

### 研究成果の刊行に関する一覧表

発表者名	論文タイトル名	発表誌	巻・号	ページ	出版年
Mori-Yoshimura M, Oya Y, Hayashi YK, Noguchi S, Nishino I, Murata M	Respiratory dysfunction in patients severely affected by GNE myopathy (distal myopathy with rimmed vacuoles).	Neuromuscul Disord	23(1)	84-88	2013
Nalini A, Gayathri N, Nishino I, Hayashi YK	GNE myopathy in India.	Neurol India	61(4)	371-374	2013
Yonekawa T, Komaki H, Okada M, Hayashi YK, Nonaka I, Sugai K, Sasaki M, Nishino I	Rapidly progressive scoliosis and respiratory deterioration in Ullrich congenital muscular dystrophy.	J Neurol Neurosurg Psychiatry.	84(9)	982-988	2013
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 (GNE myopathy).	J Neurol Neurosurg Psychiatry.	[Epub ]		2013
Murakami N, Hayashi YK, Oto Y, Shiraishi M, Itabashi H, Kudo K, Nishino I, Nonaka I, Nagai T	Congenital generalized lipodystrophy type 4 with muscular dystrophy: Clinical and pathological manifestations in early childhood.	Neuromuscul Disord.	23(5)	441-444	2013

発表者名	論文タイトル名	発表誌	巻・号	ページ	出版年
Gupta VA, et al.	Identification of KLHL41 Mutations Implicates BTB-Kelch-Mediated Ubiquitination as an Alternate Pathway to Myofibrillar Disruption in Nemaline Myopathy.	Am J Hum Genet	93・6	1108-1117	2013
Motoki T, et al.	Fatal hepatic hemorrhage by peliosis hepatis in X-linked myotubular myopathy: A case report.	Neuromuscul Disord	23・11	917-921	2013
Matsuura T, et al.	Exome sequencing as a diagnostic tool to identify a causal mutation in genetically highly heterogeneous limb-girdle muscular dystrophy.	J Hum Genet	58・8	546-565	2013
Liang WC, et al.	Limb-girdle muscular dystrophy type 2I is not rare in Taiwan.	Neuromuscul Disord.	23・8	675-681	2013
Ravenscroft G, et al.	Mutations in KLHL40 Are a Frequent Cause of Severe Autosomal-Recessive Nemaline Myopathy.	Am J Hum Genet	93・1	6-18	2013
Murakami N, et al.	Congenital generalized lipodystrophy type 4 with muscular dystrophy: Clinical and pathological manifestations in early childhood.	Neuromuscul Disord	23・5	441-444	2013

発表者名	論文タイトル名	発表誌	巻・号	ページ	出版年
須藤 章、他	<i>ACTA1</i> 遺伝子変異を有する重症乳児型ネマリンミオパチーの兄弟例 .	脳と発達	45・6	452-456	2013
林 由起子	Myofibrillar myopathy	臨床神経学	53・11	1105-1108	2013
Nakamura H, Kimura E, Mori-Yoshimura M, Komaki H, Matsuda Y, Goto K, Hayashi YK, Nishino I, Takeda SI, Kawai M.	Characteristics of Japanese Duchenne and Becker muscular dystrophy patients in a novel Japanese national registry of muscular dystrophy (Remudy).	Orphanet J Rare Dis.	8(1)	60	2013
Yonekawa T, Komaki H, Okada M, Hayashi YK, Nonaka I, Sugai K, Sasaki M, Nishino I.	Rapidly progressive scoliosis and respiratory deterioration in Ullrich congenital muscular dystrophy.	J Neurol Neurosurg Psychiatry.	84	982-988	2013

