

Pu Dai

List of Publications by Year in descending order

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85
papers

1,664
citations

331670

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#	ARTICLE	IF	CITATIONS
1	Analysis of genotype-phenotype relationships in 90 Chinese probands with Waardenburg syndrome. <i>Human Genetics</i> , 2022, 141, 839-852.	3.8	13
2	Genetic architecture and phenotypic landscape of deafness and onychodystrophy syndromes. <i>Human Genetics</i> , 2022, 141, 821-838.	3.8	1
3	Petrous bone cholesteatoma: our experience of 20 years and management of two giant cases affecting rhinopharynx. <i>European Archives of Oto-Rhino-Laryngology</i> , 2022, 279, 2791-2801.	1.6	1
4	The natural history of OTOF-related auditory neuropathy spectrum disorders: a multicenter study. <i>Human Genetics</i> , 2022, 141, 853-863.	3.8	7
5	Sheathless acoustic based flow cell sorter for enrichment of rare cells. <i>Cytometry Part A: the Journal of the International Society for Analytical Cytology</i> , 2022, 101, 311-324.	1.5	7
6	Novel CRISPR/Cas12a-based genetic diagnostic approach for SLC26A4 mutation-related hereditary hearing loss. <i>European Journal of Medical Genetics</i> , 2022, 65, 104406.	1.3	3
7	Transcriptome analysis of the early stage ifnlr1-mutant zebrafish indicates the immune response to auditory dysfunction. <i>Gene Expression Patterns</i> , 2022, 43, 119229.	0.8	0
8	Variant analysis of 92 Chinese Han families with hearing loss. <i>BMC Medical Genomics</i> , 2022, 15, 12.	1.5	6
9	A novel missense variant in <i>CEACAM16</i> gene causes autosomal dominant nonsyndromic hearing loss. <i>Annals of Human Genetics</i> , 2022, , .	0.8	1
10	Evolutionary origin of pathogenic <i>GJB2</i> alleles in China. <i>Clinical Genetics</i> , 2022, 102, 305-313.	2.0	3
11	Congenital sensorineural hearing loss as the initial presentation of <i>PTPN11</i> -associated Noonan syndrome with multiple lentigines or Noonan syndrome: clinical features and underlying mechanisms. <i>Journal of Medical Genetics</i> , 2021, 58, 465-474.	3.2	10
12	Treatment of venous pulsatile tinnitus by compression reconstruction of sigmoid sinus. <i>Acta Oto-Laryngologica</i> , 2021, 141, 242-249.	0.9	5
13	Generation of a gene corrected human isogenic iPSC line (CPGHi002-A-1) from a DDOD patient with heterozygous c.1516 C>T mutation in the <i>ATP6V1B2</i> gene. <i>Stem Cell Research</i> , 2021, 53, 102271.	0.7	1
14	Correspondence on DOORS syndrome and a recurrent truncating <i>ATP6V1B2</i> variant by Beauregard-Lacroix et al.. <i>Genetics in Medicine</i> , 2021, 23, 1578-1579.	2.4	1
15	Study of complex structural variations of X-linked deafness-2 based on single-molecule sequencing. <i>Bioscience Reports</i> , 2021, 41, .	2.4	3
16	Sequential Bilateral Cochlear Implantation in a Child with Severe External, Middle, and Inner Ear Malformations: Surgical Considerations and Practical Aspects. <i>Orl</i> , 2021, 83, 1-7.	1.1	0
17	Gene4HL: An Integrated Genetic Database for Hearing Loss. <i>Frontiers in Genetics</i> , 2021, 12, 773009.	2.3	3
18	Syndromic Deafness Gene <i>ATP6V1B2</i> Controls Degeneration of Spiral Ganglion Neurons Through Modulating Proton Flux. <i>Frontiers in Cell and Developmental Biology</i> , 2021, 9, 742714.	3.7	2

