



・ 研究成果の刊行に関する一覧表



雑誌

発表者氏名	論文タイトル名	発表誌名	巻	ページ		出版年
Yano T, Nishio S, Usami S, deafness gene study consortium	Frequency of mitochondrial mutation in non-syndromic hearing loss as well as possibly responsible variants found by whole mitochondrial genome screening.	J Hum Genet	59	100	106	2014
Miyagawa M, Nishio S, Usami S.	Mutation spectrum and genotype-phenotype correlation of hearing loss patients caused by <i>SLC26A4</i> mutations in the Japanese: a large cohort study	J Hum Genet	59	262	268	2014
Ishikawa K, Naito T, Nishio S, Iwasa Y, Nakamura K, Usami S, Ichimura K.	A Japanese family showing high-frequency hearing loss with <i>KCNQ4</i> and <i>TECTA</i> mutations.	Acta Otolaryngol	134	557	563	2014
Abe S, Nagano M, Nishio S, Kumakawa K, Usami S.	High-frequency involved hearing loss Caused by Novel Mitochondrial DNA Mutation in 16S Ribosomal RNA Gene.	Otol Neurotol	35	1087	1090	2014
Shearer AE, Eppsteiner RW, Booth KT, Ephraim SS, Gurrola J2nd, Simpson A, Black-Ziegelbein EA, Joshi S, Ravi H, Giuffre AC, Happe S, Hildebrand MS, Azaiez H, Bayazit YA, Erdai ME, Lopez-Escamez JA, Gazquez I, Tamayo ML, Gelvez NY, Leal GL, Jalas C, Ekstein J, Yang T, Usami S, Kahrizi K, Bazazzadegan N, Najmabadi H, Scheetz TE, Braun TA, Casavant TL, LeProust EM, Smith RJ.	Utilizing ethnic-specific differences in minor allele frequency to recategorize reported pathogenic deafness variants.	Am J Hum Genet	95	445	453	2014
工 穰	単一遺伝子異常と疾患難聴と眼の異常	JOHNS	30	727	732	2014
西尾信哉, 宇佐美	難聴における遺伝子医	医学のあゆみ	250	371	377	2014

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宇佐美真一	知っておきたい甲状腺 診療 4 Pendred 症候群 の診断と治療	MB ENT	172	53	58	2014
Nishio S, Hayashi Y, Watanabe M, Usami S.	Clinical Application of a Custom AmpliSeq Library and Ion Torrent PGM Sequencing to Comprehensive Mutation Screening for Deafness Genes.	Genet Test Mol Biomarkers	19	1	9	2015
Miyagawa M, Nishio S, Sakurai Y, Hattori M, Tsukada K, Moteki H, Kojima H, Usami S.	The Patients Associated with <i>TMPRSS3</i> Mutations are Good Candidates for Electric Acoustic Stimulation.	Ann Otol Rhinol Laryngol	124	193	204	2015
Tsukada K, Ichinose A, Miyagawa M, Mori K, Hattori M, Nishio S, Naito Y, Kitajiri S, Usami S.	Detailed Hearing and Vestibular Profiles in the Patients with <i>COCH</i> Mutations.	Ann Otol Rhinol Laryngol	124	100	110	2015
Miyagawa M, Nishio S, Hattori M, Takumi Y, Usami S.	Germinal Mosaicism in a Family with BO Syndrome.	Ann Otol Rhinol Laryngol	124	118	122	2015
Mori K, Moteki H, Kobayashi Y, Azaiez H, Booth KT, Nishio S, Sato H, Smith RJ, Usami S.	Mutations in <i>LOXHD1</i> Gene Cause Various Types and Severities of Hearing Loss.	Ann Otol Rhinol Laryngol	124	135	141	2015
Mori K, Miyanojara I, Moteki H, Nishio S, Kuroyo Y, Usami S.	Novel Mutations in <i>GRXCR1</i> at DFNB25 Lead to Progressive Hearing Loss and Dizziness.	Ann Otol Rhinol Laryngol	124	129	134	2015
Moteki H, Azaiez H, Booth KT, Hattori M, Sato A, Sato Y, Motobayashi M, Sloan CM, Kolbe DL, Shearer AE, Smith RJ, Usami S.	Hearing Loss Caused by a <i>P2RX2</i> Mutation Identified in a MELAS Family with a Coexisting Mitochondrial 3243AG Mutation.	Ann Otol Rhinol Laryngol	124	177	183	2015
Ichinose A, Moteki H, Hattori M, Nishio S, Usami S.	Novel Mutation in <i>LRTOMT</i> Associated with Moderate Progressive Hearing Loss in Autosomal Recessive Inheritance.	Ann Otol Rhinol Laryngol	124	142	147	2015
Nishio S, Usami S.	Deafness Gene Variations in a 1120 Nonsyndromic Hearing Loss Cohort: Molecular Epidemiology and Deafness Mutation Spectrum of Patients in	Ann Otol Rhinol Laryngol	124	49	60	2015

	Japan.					
Miyagawa M, Nishio S, Ichinose A, Iwasaki S, Murata T, Kitajiri S, Usami S.	Mutational Spectrum and Clinical Features of patients with <i>ACTG1</i> Mutations identified by Massively Parallel DNA Sequencing.	Ann Otol Rhinol Laryngol	124	84	93	2015
Sakuma N, Moteki H, Azaiez H, Booth KT, Takahashi M, Arai Y, Shearer AE, Sloan CM, Nishio S, Kolbe DL, Iwasaki S, Oridate N, Smith RJ, Usami S.	Novel <i>PTPRQ</i> Mutations Identified in Three Congenital Hearing Loss Patients with Various Types of Hearing Loss.	Ann Otol Rhinol Laryngol	124	184	192	2015
Moteki H, Shearer AE, Izumi S, Kubota Y, Azaiez H, Booth KT, Sato A, Sloan CM, Kolbe DL, Shearer AE, Smith RJ, Usami S.	De Novo Mutation in X-Linked Hearing Loss-Associated <i>POU3F4</i> in a Sporadic case of Congenital Hearing Loss.	Ann Otol Rhinol Laryngol	124	169	176	2015
Miyagawa M, Nishio S, Hattori M, Moteki H, Kobayashi Y, Saton H, Watanabe T, Naito Y, Oshikawa C, Usami S.	Mutations in the <i>MYO15A</i> Gene are a Significant Cause of Nonsyndromic Hearing Loss: Massively Parallel DNA Sequencing-Based Analysis.	Ann Otol Rhinol Laryngol	124	158	168	2015
Tsukada K, Fukuoka H, Usami S.	Vestibular Functions of Hereditary Hearing Loss Patients with <i>GJB2</i> Mutations.	Audiol Neurootol	20	147	152	2015
Miyagawa M, Nishio S, Kumakaa K, Usami S.	Massively Parallel DNA Sequencing Successfully Identified Seven Families with Deafness-Associated <i>MYO6</i> Mutations: The Mutational Spectrum and Clinical Characteristics.	Ann Otol Rhinol Laryngol	124	148	157	2015