発表者名	論文タイトル名	発表誌	巻・号	ページ	出版年
Nakata T, Ito M, Azuma Y, Otsuka K, Noguchi Y, Komaki H, Okumura A, Shiraishi K, Masuda A, Natsume J, Kojima S, Ohno K.	Mutations in the C-terminal domain of ColQ in endplate acetylcholinesterase deficiency compromise ColQ-MuSK interaction.	Hum Mutat.	34	997-1004	2013
Yonekawa T, Komaki H, Saito Y, Takashima H, Sasaki M.	Congenital hypomyelinating neuropathy attributable to a de novo p.Asp61Asn mutation of the myelin protein zero gene.	Pediatr Neurol.	48	59-62	2013
Takeuchi F, Yonemoto N, Nakamura H, Shimizu R, Komaki H, Mori-Yoshimura M, Hayashi YK, Nishino I, Kawai M, Kimura E, Takeda S.	Prednisolone improves walking in Japanese Duchenne muscular dystrophy patients.	J Neurol.	In press		

発表者名	論文タイトル名	発表誌	巻・号	ページ	出版年
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, Ohno K, Takahashi MP.	A Kir3.4 mutation causes Andersen–Tawil syndrome by an inhibitory effect on Kir2.1.	Neurology	印刷中		2014
Koebis M, Kiyatake T, Yamamura H, Nagano K, Higashihara M, Sonoo M, Hayashi Y, Negishi Y, Endo-Takahashi Y, Yanagihara D, Matsuda R, Takahashi MP, Nishino I, Ishiura S.	Ultrasound-enhanced delivery of Morpholino with Bubble liposomes ameliorates the myotonia of myotonic dystrophy model mice.	Sci Rep.	3	2242	2013
Oana K, Oma Y, Suo S, Takahashi MP, Nishino I, Takeda S, Ishiura S.	Manumycin A corrects aberrant splicing of Clcn1 in myotonic dystrophy type 1 (DM1) mice.	Sci Rep.	3	2142	2013
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	医学のあゆみ	245・9	732-739	2013
高橋正紀	周期性四肢麻痺	今日の診断指針	印刷中		

発表者名	論文タイトル名	発表誌	巻・号	ページ	出版年
Ning R de Vega S, Kurihara H, Ichikawa-Tomikawa N, Xu Z, Nonaka R, Yamada Y, Miner J, Arikawa-Hirasawa E.	Laminin $\alpha 1$ regulates age-related mesangial cell proliferation and mesangial matrix accumulation through the TGF $\beta$ pathway.	Am J Pathol	in press.		2013
Furuya N, Ikeda SI, Sato S, Soma S, Ezaki J, Trejo JA, Takeda-Ezaki M, Fujimura T, Arikawa-Hirasawa E, 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	in press.		2013
Kerever A, Mercier F, Nonaka R, de Vega S, Oda Y, Zalc B, Okada Y, Hattori N, Yamada Y, Arikawa-Hirasawa,	Perlecan is required for FGF-2 signaling in the neural stem cell niche.	Stem Cell Res	121	492-505	2013

発表者名	論文タイトル名	発表誌	巻・号	ページ	出版年
Douet V, Arikawa-Hiras awa E, Mercier F	Fractone-heparan sulfates mediate FGF-2 stimulation of cell proliferation in the adult subventricular zone	Cell Prolif	46	137-145 .	2013
Nakata T, Ito M, Azuma Y, Otsuka K, Noguchi Y, Komaki H, Okumura A, Shiraishi K, Masuda A, Natsume J, Kojima S, Ohno K.	Mutations in the C-terminal domain of ColQ in endplate acetylcholinesterase deficiency compromise ColQ-MuSK interaction	Hum Mutat	34	997-100 4	2013
Selcen D, Shen XM, Milone M, Brenngman J, Ohno K, Deymeer F, Finkel R, Rowin J, Engel AG.	Gfpt1-myasthenia: Clinical, structural, and electrophysiologic heterogeneity	Neurology	81	370-378	2013
Fujioka Y, Ishigaki S, Masuda A, Iguchi Y, Udagawa T, Watanabe H, Katsuno M, Ohno K, Sobue G.	FUS-regulated region- and cell-type-specific transcriptome is associated with cell selectivity in ALS/FTLD	Sci Rep	3	2388	2013

