Masoud Garshasbi

List of Publications by Year in Descending Order

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	Novel compound heterozygous variants in XYLT1 gene caused Desbuquois dysplasia type 2 in an aborted fetus: a case report.. *BMC Pediatrics*, **2022**, 22, 63	2.6	0
112	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.. *BMC Medical Genomics*, **2022**, 15, 78	3.7	
111	Biomarker Discovery by Imperialist Competitive Algorithm in Mass Spectrometry Data for Ovarian Cancer Prediction. *Journal of Medical Signals and Sensors*, **2021**, 11, 108-119	1	
110	Importance of miR-141-5p and miR-501-5P expression in patients with HBV infection.. *Cellular and Molecular Biology*, **2021**, 67, 184-189	1.1	
109	Whole-exome sequencing identified first homozygous frameshift variant in the COLEC10 gene in an Iranian patient causing 3MC syndrome type 3. *Molecular Genetics & Genomic Medicine*, **2021**, 9, e1834	2.3	4
108	A novel deletion variant in CLN3 with highly variable expressivity is responsible for juvenile neuronal ceroid lipofuscinoses. *Acta Neurologica Belgica*, **2021**, 121, 737-748	1.5	1
107	A novel missense variant in the LMNB2 gene causes progressive myoclonus epilepsy. *Acta Neurologica Belgica*, **2021**, 1	1.5	4
106	Novel variants in critical domains of ATP8A2 and expansion of clinical spectrum. *Human Mutation*, **2021**, 42, 491-497	4.7	0
105	A novel metabolic disorder in the degradation pathway of endogenous methanol due to a mutation in the gene of alcohol dehydrogenase. *Clinical Biochemistry*, **2021**, 90, 66-72	3.5	1
104	Novel manifestations of Warburg micro syndrome type 1 caused by a new splicing variant of RAB3GAP1: a case report. *BMC Neurology*, **2021**, 21, 180	3.1	0
103	Identification of a novel MICU1 nonsense variant causes myopathy with extrapyramidal signs in an Iranian consanguineous family. *Molecular and Cellular Pediatrics*, **2021**, 8, 6	3.3	1
102	A novel homozygous missense variant in the NAXE gene in an Iranian family with progressive encephalopathy with brain edema and leukoencephalopathy. *Acta Neurologica Belgica*, **2021**, 1	1.5	2
101	ACER3-related leukoencephalopathy: expanding the clinical and imaging findings spectrum due to novel variants. *Human Genomics*, **2021**, 15, 45	6.8	
100	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. *Mitochondrion*, **2021**, 59, 1-7	4.9	1
99	The identification of two pathogenic variants in a family with mild and severe forms of developmental delay. *Journal of Human Genetics*, **2021**, 66, 445-448	4.3	
98	Identification of a novel missense c.386G > A variant in a boy with the POMGNT1-related muscular dystrophy-dystroglycanopathy. *Acta Neurologica Belgica*, **2021**, 121, 143-151	1.5	5
97	Distribution of the most common types of HPV in Iranian women with and without cervical cancer. *Women and Health*, **2021**, 61, 73-82	1.7	4

96	Compound Heterozygous Mutations in Gene in an Iranian Child with Microcephaly, Seizures, and Developmental Delay. *Fetal and Pediatric Pathology*, **2021**, 40, 174-180	1.7	2
95	Kabuki Syndrome: Identification of Two Novel Variants in and. *Molecular Syndromology*, **2021**, 12, 118-126	1.5	2
94	The oncogenic and tumor suppressive roles of RNA-binding proteins in human cancers. *Journal of Cellular Physiology*, **2021**, 236, 6200-6224	7	9
93	Identification of a Novel Splice Site Mutation in RUNX2 Gene in a Family with Rare Autosomal Dominant Cleidocranial Dysplasia. *Iranian Biomedical Journal*, **2021**, 25, 297-302	2	1
92	Defective complex III mitochondrial respiratory chain due to a novel variant in CYC1 gene masquerades acute demyelinating syndrome or Leber hereditary optic neuropathy. *Mitochondrion*, **2021**, 60, 12-20	4.9	0
91	Nonsyndromic Early-Onset Epileptic Encephalopathies: Two Novel KCTD7 Pathogenic Variants and a Literature Review. *Developmental Neuroscience*, **2021**, 43, 348-357	2.2	
