

### 研究成果の刊行に関する一覧表

発表者名	論文タイトル	発表誌	巻・号	ページ	出版年
Nishino I, Crrillo-Carraasco N, Argov Z	GNE myopathy: current update and future therapy.	J Neurol Neurosurg Psychiatry.			2014
Yonekawa T, Nishino I	Ullrich congenital muscular dystrophy: clinicopathological features, natural history and pathomechanism(s)	Neurol Neurosurg Psychiatry.			2014
Cho A, Hayashi YK, Monma K, Oya Y, Noguchi S, Nonaka I, Nishino I	Mutation profile of the <i>GNE</i> gene in Japanese patients with distal myopathy with rimmed vacuoles ( <i>GNE</i> myopathy).	J Neurol Neurosurg Psychiatry.	85(8)	912-915	2014
Goto M, Okada M, Komaki H, Sugai K, Sasaki M, Noguchi S, Nonaka I, Nishino I, Hayashi YK	A nationwide survey on Marinesco-Sjögren syndrome in Japan.	Orphanet J Rare Diseases	9・1	58	2014
Kajino S, Ishihara K, Goto K, Ishigaki K, Noguchi S, Nonaka I, Osawa M, Nishino I, Hayashi YK	Congenital fiber type disproportion myopathy caused by LMNA mutations.	J Neurol Sci	340・ 1-2	94-98	2014

発表者名	論文タイトル	発表誌	巻・号	ページ	出版年
Miyatake S, Koshimizu E, Hayashi YK, Miya K, Shiina M, Nakashima M, Tsurusaki Y, Miyake N, Saito H, Ogata K, Nishino I, Matsumoto N.	Deep sequencing detects very-low-grade somatic mosaicism in the unaffected mother of siblings with nemaline myopathy.	Neuromuscu l Disord	24・7	642-647	2014
Uruha A, Hayashi YK, Oya Y, Mori-Yoshimur a M, Kanai M, Murata M, Kawamura M, Ogata K, Matsumura T, Suzuki S, Takahashi Y, Kondo T, Kawarabayashi T, Ishii Y, Kokubun N, Yokoi S, Yasuda R, Kira JI, Mitsuhashi S, Noguchi S, Nonaka I, Nishino I.	Necklace cytoplasmic bodies in hereditary myopathy with early respiratory failure.	J Neurol Neurosurg Psychiatry	Epub		2014

発表者名	論文タイトル	発表誌	巻・号	ページ	出版年
Mori-Yoshimura M, Oya Y, Yajima H, Yonemoto N, Kobayashi Y, Hayashi YK, Noguchi S, Nishino I, Murata M.	GNE myopathy: A prospective natural history study of disease progression.	Neuromuscul Disord	24・5	380-386	2014
Mori-Yoshimura M, Hayashi YK, Yonemoto N, Nakamura H, Murata M, Takeda SI, Nishino I, Kimura E.	Nationwide patient registry for GNE myopathy in Japan.	Orphanet J Rare Dis.	9・1	150	2014
Yuen M, Sandaradura SA, Dowling JJ, YK Hayashi, et al.	Leiomodin-3 dysfunction results in thin filament disorganization and nemaline myopathy.	J Clin Invest	125・1	456-457	2015
Matsuzaka Y, Kishi S, Aoki Y, Komaki H, Oya Y, Takeda S, Hashido K.	Three novel serum biomarkers, miR-1, miR-133a, and miR-206 for Limb-girdle muscular dystrophy, Facioscapulohumeral muscular dystrophy, and Becker muscular dystrophy.	Environ Health Prev Med.	19	452-8	2014

