

Hela Azaiez

List of Publications by Year in descending order

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Version: 2024-02-01

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	Comprehensive genetic testing in the clinical evaluation of 1119 patients with hearing loss. <i>Human Genetics</i> , 2016, 135, 441-450.	1.8	373
2	Expert specification of the ACMG/AMP variant interpretation guidelines for genetic hearing loss. <i>Human Mutation</i> , 2018, 39, 1593-1613.	1.1	312
3	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. <i>Journal of Medical Genetics</i> , 2005, 42, 588-594.	1.5	282
4	Genomic Landscape and Mutational Signatures of Deafness-Associated Genes. <i>American Journal of Human Genetics</i> , 2018, 103, 484-497.	2.6	214
5	A genotype-phenotype correlation for GJB2 (connexin 26) deafness. <i>Journal of Medical Genetics</i> , 2004, 41, 147-154.	1.5	178
6	Copy number variants are a common cause of non-syndromic hearing loss. <i>Genome Medicine</i> , 2014, 6, 37.	3.6	137
7	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
8	An international effort towards developing standards for best practices in analysis, interpretation and reporting of clinical genome sequencing results in the CLARITY Challenge. <i>Genome Biology</i> , 2014, 15, R53.	13.9	101
9	gEAR: Gene Expression Analysis Resource portal for community-driven, multi-omic data exploration. <i>Nature Methods</i> , 2021, 18, 843-844.	9.0	100
10	Genotype-phenotype correlations for SLC26A4-related deafness. <i>Human Genetics</i> , 2007, 122, 451-457.	1.8	97
11	High-Throughput Genetic Testing for Thrombotic Microangiopathies and C3 Glomerulopathies. <i>Journal of the American Society of Nephrology: JASN</i> , 2016, 27, 1245-1253.	3.0	89
12	Characterising the spectrum of autosomal recessive hereditary hearing loss in Iran. <i>Journal of Medical Genetics</i> , 2015, 52, 823-829.	1.5	87
13	Consanguinity, endogamy, and genetic disorders in Tunisia. <i>Journal of Community Genetics</i> , 2013, 4, 273-284.	0.5	79
14	<i>TBC1D24</i> Mutation Causes Autosomal-Dominant Nonsyndromic Hearing Loss. <i>Human Mutation</i> , 2014, 35, 819-823.	1.1	78
15	A novel DFNB1 deletion allele supports the existence of a distant cis-regulatory region that controls <i>GJB2</i> and <i>GJB6</i> expression. <i>Clinical Genetics</i> , 2010, 78, 267-274.	1.0	75
16	GJB2: The spectrum of deafness-causing allele variants and their phenotype. <i>Human Mutation</i> , 2004, 24, 305-311.	1.1	72
17	Founder mutations in Tunisia: implications for diagnosis in North Africa and Middle East. <i>Orphanet Journal of Rare Diseases</i> , 2012, 7, 52.	1.2	72
18	ClinGen expert clinical validity curation of 164 hearing loss gene-disease pairs. <i>Genetics in Medicine</i> , 2019, 21, 2239-2247.	1.1	67

