Masanori P Takahashi

List of Publications by Year
in descending order

73	3,452	236925 25	144013 57
papers	citations	h-index	g-index

78	78	78	3392
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Characteristics of myotonic dystrophy patients in the national registry of Japan. Journal of the Neurological Sciences, 2022, 432, 120080.	0.6	2
2	Quality of life and subjective symptom impact in Japanese patients with myotonic dystrophy type 1. BMC Neurology, 2022, 22, 55.	1.8	1
3	Mature Myotubes Generated From Human-Induced Pluripotent Stem Cells Without Forced Gene Expression. Frontiers in Cell and Developmental Biology, 2022, 10, .	3.7	3
4	The practice of active patient involvement in rare disease research using ICT: experiences and lessons from the RUDY JAPAN project. Research Involvement and Engagement, 2021, 7, 9.	2.9	10
5	Clinicopathologic Features of Oculopharyngodistal Myopathy WithLRP12CGG Repeat Expansions Compared With Other Oculopharyngodistal Myopathy Subtypes. JAMA Neurology, 2021, 78, 853.	9.0	30
6	Metabolic complications in myotonic dystrophy type 1: A cross-sectional survey using the National Registry of Japan. Journal of the Neurological Sciences, 2021, 427, 117511.	0.6	3
7	A nationwide survey of episodic ataxia in Japan. Neurology and Clinical Neuroscience, 2021, 9, 443.	0.4	0
8	Hemiplegic migraine type 2 caused by a novel variant within the P-type ATPase motif in ATP1A2 concomitant with a CACNA1A variant. Brain and Development, 2021, 43, 952-957.	1.1	0
9	Cardiac Emerinopathy. Circulation: Arrhythmia and Electrophysiology, 2020, 13, e008712.	4.8	20
10	Hypokalaemic periodic paralysis with a charge-retaining substitution in the voltage sensor. Brain Communications, 2020, 2, fcaa103.	3.3	11
11	Cardiac Conduction Disorders as Markers of Cardiac Events in Myotonic Dystrophy Type 1. Journal of the American Heart Association, 2020, 9, e015709.	3.7	12
12	Mutation spectrum and health status in skeletal muscle channelopathies in Japan. Neuromuscular Disorders, 2020, 30, 546-553.	0.6	9
13	EF handâ€like motif mutations of Nav1.4 Câ€terminus cause myotonic syndrome by impairing fast inactivation. Muscle and Nerve, 2020, 61, 808-814.	2.2	7
14	The expanding phenotype of hypokalemic periodic paralysis in a Japanese family with p.Val876Glu mutation in CACNA1S. Molecular Genetics & Genomic Medicine, 2020, 8, e1175.	1.2	4
15	A slipped-CAG DNA-binding small molecule induces trinucleotide-repeat contractions in vivo. Nature Genetics, 2020, 52, 146-159.	21.4	110
16	Analysis of the genetic background associated with sporadic periodic paralysis in Japanese patients. Journal of the Neurological Sciences, 2020, 412, 116795.	0.6	5
17	Functional analysis of a double-point mutation in the KCNJ2 gene identified in a family with Andersen-Tawil syndrome. Journal of the Neurological Sciences, 2019, 407, 116521.	0.6	2
18	The myotonic dystrophy health index: Japanese adaption and validity testing. Muscle and Nerve, 2019, 59, 577-582.	2.2	5

