有意な関連が示唆された。[27]（３）ライフスタイルの消極性と発症年齢に有意な関連が示唆された。[28]（４）運動によりSF36のメンタル・コンポーネント･サマリースコア(精神的側面のQOL)の有意な改善が示唆された。[29]（５）HD患者では、対照者に比べ、メラトニンレベルの低下や夜間の活動性の亢進が示唆された（睡眠相後退症候群など概日リズム障害)。[30, 31]

D.考察

Incidence/Prevalenceの差異（表１と２）は、Huntingtin gene haplotypesの人種差によるものであることから、やはり、日本人HD患者の自然史に関する知見が必要とされる。欧米人と日本人のHD患者で、その症状や予後に差異があるか否か、確認しておく必要があると思われる。患者・家族が最も知りたい点は治療経過や予後であるので、個人差があるとはいうものの、偏りのない日本人HD患者集団の自然史を把握しておかなければならない。また、将来、欧米でHDに有効な薬剤が開発された場合に備えて、薬剤の有効性を評価する上でも、日本人HD患者集団の自然史を把握しておくことは必須であると思われる。

COHORT-HDやREGISTRY-HDは、１年に１回、基本情報と神経学的診察所見をモニターして行く観察研究である。本邦では、新法の下、安定的な医療費の助成および療養生活の環境整備とともに、医療に関する調査及び研究を推進することが明記され、平成27年１月１日より施行されている。本邦で、COHORT-HDやREGISTRY-HDと同様の観察研究を実施することは、不可能なのであろうか？ 日本人のHD患者数は極めて少ないので、本法の下、自然史把握に必要なデータを収集し蓄積して行くのが、最も効率的で、本法の趣旨に最も適っていると思われる。

表３に示すような大規模な前向き多施設共同研究のほかに、HDと後天的要因（アルコール・ドラッグ・タバコ、カフェイン、運動、睡眠など）との関連を検討した100人程度の比較的小規模な横断的研究も実施されていた。HDの非薬物療法・新薬開発に関するヒントとなるような研究成果が報告されていた。今後は、表３で示されているような大規模プロジェクトで、さらなる検証が進められるものと思われる。

日本人のHD患者を対象とした疫学研究を進めて行くにあたり、短期的には、既存のデータ（診療録や臨床調査個人票）を用いた後ろ向きCohort研究や横断研究によって後天的要因を特定した上でRCTを実施し介入効果を評価する。長期的には、Cohort研究を立ち上げる、あるいは、既存の患者登録制度を充実させるなどして、日本人のHDの自然史の詳細を明らかにして行くことが重要だと思われる。得られる研究成果は、HD患者の治療・介護、家族へのサポート、カウンセリングを含めた地域医療連携、一般の人々へのHDに関する知見の普及に役立てることができる。

E.結論

HDは極めて稀少性の高い疾患であるため、データ収集が困難であり、それゆえに疫学研究を行うのが難しい面がある。近年、欧米では、大規模な前向き多施設共同研究プロジェクトが進んでいる。本研究では、HDに関する疫学研究の文献レビューを行うとともに、主要な海外の研究プロジェクトに関する情報を収集し、それらの知見・情報が、今後、本邦における疫学研究を実施して行く上で有用となり得るよう取りまとめた。

F.健康危険情報

なし。

G.研究発表

（発表雑誌名巻号・頁・発行年なども記入）

1. 論文発表

なし。

2.学会発表

なし。

H.知的所有権の取得状況（予定を含む）

なし。

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Table 1 Incidence studies of Huntington's disease (HD)

Study (Authors)	Country	Population	Diagnosis	Year	Incidence ($/10^5$/year)
Mercy et al.[2]	UK (Cambidgeshire)	75,600	UHDRS	2000-2006	0.80
Kokmen et al.[3]	USA (Minnesota)	106,000	Unspecified	1950-1989	0.30
Almqist et al.[4]	Canada (British Columbia)	4 million	DNA analysis	1996-1999	0.69
McCusker et al.[5]	Australia (New South Wales)	6,038,695	DNA analysis	1996	0.65
Pavoni et al.[6]	Italy (Ferrara)	370,374	FH Symptoms	1971-1987	0.11
Ramos-Arroyo et al.[7]	Spain (Navarra and Basque)	Unspecified	DNA analysis	1994-2002	0.47
Panas et al.[8]	Greece (Nationwide)	10,964,202	DNA analysis	1995-2008	0.22-0.44
Chang et al.[9]	China (Hong Kong)	5,440,000	DNA analysis	1984-1991	0.05
Chen & Lai[10]	Taiwan (Nationwide)	23,000,000	Unspecified	2007	0.08

Note: The numerical superscripts correspond to the reference numbers listed in the references. All studies used administrative database or hospital chart review to identify cases with HD. UHDRS means the Unified Huntington's Disease Rating Scale. FH means family history.

Table 2 Prevalence studies of Huntington's disease (HD)

