Hossein Najmabadi

List of Publications by Year
in descending order

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265 papers	9,847 citations	43973 48 h-index	53109 85 g-index
276 all docs	276 docs citations	276 times ranked	14351 citing authors

#	Article	IF	Citations
1	Deep sequencing reveals 50 novel genes for recessive cognitive disorders. Nature, 2011, 478, 57-63.	13.7	805
2	LRP6 Mutation in a Family with Early Coronary Disease and Metabolic Risk Factors. Science, 2007, 315, 1278-1282.	6.0	567
3	Biallelic mutations in the death domain of PIDD1 impair caspase-2 activation and are associated with intellectual disability. Translational Psychiatry, 2021, 11, 1.	2.4	334
4	Epigenetically Deregulated microRNA-375 Is Involved in a Positive Feedback Loop with Estrogen Receptor Î± in Breast Cancer Cells. Cancer Research, 2010, 70, 9175-9184.	0.4	260
5	Mutations in NSUN2 Cause Autosomal- Recessive Intellectual Disability. American Journal of Human Genetics, 2012, 90, 847-855.	2.6	243
6	Human Male Infertility Caused by Mutations in the CATSPER1 Channel Protein. American Journal of Human Genetics, 2009, 84, 505-510.	2.6	206
7	Thalassemia in Iran. Journal of Pediatric Hematology/Oncology, 2007, 29, 233-238.	0.3	181
8	A Forward Genetics Screen in Mice Identifies Recessive Deafness Traits and Reveals That Pejvakin Is Essential for Outer Hair Cell Function. Journal of Neuroscience, 2007, 27, 2163-2175.	1.7	159
9	Hexosamine Biosynthetic Pathway Mutations Cause Neuromuscular Transmission Defect. American Journal of Human Genetics, 2011, 88, 162-172.	2.6	153
10	Genetic male infertility and mutation of CATSPER ion channels. European Journal of Human Genetics, 2010, 18, 1178-1184.	1.4	139
11	A Defect in the Ionotropic Glutamate Receptor 6 Gene (GRIK2) Is Associated with Autosomal Recessive Mental Retardation. American Journal of Human Genetics, 2007, 81, 792-798.	2.6	137
12	Utilizing Ethnic-Specific Differences in Minor Allele Frequency to Recategorize Reported Pathogenic Deafness Variants. American Journal of Human Genetics, 2014, 95, 445-453.	2.6	137
13	THE Î²-THALASSEMIA MUTATION SPECTRUM IN THE IRANIAN POPULATION. Hemoglobin, 2001, 25, 285-296.	0.4	134
14	Substantial prevalence of microdeletions of the Y-chromosome in infertile men with idiopathic azoospermia and oligozoospermia detected using a sequence-tagged site-based mapping strategy. Journal of Clinical Endocrinology and Metabolism, 1996, 81, 1347-1352.	1.8	133
15	Genetics of intellectual disability in consanguineous families. Molecular Psychiatry, 2019, 24, 1027-1039.	4.1	131
16	Mutations in LOXHD1, an Evolutionarily Conserved Stereociliary Protein, Disrupt Hair Cell Function in Mice and Cause Progressive Hearing Loss in Humans. American Journal of Human Genetics, 2009, 85, 328-337.	2.6	129
17	A Defect in the TUSC3 Gene Is Associated with Autosomal Recessive Mental Retardation. American Journal of Human Genetics, 2008, 82, 1158-1164.	2.6	127
18	Identification of Mutations in TRAPPC9, which Encodes the NIK- and IKK-Î²-Binding Protein, in Nonsyndromic Autosomal-Recessive Mental Retardation. American Journal of Human Genetics, 2009, 85, 909-915.	2.6	120

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19	Ethnic-specific distribution of mutations in 716 patients with congenital adrenal hyperplasia owing to 21-hydroxylase deficiency. Molecular Genetics and Metabolism, 2007, 90, 414-421.	0.5	116
20	Iranome: A catalog of genomic variations in the Iranian population. Human Mutation, 2019, 40, 1968-1984.	1.1	116
21	Loss-of-Function Mutations of ILDR1 Cause Autosomal-Recessive Hearing Impairment DFNB42. American Journal of Human Genetics, 2011, 88, 127-137.	2.6	108
22	Sensorineural deafness and male infertility: a contiguous gene deletion syndrome. Journal of Medical Genetics, 2007, 44, 233-240.	1.5	98
