

Masoud Garshasbi

List of Publications by Citations

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

113
papers

2,981
citations

27
h-index

53
g-index

124
ext. papers

3,516
ext. citations

4
avg, IF

4.77
L-index

#	Paper	IF	Citations
113	Deep sequencing reveals 50 novel genes for recessive cognitive disorders. <i>Nature</i> , 2011 , 478, 57-63	50.4	649
112	Mutations in NSUN2 cause autosomal-recessive intellectual disability. <i>American Journal of Human Genetics</i> , 2012 , 90, 847-55	11	179
111	Somatic mutation profiles of MSI and MSS colorectal cancer identified by whole exome next generation sequencing and bioinformatics analysis. <i>PLoS ONE</i> , 2010 , 5, e15661	3.7	169
110	Cranioectodermal Dysplasia, Sensenbrenner syndrome, is a ciliopathy caused by mutations in the IFT122 gene. <i>American Journal of Human Genetics</i> , 2010 , 86, 949-56	11	154
109	A defect in the ionotropic glutamate receptor 6 gene (GRIK2) is associated with autosomal recessive mental retardation. <i>American Journal of Human Genetics</i> , 2007 , 81, 792-8	11	116
108	A defect in the TUSC3 gene is associated with autosomal recessive mental retardation. <i>American Journal of Human Genetics</i> , 2008 , 82, 1158-64	11	107
107	CA8 mutations cause a novel syndrome characterized by ataxia and mild mental retardation with predisposition to quadrupedal gait. <i>PLoS Genetics</i> , 2009 , 5, e1000487	6	99
106	Homozygosity mapping in consanguineous families reveals extreme heterogeneity of non-syndromic autosomal recessive mental retardation and identifies 8 novel gene loci. <i>Human Genetics</i> , 2007 , 121, 43-8	6.3	79
105	Mutations in the Alpha 1,2-Mannosidase Gene, MAN1B1, Cause Autosomal-Recessive Intellectual Disability. <i>American Journal of Human Genetics</i> , 2011 , 89, 348	11	78
104	A clinical and molecular genetic study of 112 Iranian families with primary microcephaly. <i>Journal of Medical Genetics</i> , 2010 , 47, 823-8	5.8	72
103	Genetic implications in the pathogenesis of rheumatoid arthritis; an updated review. <i>Gene</i> , 2019 , 702, 8-16	3.8	70
102	ST3GAL3 mutations impair the development of higher cognitive functions. <i>American Journal of Human Genetics</i> , 2011 , 89, 407-14	11	70
101	SNP array-based homozygosity mapping reveals MCPH1 deletion in family with autosomal recessive mental retardation and mild microcephaly. <i>Human Genetics</i> , 2006 , 118, 708-15	6.3	62
100	Mutation of the conserved polyadenosine RNA binding protein, ZC3H14/dNab2, impairs neural function in Drosophila and humans. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2011 , 108, 12390-5	11.5	61
99	Mutations in the alpha 1,2-mannosidase gene, MAN1B1, cause autosomal-recessive intellectual disability. <i>American Journal of Human Genetics</i> , 2011 , 89, 176-82	11	60
98	Identification of a novel candidate gene for non-syndromic autosomal recessive intellectual disability: the WASH complex member SWIP. <i>Human Molecular Genetics</i> , 2011 , 20, 2585-90	5.6	60
97	Evaluation of DNMT1 gene expression profile and methylation of its promoter region in patients with ankylosing spondylitis. <i>Clinical Rheumatology</i> , 2016 , 35, 2723-2731	3.9	46