発表者名	論文タイトル	発表誌	巻・号	ページ	出版年
Goto M, Okada M, Komaki H, Sugai K, Sasaki M, Noguchi S, Nonaka I, Nishino I, Hayashi YK.	A nationwide survey on Marinesco-Sjögren syndrome in Japan.	Orphanet J Rare Dis.	23(9)	58	2014
Yoshinaga H et al.	Phenotypic variability in childhood of skeletal muscle sodium channelopathies	Pediatric Neurology	印刷中		
久保田智哉 高橋正紀	骨格筋チャネル病の最新知見 ミオトニー症候群と周期性四肢麻痺を中心に	別冊 医学のあゆみ イオンチャネル病のすべて	pp. 38-45		2014
高橋正紀	周期性四肢麻痺	今日の整形外科治療指針 第7版	印刷中		
Furuya N, Ikeda SI, Sato S, Soma S, Ezaki J, Trejo JA, Takeda-Ezaki M, Fujimura T, <u>Arikawa-Hirasa wa E</u> , Tada N, Komatsu M, Tanaka K, Kominami E, Hattori N, Ueno T.	PARK2/Parkin-mediated mitochondrial clearance contributes to proteasome activation during slow-twitch muscle atrophy via NFE2L1 nuclear translocation.	Autophagy	Apr;10 (4)	631-41	2014

発表者名	論文タイトル	発表誌	巻・号	ページ	出版年
de Vega S, Suzuki N, Nonaka R, Sasaki T, Forcinito P, <u>Arikawa-Hirasa</u> <u>wa E</u> , Yamada Y.	A C-terminal fragment of fibulin-7 interacts with endothelial cells and inhibits their tube formation in culture.	Arch Biochem Biophys.	Mar 1;545:	148-53.	2014
Ning L, Kurihara H, de Vega S,* Ichikawa-Tomik awa n, Xu Z,Nonaka R, Kazuno S, Yamada Y, Miner JH, <u>Arikawa-Hirasa</u> <u>wa E</u>	Laminin $\alpha 1$ regulates age-related mesangial cell proliferation and mesangial matrix accumulation through the TGF $\beta$ pathway	The American Journal of Pathology	Jun:18 4(6)	1683-94	2014
Nonaka R, Iesaki T, de Vega S, Daida H, Okada T, Sasaki T, and <u>Arikawa-Hirasa</u> <u>wa E</u>	Perlecan deficiency causes endothelial dysfunction by reducing the expression of endothelial nitric oxide synthase.	Physiological Reports	Jan 27:3(1)	In press	2014

発表者名	論文タイトル	発表誌	巻・号	ページ	出版年
Kerever A, Kamagata K, Yokosawa S, Yosuke Otake Y, Ochi H Hori M, Nishikori A, Aoki S, <u>Arikawa-Hirasa wa E.</u>	High-Resolution MRI and Three-Dimensional Imaging of Cleared Mouse Brain: A Preliminary Microstructural Study in a Mouse with callosal agenesis	Magnetic Resonance in Medical Sciences in press	in press		2014
Inaguma Y, Hamada N, Tabata H, Iwamoto I, Mizuno M, Nishimura YV, Ito H, Morishita R, Suzuki M, <u>Ohno K,</u> Kumagai T, Nagata KI.	SIL1, a causative cochaperone gene of Marinesco-Sjogren syndrome, plays an essential role in establishing the architecture of the developing cerebral cortex	<i>EMBO Mol Med</i>	6	155 - 295	2014
Ohkawara B, Cabrera-Serran o M, Nakata T, Milone M, Asai N, Ito K, Ito M, Masuda A, Ito Y, Engel AG, <u>Ohno K.</u>	LRP4 third beta-propeller domain mutations cause novel congenital myasthenia by compromising agrin-mediated MuSK signaling in a position-specific manner	<i>Hum Mol Genet</i>	23	1856-1868	2014

