Hela Azaiez

List of Publications by Year
in descending order

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61 papers	3,460 citations	186209 28 h-index	149623 56 g-index
68 all docs	68 docs citations	68 times ranked	4126 citing authors

#	Article	IF	Citations
1	The natural history of OTOF-related auditory neuropathy spectrum disorders: a multicenter study. Human Genetics, 2022, 141, 853-863.	1.8	7
2	DVPred: a disease-specific prediction tool for variant pathogenicity classification for hearing loss. Human Genetics, 2022, 141, 401-411.	1.8	6
3	A synonymous variant in MYO15A enriched in the Ashkenazi Jewish population causes autosomal recessive hearing loss due to abnormal splicing. European Journal of Human Genetics, 2021, 29, 988-997.	1.4	8
4	Exome sequencing utility in defining the genetic landscape of hearing loss and novelâ€gene discovery in Iran. Clinical Genetics, 2021, 100, 59-78.	1.0	4
5	gEAR: Gene Expression Analysis Resource portal for community-driven, multi-omic data exploration. Nature Methods, 2021, 18, 843-844.	9.0	100
6	Disease-specific ACMG/AMP guidelines improve sequence variant interpretation for hearing loss. Genetics in Medicine, 2021, 23, 2208-2212.	1.1	18
7	A biallelic variant in CLRN2 causes non-syndromic hearing loss in humans. Human Genetics, 2021, 140, 915-931.	1.8	16
8	A comparative analysis of genetic hearing loss phenotypes in European/American and Japanese populations. Human Genetics, 2020, 139, 1315-1323.	1.8	12
9	DFNA5 (GSDME) c.991-15_991-13delTTC: Founder Mutation or Mutational Hotspot?. International Journal of Molecular Sciences, 2020, 21, 3951.	1.8	8
10	Novel loss-of-function mutations in COCH cause autosomal recessive nonsyndromic hearing loss. Human Genetics, 2020, 139, 1565-1574.	1.8	13
11	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
12	Insights into the pathophysiology of DFNA10 hearing loss associated with novel EYA4 variants. Scientific Reports, 2020, 10, 6213.	1.6	15
13	Is it Usher syndrome? Collaborative diagnosis and molecular genetics of patients with visual impairment and hearing loss. Ophthalmic Genetics, 2020, 41, 151-158.	0.5	7
14	ClinGen expert clinical validity curation of 164 hearing loss geneâ€“disease pairs. Genetics in Medicine, 2019, 21, 2239-2247.	1.1	67
15	Splice-altering variant in COL11A1 as a cause of nonsyndromic hearing loss DFNA37. Genetics in Medicine, 2019, 21, 948-954.	1.1	36
16	Exonic mutations and exon skipping: Lessons learned from DFNA5. Human Mutation, 2018, 39, 433-440.	1.1	44
17	CDC14A phosphatase is essential for hearing and male fertility in mouse and human. Human Molecular Genetics, 2018, 27, 780-798.	1.4	49
18	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

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19	Variants in CIB2 cause DFNB48 and not USH1J. Clinical Genetics, 2018, 93, 812-821.	1.0	46
20	Expert specification of the ACMG/AMP variant interpretation guidelines for genetic hearing loss. Human Mutation, 2018, 39, 1593-1613.	1.1	312
21	Genomic Landscape and Mutational Signatures of Deafness-Associated Genes. American Journal of Human Genetics, 2018, 103, 484-497.	2.6	214
22	Grxcr2 is required for stereocilia morphogenesis in the cochlea. PLoS ONE, 2018, 13, e0201713.	1.1	11
23	Intracellular Regulome Variability Along the Organ of Corti: Evidence, Approaches, Challenges, and Perspective. Frontiers in Genetics, 2018, 9, 156.	1.1	17
24	CIB2, defective in isolated deafness, is key for auditory hair cell mechanotransduction and survival. EMBO Molecular Medicine, 2017, 9, 1711-1731.	3.3	66
25	Advances in Molecular Genetics and the Molecular Biology of Deafness. BioMed Research International, 2016, 2016, 1-2.	0.9	1
26	Targeted genomic enrichment and massively parallel sequencing identifies novel nonsyndromic hearing impairment pathogenic variants in Cameroonian families. Clinical Genetics, 2016, 90, 288-290.	1.0	35
27	Comprehensive genetic testing with ethnicâ€specific filtering by allele frequency in a Japanese hearingâ€loss population. Clinical Genetics, 2016, 89, 466-472.	1.0	31
28	Detection and Confirmation of Deafness-Causing Copy Number Variations in the STRC Gene by Massively Parallel Sequencing and Comparative Genomic Hybridization. Annals of Otology, Rhinology and Laryngology, 2016, 125, 918-923.	0.6	28
29	Audioprofile Surfaces. Annals of Otology, Rhinology and Laryngology, 2016, 125, 361-368.	0.6	8
30	Comprehensive genetic testing in the clinical evaluation of 1119 patients with hearing loss. Human Genetics, 2016, 135, 441-450.	1.8	373
31	High-Throughput Genetic Testing for Thrombotic Microangiopathies and C3 Glomerulopathies. Journal of the American Society of Nephrology: JASN, 2016, 27, 1245-1253.	3.0	89
32	Heterogeneity of Hereditary Hearing Loss in Iran: a Comprehensive Review. Archives of Iranian Medicine, 2016, 19, 720-728.	0.2	18
33	PDZD7 and hearing loss: More than just a modifier. American Journal of Medical Genetics, Part A, 2015, 167, 2957-2965.	0.7	54
34	Estimation of Recent and Ancient Inbreeding in a Small Endogamous Tunisian Community Through Genomic Runs of Homozygosity. Annals of Human Genetics, 2015, 79, 402-417.	0.3	14
35	Hearing Loss Caused by a P2RX2 Mutation Identified in a MELAS Family With a Coexisting Mitochondrial 3243AG Mutation. Annals of Otology, Rhinology and Laryngology, 2015, 124, 177S-183S.	0.6	17
36	HOMER2, a Stereociliary Scaffolding Protein, Is Essential for Normal Hearing in Humans and Mice. PLoS Genetics, 2015, 11, e1005137.	1.5	52