96	Next generation sequencing in a family with autosomal recessive Kahrizi syndrome (OMIM 612713) reveals a homozygous frameshift mutation in SRD5A3. <i>European Journal of Human Genetics</i> , 2011 , 19, 115-7	5.3	45
95	Identification of a nonsense mutation in the very low-density lipoprotein receptor gene (VLDLR) in an Iranian family with dysequilibrium syndrome. <i>European Journal of Human Genetics</i> , 2008 , 16, 270-3	5.3	44
94	Establishment of a mouse model with misregulated chromosome condensation due to defective Mcph1 function. <i>PLoS ONE</i> , 2010 , 5, e9242	3.7	43
93	Mutations of the aminoacyl-tRNA-synthetases SARS and WARS2 are implicated in the etiology of autosomal recessive intellectual disability. <i>Human Mutation</i> , 2017 , 38, 621-636	4.7	40
92	Autosomal recessive mental retardation: homozygosity mapping identifies 27 single linkage intervals, at least 14 novel loci and several mutation hotspots. <i>Human Genetics</i> , 2011 , 129, 141-8	6.3	40
91	Promoter hypermethylation of BCL11B gene correlates with downregulation of gene transcription in ankylosing spondylitis patients. <i>Genes and Immunity</i> , 2017 , 18, 170-175	4.4	36
90	A novel nonsense mutation in TUSC3 is responsible for non-syndromic autosomal recessive mental retardation in a consanguineous Iranian family. <i>American Journal of Medical Genetics, Part A</i> , 2011 , 155A, 1976-80	2.5	35
89	Non-Coding RNAs in Cartilage Development: An Updated Review. <i>International Journal of Molecular Sciences</i> , 2019 , 20,	6.3	34
88	Expanded mutational spectrum in Cohen syndrome, tissue expression, and transcript variants of COH1. <i>Human Mutation</i> , 2009 , 30, E404-20	4.7	33
87	Cohen syndrome diagnosis using whole genome arrays. <i>Journal of Medical Genetics</i> , 2011 , 48, 136-40	5.8	28
86	PTRHD1 (C2orf79) mutations lead to autosomal-recessive intellectual disability and parkinsonism. <i>Movement Disorders</i> , 2017 , 32, 287-291	7	23
85	Identifying occult maternal malignancies from 1.93 million pregnant women undergoing noninvasive prenatal screening tests. <i>Genetics in Medicine</i> , 2019 , 21, 2293-2302	8.1	21
84	Mutations in the histamine N-methyltransferase gene, HNMT, are associated with nonsyndromic autosomal recessive intellectual disability. <i>Human Molecular Genetics</i> , 2015 , 24, 5697-710	5.6	19
83	Determination of IL1 R2, ANTXR2, CARD9, and SNAPC4 single nucleotide polymorphisms in Iranian patients with ankylosing spondylitis. <i>Rheumatology International</i> , 2016 , 36, 429-35	3.6	19
82	alpha-globin gene deletion and point mutation analysis among in Iranian patients with microcytic hypochromic anemia. <i>Haematologica</i> , 2003 , 88, 1196-7	6.6	19
81	An update on clinical, pathological, diagnostic, and therapeutic perspectives of childhood leukodystrophies. <i>Expert Review of Neurotherapeutics</i> , 2020 , 20, 65-84	4.3	17
80	miR-31 and miR-145 as Potential Non-Invasive Regulatory Biomarkers in Patients with Endometriosis. <i>Cell Journal</i> , 2018 , 20, 84-89	2.4	16
79	A defect in the CLIP1 gene (CLIP-170) can cause autosomal recessive intellectual disability. <i>European Journal of Human Genetics</i> , 2015 , 23, 331-6	5.3	15

