

# Shin-Ya Nishio

## List of Publications by Citations

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

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	Targeted exon sequencing successfully discovers rare causative genes and clarifies the molecular epidemiology of Japanese deafness patients. <i>PLoS ONE</i> , <b>2013</b> , 8, e71381	3.7	78
66	Mutation spectrum and genotype-phenotype correlation of hearing loss patients caused by SLC26A4 mutations in the Japanese: a large cohort study. <i>Journal of Human Genetics</i> , <b>2014</b> , 59, 262-8	4.3	73
65	Massively parallel DNA sequencing successfully identifies new causative mutations in deafness genes in patients with cochlear implantation and EAS. <i>PLoS ONE</i> , <b>2013</b> , 8, e75793	3.7	71
64	Ethnic-specific spectrum of GJB2 and SLC26A4 mutations: their origin and a literature review. <i>Annals of Otology, Rhinology and Laryngology</i> , <b>2015</b> , 124 Suppl 1, 61S-76S	2.1	68
63	Achievement of hearing preservation in the presence of an electrode covering the residual hearing region. <i>Acta Oto-Laryngologica</i> , <b>2011</b> , 131, 405-12	1.6	60
62	Deafness gene variations in a 1120 nonsyndromic hearing loss cohort: molecular epidemiology and deafness mutation spectrum of patients in Japan. <i>Annals of Otology, Rhinology and Laryngology</i> , <b>2015</b> , 124 Suppl 1, 49S-60S	2.1	58
61	Prevalence and clinical features of hearing loss patients with CDH23 mutations: a large cohort study. <i>PLoS ONE</i> , <b>2012</b> , 7, e40366	3.7	54
60	A Comprehensive Study on the Etiology of Patients Receiving Cochlear Implantation With Special Emphasis on Genetic Epidemiology. <i>Otology and Neurotology</i> , <b>2016</b> , 37, e126-34	2.6	43
59	Mutations in the MYO15A gene are a significant cause of nonsyndromic hearing loss: massively parallel DNA sequencing-based analysis. <i>Annals of Otology, Rhinology and Laryngology</i> , <b>2015</b> , 124 Suppl 1, 158S-68S	2.1	36
58	Comprehensive genetic screening of KCNQ4 in a large autosomal dominant nonsyndromic hearing loss cohort: genotype-phenotype correlations and a founder mutation. <i>PLoS ONE</i> , <b>2013</b> , 8, e63231	3.7	36
57	Long-term results of hearing preservation cochlear implant surgery in patients with residual low frequency hearing. <i>Acta Oto-Laryngologica</i> , <b>2017</b> , 137, 516-521	1.6	35
56	Massively parallel DNA sequencing facilitates diagnosis of patients with Usher syndrome type 1. <i>PLoS ONE</i> , <b>2014</b> , 9, e90688	3.7	34
55	Frequency and clinical features of hearing loss caused by STRC deletions. <i>Scientific Reports</i> , <b>2019</b> , 9, 44084.9	4.9	31
54	Patients with CDH23 mutations and the 1555A>G mitochondrial mutation are good candidates for electric acoustic stimulation (EAS). <i>Acta Oto-Laryngologica</i> , <b>2012</b> , 132, 377-84	1.6	31
53	OTOF mutation screening in Japanese severe to profound recessive hearing loss patients. <i>BMC Medical Genetics</i> , <b>2013</b> , 14, 95	2.1	26
52	TECTA mutations in Japanese with mid-frequency hearing loss affected by zona pellucida domain protein secretion. <i>Journal of Human Genetics</i> , <b>2012</b> , 57, 587-92	4.3	26
51	The patients associated with TMPRSS3 mutations are good candidates for electric acoustic stimulation. <i>Annals of Otology, Rhinology and Laryngology</i> , <b>2015</b> , 124 Suppl 1, 193S-204S	2.1	23

