 86.	2.7	29
5	Evaluation of miR-181b and miR-126-5p expression levels in T2DM patients compared to healthy individuals: Relationship with NF-Î°B gene expression. Endocrinologia, Diabetes Y NutriciÃ“n, 2020, 67, 454-460.	0.3	22
6	Genetic screening of Congenital Short Bowel Syndrome patients confirms CLMP as the major gene involved in the recessive form of this disorder. European Journal of Human Genetics, 2016, 24, 1627-1629.	2.8	18
7	Pontocerebellar hypoplasia due to bi-allelic variants in MINPP1. European Journal of Human Genetics, 2021, 29, 411-421.	2.8	13
8	Defective phosphatidylethanolamine biosynthesis leads to a broad ataxia-spasticity spectrum. Brain, 2021, 144, e30-e30.	7.6	12
9	Dravet phenotype in a subject with a der(4)t(4;8)(p16.3;p23.3) without the involvement of the LETM1 gene. European Journal of Medical Genetics, 2013, 56, 551-555.	1.3	11
10	Bi-allelic loss-of-function variants in BCAS3 cause a syndromic neurodevelopmental disorder. American Journal of Human Genetics, 2021, 108, 1069-1082.	6.2	8
11	Martsolf syndrome with novel mutation in the TBC1D20 gene in a family from Iran. American Journal of Medical Genetics, Part A, 2020, 182, 957-961.	1.2	6
12	Biallelic variants in <scp>ZNF142</scp> lead to a syndromic neurodevelopmental disorder. Clinical Genetics, 2022, 102, 98-109.	2.0	6
13	A Common Ancestral Asn242Ser Mutation in TMEM67 Identified in Multiple Iranian Families with Joubert Syndrome. Public Health Genomics, 2017, 20, 188-193.	1.0	5
14	A Novel Loss-of-Function Mutation in HOXB1 Associated with Autosomal Recessive Hereditary Congenital Facial Palsy in a Large Iranian Family. Molecular Syndromology, 2017, 8, 261-265.	0.8	5
15	Is there any relationship between mutation in CPS1 Gene and pregnancy loss?. International Journal of Reproductive BioMedicine, 2019, 17, .	0.9	5
16	A Novel Missense Mutation in the ALDH13 Gene Causes Anophthalmia in Two Unrelated Iranian Consanguineous Families. International Journal of Molecular and Cellular Medicine, 2017, 6, 131-134.	1.1	5
17	A Novel Mutation in the OFD1 Gene in a Family with Oral-Facial-Digital Syndrome Type 1: A Case Report. Iranian Journal of Public Health, 2016, 45, 1359-1366.	0.5	2
18	Novel variants underlying autosomal recessive neurodevelopmental disorders with intellectual disability in Iranian consanguineous families. Journal of Clinical Laboratory Analysis, 2022, 36, e24241.	2.1	2

#	Article	IF	Citations
19	Evaluation of the relationship between miR-337-3p and RAP1A gene in endometriosis. Journal of Endometriosis and Pelvic Pain Disorders, 0, , 228402652210996.	0.5	2
20	The effect of regular resistance exercise, vitamin D, and calcium supplements on the gastrocnemius muscle in rats in the post-menopausal period: An experimental study. International Journal of Reproductive BioMedicine, 2021, 19, 283-292.	0.9	1
21	A newborn with ambiguous genitalia and a complex X;Y rearrangement. Iranian Journal of Reproductive Medicine, 2014, 12, 351-6.	0.8	1
22	Newborn with Supernumerary Marker Chromosome Derived from Chromosomes 11 And 22- A Case Report. Iranian Journal of Public Health, 2016, 45, 376-80.	0.5	0
23	Co-segregation of variant NSUN2 Lue198Arg among Iranian family with intellectual disability: a case report. Egyptian Journal of Medical Human Genetics, 2022, 23, .	1.0	0