Matsui N., Takahara M., Yamazaki H., Takamatsu N., Osaki Y., Kaji R., Nishino I., Yamashita S., Izumi Y.	Case of anti-NT5c1A antibody-seropositive inclusion body myositis associated with severe dysphagia and prominent forearm weakness.	Neurol Clin Neurosci.	11	46-48	2023
Yamasaki Y., Mukaino A., Yamashita S., Takeuchi Y., Tawara N., Yoshida R., Honda Y., Yamashita T., Kakimoto A., Ueyama H., Ando Y.	Macroglossia in rapidly progressive inclusion body myositis.	Neuropathology		In press	2023
Yamashita S., Nagatoshi A., Takeuchi Y., Nishino I., Ueda M.	Myopathic changes caused by protein aggregates in adult-onset spinal muscular atrophy.	Neuropathology		In press	2023
山下賢	筋炎・ミオパチー　封入体筋炎.	下畑享良，編. 脳神経内科診断ハンドブック. 東京: 中外医学社		481-488	2022
山下賢	免疫性筋疾患　封入体筋炎.	日本臨床増刊号 免疫性神経疾患（第 2 版）-基礎・臨床の最新知見-. 東京: 日本臨床社		461-471	2022
山下賢, 村井弘之	ここまでわかった眼咽頭型筋ジストロフィーの病態と治療戦略	医学のあゆみ	283	939-943	2022
山下賢, 村井弘之	ALS に対する新規治療	難病と在宅ケア	28	41-44	2023
山下賢	封入体筋炎における自己抗体－抗 NT5C1A 抗体（cN1A）抗体の臨床的意義	リウマチ科	69	224-230	2023
Matsui N., Takahara M., Yamazaki H., Takamatsu N., Osaki Y., Kaji R., Nishino I., Yamashita S., Izumi Y.	Case of anti-NT5c1A antibody-seropositive inclusion body myositis associated with severe dysphagia and prominent forearm weakness.	Neurol Clin Neurosci.	11	46-48	2023
Mano T, Soyama S, Sugie K.	Improvement in Tongue Pressure Precedes Improvement in Dysphagia in Dermatomyositis	Clin Pract	12 (5)	797-802	2022
Shiota T, Eura N, Hasegawa A, Kiriyama T, Sugie K.	Pathological features of inflammatory myopathy as a manifestation of chronic graft-versus-host disease after allogeneic bone marrow transplantation	Neuropathology	42(4)	309-314	2022
Shiota T, Nagata R, Kikuchi S, Nanaura H, Matsubayashi M, Nakanishi M, Kobashigawa S, Isozumi N, Kiriyama T, Nagayama K, Sugie K, Yamashiro Y, Mori E.	C9orf72-Derived Proline:Arginine Poly-Dipeptides Modulate Cytoskeleton and Mechanical Stress Response	Front Cell Dev Biol	10	750829	2022
杉江和馬	筋病理から見た筋炎	皮膚科	3(1)	40-48	2023

Saito Y, Baba S, Komaki H, Nishino I.	A 7-year-old female with hypotonia and scoliosis.	Brain Pathol	32(6)	e13076	2022
Nozomi Hayashiji, Genri Kawahara, Xing Xu, Tomohiko Fukuda, Aurelien Kerever, Jianguo Gu, Yukiko K. Hayashi, Eri Arikawa-Hirasawa	α-1,6-Fucosyltransferase Is Essential for Myogenesis in Zebrafish	Cells	12(1)	144	2022
Satoshi Nakada, Yuri Yamashita, Seiya Akiba, Takeru Shima, Eri Arikawa-Hirasawa	Myocyte Culture with Decellularized Skeletal Muscle Sheet with Observable Interaction with the Extracellular Matrix	Bioengineering	9(7)	309	2022
杉江和馬	自己貪食空胞性ミオパチー:ダノン病とその類縁疾患	医学のあゆみ	283(10)	951-957	2022
杉江和馬	ステロイド･IVIgで筋力回復が不良な皮膚筋炎･壊死性筋症　難治性の皮膚筋炎・壊死性筋症	Brain and Nerve	74(5)	537-544	2022
Ishigakii K, Ikeda R, Suzuki J, Hirano-Kawamoto A, Ohta J, Kato K, Izumi R, Suzuki N, Aoki M, Kawase T, Katori Y.	Patulous Eustachian Tube Patients With Oculopharyngeal Muscular Dystrophy.	Otol Neurotol.	43	E442-445	2022
Li Y, Chen W, Ogawa K, Koide M, Takahashi T, Hagiwara Y, Itoi E, Aizawa T, Tsuchiya M, Izumi R, Suzuki N, Aoki M, Kanzaki M.	Feeder-supported in vitro exercise model using human satellite cells from patients with sporadic inclusion body myositis.	Sci Rep.	12	1082	2022
