Kevin Booth

List of Publications by Year
in descending order

44 papers	2,195 citations	304743 22 h-index	265206 42 g-index
49 all docs	49 docs citations	49 times ranked	3127 citing authors

#	Article	IF	Citations
1	Comprehensive genetic testing in the clinical evaluation of 1119 patients with hearing loss. Human Genetics, 2016, 135, 441-450.	3.8	373
2	Expert specification of the ACMG/AMP variant interpretation guidelines for genetic hearing loss. Human Mutation, 2018, 39, 1593-1613.	2.5	312
3	Genomic Landscape and Mutational Signatures of Deafness-Associated Genes. American Journal of Human Genetics, 2018, 103, 484-497.	6.2	214
4	Utilizing Ethnic-Specific Differences in Minor Allele Frequency to Recategorize Reported Pathogenic Deafness Variants. American Journal of Human Genetics, 2014, 95, 445-453.	6.2	137
5	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.	9.6	101
6	Characterising the spectrum of autosomal recessive hereditary hearing loss in Iran. Journal of Medical Genetics, 2015, 52, 823-829.	3.2	87
7	TBC1D24Mutation Causes Autosomal-Dominant Nonsyndromic Hearing Loss. Human Mutation, 2014, 35, 819-823.	2.5	78
8	ClinGen expert clinical validity curation of 164 hearing loss geneâ€“disease pairs. Genetics in Medicine, 2019, 21, 2239-2247.	2.4	67
9	CIB2, defective in isolated deafness, is key for auditory hair cell mechanotransduction and survival. EMBO Molecular Medicine, 2017, 9, 1711-1731.	6.9	66
10	PDZD7 and hearing loss: More than just a modifier. American Journal of Medical Genetics, Part A, 2015, 167, 2957-2965.	1.2	54
11	HOMER2, a Stereociliary Scaffolding Protein, Is Essential for Normal Hearing in Humans and Mice. PLoS Genetics, 2015, 11, e1005137.	3.5	52
12	Defective Tmprss3-Associated Hair Cell Degeneration in Inner Ear Organoids. Stem Cell Reports, 2019, 13, 147-162.	4.8	52
13	CDC14A phosphatase is essential for hearing and male fertility in mouse and human. Human Molecular Genetics, 2018, 27, 780-798.	2.9	49
14	Old gene, new phenotype: splice-altering variants in CEACAM16 cause recessive non-syndromic hearing impairment. Journal of Medical Genetics, 2018, 55, 555-560.	3.2	48
15	Variants in CIB2 cause DFNB48 and not USH1J. Clinical Genetics, 2018, 93, 812-821.	2.0	46
16	Exonic mutations and exon skipping: Lessons learned from DFNA5. Human Mutation, 2018, 39, 433-440.	2.5	44
17	Splice-altering variant in COL11A1 as a cause of nonsyndromic hearing loss DFNA37. Genetics in Medicine, 2019, 21, 948-954.	2.4	36
18	Targeted genomic enrichment and massively parallel sequencing identifies novel nonsyndromic hearing impairment pathogenic variants in Cameroonian families. Clinical Genetics, 2016, 90, 288-290.	2.0	35

#	Article	IF	Citations
19	Comprehensive genetic testing with ethnicâ€specific filtering by allele frequency in a Japanese hearingâ€loss population. Clinical Genetics, 2016, 89, 466-472.	2.0	31
20	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.	1.1	28
21	Biomarker pattern of ARIA-E participants in phase 3 randomized clinical trials with bapineuzumab. Neurology, 2018, 90, e877-e886.	1.1	28
22	Mutations in LOXHD1 Gene Cause Various Types and Severities of Hearing Loss. Annals of Otology, Rhinology and Laryngology, 2015, 124, 135S-141S.	1.1	24
23	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.	1.1	19
24	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.	1.1	19
25	Disease-specific ACMG/AMP guidelines improve sequence variant interpretation for hearing loss. Genetics in Medicine, 2021, 23, 2208-2212.	2.4	18
26	Heterogeneity of Hereditary Hearing Loss in Iran: a Comprehensive Review. Archives of Iranian Medicine, 2016, 19, 720-728.	0.6	18
27	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.	1.1	17
28	Intracellular Regulome Variability Along the Organ of Corti: Evidence, Approaches, Challenges, and Perspective. Frontiers in Genetics, 2018, 9, 156.	2.3	17
29	A biallelic variant in CLRN2 causes non-syndromic hearing loss in humans. Human Genetics, 2021, 140, 915-931.	3.8	16
30	Insights into the pathophysiology of DFNA10 hearing loss associated with novel EYA4 variants. Scientific Reports, 2020, 10, 6213.	3.3	15
31	Novel loss-of-function mutations in COCH cause autosomal recessive nonsyndromic hearing loss. Human Genetics, 2020, 139, 1565-1574.	3.8	13
32	A comparative analysis of genetic hearing loss phenotypes in European/American and Japanese populations. Human Genetics, 2020, 139, 1315-1323.	3.8	12
33	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.	1.1	9
34	Audioprofile Surfaces. Annals of Otology, Rhinology and Laryngology, 2016, 125, 361-368.	1.1	8
35	DFNA5 (GSDME) c.991-15_991-13delTTC: Founder Mutation or Mutational Hotspot?. International Journal of Molecular Sciences, 2020, 21, 3951.	4.1	8
36	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.	2.8	8

#	Article	IF	Citations
37	Is it Usher syndrome? Collaborative diagnosis and molecular genetics of patients with visual impairment and hearing loss. Ophthalmic Genetics, 2020, 41, 151-158.	1.2	7
38	Genetic etiology of hearing loss in Iran. Human Genetics, 2022, 141, 623-631.	3.8	6
39	DVPred: a disease-specific prediction tool for variant pathogenicity classification for hearing loss. Human Genetics, 2022, 141, 401-411.	3.8	6
40	Exome sequencing utility in defining the genetic landscape of hearing loss and novelâ€gene discovery in Iran. Clinical Genetics, 2021, 100, 59-78.	2.0	4
41	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.	2.3	2
42	Identification of Novel and Recurrent Variants in MYO15A in Ashkenazi Jewish Patients With Autosomal Recessive Nonsyndromic Hearing Loss. Frontiers in Genetics, 2021, 12, 737782.	2.3	1
43	SEQuencing a baby for an optimal outcome: a genomic future for newborn screening. Molecular Genetics and Metabolism, 2021, 132, S138.	1.1	0
44	Editorial to the Special Issue on â€œThe molecular genetics of hearing and deafnessâ€• Human Genetics, 2022, 141, 305.	3.8	0