

Hossein Najmabadi

List of Publications by Year in descending order

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Version: 2024-02-01

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	SARS-CoV-2 outbreak in Iran: The dynamics of the epidemic and evidence on two independent introductions. <i>Transboundary and Emerging Diseases</i> , 2022, 69, 1375-1386.	1.3	19
2	Anticipation Can Be More Common in Hereditary Spastic Paraplegia with <i>SPAST</i> Mutations Than It Appears. <i>Canadian Journal of Neurological Sciences</i> , 2022, 49, 651-661.	0.3	6
3	Genetic etiology of hearing loss in Iran. <i>Human Genetics</i> , 2022, 141, 623-631.	1.8	6
4	ZBTB11 dysfunction: spectrum of brain abnormalities, biochemical signature and cellular consequences. <i>Brain</i> , 2022, 145, 2602-2616.	3.7	5
5	Phenotype and genotype spectrum of variants in guanine nucleotide exchange factor genes in a broad cohort of Iranian patients. <i>Molecular Genetics & Genomic Medicine</i> , 2022, 10, e1894.	0.6	5
6	Intellectual disability associated with craniofacial dysmorphism due to <i>POLR3B</i> mutation and defect in spliceosomal machinery. <i>BMC Medical Genomics</i> , 2022, 15, 89.	0.7	1
7	Understanding the Molecular Basis of Fragile X Syndrome Using Differentiated Mesenchymal Stem Cells. <i>Iranian Journal of Child Neurology</i> , 2022, 16, 85-95.	0.2	0
8	Comprehensive genotype-phenotype correlation in <i>AP4</i> deficiency syndrome; Adding data from a large cohort of Iranian patients. <i>Clinical Genetics</i> , 2021, 99, 187-192.	1.0	2
9	The First Case of BENTA Disease (B Cell Expansion with NF- κ B and T Cell Anergy) from Iran. <i>Journal of Clinical Immunology</i> , 2021, 41, 811-813.	2.0	2
10	Biallelic mutations in the death domain of <i>PIDD1</i> impair caspase-2 activation and are associated with intellectual disability. <i>Translational Psychiatry</i> , 2021, 11, 1.	2.4	334
11	Subcellular relocalization and nuclear redistribution of the RNA methyltransferases <i>TRMT1</i> and <i>TRMT1L</i> upon neuronal activation. <i>RNA Biology</i> , 2021, 18, 1905-1919.	1.5	9
12	<i>POLRMT</i> mutations impair mitochondrial transcription causing neurological disease. <i>Nature Communications</i> , 2021, 12, 1135.	5.8	21
13	Exome sequencing utility in defining the genetic landscape of hearing loss and novel gene discovery in Iran. <i>Clinical Genetics</i> , 2021, 100, 59-78.	1.0	4
14	<i>NGLY1</i> deficiency: Novel variants and literature review. <i>European Journal of Medical Genetics</i> , 2021, 64, 104146.	0.7	15
15	Clinical and Genetic Characteristics of Splicing Variant in <i>CYP27A1</i> in an Iranian Family with Cerebrotendinous xanthomatosis. <i>Iranian Biomedical Journal</i> , 2021, 25, 132-139.	0.4	4
16	Identifying the causes of recurrent pregnancy loss in consanguineous couples using whole exome sequencing on the products of miscarriage with no chromosomal abnormalities. <i>Scientific Reports</i> , 2021, 11, 6952.	1.6	15
17	Discovery of a neuromuscular syndrome caused by biallelic variants in <i>ASCC3</i> . <i>Human Genetics and Genomics Advances</i> , 2021, 2, 100024.	1.0	1
18	<i>CEP104</i> and <i>CEP290</i> ; Genes with Ciliary Functions Cause Intellectual Disability in Multiple Families. <i>Archives of Iranian Medicine</i> , 2021, 24, 364-373.	0.2	3

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19	A Novel β^0 -Thalassemia Mutation, HBB: c.356_357delTT [Codon 118 (â€“TT)] in an Iraqi Kurd. Hemoglobin, 2021, 45, 212-214.	0.4	1