Okada Y, Izumi R, Hosaka T, Watanabe S, Shijo T, Hatchome N, Konishi R, Ichimura Y, Okiyama N, Suzuki N, Misu T, Aoki M.	Anti-NXP2 antibody-positive dermatomyositis developed after COVID-19 manifesting as type I interferonopathy.	Rheumatology (Oxford).		keab872.	2021
Konomatsu K, Izumi R, Suzuki N, Takai Y, Shirota Y, Saito R, Kuroda H, Aoki M.	A rare case of sporadic inclusion body myositis and rheumatoid arthritis exhibiting ectopic lymphoid follicle-like structures: a case report and literature review.	Neuromuscul Disord.	31	870-876	2021
Inoue-Shibui A, Niihori T, Kobayashi M, Suzuki N, Izumi R, Warita H, Hara K, Shirota M, Funayama R, Nakayama K, Nishino I, Aoki M, Aoki Y.	A novel deletion in the C-terminal region of HSPB8 in a family with rimmed vacuolar myopathy.	J Hum Genet.	66	965-972	2021
Yoshioka W, Shimizu R, Takahashi Y, Oda Y, Yoshida S, Ishihara N, Nishino I, Nakamura H, Mori-Yoshimura M	Extra-muscular manifestations in GNE myopathy patients: A nationwide repository questionnaire survey in Japan.	Clin Neurol Neurosurg	212	107057	2022
Kumutpongpanich T, Ogasawara M, Ozaki A, Ishiura H, Tsuji S, Minami N, Hayashi S, Noguchi S, Iida A, Nishino I; OPDM_LRP12 Study Group	Clinicopathologic Features of Oculopharyngodistal Myopathy With LRP12 CGG Repeat Expansions Compared With Other Oculopharyngodistal Myopathy Subtypes.	JAMA Neurol	78・7	853-863	2021
Inoue M, Saito Y, Yonekawa T, Ogawa M, Iida A, Nishino I, Noguchi S.	Causative variant profile of collagen VI-related dystrophy in Japan.	Orphanet J Rare Dis.	16・1	284	2021

Lopes Abath Neto O, Medne L, Donkervoort S, Rodríguez-García ME, Bolduc V, Hu Y, Guadagnin E, Foley AR, Brandsema JF, Glanzman AM, Tennekoon GI, Santi M, Berger JH, Megeney LA, Komaki H, Inoue M, Cotrina-Vinagre FJ, Hernández-Lain A, Martin-Hernández E, Williams L, Borell S, Schorling D, Lin K, Kolokotronis K, Lichter-Konecki U, Kirschner J, Nishino I, Banwell B, Martínez-Azorín F, Burgon PG, Bönnemann CG.	MLIP causes recessive myopathy with rhabdomyolysis, myalgia and baseline elevated serum creatine kinase.	Brain.	22;144(9)	2722-2731	2021
Awano H, Saito Y, Shimizu M, Sekiguchi K, Niijima S, Matsuo M, Maegaki Y, Izumi I, Kikuchi C, Ishibashi M, Okazaki T, Komaki H, Iijima K, Nishino I.	FKRP mutations cause congenital muscular dystrophy 1C and limb-girdle muscular dystrophy 2I in Asian patients.	J Clin Neurosci.	92	215-221	2021
Ben Yaou R, Yun P, Dabaj I, Norato G, Donkervoort S, Xiong H, Nascimento A, Maggi L, Sarkozy A, Monges S, Bertoli M, Komaki H, Mayer M, Mercuri E, Zanoteli E, Castiglioni C, Marini-Bettolo C, D'Amico A, Deconinck N, Desguerre I, Erazo-Torricelli R, Gurgel-Giannetti J, Ishiyama A, Kleinsteuber KS, Lagrue E, Laugel V, Mercier S, Messina S, Politano L, Ryan MM, Sabouraud P, Schara U, Siciliano G, Vercelli L, Voit T, Yoon G, Alvarez R, Muntoni F, Pierson TM, Gómez-Andrés D, Reghan Foley A, Quijano-Roy S, Bönnemann CG, Bonne G.	International retrospective natural history study of LMNArelated congenital muscular dystrophy.	Brain Commun.	3(3)	fcab075	2021
Tawara N, Yamashita S, 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
Inoue T, Ohkawara B, Bushra S, Kanbara S, Nakashima H, Koshimizu H, Tomita H, Ito M, Masuda A, Ishiguro N, Imagama S, Ohno K.	Zonisamide upregulates neuregulin-1 expression and enhances acetylcholine receptor clustering at the in vitro neuromuscular junction	Neuropharmacology	195	108637	2021