Study (Authors)	Country	Populaion	Diagnosis	Year	Prevalence ($/10^5$)
Japan					
Nakashima et al.[11]	Japan (Tottori)	1,387,000	DNA analysis	1993	0.65
Kanazawa et al.[12]	Japan (Ibaraki)	2,638,280	FH Symptoms	1982	1.10
Kishimoto et al.[13]	Japan (Aichi)	3,339,000	FH Symptoms	1957	3.80
Other countries					
Evans et al.[14]	UK (Nationwide)	3,515,986	Unspecified	2010	12.30
James et al.[15]	UK (Wales)	1,393,900	Unspecified	1994	6.20
Shiwach[16]	UK (England)	2,437,000	DNA analysis	1991	5.66
Watt & Seller[17]	UK (England)	2,520,000	Unspecified	1988	4.00
Morrison[18]	Ireland (Northern Ireland)	Unspecified	DNA analysis	2001	10.60
Kokmen et al.[3]	USA (Minnesota)	106,000	Unspecified	1990	1.90
Fisher & Hayden[19]	Canada (British Columbia)	4,609,659	DNA analysis	2012	10.4-13.2
McCusker et al.[5]	Australia (New South Wales)	6,038,695	DNA analysis	1996	6.29
Pridomore[20]	Australia (Tasmania)	447,000	FH Symptoms	1990	12.10
Peterlin et al.[21]	Slovenia	2,011,614	DNA analysis	2006	5.16
Pavoni et al.[6]	Italy (Ferrara)	370,374	FH Symptoms	1987	1.87
Panas et al.[8]	Greece (Nationwide)	10,964,202	DNA analysis	1995-2008	0.25-0.54
Kandil et al.[22]	Eygpt (Assiut)	42,000	Unspecified	1988-1990	21
Chang et al.[9]	China (Hong Kong)	5,440,000	DNA analysis	1984-1991	0.37
Chen and Lai[10]	Taiwan (Nationwide)	23,000,000	Unspecified	2007	0.42

Note: The numerical superscripts correspond to the reference numbers listed in the references. All but one study, door-to-door survey conducted by Kandil et al., used administrative database or hospital chart review to identify cases with HD. Evans et al. used the General Practice Research Database (GPRD). FH means family history.

Table 3 On-going projects on Huntington's disease (HD)

Projects	Countries	Founding
(1) Huntington Study Group (HSG)	USA, Canada, Europe, Australia, New Zealand, South America	1993
・COHORT-HD (Cooperative Huntington's Observational Research Trial)		
・TRACK-HD (a multi-centre, multi-national prospective, observational biomarker study of premanifest and early stage HD with no experimental treatment)		
・PREDICT-HD (Neurobiological Predictors of Huntington's Disease)		
・PHAROS-HD (Prospective Huntington At Risk Observational Study)		
	http://www.huntington-study-group.org	
(2) European HD Network (EHDN)	Europe	2003
・REGISTRY-HD (an Observational Study of the EHDN)		
	http://www.euro-hd.net/html/network	
(3) Enroll-HD	North America, Europe, Latin America, Australia/New Zealand, some countries in Asia	2012
・A Prospective Registry Study in a Global HD Cohort		
	http://www.enroll-hd.org/html/about	

Ⅲ．研究成果の刊行に関する一覧表

英文単行本

著　者　名	論　文　題　名	書　名	（編集者名）	発行社名	（発行地名）	出版西暦年	頁
Toshitaka Kawarai and Ryuji Kaji	Genetics of dystonia	DYSTONIA: A CLINICIAN'S GUIDE TO DIAGNOSIS, PATHOPHYSIOLOGY AND TREATMENT.	Duc H. Le	Future Medicine	London	2013	48-62
Hatano T, Kubo S-I, Hattori N, Mizuno Y	Movement disorders in neoplastic brain disease.	Movement disorders in neurologic and systemic disease	W. Poewe J. Jankovic	Cambridge university press	UK	2014	279-292