23	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, 43-48.	1.8	92
24	ST3GAL3 Mutations Impair the Development of Higher Cognitive Functions. American Journal of Human Genetics, 2011, 89, 407-414.	2.6	89
25	A clinical and molecular genetic study of 112 Iranian families with primary microcephaly. Journal of Medical Genetics, 2010, 47, 823-828.	1.5	87
26	Characterising the spectrum of autosomal recessive hereditary hearing loss in Iran. Journal of Medical Genetics, 2015, 52, 823-829.	1.5	87
27	GJB2 mutations: Passage through Iran. American Journal of Medical Genetics, Part A, 2005, 133A, 132-137.	0.7	77
28	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, 12390-12395.	3.3	77
29	Mutation of COL11A2 causes autosomal recessive non-syndromic hearing loss at the DFNB53 locus. Journal of Medical Genetics, 2005, 42, e61-e61.	1.5	75
30	Association of AKT1 haplotype with the risk of schizophrenia in Iranian population. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2006, 141B, 383-386.	1.1	74
31	Mutations in the Alpha 1,2-Mannosidase Gene, MAN1B1, Cause Autosomal-Recessive Intellectual Disability. American Journal of Human Genetics, 2011, 89, 176-182.	2.6	73
32	Reducing the exome search space for Mendelian diseases using genetic linkage analysis of exome genotypes. Genome Biology, 2011, 12, R85.	13.9	72
33	Congenital myasthenic syndrome with tubular aggregates caused by GFPT1 mutations. Journal of Neurology, 2012, 259, 838-850.	1.8	72
34	SNP array-based homozygosity mapping reveals MCPH1 deletion in family with autosomal recessive mental retardation and mild microcephaly. Human Genetics, 2006, 118, 708-715.	1.8	67
35	Update of the pompe disease mutation database with 60 novel GAA sequence variants and additional studies on the functional effect of 34 previously reported variants. Human Mutation, 2012, 33, 1161-1165.	1.1	67
36	CIB2, defective in isolated deafness, is key for auditory hair cell mechanotransduction and survival. EMBO Molecular Medicine, 2017, 9, 1711-1731.	3.3	66

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37	A novel homozygous missense mutation in the GNE gene of a patient with quadriceps-sparing hereditary inclusion body myopathy associated with muscle inflammation. Neuromuscular Disorders, 2003, 13, 830-834.	0.3	65
38	A catechol- O -methyltransferase that is essential for auditory function in mice and humans. Proceedings of the National Academy of Sciences of the United States of America, 2008, 105, 14609-14614.	3.3	62
39	FASN-Dependent Lipid Metabolism Links Neurogenic Stem/Progenitor Cell Activity to Learning and Memory Deficits. Cell Stem Cell, 2020, 27, 98-109.e11.	5.2	62
40	Structure and Organization of theRBMYGenes on the Human Y Chromosome: Transposition and Amplification of an Ancestral AutosomalhnRNPGGene. Genomics, 1998, 49, 283-289.	1.3	57
41	The Role of a Novel TRMT1 Gene Mutation and Rare GRM1 Gene Defect in Intellectual Disability in Two Azeri Families. PLoS ONE, 2015, 10, e0129631.	1.1	56
42	Fourteen-Year Experience of Prenatal Diagnosis of Thalassemia in Iran. Public Health Genomics, 2006, 9, 93-97.	0.6	55
43	A comprehensive study to determine heterogeneity of autosomal recessive nonsyndromic hearing loss in Iran. American Journal of Medical Genetics, Part A, 2012, 158A, 2485-2492.	0.7	55
44	Screening for MYO15A gene mutations in autosomal recessive nonsyndromic, GJB2 negative Iranian deaf population. American Journal of Medical Genetics, Part A, 2012, 158A, 1857-1864.	0.7	54
45	PDZD7 and hearing loss: More than just a modifier. American Journal of Medical Genetics, Part A, 2015, 167, 2957-2965.	0.7	54
46	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.	1.1	54
47	Redefining the MED13L syndrome. European Journal of Human Genetics, 2015, 23, 1308-1317.	1.4	53
