

# Masanori P Takahashi

## List of Publications by Year in descending order

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73  
papers

3,452  
citations

236925

25  
h-index

144013

57  
g-index

78  
all docs

78  
docs citations

78  
times ranked

3392  
citing authors

#	ARTICLE	IF	CITATIONS
1	Expanded CUG Repeats Trigger Aberrant Splicing of ClC-1 Chloride Channel Pre-mRNA and Hyperexcitability of Skeletal Muscle in Myotonic Dystrophy. <i>Molecular Cell</i> , 2002, 10, 35-44.	9.7	611
2	Misregulated alternative splicing of BIN1 is associated with T tubule alterations and muscle weakness in myotonic dystrophy. <i>Nature Medicine</i> , 2011, 17, 720-725.	30.7	299
3	Muscleblind-like 2-Mediated Alternative Splicing in the Developing Brain and Dysregulation in Myotonic Dystrophy. <i>Neuron</i> , 2012, 75, 437-450.	8.1	296
4	Misregulation of miR-1 processing is associated with heart defects in myotonic dystrophy. <i>Nature Structural and Molecular Biology</i> , 2011, 18, 840-845.	8.2	248
5	Altered mRNA splicing of the skeletal muscle ryanodine receptor and sarcoplasmic/endoplasmic reticulum Ca <sup>2+</sup> -ATPase in myotonic dystrophy type 1. <i>Human Molecular Genetics</i> , 2005, 14, 2189-2200.	2.9	247
6	Long-term safety and efficacy of eculizumab in generalized myasthenia gravis. <i>Muscle and Nerve</i> , 2019, 60, 14-24.	2.2	162
7	Splicing misregulation of SCN5A contributes to cardiac-conduction delay and heart arrhythmia in myotonic dystrophy. <i>Nature Communications</i> , 2016, 7, 11067.	12.8	155
8	A slipped-CAG DNA-binding small molecule induces trinucleotide-repeat contractions in vivo. <i>Nature Genetics</i> , 2020, 52, 146-159.	21.4	110
9	Molecular mechanisms responsible for aberrant splicing of SERCA1 in myotonic dystrophy type 1. <i>Human Molecular Genetics</i> , 2007, 16, 2834-2843.	2.9	92
10	Muscleblind-Like 1 Knockout Mice Reveal Novel Splicing Defects in the Myotonic Dystrophy Brain. <i>PLoS ONE</i> , 2012, 7, e33218.	2.5	79
11	Intracellular calcium increase induced by GABA in visual cortex of fetal and neonatal rats and its disappearance with development. <i>Neuroscience Research</i> , 1994, 20, 85-94.	1.9	74
12	rbFOX1/MBNL1 competition for CCUG RNA repeats binding contributes to myotonic dystrophy type 1/type 2 differences. <i>Nature Communications</i> , 2018, 9, 2009.	12.8	61
13	Enhanced Slow Inactivation by V445M: A Sodium Channel Mutation Associated with Myotonia. <i>Biophysical Journal</i> , 1999, 76, 861-868.	0.5	60
14	A Kir3.4 mutation causes Andersen's Tawil syndrome by an inhibitory effect on Kir2.1. <i>Neurology</i> , 2014, 82, 1058-1064.	1.1	59
15	Oral administration of erythromycin decreases <sc>RNA</sc> toxicity in myotonic dystrophy. <i>Annals of Clinical and Translational Neurology</i> , 2016, 3, 42-54.	3.7	55
16	Endoplasmic reticulum stress in myotonic dystrophy type 1 muscle. <i>Acta Neuropathologica</i> , 2007, 114, 527-535.	7.7	52
17	Induction of LTD but not LTP through metabotropic glutamate receptors in visual cortex. <i>NeuroReport</i> , 1994, 5, 1829-1832.	1.2	46
18	Altered mRNA splicing of dystrophin in type 1 myotonic dystrophy. <i>Muscle and Nerve</i> , 2007, 36, 251-257.	2.2	42

