Tatsuo Matsunaga

List of Publications by Year
in descending order

95 papers	1,817 citations	279798 23 h-index	345221 36 g-index
97 all docs	97 docs citations	97 times ranked	2226 citing authors

#	Article	IF	Citations
1	Phenotypeâ€“genotype correlation in patients with typical and atypical branchio-oto-renal syndrome. Scientific Reports, 2022, 12, 969.	3.3	6
2	Whole exome analysis of patients in Japan with hearing loss reveals high heterogeneity among responsible and novel candidate genes. Orphanet Journal of Rare Diseases, 2022, 17, 114.	2.7	3
3	Clinical genetics, practice, and research of deafblindness: From uncollected experiences to the national registry in Japan. Auris Nasus Larynx, 2021, 48, 185-193.	1.2	4
4	Disease-specific ACMG/AMP guidelines improve sequence variant interpretation for hearing loss. Genetics in Medicine, 2021, 23, 2208-2212.	2.4	18
5	Effects on cervical vestibular-evoked myogenic potentials of four clinically used head and neck measurement positions in healthy subjects. Acta Oto-Laryngologica, 2021, 141, 729-735.	0.9	1
6	Investigation of the hearing levels of siblings affected by a single GJB2 variant: Possibility of genetic modifiers. International Journal of Pediatric Otorhinolaryngology, 2021, 149, 110840.	1.0	2
7	A High Risk of Missing Congenital Cytomegalovirus-Associated Hearing Loss through Newborn Hearing Screening in Japan. Journal of Clinical Medicine, 2021, 10, 5056.	2.4	2
8	Clinical and genetic analysis of children with hearing loss and bilateral enlarged vestibular aqueducts. International Journal of Pediatric Otorhinolaryngology, 2021, 152, 110975.	1.0	0
9	A Japanese boy with double diagnoses of 2p15p16.1 microdeletion syndrome and RP2-associated retinal disorder. Human Genome Variation, 2021, 8, 46.	0.7	0
10	Systematic quantification of the anion transport function of pendrin (SLC26A4) and its diseaseâ€associated variants. Human Mutation, 2020, 41, 316-331.	2.5	16
11	Clinical Profiles of DFNA11 at Diverse Stages of Development and Aging in a Large Family Identified by Linkage Analysis. Otology and Neurotology, 2020, 41, e663-e673.	1.3	5
12	A phase I/IIa double blind single institute trial of low dose sirolimus for Pendred syndrome/DFNB4. Medicine (United States), 2020, 99, e19763.	1.0	11
13	Differences in hearing levels between siblings with hearing loss caused by GJB2 mutations. Auris Nasus Larynx, 2020, 47, 938-942.	1.2	2
14	Variants encoding a restricted carboxy-terminal domain of SLC12A2 cause hereditary hearing loss in humans. PLoS Genetics, 2020, 16, e1008643.	3.5	36
15	Clinical Features of GJB2â€•associated Hearing Loss Children. Journal of Otolaryngology of Japan, 2020, 123, 1225-1230.	0.1	2
16	Consensus interpretation of the p.Met34Thr and p.Val37Ile variants in GJB2 by the ClinGen Hearing Loss Expert Panel. Genetics in Medicine, 2019, 21, 2442-2452.	2.4	56
17	ClinGen expert clinical validity curation of 164 hearing loss geneâ€“disease pairs. Genetics in Medicine, 2019, 21, 2239-2247.	2.4	67
18	A clinical and genetic study of 16 Japanese families with Waardenburg syndrome. Gene, 2019, 704, 86-90.	2.2	17