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19	Comprehensive genetic testing of Chinese SNHL patients and variants interpretation using ACMG guidelines and ethnically matched normal controls. <i>European Journal of Human Genetics</i> , 2020, 28, 231-243.	2.8	29
20	Clinical characteristics of petrosal cholesteatoma and value of MRI-DWI in the diagnosis. <i>Acta Oto-Laryngologica</i> , 2020, 140, 281-285.	0.9	4
21	Retrospective study of Langerhans cell histiocytosis in ear, nose and neck. <i>American Journal of Otolaryngology - Head and Neck Medicine and Surgery</i> , 2020, 41, 102369.	1.3	8
22	Establishment of human induced pluripotent stem cell line (CPGHi002-A) from a 10-month-old female patient with DDOD syndrome carrying a heterozygous c.1516 C>A mutation in ATP6V1B2. <i>Stem Cell Research</i> , 2020, 48, 101986.	0.7	3
23	Hearing Phenotypes of Patients with Hearing Loss Homozygous for the <i>GJB2</i> c.235delc Mutation. <i>Neural Plasticity</i> , 2020, 2020, 1-11.	2.2	6
24	Unilateral Cochlear Implants for Severe, Profound, or Moderate Sloping to Profound Bilateral Sensorineural Hearing Loss. <i>JAMA Otolaryngology - Head and Neck Surgery</i> , 2020, 146, 942.	2.2	69
25	Prelingual Sensorineural Hearing Loss Caused by a Novel <i>GJB2</i> Dominant Mutation in a Chinese Family. <i>BioMed Research International</i> , 2020, 2020, 1-6.	1.9	4
26	Multiple synostoses syndrome: Clinical report and retrospective analysis. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 1438-1448.	1.2	6
27	Analysis of revision and reimplantation of cochlear implantations in 45 cases. <i>Clinical Otolaryngology</i> , 2019, 44, 1109-1114.	1.2	1
28	A subunit of V-ATPases, ATP6V1B2, underlies the pathology of intellectual disability. <i>EBioMedicine</i> , 2019, 45, 408-421.	6.1	28
29	Concurrent Hearing and Genetic Screening of 180,469 Neonates with Follow-up in Beijing, China. <i>American Journal of Human Genetics</i> , 2019, 105, 803-812.	6.2	66
30	Genotype-phenotype variability in Chinese cases of Treacher Collins syndrome. <i>Acta Oto-Laryngologica</i> , 2019, 139, 567-575.	0.9	8
31	Application of multiplanar reconstruction of spiral CT in the diagnosis and treatment of enlarged vestibular aqueducts. <i>Acta Oto-Laryngologica</i> , 2019, 139, 665-670.	0.9	3
32	Detecting novel mutations and combined Klinefelter syndrome in Usher syndrome cases. <i>Acta Oto-Laryngologica</i> , 2019, 139, 479-486.	0.9	4
33	Mutation of <i>IFNL1</i> , an interferon lambda receptor 1, is associated with autosomal-dominant non-syndromic hearing loss. <i>Journal of Medical Genetics</i> , 2018, 55, 298-306.	3.2	16
34	Genetic mutations in non-syndromic deafness patients in Hainan Province have a different mutational spectrum compared to patients from Mainland China. <i>International Journal of Pediatric Otorhinolaryngology</i> , 2018, 108, 49-54.	1.0	8
35	Mutation spectra and founder effect of <i>TMC1</i> in patients with non-syndromic deafness in Xiamen area, China. <i>American Journal of Medical Genetics Part B: Neuropsychiatric Genetics</i> , 2018, 177, 301-307.	1.7	6
36	Cochlear implantation in patients with canal wall down mastoidectomy cavities. <i>Acta Oto-Laryngologica</i> , 2018, 138, 993-997.	0.9	3

