Shin-Ya Nishio

List of Publications by Year
in descending order

69	1,867	236612	301761
papers	citations	25 h-index	39 g-index

70	70	70	1833
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Mutation spectrum and genotypeâ€“phenotype correlation of hearing loss patients caused by SLC26A4 mutations in the Japanese: a large cohort study. Journal of Human Genetics, 2014, 59, 262-268.	1.1	113
2	Ethnic-Specific Spectrum of GJB2 and SLC26A4 Mutations. Annals of Otology, Rhinology and Laryngology, 2015, 124, 61S-76S.	0.6	91
3	Targeted Exon Sequencing Successfully Discovers Rare Causative Genes and Clarifies the Molecular Epidemiology of Japanese Deafness Patients. PLoS ONE, 2013, 8, e71381.	1.1	88
4	Massively Parallel DNA Sequencing Successfully Identifies New Causative Mutations in Deafness Genes in Patients with Cochlear Implantation and EAS. PLoS ONE, 2013, 8, e75793.	1.1	83
5	Achievement of hearing preservation in the presence of an electrode covering the residual hearing region. Acta Oto-Laryngologica, 2011, 131, 405-412.	0.3	70
6	Deafness Gene Variations in a 1120 Nonsyndromic Hearing Loss Cohort. Annals of Otology, Rhinology and Laryngology, 2015, 124, 49S-60S.	0.6	68
7	Prevalence and Clinical Features of Hearing Loss Patients with CDH23 Mutations: A Large Cohort Study. PLoS ONE, 2012, 7, e40366.	1.1	61
8	A Comprehensive Study on the Etiology of Patients Receiving Cochlear Implantation With Special Emphasis on Genetic Epidemiology. Otology and Neurotology, 2016, 37, e126-e134.	0.7	61
9	Frequency and clinical features of hearing loss caused by STRC deletions. Scientific Reports, 2019, 9, 4408.	1.6	56
10	Comprehensive Genetic Screening of KCNQ4 in a Large Autosomal Dominant Nonsyndromic Hearing Loss Cohort: Genotype-Phenotype Correlations and a Founder Mutation. PLoS ONE, 2013, 8, e63231.	1.1	47
11	Gene Expression Profiles of the Cochlea and Vestibular Endorgans. Annals of Otology, Rhinology and Laryngology, 2015, 124, 6S-48S.	0.6	44
12	Mutations in the MYO15A Gene Are a Significant Cause of Nonsyndromic Hearing Loss. Annals of Otology, Rhinology and Laryngology, 2015, 124, 158S-168S.	0.6	42
13	Long-term results of hearing preservation cochlear implant surgery in patients with residual low frequency hearing. Acta Oto-Laryngologica, 2017, 137, 516-521.	0.3	40
14	Patients withCDH23mutations and the 1555A>G mitochondrial mutation are good candidates for electric acoustic stimulation (EAS). Acta Oto-Laryngologica, 2012, 132, 377-384.	0.3	39
15	Clinical Application of a Custom AmpliSeq Library and Ion Torrent PGM Sequencing to Comprehensive Mutation Screening for Deafness Genes. Genetic Testing and Molecular Biomarkers, 2015, 19, 209-217.	0.3	37
16	Massively Parallel DNA Sequencing Facilitates Diagnosis of Patients with Usher Syndrome Type 1. PLoS ONE, 2014, 9, e90688.	1.1	36
17	Outcomes of cochlear implantation for the patients with specific genetic etiologies: a systematic literature review. Acta Oto-Laryngologica, 2017, 137, 730-742.	0.3	35
18	OTOF mutation screening in Japanese severe to profound recessive hearing loss patients. BMC Medical Genetics, 2013, 14, 95.	2.1	34