Takeda J*, Fukami S*, Tamura A, Shibata A, Ohno K. *Equal contributions.	Intsplice2: Prediction of the splicing effects of intronic single-nucleotide variants using lightGBM modeling	Front Genet	12	701076	2021
Kawachi T, Masuda A, Yamashita Y, Takeda J, Ohkawara B, Ito M, Ohno K.	Regulated splicing of large exons is linked to phase-separation of vertebrate transcription factors	EMBO J	40(22)	e107485	2021
Takemoto G, Matsushita M, Okamoto T, Ito T, Matsuura Y, Takashima C, Chen-Yoshikawa TF, Ebi H, Imagama S, Kitoh H, Ohno K, Hosono Y.	Meclozine attenuates the mark pathway in mammalian chondrocytes and ameliorates fgf2-induced bone hyperossification in larval zebrafish	Front Cell Dev Biol	9	694018	2021
Kawamura Y, Hida T, Ohkawara B, Matsushita M, Kobayashi T, Ishizuka S, Hiraiwa H, Tanaka S, Tsushima M, Nakashima H, Ito K, Imagama S, Ito M, Masuda A,	Meclozine ameliorates skeletal muscle pathology and increases muscle forces in mdx mice	Biochem Biophys Res Commun	592	87-92	2022

Ishiguro N, Ohno K.					
Noto K, Ohkawara B, Ishii H, Ito M, Masuda A, Hirata H, Ohno K.	Screening of signaling pathways for agrin-induced AChR clustering in C2C12 myotubes	Genes Cells		in press.	2022
Sakaguchi T, Miyamoto K, Ohkawara B, Kisimoto Y, Ishizuka S, Hiraiwa H, Imagama S, Ishiguro N, Ohno K.	Promethazine downregulates Wnt/β-catenin signaling and increases biomechanical forces of injured Achilles tendon in early stage of healing	Am J Sport Med		in press.	2022
Sugie K.	Autophagy dysfunction in skeletal myopathies: Inclusion body myositis and Danon disease.	Autophagy Dysfunction in Alzheimer's Disease and Dementia (Elsevier)	In press		2022
Sugie K.	Autophagic vacuolar myopathy: Danon disease and related myopathies.	Neurol Clin Neurosci	Online ahead of print		2022
Sugie K.	Editorial commentary: Highlighting the ray of hope in Danon disease research after 40 years.	Trends Cardiovasc Med	Online ahead of print		2021
Nanaura H, Kawamukai H, Fujiwara A, Uehara T, Aiba Y, Nakanishi M, Shiota T, Hibino M, Wiriyasermkul P, Kikuchi S, Nagata R, Matsubayashi M, Shinkai Y, Niwa T, Mannen T, Morikawa N, Iguchi N, Kiriyama T, Morishima K, Inoue R, Sugiyama M, Oda T, Kodera N, Toma-Fukai S, Sato M, Taguchi H, Nagamori S, Shoji O, Ishimori K, Matsumura H, Sugie K, Saio T, Yoshizawa T, Mori E.	C9orf72-derived arginine-rich poly-dipeptides impede phase modifiers	Nat Commun	12(1)	5301	2021
Yasui T, Nagaoka U, Oya Y, Uruha A, Karashima J, Funai A, Miyamoto K, Matsubara S, Sugaya K, Takahashi K, Inoue M, Okubo M, Sugie K, Nishino I.	Mild form of Danon dsease: two case reports.	Neuromuscul Disord	31(11)	1207-1211	2021
Iwasa N, Matsui TK, Iguchi N, Kinugawa K, Morikawa N, Sakaguchi YM, Shiota T, Kobashigawa S, Nakanishi M, Matsubayashi M, Nagata R, Kikuchi S, Tanaka T, Eura N, Kiriyama T, Izumi T, Saito K, Kataoka H, Saito Y, Kimura W, Wanaka A, Nishimura Y, Mori E, Sugie K.	Gene Expression Profiles of Human Cerebral Organoids Identify PPAR Pathway and PKM2 as Key Markers for Oxygen-Glucose Deprivation and Reoxygenation	Front Cell Neurosci	15	605030	2021
杉江和馬	先天性ミオパチー	脳神経内科学レビュー（総合医学社）		309-314	2022

杉江和馬	希少性筋疾患	脳神経内科診断ハンドブック（中外医学社）		501-509	2021
杉江和馬	筋疾患	臨床遺伝学成人領域（診断と治療社）		29-42	2021
