

## 研究成果の刊行に関する一覧表

### 雑誌

| 発表者氏名   | 論文タイトル名   | 発表誌名                                       | 巻号 | ページ       | 出版年  |
|---|---|--|----|-----------|------|
| 木村 円, 中村 治雅, 三橋 里美, 竹内 芙実, 森 まどか, 清水 玲子, 小牧 宏文, 林 由起子, 西野 一三, 川井 充, 武田 伸一   | 筋ジストロフィーの臨床開発を推進する研究基盤:<br>RemudyとMDCTN   | <b>臨床神経</b>                                | 54 | 1069-1070 | 2014 |
| 高橋 正紀, 中森 雅之, 望月 秀樹   | 筋強直性ジストロフィー症の治療開発   | <b>臨床神経</b>                                | 54 | 1077-79   | 2014 |
| Matsumura T, Kimura T, Kokunai Y, et al.  | A simple questionnaire for screening patients with myotonic dystrophy type 1                  | <b>Neurology and Clinical Neuroscience</b> | 2  | 87-103    | 2014 |
| Nakayama T, Nakamura H, Oya Y, Kimura T, Imahuku I, <u>Ohno K</u> , <u>Nishino I</u> , Abe K, <u>Matsuura T</u> . | Clinical and genetic analysis of the first known Asian family with myotonic dystrophy type 2. | <b>J Hum Genet</b>                         | 59 | 129-33    | 2014 |
| 高橋 俊明 他   | 舞踏運動を呈した dysferlin異常症の1例  | <b>JMDD</b>                                | 24 | 51-54     | 2014 |

| 発表者氏名  | 論文タイトル名   | 発表誌名  | 巻号          | ページ     | 出版年  |
|--|---|---|-------------|---------|------|
| <u>Mori-Yoshimura M</u> ,<br><u>Hayashi YK</u> , <u>Yonemoto N</u> , Nakamura H,<br>Murata M, <u>Takeda S</u> ,<br><u>Nishino I</u> , <u>Kimura E</u> .    | Nationwide patient registry for GNE myopathy in Japan   | <i>Orphanet J Rare Diseases</i>             | 9(1)        | 150     | 2014 |
| Goto M, Okada M,<br><u>Komaki H</u> , Sugai K,<br>Sasaki M, <u>Noguchi S</u> ,<br>Nonaka I, <u>Nishino I</u> ,<br><u>Hayashi YK</u> .                      | A nationwide survey on Marinesco-Sjögren syndrome in Japan.   | <i>Orphanet J Rare Diseases</i>             | 9(1)        | 58      | 2014 |
| Hori H, <u>Yamashita S</u> ,<br>Tawara N, Hirahara T,<br>Kawakami K, Nishikami T,<br>Maeda Y, Ando Y.  | Clinical features of Japanese patients with inclusion body myositis   | <i>Journal of the Neurological Sciences</i> | 34<br>(1-2) | 133-137 | 2014 |
| Izumi R, Niihori T,<br>Takahashi T, Suzuki N,<br>Tateyama M, Watanabe C,<br>Sugie K, Nakanishi H,<br>Sobue G, Kato M,<br>Warita H, Aoki Y, <u>Aoki M</u> . | Genetic profile for suspected dysferlinopathy identified by targeted next-generation sequencing.  | <i>Neurol Genet</i>                         | 1           | e36     | 2015 |
| <u>Yamashita S</u> , Mori A,<br>Nishida Y, Kurisaki R,<br>Tawara N, Nishikami T,<br>Misumi Y, Ueyama H, <i>et al.</i>                                      | Clinicopathological features of the first Asian family having vocal cord and pharyngeal weakness with distal myopathy due to a <i>MATR3</i> mutation. | <i>Neuropathol Appl Neurobiol</i>           | 41          | 391-398 | 2015 |