20	The recurrent missense mutation p.(Arg367Trp) in YARS1 causes a distinct neurodevelopmental phenotype. Journal of Molecular Medicine, 2021, 99, 1755-1768.	1.7	3
21	The <i>PTRHD1</i> Mutation in Intellectual Disability. Archives of Iranian Medicine, 2021, 24, 747-751.	0.2	3
22	A novel variant of C12orf4 linked to autosomal recessive intellectual disability type 66 with phenotype expansion. Journal of Gene Medicine, 2021, , e3406.	1.4	0
23	Novel variants in Iranian individuals suspected to have inherited red blood cell disorders, including bone marrow failure syndromes. Haematologica, 2020, 105, e1-e4.	1.7	3
24	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
25	Heterozygosity for the Novel HBA2: c.*91_*92delTA Polyadenylation Site Variant on the β^2 -Globin Gene Expanding the Genetic Spectrum of β^+ -Thalassemia in Iran. Hemoglobin, 2020, 44, 423-426.	0.4	2
26	YIF1B mutations cause a post-natal neurodevelopmental syndrome associated with Golgi and primary cilium alterations. Brain, 2020, 143, 2911-2928.	3.7	13
27	Whole genome sequencing identifies a duplicated region encompassing Xq13.2q13.3 in a large Iranian family with intellectual disability. Molecular Genetics & Genomic Medicine, 2020, 8, e1418.	0.6	1
28	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
29	Adult-onset very-long-chain acyl-CoA dehydrogenase deficiency (VLCADD). European Journal of Neurology, 2020, 27, 2257-2266.	1.7	6
30	When transcripts matter: delineating between non-syndromic hearing loss DFNB32 and hearing impairment infertile male syndrome (HIIMS). Journal of Human Genetics, 2020, 65, 609-617.	1.1	2
31	A novel PTC mutation in the BTB domain of KLHL7 gene in two patients with Bohring-Opitz syndrome-like features. European Journal of Medical Genetics, 2020, 63, 103849.	0.7	3
32	CAPN1 and hereditary spastic paraplegia: a novel variant in an Iranian family and overview of the genotype-phenotype correlation. International Journal of Neuroscience, 2020, 131, 1-13.	0.8	7
33	Limbic System Associated Membrane Protein Mutation in an Iranian Family Diagnosed with $\text{M}^{\text{C}}\text{ni}^{\text{r}}\text{re}^{\text{TM}}\text{s}$ Disease. Archives of Iranian Medicine, 2020, 23, 319-325.	0.2	6
34	Novel Mutation in <i>LARP7</i> in Two Iranian Consanguineous Families with Syndromic Intellectual Disability and Facial Dysmorphism. Archives of Iranian Medicine, 2020, 23, 842-847.	0.2	2
35	Molecular Diagnosis of Hereditary Neuropathies by Whole Exome Sequencing and Expanding the Phenotype Spectrum. Archives of Iranian Medicine, 2020, 23, 426-433.	0.2	6
36	<i>GPR126</i> : A novel candidate gene implicated in autosomal recessive intellectual disability. American Journal of Medical Genetics, Part A, 2019, 179, 13-19.	0.7	12

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37	G130V de novo mutation in an Iranian pedigree with nonsyndromic hearing loss without palmoplantar keratoderma. <i>International Journal of Pediatric Otorhinolaryngology</i> , 2019, 126, 109607.	0.4	1
38	Iranome: A catalog of genomic variations in the Iranian population. <i>Human Mutation</i> , 2019, 40, 1968-1984.	1.1	116
39	Bi-allelic Pathogenic Variants in TUBGCP2 Cause Microcephaly and Lissencephaly Spectrum Disorders. <i>American Journal of Human Genetics</i> , 2019, 105, 1005-1015.	2.6	24
40	Distinct genetic variation and heterogeneity of the Iranian population. <i>PLoS Genetics</i> , 2019, 15, e1008385.	1.5	34
41	Chromosomal aberrations in pregnancy and fetal loss: Insight on the effect of consanguinity, review of 1625 cases. <i>Molecular Genetics & Genomic Medicine</i> , 2019, 7, e820.	0.6	10