78	An autosomal recessive syndrome of severe mental retardation, cataract, coloboma and kyphosis maps to the pericentromeric region of chromosome 4. <i>European Journal of Human Genetics</i> , 2009 , 17, 125-8	5.3	15
77	Missense variants in AIMP1 gene are implicated in autosomal recessive intellectual disability without neurodegeneration. <i>European Journal of Human Genetics</i> , 2016 , 24, 392-9	5.3	14
76	A novel ALDH5A1 mutation is associated with succinic semialdehyde dehydrogenase deficiency and severe intellectual disability in an Iranian family. <i>American Journal of Medical Genetics, Part A</i> , 2013 , 161A, 1915-22	2.5	14
75	Identification of miR-24 and miR-137 as novel candidate multiple sclerosis miRNA biomarkers using multi-staged data analysis protocol. <i>Molecular Biology Research Communications</i> , 2017 , 6, 127-140	1.6	14
74	Aberrant expression of a five-microRNA signature in breast carcinoma as a promising biomarker for diagnosis. <i>Journal of Clinical Laboratory Analysis</i> , 2020 , 34, e23063	3	14
73	New kid on the ID block: neural functions of the Nab2/ZC3H14 class of Cys ² His tandem zinc-finger polyadenosine RNA binding proteins. <i>RNA Biology</i> , 2012 , 9, 555-62	4.8	13
72	Whole-exome sequencing identifies R1279X of MYH6 gene to be associated with congenital heart disease. <i>BMC Cardiovascular Disorders</i> , 2018 , 18, 137	2.3	12
71	RNASET2-deficient leukoencephalopathy mimicking congenital CMV infection and Aicardi-Goutieres syndrome: a case report with a novel pathogenic variant. <i>Orphanet Journal of Rare Diseases</i> , 2019 , 14, 184	4.2	10
70	Human papillomavirus (HPV) prevalence and types among women attending regular gynecological visit in Tehran, Iran. <i>Clinical Laboratory</i> , 2014 , 60, 267-73	2	10
69	Clinical Application of Cell-Free DNA Sequencing-Based Noninvasive Prenatal Testing for Trisomies 21, 18, 13 and Sex Chromosome Aneuploidy in a Mixed-Risk Population in Iran. <i>Fetal Diagnosis and Therapy</i> , 2020 , 47, 220-227	2.4	10
68	Identification of novel loss of function variants in MBOAT7 resulting in intellectual disability. <i>Genomics</i> , 2020 , 112, 4072-4077	4.3	9
67	The oncogenic and tumor suppressive roles of RNA-binding proteins in human cancers. <i>Journal of Cellular Physiology</i> , 2021 , 236, 6200-6224	7	9
66	Isolated Congenital Anosmia and CNGA2 Mutation. <i>Scientific Reports</i> , 2017 , 7, 2667	4.9	8
65	Novel neuroclinical findings of autosomal recessive primary microcephaly 15 in a consanguineous Iranian family. <i>European Journal of Medical Genetics</i> , 2020 , 63, 104096	2.6	8
64	Homozygous in-frame variant of SCL6A3 causes dopamine transporter deficiency syndrome in a consanguineous family. <i>Annals of Human Genetics</i> , 2020 , 84, 315-323	2.2	7
63	Novel compound heterozygote mutations in the ATP7B gene in an Iranian family with Wilson disease: a case report. <i>Journal of Medical Case Reports</i> , 2018 , 12, 68	1.2	7
62	S3440P Substitution in C-Terminal Region of Human Reelin Dramatically Impairs Secretion of Reelin from HEK 293T cells. <i>Cellular and Molecular Biology</i> , 2019 , 65, 12	1.1	7
61	Identification of RELN variant p.(Ser2486Gly) in an Iranian family with ankylosing spondylitis; the first association of RELN and AS. <i>European Journal of Human Genetics</i> , 2020 , 28, 754-762	5.3	7

