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	Genetic etiology of hearing loss in Iran. Human Genetics, 2022, 141, 623-631.	3.8	6
2	DVPred: a disease-specific prediction tool for variant pathogenicity classification for hearing loss. Human Genetics, 2022, 141, 401-411.	3.8	6
3	Editorial to the Special Issue on â€œThe molecular genetics of hearing and deafnessâ€. Human Genetics, 2022, 141, 305.	3.8	0
4	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
5	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
6	SEQuencing a baby for an optimal outcome: a genomic future for newborn screening. Molecular Genetics and Metabolism, 2021, 132, S138.	1.1	0
7	Disease-specific ACMG/AMP guidelines improve sequence variant interpretation for hearing loss. Genetics in Medicine, 2021, 23, 2208-2212.	2.4	18
8	A biallelic variant in CLRN2 causes non-syndromic hearing loss in humans. Human Genetics, 2021, 140, 915-931.	3.8	16
9	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
10	A comparative analysis of genetic hearing loss phenotypes in European/American and Japanese populations. Human Genetics, 2020, 139, 1315-1323.	3.8	12
11	DFNA5 (GSDME) c.991-15_991-13delTTC: Founder Mutation or Mutational Hotspot?. International Journal of Molecular Sciences, 2020, 21, 3951.	4.1	8
12	Novel loss-of-function mutations in COCH cause autosomal recessive nonsyndromic hearing loss. Human Genetics, 2020, 139, 1565-1574.	3.8	13
13	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
14	Insights into the pathophysiology of DFNA10 hearing loss associated with novel EYA4 variants. Scientific Reports, 2020, 10, 6213.	3.3	15
15	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
16	Defective Tmprss3-Associated Hair Cell Degeneration in Inner Ear Organoids. Stem Cell Reports, 2019, 13, 147-162.	4.8	52
17	ClinGen expert clinical validity curation of 164 hearing loss geneâ€“disease pairs. Genetics in Medicine, 2019, 21, 2239-2247.	2.4	67
18	Splice-altering variant in COL11A1 as a cause of nonsyndromic hearing loss DFNA37. Genetics in Medicine, 2019, 21, 948-954.	2.4	36

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19	Biomarker pattern of ARIA-E participants in phase 3 randomized clinical trials with bapineuzumab. Neurology, 2018, 90, e877-e886.	1.1	28
20	Exonic mutations and exon skipping: Lessons learned from DFNA5. Human Mutation, 2018, 39, 433-440.	2.5	44
21	CDC14A phosphatase is essential for hearing and male fertility in mouse and human. Human Molecular Genetics, 2018, 27, 780-798.	2.9	49
22	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
23	Variants in CIB2 cause DFNB48 and not USH1J. Clinical Genetics, 2018, 93, 812-821.	2.0	46
24	Expert specification of the ACMG/AMP variant interpretation guidelines for genetic hearing loss. Human Mutation, 2018, 39, 1593-1613.	2.5	312
25	Genomic Landscape and Mutational Signatures of Deafness-Associated Genes. American Journal of Human Genetics, 2018, 103, 484-497.	6.2	214
26	Intracellular Regulome Variability Along the Organ of Corti: Evidence, Approaches, Challenges, and Perspective. Frontiers in Genetics, 2018, 9, 156.	2.3	17
27	CIB2, defective in isolated deafness, is key for auditory hair cell mechanotransduction and survival. EMBO Molecular Medicine, 2017, 9, 1711-1731.	6.9	66
28	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
29	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
30	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
31	Audioprofile Surfaces. Annals of Otology, Rhinology and Laryngology, 2016, 125, 361-368.	1.1	8
32	Comprehensive genetic testing in the clinical evaluation of 1119 patients with hearing loss. Human Genetics, 2016, 135, 441-450.	3.8	373
33	Heterogeneity of Hereditary Hearing Loss in Iran: a Comprehensive Review. Archives of Iranian Medicine, 2016, 19, 720-728.	0.6	18
34	PDZD7 and hearing loss: More than just a modifier. American Journal of Medical Genetics, Part A, 2015, 167, 2957-2965.	1.2	54
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.	1.1	17
36	HOMER2, a Stereociliary Scaffolding Protein, Is Essential for Normal Hearing in Humans and Mice. PLoS Genetics, 2015, 11, e1005137.	3.5	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.	1.1	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.	1.1	19
39	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
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.	1.1	9
41	Characterising the spectrum of autosomal recessive hereditary hearing loss in Iran. Journal of Medical Genetics, 2015, 52, 823-829.	3.2	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.	9.6	101
43	TBC1D24Mutation Causes Autosomal-Dominant Nonsyndromic Hearing Loss. Human Mutation, 2014, 35, 819-823.	2.5	78
44	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