発表者名	論文タイトル名	発表誌	巻・号	ページ	出版年
Rahman MA, Masuda A, Ohe K, Ito M, Hutchinson DO, Mayeda A, Engel AG, Ohno K.	HnRNP L and hnRNP LL antagonistically modulate PTB-mediated splicing suppression of CHRNA1 pre-mRNA	Sci Rep	3	2931	2013
Ohno K, Ito M, Kawakami Y, Krejci E, Engel AG.	Specific binding of collagen Q to the neuromuscular junction is exploited to cure congenital myasthenia and to explore bases of myasthenia gravis	Chem Biol Interact	203	pp 335-340 (査読有)	2013
Ohno K, Ito M, Kawakami Y.	Collagen Q is a key player for developing rational therapy for congenital myasthenia and for dissecting the mechanisms of anti-MuSK myasthenia gravis	J Mol Neurosci, Springer, New York		DOI 10.1007 /s12031 -013-01 70-x, 3 pages (査読有)	2013
Ohkawara B, Cabrera Serrano M, Nakata T, Milone M, Asai N, Ito K, Ito M, Masuda A, Ito Y, Engel AG, Ohno K.	LRP4 third $\beta$ -propeller domain mutations cause novel congenital myasthenia by compromising agrin-mediated MuSK signaling in a position-specific manner	Hum Mol Genet		in press	
Ohno K, Ohkawara B, Ito M, Engel AG.	Molecular Genetics of Congenital Myasthenic Syndromes	eLS. John Wiley & Sons, Inc.		in press (査読有)	



発表者名	論文タイトル名	発表誌	巻・号	ページ	出版年
Sato T, Hayashi YK, Oya Y, Kondo T, Sugie K, Kaneda D, Houzen H, Yabe I, Sasaki H, Noguchi S, Nonaka I, Osawa M, Nishino I.	DNAJB6 myopathy in an Asian cohort and cytoplasmic/nuclear inclusions.	Neuromuscul Disord.	23(3)	269-276	2013
Kataoka H, Saeki K, Kobayashi Y, Kiriyaama T, Sugie K, Ueno S.	Predictors of outcomes in acyclovir-treated limbic encephalitis.	J Infect	66(2)	201-205	2013
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	印刷中	2014
Yamashita S, Kimura E, Tawara N, et al.	Optineurin is potentially associated with TDP-43 and involved in the pathogenesis of inclusion body myositis.	Neuropathol Appl Neurobiol.	39 (4)	406-416	2013
Uchino M, Yamashita S, Uchino K, et al.	Muscle biopsy findings predictive of malignancy in rare infiltrative dermatomyositis.	Clin Neurol Neurosurg.	115 (5)	603-606	2013
Tanaka A, Woltjen K, Miyake K, et al.	Efficient and Reproducible Myogenic Differentiation from Human iPS Cells: Prospects for Modeling Miyoshi Myopathy In Vitro.	PLoS One	8 (4)	e61540	2013

発表者名	論文タイトル名	発表誌	巻・号	ページ	出版年
Mori-Yoshimura M, Momma K, Suzuki N, et al.	Heterozygous UDP-GlcNAc 2-epimerase and N-acetylmannosamine kinase domain mutations in the GNE gene result in a less severe GNE myopathy phenotype compared to homozygous N-acetylmannosamine kinase domain mutations	Journal of the Neurological Sciences	318(1-2)	100-105	2012
Nakamura S, Kaneko S, Shinde A, Morita J, Fujita K, Nakano S, Kusaka H.	Prednisolone-sparing effect of cyclosporin A therapy for very elderly patients with myasthenia gravis.	Neuromuscul Disord.	23(2)	176-179	2013
Nakamura M, Kaneko S, Ito H, Jiang S, Fujita K, Wate R, Nakano S, Fujisawa J, Kusaka H.	Activation of transforming growth factor- $\beta$ /Smad signaling reduces aggregate formation of mislocalized TAR DNA-binding protein-43.	Neurodegener Dis.	11(4)	182-193	2013
中野 智	先天性ミオトニー	今日の神経疾患治療指針 第2版 水澤英洋、鈴木側宏、梶龍兒 他編集	医学書院、東京	786-788	2013

