

# Huijun Yuan

## List of Publications by Year in descending order

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23  
papers

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citations

1163117

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677142

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#	ARTICLE	IF	CITATIONS
1	The natural history of OTOF-related auditory neuropathy spectrum disorders: a multicenter study. <i>Human Genetics</i> , 2022, 141, 853-863.	3.8	7
2	Rbm24 regulates inner-ear-specific alternative splicing and is essential for maintaining auditory and motor coordination. <i>RNA Biology</i> , 2021, 18, 468-480.	3.1	11
3	Two novel <i>BTD</i> mutations causing profound biotinidase deficiency in a Chinese patient. <i>Molecular Genetics &amp; Genomic Medicine</i> , 2021, 9, e1591.	1.2	3
4	Identification of novel <i>MITF</i> mutations in Chinese families with Waardenburg syndrome type II. <i>Molecular Genetics &amp; Genomic Medicine</i> , 2021, 9, e1770.	1.2	3
5	A follow-up study of a Chinese family with Waardenburg syndrome type II caused by a truncating mutation of <i>MITF</i> gene. <i>Molecular Genetics &amp; Genomic Medicine</i> , 2020, 8, e1520.	1.2	0
6	The <i>slc4a2b</i> gene is required for hair cell development in zebrafish. <i>Aging</i> , 2020, 12, 18804-18821.	3.1	41
7	Identification of a complex genomic rearrangement in <i>TMPRSS3</i> by massively parallel sequencing in Chinese cases with prelingual hearing loss. <i>Molecular Genetics &amp; Genomic Medicine</i> , 2019, 7, e685.	1.2	3
8	Identification of a novel <i>TCOF1</i> mutation in a Chinese family with Treacher Collins syndrome. <i>Experimental and Therapeutic Medicine</i> , 2018, 16, 2645-2650.	1.8	10
9	Identification of Pathogenic Genes of Nonsyndromic Hearing Loss in Uyghur Families Using Massively Parallel DNA Sequencing Technique. <i>Disease Markers</i> , 2018, 2018, 1-9.	1.3	5
10	Novel <i>OTOF</i> gene mutations identified using a massively parallel DNA sequencing technique in DFNB9 deafness. <i>Acta Oto-Laryngologica</i> , 2018, 138, 865-870.	0.9	6
11	Identification of <i>KCNQ1</i> compound heterozygous mutations in three Chinese families with Jervell and Lange-Nielsen Syndrome. <i>Acta Oto-Laryngologica</i> , 2017, 137, 522-528.	0.9	5
12	Gene screening facilitates diagnosis of complicated symptoms: A case report. <i>Molecular Medicine Reports</i> , 2017, 16, 7915-7922.	2.4	2
13	Mutation in <i>SSUH2</i> Causes Autosomal-Dominant Dentin Dysplasia Type I. <i>Human Mutation</i> , 2017, 38, 95-104.	2.5	26
14	Application of SNPscan in Genetic Screening for Common Hearing Loss Genes. <i>PLoS ONE</i> , 2016, 11, e0165650.	2.5	1
15	Identification of two recurrent mutations of <i>COL1A1</i> gene in Chinese Van der Hoeve syndrome patients. <i>Acta Oto-Laryngologica</i> , 2016, 136, 786-791.	0.9	1
16	A missense mutation in <i>TMEM67</i> causes Meckel-Gruber syndrome type 3 (MKS3): a family from China. <i>International Journal of Clinical and Experimental Pathology</i> , 2015, 8, 5379-86.	0.5	3
17	De novo mutation in <i>ATP6V1B2</i> impairs lysosome acidification and causes dominant deafness-onychodystrophy syndrome. <i>Cell Research</i> , 2014, 24, 1370-1373.	12.0	52
18	Resolving the genetic heterogeneity of prelingual hearing loss within one family: Performance comparison and application of two targeted next generation sequencing approaches. <i>Journal of Human Genetics</i> , 2014, 59, 599-607.	2.3	16

#	ARTICLE	IF	CITATIONS
19	A rapid method for simultaneous multi-gene mutation screening in children with nonsyndromic hearing loss. <i>Genomics</i> , 2014, 104, 264-270.	2.9	46
20	Exome Sequencing Identifies a Novel Frameshift Mutation of <i>MYO6</i> as the Cause of Autosomal Dominant Nonsyndromic Hearing Loss in a Chinese Family. <i>Annals of Human Genetics</i> , 2014, 78, 410-423.	0.8	10
21	GJB2 mutation spectrum in 2063 Chinese patients with nonsyndromic hearing impairment. <i>Journal of Translational Medicine</i> , 2009, 7, 26.	4.4	157
22	Coexistence of mitochondrial 12S rRNA C1494T and COI/tRNASer(UCN) G7444A mutations in two Han Chinese pedigrees with aminoglycoside-induced and non-syndromic hearing loss. <i>Biochemical and Biophysical Research Communications</i> , 2007, 362, 94-100.	2.1	39
23	Cosegregation of the G7444A mutation in the mitochondrial COI/tRNASer(UCN) genes with the 12S rRNA A1555C mutation in a Chinese family with aminoglycoside-induced and nonsyndromic hearing loss. <i>American Journal of Medical Genetics, Part A</i> , 2005, 138A, 133-140.	1.2	79