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19	A family with both X-linked dominant Charcot-Marie-Tooth disease and myotonic dystrophy type 1 mutations with phenotypic variations. Neurology and Clinical Neuroscience, 2019, 7, 88-90.	0.4	0
20	Longâ€term safety and efficacy of eculizumab in generalized myasthenia gravis. Muscle and Nerve, 2019, 60, 14-24.	2.2	162
21	Transient thyrotoxicosisâ€aggravated attacks of paralysis in a patient with hereditary hypokalemic periodic paralysis type 2. Neurology and Clinical Neuroscience, 2019, 7, 354-356.	0.4	0
22	Straightjacket/Î±2Î´3 deregulation is associated with cardiac conduction defects in myotonic dystrophy type 1. ELife, 2019, 8, .	6.0	8
23	Macroscopic and microscopic diversity of missplicing in the central nervous system of patients with myotonic dystrophy type 1. NeuroReport, 2018, 29, 235-240.	1.2	8
24	Validation of The Individualized Neuromuscular Quality of Life in Japanese patients with myotonic dystrophy. Muscle and Nerve, 2018, 58, 56-63.	2.2	7
25	GNE myopathy caused by a synonymous mutation leading to aberrant mRNA splicing. Neuromuscular Disorders, 2018, 28, 154-157.	0.6	9
26	Cognitive impairment and quality of life in patients with myotonic dystrophy type 1. Muscle and Nerve, 2018, 57, 742-748.	2.2	29
27	Hypokalemic Periodic Paralysis Cases with Substitutions from Arginine to Lysine in the Voltage Sensor. Biophysical Journal, 2018, 114, 636a.	0.5	0
28	Clinical Features of Skeletal Muscle and Their Underlying Molecular Mechanism. , 2018, , 45-61.		0
29	Eight years after an international workshop on myotonic dystrophy patient registries: case study of a global collaboration for a rare disease. Orphanet Journal of Rare Diseases, 2018, 13, 155.	2.7	19
30	rbFOX1/MBNL1 competition for CCUG RNA repeats bindingÂcontributes to myotonic dystrophy typeÂ1/typeÂ2 differences. Nature Communications, 2018, 9, 2009.	12.8	61
31	Myotonic dystrophy type 1 patient-derived iPSCs for the investigation of CTG repeat instability. Scientific Reports, 2017, 7, 42522.	3.3	34
32	Aberrant Myokine Signaling in Congenital Myotonic Dystrophy. Cell Reports, 2017, 21, 1240-1252.	6.4	40
33	An elderly-onset limb girdle muscular dystrophy type 1B (LGMD1B) with pseudo-hypertrophy of paraspinal muscles. Neuromuscular Disorders, 2016, 26, 593-597.	0.6	1
34	Oral administration of erythromycin decreases <scp>RNA</scp> toxicity in myotonic dystrophy. Annals of Clinical and Translational Neurology, 2016, 3, 42-54.	3.7	55
35	A case of non-dystrophic myotonia with concomitant mutations in the SCN4A and CLCN1 genes. Journal of the Neurological Sciences, 2016, 369, 254-258.	0.6	19
36	Splicing misregulation of SCN5A contributes to cardiac-conduction delay and heart arrhythmia in myotonic dystrophy. Nature Communications, 2016, 7, 11067.	12.8	155

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37	Using digital technologies to engage with medical research: views of myotonic dystrophy patients in Japan. BMC Medical Ethics, 2016, 17, 51.	2.4	19
38	Myotonic Dystrophy. , 2016, , 39-61.		4
39	Large expansion of CTGâ€¢CAG repeats is exacerbated by MutSÎ² in human cells. Scientific Reports, 2015, 5, 11020.	3.3	37
40	Phenotypic Variability in Childhood of Skeletal Muscle Sodium Channelopathies. Pediatric Neurology, 2015, 52, 504-508.	2.1	12
41	Efficacy of deep brain stimulation in an adolescent patient with <scp>DYT</scp>11 myoclonusâ€dystonia. Neurology and Clinical Neuroscience, 2014, 2, 57-59.	0.4	3
42	Simple questionnaire for screening patients with myotonic dystrophy type 1. Neurology and Clinical Neuroscience, 2014, 2, 97-103.	0.4	4
43	A Kir3.4 mutation causes Andersenâ€“Tawil syndrome by an inhibitory effect on Kir2.1. Neurology, 2014, 82, 1058-1064.	1.1	59
44	Manumycin A corrects aberrant splicing of Clcn1 in myotonic dystrophy type 1 (DM1) mice. Scientific Reports, 2013, 3, 2142.	3.3	21
45	Ultrasound-enhanced delivery of Morpholino with Bubble liposomes ameliorates the myotonia of myotonic dystrophy model mice. Scientific Reports, 2013, 3, 2242.	3.3	23
46	A sodium channel myotonia due to a novel SCN4A mutation accompanied by acquired autoimmune myasthenia gravis. Neuroscience Letters, 2012, 519, 67-72.	2.1	9
47	A novel mutation in SCN4A causes severe myotonia and school-age-onset paralytic episodes. Journal of the Neurological Sciences, 2012, 315, 15-19.	0.6	16
48	Muscleblind-like 2-Mediated Alternative Splicing in the Developing Brain and Dysregulation in Myotonic Dystrophy. Neuron, 2012, 75, 437-450.	8.1	296
49	Muscleblind-Like 1 Knockout Mice Reveal Novel Splicing Defects in the Myotonic Dystrophy Brain. PLoS ONE, 2012, 7, e33218.	2.5	79
50	A novel mutation in the calcium channel gene in a family with hypokalemic periodic paralysis. Journal of the Neurological Sciences, 2011, 309, 9-11.	0.6	13
51	Misregulation of miR-1 processing is associated with heart defects in myotonic dystrophy. Nature Structural and Molecular Biology, 2011, 18, 840-845.	8.2	248
52	Misregulated alternative splicing of BIN1 is associated with T tubule alterations and muscle weakness in myotonic dystrophy. Nature Medicine, 2011, 17, 720-725.	30.7	299
53	A mutation in a rare type of intron in a sodium-channel gene results in aberrant splicing and causes myotonia. Human Mutation, 2011, 32, 773-782.	2.5	20
54	The Role of Alpha-Dystrobrevin in Striated Muscle. International Journal of Molecular Sciences, 2011, 12, 1660-1671.	4.1	27