90	Prevalence and Genotype Distribution of Human Papillomavirus Infection among 12 076 Iranian Women. *International Journal of Infectious Diseases*, **2021**, 111, 295-302	10.5	1
89	Evidence of involvement of a novel VUS variant in the CHKB gene to congenital muscular dystrophy affection. *Meta Gene*, **2020**, 24, 100698	0.7	
88	Three Novel Variants identified in FBN1 and TGFBR2 in seven Iranian families with suspected Marfan syndrome. *Molecular Genetics & Genomic Medicine*, **2020**, 8, e1274	2.3	3
87	Identification of novel loss of function variants in MBOAT7 resulting in intellectual disability. *Genomics*, **2020**, 112, 4072-4077	4.3	9
86	Homozygous in-frame variant of SCL6A3 causes dopamine transporter deficiency syndrome in a consanguineous family. *Annals of Human Genetics*, **2020**, 84, 315-323	2.2	7
85	Whole-exome sequencing identified a novel variant in an Iranian patient affected by pycnodysostosis. *Molecular Genetics & Genomic Medicine*, **2020**, 8, e1118	2.3	5
84	A novel missense variant in GPT2 causes non-syndromic autosomal recessive intellectual disability in a consanguineous Iranian family. *European Journal of Medical Genetics*, **2020**, 63, 103853	2.6	4
83	Functional Analysis of S2486G Mutation and its Contribution to Pathogenesis of Ankylosing Spondylitis. *Archives of Iranian Medicine*, **2020**, 23, 688-696	2.4	1
82	A Novel Locus and Candidate Gene for Familial Developmental Dyslexia on Chromosome 4q. *Zeitschrift F Kinder- Und Jugendpsychiatrie Und Psychotherapie*, **2020**, 48, 478-489	1.8	2
81	Identification of RELN variant p.(Ser2486Gly) in an Iranian family with ankylosing spondylitis; the first association of RELN and AS. *European Journal of Human Genetics*, **2020**, 28, 754-762	5.3	7
80	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. *Fetal Diagnosis and Therapy*, **2020**, 47, 220-227	2.4	10
79	An update on clinical, pathological, diagnostic, and therapeutic perspectives of childhood leukodystrophies. *Expert Review of Neurotherapeutics*, **2020**, 20, 65-84	4.3	17

78	A novel variant of ST3GAL3 causes non-syndromic autosomal recessive intellectual disability in Iranian patients. *Journal of Gene Medicine*, **2020**, 22, e3253	3.5	4
77	Expanding the clinical and neuroimaging features of NKX6-2-related hereditary spastic ataxia type 8. *European Journal of Medical Genetics*, **2020**, 63, 103868	2.6	3
76	Novel homozygous variants in the TMC1 and CDH23 genes cause autosomal recessive nonsyndromic hearing loss. *Molecular Genetics & Genomic Medicine*, **2020**, 8, e1550	2.3	2
75	Novel neuroclinical findings of autosomal recessive primary microcephaly 15 in a consanguineous Iranian family. *European Journal of Medical Genetics*, **2020**, 63, 104096	2.6	8
74	Leukoencephalopathy in Al-Raqad syndrome: Expanding the clinical and neuroimaging features caused by a biallelic novel missense variant in DCPS. *American Journal of Medical Genetics, Part A*, **2020**, 182, 2391-2398	2.5	0
73	Novel imaging and clinical phenotypes of CONDSIAS disorder caused by a homozygous frameshift variant of ADPRHL2: a case report. *BMC Neurology*, **2020**, 20, 291	3.1	6
72	Identification of a novel Mutation in an Iranian Family with suspected patient to GM2-gangliosidoses. *Clinical Case Reports (discontinued)*, **2020**, 8, 2583-2591	0.7	
71	A homozygote variant in the tRNA splicing endonuclease subunit 54 causes pontocerebellar hypoplasia in a consanguineous Iranian family. *Molecular Genetics & Genomic Medicine*, **2020**, 8, e1413	2.3	5
70	Identification of novel variants in Iranian consanguineous pedigrees with nonsyndromic hearing loss by next-generation sequencing. *Journal of Clinical Laboratory Analysis*, **2020**, 34, e23544	3	2