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19	WFS1 mutation screening in a large series of Japanese hearing loss patients: Massively parallel DNA sequencing-based analysis. PLoS ONE, 2018, 13, e0193359.	1.1	33
20	The Patients Associated With TMPRSS3 Mutations Are Good Candidates for Electric Acoustic Stimulation. Annals of Otology, Rhinology and Laryngology, 2015, 124, 193S-204S.	0.6	32
21	OTOF mutation analysis with massively parallel DNA sequencing in 2,265 Japanese sensorineural hearing loss patients. PLoS ONE, 2019, 14, e0215932.	1.1	31
22	POU4F3 mutation screening in Japanese hearing loss patients: Massively parallel DNA sequencing-based analysis identified novel variants associated with autosomal dominant hearing loss. PLoS ONE, 2017, 12, e0177636.	1.1	31
23	TECTA mutations in Japanese with mid-frequency hearing loss affected by zona pellucida domain protein secretion. Journal of Human Genetics, 2012, 57, 587-592.	1.1	29
24	An effective screening strategy for deafness in combination with a next-generation sequencing platform: a consecutive analysis. Journal of Human Genetics, 2016, 61, 253-261.	1.1	29
25	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.	0.6	28
26	Massively Parallel DNA Sequencing Successfully Identified Seven Families With Deafness-Associated MYO6 Mutations. Annals of Otology, Rhinology and Laryngology, 2015, 124, 148S-157S.	0.6	27
27	Cochlear Implantation From the Perspective of Genetic Background. Anatomical Record, 2020, 303, 563-593.	0.8	27
28	Detailed Hearing and Vestibular Profiles in the Patients with COCH Mutations. Annals of Otology, Rhinology and Laryngology, 2015, 124, 100S-110S.	0.6	25
29	Simple and efficient germline copy number variant visualization method for the Ion AmpliSeqâ„¢ custom panel. Molecular Genetics & Genomic Medicine, 2018, 6, 678-686.	0.6	25
30	The genetic etiology of hearing loss in Japan revealed by the social health insurance-based genetic testing of 10K patients. Human Genetics, 2022, 141, 665-681.	1.8	25
31	Mutations in LOXHD1 Gene Cause Various Types and Severities of Hearing Loss. Annals of Otology, Rhinology and Laryngology, 2015, 124, 135S-141S.	0.6	24
32	Mutational Spectrum and Clinical Features of Patients With ACTG1 Mutations Identified by Massively Parallel DNA Sequencing. Annals of Otology, Rhinology and Laryngology, 2015, 124, 84S-93S.	0.6	23
33	Social Health Insurance-Based Simultaneous Screening for 154 Mutations in 19 Deafness Genes Efficiently Identified Causative Mutations in Japanese Hearing Loss Patients. PLoS ONE, 2016, 11, e0162230.	1.1	22
34	Frequency of mitochondrial mutations in non-syndromic hearing loss as well as possibly responsible variants found by whole mitochondrial genome screening. Journal of Human Genetics, 2014, 59, 100-106.	1.1	21
35	Genetic testing has the potential to impact hearing preservation following cochlear implantation. Acta Oto-Laryngologica, 2020, 140, 438-444.	0.3	21
36	Prevalence and clinical features of hearing loss caused by EYA4 variants. Scientific Reports, 2020, 10, 3662.	1.6	20

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37	Laser-capture micro dissection combined with next-generation sequencing analysis of cell type-specific deafness gene expression in the mouse cochlea. Hearing Research, 2017, 348, 87-97.	0.9	19
38	Comprehensive analysis of syndromic hearing loss patients in Japan. Scientific Reports, 2019, 9, 11976.	1.6	19
39	Haplotype Analysis of GJB2 Mutations: Founder Effect or Mutational Hot Spot?. Genes, 2020, 11, 250.	1.0	19
40	Genetic background of candidates for EAS (Electric-Acoustic Stimulation). Audiological Medicine, 2010, 8, 28-32.	0.4	18
41	Feasibility of hearing preservation for residual hearing with longer cochlear implant electrodes. Acta Oto-Laryngologica, 2018, 138, 1080-1085.	0.3	18
42	The Prevalence and Clinical Characteristics of TECTA-Associated Autosomal Dominant Hearing Loss. Genes, 2019, 10, 744.	1.0	17
43	Comprehensive Genetic Analysis of Japanese Autosomal Dominant Sensorineural Hearing Loss Patients. PLoS ONE, 2016, 11, e0166781.	1.1	17
44	Sensorineural hearing loss and mild cardiac phenotype caused by an EYA4 mutation. Human Genome Variation, 2018, 5, 23.	0.4	15
45	Mid-Frequency Hearing Loss Is Characteristic Clinical Feature of OTOA-Associated Hearing Loss. Genes, 2019, 10, 715.	1.0	15
46	Mutational Spectrum and Clinical Features of Patients with LOXHD1 Variants Identified in an 8074 Hearing Loss Patient Cohort. Genes, 2019, 10, 735.	1.0	15
47	Novel ACTG1 mutations in patients identified by massively parallel DNA sequencing cause progressive hearing loss. Scientific Reports, 2020, 10, 7056.	1.6	15
48	Replication initiator protein mRNA of ColE2 plasmid and its antisense regulator RNA are under the control of different degradation pathways. Plasmid, 2008, 59, 102-110.	0.4	12
49	The Clinical Next-Generation Sequencing Database: A Tool for the Unified Management of Clinical Information and Genetic Variants to Accelerate Variant Pathogenicity Classification. Human Mutation, 2017, 38, 252-259.	1.1	12
50	Prevalence of the mitochondrial 1555 A>G and 1494 C>T mutations in a community-dwelling population in Japan. Human Genome Variation, 2020, 7, 27.	0.4	12
51	A comparative analysis of genetic hearing loss phenotypes in European/American and Japanese populations. Human Genetics, 2020, 139, 1315-1323.	1.8	12
52	Clinical Characteristics and In Vitro Analysis of MYO6 Variants Causing Late-onset Progressive Hearing Loss. Genes, 2020, 11, 273.	1.0	12
53	Human deafness-associated variants alter the dynamics of key molecules in hair cell stereocilia F-actin cores. Human Genetics, 2022, 141, 363-382.	1.8	12
54	An Usher syndrome type 1 patient diagnosed before the appearance of visual symptoms by MYO7A mutation analysis. International Journal of Pediatric Otorhinolaryngology, 2013, 77, 298-302.	0.4	11