42	Homozygous variants in the gene <i>SCAPER</i> cause syndromic intellectual disability. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 1214-1225.	0.7	10
43	Identification of disease-causing variants in the <i>EXOSC</i> gene family underlying autosomal recessive intellectual disability in Iranian families. <i>Clinical Genetics</i> , 2019, 95, 718-725.	1.0	5
44	Whole-Transcriptome Analysis Reveals Dysregulation of Actin-Cytoskeleton Pathway in Intellectual Disability Patients. <i>Neuroscience</i> , 2019, 404, 423-444.	1.1	4
45	A splice-altering variant in LARP7 gene leads to exon exclusion. <i>Gene Reports</i> , 2019, 15, 100375.	0.4	1
46	Homozygous Null TBX4 Mutations Lead to Posterior Amelia with Pelvic and Pulmonary Hypoplasia. <i>American Journal of Human Genetics</i> , 2019, 105, 1294-1301.	2.6	17
47	High expression of miR-510 was associated with CGG expansion located at upstream of FMR1 into full mutation. <i>Journal of Cellular Biochemistry</i> , 2019, 120, 1916-1923.	1.2	6
48	SZT2 mutation in a boy with intellectual disability, seizures and autistic features. <i>European Journal of Medical Genetics</i> , 2019, 62, 103556.	0.7	12
49	Effect of inbreeding on intellectual disability revisited by trio sequencing. <i>Clinical Genetics</i> , 2019, 95, 151-159.	1.0	49
50	Novel mutations in MYTH4-FERM domains of myosin 15 are associated with autosomal recessive nonsyndromic hearing loss. <i>International Journal of Pediatric Otorhinolaryngology</i> , 2019, 117, 115-126.	0.4	9
51	Genetics of intellectual disability in consanguineous families. <i>Molecular Psychiatry</i> , 2019, 24, 1027-1039.	4.1	131
52	Investigation of Chromosomal Abnormalities and Microdeletion/ Microduplication(s) in Fifty Iranian Patients with Multiple Congenital Anomalies. <i>Cell Journal</i> , 2019, 21, 337-349.	0.2	1
53	Brief Report of Variants Detected in Hereditary Hearing Loss Cases in Iran over a 3-Year Period. <i>Iranian Journal of Public Health</i> , 2019, 48, 1910-1915.	0.3	1
54	Novel Mutations in KCNQ4, LHFPL5 and COCH Genes in Iranian Families with Hearing Impairment. <i>Archives of Iranian Medicine</i> , 2019, 22, 189-197.	0.2	9

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55	Contribution of Iran in Elucidating the Genetic Causes of Autosomal Recessive Intellectual Disability. Archives of Iranian Medicine, 2019, 22, 461-471.	0.2	0
56	Novel mutations in mitochondrial carrier family gene SLC25A38, causing congenital sideroblastic anemia in Iranian families, identified by whole exome sequencing. Blood Cells, Molecules, and Diseases, 2018, 71, 39-44.	0.6	7
57	Exonic mutations and exon skipping: Lessons learned from <i>DFNA5</i> . Human Mutation, 2018, 39, 433-440.	1.1	44
58	Genotype and phenotype analysis of 43 Iranian facioscapulohumeral muscular dystrophy patients; Evidence for anticipation. Neuromuscular Disorders, 2018, 28, 303-314.	0.3	6
59	SLC52A2 mutations cause SCABD2 phenotype: A second report. International Journal of Pediatric Otorhinolaryngology, 2018, 104, 195-199.	0.4	9
60	CDC14A phosphatase is essential for hearing and male fertility in mouse and human. Human Molecular Genetics, 2018, 27, 780-798.	1.4	49
61	Old gene, new phenotype: splice-altering variants in <i>CEACAM16</i> cause recessive non-syndromic hearing impairment. Journal of Medical Genetics, 2018, 55, 555-560.	1.5	48
62	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
63	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
64	Variants in <i>CIB2</i> cause DFNB48 and not USH1J. Clinical Genetics, 2018, 93, 812-821.	1.0	46
