Huijun Yuan

List of Publications by Year in descending order

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ΗΠΠΙΝ ΥΠΑΝ

#	Article	IF	CITATIONS
1	GJB2 mutation spectrum in 2063 Chinese patients with nonsyndromic hearing impairment. Journal of Translational Medicine, 2009, 7, 26.	4.4	157
2	Cosegregation of the G7444A mutation in the mitochondrial COI/tRNASer(UCN) genes with the 12S rRNA A1555G mutation in a Chinese family with aminoglycoside-induced and nonsyndromic hearing loss. American Journal of Medical Genetics, Part A, 2005, 138A, 133-140.	1.2	79
3	De novo mutation in ATP6V1B2 impairs lysosome acidification and causes dominant deafness-onychodystrophy syndrome. Cell Research, 2014, 24, 1370-1373.	12.0	52
4	A rapid method for simultaneous multi-gene mutation screening in children with nonsyndromic hearing loss. Genomics, 2014, 104, 264-270.	2.9	46
5	The <i>slc4a2b</i> gene is required for hair cell development in zebrafish. Aging, 2020, 12, 18804-18821.	3.1	41
6	Coexistence of mitochondrial 12S rRNA C1494T and CO1/tRNASer(UCN) G7444A mutations in two Han Chinese pedigrees with aminoglycoside-induced and non-syndromic hearing loss. Biochemical and Biophysical Research Communications, 2007, 362, 94-100.	2.1	39
7	Mutation in <i>SSUH2</i> Causes Autosomal-Dominant Dentin Dysplasia Type I. Human Mutation, 2017, 38, 95-104.	2.5	26
8	Resolving the genetic heterogeneity of prelingual hearing loss within one family: Performance comparison and application of two targeted next generation sequencing approaches. Journal of Human Genetics, 2014, 59, 599-607.	2.3	16
9	Rbm24 regulates inner-ear-specific alternative splicing and is essential for maintaining auditory and motor coordination. RNA Biology, 2021, 18, 468-480.	3.1	11
10	Exome Sequencing Identifies a Novel Frameshift Mutation of <i>MYO6</i> as the Cause of Autosomal Dominant Nonsyndromic Hearing Loss in a Chinese Family. Annals of Human Genetics, 2014, 78, 410-423.	0.8	10
11	Identification of a novel TCOF1 mutation in a Chinese family with Treacher Collins syndrome. Experimental and Therapeutic Medicine, 2018, 16, 2645-2650.	1.8	10
12	The natural history of OTOF-related auditory neuropathy spectrum disorders: a multicenter study. Human Genetics, 2022, 141, 853-863.	3.8	7
13	Novel <i>OTOF</i> gene mutations identified using a massively parallel DNA sequencing technique in DFNB9 deafness. Acta Oto-Laryngologica, 2018, 138, 865-870.	0.9	6
14	ldentification of KCNQ1 compound heterozygous mutations in three Chinese families with Jervell and Lange-Nielsen Syndrome. Acta Oto-Laryngologica, 2017, 137, 522-528.	0.9	5
15	Identification of Pathogenic Genes of Nonsyndromic Hearing Loss in Uyghur Families Using Massively Parallel DNA Sequencing Technique. Disease Markers, 2018, 2018, 1-9.	1.3	5
16	ldentification of a complex genomic rearrangement in <i>TMPRSS3</i> by massively parallel sequencing in Chinese cases with prelingual hearing loss. Molecular Genetics & Genomic Medicine, 2019, 7, e685.	1.2	3
17	Two novel <i>BTD</i> mutations causing profound biotinidase deficiency in a Chinese patient. Molecular Genetics & Genomic Medicine, 2021, 9, e1591.	1.2	3
18	Identification of novel <i>MITF</i> mutations in Chinese families with Waardenburg syndrome type II. Molecular Genetics & Genomic Medicine, 2021, 9, e1770.	1.2	3

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#	Article	IF	CITATIONS
19	A missense mutation in TMEM67 causes Meckel-Gruber syndrome type 3 (MKS3): a family from China. International Journal of Clinical and Experimental Pathology, 2015, 8, 5379-86.	0.5	3
20	Gene screening facilitates diagnosis of complicated symptoms: A case report. Molecular Medicine Reports, 2017, 16, 7915-7922.	2.4	2
21	Application of SNPscan in Genetic Screening for Common Hearing Loss Genes. PLoS ONE, 2016, 11, e0165650.	2.5	1
22	ldentification of two recurrent mutations of COL1A1 gene in Chinese Van der Hoeve syndrome patients. Acta Oto-Laryngologica, 2016, 136, 786-791.	0.9	1
23	A followâ€up study of a Chinese family with Waardenburg syndrome type II caused by a truncating mutation of <i>MITF</i> gene. Molecular Genetics & Genomic Medicine, 2020, 8, e1520.	1.2	0