発表者名	論文タイトル名	発表誌	巻・号	ページ	出版年
中野 智	11 . 筋疾患	わかりやすい内科学 第4版 井村裕夫 他 編集	文光堂、東京	646-653	2014
Rumiko Izumi, Tetsuya Niihori, Yoko Aoki1, Naoki Suzuki, Masaaki Kato, Hitoshi Warita, Toshiaki Takahashi, Maki Tateyama, Takeshi Nagashima, Ryo Funayama, Koji Abe, Keiko Nakayama, Masashi Aoki and Yoichi Matsubara	Exome sequencing identifies a novel TTN mutation in a family with hereditary myopathy with early respiratory failure	Journal of Human Genetics		1-8	2013
Mori-Yoshimura M, Oya Y, Hayashi YK, Noguchi S, Nishino I, Murata M	Respiratory dysfunction in patients severely affected by GNE myopathy (distal myopathy with rimmed vacuoles).	Neuromuscul Disord.	23(1)	84-88	2013
Ken-ya Murata, Ken Kouda, Fumihiro Tajima, Tomoyoshi Kondo	Balloon Dilation in Sporadic Inclusion Body Myositis Patients with Dysphagia	Clinical Medicine Insights		1-7	2013
Yamashita S, Kimura E, Tawara N, et al.	Optineurin is potentially associated with TDP-43 and involved in the pathogenesis of inclusion body myositis.	Neuropathol Appl Neurobiol.		In press	2013

発表者名	論文タイトル名	発表誌	巻・号	ページ	出版年
Uchino M, Yamashita S, Uchino K, et al.	Muscle biopsy findings predictive of malignancy in rare infiltrative dermatomyositis.	Clin Neurol Neurosurg.		In press	2013
Ramachandran N, Munteanu I, Wang P, Ruggieri A, Rilstone JJ, Israelian N, Naranian T, Paroutis P,Guo R, Ren ZP, Nishino I, Chabrol B, Pellissier JF, Minetti C, Udd B, Fardeau M, Tailor CS,Mahuran DJ, Kissel JT, Kalimo H,Levy N, Manolson MF, Ackerley CA, Minassian BA	VMA21 deficiency Prevents vacuolar ATPase assembly and Causes autophagic vacuolar myopathy.	Acta Neuropatho l.		Epub ahead of print	
Furuta A, Wakabayashi K, Haratake J, Kikuchi H, Kabuta T, Mori F, Tokonami F, Katsumi Y, Tanioka F, Uchiyama Y, Nishino I, Wada K	Lysosomal storage and advanced senescence in the brain of LAMP-2 deficient Danon Disease.	Acta Neuropatho l.		Epub ahead of print	

発表者名	論文タイトル名	発表誌	巻・号	ページ	出版年
Neumann M, Valori CF, Ansorge O, Kretzschmar HA, Munoz DG, Kusaka H, Yokota O, Ishihara K, Ang LC, Bilbao JM, Mackenzie IR.	Transportin 1 accumulates specifically with FET proteins but no other transportin cargos in FTLD-FUS and is absent in FUS inclusions in ALS with FUS mutations.	Acta neuropathol ologica	124(5)	705-716	2012
Nakamura S, Nakano S, Nishii M, Kaneko S, Kusaka H.	Localization of O-GlcNAc-modified proteins in neuromuscular diseases.	Medical molecular morphology	45(2)	86-90	2012
中野 智, 日下 博文	「封入体筋炎における核遺残 物を含んだ空胞」特集 細胞 の分子構造と機能—核以外の 細胞小器官 8. 膜小胞と封入 体	生体の科学	63:53 4-535		2012
Inamori Y, Higuchi I, Inoue T, Sakiyama Y, Hashiguchi A, Higashi K, Shiraishi T, Okubo R, Arimura K, Mitsuyama Y, Takashima H.	Inclusion body myositis coexisting with hypertrophic cardiomyopathy: an autopsy study.	Neuromuscu lar Disorders	22	747-754	2012
Uchino M, Yamashita S, Uchino K, et al.	Long-term outcome of polymyositis treated with high single-dose alternate-day prednisolone therapy.	Eur Neurol.	68(2)	117-21	2012
高松直子、寺澤 由佳、酒井和香、 宮本亮介、宮城 愛、島谷佳光、 佐藤健太、松井 尚子、 和泉唯信、梶龍 兒	筋超音波所見を契機として確 定診断できたサルコイドーシ スの一例	Neurosonol ogy	25(1)	13-16	2012