69	The Identification and Stereochemistry Analysis of a Novel Mutation p.(D367Tfs*61) in the CYP1B1 Gene: A Case Report. *Journal of Current Ophthalmology*, **2020**, 32, 114-118	2	
68	Leukoencephalopathy in RIN2 syndrome: Novel mutation and expansion of clinical spectrum. *European Journal of Medical Genetics*, **2020**, 63, 103629	2.6	3
67	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. *Acta Neurologica Belgica*, **2020**, 120, 511-516	1.5	5
66	Aberrant expression of a five-microRNA signature in breast carcinoma as a promising biomarker for diagnosis. *Journal of Clinical Laboratory Analysis*, **2020**, 34, e23063	3	14
65	Possible dual contribution of a novel GUCY2D mutation in the development of retinal degeneration in a consanguineous population. *European Journal of Medical Genetics*, **2020**, 63, 103750	2.6	
64	Non-Coding RNAs in Cartilage Development: An Updated Review. *International Journal of Molecular Sciences*, **2019**, 20,	6.3	34
63	Heterozygosity mapping for human dominant trait variants. *Human Mutation*, **2019**, 40, 996-1004	4.7	3
62	Genetic implications in the pathogenesis of rheumatoid arthritis; an updated review. *Gene*, **2019**, 702, 8-16	3.8	70
61	Identifying occult maternal malignancies from 1.93 million pregnant women undergoing noninvasive prenatal screening tests. *Genetics in Medicine*, **2019**, 21, 2293-2302	8.1	21

60	A novel deletion mutation in GUCY2D gene may be responsible for Leber congenital amaurosis-1 disease: A case report. *Journal of Current Ophthalmology*, **2019**, 31, 458-462	2	0
59	A novel ISCA2 variant responsible for an early-onset neurodegenerative mitochondrial disorder: a case report of multiple mitochondrial dysfunctions syndrome 4. *BMC Neurology*, **2019**, 19, 153	3.1	4
58	RNASET2-deficient leukoencephalopathy mimicking congenital CMV infection and Aicardi-Goutieres syndrome: a case report with a novel pathogenic variant. *Orphanet Journal of Rare Diseases*, **2019**, 14, 184	4.2	10
57	S3440P Substitution in C-Terminal Region of Human Reelin Dramatically Impairs Secretion of Reelin from HEK 293T cells. *Cellular and Molecular Biology*, **2019**, 65, 12	1.1	7
56	Molecular genetic analysis of polycystic kidney disease 1 and polycystic kidney disease 2 mutations in pedigrees with autosomal dominant polycystic kidney disease. *Journal of Research in Medical Sciences*, **2019**, 24, 44	1.6	0
55	S3440P Substitution in C-Terminal Region of Human Reelin Dramatically Impairs Secretion of Reelin from HEK 293T cells. *Cellular and Molecular Biology*, **2019**, 65, 12-16	1.1	5
54	Novel compound heterozygote mutations in the ATP7B gene in an Iranian family with Wilson disease: a case report. *Journal of Medical Case Reports*, **2018**, 12, 68	1.2	7
53	Evaluation of the association of single nucleotide polymorphisms in DDP4 and CDK5RAP2 genes with rheumatoid arthritis susceptibility in Iranian population. *Egyptian Journal of Medical Human Genetics*, **2018**, 19, 185-189	2	2
52	Whole-exome sequencing identifies R1279X of MYH6 gene to be associated with congenital heart disease. *BMC Cardiovascular Disorders*, **2018**, 18, 137	2.3	12
51	Improving Classification of Cancer and Mining Biomarkers from Gene Expression Profiles Using Hybrid Optimization Algorithms and Fuzzy Support Vector Machine. *Journal of Medical Signals and Sensors*, **2018**, 8, 1-11	1	1
50	Association between rs6759298 and Ankylosing Spondylitis in Iranian Population. *Avicenna Journal of Medical Biotechnology*, **2018**, 10, 178-182	1.4	
49	The first case of NSHL by direct impression on gene and identification of one novel mutation in in the Iranian families. *Iranian Journal of Basic Medical Sciences*, **2018**, 21, 333-341	1.8	5
48	miR-31 and miR-145 as Potential Non-Invasive Regulatory Biomarkers in Patients with Endometriosis. *Cell Journal*, **2018**, 20, 84-89	2.4	16
