

【論文】

発表者名	論文タイトル名	発表誌	巻・号	ページ	出版年
Inoue-Shibui A, Niihori T, Kobayashi M, Suzuki N, Izumi R, Warita H, Hara K, Shirota M, Funayama R, Nakayama K, Nishino I, <u>Aoki M</u> , Aoki Y.	A novel deletion in the C-terminal region of HSPB8 in a family with rimmed vacuolar myopathy.	J Hum Genet	doi: 10.1038/ /s10038- 021- 00916-y.		2021
Amato AA, Hanna MG, Machado PM, Badrising UA, Chinoy H, Benveniste O, Karanam AK, Wu M, Tankó LB, Schubert-Tennigkeit AA, Papanicolaou DA, Lloyd TE, Needham M, Liang C, Reardon KA, de Visser M, Ascherman DP, Barohn RJ, Dimachkie MM, Miller JAL, Kissel JT, Oskarsson B, Joyce NC, Van den Bergh P, Baets J, De Bleecker JL, Karam C, David WS, Mirabella M, Nations SP, Jung HH, Pegoraro E, Maggi L, Rodolico C, Filosto M, Shaibani AI, Sivakumar K, Goyal NA, Mori-Yoshimura M, Yamashita S, Suzuki N, <u>Aoki M</u> , Katsuno M, Morihata H, Murata K, Nodera H, Nishino I, Romano CD, Williams VSL, Vissing J, Zhang Auberson L.	RESILIENT Study Extension Group. Efficacy and Safety of Bimagrumab in Sporadic Inclusion Body Myositis: Long-term Extension of RESILIENT.	Neurology	96・12	e1595- e1607	2021

Oikawa Y, Izumi R, Koide M, Hagiwara Y, Kanzaki M, Suzuki N, Kikuchi K, Matsuhashi T, Akiyama Y, Ichijo M, Watanabe S, Toyohara T, Suzuki T, Mishima E, Akiyama Y, Ogata Y, Suzuki C, Hayashi H, Kodama EN, Hayashi KI, Itoi E, <u>Aoki M</u> , Kure S, Abe T.	Mitochondrial dysfunction underlying sporadic inclusion body myositis is ameliorated by the mitochondrial homing drug MA-5.	PLoS One	15•12	e0231064	2020
Suzuki N, Soga T, Izumi R, Toyoshima M, Shibasaki M, Sato I, Kudo Y, <u>Aoki M</u> , Kato M.	Hybrid Assistive Limb® for sporadic inclusion body myositis: A case series.	J Clin Neurosci	81	92-94	2020
Kitajima Y, Suzuki N, Yoshioka K, Izumi R, Tateyama M, Tashiro Y, Takahashi R, <u>Aoki M</u> , Ono Y.	Inducible Rpt3, a Proteasome Component, Knockout in Adult Skeletal Muscle Results in Muscle Atrophy.	Front Cell Dev Biol.	8	859	2020
Samukawa M, Nakamura N, Hirano M, Morikawa M, Sakata H, Nishino I, Izumi R, Suzuki N, Kuroda H, Shiga K, Saigoh K, <u>Aoki M</u> , Kusunoki S.	Neutral Lipid Storage Disease Associated with the PNPLA2 Gene: Case Report and Literature Review.	Eur Neurol	83	317-322	2020
Izumi R, Takahashi T, Suzuki N, Niihori T, Ono H, Nakamura N, Katada S, Kato M, Warita H, Tateyama M, Aoki Y, <u>Aoki M</u> .	The genetic profile of dysferlinopathy in a cohort of 209 cases: Genotype-phenotype relationship and a hotspot on the inner DysF domain.	Hum Mutat	41	1540-1554	2020