発表者名	論文タイトル	発表誌	巻・号	ページ	出版年
Nakayama T, Nakamura H, Oya Y, Kimura T, Imahuku I, <u>Ohno K</u> , Nishino I, Abe K, Matsuura T.	Clinical and genetic analysis of the first known Asian family with myotonic dystrophy type 2	<i>J Hum Genet</i>	59	129-133	2014
Kokunai Y*, Nakata T*, Furuta M*, Sakata S, Kimura H, Aiba T, Yoshinaga M, Osaki Y, Nakamori M, Itoh H, Sato T, Kubota T, Kadota K, Shindo K, Mochizuki H, Shimizu W, Horie M, Okamura Y, <u>Ohno K</u> , Takahashi M. *Equal contribution.	A Kir3.4 mutation causes Andersen-Tawil syndrome by an inhibitory effect on Kir2.1	<i>Neurology</i>	82	1058-1064	2014

発表者名	論文タイトル	発表誌	巻・号	ページ	出版年
Kobayashi M, Ohno T, Ihara K, Murai A, Kumazawa M, Hoshino H, Iwanaga K, Iwai H, Hamana Y, Ito M, <u>Ohno K</u> , Horio F.	Searching for genomic region of high-fat diet-induced type 2 diabetes in mouse chromosome 2 by analysis of congenic strains	<i>PLoS ONE</i>	9	e96271	2014
Yamashita Y*, Matsuura T*, Kurosaki T, Amakusa Y, Kinoshita M, Ibi T, Sahashi K, <u>Ohno K</u> . *Equal contribution.	LDB3 splicing abnormalities are specific to skeletal muscles of patients with myotonic dystrophy type 1 and alter its PKC binding affinity	<i>Neurobiol Dis</i>	69	200-205	2014
Nasrin F, Rahman MA, Masuda A, Ohe K, Takeda J, <u>Ohno K</u> .	HnRNP C, YB-1 and hnRNP L coordinately enhance skipping of human MUSK exon 10 to generate a Wnt-insensitive MuSK isoform	<i>Sci Rep</i>	4	6841	2014



発表者名	論文タイトル	発表誌	巻・号	ページ	出版年
Azuma Y, Nakata T, Tanaka M, Shen XM, Ito M, Iwata S, Okuno T, Nomura Y, Ando N, Ishigaki K, Ohkawara B, Masuda A, Natsume J, Kojima S, Sokabe M, <u>Ohno K.</u>	Congenital myasthenic syndrome in Japan: Ethnically unique mutations in muscle nicotinic acetylcholine receptor subunits	<i>Neuromuscu lar Disorders</i>	25	60-69	2015
<u>Ohno K.</u> , Ohkawara B, Ito M, Engel AG.	Molecular Genetics of Congenital Myasthenic Syndromes	<i>eLS. John Wiley &amp; Sons, Inc., Chichester (査読有)</i>		<a href="http://www.els.net">http://www .els.net</a> [doi: 10.1002/97 804700159 02.a00243 14].	2014
<u>Ohno K.</u> , Ito M, Kawakami Y, Ohtsuka K.	Collagen Q is a key player for developing rational therapy for congenital myasthenia and for dissecting the mechanisms of anti-MuSK myasthenia gravis	<i>J Mol Neurosci</i> (査 読有)	53	359-361	2014
Rahman MA, Nasrin F, Masuda A, <u>Ohno K.</u>	Decoding abnormal splicing code in human diseases	<i>J Invest Genomics</i> (査 読有)	2(1)	00016	2015

発表者名	論文タイトル	発表誌	巻・号	ページ	出版年
Sugie K, Nishino I.	Lysosomal Membrane Disorders: LAMP-2 Deficiency.	Rosenberg's Molecular and Genetic Basis of Neurological and Psychiatric Disease (5th Edition, Elsevier)		411-417	2014
Eura N, Sugie K, Kiriyaama T, Ueno S.	Characteristic dysphagia as a manifestation of dermatomyositis on oropharyngeal muscle imaging.	Journal of Clinical Rheumatology			In press
杉江和馬 .	ライソゾーム膜の異常：ダノン病 . 神経症候群 III( 第 2 版 ) - その他の神経疾患を含めて - .	別冊日本臨床新領域別症候群シリーズ	28	839-843	2014
Madoka Mori-Yoshimura , Yasushi Oya, Hiroyuki Yajima, Naohiro Yonemoto, Yoko Kobayashi, Yukiko K. Hayashi, Satoru Noguchi, Ichizo Nishino, Miho Murata	GNE myopathy: a prospective natural history study of disease progression.	Neuromuscular Disord.	24(5)	380-386	2014