48	Elucidating the spectrum of Â-thalassemia mutations in Iran. Haematologica, 2007, 92, 992-993.	1.7	52
49	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, 115-117.	1.4	52
50	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, 270-273.	1.4	50
51	Mutations in Grxcr1 Are The Basis for Inner Ear Dysfunction in the Pirouette Mouse. American Journal of Human Genetics, 2010, 86, 148-160.	2.6	49
52	Hydroxyurea responsiveness in Â-thalassemic patients is determined by the stress response adaptation of erythroid progenitors and their differentiation propensity. Haematologica, 2013, 98, 696-704.	1.7	49
53	CDC14A phosphatase is essential for hearing and male fertility in mouse and human. Human Molecular Genetics, 2018, 27, 780-798.	1.4	49
54	Effect of inbreeding on intellectual disability revisited by trio sequencing. Clinical Genetics, 2019, 95, 151-159.	1.0	49

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55	Mutations in the first MyTH4 domain of MYO15A are a common cause of DFNB3 hearing loss. Laryngoscope, 2009, 119, 727-733.	1.1	48
56	Old gene, new phenotype: splice-altering variants in CEACAM16 cause recessive non-syndromic hearing impairment. Journal of Medical Genetics, 2018, 55, 555-560.	1.5	48
57	NPHP4 Variants Are Associated With Pleiotropic Heart Malformations. Circulation Research, 2012, 110, 1564-1574.	2.0	46
58	Variants in CIB2 cause DFNB48 and not USH1J. Clinical Genetics, 2018, 93, 812-821.	1.0	46
59	Autosomal recessive mental retardation: homozygosity mapping identifies 27 single linkage intervals, at least 14 novel loci and several mutation hotspots. Human Genetics, 2011, 129, 141-148.	1.8	45
60	Genetics of non-syndromic hearing loss in the Middle East. International Journal of Pediatric Otorhinolaryngology, 2014, 78, 2026-2036.	0.4	45
61	De novo and inherited mutations in the X-linked gene CLCN4 are associated with syndromic intellectual disability and behavior and seizure disorders in males and females. Molecular Psychiatry, 2018, 23, 222-230.	4.1	45
62	Expanded mutational spectrum in Cohen syndrome, tissue expression, and transcript variants ofCOH1. Human Mutation, 2009, 30, E404-E420.	1.1	44
63	Exonic mutations and exon skipping: Lessons learned from DFNA5. Human Mutation, 2018, 39, 433-440.	1.1	44
64	Identification of three novelTECTA mutations in Iranian families with autosomal recessive nonsyndromic hearing impairment at the DFNB21 locus. American Journal of Medical Genetics, Part A, 2007, 143A, 1623-1629.	0.7	43
65	Î±-Thalassemia Mutation Analyses in Mazandaran Province, North Iran. Hemoglobin, 2009, 33, 115-123.	0.4	43
66	A novel nonsense mutation in TUSC3 is responsible for nonâ€syndromic autosomal recessive mental retardation in a consanguineous Iranian family. American Journal of Medical Genetics, Part A, 2011, 155, 1976-1980.	0.7	43
67	Molecular Characterization of Î±-Thalassemia in the Dohuk Region of Iraq. Hemoglobin, 2009, 33, 37-44.	0.4	41
68	Improved diagnostic yield of neuromuscular disorders applying clinical exome sequencing in patients arising from a consanguineous population. Clinical Genetics, 2017, 91, 386-402.	1.0	41
69	GJB2 mutations in Iranians with autosomal recessive non-syndromic sensorineural hearing loss. Human Mutation, 2002, 19, 572-572.	1.1	40
70	Homozygous THAP1 mutations as cause of earlyâ€onset generalized dystonia. Movement Disorders, 2011, 26, 858-861.	2.2	39
71	Homozygous Truncating Variants in TBC1D23 Cause Pontocerebellar Hypoplasia and Alter Cortical Development. American Journal of Human Genetics, 2017, 101, 428-440.	2.6	39
72	The spectrum of GJB2 mutations in the Iranian population with non-syndromic hearing lossâ€”A twelve year study. International Journal of Pediatric Otorhinolaryngology, 2012, 76, 1164-1174.	0.4	38

