

# Alireza Haghghi

## 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	Titin mutations in iPS cells define sarcomere insufficiency as a cause of dilated cardiomyopathy. <i>Science</i> , 2015, 349, 982-986.	6.0	508
2	Sengers syndrome: six novel AGK mutations in seven new families and review of the phenotypic and mutational spectrum of 29 patients. <i>Orphanet Journal of Rare Diseases</i> , 2014, 9, 119.	1.2	77
3	Genetic and Phenotypic Landscape of Peripartum Cardiomyopathy. <i>Circulation</i> , 2021, 143, 1852-1862.	1.6	65
4	Characterization of CSF2RA mutation related juvenile pulmonary alveolar proteinosis. <i>Orphanet Journal of Rare Diseases</i> , 2014, 9, 171.	1.2	61
5	The clinical, biochemical and genetic features associated with <i>RMND1</i> -related mitochondrial disease. <i>Journal of Medical Genetics</i> , 2016, 53, 768-775.	1.5	35
6	Mechanisms of Congenital Heart Disease Caused by NAA15 Haploinsufficiency. <i>Circulation Research</i> , 2021, 128, 1156-1169.	2.0	27
7	Pharmacogenomics in diabetes: outcomes of thiamine therapy in TRMA syndrome. <i>Diabetologia</i> , 2018, 61, 1027-1036.	2.9	26
8	A gene-centric strategy for identifying disease-causing rare variants in dilated cardiomyopathy. <i>Genetics in Medicine</i> , 2019, 21, 133-143.	1.1	25
9	Expanding the clinical and genetic spectrum of ALPK3 variants: Phenotypes identified in pediatric cardiomyopathy patients and adults with heterozygous variants. <i>American Heart Journal</i> , 2020, 225, 108-119.	1.2	25
10	An integrated clinical program and crowdsourcing strategy for genomic sequencing and Mendelian disease gene discovery. <i>Npj Genomic Medicine</i> , 2018, 3, 21.	1.7	24
11	Genetic Determinants of Sudden Unexpected Death in Pediatrics. <i>Genetics in Medicine</i> , 2022, 24, 839-850.	1.1	20
12	Genetics of GNE myopathy in the non-Jewish Persian population. <i>European Journal of Human Genetics</i> , 2016, 24, 243-251.	1.4	18
13	Homozygosity Mapping and Whole Exome Sequencing Reveal a Novel Homozygous COL18A1 Mutation Causing Knobloch Syndrome. <i>PLoS ONE</i> , 2014, 9, e112747.	1.1	15
14	Contribution of Noncanonical Splice Variants to <i>TTN</i> Truncating Variant Cardiomyopathy. <i>Circulation Genomic and Precision Medicine</i> , 2021, 14, e003389.	1.6	15
15	Identification of a c.601C>G mutation in the CCM1 gene in a kindred with multiple skin, spinal and cerebral cavernous malformations. <i>Journal of the Neurological Sciences</i> , 2013, 334, 97-101.	0.3	13
16	Missense mutation outside the forkhead domain of FOXL2 causes a severe form of BPES type II. <i>Molecular Vision</i> , 2012, 18, 211-8.	1.1	13
17	Tay-Sachs disease in an Arab family due to c.78G>A HEXA nonsense mutation encoding a p.W26X early truncation enzyme peptide. <i>Molecular Genetics and Metabolism</i> , 2011, 104, 700-702.	0.5	8
18	Molecular analysis of dilated and left ventricular noncompaction cardiomyopathies in Egyptian children. <i>Cardiology in the Young</i> , 2021, , 1-6.	0.4	7

#	ARTICLE	IF	CITATIONS
19	A Novel Missense Mutation in Oncostatin M Receptor Beta Causing Primary Localized Cutaneous Amyloidosis. <i>BioMed Research International</i> , 2014, 2014, 1-6.	0.9	6
20	Identification of two HEXA mutations causing infantile-onset Tayâ€“Sachs disease in the Persian population. <i>Journal of Human Genetics</i> , 2011, 56, 682-684.	1.1	5
21	Senior-Loken syndrome secondary to NPHP5/IQCB1 mutation in an Iranian family. <i>CKJ: Clinical Kidney Journal</i> , 2011, 4, 421-423.	1.4	5
22	Genetic study of pediatric hypertrophic cardiomyopathy in Egypt. <i>Cardiology in the Young</i> , 2020, 30, 1910-1916.	0.4	4
23	Homozygous p.R284* mutation in HEXB gene causing Sandhoff disease with nystagmus. <i>European Journal of Paediatric Neurology</i> , 2014, 18, 399-403.	0.7	3
24	Consanguinity and the Risk of Hashimoto's Thyroiditis. <i>Thyroid</i> , 2017, 27, 390-395.	2.4	2
25	Association of Parental Consanguinity With Papillary Thyroid Carcinoma: A Case-Control Study. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2021, 106, e2769-e2774.	1.8	0