47	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. *Journal of Medical Case Reports*, **2018**, 12, 281	1.2	5
46	Molecular Genetic Analysis of PKHD1 Mutations in Pedigrees With Autosomal Recessive Polycystic Kidney Disease. *Iranian Journal of Kidney Diseases*, **2018**, 12, 350-358	0.9	2
45	Mutations of the aminoacyl-tRNA-synthetases SARS and WARS2 are implicated in the etiology of autosomal recessive intellectual disability. *Human Mutation*, **2017**, 38, 621-636	4.7	40
44	Identification of a novel mutation in the gene associated with ataxia-oculomotor apraxia. *Journal of Physical Education and Sports Management*, **2017**, 3,	2.8	1
43	Isolated Congenital Anosmia and CNGA2 Mutation. *Scientific Reports*, **2017**, 7, 2667	4.9	8

42	Promoter hypermethylation of BCL11B gene correlates with downregulation of gene transcription in ankylosing spondylitis patients. *Genes and Immunity*, **2017**, 18, 170-175	4.4	36
41	PTRHD1 (C2orf79) mutations lead to autosomal-recessive intellectual disability and parkinsonism. *Movement Disorders*, **2017**, 32, 287-291	7	23
40	Identification of miR-24 and miR-137 as novel candidate multiple sclerosis miRNA biomarkers using multi-staged data analysis protocol. *Molecular Biology Research Communications*, **2017**, 6, 127-140	1.6	14
39	Pharmacogenetics and Personalized Medicine in Pancreatic Cancer. *Acta Medica Iranica*, **2017**, 55, 194-199		1
38	Missense variants in AIMP1 gene are implicated in autosomal recessive intellectual disability without neurodegeneration. *European Journal of Human Genetics*, **2016**, 24, 392-9	5.3	14
37	Evaluation of DNMT1 gene expression profile and methylation of its promoter region in patients with ankylosing spondylitis. *Clinical Rheumatology*, **2016**, 35, 2723-2731	3.9	46
36	Determination of IL1 R2, ANTXR2, CARD9, and SNAPC4 single nucleotide polymorphisms in Iranian patients with ankylosing spondylitis. *Rheumatology International*, **2016**, 36, 429-35	3.6	19
35	Lack of association between btb domain and cnc homolog 2 polymorphism and susceptibility to rheumatoid arthritis in Iranian population. *Indian Journal of Rheumatology*, **2016**, 11, 197	0.5	2
34	Mutations in the histamine N-methyltransferase gene, HNMT, are associated with nonsyndromic autosomal recessive intellectual disability. *Human Molecular Genetics*, **2015**, 24, 5697-710	5.6	19
33	A defect in the CLIP1 gene (CLIP-170) can cause autosomal recessive intellectual disability. *European Journal of Human Genetics*, **2015**, 23, 331-6	5.3	15
32	Biomarker Discovery Based on Hybrid Optimization Algorithm and Artificial Neural Networks on Microarray Data for Cancer Classification. *Journal of Medical Signals and Sensors*, **2015**, 5, 88-96	1	4
31	Double Heterozygosity of BRCA2 and STK11 in Familial Breast Cancer Detected by Exome Sequencing. *Iranian Journal of Public Health*, **2015**, 44, 1348-52	0.7	3
30	Application and effectiveness of ontology on e-Health **2014**,		1
29	Human papillomavirus (HPV) prevalence and types among women attending regular gynecological visit in Tehran, Iran. *Clinical Laboratory*, **2014**, 60, 267-73	2	10
28	A novel ALDH5A1 mutation is associated with succinic semialdehyde dehydrogenase deficiency and severe intellectual disability in an Iranian family. *American Journal of Medical Genetics, Part A*, **2013**, 161A, 1915-22	2.5	14
27	Mutations in NSUN2 cause autosomal-recessive intellectual disability. *American Journal of Human Genetics*, **2012**, 90, 847-55	11	179
26	New kid on the ID block: neural functions of the Nab2/ZC3H14 class of CysHis tandem zinc-finger polyadenosine RNA binding proteins. *RNA Biology*, **2012**, 9, 555-62	4.8	13
25	Deep sequencing reveals 50 novel genes for recessive cognitive disorders. *Nature*, **2011**, 478, 57-63	50.4	649

24	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, 115-7	5.3	45