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73	Variable hearing impairment in a DFNB2 family with a novel MYO7A missense mutation. Clinical Genetics, 2010, 77, 563-571.	1.0	37
74	A large deletion in GPR98 causes type IIC Usher syndrome in male and female members of an Iranian family. Journal of Medical Genetics, 2009, 46, 272-276.	1.5	36
75	A Mutation Causes MuSK Reduced Sensitivity to Agrin and Congenital Myasthenia. PLoS ONE, 2013, 8, e53826.	1.1	36
76	The XmnI andBCL11ASingle Nucleotide Polymorphisms May Help Predict Hydroxyurea Response in Iranian Î²-Thalassemia Patients. Hemoglobin, 2012, 36, 371-380.	0.4	35
77	The frequency of GJB2 mutations and the Î” (GJB6-D13S1830) deletion as a cause of autosomal recessive non-syndromic deafness in the Kurdish population. Clinical Genetics, 2004, 65, 506-508.	1.0	34
78	NDST1 missense mutations in autosomal recessive intellectual disability. American Journal of Medical Genetics, Part A, 2014, 164, 2753-2763.	0.7	34
79	Distinct genetic variation and heterogeneity of the Iranian population. PLoS Genetics, 2019, 15, e1008385.	1.5	34
80	The second mutation of SYCE1 gene associated with autosomal recessive nonobstructive azoospermia. Journal of Assisted Reproduction and Genetics, 2020, 37, 451-458.	1.2	34
81	Validation of a reverse-hybridization StripAssay for the simultaneous analysis of common Î±-thalassemia point mutations and deletions. Clinical Chemistry and Laboratory Medicine, 2007, 45, 605-10.	1.4	33
82	Mutation profile of BBS genes in Iranian patients with Bardetâ€“Biedl syndrome: genetic characterization and report of nine novel mutations in five BBS genes. Journal of Human Genetics, 2014, 59, 368-375.	1.1	33
83	Association of the dopamine transporter gene (DAT1) core promoter polymorphism ?67T variant with schizophrenia. American Journal of Medical Genetics Part A, 2004, 129B, 10-12.	2.4	32
84	Association between the DRD2 A1 allele and opium addiction in the Iranian population. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2005, 134B, 39-41.	1.1	32
85	The Cypriot and Iranian National Mutation Frequency Databases. Human Mutation, 2006, 27, 598-599.	1.1	32
86	Association analysis of the dopamine transporter (DAT1)-67A/T polymorphism in bipolar disorder. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2005, 135B, 47-49.	1.1	31
87	Î±-Thalassemia Mutations in Khuzestan Province, Southwest Iran. Hemoglobin, 2008, 32, 546-552.	0.4	31
88	Integrated Sequence Analysis Pipeline Provides One-Stop Solution for Identifying Disease-Causing Mutations. Human Mutation, 2014, 35, 1427-1435.	1.1	31
89	Î±-Thalassemia Mutations in Gilan Province, North Iran. Hemoglobin, 2009, 33, 235-241.	0.4	30
90	Identification of SLC26A4 gene mutations in Iranian families with hereditary hearing impairment. European Journal of Pediatrics, 2009, 168, 651-653.	1.3	30

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91	A novel mutation adjacent to the Bth mouse mutation in the TMC1 gene makes this mouse an excellent model of human deafness at the DFNA36 locus. Clinical Genetics, 2010, 77, 395-398.	1.0	29
92	Mutations in TMC1 are a Common Cause of DFNB7/11 Hearing Loss in the Iranian Population. Annals of Otology, Rhinology and Laryngology, 2010, 119, 830-835.	0.6	29
93	The Coxsackievirus and Adenovirus Receptor: A new adhesion protein in cochlear development. Hearing Research, 2006, 215, 1-9.	0.9	28
94	Î²-Thalassemia Mutations in the Kurdish Population of Northeastern Iraq. Hemoglobin, 2010, 34, 469-476.	0.4	27
95	Homozygous SLC6A17 Mutations Cause Autosomal-Recessive Intellectual Disability with Progressive Tremor, Speech Impairment, and Behavioral Problems. American Journal of Human Genetics, 2015, 96, 386-396.	2.6	27