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55	The effects of RNA degradation enzymes on antisense RNAI controlling ColE2 plasmid copy number. Plasmid, 2008, 60, 174-180.	0.4	9
56	The advantages of sound localization and speech perception of bilateral electric acoustic stimulation. Acta Oto-Laryngologica, 2015, 135, 147-153.	0.3	9
57	Genetic background in late-onset sensorineural hearing loss patients. Journal of Human Genetics, 2022, 67, 223-230.	1.1	9
58	Arginine-rich RNA binding domain and protein scaffold domain of RNase E are important for degradation of RNAI but not for that of the Rep mRNA of the ColE2 plasmid. Plasmid, 2009, 62, 83-87.	0.4	8
59	Milestones toward cochlear gene therapy for patients with hereditary hearing loss. Laryngoscope Investigative Otolaryngology, 2021, 6, 958-967.	0.6	8
60	Frequency of the STRC-CATSPER2 deletion in STRC-associated hearing loss patients. Scientific Reports, 2022, 12, 634.	1.6	6
61	Variants in CDH23 cause a broad spectrum of hearing loss: from non-syndromic to syndromic hearing loss as well as from congenital to age-related hearing loss. Human Genetics, 2022, 141, 903-914.	1.8	6
62	Prevalence and Clinical Characteristics of Hearing Loss Caused by MYH14 Variants. Genes, 2021, 12, 1623.	1.0	5
63	Detailed Clinical Features of Deafness Caused by a Claudin-14 Variant. International Journal of Molecular Sciences, 2019, 20, 4579.	1.8	4
64	Identification of a Novel Copy Number Variation of EYA4 Causing Autosomal Dominant Non-syndromic Hearing Loss. Otology and Neurotology, 2021, 42, e866-e874.	0.7	4
65	Etiology of hearing loss affects auditory skill development and vocabulary development in pediatric cochlear implantation cases. Acta Oto-Laryngologica, 2022, 142, 308-315.	0.3	4
66	Phylogeny and biogeography of arcticâ€alpine butterflies of the genus Oeneis (Nymphalidae: Satyrinae). Entomological Science, 2021, 24, 183-195.	0.3	3
67	Improvement of a Rapid and Highly Sensitive Method for the Diagnosis of the Mitochondrial m.1555A>G Mutation Based on a Single-Stranded Tag Hybridization Chromatographic Printed-Array Strip. Genetic Testing and Molecular Biomarkers, 2021, 25, 79-83.	0.3	2
68	Advances in Molecular Genetics and the Molecular Biology of Deafness. BioMed Research International, 2016, 2016, 1-2.	0.9	1
69	Genetic Counseling for Patients with GJB2-Associated Hearing Loss. Practica Otologica, 2020, 113, 223-233.	0.0	0