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19	A Familial Case of a Whole Germline <i>CDC73</i> Deletion Discordant for Primary Hyperparathyroidism. Hormone Research in Paediatrics, 2019, 92, 56-63.	1.8	1
20	Estimating the concentration of therapeutic range using disease-specific iPS cells: Low-dose rapamycin therapy for Pendred syndrome. Regenerative Therapy, 2019, 10, 54-63.	3.0	17
21	Deterioration in Distortion Product Otoacoustic Emissions in Auditory Neuropathy Patients With Distinct Clinical and Genetic Backgrounds. Ear and Hearing, 2019, 40, 184-191.	2.1	17
22	Autosomal dominant optic atrophy with gene mutations accompanied by auditory neuropathy and other systemic complications in a Japanese cohort. Molecular Vision, 2019, 25, 559-573.	1.1	5
23	Clinically diverse phenotypes and genotypes of patients with branchio-oto-renal syndrome. Journal of Human Genetics, 2018, 63, 647-656.	2.3	31
24	High-level heteroplasmy for the m.7445A>G mitochondrial DNA mutation can cause progressive sensorineural hearing loss in infancy. International Journal of Pediatric Otorhinolaryngology, 2018, 108, 125-131.	1.0	6
25	IMSindel: An accurate intermediate-size indel detection tool incorporating de novo assembly and gapped global-local alignment with split read analysis. Scientific Reports, 2018, 8, 5608.	3.3	20
26	Spontaneous intramural duodenal hematoma as the manifestation of Noonan syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 496-498.	1.2	4
27	Homozygous EDNRB mutation in a patient with Waardenburg syndrome type 1. Auris Nasus Larynx, 2018, 45, 222-226.	1.2	17
28	Japanese pathogenic variant database: DPV. Translational Science of Rare Diseases, 2018, 3, 133-137.	1.5	1
29	Elongated EABR wave latencies observed in patients with auditory neuropathy caused by OTOF mutation. Laryngoscope Investigative Otolaryngology, 2018, 3, 388-393.	1.5	7
30	A case report of reversible generalized seizures in a patient with Waardenburg syndrome associated with a novel nonsense mutation in the penultimate exon of SOX10. BMC Pediatrics, 2018, 18, 171.	1.7	14
31	Gene expression dataset for whole cochlea of Macaca fascicularis. Scientific Reports, 2018, 8, 15554.	3.3	5
32	Response to â€œinfantile-onset deafness in m.7445A>G carriers may be multicausalâ€• International Journal of Pediatric Otorhinolaryngology, 2018, 111, 194.	1.0	0
33	The clinical features and prognosis of mumps-associated hearing loss: a retrospective, multi-institutional investigation in Japan. Acta Oto-Laryngologica, 2017, 137, S44-S47.	0.9	25
34	Differences between acoustic trauma and other types of acute noise-induced hearing loss in terms of treatment and hearing prognosis. Acta Oto-Laryngologica, 2017, 137, S48-S52.	0.9	14
35	Nationwide epidemiological survey of idiopathic sudden sensorineural hearing loss in Japan. Acta Oto-Laryngologica, 2017, 137, S8-S16.	0.9	52
36	Epidemiological survey of acute low-tone sensorineural hearing loss. Acta Oto-Laryngologica, 2017, 137, S34-S37.	0.9	22

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37	Idiopathic sudden sensorineural hearing loss and acute low-tone sensorineural hearing loss: a comparison of the results of a nationwide epidemiological survey in Japan. Acta Oto-Laryngologica, 2017, 137, S38-S43.	0.9	30
38	WFS1 and GJB2 mutations in patients with bilateral lowâ€frequency sensorineural hearing loss. Laryngoscope, 2017, 127, E324-E329.	2.0	14
39	The effect of initial treatment on hearing prognosis in idiopathic sudden sensorineural hearing loss: a nationwide survey in Japan. Acta Oto-Laryngologica, 2017, 137, S30-S33.	0.9	25
40	Relationships among drinking and smoking habits, history of diseases, body mass index and idiopathic sudden sensorineural hearing loss in Japanese patients. Acta Oto-Laryngologica, 2017, 137, S17-S23.	0.9	11
41	Cochlear Cell Modeling Using Disease-Specific iPSCs Unveils a Degenerative Phenotype and Suggests Treatments for Congenital Progressive Hearing Loss. Cell Reports, 2017, 18, 68-81.	6.4	63
42	The first sporadic case of DFNA11 identified by next-generation sequencing. International Journal of Pediatric Otorhinolaryngology, 2017, 100, 183-186.	1.0	6
43	A case of auditory neuropathy revealed by OTOF gene mutation analysis in a junior high school girl. Journal of Otology, 2017, 12, 202-206.	1.0	1
44	Mitochondrial mutations in maternally inherited hearing loss. BMC Medical Genetics, 2017, 18, 32.	2.1	19
45	Frequency and specific characteristics of the incomplete partition type III anomaly in children. Laryngoscope, 2017, 127, 1663-1669.	2.0	14
46	Prevalence of TECTA mutation in patients with mid-frequency sensorineural hearing loss. Orphanet Journal of Rare Diseases, 2017, 12, 157.	2.7	19
47	Genetics of Inner Ear Malformation and Cochlear Nerve Deficiency. , 2017, , 47-59.		1
48	The auditory phenotype of children harboring mutations in the prestin gene. Acta Oto-Laryngologica, 2016, 136, 397-401.	0.9	5
49	A novel nonsense mutation in the NOG gene causes familial NOG-related symphalangism spectrum disorder. Human Genome Variation, 2016, 3, 16023.	0.7	20
50	Molecular Impairment Mechanisms of Novel OPA1 Mutations Predicted by Molecular Modeling in Patients With Autosomal Dominant Optic Atrophy and Auditory Neuropathy Spectrum Disorder. Otology and Neurotology, 2016, 37, 394-402.	1.3	9
51	Recovery of endocochlear potential after severe damage to lateral wall fibrocytes following acute cochlear energy failure. NeuroReport, 2016, 27, 1159-1166.	1.2	3
52	Late onset and high-frequency dominant hearing loss in a family with MYH9 disorder. European Archives of Oto-Rhino-Laryngology, 2016, 273, 3547-3552.	1.6	6
53	A novel frameshift variant of COCH supports the hypothesis that haploinsufficiency is not a cause of autosomal dominant nonsyndromic deafness 9. Biochemical and Biophysical Research Communications, 2016, 469, 270-274.	2.1	9
54	Clinical characteristics of a Japanese family with hearing loss accompanied by compound heterozygous mutations in LOXHD1. Auris Nasus Larynx, 2016, 43, 609-613.	1.2	10