65	<i>CNKSR1</i> gene defect can cause syndromic autosomal recessive intellectual disability. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2018, 177, 691-699.	1.1	4
66	Frequency of α -Globin Gene Triplications and Coinheritance with β -Globin Gene Mutations in the Iranian Population. Hemoglobin, 2018, 42, 252-256.	0.4	7
67	Grxcr2 is required for stereocilia morphogenesis in the cochlea. PLoS ONE, 2018, 13, e0201713.	1.1	11
68	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
69	Biallelic missense variants in ZBTB11 can cause intellectual disability in humans. Human Molecular Genetics, 2018, 27, 3177-3188.	1.4	19
70	Intellectual Disability and Ataxia: Genetic Collisions. Archives of Iranian Medicine, 2018, 21, 29-40.	0.2	3
71	Calpains: Diverse Functions but Enigmatic. Archives of Iranian Medicine, 2018, 21, 170-179.	0.2	4
72	tRNA Methyltransferase Defects and Intellectual Disability. Archives of Iranian Medicine, 2018, 21, 478-485.	0.2	5

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73	Improved diagnostic yield of neuromuscular disorders applying clinical exome sequencing in patients arising from a consanguineous population. <i>Clinical Genetics</i> , 2017, 91, 386-402.	1.0	41
74	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.	1.1	54
75	Homozygous Truncating Variants in TBC1D23 Cause Pontocerebellar Hypoplasia and Alter Cortical Development. <i>American Journal of Human Genetics</i> , 2017, 101, 428-440.	2.6	39
76	CIB2, defective in isolated deafness, is key for auditory hair cell mechanotransduction and survival. <i>EMBO Molecular Medicine</i> , 2017, 9, 1711-1731.	3.3	66
77	LGMD2E is the most common type of sarcoglycanopathies in the Iranian population. <i>Journal of Neurogenetics</i> , 2017, 31, 161-169.	0.6	18
78	Copy Number Variants in Patients with Autism and Additional Clinical Features: Report of VIPR2 Duplication and a Novel Microduplication Syndrome. <i>Molecular Neurobiology</i> , 2017, 54, 7019-7027.	1.9	20
79	De novo Mutation in CACNA1S Gene in a 20-Year-Old Man Diagnosed with Metabolic Myopathy. <i>Archives of Iranian Medicine</i> , 2017, 20, 617-620.	0.2	1
80	Whole Genome Linkage Analysis Followed by Whole Exome Sequencing Identifies Nicastrin (NCSTN) as a Causative Gene in a Multiplex Family with I ³ -Secretase Spectrum of Autoinflammatory Skin Phenotypes. <i>Journal of Investigative Dermatology</i> , 2016, 136, 1283-1286.	0.3	17
81	Prevalence of common MEFV mutations and carrier frequencies in a large cohort of Iranian populations. <i>Journal of Genetics</i> , 2016, 95, 667-674.	0.4	19
82	Investigating the <i>CFH</i> Gene Polymorphisms as a Risk Factor for Age-related Macular Degeneration in an Iranian Population. <i>Ophthalmic Genetics</i> , 2016, 37, 144-149.	0.5	12
83	Characterization of Homozygous Hb Setif (<i>HBA2</i> : c.283G>T) in the Iranian Population. <i>Hemoglobin</i> , 2016, 40, 53-55.	0.4	2
84	Report of limb girdle muscular dystrophy type 2a in 6 Iranian patients, one with a novel deletion in CAPN3 gene. <i>Neuromuscular Disorders</i> , 2016, 26, 277-282.	0.3	5
85	First Report of a Dominantly Inherited $\hat{\iota}^2$ -Thalassemia Caused by a Novel Elongated $\hat{\iota}^2$ -Globin Chain. <i>Hemoglobin</i> , 2016, 40, 102-107.	0.4	6
86	Point mutations which should not be overlooked in Hb H disease. <i>Expert Review of Hematology</i> , 2016, 9, 107-113.	1.0	6
87	Identification of Mutations Causing Aberrant Termination and Deficient Splice Donor Site on the <i>HBA1</i> Gene. <i>Hemoglobin</i> , 2016, 40, 38-43.	0.4	2
