Alireza Haghighi

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/7508032/publications.pdf

Version: 2024-02-01

25 papers	1,007 citations	686830 13 h-index	610482 24 g-index
25 all docs	25 docs citations	25 times ranked	2222 citing authors

#	Article	IF	Citations
1	Genetic Determinants of Sudden Unexpected Death in Pediatrics. Genetics in Medicine, 2022, 24, 839-850.	1.1	20
2	Association of Parental Consanguinity With Papillary Thyroid Carcinoma: A Case-Control Study. Journal of Clinical Endocrinology and Metabolism, 2021, 106, e2769-e2774.	1.8	0
3	Mechanisms of Congenital Heart Disease Caused by NAA15 Haploinsufficiency. Circulation Research, 2021, 128, 1156-1169.	2.0	27
4	Genetic and Phenotypic Landscape of Peripartum Cardiomyopathy. Circulation, 2021, 143, 1852-1862.	1.6	65
5	Molecular analysis of dilated and left ventricular noncompaction cardiomyopathies in Egyptian children. Cardiology in the Young, 2021, , 1-6.	0.4	7
6	Contribution of Noncanonical Splice Variants toTTNTruncating Variant Cardiomyopathy. Circulation Genomic and Precision Medicine, 2021, 14, e003389.	1.6	15
7	Genetic study of pediatric hypertrophic cardiomyopathy in Egypt. Cardiology in the Young, 2020, 30, 1910-1916.	0.4	4
8	Expanding the clinical and genetic spectrum of ALPK3 variants: Phenotypes identified in pediatric cardiomyopathy patients and adults with heterozygous variants. American Heart Journal, 2020, 225, 108-119.	1.2	25
9	A gene-centric strategy for identifying disease-causing rare variants in dilated cardiomyopathy. Genetics in Medicine, 2019, 21, 133-143.	1.1	25
10	Pharmacogenomics in diabetes: outcomes of thiamine therapy in TRMA syndrome. Diabetologia, 2018, 61, 1027-1036.	2.9	26
11	An integrated clinical program and crowdsourcing strategy for genomic sequencing and Mendelian disease gene discovery. Npj Genomic Medicine, 2018, 3, 21.	1.7	24
12	Consanguinity and the Risk of Hashimoto's Thyroiditis. Thyroid, 2017, 27, 390-395.	2.4	2
13	The clinical, biochemical and genetic features associated withRMND1-related mitochondrial disease. Journal of Medical Genetics, 2016, 53, 768-775.	1.5	35
14	Genetics of GNE myopathy in the non-Jewish Persian population. European Journal of Human Genetics, 2016, 24, 243-251.	1.4	18
15	Titin mutations in iPS cells define sarcomere insufficiency as a cause of dilated cardiomyopathy. Science, 2015, 349, 982-986.	6.0	508
16	Homozygosity Mapping and Whole Exome Sequencing Reveal a Novel Homozygous COL18A1 Mutation Causing Knobloch Syndrome. PLoS ONE, 2014, 9, e112747.	1.1	15
17	A Novel Missense Mutation in Oncostatin M Receptor Beta Causing Primary Localized Cutaneous Amyloidosis. BioMed Research International, 2014, 2014, 1-6.	0.9	6
18	Sengers syndrome: six novel AGK mutations in seven new families and review of the phenotypic and mutational spectrum of 29 patients. Orphanet Journal of Rare Diseases, 2014, 9, 119.	1.2	77

#	Article	IF	Citations
19	Characterization of CSF2RA mutation related juvenile pulmonary alveolar proteinosis. Orphanet Journal of Rare Diseases, 2014, 9, 171.	1.2	61
20	Homozygous p.R284* mutation in HEXB gene causing Sandhoff disease with nystagmus. European Journal of Paediatric Neurology, 2014, 18, 399-403.	0.7	3
21	Identification of a c.601C>G mutation in the CCM1 gene in a kindred with multiple skin, spinal and cerebral cavernous malformations. Journal of the Neurological Sciences, 2013, 334, 97-101.	0.3	13
22	Missense mutation outside the forkhead domain of FOXL2 causes a severe form of BPES type II. Molecular Vision, 2012, 18, 211-8.	1.1	13
23	Tay-Sachs disease in an Arab family due to c.78G>A HEXA nonsense mutation encoding a p.W26X early truncation enzyme peptide. Molecular Genetics and Metabolism, 2011, 104, 700-702.	0.5	8
24	Identification of two HEXA mutations causing infantile-onset Tayâ€“Sachs disease in the Persian population. Journal of Human Genetics, 2011, 56, 682-684.	1.1	5
25	Senior-Loken syndrome secondary to NPHP5/IQCB1 mutation in an Iranian family. CKJ: Clinical Kidney Journal, 2011, 4, 421-423.	1.4	5