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37	De Novo Mutation in X-Linked Hearing Lossâ€“Associated POU3F4 in a Sporadic Case of Congenital Hearing Loss. Annals of Otology, Rhinology and Laryngology, 2015, 124, 169S-176S.	0.6	19
38	Novel PTPRQ Mutations Identified in Three Congenital Hearing Loss Patients With Various Types of Hearing Loss. Annals of Otology, Rhinology and Laryngology, 2015, 124, 184S-192S.	0.6	19
39	Mutations in LOXHD1 Gene Cause Various Types and Severities of Hearing Loss. Annals of Otology, Rhinology and Laryngology, 2015, 124, 135S-141S.	0.6	24
40	USH2 Caused by GPR98 Mutation Diagnosed by Massively Parallel Sequencing in Advance of the Occurrence of Visual Symptoms. Annals of Otology, Rhinology and Laryngology, 2015, 124, 123S-128S.	0.6	9
41	Characterising the spectrum of autosomal recessive hereditary hearing loss in Iran. Journal of Medical Genetics, 2015, 52, 823-829.	1.5	87
42	An international effort towards developing standards for best practices in analysis, interpretation and reporting of clinical genome sequencing results in the CLARITY Challenge. Genome Biology, 2014, 15, R53.	13.9	101
43	TBC1D24Mutation Causes Autosomal-Dominant Nonsyndromic Hearing Loss. Human Mutation, 2014, 35, 819-823.	1.1	78
44	High frequency of exon 15 deletion in the <scp>FANCA</scp> gene in <scp>T</scp>unisian patients affected with <scp>F</scp>anconi anemia disease: implication for diagnosis. Molecular Genetics & Genomic Medicine, 2014, 2, 160-165.	0.6	8
45	Cordova: Web-based management of genetic variation data. Bioinformatics, 2014, 30, 3438-3439.	1.8	3
46	Copy number variants are a common cause of non-syndromic hearing loss. Genome Medicine, 2014, 6, 37.	3.6	137
47	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
48	Consanguinity, endogamy, and genetic disorders in Tunisia. Journal of Community Genetics, 2013, 4, 273-284.	0.5	79
49	Founder mutations in Tunisia: implications for diagnosis in North Africa and Middle East. Orphanet Journal of Rare Diseases, 2012, 7, 52.	1.2	72
50	Adult gaucher disease in southern Tunisia: report of three cases. Diagnostic Pathology, 2012, 7, 4.	0.9	7
51	Severe phenotypes in two Tunisian families with novel XPA mutations: evidence for a correlation between mutation location and disease severity. Archives of Dermatological Research, 2012, 304, 171-176.	1.1	20
52	A novel POLH gene mutation in a xeroderma pigmentosum-V Tunisian patient: phenotypeâ€“genotype correlation. Journal of Genetics, 2011, 90, 483-487.	0.4	16
53	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
54	A novel DFNB1 deletion allele supports the existence of a distant cisâ€regulatory region that controls GJB2 and GJB6 expression. Clinical Genetics, 2010, 78, 267-274.	1.0	75

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55	In Reference toTemporal Bone Imaging inGJB2Deafness. Laryngoscope, 2007, 117, 1127-1127.	1.1	11
56	In Reference toTemporal Bone Imaging inGJB2Deafness. Laryngoscope, 2007, 117, 1127-1129.	1.1	6
57	Genotypeâ€“phenotype correlations for SLC26A4-related deafness. Human Genetics, 2007, 122, 451-457.	1.8	97
58	Connexins and Deafness: From Molecules to Disease. Seminars in Hearing, 2006, 27, 148-159.	0.5	3
59	A novel deletion involving the connexin-30 gene, del(GJB6-d13s1854), found in trans with mutations in the GJB2 gene (connexin-26) in subjects with DFNB1 non-syndromic hearing impairment. Journal of Medical Genetics, 2005, 42, 588-594.	1.5	282
60	GJB2: The spectrum of deafness-causing allele variants and their phenotype. Human Mutation, 2004, 24, 305-311.	1.1	72
61	A genotype-phenotype correlation for GJB2 (connexin 26) deafness. Journal of Medical Genetics, 2004, 41, 147-154.	1.5	178