88	Missense variants in AIMP1 gene are implicated in autosomal recessive intellectual disability without neurodegeneration. <i>European Journal of Human Genetics</i> , 2016, 24, 392-399.	1.4	17
89	BOD1 Is Required for Cognitive Function in Humans and Drosophila. <i>PLoS Genetics</i> , 2016, 12, e1006022.	1.5	18
90	Heterogeneity of Hereditary Hearing Loss in Iran: a Comprehensive Review. <i>Archives of Iranian Medicine</i> , 2016, 19, 720-728.	0.2	18

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91	Genetic Studies in Intellectual Disability and Behavioral Impairment. Archives of Iranian Medicine, 2016, 19, 363-75.	0.2	10
92	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
93	<i>PDZD7</i> and hearing loss: More than just a modifier. American Journal of Medical Genetics, Part A, 2015, 167, 2957-2965.	0.7	54
94	Diagnostic pitfalls of less well recognized HbH disease. Blood Cells, Molecules, and Diseases, 2015, 55, 387-395.	0.6	13
95	Association Study of the <i>TREM2</i> 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
96	Finding mutation within non-coding region of GJB2 reveals its importance in genetic testing of Hearing Loss in Iranian population. International Journal of Pediatric Otorhinolaryngology, 2015, 79, 136-138.	0.4	4
97	Mutation Screening of the <i>Krüppel-Like Factor 1</i> Gene Using Single-Strand Conformational Polymorphism in a Cohort of Iranian β -Thalassemia Patients. Hemoglobin, 2015, 39, 24-29.	0.4	9
98	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
99	Redefining the MED13L syndrome. European Journal of Human Genetics, 2015, 23, 1308-1317.	1.4	53
100	Mutations in the histamine <i>N</i> -methyltransferase gene, <i>HNMT</i> , are associated with nonsyndromic autosomal recessive intellectual disability. Human Molecular Genetics, 2015, 24, 5697-5710.	1.4	27
101	Interaction of an α -Globin Gene Triplication with β -Globin Gene Mutations in Iranian Patients with β -Thalassemia Intermedia. Hemoglobin, 2015, 39, 201-206.	0.4	21
102	Copy number variations of six and seven α -globin genes in a family with intermedia and major thalassemia phenotypes. Expert Review of Hematology, 2015, 8, 693-698.	1.0	9
103	Characterising the spectrum of autosomal recessive hereditary hearing loss in Iran. Journal of Medical Genetics, 2015, 52, 823-829.	1.5	87
104	Hb Dartmouth (<i>HBA2</i> : c.200T>C): An α -Globin Gene Associated with Hb H Disease in One Homozygous Patient. Hemoglobin, 2015, 39, 152-155.	0.4	8
105	A 21 Nucleotide Duplication on the α - and β -Globin Genes Involves a Variety of Hypochromic Microcytic Anemias, From Mild to Hb H Disease. Hemoglobin, 2015, 39, 196-200.	0.4	2
106	Mutations on the α -Globin Gene That May Trigger α -Thalassemia. Hemoglobin, 2015, 39, 398-402.	0.4	3
107	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
108	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

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109	Homozygosity for the AATAAA>AATA- Polyadenylation Site Mutation on the β -2-Globin Gene Causing Transfusion-Dependent Hb H Disease in an Iranian Patient: A Case Report. <i>Hemoglobin</i> , 2015, 39, 355-8.	0.4	8
110	Report of a patient with limb-girdle muscular dystrophy, ptosis and ophthalmoparesis caused by plectinopathy. <i>Archives of Iranian Medicine</i> , 2015, 18, 60-4.	0.2	8
111	New evidence for the role of calpain 10 in autosomal recessive intellectual disability: identification of two novel nonsense variants by exome sequencing in Iranian families. <i>Archives of Iranian Medicine</i> , 2015, 18, 179-84.	0.2	6