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55	An Intronic Mutation of SCN4A Associated with Myotonia Raises an Aberrantly Spliced Isoform with Disrupted Fast Inactivation. Biophysical Journal, 2010, 98, 311a-312a.	0.5	0
56	New mutation of the Na channel in the severe form of potassiumâ€aggravated myotonia. Muscle and Nerve, 2009, 39, 666-673.	2.2	41
57	A cross-sectional study for glucose intolerance of myotonic dystrophy. Journal of the Neurological Sciences, 2009, 276, 60-65.	0.6	35
58	Molecular mechanisms responsible for aberrant splicing of SERCA1 in myotonic dystrophy type 1. Human Molecular Genetics, 2007, 16, 2834-2843.	2.9	92
59	Altered mRNA splicing of dystrophin in type 1 myotonic dystrophy. Muscle and Nerve, 2007, 36, 251-257.	2.2	42
60	Endoplasmic reticulum stress in myotonic dystrophy type 1 muscle. Acta Neuropathologica, 2007, 114, 527-535.	7.7	52
61	Class Ic antiarrhythmics block human skeletal muscle Na channel during myotonia-like stimulation. European Journal of Pharmacology, 2006, 532, 24-31.	3.5	10
62	Altered mRNA splicing of the skeletal muscle ryanodine receptor and sarcoplasmic/endoplasmic reticulum Ca2+-ATPase in myotonic dystrophy type 1. Human Molecular Genetics, 2005, 14, 2189-2200.	2.9	247
63	Expression and distribution of a small-conductance calcium-activated potassium channel (SK3) protein in skeletal muscles from myotonic muscular dystrophy patients and congenital myotonic mice. Neuroscience Letters, 2003, 347, 191-195.	2.1	13
64	Expanded CUG Repeats Trigger Aberrant Splicing of ClC-1 Chloride Channel Pre-mRNA and Hyperexcitability of Skeletal Muscle in Myotonic Dystrophy. Molecular Cell, 2002, 10, 35-44.	9.7	611
65	The expression of ion channel mRNAs in skeletal muscles from patients with myotonic muscular dystrophy. Neuroscience Letters, 2000, 295, 93-96.	2.1	34
66	Calcium increase in mouse skeletal muscles by triparanol: a drug to induce myotonic dystrophy-like clinical manifestations. Neuroscience Letters, 1999, 272, 87-90.	2.1	4
67	Enhanced Slow Inactivation by V445M: A Sodium Channel Mutation Associated with Myotonia. Biophysical Journal, 1999, 76, 861-868.	0.5	60
68	Lambert-Eaton syndrome antibodies inhibit acetylcholine release and P/Q-type Ca2+channels in electric ray nerve endings. Journal of Physiology, 1998, 508, 427-438.	2.9	11
69	Synaptic input-induced increase in intraneuronal Ca2+ in the medial vestibular nucleus of young rats. Neuroscience Research, 1994, 21, 59-69.	1.9	17
70	Intracellular calcium increase induced by GABA in visual cortex of fetal and neonatal rats and its disappearance with development. Neuroscience Research, 1994, 20, 85-94.	1.9	74
71	Induction of LTD but not LTP through metabotropic glutamate receptors in visual cortex. NeuroReport, 1994, 5, 1829-1832.	1.2	46
72	Laminar difference in tetanus-induced increase of intracellular Ca2+ in visual cortex of young rats. Neuroscience Research, 1993, 17, 217-228.	1.9	14

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73	Contribution of NMDA receptors to tetanus-induced increase in postsynaptic Ca2+ in visual cortex of young rats. Neuroscience Research, 1993, 17, 229-239.	1.9	14