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55	New Trends in Research and Clinical Practice for Congenital Hearing Loss Caused by Gene Mutations. Japan Journal of Logopedics and Phoniatrics, 2015, 56, 219-225.	0.1	0
56	Neuroprotective effects of cutamesine, a ligand of the sigmaâ€1 receptor chaperone, against noiseâ€induced hearing loss. Journal of Neuroscience Research, 2015, 93, 788-795.	2.9	10
57	Attenuation of Progressive Hearing Loss in DBA/2J Mice by Reagents that Affect Epigenetic Modifications Is Associated with Up-Regulation of the Zinc Importer Zip4. PLoS ONE, 2015, 10, e0124301.	2.5	11
58	High prevalence of CDH23 mutations in patients with congenital high-frequency sporadic or recessively inherited hearing loss. Orphanet Journal of Rare Diseases, 2015, 10, 60.	2.7	34
59	Gene expression profiling of DBA/2J mice cochleae treated with l-methionine and valproic acid. Genomics Data, 2015, 5, 323-325.	1.3	1
60	A novel frameshift mutation in KCNQ4 in a family with autosomal recessive non-syndromic hearing loss. Biochemical and Biophysical Research Communications, 2015, 463, 582-586.	2.1	13
61	Pharmacological Inhibition of Cochlear Mitochondrial Respiratory Chain Induces Secondary Inflammation in the Lateral Wall: A Potential Therapeutic Target for Sensorineural Hearing Loss. PLoS ONE, 2014, 9, e90089.	2.5	26
62	Subgroups of enlarged vestibular aqueduct in relation to SLC26A4 mutations and hearing loss. Laryngoscope, 2014, 124, E134-40.	2.0	24
63	A mutation in the heparin-binding site of noggin as a novel mechanism of proximal symphalangism and conductive hearing loss. Biochemical and Biophysical Research Communications, 2014, 447, 496-502.	2.1	23
64	Chronic constipation recognized as a sign of a SOX10 mutation in a patient with Waardenburg syndrome. Gene, 2014, 540, 258-262.	2.2	6
65	GJB2-associated hearing loss undetected by hearing screening of newborns. Gene, 2013, 532, 41-45.	2.2	41
66	Moderate hearing loss associated with a novel KCNQ4 non-truncating mutation located near the N-terminus of the pore helix. Biochemical and Biophysical Research Communications, 2013, 432, 475-479.	2.1	10
67	High prevalence of inner-ear and/or internal auditory canal malformations in children with unilateral sensorineural hearing loss. International Journal of Pediatric Otorhinolaryngology, 2013, 77, 228-232.	1.0	64
68	Long-lasting changes in the cochlear K+ recycling structures after acute energy failure. Neuroscience Research, 2013, 77, 33-41.	1.9	4
69	Genetic analysis ofPAX3for diagnosis of Waardenburg syndrome type I. Acta Oto-Laryngologica, 2013, 133, 345-351.	0.9	15
70	Diverse spectrum of rare deafness genes underlies early-childhood hearing loss in Japanese patients: a cross-sectional, multi-center next-generation sequencing study. Orphanet Journal of Rare Diseases, 2013, 8, 172.	2.7	84
71	Cochlear Nerve Deficiency and Associated Clinical Features in Patients With Bilateral and Unilateral Hearing Loss. Otology and Neurotology, 2013, 34, 554-558.	1.3	52
72	A Novel Animal Model of Hearing Loss Caused by Acute Endoplasmic Reticulum Stress in the Cochlea. Journal of Pharmacological Sciences, 2012, 118, 363-372.	2.5	33

