Shin-Ya Nishio

List of Publications by Year in Descending Order

67 papers	1,323 citations	22 h-index	33 g-index
70 ext. papers	1,608 ext. citations	3.1 avg, IF	4.73 L-index

#	Paper	IF	Citations
67	Frequency of the STRC-CATSPER2 deletion in STRC-associated hearing loss patients.. *Scientific Reports*, **2022**, 12, 634	4.9	1
66	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**, 1	6.3	2
65	Etiology of hearing loss affects auditory skill development and vocabulary development in pediatric cochlear implantation cases.. *Acta Oto-Laryngologica*, **2022**, 1-8	1.6	0
64	Phylogeny and biogeography of arctic-alpine butterflies of the genus Oeneis (Nymphalidae: Satyrinae). *Entomological Science*, **2021**, 24, 183-195	1.1	1
63	Identification of a Novel Copy Number Variation of EYA4 Causing Autosomal Dominant Non-syndromic Hearing Loss. *Otology and Neurotology*, **2021**, 42, e866-e874	2.6	1
62	Human deafness-associated variants alter the dynamics of key molecules in hair cell stereocilia F-actin cores. *Human Genetics*, **2021**, 1	6.3	2
61	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	1.6	1
60	Milestones toward cochlear gene therapy for patients with hereditary hearing loss. *Laryngoscope Investigative Otolaryngology*, **2021**, 6, 958-967	2.8	1
59	The genetic etiology of hearing loss in Japan revealed by the social health insurance-based genetic testing of 10K patients. *Human Genetics*, **2021**, 1	6.3	7
58	A comparative analysis of genetic hearing loss phenotypes in European/American and Japanese populations. *Human Genetics*, **2020**, 139, 1315-1323	6.3	8
57	Clinical Characteristics and In Vitro Analysis of Variants Causing Late-Onset Progressive Hearing Loss. *Genes*, **2020**, 11,	4.2	5
56	Genetic testing has the potential to impact hearing preservation following cochlear implantation. *Acta Oto-Laryngologica*, **2020**, 140, 438-444	1.6	10
55	Haplotype Analysis of Mutations: Founder Effect or Mutational Hot Spot?. *Genes*, **2020**, 11,	4.2	10
54	Cochlear Implantation From the Perspective of Genetic Background. *Anatomical Record*, **2020**, 303, 563-593	2.3	14
53	Prevalence and clinical features of hearing loss caused by EYA4 variants. *Scientific Reports*, **2020**, 10, 3662	4.9	9
52	Novel ACTG1 mutations in patients identified by massively parallel DNA sequencing cause progressive hearing loss. *Scientific Reports*, **2020**, 10, 7056	4.9	6
51	Genetic Counseling for Patients with GJB2-Associated Hearing Loss. *Practica Otologica*, **2020**, 113, 223-233		

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	1.8	5
49	Mid-Frequency Hearing Loss Is Characteristic Clinical Feature of -Associated Hearing Loss. *Genes*, **2019**, 10,	4.2	12
48	Mutational Spectrum and Clinical Features of Patients with Variants Identified in an 8074 Hearing Loss Patient Cohort. *Genes*, **2019**, 10,	4.2	11
47	The Prevalence and Clinical Characteristics of -Associated Autosomal Dominant Hearing Loss. *Genes* , **2019**, 10,	4.2	8
46	Detailed Clinical Features of Deafness Caused by a Claudin-14 Variant. *International Journal of Molecular Sciences*, **2019**, 20,	6.3	1
45	OTOF mutation analysis with massively parallel DNA sequencing in 2,265 Japanese sensorineural hearing loss patients. *PLoS ONE*, **2019**, 14, e0215932	3.7	17
44	Frequency and clinical features of hearing loss caused by STRC deletions. *Scientific Reports*, **2019**, 9, 4408	4.9	31
43	Comprehensive analysis of syndromic hearing loss patients in Japan. *Scientific Reports*, **2019**, 9, 11976	4.9	13
42	Simple and efficient germline copy number variant visualization method for the Ion AmpliSeq custom panel. *Molecular Genetics & Genomic Medicine*, **2018**, 6, 678	2.3	15
41	WFS1 mutation screening in a large series of Japanese hearing loss patients: Massively parallel DNA sequencing-based analysis. *PLoS ONE*, **2018**, 13, e0193359	3.7	22
40	Feasibility of hearing preservation for residual hearing with longer cochlear implant electrodes. *Acta Oto-Laryngologica*, **2018**, 138, 1080-1085	1.6	14
39	Sensorineural hearing loss and mild cardiac phenotype caused by an mutation. *Human Genome Variation*, **2018**, 5, 23	1.8	10
38	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	3.9	11
37	Outcomes of cochlear implantation for the patients with specific genetic etiologies: a systematic literature review. *Acta Oto-Laryngologica*, **2017**, 137, 730-742	1.6	20
36	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	4.7	10
35	Long-term results of hearing preservation cochlear implant surgery in patients with residual low frequency hearing. *Acta Oto-Laryngologica*, **2017**, 137, 516-521	1.6	35
34	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	3.7	23
33	An effective screening strategy for deafness in combination with a next-generation sequencing platform: a consecutive analysis. *Journal of Human Genetics*, **2016**, 61, 253-61	4.3	21