発表者名	論文タイトル名	発表誌	巻・号	ページ	出版年
Keduka E, <u>Hayashi YK</u> , Shalaby S, Mitsuhashi H, Noguchi S, Nonaka I, Nishino I	<i>In Vivo</i> Characterization of Mutant Myotilins.	Am J Pathol.	180・4	1570-1580	2012
Tsuburaya RS, Monma K, Oya Y, Nakayama T, Fukuda T, Sugie H, <u>Hayashi YK</u> , Nonaka I, Nishino I	Acid phosphatase-positive globular inclusions is a good diagnostic marker for two patients with adult-onset Pompe disease lacking disease specific pathology.	Neuromuscul Disord.	22・5	389-393	2012
Suzuki S, <u>Hayashi YK</u> , Kuwana M, Tsuburaya R, Suzuki N, Nishino I	Myopathy associated with antibodies to signal recognition particle: disease progression and neurological outcome.	Arch Neurol.	69・6	728-732	2012
Yoshinaga Y. Sakoda S-I, Good JM, Takahashi MP, Kubota T, Arikawa-Hirasawa E, Nakata T, Ohno K, Kitamura T, Kobayashi K, Ohtsuka Y.	A novel mutation in SCN4A causes severe myotonia and school-age-onset paralytic episodes.	J Neurol Sci.	315・1-2	15-19	2012
Kokunai Y, Goto K, Kubota T, Fukuoka T, Sakoda S, Ibi T, Doyu M, Mochizuki H, Sahashi K, Takahashi MP.	A sodium channel myotonia due to a novel SCN4A mutation accompanied by acquired autoimmune myasthenia gravis.	Neurosci Lett.	519・1	67-72	2012

発表者名	論文タイトル名	発表誌	巻・号	ページ	出版年
Muneaki Ishijima , Nobuharu Suzuki, Kentaro Hozumi, Tomoya Matsunobu, Keisuke Kosaki , Haruka Kaneko, JohnR. Hassell,Eri Arikawa-Hiras awa , Yoshihiko Yamada ,	Perlecan modulates VEGF signaling and is essential for vascularization in endochondral bone formation	Matrix Biology	12138	In Press (1-5)	2012
Harumi Yoshinaga , Shunichi Sakoda, Jean-Marc Good, Masanori P. Takahashi , Tomoya Kubota , Eri Arikawa-Hiras awa , Tomohiko Nakata, Kinji Ohno , Tetsuro Kitamura, Katsuhiko Kobayashi, Yoko Ohtsuka	A novel mutation in SCN4A causes severe myotonia and school-age-onset paralytic episodes	Journal of the Neurologica l Sciences	12138	In Press (1-5)	2012