英文原著・症例報告

著者名	論文題名	雑誌名	巻	頁	出版西暦年	GRANTへの謝辞の有無
Kenichiro Tanaka, Kenji Wada-Isoe, Mikie Yamamoto, Shugo Tagashira, Yuki Tajiri, Satoko Nakashita & Kenji Nakashima	Clinical evaluation of fatigue in Japanese patients with Parkinson's disease	Brain and Behavior		1-7		
Adachi T, Kitayama M, Nakano T, Adachi Y, Kato S, Nakashima K	Autopsy case of spinocerebellar ataxia type 31 with severe dementia at the terminal stage.	Neuropathology				
Kurashiki-Osaka T, Adachi T, Nakayasu H, Nakashima K.	Alpha-synuclein accumulation in a patient with Auerbach's plexus of pure autonomic failure.	Intern Med.	53	2261		
Miyashita A, Wen Y, Kitamura N, Matsubara E, Kawarabayashi T, Shoji M, Tomita N, Furukawa K, Arai H, Asada T, Harigaya Y, Ikeda M, Amari M, Hanyu H, Higuchi S, Nishizawa M, Suga M, Kawase Y, Akatsu H, Imagawa M, Hamaguchi T, Yamada M, Morihara T, Takeda M, Takao T, Nakata K, Sasaki K, Watanabe K, Nakashima K, Urakami K, Ooya T, Takahashi M, Yuzuriha T, Serikawa K, Yoshimoto S, Nakagawa R, Saito Y, Hatsuta H, Murayama S, Kakita A, Takahashi H, Yamaguchi H, Akazawa K, Kanazawa I, Ihara Y, Ikeuchi T, Kuwano R.	Lack of genetic association between TREM2 and late-onset Alzheimer's disease in a Japanese population.	J Alzheimers Dis.	41	1031-8		
Yasui K, Yabe I, Yoshida K, Kanai K, Arai K, Ito M, Onodera O, Koyano S, Isozaki E, Sawai S, Adachi Y, Sasaki H, Kuwabara S, Hattori T, Sobue G, Mizusawa H, Tsuji S, Nishizawa M, Nakashima K.	A 3-year cohort study of the natural history of spinocerebellar ataxia type 6 in Japan.	Orphanet J Rare Dis.	9	118		
Watanabe H, Atsuta N, Nakamura R, Hirakawa A, Watanabe H, Ito M, Senda J, Katsuno M, Izumi Y, Morita M, Tomiyama H, Taniguchi A, Aiba I, Abe K, Mizoguchi K, Oda M, Kano O, Okamoto K, Kuwabara S, Hasegawa K, Imai T, Aoki M, Tsuji S, Nakano I, Kaji R, Sobue G.	Factors affecting longitudinal functional decline and survival in amyotrophic lateral sclerosis patients.	Amyotroph Lateral Scler Frontotemporal Degener.	Epub ahead of print		2014	有
Riku Y, Watanabe H, Yoshida M, Tatsumi S, Mimuro M, Iwasaki Y, Katsuno M, Iguchi Y, Masuda M, Senda J, Ishigaki S, Udagawa T, Sobue G.	Lower motor neuron involvement in TAR DNA-binding protein of 43 kDa-related frontotemporal lobar degeneration and amyotrophic lateral sclerosis.	JAMA Neurol.	71	172-9	2014	有
Riku Y, Atsuta N, Yoshida M, Tatsumi S, Iwasaki Y, Mimuro M, Watanabe H, Ito M, Senda J, Nakamura R, Koike H, Sobue G.	Differential motor neuron involvement in progressive muscular atrophy: a comparative study with amyotrophic lateral sclerosis.	BMJ Open.	4	e005213	2014	有
Iida M, Katsuno M, Nakatsuji H, Adachi H, Kondo N, Miyazaki Y, Tohnai G, Ikenaka K, Watanabe H, Yamamoto M, Kishida K, Sobue G.	Pioglitazone suppresses neuronal and muscular degeneration caused by polyglutamine-expanded androgen receptors.	Hum Mol Genet.	Epub ahead of print		2014	有
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Tohnai G, Adachi H, Katsuno M, Doi H, Matsumoto S, Kondo N, Miyazaki Y, Iida M, Nakatsuji H, Qiang Q, Ding Y, Watanabe H, Yamamoto M, Ohtsuka K, Sobue G.	Paeoniflorin eliminates a mutant AR via NF-YA-dependent proteolysis in spinal and bulbar muscular atrophy.	Hum Mol Genet.	23	3552-65	2014	有
Yokoyama T, Ishiyama M, Hasegawa K, Uchihara T, Yagishita	Novel neuronal cytoplasmic inclusions in a patient carrying SCA8 expansion mutation.	Neuropathology	0:00	27-31	2014	無
Yokoyama T, Nakamura S, Horiuchi E, Ishiyama M, Kawashima R, Nakamura K, Hasegawa K, Yagishita S	Late onset GM2 gangliosidosis presenting with motor neuron disease: an autopsy case.	Neuropathology	34	308-313	2014	無
Kenichi Kashihara, Tomoyoshi Kondo, Yoshikuni Mizuno, Seiji Kikuchi, Sadako Kuno, Kazuko Hasegawa, Nobutaka Hattori, Hideki Mochizuki, Hideo Mori, Miho Murata, Masahiro Nomoto, Ryosuke Takahashi, Atsushi Takeda, Yoshio Tsuboi, Yoshikazu Ugawa, Mitsutoshi Yamanmoto, Fusako Yokochi, Fumihito Yoshii, Glenn T. Stebbins, Barbara C. Tilley, Sheng Luo, Lu Wang, Nancy R. LaPelle, Christopher G. Goetz, MDS-UPDRS Japanese Validation Study Group	Official Japanese Version of the International Parkinson and Movement Disorder Society-Unified Parkinson's Disease Rating Scale: Validation Against the Original English Version.	Mov Disord			2014	無
Yoshikuni Mizuno, MD, PhD; Masahiro Nomoto, MD; Kazuko Hasegawa, MD; Nobutaka Hattori, MD; Tomoyoshi Kondo, MD; Miho Murata, MD; Masahiro Takeuchi, ScD; Masayoshi Takahashi, MSc; Takayuki Tomita, MSc	Rotigotine vs ropinirole in advanced stage Parkinson's disease: a double-blind study	Parkinsonism related disorders			2014	無