Ono H, Suzuki N, Kanno SI, Kawahara G, Izumi R, Takahashi T, Kitajima Y, Osana S, Nakamura N, Akiyama T, Ikeda K, Shijo T, Mitsuzawa S, Nagatomi R, Araki N, Yasui A, Warita H, Hayashi YK, Miyake K, <u>Aoki M</u> .	AMPK Complex Activation Promotes Sarcolemmal Repair in Dysferlinopathy.	Mol Ther	28	1133-1153	2020
Samukawa M, Nakamura N, Hirano M, Morikawa M, Sakata H, <u>Nishino I</u> , Izumi R, Suzuki N, Kuroda H, Shiga K, Saigoh K, Aoki M, Kusunoki S.	Neutral Lipid Storage Disease Associated With the PNPLA2 Gene: Case Report and Literature Review.	Eur Neurol	83 • 3	317-322	2020
Yoshioka W, Miyasaka N, Okubo R, Shimizu R, Takahashi Y, Oda Y, <u>Nishino I</u> , Nakamura H, Mori-Yoshimura M.	Pregnancy in GNE myopathy patients: a nationwide repository survey in Japan.	Orphanet J Rare Dis	15 • 1	245	2020
Ogasawara, M, Iida, A, Kumutponpanich, T, Ozaki A, Oya Y, Konishi, H, Nakamura, A, Abe, R, Takai, H, Hanajima, R, Doi, H, Tanaka F, Nakamura H, Nonaka I, Wang, Z, Hayashi S. Noguchi S, <u>Nishino I</u> .	CGG expansion in NOTCH2NLC is associated with oculopharyngodistal myopathy with neurological manifestations.	Acta Neuropathol Commun	8 • 1	204	2020
Deng, J, Yu, J, Li, P, Luan, X, Cao, L, Zhao, J, Yu, M, Zhang, W, Lv, H, Xie, Z, Meng, L, Zheng, Y, Zhao, Y, Gang, Q, Wang, Q, Liu, J, Zhu, M, Guo, X, Su, Y, Liang, Y, Liang, F, Hayashi, T,	Expansion of GGC Repeat in GIPC1 Is Associated with Oculopharyngodistal Myopathy.	Am J Hum Genet	106 • 1	793-804	2020

Maeda, M. H, Sato, T, Ura, S, Oya, Y, Ogasawara, M, Iida, A, <u>Nishino, I</u> , Zhou, C, Yan, C, Yuan, Y, Hong, D, Wang, Z.					
Ono H, Suzuki N, Kanno S, Kawahara G, Izumi R, Takahashi T, Kitajima Y, Osana S, Nakamura N, Akiyama T, Ikeda K, Shijo T, Mitsuzawa S, Nagatomi R, Araki N, Yasui A, Warita H, <u>Hayashi YK</u> , Miyake K, Aoki M.	AMPK complex activation promotes sarcolemmal repair in dysferlinopathy.	Mol Ther	28 · 4	1133-1153	2020
Ueta Y, Akiba Y, Yamazaki J, Okubo Y, Taguchi T, Terashi H, <u>Hayashi YK</u> , Aizawa H.	Cerebral infarction and myalgia in a 75-year-old man with eosinophilic granulomatosis with polyangiitis.	Intern Med	59 · 23	3089-3092	2020
Hamakawa N, Kogetsu A, Isono M, Yamasaki C, Manabe S, Takeda T, Iwamoto K, Kubota T, Barrett J, Gray N, Turner A, Teare H, Imamura Y, Yamamoto BA, Kaye J, Hide M, <u>Takahashi MP</u> , Matsumura Y, Javaid MK, Kato K.	The practice of active patient involvement in rare disease research using ICT: experiences and lessons from the RUDY JAPAN project.	Res Involv Engagem	7 · 1	9	2021
Kubota T, Wu F, Vicart S, Nakaza M, Sternberg D, Watanabe D, Furuta M, Kokunai Y, Abe T, Kokubun N, Fontaine B, Cannon SC, <u>Takahashi MP</u> .	Hypokalaemic periodic paralysis with a charge-retaining substitution in the voltage sensor.	Brain Commun	2 · 2	Fcaa103	2020

Sasaki R, Nakaza M, Furuta M, Fujino H, Kubota T, <u>Takahashi MP</u> .	Mutation spectrum and health status in skeletal muscle channelopathies in Japan.	Neuromuscul Disord	30・7	546-553	2020
Nakaza M, Kitamura Y, Furuta M, Kubota T, Sasaki R, <u>Takahashi MP</u> .	Analysis of the genetic background associated with sporadic periodic paralysis in Japanese patients.	J Neurol Sci	412	116795	2020
Horie R, Kubota T, Koh J, Tanaka R, Nakamura Y, Sasaki R, Ito H, <u>Takahashi MP</u> .	EF hand-like motif mutations of Nav1.4 C-terminus cause myotonic syndrome by impairing fast inactivation.	Muscle Nerve	61・6	808-814	2020
Kurokawa M, Torio M, Ohkubo K, Tocan V, Ohyama N, Toda N, Ishii K, Nishiyama K, Mushimoto Y, Sakamoto R, Nakaza M, Horie R, Kubota T, <u>Takahashi MP</u> , Sakai Y, Nomura M, Ohga S.	The expanding phenotype of hypokalemic periodic paralysis in a Japanese family with p.Val876Glu mutation in CACNA1S.	Mol Genet Genomic Med	8・4	e1175	2020
Satoshi Nakada , Yuri Yamashita , Shuichi Machida , Yuko Miyagoe-Suzuki, <u>Eri Arikawa-Hirasawa</u> .	Perlecan Facilitates Neuronal Nitric Oxide Synthase Delocalization in Denervation-Induced Muscle Atrophy.	Cells	9・11	1094-1104	2020
<u>平澤 恵理</u> , 山下 由莉	難治性疾患(難病)を学ぶ シュワルツ・ヤンペル症候群	遺伝子医学	10・3	98-100	2020
吉村 祐輔, 石島 旨章, 金子 晴香, 長尾 雅史, 石橋 雅義, <u>平澤 恵理[有川]</u> , 町田 修一, 内藤 久士, 金子 和夫, 木南 英紀	青壮年のロコモと活動性の関連 Research kit アプリ「ロコモニタープラス」を用いた全国規模調査	日本整形外科学会雑誌	94・3	S525	2020