発表者名	論文タイトル名	発表誌	巻・号	ページ	出版年
Takenori Inomata, Nobuyuki, Ebihara, Toshinari Funaki, Akira Matsuda, Yasuo Watanabe, Liang Ning, Zhuo Xu, Akira Murakami, and Eri Arikawa-Hirasawa,	Perlecan-Deficient Mutation Impairs Corneal Epithelial Structure	IOVS	Vol.53, No. 3	1277-1284	2012
Masuda A, Andersen HS, Doktor TK, Okamoto T, Ito M, Andresen BS, Ohno K.	CUGBP1 and MBNL1 preferentially bind to 3' UTRs and facilitate mRNA decay	<i>Sci Rep</i>	2	209	2012
Ito M, Suzuki Y, Okada T, Fukudome T, Yoshimura T, Masuda A, Takeda S, Krejci E, Ohno K.	Protein-anchoring strategy for delivering acetylcholinesterase to the neuromuscular junction	<i>Mol Ther</i>	20	1384-1392	2012
Ishigaki S, Masuda A, Fujioka Y, Iguchi Y, Katsuno M, Shibata A, Urano F, Sobue G, <u>Ohno K.</u>	Position-dependent fus-rna interactions regulate alternative splicing events and transcriptions	<i>Sci Rep</i>	2	529	2012
Sugie K, Hayashi YK, Goto K, Nishino I, Ueno S.	Unusual presentation: Unilateral arm and contralateral leg amyotrophy in FSHD.	Neurology	79(5)	e46	2012



発表者名	論文タイトル名	発表誌	巻・号	ページ	出版年
Sugie K, Tonomura Y, Ueno S.	Characterization of dermatomyositis with coexistence of anti-Jo-1 and anti-SRP antibodies.	Intern Med	51(7)	799-802	2012
杉江和馬 .	ライソゾーム病：ダノン病 . 先天代謝異常症候群 - 病因・ 病態研究、診断・治療の進歩 - .	日本臨床	20	588-592	2012
Momma K, Noguchi S, Malicdan MC, Hayashi YK, Minami N, Kamakura K, Nonaka I, Nishino I	Rimmed vacuoles in Becker muscular dystrophy have similar features with inclusion myopathies.	PLoS One.	7(12)	e52002	2012
Komagamine T, Kawai M, Kokubun N, Miyatake S, Ogata K, Hayashi YK, Nishino I, Hirata K	Selective muscle involvement in a family affected by a second LIM domain mutation of fhl1: An imaging study using computed tomography.	J Neurol Sci.	318(2012)	163-167	2012
Mori-Yoshimura M, Monma K, Suzuki N, Aoki M, Kumamoto T, Tanaka K, Tomimitsu H, Nakano S, Sonoo M, Shimizu J, Sugie K, Nakamura H, Oya Y, Hayashi YK, Malicdan MC, Noguchi S, Murata M, Nishino I	Heterozygous UDP-GlcNAc 2-epimerase and N-acetylmannosamine kinase domain mutations in the GNE gene result in a less severe GNE myopathy phenotype compared to homozygous N-acetylmannosamine kinase domain mutations.	J Neurol Sci.	318(2012):	100-105	2012

発表者名	論文タイトル名	発表誌	巻・号	ページ	出版年
Nakamura M, Kaneko S, Ito H, Jiang S, Fujita K, Wate R, Nakano S, Fujisawa JI, Kusaka H.	Activation of Transforming Growth Factor-β/Smad Signaling Reduces Aggregate Formation of Mislocalized TAR DNA-Binding Protein-43.	Neuro-degenerative diseases	:Jul 10.	[Epub ahead of print]	2012
Nakamura S, Kaneko S, Shinde A, Morita JI, Fujita K, Nakano S, Kusaka H.	Prednisolone-sparing effect of cyclosporin A therapy for very elderly patients with myasthenia gravis.	Neuromuscular disorders : NMD	:Dec 10.	[Epub ahead of print]	2012
Nakamura M, Kaneko S, Wate R, Asayama S, Nakamura Y, Fujita K, Ito H, Kusaka H.	Regionally different immunoreactivity for Smurf2 and pSmad2/3 in TDP-43-positive inclusions of amyotrophic lateral sclerosis.	Neuropathology and applied neurobiology	:Mar 21.	[Epub ahead of print]	2012
Yamashita S, Mori A, Sakaguchi H, et al.	Sporadic juvenile amyotrophic lateral sclerosis caused by mutant FUS/TLS: possible association of mental retardation with this mutation.	J Neurol.	259(6)	1039-44	2012
Yamashita S, Sakaguchi H, Mori A, et al.	Significant CMAP decrement by repetitive nerve stimulation is more frequent in median than ulnar nerves of patients with amyotrophic lateral sclerosis.	Muscle Nerve	45(3)	426-8	2012
小牧宏文	小児の診療手技 100 筋生検	小児科診療	75	276-278	2012