| 発表者氏名  | 論文タイトル名  | 発表誌名                           | 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>Neuromuscul Disord</i>      | 25 | 60-69   | 2015 |
| Selcen D, Ohkawara B, Shen XM, McEvoy K, <u>Ohno K</u> , Engel AG.   | Impaired Synaptic Development, Maintenance, and Neuromuscular Transmission in LRP4-Related Myasthenia  | <i>JAMA Neurol</i>             | 72 | 889-896 | 2015 |
| Rahman MA, Azuma Y, Nasrin F, Takeda J, Nazim M, Ahsan KB, Masuda A, Engel AG, <u>Ohno K.</u>  | SRSF1 and hnRNP H antagonistically regulate splicing of COLQ exon 16 in a congenital myasthenic syndrome   | <i>Sci Rep</i>                 | 5  | 13208   | 2015 |
| <u>Watanabe N</u> , Horikoshi M, Yamada M, Shimodera S, Akechi T, Miki K, Inagaki M, <u>Yonemoto N</u> , <i>et al.</i>   | Adding smartphone-based cognitive-behavior therapy to pharmacotherapy for major depression (FLATT project): study protocol for a randomized controlled trial | <i>Trials</i>                  | 16 | 293     | 2015 |
| <u>Watanabe N</u> , Furukawa TA, Shimodera S, Katsuki F, Fujita H, Sasaki M, Sado M, Perlis ML.  | Cost-effectiveness of cognitive behavioral therapy for insomnia comorbid with depression: Analysis of a randomized controlled trial                          | <i>Psychiatry Clin Neurosc</i> | 69 | 335-343 | 2015 |

| 発表者氏名   | 論文タイトル名  | 発表誌名                          | 巻号        | ページ     | 出版年  |
|---|--|-------------------------------|-----------|---------|------|
| Kuraoka M, <u>Kimura E</u> , Nagata T, Okada T, Aoki Y, Tachimori H, <u>Yonemoto N</u> , Imamura M, Takeda S  | Serum Osteopontin as a Novel Biomarker for Muscle Regeneration in Duchenne Muscular Dystrophy  | <i>Am J Pathol</i>            | 186(5)    | 1302-12 | 2016 |
| Shimizu R, <u>Ogata K</u> , Tamaura A, <u>Kimura E</u> , Ohata M, Takeshita E, Nakamura H, Takeda S, Komaki H   | Clinical trial network for the promotion of clinical research for rare diseases in Japan: muscular dystrophy clinical trial network.                                   | <i>BMC Health Serv Res</i>    | 16        | 241     | 2016 |
| Nishikawa A, <u>Mori-Yoshimura M</u> , Segawa K, Hayashi YK, Takahashi T, Saito Y, Nonaka I, Krahn M, Levy N, Shimizu J, Mitsui J, <u>Kimura E</u> , Goto J, <u>Yonemoto N</u> , <u>Aoki M</u> , et al. | Respiratory and cardiac function in Japanese patients with dysferlinopathy.  | <i>Muscle Nerve</i>           | 53<br>(3) | 394-401 | 2016 |
| Coathup V, Teare HJ, Minari J, Yoshizawa G, Kaye J, <u>Takahashi MP</u> , Kato K.   | Using Digital Technologies to Engage with Medical Research: Views of Myotonic Dystrophy Patients in Japan.   | <i>BMC Medical Ethics</i>     | 17(1)     | 51      | 2016 |
| <u>高橋正紀</u> , <u>松村 剛</u> ,<br><u>木村 円</u>  | 筋強直性ジストロフィー—患者レジストリーと治験・臨床研究   | <b>神経内科</b>                   | 86(6)     | 646-651 | 2016 |
| Okumura K., Yamashita T., Masuda T., Misumi Y., Ueda A., Ueda M., Obayashi K., Jono H., <u>Yamashita S.</u> , Inomata Y., Ando Y.   | Long-term outcome of patients with hereditary transthyretin V30M amyloidosis with polyneuropathy after liver transplantation.  | <i>Amyloid</i>                | 23(1)     | 39-45   | 2016 |
| Azuma M., Hirai T., Yamada K., <u>Yamashita S.</u> , Ando Y., Tateishi M., Iryo Y., Yoneda T., Kitajima M., Wang Y., Yamashita Y.   | Lateral asymmetry and spatial difference of iron deposition in the substantia nigra of Parkinson's disease patients measured with quantitative susceptibility mapping. | <i>AJNR Am J Neuroradiol.</i> | 37(5)     | 782-788 | 2016 |