Abe K, Hirayama M, <u>Ohno K</u> , Shimamura T.	ENIGMA: an enterotype-like unigram mixture model for microbial association analysis.	BMC Genomics	20(Suppl 2)	191	2019
Huang K, Li J, Ito M, Takeda JI, Ohkawara B, Ogi T, Masuda A, <u>Ohno K</u> .	Gene Expression Profile at the Motor Endplate of the Neuromuscular Junction of Fast-Twitch Muscle.	Front Mol Neurosci	13	154	2020
Huang K, Masuda A, Chen G, Bushra S, Kamon M, Araki T, Kinoshita M, Ohkawara B, Ito M, <u>Ohno K</u> .	Inhibition of cyclooxygenase-1 by nonsteroidal anti-inflammatory drugs demethylates MeR2 enhancer and promotes Mbn1 transcription in myogenic cells.	Sci Rep	10 · 1	2558	2020
Kanbara S, Ohkawara B, Nakashima H, Ohta K, Koshimizu H, Inoue T, Tomita H, Ito M, Masuda A, Ishiguro N, Imagama S, <u>Ohno K</u> .	Zonisamide ameliorates progression of cervical spondylotic myelopathy in a rat model.	Sci Rep	10 · 1	13138	2020
Masuda A, Kawachi T, Takeda JI, Ohkawara B, Ito M, <u>Ohno K</u> .	tRIP-seq reveals repression of premature polyadenylation by co-transcriptional FUS-U1 snRNP assembly.	EMBO Rep	21 · 5	e49890	2020
Nakazawa Y, Hara Y, Oka Y, Komine O, van den Heuvel D, Guo C, Daigaku Y, Isono M, He Y, Shimada M, Kato K, Jia N, Hashimoto S, Kotani Y, Miyoshi Y, Tanaka M, Sobue A, Mitsutake N, Suganami T, Masuda A, <u>Ohno K</u> , Nakada S, Mashimo	Ubiquitination of DNA Damage-Stalled RNAPII Promotes Transcription-Coupled Repair.	Cell	180 · 6	1228-1244 e1224	2020

T, Yamanaka K, Luijsterburg MS, Ogi T.					
Nishiwaki H, Hamaguchi T, Ito M, Ishida T, Maeda T, Kashihara K, Tsuboi Y, Ueyama J, Shimamura T, Mori H, Kurokawa K, Katsuno M, Hirayama M, <u>Ohno K.</u>	Short-Chain Fatty Acid- Producing Gut Microbiota Is Decreased in Parkinson's Disease but Not in Rapid- Eye-Movement Sleep Behavior Disorder.	mSystems	5 • 6	e00797- 00720	2020
Nishiwaki H, Ito M, Ishida T, Hamaguchi T, Maeda T, Kashihara K, Tsuboi Y, Ueyama J, Shimamura T, Mori H, Kurokawa K, Katsuno M, Hirayama M, <u>Ohno K.</u>	Meta-Analysis of Gut Dysbiosis in Parkinson's Disease.	Mov Disord	35 • 9	1626- 1635	2020
Ohkawara B, Kobayakawa A, Kanbara S, Hattori T, Kubota S, Ito M, Masuda A, Takigawa M, Lyons KM, Ishiguro N, <u>Ohno K.</u>	CTGF/CCN2 facilitates LRP4-mediated formation of the embryonic neuromuscular junction.	EMBO Rep	21 • 8	e48462	2020
Ohkawara B, Shen X, Selcen D, Nazim M, Brill V, Tarnopolsky MA, Brady L, Fukami S, Amato AA, Yis U, <u>Ohno K</u> , Engel AG.	Congenital myasthenic syndrome-associated agrin variants affect clustering of acetylcholine receptors in a domain-specific manner.	JCI Insight	5 • 7	e13202 3	2020
Takeda J-i, Nanatsue K, Yamagishi R, Ito M, Haga N, Hirata H, Ogi T, <u>Ohno K.</u>	InMeRF: prediction of pathogenicity of missense variants by individual modeling for each amino acid substitution.	NAR Genomics and Bioinformatic s	2 • 2	lqaa038	2020
Takeuchi A, Takahashi Y, Iida K, Hosokawa M, Irie K, Ito M, Brown JB, <u>Ohno K</u> , Nakashima K, Hagiwara M.	Identification of Qk as a Glial Precursor Cell Marker that Governs the Fate Specification of Neural Stem Cells to a Glial Cell Lineage.	Stem Cell Reports	15 • 4	883-897	2020