50	Clinical application of a custom AmpliSeq library and ion torrent PGM sequencing to comprehensive mutation screening for deafness genes. <i>Genetic Testing and Molecular Biomarkers</i> , <b>2015</b> , 19, 209-17	1.6	23
49	POU4F3 mutation screening in Japanese hearing loss patients: Massively parallel DNA sequencing-based analysis identified novel variants associated with autosomal dominant hearing loss. <i>PLoS ONE</i> , <b>2017</b> , 12, e0177636	3.7	23
48	Massively parallel DNA sequencing successfully identified seven families with deafness-associated MYO6 mutations: the mutational spectrum and clinical characteristics. <i>Annals of Otology, Rhinology and Laryngology</i> , <b>2015</b> , 124 Suppl 1, 148S-57S	2.1	22
47	Mutations in LOXHD1 gene cause various types and severities of hearing loss. <i>Annals of Otology, Rhinology and Laryngology</i> , <b>2015</b> , 124 Suppl 1, 135S-41S	2.1	22
46	WFS1 mutation screening in a large series of Japanese hearing loss patients: Massively parallel DNA sequencing-based analysis. <i>PLoS ONE</i> , <b>2018</b> , 13, e0193359	3.7	22
45	Detailed hearing and vestibular profiles in the patients with COCH mutations. <i>Annals of Otology, Rhinology and Laryngology</i> , <b>2015</b> , 124 Suppl 1, 100S-10S	2.1	21
44	Gene expression profiles of the cochlea and vestibular endorgans: localization and function of genes causing deafness. <i>Annals of Otology, Rhinology and Laryngology</i> , <b>2015</b> , 124 Suppl 1, 6S-48S	2.1	21
43	An effective screening strategy for deafness in combination with a next-generation sequencing platform: a consecutive analysis. <i>Journal of Human Genetics</i> , <b>2016</b> , 61, 253-61	4.3	21
42	Detection and Confirmation of Deafness-Causing Copy Number Variations in the STRC Gene by Massively Parallel Sequencing and Comparative Genomic Hybridization. <i>Annals of Otology, Rhinology and Laryngology</i> , <b>2016</b> , 125, 918-923	2.1	21
41	Outcomes of cochlear implantation for the patients with specific genetic etiologies: a systematic literature review. <i>Acta Oto-Laryngologica</i> , <b>2017</b> , 137, 730-742	1.6	20
40	Mutational spectrum and clinical features of patients with ACTG1 mutations identified by massively parallel DNA sequencing. <i>Annals of Otology, Rhinology and Laryngology</i> , <b>2015</b> , 124 Suppl 1, 84S-93S	2.1	18
39	OTOF mutation analysis with massively parallel DNA sequencing in 2,265 Japanese sensorineural hearing loss patients. <i>PLoS ONE</i> , <b>2019</b> , 14, e0215932	3.7	17
38	Frequency of mitochondrial mutations in non-syndromic hearing loss as well as possibly responsible variants found by whole mitochondrial genome screening. <i>Journal of Human Genetics</i> , <b>2014</b> , 59, 100-6	4.3	17
37	Simple and efficient germline copy number variant visualization method for the Ion AmpliSeq <sup>®</sup> custom panel. <i>Molecular Genetics &amp; Genomic Medicine</i> , <b>2018</b> , 6, 678	2.3	15
36	Social Health Insurance-Based Simultaneous Screening for 154 Mutations in 19 Deafness Genes Efficiently Identified Causative Mutations in Japanese Hearing Loss Patients. <i>PLoS ONE</i> , <b>2016</b> , 11, e0162230	3.7	15
35	Cochlear Implantation From the Perspective of Genetic Background. <i>Anatomical Record</i> , <b>2020</b> , 303, 563-593		14
34	Genetic background of candidates for EAS (Electric-Acoustic Stimulation). <i>Audiological Medicine</i> , <b>2010</b> , 8, 28-32		14
33	Feasibility of hearing preservation for residual hearing with longer cochlear implant electrodes. <i>Acta Oto-Laryngologica</i> , <b>2018</b> , 138, 1080-1085	1.6	14