| 発表者氏名   | 論文タイトル名  | 発表誌名                         | 巻号         | ページ         | 出版年  |
|---|--|------------------------------|------------|-------------|------|
| Mori A., <u>Yamashita S.</u> , Nakajima M., Hori H., Tawara A., Matsuo Y., Misumi Y., Ando Y.   | CMAP decrement as a potential diagnostic marker for ALS.   | <i>Acta Neurol. Scand.</i>   | 134<br>(1) | 49-53       | 2016 |
| Yamashita T., Ueda M., Saga N., Nanto K., Tasaki M., Masuda T., Misumi Y., Oda S., Fujimoto A., Amano T., Takamatsu K., <u>Yamashita S.</u> , Obayashi K., Matsui H., Ando Y. | Hereditary amyloidosis with cardiomyopathy caused by the novel variant transthyretin A36D.   | <i>Amyloid</i>               | 23(3)      | 207-208     | 2016 |
| Mori Y., <u>Yamashita S.</u> , Kato M., Masuda T., Takamatsu K., Kumamoto T., Sasaki R., Ando Y.  | Thomsen disease with ptosis and abnormal MR findings.  | <i>Neuromuscul. Disord.</i>  | 26(11)     | 805-808     | 2016 |
| <u>山下賢</u> , 安東由喜雄  | 眼咽頭遠位型ミオパチー  | <i>Clinical Neuroscience</i> | 34(3)      | 332-333     | 2016 |
| Chen G, Masuda A, Konishi H, Ohkawara B, Ito M, Kinoshita M, Kiyama H, <u>Matsuura T.</u> , <u>Ohno K.</u>  | Phenylbutazone induces expression of MBNL1 and suppresses formation of MBNL1-CUG RNA foci in a mouse model of myotonic dystrophy.  | <i>Sci Rep</i>               | 6          | 25317       | 2016 |
| Mashiko T, Sakashita E, Kasashima K, Tominaga K, Kuroiwa K, Nozaki Y, <u>Matsuura T.</u> , Hamamoto T, Endo H.  | Developmentally-regulated RNA-binding Protein 1 (Drb1)/RNA-binding Motif Protein 45 (RBM45), a Nuclear-cytoplasmic Trafficking Protein, Forms TAR DNA-binding Protein 43 (TDP-43)-mediated Cytoplasmic Aggregates. | <i>J Biol Chem</i>           | 291        | 14996-15007 | 2016 |

| 発表者氏名   | 論文タイトル名   | 発表誌名                      | 巻号    | ページ     | 出版年  |
|---|---|---------------------------|-------|---------|------|
| Harris E , Bladen CL , Mayhew A , James M , Bettinson K , Moore Y , Smith FE , Rufibach L , Cnaan A , Bharucha-Goebel DX , Blamire AM , Bravver E , Carlier PG , Day JW , Diaz-Manera J , Eagle M , Grieben U , Harms M , Jones KJ , Lochmüller H , Mendell JR , <u>Mori-Yoshimura M</u> , Paradas C , Pegoraro E , Pestronk A , Salort-Campana E , Schreiber-Katz O , Semplicini C , Spuler S , Stojkovic T , Straub V , Takeda S , Rocha CT , Walter MC , Bushby K ; Jain COS Consortium: | The Clinical Outcome Study for dysferlinopathy :An international multicenter study.               | <i>Neurol Genet .</i>     | 2(4)  | e89     | 2016 |
| Suzuki N, <u>Mori-Yoshimura M</u> , <u>Yamashita S</u> , Nakano S, Murata KY, Inamori Y, Matsui N, <u>Kimura E</u> , Kusaka H, Kondo T, Higuchi I, Kaji R, Tateyama M, Izumi R, Ono H, Kato M, Warita H, Takahashi T, Nishino I, <u>Aoki M</u>  | Multicenter questionnaire survey for sporadic inclusion body myositis in Japan.                   | <i>Orphanet J RareDis</i> | 11(1) | 146     | 2016 |
| Itani O, <u>Watanabe N</u> , et al.   | Short sleep duration and health outcomes: a systematic review, meta-analysis, and meta-regression | <i>Sleep Med.</i>         | 32    | 246-256 | 2016 |