発表者名	論文タイトル名	発表誌	巻・号	ページ	出版年
小牧宏文	症候・疾患と検査・診断 神経筋疾患の診断	小児神経学の進歩	41	69-77	2012
石山昭彦, 小牧宏文	小児慢性疾患の生活指導 - 最新の知見から - 10.神経・筋疾患 2) 先天性ミオパチー	小児科臨床	65巻4号	839-846	2012
石山昭彦, 藤義朗	先天性筋ジストロフィー	小児内科	44巻増刊号	794-795	2012
佐々木良元、高橋正紀、穀内洋介、平山正昭、衣斐 達、富本秀和、望月秀樹、佐橋 功	骨格筋型塩化物イオンチャネル遺伝子 (CLCN1) の複合ヘテロ接合体変異で重症化した Thomsen 病	臨床神経学	印刷中		
Yoshinaga H, Sakoda S, Good J M, Takahashi M P, Kubota T, Arikawa-Hirasawa E, Nakata T, Ohno K, Kitamura T, Kobayashi K, Ohtsuka Y.	A novel mutation in <i>SCN4A</i> causes severe myotonia and school-age-onset paralytic episodes	<i>J Neurol Sci</i>	315	15-19	2012
Matsuura T, Minami N, Arahata H, Ohno K, Abe K, Hayashi YK, Nishino I.	Myotonic dystrophy type 2 is rare in the Japanese population	<i>J Hum Genet</i>	57	219-220	2012

発表者名	論文タイトル名	発表誌	巻・号	ページ	出版年
Yamashita Y*, Matsuura T*, Shinmi J, Amakusa Y, Masuda A, Ito M, Kinoshita M, Furuya H, Abe K, Ibi T, Sahashi K, Ohno K.	Four parameters increase the sensitivity and specificity of the exon array analysis and disclose twenty-five novel aberrantly spliced exons in myotonic dystrophy	<i>J Hum Genet</i>	57	368-374	2012
Yamamoto R, Matsushita M, Kitoh H, Masuda A, Ito M, Katagiri T, Kawai T, Ishiguro N, Ohno K.	Clinically applicable antianginal agents suppress osteoblastic transformation of myogenic cells and heterotopic ossifications in mice	<i>J Bone Miner Metab</i>	31	26-33	2012
Ohe K, Masuda A, Ohno K.	Intronic and exonic nucleotide variations that affect rna splicing in humans	<i>Introduction to Sequence and Genome Analysis.</i> iConcept Press, Hong Kong		in press	2012

発表者名	論文タイトル名	発表誌	巻・号	ページ	出版年
Mori-Yoshimura M, Monma K, Suzuki N, Aoki M, Kumamoto T, Tanaka K, Tomimitsu H, Nakano S, Sonoo M, Shimizu J, Sugie K, Nakamura H, Oya Y, Hayashi YK, Malicdan MC, Noguchi S, Murata M, Nishino I.	Heterozygous UDP-GlcNAc 2-epimerase and N-acetylmannosamine kinase domain mutations in the GNE gene result in a less severe GNE myopathy phenotype compared to homozygous N-acetylmannosamine kinase domain mutations.	J Neurol Sci	318(1-2)	100-105	2012
Sawa N, Kataoka H, Sugie K, Kawahara M, Horikawa H, Kusunoki S, Ueno S.	Clinical analysis and outcomes of amyotrophic lateral sclerosis with demyelinating polyneuropathy.	Amyotroph Lateral Scler	13(1)	125-31	2012
杉江和馬 .	顔面肩甲上腕型筋ジストロフィーの骨格筋障害の分布 .	難病と在宅ケア	17(10)	53-55	2012