杉江和馬	進化する筋疾患診療の最前線	BIO Clinica	36(9)	826-827	2021
杉江和馬	オートファジー関連筋疾患の新展開：ポンペ病とダノン病	BIO Clinica	36(9)	848-852	2021
杉江和馬	COVID-19 と筋障害	脳神経内科	95(3)	342-350	2021
杉江和馬	自己貪食空胞性ミオパチー	新薬と臨床	70（6）	681-687	2021
Taira K, Mori-Yoshimura M.	Regarding Cricopharyngeal Myotomy in IBM: Comparison of Endoscopic and Transcervical Approaches.	Laryngoscope.	131(6)	E1998	2021
Taira K, Mori-Yoshimura M, 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	422	117327	2021
Taira K, Yamamoto T, Mori-Yoshimura M, Sajima K, Takizawa H, Shinmi J, Oya Y, Nishino I, Takahashi Y.	Cricopharyngeal bar on videofluoroscopy: high specificity for inclusion body myositis.	J Neurol.	268(3)	1016-1024	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, Maggi L, Rodolico C, Filosto M, Shaibani AI, Sivakumar K, Goyal NA, Mori-Yoshimura M, Yamashita S, 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.	Efficacy and safety of bimagrumab in sporadic inclusion body myositis: Long-term extension of RESILIENT.	Neurology	96	e1595-e1607	2021
Tawara N, Yamashita S, 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	E30-E32	2021
Yamashita S.	Recent Progress in Oculopharyngeal Muscular Dystrophy.	J Clin Med	10	1375	2021

Tawara N, Yamashita S, Nagatoshi C, Nakajima M, Ichimura Y, Okiyama N, Ando Y.	Anti-NXP2 antibody-positive dermatomyositis with aortic thrombus in normal aortic wall.	Rheumatology (Oxford)	60	e159-e161	2021
Kumai Y, Miyamoto T, Matsubara K, Satoh C, Yamashita S, Orita Y.	Swallowing dysfunction in myasthenia gravis patients examined with high-resolution manometry.	Auris Nasus Larynx	48	1135-1139	2021
Tawara N, Nakane S, Kudo N, Kosaka T, Takamatsu K, Wada K, Kobayashi A, Yamashita S, Funagura N, Inoue T, Ando Y.	Binasal hemianopia caused by bilateral optic perineuritis due to sarcoidosis.	eNeurologicalSci	24	100354	2021
山下賢, 青木正志	運動ニューロン疾患.	Clinical Neuroscience	39	91-94	2021
山下賢.	遺伝子変異から見た多系統蛋白質症.	脳神経内科	95	104-111	2021
山下賢.	多系統蛋白質症における前頭側頭型認知症.	Dementia Japan	35	295-303	2021
山下賢.	筋炎・ミオパチー　封入体筋炎.	脳神経内科診断ハンドブック. 下畑享良,編		481-488	2022
Kubota T, Hama M, Sugiura Y, Takahashi Y, Ishikawa K, Mizusawa H, Takahashi MP.	A nationwide survey of episodic ataxia in Japan.	Neurology and Clinical Neuroscience.	9	443‐451	2021
Kubota T, Nabatame S, Sato R, Hama M, Nishiike U, Mochizuki H, Takahashi MP, Takeshima T.	Hemiplegic migraine type 2 caused by a novel variant within the P-type ATPase motif in ATP1A2 concomitant with a CACNA1A variant.	Brain Dev.	43・9	952-957	2021
Kumutpongpanich T, Ogasawara M, Ozaki A, Ishiura H, Tsuji S, Minami N, Hayashi S, Noguchi S, Iida A, Nishino I; OPDM_LRP12 Study Group, Mori-Yoshimura M, Oya Y, Ono K, Shimizu T, Kawata A, Shimohama S, Toyooka K, Endo K, Toru S, Sasaki O, Isahaya K, Takahashi MP, Iwasa K, Kira JI, Yamamoto T, Kawamoto M, Hamano T, Sugie K, Eura N, Shiota T, Koide M, Sekiya K, Kishi H, Hideyama T, Kawai S, Yanagimoto S, Sato H, Arahata H, Murayama S, Saito K, Hara H, Kanda T, Yaguchi H, Imai N, Kawagashira Y, Sanada M, Obara K, Kaido M, Furuta M, Kurashige T, Hara W, Kuzume D, Yamamoto M, Tsugawa J, Kishida H, Ishizuka N, Morimoto K, Tsuji Y, Tsuneyama A, Matsuno A, Sasaki R, Tamakoshi D, Abe E, Yamada S, Uzawa A.	Clinicopathologic Features of Oculopharyngodistal Myopathy With LRP12 CGG Repeat Expansions Compared With Other Oculopharyngodistal Myopathy Subtypes.	JAMA Neurol.	78・7	853-863	2021