32	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	3.7	15
31	Comprehensive Genetic Analysis of Japanese Autosomal Dominant Sensorineural Hearing Loss Patients. *PLoS ONE*, **2016**, 11, e0166781	3.7	8
30	Advances in Molecular Genetics and the Molecular Biology of Deafness. *BioMed Research International*, **2016**, 2016, 5629093	3	1
29	A Comprehensive Study on the Etiology of Patients Receiving Cochlear Implantation With Special Emphasis on Genetic Epidemiology. *Otology and Neurotology*, **2016**, 37, e126-34	2.6	43
28	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	2.1	21
27	Mutational spectrum and clinical features of patients with ACTG1 mutations identified by massively parallel DNA sequencing. *Annals of Otology, Rhinology and Laryngology*, **2015**, 124 Suppl 1, 84S-93S	2.1	18
26	Detailed hearing and vestibular profiles in the patients with COCH mutations. *Annals of Otology, Rhinology and Laryngology*, **2015**, 124 Suppl 1, 100S-10S	2.1	21
25	Massively parallel DNA sequencing successfully identified seven families with deafness-associated MYO6 mutations: the mutational spectrum and clinical characteristics. *Annals of Otology, Rhinology and Laryngology*, **2015**, 124 Suppl 1, 148S-57S	2.1	22
24	The advantages of sound localization and speech perception of bilateral electric acoustic stimulation. *Acta Oto-Laryngologica*, **2015**, 135, 147-53	1.6	8
23	The patients associated with TMPRSS3 mutations are good candidates for electric acoustic stimulation. *Annals of Otology, Rhinology and Laryngology*, **2015**, 124 Suppl 1, 193S-204S	2.1	23
22	Mutations in LOXHD1 gene cause various types and severities of hearing loss. *Annals of Otology, Rhinology and Laryngology*, **2015**, 124 Suppl 1, 135S-41S	2.1	22
21	Gene expression profiles of the cochlea and vestibular endorgans: localization and function of genes causing deafness. *Annals of Otology, Rhinology and Laryngology*, **2015**, 124 Suppl 1, 6S-48S	2.1	21
20	Mutations in the MYO15A gene are a significant cause of nonsyndromic hearing loss: massively parallel DNA sequencing-based analysis. *Annals of Otology, Rhinology and Laryngology*, **2015**, 124 Suppl 1, 158S-68S	2.1	36
19	Deafness gene variations in a 1120 nonsyndromic hearing loss cohort: molecular epidemiology and deafness mutation spectrum of patients in Japan. *Annals of Otology, Rhinology and Laryngology*, **2015**, 124 Suppl 1, 49S-60S	2.1	58
18	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, 209-17	1.6	23
17	Ethnic-specific spectrum of GJB2 and SLC26A4 mutations: their origin and a literature review. *Annals of Otology, Rhinology and Laryngology*, **2015**, 124 Suppl 1, 61S-76S	2.1	68
16	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, 100-6	4.3	17
15	Mutation spectrum and genotype-phenotype correlation of hearing loss patients caused by SLC26A4 mutations in the Japanese: a large cohort study. *Journal of Human Genetics*, **2014**, 59, 262-8	4.3	73

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14	Massively parallel DNA sequencing facilitates diagnosis of patients with Usher syndrome type 1. *PLoS ONE*, **2014**, 9, e90688	3.7	34
13	OTOF mutation screening in Japanese severe to profound recessive hearing loss patients. *BMC Medical Genetics*, **2013**, 14, 95	2.1	26
12	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	1.7	10
11	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	3.7	36
10	Targeted exon sequencing successfully discovers rare causative genes and clarifies the molecular epidemiology of Japanese deafness patients. *PLoS ONE*, **2013**, 8, e71381	3.7	78
9	Massively parallel DNA sequencing successfully identifies new causative mutations in deafness genes in patients with cochlear implantation and EAS. *PLoS ONE*, **2013**, 8, e75793	3.7	71
8	Prevalence and clinical features of hearing loss patients with CDH23 mutations: a large cohort study. *PLoS ONE*, **2012**, 7, e40366	3.7	54
7	Patients with CDH23 mutations and the 1555A>G mitochondrial mutation are good candidates for electric acoustic stimulation (EAS). *Acta Oto-Laryngologica*, **2012**, 132, 377-84	1.6	31
6	TECTA mutations in Japanese with mid-frequency hearing loss affected by zona pellucida domain protein secretion. *Journal of Human Genetics*, **2012**, 57, 587-92	4.3	26
5	Achievement of hearing preservation in the presence of an electrode covering the residual hearing region. *Acta Oto-Laryngologica*, **2011**, 131, 405-12	1.6	60
4	Genetic background of candidates for EAS (Electric-Acoustic Stimulation). *Audiological Medicine*, **2010**, 8, 28-32		14
3	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, 83-7	3.3	5
2	Replication initiator protein mRNA of ColE2 plasmid and its antisense regulator RNA are under the control of different degradation pathways. *Plasmid*, **2008**, 59, 102-10	3.3	11
1	The effects of RNA degradation enzymes on antisense RNAI controlling ColE2 plasmid copy number. *Plasmid*, **2008**, 60, 174-80	3.3	9