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19	New mutation of the Na channel in the severe form of potassium-aggravated myotonia. <i>Muscle and Nerve</i> , 2009, 39, 666-673.	2.2	41
20	Aberrant Myokine Signaling in Congenital Myotonic Dystrophy. <i>Cell Reports</i> , 2017, 21, 1240-1252.	6.4	40
21	Large expansion of CTG-CAG repeats is exacerbated by MutS <sup>2</sup> in human cells. <i>Scientific Reports</i> , 2015, 5, 11020.	3.3	37
22	A cross-sectional study for glucose intolerance of myotonic dystrophy. <i>Journal of the Neurological Sciences</i> , 2009, 276, 60-65.	0.6	35
23	The expression of ion channel mRNAs in skeletal muscles from patients with myotonic muscular dystrophy. <i>Neuroscience Letters</i> , 2000, 295, 93-96.	2.1	34
24	Myotonic dystrophy type 1 patient-derived iPSCs for the investigation of CTG repeat instability. <i>Scientific Reports</i> , 2017, 7, 42522.	3.3	34
25	Clinicopathologic Features of Oculopharyngodistal Myopathy With LRP12 CCG Repeat Expansions Compared With Other Oculopharyngodistal Myopathy Subtypes. <i>JAMA Neurology</i> , 2021, 78, 853.	9.0	30
26	Cognitive impairment and quality of life in patients with myotonic dystrophy type 1. <i>Muscle and Nerve</i> , 2018, 57, 742-748.	2.2	29
27	The Role of Alpha-Dystrobrevin in Striated Muscle. <i>International Journal of Molecular Sciences</i> , 2011, 12, 1660-1671.	4.1	27
28	Ultrasound-enhanced delivery of Morpholino with Bubble liposomes ameliorates the myotonia of myotonic dystrophy model mice. <i>Scientific Reports</i> , 2013, 3, 2242.	3.3	23
29	Manumycin A corrects aberrant splicing of Clcn1 in myotonic dystrophy type 1 (DM1) mice. <i>Scientific Reports</i> , 2013, 3, 2142.	3.3	21
30	A mutation in a rare type of intron in a sodium-channel gene results in aberrant splicing and causes myotonia. <i>Human Mutation</i> , 2011, 32, 773-782.	2.5	20
31	Cardiac Emerinopathy. <i>Circulation: Arrhythmia and Electrophysiology</i> , 2020, 13, e008712.	4.8	20
32	A case of non-dystrophic myotonia with concomitant mutations in the SCN4A and CLCN1 genes. <i>Journal of the Neurological Sciences</i> , 2016, 369, 254-258.	0.6	19
33	Using digital technologies to engage with medical research: views of myotonic dystrophy patients in Japan. <i>BMC Medical Ethics</i> , 2016, 17, 51.	2.4	19
34	Eight years after an international workshop on myotonic dystrophy patient registries: case study of a global collaboration for a rare disease. <i>Orphanet Journal of Rare Diseases</i> , 2018, 13, 155.	2.7	19
35	Synaptic input-induced increase in intraneuronal Ca <sup>2+</sup> in the medial vestibular nucleus of young rats. <i>Neuroscience Research</i> , 1994, 21, 59-69.	1.9	17
36	A novel mutation in SCN4A causes severe myotonia and school-age-onset paralytic episodes. <i>Journal of the Neurological Sciences</i> , 2012, 315, 15-19.	0.6	16

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37	Laminar difference in tetanus-induced increase of intracellular Ca <sup>2+</sup> in visual cortex of young rats. <i>Neuroscience Research</i> , 1993, 17, 217-228.	1.9	14
38	Contribution of NMDA receptors to tetanus-induced increase in postsynaptic Ca <sup>2+</sup> in visual cortex of young rats. <i>Neuroscience Research</i> , 1993, 17, 229-239.	1.9	14
39	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. <i>Neuroscience Letters</i> , 2003, 347, 191-195.	2.1	13
40	A novel mutation in the calcium channel gene in a family with hypokalemic periodic paralysis. <i>Journal of the Neurological Sciences</i> , 2011, 309, 9-11.	0.6	13
41	Phenotypic Variability in Childhood of Skeletal Muscle Sodium Channelopathies. <i>Pediatric Neurology</i> , 2015, 52, 504-508.	2.1	12
42	Cardiac Conduction Disorders as Markers of Cardiac Events in Myotonic Dystrophy Type 1. <i>Journal of the American Heart Association</i> , 2020, 9, e015709.	3.7	12
43	Lambert-Eaton syndrome antibodies inhibit acetylcholine release and P/Q-type Ca <sup>2+</sup> channels in electric ray nerve endings. <i>Journal of Physiology</i> , 1998, 508, 427-438.	2.9	11
44	Hypokalaemic periodic paralysis with a charge-retaining substitution in the voltage sensor. <i>Brain Communications</i> , 2020, 2, fcaa103.	3.3	11
45	Class Ic antiarrhythmics block human skeletal muscle Na channel during myotonia-like stimulation. <i>European Journal of Pharmacology</i> , 2006, 532, 24-31.	3.5	10
46	The practice of active patient involvement in rare disease research using ICT: experiences and lessons from the RUDY JAPAN project. <i>Research Involvement and Engagement</i> , 2021, 7, 9.	2.9	10
47	A sodium channel myotonia due to a novel SCN4A mutation accompanied by acquired autoimmune myasthenia gravis. <i>Neuroscience Letters</i> , 2012, 519, 67-72.	2.1	9
48	GNE myopathy caused by a synonymous mutation leading to aberrant mRNA splicing. <i>Neuromuscular Disorders</i> , 2018, 28, 154-157.	0.6	9
49	Mutation spectrum and health status in skeletal muscle channelopathies in Japan. <i>Neuromuscular Disorders</i> , 2020, 30, 546-553.	0.6	9
50	Macroscopic and microscopic diversity of missplicing in the central nervous system of patients with myotonic dystrophy type 1. <i>NeuroReport</i> , 2018, 29, 235-240.	1.2	8
51	Straightjacket/±2 <sup>3</sup> deregulation is associated with cardiac conduction defects in myotonic dystrophy type 1. <i>ELife</i> , 2019, 8, .	6.0	8
52	Validation of The Individualized Neuromuscular Quality of Life in Japanese patients with myotonic dystrophy. <i>Muscle and Nerve</i> , 2018, 58, 56-63.	2.2	7
53	EF hand-like motif mutations of Nav1.4 C-terminus cause myotonic syndrome by impairing fast inactivation. <i>Muscle and Nerve</i> , 2020, 61, 808-814.	2.2	7
54	The myotonic dystrophy health index: Japanese adaption and validity testing. <i>Muscle and Nerve</i> , 2019, 59, 577-582.	2.2	5