発表者名	論文タイトル	発表誌	巻・号	ページ	出版年
Madoka Mori-Yoshimura, Yukiko K Hayashi, Naohiro Yonemoto, Harumasa Nakamura, Miho Murata, Shin'ichi Takeda, Ichizo Nishino and En Kimura	Nationwide patient registry for GNE myopathy in Japan.	Orphanet J Rare Dis.	9	150	2014
Ayaki T, Ito H, Fukushima H, (7名) Nakano S, Kusaka H, (7名)	Immunoreactivity of valosin-containing protein in sporadic amyotrophic lateral sclerosis and in a case of its novel mutant.	Acta Neuropathol Commun.	2(1).	172 [Epub ahead of print]	2014
辰野健太郎, 中 村聖香, 朝山 知 子, 中野智	筋症状のみを呈した慢 性ミオパチー型筋サル コイドーシスの 1 例	臨床神経	54	313-316	2014
村田顕也, 伊東秀文	封入体筋炎の病態と原 因	Brain and Nerve	66	1385-1394	2014

発表者名	論文タイトル	発表誌	巻・号	ページ	出版年
Nakamura M, Hachiya N, <u>Murata K</u> , Nakanishi I, Kondo T, Yasutake A, Miyamoto KI, Ser PH, Omi S, Furusawa H, Watanabe C, Usuki F, Sakamoto M.	Methylmercury exposure and neurological outcomes in Taiji residents accustomed to consuming whale meat.	Environ Int	68	25-32	2014
Watanabe Y, Suzuki S, Nishimura H, <u>Murata K</u> , Kurashige T, Ikawa M, Asahi M, Konishi H, Mitsuma S, Kawabata S, Suzuki N, Nishino I	Statins and myotoxic effects associated with anti-3-hydroxy-3-methylglutaryl-coenzyme A reductase autoantibodies: an observational study in Japan.	Medicine	94	e416	
Nakane S, Higuchi O, Koga M, Kanda T, <u>Murata K</u> , Suzuki T, Kuroh H, Kunimoto M, Kaida K, Mukaino A, Sakai W, Maede Y and Matsuo H	Clinical features of autoimmune autonomic ganglionopathy and the detection of subunit-specific autoantibodies to the ganglionic acetylcholine receptor in Japanese patients	PLos one			in press

発表者名	論文タイトル	発表誌	巻・号	ページ	出版年
村田 顕也	偽性髄膜瘤	神経症候群 (2版) V		817-821	2014
村田 顕也	巣状筋炎	骨格筋症候群 上 (第2版)		印刷中	
村田 顕也	増殖性筋炎	骨格筋症候群 上 (第2版)		印刷中	
村田 顕也	微小管障害性ミオパチー	骨格筋症候群 下 (第2版)		印刷中	
Hori H, Yamashita S, Tawara N, Hirahara T, Kawakami K, Nishikami T, Maeda Y, Ando Y.	Clinical features of Japanese patients with inclusion body myositis.	Journal of the Neurological Sciences	346 (1-2)	133-137	2014
Yamashita S, Mori A, Nishida Y, Kurisaki R, Tawara N, Nishikami T, Misumi Y, Ueyama H, Imamura S, Higuchi Y, Hashiguchi A, Higuchi I, Morishita S, Yoshimura J, Uchino M, Takashima H, Tsuji S, Ando Y.	Clinicopathological features of the first Asian family having vocal cord and pharyngeal weakness with distal myopathy due to a MATR3 mutation.	Neuropathol ogy and Applied Neurobiology .		In press	2015