60	Structural model of the OPA1 GTPase domain may explain the molecular consequences of a novel mutation in a family with autosomal dominant optic atrophy. <i>Experimental Eye Research</i> , 2006 , 83, 702-6 ³⁻⁷	3.7	6
59	Novel imaging and clinical phenotypes of CONDSIAS disorder caused by a homozygous frameshift variant of ADPRHL2: a case report. <i>BMC Neurology</i> , 2020 , 20, 291	3.1	6
58	Whole-exome sequencing identified a novel variant in an Iranian patient affected by pycnodysostosis. <i>Molecular Genetics & Genomic Medicine</i> , 2020 , 8, e1118	2.3	5
57	The first case of NSHL by direct impression on gene and identification of one novel mutation in in the Iranian families. <i>Iranian Journal of Basic Medical Sciences</i> , 2018 , 21, 333-341	1.8	5
56	A homozygote variant in the tRNA splicing endonuclease subunit 54 causes pontocerebellar hypoplasia in a consanguineous Iranian family. <i>Molecular Genetics & Genomic Medicine</i> , 2020 , 8, e1413 ²⁻³	2.3	5
55	Primary creatine deficiency syndrome as a potential missed diagnosis in children with psychomotor delay and seizure: case presentation with two novel variants and literature review. <i>Acta Neurologica Belgica</i> , 2020 , 120, 511-516	1.5	5
54	Identification of a novel missense c.386G > A variant in a boy with the POMGNT1-related muscular dystrophy-dystroglycanopathy. <i>Acta Neurologica Belgica</i> , 2021 , 121, 143-151	1.5	5
53	Novel in-frame deletion in MFSD8 gene revealed by trio whole exome sequencing in an Iranian affected with neuronal ceroid lipofuscinosis type 7: a case report. <i>Journal of Medical Case Reports</i> , 2018 , 12, 281	1.2	5
52	S3440P Substitution in C-Terminal Region of Human Reelin Dramatically Impairs Secretion of Reelin from HEK 293T cells. <i>Cellular and Molecular Biology</i> , 2019 , 65, 12-16	1.1	5
51	A novel missense variant in GPT2 causes non-syndromic autosomal recessive intellectual disability in a consanguineous Iranian family. <i>European Journal of Medical Genetics</i> , 2020 , 63, 103853	2.6	4
50	A novel ISCA2 variant responsible for an early-onset neurodegenerative mitochondrial disorder: a case report of multiple mitochondrial dysfunctions syndrome 4. <i>BMC Neurology</i> , 2019 , 19, 153	3.1	4
49	Alopecia-mental retardation syndrome: clinical and molecular characterization of four patients. <i>British Journal of Dermatology</i> , 2008 , 159, 748-51	4	4
48	Biomarker Discovery Based on Hybrid Optimization Algorithm and Artificial Neural Networks on Microarray Data for Cancer Classification. <i>Journal of Medical Signals and Sensors</i> , 2015 , 5, 88-96	1	4
47	Whole-exome sequencing identified first homozygous frameshift variant in the COLEC10 gene in an Iranian patient causing 3MC syndrome type 3. <i>Molecular Genetics & Genomic Medicine</i> , 2021 , 9, e1834	2.3	4
46	A novel variant of ST3GAL3 causes non-syndromic autosomal recessive intellectual disability in Iranian patients. <i>Journal of Gene Medicine</i> , 2020 , 22, e3253	3.5	4
45	A novel missense variant in the LMNB2 gene causes progressive myoclonus epilepsy. <i>Acta Neurologica Belgica</i> , 2021 , 1	1.5	4
44	Distribution of the most common types of HPV in Iranian women with and without cervical cancer. <i>Women and Health</i> , 2021 , 61, 73-82	1.7	4
43	Heterozygosity mapping for human dominant trait variants. <i>Human Mutation</i> , 2019 , 40, 996-1004	4.7	3