久保田智哉、高橋正紀	筋チャネル病の新たな病態	生体の科学	72・6	555-559	2021
高橋正紀	周期性四肢麻痺	福井次矢、高木誠、小室一成総編集「今日の治療指針　2022 年版―私はこう治療している」		1023	2022
高橋正紀、青木正志	筋チャネル病､遠位型ミオパチー	下畑享良編「脳神経内科診断ハンドブック」		489-498	2021
Inoue-Shibui A, Niihori T, Kobayashi M, Suzuki N, Izumi R, Warita H, Hara K, Shirota M, Funayama R, Nakayama K, Nishino I, Aoki M, 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/s 10038-021-00916-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, Aoki M, 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, Aoki M, 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, Aoki M, 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, Aoki M, 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, Aoki M, 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, Aoki M.	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, Aoki M.	AMPK Complex Activation Promotes Sarcolemmal Repair in Dysferlinopathy.	Mol Ther	28	1133-1153	2020

Samukawa M, Nakamura N, Hirano M, Morikawa M, Sakata H, Nishino I, 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, Nishino I, 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, Kumutpongpanich, 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, Nishino I.	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, Maeda, M. H, Sato, T, Ura, S, Oya, Y, Ogasawara, M, Iida, A, Nishino, I, Zhou, C, Yan, C, Yuan, Y, Hong, D, Wang, Z.	Expansion of GGC Repeat in GIPC1 Is Associated with Oculopharyngodistal Myopathy.	Am J Hum Genet	106・1	793-804	2020
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, Hayashi YK, 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, Hayashi YK, 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, Takahashi MP, 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, Takahashi MP.	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, Takahashi MP.	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, Takahashi MP.	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, Takahashi MP.	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, Takahashi MP, 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, Eri Arikawa-Hirasawa.	Perlecan Facilitates Neuronal Nitric Oxide Synthase Delocalization in Denervation-Induced Muscle Atrophy.	Cells	9・11	1094-1104	2020
平澤 恵理, 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, 平澤 恵理[有川], 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, Ohno K, 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, Ohno K.	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, Ohno K.	Inhibition of cyclooxygenase-1 by nonsteroidal anti-inflammatory drugs demethylates MeR2 enhancer and promotes Mbnl1 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, Ohno K.	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, Ohno K.	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, Ohno K, Nakada S, Mashimo T, Yamanaka K, Luijsterburg MS, Ogi T.	Ubiquitination of DNA Damage-Stalled RNAPII Promotes Transcription-Coupled Repair.	Cell	180・6	1228-1244 e1224	2020
Nishiwaki H, Hamaguchi T, Ito M, Ishida T, Maeda T, Kashihara K, Tsuboi Y, Ueyama J, Shimamura T, Mori H, Kurokawa K, Katsuno M,	Short-Chain Fatty Acid-Producing Gut Microbiota Is Decreased in Parkinson's Disease but Not in Rapid-Eye-Movement Sleep	mSystems	5・6	e00797-00720	2020