96	Mutations in the histamineN-methyltransferase gene,HNMT, are associated with nonsyndromic autosomal recessive intellectual disability. Human Molecular Genetics, 2015, 24, 5697-5710.	1.4	27
97	A novel DFNA5 mutation does not cause hearing loss in an Iranian family. Journal of Human Genetics, 2007, 52, 549-552.	1.1	26
98	Rare and unexpected mutations among Iranian beta-thalassemia patients and prenatal samples discovered by reverse-hybridization and DNA sequencing. Haematologica, 2002, 87, 1113-4.	1.7	26
99	The Iranian Human Mutation Gene Bank: A data and sample resource for worldwide collaborative genetics research. Human Mutation, 2003, 21, 146-150.	1.1	25
100	Core promoter STRs: Novel mechanism for inter-individual variation in gene expression in humans. Gene, 2012, 492, 195-198.	1.0	25
101	Spectrum of GJB2 (Cx26) gene mutations in Iranian Azeri patients with nonsyndromic autosomal recessive hearing loss. International Journal of Pediatric Otorhinolaryngology, 2012, 76, 268-271.	0.4	24
102	The power of the Mediator complex-Expanding the genetic architecture and phenotypic spectrum of MED12 -related disorders. Clinical Genetics, 2018, 94, 450-456.	1.0	24
103	Bi-allelic Pathogenic Variants in TUBGCP2 Cause Microcephaly and Lissencephaly Spectrum Disorders. American Journal of Human Genetics, 2019, 105, 1005-1015.	2.6	24
104	C677T Methylentetrahydrofulate Reductase and Angiotensin Converting Enzyme Gene Polymorphisms in Patients with Alzheimerâ€™s Disease in Iranian Population. Neurochemical Research, 2006, 31, 1079-1083.	1.6	22
105	A defect in the CLIP1 gene (CLIP-170) can cause autosomal recessive intellectual disability. European Journal of Human Genetics, 2015, 23, 331-336.	1.4	22
106	Determining the cause of patchwork HBA1 and HBA2 genes: recurrent gene conversion or crossing over fixation events. Haematologica, 2006, 91, 297-302.	1.7	22
107	High Prevalence of the â^'Î±3.7Deletion Among Thalassemia Patients in Iran. Hemoglobin, 2003, 27, 53-55.	0.4	21
108	Molecular Mechanisms Underlying Thalassemia Intermedia in Iran. Genetic Testing and Molecular Biomarkers, 2008, 12, 549-556.	1.7	21

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109	Interaction of anÎ±-Globin Gene Triplication withÎ²-Globin Gene Mutations in Iranian Patients withÎ²-Thalassemia Intermedia. Hemoglobin, 2015, 39, 201-206.	0.4	21
110	POLRMT mutations impair mitochondrial transcription causing neurological disease. Nature Communications, 2021, 12, 1135.	5.8	21
111	Attention-deficit/hyperactivity disorder (ADHD) association with the DAT1 core promoter â^'67 T allele. Brain Research, 2006, 1101, 1-4.	1.1	20
112	Frequency of Positive XmnI GÎ³ polymorphism and coinheritance of common alpha thalassemia mutations do not show statistically significant difference between thalassemia major and intermedia cases with homozygous IVSII-1 mutation. Blood Cells, Molecules, and Diseases, 2010, 44, 95-99.	0.6	20
113	Genotype-Phenotype Correlation In Iranian Patients With Hb H Disease. Hemoglobin, 2011, 35, 40-46.	0.4	20
114	Retinitis Pigmentosa, Cutis Laxa, and Pseudoxanthoma Elasticumâ€"Like Skin Manifestations Associated with GGCX Mutations. Journal of Investigative Dermatology, 2014, 134, 2331-2338.	0.3	20
115	Copy Number Variants in Patients with Autism and Additional Clinical Features: Report of VIPR2 Duplication and a Novel Microduplication Syndrome. Molecular Neurobiology, 2017, 54, 7019-7027.	1.9	20
116	Exceptional human core promoter nucleotide compositions. Gene, 2011, 475, 79-86.	1.0	19
117	Association Study of the TREM2 Gene and Identification of a Novel Variant in Exon 2 in Iranian Patients with Late-Onset Alzheimer's Disease. Medical Principles and Practice, 2015, 24, 351-354.	1.1	19
118	Prevalence of common MEFV mutations and carrier frequencies in a large cohort of Iranian populations. Journal of Genetics, 2016, 95, 667-674.	0.4	19