42	Three Novel Variants identified in FBN1 and TGFBR2 in seven Iranian families with suspected Marfan syndrome. <i>Molecular Genetics & Genomic Medicine</i> , 2020 , 8, e1274	2.3	3
41	Double Heterozygosity of BRCA2 and STK11 in Familial Breast Cancer Detected by Exome Sequencing. <i>Iranian Journal of Public Health</i> , 2015 , 44, 1348-52	0.7	3
40	Expanding the clinical and neuroimaging features of NKX6-2-related hereditary spastic ataxia type 8. <i>European Journal of Medical Genetics</i> , 2020 , 63, 103868	2.6	3
39	Leukoencephalopathy in RIN2 syndrome: Novel mutation and expansion of clinical spectrum. <i>European Journal of Medical Genetics</i> , 2020 , 63, 103629	2.6	3
38	Evaluation of the association of single nucleotide polymorphisms in DDP4 and CDK5RAP2 genes with rheumatoid arthritis susceptibility in Iranian population. <i>Egyptian Journal of Medical Human Genetics</i> , 2018 , 19, 185-189	2	2
37	Lack of association between btb domain and cnc homolog 2 polymorphism and susceptibility to rheumatoid arthritis in Iranian population. <i>Indian Journal of Rheumatology</i> , 2016 , 11, 197	0.5	2
36	A Novel Locus and Candidate Gene for Familial Developmental Dyslexia on Chromosome 4q. <i>Zeitschrift Für Kinder- Und Jugendpsychiatrie Und Psychotherapie</i> , 2020 , 48, 478-489	1.8	2
35	Novel homozygous variants in the TMC1 and CDH23 genes cause autosomal recessive nonsyndromic hearing loss. <i>Molecular Genetics & Genomic Medicine</i> , 2020 , 8, e1550	2.3	2
34	Identification of novel variants in Iranian consanguineous pedigrees with nonsyndromic hearing loss by next-generation sequencing. <i>Journal of Clinical Laboratory Analysis</i> , 2020 , 34, e23544	3	2
33	A novel homozygous missense variant in the NAXE gene in an Iranian family with progressive encephalopathy with brain edema and leukoencephalopathy. <i>Acta Neurologica Belgica</i> , 2021 , 1	1.5	2
32	Compound Heterozygous Mutations in Gene in an Iranian Child with Microcephaly, Seizures, and Developmental Delay. <i>Fetal and Pediatric Pathology</i> , 2021 , 40, 174-180	1.7	2
31	Kabuki Syndrome: Identification of Two Novel Variants in and. <i>Molecular Syndromology</i> , 2021 , 12, 118-126	5	2
30	Molecular Genetic Analysis of PKHD1 Mutations in Pedigrees With Autosomal Recessive Polycystic Kidney Disease. <i>Iranian Journal of Kidney Diseases</i> , 2018 , 12, 350-358	0.9	2
29	Identification of a novel mutation in the gene associated with ataxia-oculomotor apraxia. <i>Journal of Physical Education and Sports Management</i> , 2017 , 3,	2.8	1
28	Application and effectiveness of ontology on e-Health 2014 ,		1
27	Improving Classification of Cancer and Mining Biomarkers from Gene Expression Profiles Using Hybrid Optimization Algorithms and Fuzzy Support Vector Machine. <i>Journal of Medical Signals and Sensors</i> , 2018 , 8, 1-11	1	1
26	Functional Analysis of S2486G Mutation and its Contribution to Pathogenesis of Ankylosing Spondylitis. <i>Archives of Iranian Medicine</i> , 2020 , 23, 688-696	2.4	1
25	A novel deletion variant in CLN3 with highly variable expressivity is responsible for juvenile neuronal ceroid lipofuscinoses. <i>Acta Neurologica Belgica</i> , 2021 , 121, 737-748	1.5	1