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55	Analysis of the genetic background associated with sporadic periodic paralysis in Japanese patients. <i>Journal of the Neurological Sciences</i> , 2020, 412, 116795.	0.6	5
56	Calcium increase in mouse skeletal muscles by triparanol: a drug to induce myotonic dystrophy-like clinical manifestations. <i>Neuroscience Letters</i> , 1999, 272, 87-90.	2.1	4
57	Simple questionnaire for screening patients with myotonic dystrophy type 1. <i>Neurology and Clinical Neuroscience</i> , 2014, 2, 97-103.	0.4	4
58	The expanding phenotype of hypokalemic periodic paralysis in a Japanese family with p.Val876Glu mutation in <i>CACNA1S</i> . <i>Molecular Genetics &amp; Genomic Medicine</i> , 2020, 8, e1175.	1.2	4
59	Myotonic Dystrophy. , 2016, , 39-61.		4
60	Efficacy of deep brain stimulation in an adolescent patient with <i>DYT</i> 11 myoclonus&dystonia. <i>Neurology and Clinical Neuroscience</i> , 2014, 2, 57-59.	0.4	3
61	Metabolic complications in myotonic dystrophy type 1: A cross-sectional survey using the National Registry of Japan. <i>Journal of the Neurological Sciences</i> , 2021, 427, 117511.	0.6	3
62	Mature Myotubes Generated From Human-Induced Pluripotent Stem Cells Without Forced Gene Expression. <i>Frontiers in Cell and Developmental Biology</i> , 2022, 10, .	3.7	3
63	Functional analysis of a double-point mutation in the <i>KCNJ2</i> gene identified in a family with Andersen-Tawil syndrome. <i>Journal of the Neurological Sciences</i> , 2019, 407, 116521.	0.6	2
64	Characteristics of myotonic dystrophy patients in the national registry of Japan. <i>Journal of the Neurological Sciences</i> , 2022, 432, 120080.	0.6	2
65	An elderly-onset limb girdle muscular dystrophy type 1B (LGMD1B) with pseudo-hypertrophy of paraspinal muscles. <i>Neuromuscular Disorders</i> , 2016, 26, 593-597.	0.6	1
66	Quality of life and subjective symptom impact in Japanese patients with myotonic dystrophy type 1. <i>BMC Neurology</i> , 2022, 22, 55.	1.8	1
67	An Intronic Mutation of <i>SCN4A</i> Associated with Myotonia Raises an Aberrantly Spliced Isoform with Disrupted Fast Inactivation. <i>Biophysical Journal</i> , 2010, 98, 311a-312a.	0.5	0
68	Hypokalemic Periodic Paralysis Cases with Substitutions from Arginine to Lysine in the Voltage Sensor. <i>Biophysical Journal</i> , 2018, 114, 636a.	0.5	0
69	Clinical Features of Skeletal Muscle and Their Underlying Molecular Mechanism. , 2018, , 45-61.		0
70	A family with both X-linked dominant Charcot-Marie-Tooth disease and myotonic dystrophy type 1 mutations with phenotypic variations. <i>Neurology and Clinical Neuroscience</i> , 2019, 7, 88-90.	0.4	0
71	Transient thyrotoxicosis&aggravated attacks of paralysis in a patient with hereditary hypokalemic periodic paralysis type 2. <i>Neurology and Clinical Neuroscience</i> , 2019, 7, 354-356.	0.4	0
72	A nationwide survey of episodic ataxia in Japan. <i>Neurology and Clinical Neuroscience</i> , 2021, 9, 443.	0.4	0

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73	Hemiplegic migraine type 2 caused by a novel variant within the P-type ATPase motif in ATP1A2 concomitant with a CACNA1A variant. <i>Brain and Development</i> , 2021, 43, 952-957.	1.1	0