Tawara N, Yamashita S, Takamatsu K, Yamasaki Y, Mukaino A, Nakane S, Farshadyeganeh P, <u>Ohno K</u> , Ando Y.	Efficacy of salbutamol monotherapy in slow-channel congenital myasthenic syndrome caused by a novel mutation in <i>CHRND</i> .	Muscle & Nerve			in press
<u>Sugie K</u> , Nishino I.	Lysosomal Membrane Disorders: LAMP-2 Deficiency.	Rosenberg's Molecular and Genetic Basis of Neurological and Psychiatric Disease (6th Edition, Elsevier)		567-574	2020
Eura N, Matsui TK, Luginbühl J, Matsubayashi M, Nanaura H, Shiota T, Kinugawa K, Iguchi N, Kiriya T, Zheng C, Kouno T, Lan YJ, Kongpracha P, Wiriyaermskul P, Sakaguchi YM, Nagata R, Komeda T, Morikawa N, Kitayoshi F, Jong M, Kobashigawa S, Nakanishi M, Hasegawa M, Saito Y, Shiromizu T, Nishimura Y, Kasai T, Takeda M, Kobayashi H, Inagaki Y, Tanaka Y, Makinodan M, Kishimoto T, Kuniyasu H, Nagamori S, Muotri AR, Shin JW, <u>Sugie K</u> , Mori E.	Brainstem Organoids From Human Pluripotent Stem Cells.	Front Neurosci	14	538	2020

Izumi T, Nanaura H, Iguchi N, Ozaki M, <u>Sugie K.</u>	Low Serum Eicosapentaenoic Acid Levels in Cryptogenic Stroke with Active Cancer.	J Stroke Cerebrovasc Dis	29 · 8	104892	2020
Kurashige T, Takahashi T, Nagano Y, <u>Sugie K.</u> , Maruyama H.	Krebs von den Lungen 6 decreased in the serum and muscle of GNE myopathy patients.	Neuropathology	41 · 1	29-36	2021
Ayaki T, Murata K, Kanazawa N, Uruha A, Ohmura K, <u>Sugie K.</u> , Kasagi S, Li F, Mori M, Nakajima R, Sasai T, Nishino I, Satoshi U, Makoto U, Fukumi F, Ito H, Takahashi R.	Myositis with sarcoplasmic inclusions in Nakajo-Nishimura syndrome: a genetic inflammatory myopathy.	Neuropathol Appl Neurobiol	46 · 6	579-587	2020
Kataoka H, Sawada Y, Shimozato N, Inatomi S, Yoshiji H, <u>Sugie K.</u>	Levodopa-responsive retrocollis on the background of choreic dyskinesia.	Int J Neurosci	130 · 5	461-463	2020
Kataoka H, <u>Sugie K.</u>	Serum adiponectin levels between patients with Parkinson's disease and those with PSP.	Neurol Sci	41 · 5	1125-1131	2020
<u>杉江和馬</u>	筋サルコイドーシス	Brain and Nerve	72 · 8	863-870	2020
<u>杉江和馬</u>	シュワルツ・ヤンペル症候群	新薬と臨床	70 · 2	201-204	2021
Nakahara K, Ikeda T, Takamatsu K, Tawara N, Hara K, Enokida Y, Tanoue N, Narita S, Fujii A, Yamanouchi Y, Morinaga J, <u>Yamashita S.</u>	A randomized phase 2 trial of antibiotic prophylaxis versus no intervention for muscle biopsy in department of neurology.	Acta Med. Okayama	74	261-264	2020