英文原著・症例報告

著　者　名	論　文　題　名	雑　誌　名	巻	頁	出版西暦年	GRANTへの謝辞の有無
LV Kalia[1], AE Lang[1], L Hazrati[1], S Fujioka[2], ZK Wszolek[2], DW Dickson[2], OA Ross[2], V-M Van Deerlin[3], JQ Trojanowski[3], HI Hurtig[3], RN Alcalay[4], KS Marder[4], LN Clark[4], C Gaig[5], E Tolosa[5], J Ruiz-Martinez[6], J Marti Masso[6], I Ferrer[7], A Lopez de Munain[6], SM Goldman[8], B Schüle[8], J Langston[8], J Aasly[9], MT Giordana[10], V Bonifatill, A Puschmann[12], M Canesi[13], G Pezzoli[13], A Maues de Paula[14], K Hasegawa[15], C Duyckaerts[16], A Brice[16], C Marras[1]	Clinical correlations with Lewy body pathology in LRRK2-related Parkinson's disease.	JAMA neurol			2014	無
Kitajima Y, Tashiro Y, Suzuki N, Warita H, Kato M, Tateyama M, Ando R, Izumi R, Yamazaki M, Abe M, Sakimura K, Ito H, Urushitani M, Nagatomi R, Takahashi R, Aoki M.	Proteasome dysfunction induces muscle growth defects and protein aggregation.	J Cell Sci	127	5204-17	2014	無
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Nishiyama A, Sugeno N, Tateyama M, Nishiyama S, Kato M, Aoki M.	Postural leg tremor in X-linked spinal and bulbar muscular atrophy.	J Clin Neurosci	21	799-802	2014	無
○ Sato K, Morimoto N, Deguchi K, Ikeda Y, Matsuura T, Abe K.	Seven amyotrophic lateral sclerosis patients diagnosed only after development of respiratory failure.	J Clin Neurosci.	21(8)	1341-1343	2014	無
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○ Abe K, Itoyama Y, Sobue G, Tsuji S, Aoki M, Doyu M, Hamada C, Kondo K, Yoneoka T, Akimoto M, Yoshino H; Edaravone ALS Study Group.	Confirmatory double-blind, parallel-group, placebo-controlled study of efficacy and safety of edaravone (MCI-186) in amyotrophic lateral sclerosis patients	Amyotroph Lateral Scler Frontotemporal Degener.	15(7-8)	610-617	2014	有
Kawahara Y, Ikeda M, Deguchi K, Hishikawa N, Kono S, Omote Y, Matsuzono K, Yamashita T, Ikeda Y, Abe K.	Cognitive and affective assessments of multiple sclerosis (MS) and neuromyelitis optica (NMO) patients utilizing computerized touch panel-type screening tests.	Intern Med.	53(20	2281-2290	2014	有
Konno T, Tada M, Shiga A, Tsujino A, Eguchi H, Masuda-Suzukake M, Hasegawa M, Nishizawa M, Onodera O, Kakita A, Takahashi H.	C9ORF72 repeat-associated non-ATG-translated polypeptides are distributed independently of TDP-43 in a Japanese patient with c9ALS.	Neuropathol Appl Neurobiol.	40	783-788	2014	無
Akimoto C, Volk AE, van Blitterswijk M, Van den Broeck M, Leblond CS, Lumbroso S, Camu W, Neitzel B, Onodera O, van Rheenen W, Pinto S, Weber M, Smith B, Proven M, Talbot K, Keagle P, Chesi A, Ratti A, van der Zee J, Alstermark H, Birve A, Calini D, Nordin A, Tradowsky DC, Just W, Daoud H, Angerbauer S, DeJesus-Hernandez M, Konno T, Lloyd-Jani A, de Carvalho M, Mouzat K, Landers JE, Veldink JH, Silani V, Gitler AD, Shaw CE, Rouleau GA, van den Berg LH, Van Broeckhoven C, Rademakers R, Andersen PM, Kubisch C.	A blinded international study on the reliability of genetic testing for GGGGCC-repeat expansions in C9orf72 reveals marked differences in results among 14 laboratories.	J Med Genet.	51	419-424	2014	無
Kimura T, Jiang H, Konno T, Seto M, Iwanaga K, Tsujihata M, Satoh A, Onodera O, Kakita A, Takahashi H.	Bunina bodies in motor and non-motor neurons revisited:a pathological study of an ALS patient after long-term survival on a respirator. Neuropathology.	Neuropathology.	34	392-397	2014	有

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Mure H, Morigaki R, Koizumi H, Okita S, Kawarai T, Miyamoto R, Kaji R, Nagahiro S, Goto S.	Deep Brain Stimulation of the Thalamic Ventral Lateral Anterior Nucleus for DYT6 Dystonia.	Stereotact Funct Neurosurg.	92	393-396	2014	無
Kimura Y, Mihara M, Kawarai T, Kishima H, Sakai N, Takahashi PM and Mochizuki H.	Efficiency of deep brain stimulation in an adolescent patient with DYT11 myoclonus-dustonia.	Neurology and Clinical Neuroscience	2	57-59	2014	有
Kishore R Kumar, Katja Lohmann, Ikuo Masuho, Ryosuke Miyamoto, Andreas Ferbert, Thora Lohnau, Meike Kasten, Johann Hagenah, Norbert Brüggemann, Julia Graf, Alexander Münchau, Vladimir S Kostic, Carolyn M Sue, Aloysius R Domingo, Raymond L Rosales, Lilian V Lee, Karen Freimann, Ana Westenberger, Youhei Mukai, Toshitaka Kawarai, Ryuji Kaji, Christine Klein, Kirill A Martemyanov and Alexander Schmidt.	Mutations in GNAL: a novel cause of craniocervical dystonia.	JAMA Neurology,	71	490-494	2014	有
Morigaki R, Nakataki M, Kawarai T, Lee LV, Teleg RA, Tabuena MD, Mure H, Sako W, Pasco PM, Nagahiro S, Iga J, Ohmori T, Goto S, Kaji R.	Depression in X-linked dystonia-parkinsonism: A case-control study.	Parkinsonism Relat Disord.	19	844-846	2013	有
Goto S, Kawarai T, Morigaki R, Okita S, Koizumi H, Nagahiro S, Munoz EL, Lee LV, Kaji R.	Defects in the striatal neuropeptide Y system in X-linked dystonia-parkinsonism.	Brain	136	1555-1567	2013	有
Toshitaka Kawarai, Paul Matthew D. Pasco, Rosalia A. Teleg, Masaki Kamada, Waka Sakai, Komei Shimozono, Makoto Mizuguchi, Daisy Tabuena , Antonio Orlacchio, Yuishin Izumi, Satoshi Goto, Lillian V. Lee, Ryuji Kaji.	Application of long-range polymerase chain reaction in the diagnosis of X-linked dystonia parkinsonism.	Neurogenetics.	14	167-169	2013	有
Toshitaka Kawarai, Ai Miyashiro, Katsunobu Sugihara, Ryosuke Miyamoto, Yuishin Izumi, Hiroyuki Morino, Hirofumi Maruyama, Antonio Orlacchio, Hideshi Kawakami, and Ryuji Kaji.	Oromandibular dystonia associated with SCA36.	MovDisord.	28	558-559	2013	無
Kobayakawa Y, Sakuni K, Kajitani K, Poirier F, Kadoya T, Horie H, Kira J, Nakabeppu Y	Galactin-1 deficiency improves axonal swelling of motor neurons in SOD1G93A transgenic mice	Neuropathol Appl Neurobiol		in press		有
Kiyohito Okumiya, Taizo Wada, Michiko Fujisawa, Masayuki Ishine, Eva Garcia del Saz, Yutaka Hirata, Shigeki Kuzuhara, Yasumasa Kokubo, Harumichi Seguchi, Ryota Sakamoto, Indrajaya Manuaba, Paulina Watofall, Andreas L Rantetampang, Kozo Matsubayashi	Amyotrophic lateral sclerosis and parkinsonism in Papua, Indonesia: 2001-2012 survey results	BMJ Open 2014;4:e004353 http://bmjopen.bmj.com/content/4/4/e004353.full?keytype=ref&ijkey=p0BX815JVOYSFWs	4	e004353	2014	有または無
Kenya Nishioka, Manabu Funayama, Carles Vilarino-Guell, Kotaro Ogaki, Yuanzhe Li, Ryogen Sasaki, Yasumasa Kokubo, Shigeki Kuzuhara, Jennifer Kachergus, Stephanie Cobb, Hirohide Takahashi, Yoshikuni Mizuno, Matthew Farrer, Owen A. Ross, PhD; Nobutaka Hattori	EIF4G1 gene mutations are not a common cause of Parkinson's disease in the Japanese population	Parkinsonism and Related Disorders	20	659-661	2014	
Akihiro Shindo, Ukito Ueda, Shigeki Kuzuhara and Yasumasa Kokubo	Neutron activation analysis of scalp hair from ALS patients and residents in the Kii Peninsula, Japan	BMC Neurology http://www.biomedcentral.com/1471-2377/14/151/abstract	14	151	2014	
Tameko Kihira, Iori Sakurai, Sohei Yoshida, Ikuro Wakayama1, Koichi Takamiya, Ryo Okumura, Yuhto Iinuma, Keiko Iwai, Yoshinori Kajimoto, Yasuhiro Hiwatani, Junko Kohmoto, Kazushi Okamoto, Yasumasa Kokubo, Shigeki Kuzuhara	Neutron activation analysis of scalp hair from ALS patients and residents in the Kii Peninsula, Japan	Biological Trace Element Research		in print	2014	