112	Carrier Testing in Known Autosomal Recessive Intellectual Disability Genes in an Iranian Healthy Individual Using Exome Sequencing. <i>Archives of Iranian Medicine</i> , 2015, 18, 643-69.	0.2	2
113	Exome Sequencing and Linkage Analysis Identified Novel Candidate Genes in Recessive Intellectual Disability Associated with Ataxia. <i>Archives of Iranian Medicine</i> , 2015, 18, 670-82.	0.2	4
114	Genetic Investigation of an Iranian Supercentenarian by Whole Exome Sequencing. <i>Archives of Iranian Medicine</i> , 2015, 18, 688-97.	0.2	6
115	Impact of whole exome sequencing among Iranian patients with autosomal recessive retinitis pigmentosa. <i>Archives of Iranian Medicine</i> , 2015, 18, 776-85.	0.2	15
116	Evaluation of chromosomal aberrations caused by air pollutants in some taxi drivers from two polluted districts of urban Tehran and its comparison with drivers from rural areas of Lahijan: a pilot study. <i>Journal of Environmental Health Science & Engineering</i> , 2014, 12, 144.	1.4	6
117	Retinitis Pigmentosa, Cutis Laxa, and Pseudoxanthoma Elasticum-Like Skin Manifestations Associated with GGCX Mutations. <i>Journal of Investigative Dermatology</i> , 2014, 134, 2331-2338.	0.3	20
118	Integrated Sequence Analysis Pipeline Provides One-Stop Solution for Identifying Disease-Causing Mutations. <i>Human Mutation</i> , 2014, 35, 1427-1435.	1.1	31
119	A novel mutation of the USH2C (GPR98) gene in an Iranian family with Usher syndrome type II. <i>Journal of Genetics</i> , 2014, 93, 837-841.	0.4	14
120	Pattern of immunoglobulin and T-cell receptor- γ/δ gene rearrangements in Iranian children with B-precursor acute lymphoblastic leukemia. <i>Hematology</i> , 2014, 19, 259-266.	0.7	7
121	Investigation of ATP6V1B1 and ATP6V0A4 genes causing hereditary hearing loss associated with distal renal tubular acidosis in Iranian families. <i>Journal of Laryngology and Otology</i> , 2014, 128, 1056-1059.	0.4	5
122	Genetics of non-syndromic hearing loss in the Middle East. <i>International Journal of Pediatric Otorhinolaryngology</i> , 2014, 78, 2026-2036.	0.4	45
123	NDST1 missense mutations in autosomal recessive intellectual disability. <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 2753-2763.	0.7	34
124	Mutation profile of BBS genes in Iranian patients with Bardet-Biedl syndrome: genetic characterization and report of nine novel mutations in five BBS genes. <i>Journal of Human Genetics</i> , 2014, 59, 368-375.	1.1	33
125	Characterizing a Cohort of β -Thalassemia Couples Collected During Screening for Hemoglobinopathies: 14 Years of an Iranian Experience. <i>Hemoglobin</i> , 2014, 38, 153-157.	0.4	9
126	Utilizing Ethnic-Specific Differences in Minor Allele Frequency to Recategorize Reported Pathogenic Deafness Variants. <i>American Journal of Human Genetics</i> , 2014, 95, 445-453.	2.6	137

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127	Identification of a founder mutation for Pendred syndrome in families from northwest Iran. <i>International Journal of Pediatric Otorhinolaryngology</i> , 2014, 78, 1828-1832.	0.4	9
128	Optimizing A Lipocomplex-Based Gene Transfer Method into HeLa Cell Line. <i>Cell Journal</i> , 2014, 15, 372-7.	0.2	5
129	Association of polymorphisms at LDLR locus with coronary artery disease independently from lipid profile. <i>Acta Medica Iranica</i> , 2014, 52, 352-9.	0.8	11
130	Utility Of The Multivariate Approach In Predicting $\hat{\alpha}^2$ -Thalassemia Intermedia Or $\hat{\alpha}^2$ -Thalassemia Major Types In Iranian Patients. <i>Hemoglobin</i> , 2013, 37, 413-422.	0.4	12