| 発表者氏名   | 論文タイトル名  | 発表誌名                                     | 巻号    | ページ      | 出版年  |
|---|--|--|-------|----------|------|
| Furukawa TA, <u>Watanabe N</u> , et al  | Cognitive-Behavioural Analysis System of Psychotherapy (CBASP), a drug, or their combination: differential therapeutics for persistent depressive disorder: a study protocol of an individual participant data network meta-analysis | <b><i>BMJ Open</i></b>                   | 6     | e011769  | 2016 |
| 小林道雄、石崎雅俊、足立克仁、 <u>米本直裕</u> 、松村剛、豊島至、 <u>木村円</u>  | ジストロフィン異常症保因者の遺伝カウンセリング・健康管理の実態に関する調査  | <b><i>臨床神経学</i></b>                      | 56(6) | 407-12   | 2016 |
| Takeuchi F, Komaki H, Nakamura H, <u>Yonemoto N</u> , Kashiwabara K, <u>Kimura E</u> , Takeda S.  | Trends in steroid therapy for Duchenne muscular dystrophy in Japan.  | <b><i>Muscle Nerve.</i></b>              | 54(4) | 673-80   | 2016 |
| De Crescenzo F, <u>Watanabe N</u> , et al   | Comparative efficacy and acceptability of pharmacological treatments for insomnia in adults: a systematic review and network meta-analysis [protocol]  | <b><i>Cochrane Database Syst Rev</i></b> | 9     | CD012364 | 2016 |
| Saito T, <u>Kawai M</u> , <u>Kimura E</u> , <u>Ogata K</u> , Takahashi T, Kobayashi M, Takada H, Kuru S, Mikata T, Matsumura T, <u>Yonemoto N</u> , Fujimura H, Sakoda S. | Study of Duchenne muscular dystrophy long-term survivors aged 40 years and older living in specialized institutions in Japan.  | <b><i>Neuromuscul Disord.</i></b>        | 27(2) | 107-114  | 2017 |
| <u>高橋正紀</u>   | 稀少遺伝性難病の開発研究の現状と課題   | <b><i>生産と技術</i></b>                      | 69(1) | 79-83    | 2017 |

| 発表者氏名  | 論文タイトル名   | 発表誌名                        | 巻号    | ページ     | 出版年  |
|--|---|-----------------------------|-------|---------|------|
| Ishizaki M., Kedoin C., Ueyama H., Maeda Y., <u>Yamashita S.</u> , Ando Y.   | Utility of skinfold thickness measurement in non-ambulatory patients with Duchenne muscular dystrophy.    | <i>Neuromuscul. Disord.</i> | 27(1) | 24-28   | 2017 |
| Zhu W, Mitsuhashi S, Yonekawa T, Noguchi S, Huei JC, Nalini A, Preethish-Kumar V, Yamamoto M, Murakata K, <u>Mori-Yoshimura M</u> , Kamada S, Yahikozawa H, Karasawa M, Kimura S, Yamashita F, Nishino I | Missing genetic variations in GNE myopathy: rearrangement hotspots encompassing 5'UTR and founder allele. | <i>J Hum Genet.</i>         | 62(2) | 159-166 | 2017 |

### 書籍

| 著者氏名        | 論文タイトル名        | 書籍全体の編集者名  | 書籍名                             | 出版社名      | 出版地 | 出版年            | ページ    |
|-------------|----------------|------------|---------------------------------|-----------|-----|----------------|--------|
| <u>大野欽司</u> | 先天性筋無力症候群      | 戸田達史<br>監修 | 医学のあゆみ Vol. 259, No.1           | 医歯薬出版株式会社 | 東京  | 2016<br>(10月号) | 80-86  |
| <u>大野欽司</u> | 先天性筋無力症候群の治療研究 | 武田伸一<br>監修 | CLINICAL CALCIUM Vol. 27, No. 3 | 医薬ジャーナル社  | 大阪  | 2017<br>(3月号)  | 97-104 |