119	Biallelic missense variants in ZBTB11 can cause intellectual disability in humans. Human Molecular Genetics, 2018, 27, 3177-3188.	1.4	19
120	SARSâ€CoVâ€2 outbreak in Iran: The dynamics of the epidemic and evidence on two independent introductions. Transboundary and Emerging Diseases, 2022, 69, 1375-1386.	1.3	19
121	alpha-globin gene deletion and point mutation analysis among in Iranian patients with microcytic hypochromic anemia. Haematologica, 2003, 88, 1196-7.	1.7	19
122	A point mutation at the calreticulin gene core promoter conserved sequence in a case of schizophrenia. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2006, 141B, 294-295.	1.1	18
123	A novel splice site mutation in the RDX gene causes DFNB24 hearing loss in an Iranian family. American Journal of Medical Genetics, Part A, 2009, 149A, 555-558.	0.7	18
124	Carrier frequency of SMA by quantitative analysis of the SMN1 deletion in the Iranian population. European Journal of Neurology, 2010, 17, 160-162.	1.7	18
125	Reversion of the human calreticulin gene promoter to the ancestral type as a result of a novel psychosis-associated mutation. Progress in Neuro-Psychopharmacology and Biological Psychiatry, 2011, 35, 541-544.	2.5	18
126	Prevalence of GJB2-associated deafness and outcomes of cochlear implantation in Iran. Journal of Laryngology and Otology, 2011, 125, 455-459.	0.4	18

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127	LGMD2E is the most common type of sarcoglycanopathies in the Iranian population. Journal of Neurogenetics, 2017, 31, 161-169.	0.6	18
128	BOD1 Is Required for Cognitive Function in Humans and Drosophila. PLoS Genetics, 2016, 12, e1006022.	1.5	18
129	Heterogeneity of Hereditary Hearing Loss in Iran: a Comprehensive Review. Archives of Iranian Medicine, 2016, 19, 720-728.	0.2	18
130	Gender dimorphism in the DAT1 âˆ'67 T-allele homozygosity and predisposition to bipolar disorder. Brain Research, 2007, 1144, 142-145.	1.1	17
131	GJB2 mutations in Baluchi population. Journal of Genetics, 2008, 87, 195-197.	0.4	17
132	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, 125-128.	1.4	17
133	Whole Genome Linkage Analysis Followed by Whole Exome Sequencing Identifies Nicastrin (NCSTN) as a Causative Gene in a Multiplex Family with Î³-Secretase Spectrum of Autoinflammatory Skin Phenotypes. Journal of Investigative Dermatology, 2016, 136, 1283-1286.	0.3	17
134	Missense variants in AIMP1 gene are implicated in autosomal recessive intellectual disability without neurodegeneration. European Journal of Human Genetics, 2016, 24, 392-399.	1.4	17
135	Homozygous Null TBX4 Mutations Lead to Posterior Amelia with Pelvic and Pulmonary Hypoplasia. American Journal of Human Genetics, 2019, 105, 1294-1301.	2.6	17
136	A novel polymorphic purine complex at the 1.5 kb upstream region of the human caveolinâ€1 gene and risk of Alzheimer's disease; Extraâ€short alleles and accumulated allele homozygosity. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2009, 150B, 248-253.	1.1	16
137	Diagnostic values of GHSR DNA methylation pattern in breast cancer. Breast Cancer Research and Treatment, 2012, 135, 705-713.	1.1	16
138	Investigation of primary microcephaly in Bushehr province of Iran: novel <scp>STIL</scp> andÂ<scp>ASPM</scp> mutations. Clinical Genetics, 2013, 83, 488-490.	1.0	16
139	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, 161, 1915-1922.	0.7	16
140	Support for down-tuning of the calreticulin gene in the process of human evolution. Progress in Neuro-Psychopharmacology and Biological Psychiatry, 2011, 35, 1770-1773.	2.5	15
141	Did the GJB2 35delG mutation originate in Iran?. American Journal of Medical Genetics, Part A, 2011, 155, 2453-2458.	0.7	15
142	NGLY1 deficiency: Novel variants and literature review. European Journal of Medical Genetics, 2021, 64, 104146.	0.7	15