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19	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
20	<i>PDZD7</i> and hearing loss: More than just a modifier. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 2957-2965.	0.7	54
21	HOMER2, a Stereociliary Scaffolding Protein, Is Essential for Normal Hearing in Humans and Mice. <i>PLoS Genetics</i> , 2015, 11, e1005137.	1.5	52
22	Mutations in <i>Grxcr1</i> Are The Basis for Inner Ear Dysfunction in the Pirouette Mouse. <i>American Journal of Human Genetics</i> , 2010, 86, 148-160.	2.6	49
23	CDC14A phosphatase is essential for hearing and male fertility in mouse and human. <i>Human Molecular Genetics</i> , 2018, 27, 780-798.	1.4	49
24	Old gene, new phenotype: splice-altering variants in <i>CEACAM16</i> cause recessive non-syndromic hearing impairment. <i>Journal of Medical Genetics</i> , 2018, 55, 555-560.	1.5	48
25	Variants in <i>CIB2</i> cause DFNB48 and not USH1J. <i>Clinical Genetics</i> , 2018, 93, 812-821.	1.0	46
26	Exonic mutations and exon skipping: Lessons learned from <i>DFNA5</i> . <i>Human Mutation</i> , 2018, 39, 433-440.	1.1	44
27	Splice-altering variant in <i>COL11A1</i> as a cause of nonsyndromic hearing loss DFNA37. <i>Genetics in Medicine</i> , 2019, 21, 948-954.	1.1	36
28	Targeted genomic enrichment and massively parallel sequencing identifies novel nonsyndromic hearing impairment pathogenic variants in Cameroonian families. <i>Clinical Genetics</i> , 2016, 90, 288-290.	1.0	35
29	Comprehensive genetic testing with ethnic-specific filtering by allele frequency in a Japanese hearing-loss population. <i>Clinical Genetics</i> , 2016, 89, 466-472.	1.0	31
30	Detection and Confirmation of Deafness-Causing Copy Number Variations in the <i>STRC</i> Gene by Massively Parallel Sequencing and Comparative Genomic Hybridization. <i>Annals of Otology, Rhinology and Laryngology</i> , 2016, 125, 918-923.	0.6	28
31	Mutations in <i>LOXHD1</i> Gene Cause Various Types and Severities of Hearing Loss. <i>Annals of Otology, Rhinology and Laryngology</i> , 2015, 124, 135S-141S.	0.6	24
32	Severe phenotypes in two Tunisian families with novel XPA mutations: evidence for a correlation between mutation location and disease severity. <i>Archives of Dermatological Research</i> , 2012, 304, 171-176.	1.1	20
33	De Novo Mutation in X-Linked Hearing Loss Associated <i>POU3F4</i> in a Sporadic Case of Congenital Hearing Loss. <i>Annals of Otology, Rhinology and Laryngology</i> , 2015, 124, 169S-176S.	0.6	19
34	Novel <i>PTPRQ</i> Mutations Identified in Three Congenital Hearing Loss Patients With Various Types of Hearing Loss. <i>Annals of Otology, Rhinology and Laryngology</i> , 2015, 124, 184S-192S.	0.6	19
35	Disease-specific ACMG/AMP guidelines improve sequence variant interpretation for hearing loss. <i>Genetics in Medicine</i> , 2021, 23, 2208-2212.	1.1	18
36	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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37	Hearing Loss Caused by a <i>P2RX2</i> Mutation Identified in a MELAS Family With a Coexisting Mitochondrial 3243AG Mutation. <i>Annals of Otolaryngology, Rhinology and Laryngology</i> , 2015, 124, 177S-183S.	0.6	17
38	Intracellular Regulome Variability Along the Organ of Corti: Evidence, Approaches, Challenges, and Perspective. <i>Frontiers in Genetics</i> , 2018, 9, 156.	1.1	17
39	A novel POLH gene mutation in a xeroderma pigmentosum-V Tunisian patient: phenotypeâ€“genotype correlation. <i>Journal of Genetics</i> , 2011, 90, 483-487.	0.4	16
40	A biallelic variant in CLRN2 causes non-syndromic hearing loss in humans. <i>Human Genetics</i> , 2021, 140, 915-931.	1.8	16
41	Insights into the pathophysiology of DFNA10 hearing loss associated with novel EYA4 variants. <i>Scientific Reports</i> , 2020, 10, 6213.	1.6	15
42	Estimation of Recent and Ancient Inbreeding in a Small Endogamous Tunisian Community Through Genomic Runs of Homozygosity. <i>Annals of Human Genetics</i> , 2015, 79, 402-417.	0.3	14
43	Novel loss-of-function mutations in COCH cause autosomal recessive nonsyndromic hearing loss. <i>Human Genetics</i> , 2020, 139, 1565-1574.	1.8	13
44	A comparative analysis of genetic hearing loss phenotypes in European/American and Japanese populations. <i>Human Genetics</i> , 2020, 139, 1315-1323.	1.8	12
45	In Reference to Temporal Bone Imaging in GJB2 Deafness. <i>Laryngoscope</i> , 2007, 117, 1127-1127.	1.1	11
46	Grxcr2 is required for stereocilia morphogenesis in the cochlea. <i>PLoS ONE</i> , 2018, 13, e0201713.	1.1	11
47	USH2 Caused by <i>GPR98</i> Mutation Diagnosed by Massively Parallel Sequencing in Advance of the Occurrence of Visual Symptoms. <i>Annals of Otolaryngology, Rhinology and Laryngology</i> , 2015, 124, 123S-128S.	0.6	9
48	High frequency of exon 15 deletion in the <i>FANCA</i> gene in Tunisian patients affected with Fanconi anemia disease: implication for diagnosis. <i>Molecular Genetics & Genomic Medicine</i> , 2014, 2, 160-165.	0.6	8
49	Audioprofile Surfaces. <i>Annals of Otolaryngology, Rhinology and Laryngology</i> , 2016, 125, 361-368.	0.6	8
50	DFNA5 (GSDME) c.991-15_991-13delTTC: Founder Mutation or Mutational Hotspot?. <i>International Journal of Molecular Sciences</i> , 2020, 21, 3951.	1.8	8
51	A synonymous variant in MYO15A enriched in the Ashkenazi Jewish population causes autosomal recessive hearing loss due to abnormal splicing. <i>European Journal of Human Genetics</i> , 2021, 29, 988-997.	1.4	8
52	Adult gaucher disease in southern Tunisia: report of three cases. <i>Diagnostic Pathology</i> , 2012, 7, 4.	0.9	7
53	Is it Usher syndrome? Collaborative diagnosis and molecular genetics of patients with visual impairment and hearing loss. <i>Ophthalmic Genetics</i> , 2020, 41, 151-158.	0.5	7
54	The natural history of OTOF-related auditory neuropathy spectrum disorders: a multicenter study. <i>Human Genetics</i> , 2022, 141, 853-863.	1.8	7

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55	In Reference to Temporal Bone Imaging in GJB2 Deafness. <i>Laryngoscope</i> , 2007, 117, 1127-1129.	1.1	6
56	DVPred: a disease-specific prediction tool for variant pathogenicity classification for hearing loss. <i>Human Genetics</i> , 2022, 141, 401-411.	1.8	6
57	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
58	Connexins and Deafness: From Molecules to Disease. <i>Seminars in Hearing</i> , 2006, 27, 148-159.	0.5	3
59	Cordova: Web-based management of genetic variation data. <i>Bioinformatics</i> , 2014, 30, 3438-3439.	1.8	3
60	When transcripts matter: delineating between non-syndromic hearing loss DFNB32 and hearing impairment infertile male syndrome (HIIMS). <i>Journal of Human Genetics</i> , 2020, 65, 609-617.	1.1	2
61	Advances in Molecular Genetics and the Molecular Biology of Deafness. <i>BioMed Research International</i> , 2016, 2016, 1-2.	0.9	1