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73	A prevalent founder mutation and genotypeâ€“phenotype correlations of OTOF in Japanese patients with auditory neuropathy. Clinical Genetics, 2012, 82, 425-432.	2.0	68
74	Comorbidity of GJB2 and WFS1 mutations in one family. Gene, 2012, 501, 193-197.	2.2	7
75	In silico modeling of the pore region of a KCNQ4 missense mutant from a patient with hearing loss. BMC Research Notes, 2012, 5, 145.	1.4	11
76	Functional interaction between mesenchymal stem cells and spiral ligament fibrocytes. Journal of Neuroscience Research, 2012, 90, 1713-1722.	2.9	14
77	Late-phase recovery in the cochlear lateral wall following severe degeneration by acute energy failure. Brain Research, 2011, 1419, 1-11.	2.2	14
78	Acoustic overstimulation-induced apoptosis in fibrocytes of the cochlear spiral limbus of mice. European Archives of Oto-Rhino-Laryngology, 2011, 268, 973-978.	1.6	13
79	Systematic analysis of mitochondrial genes associated with hearing loss in the Japanese population: dHPLC reveals a new candidate mutation. BMC Medical Genetics, 2011, 12, 135.	2.1	13
80	Balance dysfunction resulting from acute inner ear energy failure is caused primarily by vestibular hair cell damage. Journal of Neuroscience Research, 2010, 88, 1262-1272.	2.9	10
81	Vestibular dysfunction in a Japanese patient with a mutation in the gene OPA1. Journal of the Neurological Sciences, 2010, 293, 23-28.	0.6	14
82	Enhanced expression of C/EBP homologous protein (CHOP) precedes degeneration of fibrocytes in the lateral wall after acute cochlear mitochondrial dysfunction induced by 3-nitropropionic acid. Neurochemistry International, 2010, 56, 487-494.	3.8	21
83	Value of Genetic Testing in the Otological Approach for Sensorineural Hearing Loss. Keio Journal of Medicine, 2009, 58, 216-222.	1.1	32
84	Mutations inMYH9Exons 1, 16, 26, and 30 Are Infrequently Found in Japanese Patients with Nonsyndromic Deafness. Genetic Testing and Molecular Biomarkers, 2009, 13, 705-707.	0.7	3
85	Expression of Pou3f3/Brnâ€1 and its genomic methylation in developing auditory epithelium. Developmental Neurobiology, 2009, 69, 913-930.	3.0	20
86	Trends in Genetic Research on Auditory Neuropathy. , 2009, , 43-50.		2
87	Caspase inhibitor facilitates recovery of hearing by protecting the cochlear lateral wall from acute cochlear mitochondrial dysfunction. Journal of Neuroscience Research, 2008, 86, 215-222.	2.9	23
88	Phenotypic consequences in a Japanese family having branchio-oto-renal syndrome with a novel frameshift mutation in the geneEYA1. Acta Oto-Laryngologica, 2007, 127, 98-104.	0.9	21
89	Mesenchymal Stem Cell Transplantation Accelerates Hearing Recovery through the Repair of Injured Cochlear Fibrocytes. American Journal of Pathology, 2007, 171, 214-226.	3.8	77
90	Clinical Course of Hearing and Language Development in GJB2 and Non-GJB2 Deafness following Habilitation with Hearing Aids. Audiology and Neuro-Otology, 2006, 11, 59-68.	1.3	21

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91	Clinical features of patients with GJB2 (connexin 26) mutations: severity of hearing loss is correlated with genotypes and protein expression patterns. Journal of Human Genetics, 2005, 50, 76-83.	2.3	79
92	Audiological Features and Mitochondrial DNA Sequence in a Large Family Carrying Mitochondrial A1555G Mutation without Use of Aminoglycoside. Annals of Otology, Rhinology and Laryngology, 2005, 114, 153-160.	1.1	15
93	Permanent Threshold Shift Caused by Acute Cochlear Mitochondrial Dysfunction Is Primarily Mediated by Degeneration of the Lateral Wall of the Cochlea. Audiology and Neuro-Otology, 2005, 10, 220-233.	1.3	40
94	Deafness Due to A1555G Mitochondrial Mutation Without Use of Aminoglycoside. Laryngoscope, 2004, 114, 1085-1091.	2.0	27
95	A novel animal model of acute cochlear mitochondrial dysfunction. NeuroReport, 2004, 15, 1597-1600.	1.2	50