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Yamamoto T, Sato H, Lai PS, Nurputra DK, Harahap NI, Morikawa S, Nishimura N, Kurashige T, Ohshita T, Nakajima H, Yamada H, Nishida Y, Toda S, Takanashi J, Takeuchi A, Tohyama Y, Kubo Y, Saito K, Takeshima Y, Matsuo M, Nishio H	Intragenic mutations in SMN1 may contribute more significantly to clinical severity than SMN2 copy numbers in some spinal muscular atrophy (SMA) patients	Brain Dev	36(10)	914-920	2014	無
Arakawa M, Arakawa R, Tatsumi S, Aoki R, Saito K, Nomoto A	A novel evaluation method of survival motor neuron protein as a biomarker of spinal muscular atrophy by imaging flow cytometry	Biochem Biophys Res Commun	453(3)	368-374	2014	無
Saito T, Nurputra DK, Harahap NI, Indra S.K.Harahap, Yamamoto H, Muneshige E, Nishizono H, Matsumura T, Fujimura H, Sakoda S, Saito K, Nishio H	A study of valproic acid for patients with spinal muscular atrophy	Neurology and Clinical Neuroscience		1-9	2014	無
Kato N, Sa'adah N, Rochmah MA, Harahap NI, Nurputra DK, Sato H, Nishimura N, Sadewa AH, Astuti I, Haryana SM, Saito T, Saito K, Nishio H, Takeuchi A	SMA Screening System Using Dried Blood Spots on Filter Paper : Application of COP-PCR to the SMN1 Deletion Test	Kobe J Med Sci			Epub ahead of print	無
Harahap NI, Takeuchi A, Yusoff S, Tominaga K, Okinaga T, Kitai Y, Takarada T, Kubo Y, Saito K, Sa'adah N, Nurputra DK, Nishimura N, Saito T, Nishio H	Trinucleotide insertion in the SMN2 promoter may not be related to the clinical phenotype of SMA	Brain Dev			Epub ahead of print	無
Hama Y, Yabe I, Wakabayashi K, Kano T, Hirotani M, Iwakura Y, Utsumi J, Sasaki H.	Level of plasma neuregulin-1 SMDF is reduced in patients with idiopathic Parkinson's disease	Neurosci Lett	(in press)		2014	有
Wakabayashi K, Mori F, Kakita A, Takahashi H, Utsumi J, Sasaki H	Analysis of microRNA from archived formalin-fixed paraffin-embedded specimens of amyotrophic lateral sclerosis	Acta Neuropathol Commun	(in press)		2014	有
Yabe I, Tanino M, Yaguchi H, Takiyama A, Cai H, Kanno H, Takahashi I, Hayashi Y, Watanabe M, Takahashi H, Hatakeyama S, Tanaka S, Sasaki H	Pathology of frontotemporal dementia with limb girdle muscular dystrophy caused by a DNAJB6 mutation.	Clin Neurol Nuerosurg	127	10-12	2014	有
Koh K, Ishiura H, Miwa M, Doi K, Yoshimura J, Mitsui J, Goto J, Morishita S, Tsuji S and Takiyama Y.	Exome sequencing shows a novel de novo mutation in ATL1.	Neurol Clin Neurosci	2	1-4	2014	無
Ishiura H, Takahashi Y, Hayashi T, Saito K, Furuya H, Watanabe M, Murata M, Suzuki M, Sugiura A, Sawai S, Shibuya K, Ueda N, Ichikawa Y, Kanazawa I, Goto J and Tsuji S.	Molecular epidemiology and clinical spectrum of hereditary spastic paraplegia in the Japanese population based on comprehensive mutational analyses.	J Hum Genet	59	163-72	2014	無
Yamada M, Tanaka M, Takagi M, Kobayashi S, Taguchi Y, Takashima S, Tanaka K, Touge T, Hatsuta H, Murayama S, Hayashi Y, Kaneko M, Ishiura H, Mitsui J, Astuta N, Sobue G, Shimozawa N, Inuzuka T, Tsuji S and Hozumi I.	Evaluation of SLC20A2 mutations that cause idiopathic basal ganglia calcification in Japan.	Neurology	82	705-12	2014	無
Yuriko Doi, Naoki Atsuta, Gen Sobue, Mitsuya Morita, Imaharu Nakano, for Research Committee of CNS Degenerative Diseases of Japan	Prevalence and Incidence of Amyotrophic Lateral Sclerosis in Japan	J Epidemiol	24(6)	494-499	2014	有