Shimazaki R, Uruha A, Kimura H, Nagaoka U, Kawazoe T, <u>Yamashita S</u> , Komori T, Miyamoto K, Matsubara S, Sugaya K, Nagao M, Isozaki E.	Rimmed vacuoles in myositis associated with anti-mitochondrial antibody.	J. Clin. Neurol	16	510-512	2020
Oyama M, Ohnuki Y, Inoue M, Uruha A, <u>Yamashita S</u> , Yutani S, Tanboon J, Nakahara J, Suzuki S, Shiina T, Nishino I, Suzuki S.	HLA-DRB1 allele and autoantibody profiles in Japanese patients with inclusion body myositis.	PLoS One	15	e02378 90	2020
Hara K, Nozaki K, Matsuo Y, Tawara N, <u>Yamashita S</u> .	Biological significance of target fibers in amyotrophic lateral sclerosis.	J. Neurol. Neurosurg. Psychiatry	91	1241- 1242	2020
Tawara N, <u>Yamashita S</u> , Nagatoshi C, Nakajima M, Ichimura Y, Okiyama N, Ando Y.	Anti-NXP2 antibody-positive dermatomyositis with aortic thrombus in normal aortic wall.	Rheumatology (Oxford)		In press	2021
Amato AA, Hanna MG, Machado PM, Badrising UA, Chinoy H, Benveniste O, Karanam AK, Wu M, Tankó LB, Schubert-Tennigkeit AA, Papanicolaou DA, Lloyd TE, Needham M, Liang C, Reardon KA, de Visser M, Ascherman DP, Barohn RJ, Dimachkie MM, Miller JAL, Kissel JT, Oskarsson B, Joyce NC, den Bergh PV, Baets J, De Bleecker JL, Karam C, David WS, Mirabella M, Nations SP, Jung HH, Pegoraro E,	Efficacy and safety of bimagrumab in sporadic inclusion body myositis: Long-term extension of RESILIENT.	Neurology		In press	2021

Maggi L, Rodolico C, Filosto M, Shaibani AI, Sivakumar K, Goyal NA, Mori-Yoshimura M, <u>Yamashita S</u> , Suzuki N, Aoki M, Katsuno M, Morihata H, Murata K, Nodera H, Nishino I, Romano CD, Williams VSL, Vissing J, Auberson LZ, the RESILIENT Study Extension Group.					
Tawara N, <u>Yamashita S</u> , Takamatsu K, Yamasaki Y, Mukaino A, Nakane S, Farshadyeganeh P, Ohno K, Ando Y.	Efficacy of salbutamol monotherapy in slow-channel congenital myasthenic syndrome caused by a novel mutation in CHRND.	Muscle Nerve	63・4	E30-E32	2021
山下賢	変性と炎症がクロストークする封入体筋炎の病態メカニズム	難病と在宅ケア	26・4	43-46	2020
山下賢	指定難病最前線 封入体筋炎	新薬と臨床	69	80-86	2020
山下賢	眼咽頭型筋ジストロフィーと患者レジストリの意義	難病と在宅ケア	26・9	26-29	2020
山下賢, 青木正志	運動ニューロン疾患	Clinical Neuroscience	39・1	91-94	2021
Taira K, <u>Mori-Yoshimura M</u> , Yamamoto T, Sajima K, Takizawa H, Shinmi J, Oya Y, Nito T, Nishino I, Takahashi Y.	More prominent fibrosis of the cricopharyngeal muscle in inclusion body myositis.	J Neurol Sci			2021 Jan Online ahead of print.

Taira K, Yamamoto T, <u>Mori-Yoshimira M</u> , Sajima K, Takizawa H, Shinmi J, Oya Y, Nishino I, Takahashi Y.	Cricopharyngeal bar on videofluoroscopy: high specificity for inclusion body myositis.	J Neurol			2020 Sep Online ahead of print.
Ayaki T, <u>Murata K</u> , et.al.	Myositis with sarcoplasmic inclusions in Nakajo-Nishimura syndrome: a genetic inflammatory myopathy.	Neuropathol Appl Neurobiol	46•6	579-587	2020
Koh J, Kaneoke Y, Donishi T, Ishida T, Sakata M, Hiwatani Y, Nakayama Y, Yasui M, Ishiguchi H, Hironishi M, <u>Murata KY</u> ,et.al.	Increased large-scale inter-network connectivity in relation to impulsivity in Parkinson's disease.	Sci Rep	10•1	11418. doi: 10.1038/s41598-020-68266-x.PMID : 32651411	2020
森めぐみ、金澤伸雄、 <u>村田顕也</u> 、伊東秀文	筋炎と遺伝性筋疾患の間 -中條-西村症候群-	神経治療	37	162-165	2020