32	Comprehensive analysis of syndromic hearing loss patients in Japan. <i>Scientific Reports</i> , <b>2019</b> , 9, 11976	4.9	13
31	Mid-Frequency Hearing Loss Is Characteristic Clinical Feature of -Associated Hearing Loss. <i>Genes</i> , <b>2019</b> , 10,	4.2	12
30	Laser-capture micro dissection combined with next-generation sequencing analysis of cell type-specific deafness gene expression in the mouse cochlea. <i>Hearing Research</i> , <b>2017</b> , 348, 87-97	3.9	11
29	Mutational Spectrum and Clinical Features of Patients with Variants Identified in an 8074 Hearing Loss Patient Cohort. <i>Genes</i> , <b>2019</b> , 10,	4.2	11
28	Replication initiator protein mRNA of Cole2 plasmid and its antisense regulator RNA are under the control of different degradation pathways. <i>Plasmid</i> , <b>2008</b> , 59, 102-10	3.3	11
27	The Clinical Next-Generation Sequencing Database: A Tool for the Unified Management of Clinical Information and Genetic Variants to Accelerate Variant Pathogenicity Classification. <i>Human Mutation</i> , <b>2017</b> , 38, 252-259	4.7	10
26	Genetic testing has the potential to impact hearing preservation following cochlear implantation. <i>Acta Oto-Laryngologica</i> , <b>2020</b> , 140, 438-444	1.6	10
25	Haplotype Analysis of Mutations: Founder Effect or Mutational Hot Spot?. <i>Genes</i> , <b>2020</b> , 11,	4.2	10
24	An Usher syndrome type 1 patient diagnosed before the appearance of visual symptoms by MYO7A mutation analysis. <i>International Journal of Pediatric Otorhinolaryngology</i> , <b>2013</b> , 77, 298-302	1.7	10
23	Sensorineural hearing loss and mild cardiac phenotype caused by an mutation. <i>Human Genome Variation</i> , <b>2018</b> , 5, 23	1.8	10
22	Prevalence and clinical features of hearing loss caused by EYA4 variants. <i>Scientific Reports</i> , <b>2020</b> , 10, 3662	4.9	9
21	The effects of RNA degradation enzymes on antisense RNAi controlling Cole2 plasmid copy number. <i>Plasmid</i> , <b>2008</b> , 60, 174-80	3.3	9
20	The Prevalence and Clinical Characteristics of -Associated Autosomal Dominant Hearing Loss. <i>Genes</i> , <b>2019</b> , 10,	4.2	8
19	The advantages of sound localization and speech perception of bilateral electric acoustic stimulation. <i>Acta Oto-Laryngologica</i> , <b>2015</b> , 135, 147-53	1.6	8
18	A comparative analysis of genetic hearing loss phenotypes in European/American and Japanese populations. <i>Human Genetics</i> , <b>2020</b> , 139, 1315-1323	6.3	8
17	Comprehensive Genetic Analysis of Japanese Autosomal Dominant Sensorineural Hearing Loss Patients. <i>PLoS ONE</i> , <b>2016</b> , 11, e0166781	3.7	8
16	The genetic etiology of hearing loss in Japan revealed by the social health insurance-based genetic testing of 10K patients. <i>Human Genetics</i> , <b>2021</b> , 1	6.3	7
15	Novel ACTG1 mutations in patients identified by massively parallel DNA sequencing cause progressive hearing loss. <i>Scientific Reports</i> , <b>2020</b> , 10, 7056	4.9	6

14	Clinical Characteristics and In Vitro Analysis of Variants Causing Late-Onset Progressive Hearing Loss. <i>Genes</i> , <b>2020</b> , 11,	4.2	5
13	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. <i>Plasmid</i> , <b>2009</b> , 62, 83-7	3.3	5
12	Prevalence of the mitochondrial 1555 A>G and 1494 C>T mutations in a community-dwelling population in Japan. <i>Human Genome Variation</i> , <b>2020</b> , 7, 27	1.8	5
11	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.. <i>Human Genetics</i> , <b>2022</b> , 1	6.3	2
10	Human deafness-associated variants alter the dynamics of key molecules in hair cell stereocilia F-actin cores. <i>Human Genetics</i> , <b>2021</b> , 1	6.3	2
9	Detailed Clinical Features of Deafness Caused by a Claudin-14 Variant. <i>International Journal of Molecular Sciences</i> , <b>2019</b> , 20,	6.3	1
8	Frequency of the STRC-CATSPER2 deletion in STRC-associated hearing loss patients.. <i>Scientific Reports</i> , <b>2022</b> , 12, 634	4.9	1
7	Phylogeny and biogeography of arctic-alpine butterflies of the genus <i>Oeneis</i> (Nymphalidae: Satyrinae). <i>Entomological Science</i> , <b>2021</b> , 24, 183-195	1.1	1
6	Identification of a Novel Copy Number Variation of EYA4 Causing Autosomal Dominant Non-syndromic Hearing Loss. <i>Otology and Neurotology</i> , <b>2021</b> , 42, e866-e874	2.6	1
5	Advances in Molecular Genetics and the Molecular Biology of Deafness. <i>BioMed Research International</i> , <b>2016</b> , 2016, 5629093	3	1
4	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. <i>Genetic Testing and Molecular Biomarkers</i> , <b>2021</b> , 25, 79-83	1.6	1
3	Milestones toward cochlear gene therapy for patients with hereditary hearing loss. <i>Laryngoscope Investigative Otolaryngology</i> , <b>2021</b> , 6, 958-967	2.8	1
2	Etiology of hearing loss affects auditory skill development and vocabulary development in pediatric cochlear implantation cases.. <i>Acta Oto-Laryngologica</i> , <b>2022</b> , 1-8	1.6	0
1	Genetic Counseling for Patients with GJB2-Associated Hearing Loss. <i>Practica Otologica</i> , <b>2020</b> , 113, 223-233		