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37	Clinical and molecular characterization of POU3F4 mutations in multiple DFNX2 Chinese families. BMC Medical Genetics, 2018, 19, 157.	2.1	10
38	A Missense Mutation in <i>POU4F3</i> Causes Midfrequency Hearing Loss in a Chinese ADNSHL Family. BioMed Research International, 2018, 2018, 1-7.	1.9	9
39	Skewed X-chromosome inactivation and next-generation sequencing to identify a novel SMPX variants associated with X-linked hearing loss in a Chinese family. International Journal of Pediatric Otorhinolaryngology, 2018, 113, 88-93.	1.0	8
40	A novel pore-region mutation, c.887G>A (p.G296D) in KCNQ4, causing hearing loss in a Chinese family with autosomal dominant non-syndromic deafness 2. BMC Medical Genetics, 2017, 18, 36.	2.1	10
41	A quantitative cSMART assay for noninvasive prenatal screening of autosomal recessive nonsyndromic hearing loss caused by GJB2 and SLC26A4 mutations. Genetics in Medicine, 2017, 19, 1309-1316.	2.4	33
42	The relationship between the GJB3 c.538>T variant and hearing phenotype in the Chinese population. International Journal of Pediatric Otorhinolaryngology, 2017, 102, 67-70.	1.0	14
43	Modified Friedman stage in predicting glossopharyngeal obstruction in obstructive sleep apnea. Acta Oto-Laryngologica, 2017, 137, 78-81.	0.9	0
44	Novel Mutations and Mutation Combinations of <i>TMPRSS3</i> Cause Various Phenotypes in One Chinese Family with Autosomal Recessive Hearing Impairment. BioMed Research International, 2017, 2017, 1-8.	1.9	11
45	Identification of <i>TMPRSS3</i> as a Significant Contributor to Autosomal Recessive Hearing Loss in the Chinese Population. Neural Plasticity, 2017, 2017, 1-8.	2.2	15
46	Identification of Adeno-Associated Viral Vectors That Target Neonatal and Adult Mammalian Inner Ear Cell Subtypes. Human Gene Therapy, 2016, 27, 687-699.	2.7	79
47	A novel PIK3CD C896T mutation detected in bilateral sudden sensorineural hearing loss using next generation sequencing: An indication of primary immunodeficiency. Journal of Otology, 2016, 11, 78-83.	1.0	6
48	Effects of cochlear implant surgical technique on post-operative electrode impedance. Acta Oto-Laryngologica, 2016, 136, 677-681.	0.9	4
49	Phenotypic Heterogeneity in a DFNA20/26 family segregating a novel ACTG1 mutation. BMC Genetics, 2016, 17, 33.	2.7	19
50	Myc and Fgf Are Required for Zebrafish Neuromast Hair Cell Regeneration. PLoS ONE, 2016, 11, e0157768.	2.5	22
51	Targeted gene capture and massively parallel sequencing identify <i>TMC1</i> as the causative gene in a six-generation Chinese family with autosomal dominant hearing loss. American Journal of Medical Genetics, Part A, 2015, 167, 2357-2365.	1.2	15
52	Identification of Two Novel Compound Heterozygous PTPRQ Mutations Associated with Autosomal Recessive Hearing Loss in a Chinese Family. PLoS ONE, 2015, 10, e0124757.	2.5	11
53	A novel mutation in POU3F4 in a Chinese family with X-linked non-syndromic hearing loss. Journal of Otology, 2015, 10, 78-82.	1.0	5
54	A novel EYA4 mutation causing hearing loss in a Chinese DFNA family and genotype-phenotype review of EYA4 in deafness. Journal of Translational Medicine, 2015, 13, 154.	4.4	14

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55	Rapid identification of aminoglycoside-induced deafness gene mutations using multiplex real-time polymerase chain reaction. <i>International Journal of Pediatric Otorhinolaryngology</i> , 2015, 79, 1067-1072.	1.0	4
56	Correlation analysis of phenotype and genotype of GJB2 in patients with non-syndromic hearing loss in China. <i>Gene</i> , 2015, 570, 272-276.	2.2	23
57	The Relationship between the p.V37I Mutation in GJB2 and Hearing Phenotypes in Chinese Individuals. <i>PLoS ONE</i> , 2015, 10, e0129662.	2.5	21
58	Mutation Spectrum of Common Deafness-Causing Genes in Patients with Non-Syndromic Deafness in the Xiamen Area, China. <i>PLoS ONE</i> , 2015, 10, e0135088.	2.5	29
59	Novel Compound Heterozygous Mutations in MYO7A Associated with Usher Syndrome 1 in a Chinese Family. <i>PLoS ONE</i> , 2014, 9, e103415.	2.5	7
60	De novo mutation in ATP6V1B2 impairs lysosome acidification and causes dominant deafness-onychodystrophy syndrome. <i>Cell Research</i> , 2014, 24, 1370-1373.	12.0	52
61	Congenital Middle Ear Abnormalities With Absence of the Oval Window. <i>Otology and Neurotology</i> , 2014, 35, 1191-1195.	1.3	20
62	Impact of next-generation sequencing on molecular diagnosis of inherited non-syndromic hearing loss. <i>Journal of Otology</i> , 2014, 9, 122-125.	1.0	12
63	Hearing loss associated with an unusual mutation combination in the gap junction beta 2 (GJB2) gene in a Chinese family. <i>International Journal of Pediatric Otorhinolaryngology</i> , 2014, 78, 599-603.	1.0	4
64	Noninvasive prenatal testing for autosomal recessive conditions by maternal plasma sequencing in a case of congenital deafness. <i>Genetics in Medicine</i> , 2014, 16, 972-976.	2.4	47
65	A Novel Mutation in the TECTA Gene in a Chinese Family with Autosomal Dominant Nonsyndromic Hearing Loss. <i>PLoS ONE</i> , 2014, 9, e89240.	2.5	5
66	KCNJ10 May Not Be a Contributor to Nonsyndromic Enlargement of Vestibular Aqueduct (NSEVA) in Chinese Subjects. <i>PLoS ONE</i> , 2014, 9, e108134.	2.5	20
67	Genetic mutations in nonsyndromic deafness patients of Chinese minority and han ethnicities in Yunnan, China. <i>Journal of Translational Medicine</i> , 2013, 11, 312.	4.4	42
68	Novel compound heterozygous mutations in the MYO15A gene in autosomal recessive hearing loss identified by whole-exome sequencing. <i>Journal of Translational Medicine</i> , 2013, 11, 284.	4.4	26
69	Intraoperative CT-guided cochlear implantation in congenital ear deformity. <i>Acta Oto-Laryngologica</i> , 2012, 132, 951-958.	0.9	15
70	Identification of a p.R143Q dominant mutation in the gap junction beta-2 gene in three Chinese patients with different hearing phenotypes. <i>Acta Oto-Laryngologica</i> , 2012, 133, 55-58.	0.9	4
71	De novo dominant mutation of GJB2 in two Chinese families with nonsyndromic hearing loss. <i>International Journal of Pediatric Otorhinolaryngology</i> , 2011, 75, 1333-1336.	1.0	5
72	Loss-of-Function Mutations in the PRPS1 Gene Cause a Type of Nonsyndromic X-linked Sensorineural Deafness, DFN2. <i>American Journal of Human Genetics</i> , 2010, 86, 65-71.	6.2	88