131	The influence of the BCL11A polymorphism on the phenotype of patients with beta thalassemia could be affected by the beta globin locus control region and/or the Xmn1-HBG2 genotypic background. <i>Blood Cells, Molecules, and Diseases</i> , 2013, 51, 80-84.	0.6	11
132	The Spectrum of $\hat{\alpha}^{\pm}$ -Thalassemia Mutations in the Kurdish Population of Northeastern Iraq. <i>Hemoglobin</i> , 2013, 37, 56-64.	0.4	11
133	Hydroxyurea responsiveness in $\hat{\alpha}$ -thalassemic patients is determined by the stress response adaptation of erythroid progenitors and their differentiation propensity. <i>Haematologica</i> , 2013, 98, 696-704.	1.7	49
134	Investigation of primary microcephaly in Bushehr province of Iran: novel $\langle i \rangle \langle scp \rangle$ STIL $\langle /scp \rangle \langle /i \rangle$ and $\langle i \rangle \langle scp \rangle$ ASPM $\langle /scp \rangle \langle /i \rangle$ mutations. <i>Clinical Genetics</i> , 2013, 83, 488-490.	1.0	16
135	A novel $\langle i \rangle$ ALDH5A1 $\langle /i \rangle$ 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, 161, 1915-1922.	0.7	16
136	Genomic characterization of some Iranian children with idiopathic mental retardation using array comparative genomic hybridization. <i>Indian Journal of Human Genetics</i> , 2013, 19, 443.	0.7	1
137	A Mutation Causes MuSK Reduced Sensitivity to Agrin and Congenital Myasthenia. <i>PLoS ONE</i> , 2013, 8, e53826.	1.1	36
138	Identification of Chromosome Abnormalities in Subtelomeric Regions Using Multiplex Ligation Dependent Probe Amplification (MLPA) Technique in 100 Iranian Patients With Idiopathic Mental Retardation. <i>Iranian Red Crescent Medical Journal</i> , 2013, 15, e8221.	0.5	3
139	A Novel Deletion Mutation in ASPM Gene in an Iranian Family with Autosomal Recessive Primary Microcephaly. <i>Iranian Journal of Child Neurology</i> , 2013, 7, 23-30.	0.2	2
140	Novel mutations responsible for $\hat{\alpha}^{\pm}$ -thalassemia in Iranian families. <i>Hemoglobin</i> , 2013, 37, 148-59.	0.4	12
141	$\langle i \rangle$ NPHP4 $\langle /i \rangle$ Variants Are Associated With Pleiotropic Heart Malformations. <i>Circulation Research</i> , 2012, 110, 1564-1574.	2.0	46
142	Diagnostic values of GHSR DNA methylation pattern in breast cancer. <i>Breast Cancer Research and Treatment</i> , 2012, 135, 705-713.	1.1	16
143	The modifying effect of Xmn1-HBG2 on thalassemic phenotype is associated with its linked elements in the beta globin locus control region, including the palindromic site at 5 $\hat{\alpha}^2$ HS4. <i>Blood Cells, Molecules, and Diseases</i> , 2012, 48, 1-5.	0.6	10
144	The spectrum of GJB2 mutations in the Iranian population with non-syndromic hearing lossâ€”A twelve year study. <i>International Journal of Pediatric Otorhinolaryngology</i> , 2012, 76, 1164-1174.	0.4	38

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145	Spectrum of GJB2 (Cx26) gene mutations in Iranian Azeri patients with nonsyndromic autosomal recessive hearing loss. <i>International Journal of Pediatric Otorhinolaryngology</i> , 2012, 76, 268-271.	0.4	24
146	Two novel SLC26A4 mutations in Iranian families with autosomal recessive hearing loss. <i>International Journal of Pediatric Otorhinolaryngology</i> , 2012, 76, 845-850.	0.4	13
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152	Congenital myasthenic syndrome with tubular aggregates caused by GFPT1 mutations. <i>Journal of Neurology</i> , 2012, 259, 838-850.	1.8	72
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