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著者名	論文題名	雑誌名	巻	頁	出版西暦年	GRANTへの謝辞の有無
Heckman MG, Elbaz A, Soto-Ortolaza AI, Serie DJ, Aasly JO, Annesi G, Auburger G, Bacon JA, Boczarska-Jedynak M, Bozi M, Brighina L, Chartier-Harlin MC, Dardiotis E, Destée A, Ferrarese C, Ferraris A, Fiske B, Gispert S, Hadjigeorgiou GM, Hattori N, Ioannidis JP, Jasinska-Myga B, Jeon BS, Kim YJ, Klein C, Kruger R, Kyratzi E, Lin CH, Lohmann K, Loriot MA, Lynch T, Mellick GD, Mutez E, Opala G, Park SS, Petrucci S, Quattrone A, Sharma M, Silburn PA, Sohn YH, Stefanis L, Tadic V, Tomiyama H, Uitti RJ, Valente EM, Vassilatis DK, Vilariño-Güell C, White LR, Wirdefeldt K, Wszolek ZK, Wu RM, Xiromerisiou G, Maraganore DM, Farrer MJ, Ross OA; Genetic Epidemiology Of Parkinson's Disease (GEO-PD) Consortium.（上記Genetic Epidemiology Of Parkinson's Disease (GEO-PD) Consortium.に参画）	The protective effect of LRRK2 p.R1398H on risk of Parkinson's disease is independent of MAPT and SNCA variants.	Neurobiol Aging.	35	266.e5-14	2014	無
Shirafuji T, Ueyama T, Yoshino K, Takahashi H, Adachi N, Ago Y, Koda K, Nashida T, Hiramatsu N, Matsuda T, Toda T, Sakai N, Saito N.	The role of Pak-interacting exchange factor-β phosphorylation at serines 340 and 583 by PKC γ in dopamine release.	J Neurosci	34:	9268-9280	2014	無
Sekiguchi T, Kanouchi T, Shibuya K, Noto Y, Yagi Y, Inaba A, Abe K, Misawa S, Orimo S, Kobayashi T, Kamata T, Nakagawa M, Kuwabara S, Mizusawa H, Yokota T.	Spreading of amyotrophic lateral sclerosis lesions-multifocal hits and local propagation?	J Neurol Neurosurg Psychiatry	85(1)	85-91	2014	無
Azuma Y, Tokuda T, Shimamura M, Kyotani A, Sasayama H, Yoshida T, Mizuta I, Mizuno T, Nakagawa M, Fujikake N, Ueyama M, Nagai Y, Yamaguchi M.	Identification of ter94, Drosophila VCP, as a strong modulator of motor neuron degeneration induced by knockdown of Caz, Drosophila FUS.	Hum Mol Genet.	23(13)	3467-380	2014	無
Shimamura M, Kyotani A, Azuma Y, Yoshida H, Binh Nguyen T, Mizuta I, Yoshida T, Mizuno T, Nakagawa M, Tokuda T, Yamaguchi M.	Genetic link between Cabeza, a Drosophila homologue of Fused in Sarcoma (FUS), and the EGFR signaling pathway.	Exp Cell Res	326(1)	36-45	2014	無
Noto YI, Shiga K, Tsuji Y, Mizuta I, Higuchi Y, Hashiguchi A, Takashima H, Nakagawa M, Mizuno T.	Nerve ultrasound depicts peripheral nerve enlargement in patients with genetically distinct Charcot-Marie-Tooth disease.	J Neurol Neurosurg Psychiatry.	on line		2014	無
Tomoaki Tujii, Win Thiri Kyaw, Hirotaka Iwaki, Noriko Nishikawa, Masahiro Nagai, Madoka Kubo, Masahiro Nomoto.	Evaluation of the effect of pregabalin on simulated driving ability using a arriving simulator in healty male volunteers.	International Journal of General Medicine	7	103-108	2014	有
Masahiro Nomoto, Yoshikuni Mizuno, Tomoyoshi Kondo, Kazuko Hasegawa, Miho Murata, Masahiro Takeuchi, Junji Ikeda, Takayuki Tomida, Nobutaka Hattori.	Ransdermal rotigotine in advanced Parkinson's desease: a randomized, double-blind, placebo-controlled trial.	J Neurol	261	1887-1893	2014	無
Hatano T, Funayama M, Kubo SI, Mata IF, Oji Y, Mori A, Zabetian CP, Waldherr SM, Yoshino H, Oyama G, Shimo Y, Fujimoto KI, Oshima H, Kunii Y, Yabe H, Mizuno Y, Hattori N.	Identification of a Japanese family with LRRK2 p.R1441G-related Parkinson's disease.	Neurobiology of Aging	35	2656.e17-23	2014	無
Shiba-Fukushima K, Arano T, Matsumoto G, Inoshita T, Yoshida S, Ishihama Y, Ryu KY, Nukina N, Hattori N, Imai Y.	Phosphorylation of Mitochondrial Polyubiquitin by PINK1 Promotes Parkin Mitochondrial Tethering.	PLoS Genet.	10	e1004861.	2014	無
Kamagata K, Tomiyama H, Hatano T, Motoi Y, Abe O, Shimoji K, Suzuki M, Hori M, Yoshida M, Hattori N, Aoki S.	A preliminary diffusional kurtosis imaging study of Parkinson disease: comparison with conventional diffusion tensor imaging.	Neuroradiology	56	251-258	2014	無
Nishioka K, Tanaka R, Shimura H, Hirano K, Hatano T, Miyakawa K, Arai H, Hattori N, Urabe T.	Quantitative evaluation of electroconvulsive therapy for Parkinson's disease with refractory psychiatric symptoms.	Jounral of Neural Transmission	121	1405-1410	2014	無