Hirayama M, Ohno K.	Behavior Disorder.				
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, Ohno K.	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, Ohno K.	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, Bril V, Tarnopolsky MA, Brady L, Fukami S, Amato AA, Yis U, Ohno K, Engel AG.	Congenital myasthenic syndrome-associated agrin variants affect clustering of acetylcholine receptors in a domain-specific manner.	JCI Insight	5・7	e132023	2020
Takeda J-i, Nanatsue K, Yamagishi R, Ito M, Haga N, Hirata H, Ogi T, Ohno K.	InMeRF: prediction of pathogenicity of missense variants by individual modeling for each amino acid substitution.	NAR Genomics and Bioinformatics	2・2	lqaa038	2020
Takeuchi A, Takahashi Y, Iida K, Hosokawa M, Irie K, Ito M, Brown JB, Ohno K, 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, Ohno K, Ando Y.	Efficacy of salbutamol monotherapy in slow-channel congenital myasthenic syndrome caused by a novel mutation in CHRND.	Muscle & Nerve			in press
Sugie K, 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, Kiriyama T, Zheng C, Kouno T, Lan YJ, Kongpracha P, Wiriyasermkul 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, Sugie K, Mori E.	Brainstem Organoids From Human Pluripotent Stem Cells.	Front Neurosci	14	538	2020
Izumi T, Nanaura H, Iguchi N, Ozaki M, Sugie K.	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, Sugie K, 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, Sugie K, 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, Sugie K.	Levodopa-responsive retrocollis on the background of choreic dyskinesia.	Int J Neurosci	130・5	461-463	2020
Kataoka H, Sugie K.	Serum adiponectin levels between patients with Parkinson's disease and those with PSP.	Neurol Sci	41・5	1125-1131	2020
杉江和馬	筋サルコイドーシス	Brain and Nerve	72・8	863-870	2020
杉江和馬	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, Yamashita S.	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, Yamashita S, 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, Yamashita S, 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	e0237890	2020
Hara K, Nozaki K, Matsuo Y, Tawara N, Yamashita S.	Biological significance of target fibers in amyotrophic lateral sclerosis.	J. Neurol. Neurosurg. Psychiatry	91	1241-1242	2020
Tawara N, Yamashita S, 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, Maggi L, Rodolico C, Filosto M, Shaibani AI, Sivakumar K, Goyal NA, Mori-Yoshimura M, Yamashita S, Suzuki N, Aoki M, Katsuno M, Morihata H, Murata K, Nodera H,	Efficacy and safety of bimagrumab in sporadic inclusion body myositis: Long-term extension of RESILIENT.	Neurology		In press	2021

Nishino I, Romano CD, Williams VSL, Vissing J, Auberson LZ, the RESILIENT Study Extension Group.					
Tawara N, Yamashita S, 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, Mori-Yoshimura M, 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, Mori-Yoshimira M, 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, Murata K, 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, Murata KY,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、村田顕也、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