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73	Identification of a novel mutation in POU3F4 for prenatal diagnosis in a Chinese family with X-linked nonsyndromic hearing loss. <i>Journal of Genetics and Genomics</i> , 2010, 37, 787-793.	3.9	21
74	Distinct and novel <i>SLC26A4/Pendrin</i> mutations in Chinese and U.S. patients with nonsyndromic hearing loss. <i>Physiological Genomics</i> , 2009, 38, 281-290.	2.3	61
75	A de novo <i>GJB2</i> (connexin 26) mutation, R75W, in a Chinese pedigree with hearing loss and palmoplantar keratoderma. <i>American Journal of Medical Genetics, Part A</i> , 2009, 149A, 689-692.	1.2	20
76	GJB2 mutation spectrum in 2063 Chinese patients with nonsyndromic hearing impairment. <i>Journal of Translational Medicine</i> , 2009, 7, 26.	4.4	157
77	Molecular Etiology of Hearing Impairment in Inner Mongolia: mutations in <i>SLC26A4</i> gene and relevant phenotype analysis. <i>Journal of Translational Medicine</i> , 2008, 6, 74.	4.4	46
78	<i>SLC26A4</i> c.919-2A>G varies among Chinese ethnic groups as a cause of hearing loss. <i>Genetics in Medicine</i> , 2008, 10, 586-592.	2.4	57
79	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. <i>Biochemical and Biophysical Research Communications</i> , 2007, 362, 94-100.	2.1	39
80	The prevalence of the 235delC <i>GJB2</i> mutation in a Chinese deaf population. <i>Genetics in Medicine</i> , 2007, 9, 283-289.	2.4	68
81	Extremely low penetrance of deafness associated with the mitochondrial 12S rRNA mutation in 16 Chinese families: Implication for early detection and prevention of deafness. <i>Biochemical and Biophysical Research Communications</i> , 2006, 340, 194-199.	2.1	52
82	Extremely low penetrance of deafness associated with the mitochondrial 12S rRNA T1095C mutation in three Chinese families. <i>Biochemical and Biophysical Research Communications</i> , 2006, 348, 200-205.	2.1	11
83	Correlation of Cochlear Blood Supply with Mitochondrial DNA Common Deletion in Presbycusis. <i>Acta Oto-Laryngologica</i> , 2004, 124, 130-136.	0.9	50
84	Stereo morphology of temporal bone and ear. <i>Chinese Medical Journal</i> , 2004, 117, 733-7.	2.3	3
85	Computer-Aided Three-Dimensional Reconstruction and Measurement of the Optic Canal and Intracanalicular Structures. <i>Laryngoscope</i> , 1999, 109, 1499-1502.	2.0	14