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Hatano T, Hattori N, Kawanabe T, Terayama Y, Suzuki N, Iwasaki Y, Fujioka T, On behalf of Yokukansan Parkinson's Disease Study Group.	An exploratory study of the efficacy and safety of yokukansan for neuropsychiatric symptoms in patients with Parkinson's disease.	Jounral of Neural Transmission	122	275-279	2014	無
Shimo Y, Nakajima A, Hattori N	Dopamine agonist withdrawal syndrome in a patient with restless legs syndrome without impulse control disorder or drug abuse.	Neurological Sciences	[Epub ahead of print]		2014	無
Shimo Y, Hattori N.	Underlying mechanisms of impulse control disorders and dopamine agonist syndrome in Parkinson's disease.	Journoul of neurological disorders and stroke	2	1062	2014	無
Shimo Y, Natori S, Oyama G, Nakajima M, Ishii H, Arai H, Hattori N.	Subthalamic deep brain stimulation for a Parkinson's disease patient with duplication of SNCA.	Neuromodulation	17	102-103	2014	無
Li Y, Sekine T, Funayama M, Li L, Yoshino H, Nishioka K, Tomiyama H, Hattori N	Clinicogenetic study of GBA mutations in patients with familial Parkinson's disease	Neurobiol Aging.	35	935.e3-8.	2014	無
Shen Q, Yamano K, Head BP, Kawajiri S, Cheung JT, Wang C, Cho JH, Hattori N, Youle RJ, van der Bliek AM	Mutations in Fis1 disrupt orderly disposal of defective mitochondria.	Mol Biol Cell.	25	145-59	2014	無
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	10	631-41	2014	無
Yamashita C, Tomiyama H, Funayama M, Inamizu S, Ando M, Li Y, Yoshino H, Araki T, Ichikawa T, Ehara Y, Ishikawa K, Mizusawa H, Hattori N	The evaluation of polyglutamine repeats in autosomal dominant Parkinson's disease	Neurobiol Aging.	35	1779.e17-21.	2014	無
Miyazaki H, Oyama F, Inoue R, Aosaki T, Abe T, Kiyonari H, Kino Y, Kurosawa M, Shimizu J, Ogiwara I, Yamakawa K, Koshimizu Y, Fujiyama F, Kaneko T, Shimizu H, Nagatomo K, Yamada K, Shimogori T, Hattori N, Miura M, Nukina N.	Singular localization of sodium channel β4 subunit in unmyelinated fibres and its role in the striatum.	Nat Commun.	5	5525	2014	無
Maraschi A, Ciammola A, Folci A, Sassone F, Ronzitti G, Cappelletti G, Silani V, Sato S, Hattori N, Mazzanti M, Chieregatti E, Mulle C, Passafaro M, Sassone J.	Parkin regulates kainate receptors by interacting with the GluK2 subunit.	Nat Commun.	5	5182	2014	無
Saitoh Y, Fujikake N, Okamoto Y, Popiel HA, Hatanaka Y, Ueyama M, Suzuki M, Gaumer S, Murata M, Wada K, Nagai Y.	P62 Plays a Protective Role in the Autophagic Degradation of Polyglutamine Protein Oligomers in Polyglutamine Disease Model Flies.	J Biol Chem.			2014 Dec 5 [Epub ahead of print]	無
Mizuno Y, Nomoto M, Hasegawa K, Hatttori N, Kondo T, Murata M, Takeuchi M, Takahashi M, Tomida T; on behalf of the Rotigotine Trial Group.	Rotigotine vs ropinirole in advanced stage Parkinson's disease: A double-blind study.	Parkinsonism Relat Disord.	20 (12)	1388-1393	2014	無
Kashihara K, Kondo T, Mizuno Y, Kikuchi S, Kuno S, Hasegawa K, Hattori N, Mochizuki H, Mori H, Murata M, Nomoto M, Takahashi R, Takeda A, Tsuboi Y, Ugawa Y, Yamamoto M, Yokochi F, Yoshii F, Stebbins GT, Tilley BC, Luo S, Wang L, Lapelle NR, Goetz CG, MDS-UPDRS Japanese Validation Study Group.	Official Japanese Version of the Movement Disorder Society-Unified Parkinson's Disease Rating Scale:validation against the original English version.	Mov. Disord Clin Pract(Hoboken).	1 (3)	200-212	2014	無
Mori-Yoshimura M, Hayashi YK, Yonemoto N, Nakamura H, Murata M, Takeda S, Nishino I, Kimura E.	Nationwide patient registry for GNE myopathy in Japan.	Orphanet J Rare Dis.	9	150	2014	無
Uruha A, Hayashi K.Y, Oya Y, Mori-Yoshimura 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.			2014 Sep 24 [Epub ahead of print]	無