24	A novel metabolic disorder in the degradation pathway of endogenous methanol due to a mutation in the gene of alcohol dehydrogenase. <i>Clinical Biochemistry</i> , 2021 , 90, 66-72	3.5	1
23	Identification of a novel MICU1 nonsense variant causes myopathy with extrapyramidal signs in an Iranian consanguineous family. <i>Molecular and Cellular Pediatrics</i> , 2021 , 8, 6	3.3	1
22	Crystallographic modeling of the PNPT1:c.1453A>G variant as a cause of mitochondrial dysfunction and autosomal recessive deafness; expanding the neuroimaging and clinical features. <i>Mitochondrion</i> , 2021 , 59, 1-7	4.9	1
21	Identification of a Novel Splice Site Mutation in RUNX2 Gene in a Family with Rare Autosomal Dominant Cleidocranial Dysplasia. <i>Iranian Biomedical Journal</i> , 2021 , 25, 297-302	2	1
20	Prevalence and Genotype Distribution of Human Papillomavirus Infection among 12 076 Iranian Women. <i>International Journal of Infectious Diseases</i> , 2021 , 111, 295-302	10.5	1
19	Pharmacogenetics and Personalized Medicine in Pancreatic Cancer. <i>Acta Medica Iranica</i> , 2017 , 55, 194-199		1
18	A novel deletion mutation in GUCY2D gene may be responsible for Leber congenital amaurosis-1 disease: A case report. <i>Journal of Current Ophthalmology</i> , 2019 , 31, 458-462	2	0
17	Novel compound heterozygous variants in XYLT1 gene caused Desbuquois dysplasia type 2 in an aborted fetus: a case report.. <i>BMC Pediatrics</i> , 2022 , 22, 63	2.6	0
16	Molecular genetic analysis of polycystic kidney disease 1 and polycystic kidney disease 2 mutations in pedigrees with autosomal dominant polycystic kidney disease. <i>Journal of Research in Medical Sciences</i> , 2019 , 24, 44	1.6	0
15	Leukoencephalopathy in Al-Raqad syndrome: Expanding the clinical and neuroimaging features caused by a biallelic novel missense variant in DCPS. <i>American Journal of Medical Genetics, Part A</i> , 2020 , 182, 2391-2398	2.5	0
14	Novel variants in critical domains of ATP8A2 and expansion of clinical spectrum. <i>Human Mutation</i> , 2021 , 42, 491-497	4.7	0
13	Novel manifestations of Warburg micro syndrome type 1 caused by a new splicing variant of RAB3GAP1: a case report. <i>BMC Neurology</i> , 2021 , 21, 180	3.1	0
12	Defective complex III mitochondrial respiratory chain due to a novel variant in CYC1 gene masquerades acute demyelinating syndrome or Leber hereditary optic neuropathy. <i>Mitochondrion</i> , 2021 , 60, 12-20	4.9	0
11	Evidence of involvement of a novel VUS variant in the CHKB gene to congenital muscular dystrophy affection. <i>Meta Gene</i> , 2020 , 24, 100698	0.7	
10	Association between rs6759298 and Ankylosing Spondylitis in Iranian Population. <i>Avicenna Journal of Medical Biotechnology</i> , 2018 , 10, 178-182	1.4	
9	Biomarker Discovery by Imperialist Competitive Algorithm in Mass Spectrometry Data for Ovarian Cancer Prediction. <i>Journal of Medical Signals and Sensors</i> , 2021 , 11, 108-119	1	
8	Importance of miR-141-5p and miR-501-5P expression in patients with HBV infection.. <i>Cellular and Molecular Biology</i> , 2021 , 67, 184-189	1.1	
7	Identification of a novel Mutation in an Iranian Family with suspected patient to GM2-gangliosidosis. <i>Clinical Case Reports (discontinued)</i> , 2020 , 8, 2583-2591	0.7	

6	ACER3-related leukoencephalopathy: expanding the clinical and imaging findings spectrum due to novel variants. <i>Human Genomics</i> , 2021 , 15, 45	6.8
5	The Identification and Stereochemistry Analysis of a Novel Mutation p.(D367Tfs*61) in the CYP1B1 Gene: A Case Report. <i>Journal of Current Ophthalmology</i> , 2020 , 32, 114-118	2
4	Possible dual contribution of a novel GUCY2D mutation in the development of retinal degeneration in a consanguineous population. <i>European Journal of Medical Genetics</i> , 2020 , 63, 103750	2.6
3	The identification of two pathogenic variants in a family with mild and severe forms of developmental delay. <i>Journal of Human Genetics</i> , 2021 , 66, 445-448	4.3
2	Nonsyndromic Early-Onset Epileptic Encephalopathies: Two Novel KCTD7 Pathogenic Variants and a Literature Review. <i>Developmental Neuroscience</i> , 2021 , 43, 348-357	2.2
1	Neurodevelopmental disorder with microcephaly, hypotonia, and variable brain anomalies in a consanguineous Iranian family is associated with a homozygous start loss variant in the PRUNE1 gene.. <i>BMC Medical Genomics</i> , 2022 , 15, 78	3.7