23	Mutations in the alpha 1,2-mannosidase gene, MAN1B1, cause autosomal-recessive intellectual disability. *American Journal of Human Genetics*, **2011**, 89, 176-82	11	60
22	Mutations in the Alpha 1,2-Mannosidase Gene, MAN1B1, Cause Autosomal-Recessive Intellectual Disability. *American Journal of Human Genetics*, **2011**, 89, 348	11	78
21	ST3GAL3 mutations impair the development of higher cognitive functions. *American Journal of Human Genetics*, **2011**, 89, 407-14	11	70
20	Autosomal recessive mental retardation: homozygosity mapping identifies 27 single linkage intervals, at least 14 novel loci and several mutation hotspots. *Human Genetics*, **2011**, 129, 141-8	6.3	40
19	A novel nonsense mutation in TUSC3 is responsible for non-syndromic autosomal recessive mental retardation in a consanguineous Iranian family. *American Journal of Medical Genetics, Part A*, **2011**, 155A, 1976-80	2.5	35
18	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, 12390-5	11.5	61
17	Identification of a novel candidate gene for non-syndromic autosomal recessive intellectual disability: the WASH complex member SWIP. *Human Molecular Genetics*, **2011**, 20, 2585-90	5.6	60
16	Cohen syndrome diagnosis using whole genome arrays. *Journal of Medical Genetics*, **2011**, 48, 136-40	5.8	28
15	Establishment of a mouse model with misregulated chromosome condensation due to defective Mcph1 function. *PLoS ONE*, **2010**, 5, e9242	3.7	43
14	Somatic mutation profiles of MSI and MSS colorectal cancer identified by whole exome next generation sequencing and bioinformatics analysis. *PLoS ONE*, **2010**, 5, e15661	3.7	169
13	A clinical and molecular genetic study of 112 Iranian families with primary microcephaly. *Journal of Medical Genetics*, **2010**, 47, 823-8	5.8	72
12	Cranioectodermal Dysplasia, Sensenbrenner syndrome, is a ciliopathy caused by mutations in the IFT122 gene. *American Journal of Human Genetics*, **2010**, 86, 949-56	11	154
11	CA8 mutations cause a novel syndrome characterized by ataxia and mild mental retardation with predisposition to quadrupedal gait. *PLoS Genetics*, **2009**, 5, e1000487	6	99
10	Expanded mutational spectrum in Cohen syndrome, tissue expression, and transcript variants of COH1. *Human Mutation*, **2009**, 30, E404-20	4.7	33
9	An autosomal recessive syndrome of severe mental retardation, cataract, coloboma and kyphosis maps to the pericentromeric region of chromosome 4. *European Journal of Human Genetics*, **2009**, 17, 125-8	5.3	15
8	Identification of a nonsense mutation in the very low-density lipoprotein receptor gene (VLDLR) in an Iranian family with dysequilibrium syndrome. *European Journal of Human Genetics*, **2008**, 16, 270-3	5.3	44
7	Alopecia-mental retardation syndrome: clinical and molecular characterization of four patients. *British Journal of Dermatology*, **2008**, 159, 748-51	4	4

6	A defect in the TUSC3 gene is associated with autosomal recessive mental retardation. *American Journal of Human Genetics*, **2008**, 82, 1158-64	11	107
5	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, 43-8	6.3	79
4	A defect in the ionotropic glutamate receptor 6 gene (GRIK2) is associated with autosomal recessive mental retardation. *American Journal of Human Genetics*, **2007**, 81, 792-8	11	116
3	Structural model of the OPA1 GTPase domain may explain the molecular consequences of a novel mutation in a family with autosomal dominant optic atrophy. *Experimental Eye Research*, **2006**, 83, 702-6	3.7	6
2	SNP array-based homozygosity mapping reveals MCPH1 deletion in family with autosomal recessive mental retardation and mild microcephaly. *Human Genetics*, **2006**, 118, 708-15	6.3	62
1	alpha-globin gene deletion and point mutation analysis among in Iranian patients with microcytic hypochromic anemia. *Haematologica*, **2003**, 88, 1196-7	6.6	19