143	Identifying the causes of recurrent pregnancy loss in consanguineous couples using whole exome sequencing on the products of miscarriage with no chromosomal abnormalities. Scientific Reports, 2021, 11, 6952.	1.6	15
144	Impact of whole exome sequencing among Iranian patients with autosomal recessive retinitis pigmentosa. Archives of Iranian Medicine, 2015, 18, 776-85.	0.2	15

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145	Prenatal Diagnosis for Î²-Thalassemia Major in the Iranian Province of Hormozgan. Hemoglobin, 2008, 32, 539-545.	0.4	14
146	M-banding characterization of a 16p11.2p13.1 tandem duplication in a child with autism, neurodevelopmental delay and dysmorphism. European Journal of Medical Genetics, 2008, 51, 608-614.	0.7	14
147	miRNA mutations are not a common cause of deafness. American Journal of Medical Genetics, Part A, 2010, 152A, 646-652.	0.7	14
148	A novel mutation of the USH2C (GPR98) gene in an Iranian family with Usher syndrome type II. Journal of Genetics, 2014, 93, 837-841.	0.4	14
149	Two novel mutations in ILDR1 gene cause autosomal recessive nonsyndromic hearing loss in consanguineous Iranian families. Journal of Genetics, 2015, 94, 483-487.	0.4	14
150	A novel splicing variant in FLNC gene responsible for a highly penetrant familial dilated cardiomyopathy in an extended Iranian family. Gene, 2018, 659, 160-167.	1.0	14
151	Genomic structure of a Y-specific ribonucleic acid binding motif- containing gene: a putative candidate for a subset of male infertility. Journal of Clinical Endocrinology and Metabolism, 1996, 81, 2159-2164.	1.8	14
152	Mutation analysis of the DBC2 gene in sporadic and familial breast cancer. Acta OncolÃ³gica, 2007, 46, 770-772.	0.8	13
153	Fragile X syndrome screening of families with consanguineous and non-consanguineous parents in the Iranian population. European Journal of Medical Genetics, 2009, 52, 170-173.	0.7	13
154	Two novel SLC26A4 mutations in Iranian families with autosomal recessive hearing loss. International Journal of Pediatric Otorhinolaryngology, 2012, 76, 845-850.	0.4	13
155	Diagnostic pitfalls of less well recognized HbH disease. Blood Cells, Molecules, and Diseases, 2015, 55, 387-395.	0.6	13
156	YIF1B mutations cause a post-natal neurodevelopmental syndrome associated with Golgi and primary cilium alterations. Brain, 2020, 143, 2911-2928.	3.7	13
157	Utility Of The Multivariate Approach In Predicting Î²-Thalassemia Intermedia Or Î²-Thalassemia Major Types In Iranian Patients. Hemoglobin, 2013, 37, 413-422.	0.4	12
158	Investigating the CFH Gene Polymorphisms as a Risk Factor for Age-related Macular Degeneration in an Iranian Population. Ophthalmic Genetics, 2016, 37, 144-149.	0.5	12
159	GPR126: A novel candidate gene implicated in autosomal recessive intellectual disability. American Journal of Medical Genetics, Part A, 2019, 179, 13-19.	0.7	12
160	SZT2 mutation in a boy with intellectual disability, seizures and autistic features. European Journal of Medical Genetics, 2019, 62, 103556.	0.7	12
161	Sensorineural deafness and male infertility: a contiguous gene deletion syndrome. BMJ Case Reports, 2009, 2009, bcr0820080645-bcr0820080645.	0.2	12
162	Novel mutations responsible for Î±-thalassemia in Iranian families. Hemoglobin, 2013, 37, 148-59.	0.4	12

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163	No association between the DAT1 10-repeat allele and ADHD in the Iranian population. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2008, 147B, 110-111.	1.1	11
164	Novel extreme homozygote haplotypes at the human caveolin 1 gene upstream purine complex in sporadic Alzheimer's disease. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2010, 153B, 347-349.	1.1	11
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