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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. 2014 ; 24(5) : 380-386.		24 (5)	380-386	2014	無
Nomoto M, Mizuno Y, Kondo T, Hasegawa K, Murata M, Takeuchi M, Ikeda J, Tomida T, Hattori N.	Transdermal rotigotine in advanced Parkinson's disease:a randomized, double-blind, placebo-controlled trial.	J Neurol.	261(10)	1887-93	2014	無
Araki M, Matsuoka T, Miyamoto K, Kusunoki S, Okamoto T, Murata M, Miyake S Aranami T, Yamamura T.	Efficacy of the anti-IL-6 receptor antibody tocilizumab in neuromyelitis optica: a pilot study.	Neurology	82(15)	1302-1306	2014	無
Nakamura M, Matsuoka T, Chihara N, Miyake S, Sato W, Araki M, Okamoto T, Lin Y, Ogawa M, Murata M, Aranami T, Yamamura T.	Differential effects of fingolimod on B-cell populations in multiple sclerosis.	Mult Scler.	20(10)	137-80	2014	無
Ishiura H, Takahashi Y, Hayashi T, Saito K, Furuya H, Watanabe M, Murata M, Suzuki M, Sugiura A, Sawai S, Shibuya K, Ueda N, Ichikawa Y, Kanazawa I, Goto J, Tsuji S.	Molecular epidemiology and clinical spectrum of hereditary spastic paraplegia in the Japanese population based on comprehensive mutational analyses.	J Hum Genet.	59(3)	163-172	2014	無
Hasegawa, H., Liu, L., Tooyama, I., Murayama, S., Nishimura, M.	The FAM3 superfamily member ILEI ameliorates Alzheimer's disease-like pathology by destabilizing the penultimate amyloid-beta precursor.	Nat Commun	5	3917	2014	無
Hasegawa, M., Watanabe, S., Kondo, H., Akiyama, H., Mann, D.M., Saito, Y., and Murayama, S.	3R and 4R tau isoforms in paired helical filaments in Alzheimer's disease.	Acta Neuropathol	127	303-305	2014	無
Ishibashi, K., Ishiwata, K., Toyohara, J., Murayama, S. and Ishii, K.	Regional analysis of striatal and cortical amyloid deposition in patients with Alzheimer's disease.	Eur J Neurosci	40	2701-2706	2014	無
Ito, S., Takao, M., Hatsuta, H., Kanemaru, K., Arai, T., Saito, Y., Fukayama, M. and Murayama M.	Alpha-synuclein immunohistochemistry of gastrointestinal and biliary surgical specimens for diagnosis of Lewy body disease.	Int J Clin Exp Pathol	7	1714-1723	2014	無
Iwata, A., K. Nagata, Hatsuta, H. Takuma, H. Bundo, M. Iwamoto, K.,Tamaoka, A., Murayama, S., Saido, T. and Tsuji, S.	Altered CpG methylation in sporadic Alzheimer's disease is associated with APP and MAPT dysregulation.	Hum Mol Genet	23	648-656	2014	無
Matsumoto, H., R. Sengoku, Y. Saito, Y. Kakuta, Murayama, S. and I. Imafuku	Sudden death in Parkinson's disease: a retrospective autopsy study.	J Neurol Sci	343	149-152	2014	無
Miyashita, A., Y. Wen, N. Kitamura, E. Matsubara, T. Kawarabayashi, M. Shoji, N. Tomita, K. Furukawa, H. Arai, T. Asada, Y. Harigaya, M. Ikeda, M. Amari, H. Hanyu, S. Higuchi, M. Nishizawa, M. Suga, Y. Kawase, H. Akatsu, M. Imagawa, T. Hamaguchi, M. Yamada, T. Morihara, M. Takeda, T. Takao, K. Nakata, K. Sasaki, K. Watanabe, K. Nakashima, K. Urakami, T. Ooya, M. Takahashi, T. Yuzuriha, K. Serikawa, S. Yoshimoto, R. Nakagawa, Y. Saito, H. Hatsuta, Murayama, S., A. Kakita, H. Takahashi, H. Yamaguchi, K. Akazawa, I. Kanazawa, Y. Ihara, T. Ikeuchi and R. Kuwano	Lack of genetic association between TREM2 and late-onset Alzheimer's disease in a Japanese population.	J Alzheimers Dis	41	1031-1038	2014	無
Nagao, S., O. Yokota, C. Ikeda, N. Takeda, H. Ishizu, S. Kuroda, K. Sudo, S. Terada, Murayama, S and Y. Uchitomi	Argyrophilic grain disease as a neurodegenerative substrate in late-onset schizophrenia and delusional disorders.	Eur Arch Psychiatry Clin Neurosci	264	317-331	2014	無
Oikawa, N., H. Hatsuta, Murayama, S., A. Suzuki and K. Yanagisawa	Influence of APOE genotype and the presence of Alzheimer's pathology on synaptic membrane lipids of human brains.	J Neurosci Res	92	641-650	2014	無
Qina, T., N. Sanjo, M. Hizume, M. Higuma, M. Tomita, R. Atarashi, K. Satoh, I. Nozaki, T. Hamaguchi, Y. Nakamura, A. Kobayashi, T. Kitamoto, Murayama, S., H. Murai, M. Yamada and H. Mizusawa	Clinical features of genetic Creutzfeldt-Jakob disease with V180I mutation in the prion protein gene.	BMJ Open	4	e004968	2014	無

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著　者　名	論　文　題　名	雑　誌　名	巻	頁	出版西暦年	GRANTへの謝辞の有無
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Xie, C., T. Miyasaka, S. Yoshimura, H. Hatsuta, S. Yoshina, E. Kage-Nakadai, S. Mitani, Murayama, S. and Y. Ihara	The homologous carboxyl-terminal domains of microtubule-associated protein 2 and TAU induce neuronal dysfunction and have differential fates in the evolution of neurofibrillary tangles.	PLoS One	9	e89796	2014	無
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