

## 研究成果の刊行に関する一覧表

## 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
東山雄一、田中章景	本年の動向 小脳と認知機能	鈴木則宏編	Annual Review 神経	中外医学社	東京	2020	91-98
瀧山嘉久	痙性対麻痺	永井良三	今日の診断指針 第8版	医学書院	東京	2020	684-650
田中章景ら	脊髄小脳変性症	技術情報協会	疾患原因遺伝子・タンパク質の解析技術と創薬/診断技術への応用	技術情報協会	東京	2022	第6章 第5節
吉田邦広	多系統萎縮症, 特発性小脳失調症	下畑享良	脳神経内科ハンドブック	中外医学社	東京	2022	170-176
瀧山嘉久	痙性対麻痺 (HAMを含む)	福井次矢ほか	今日の治療指針	医学書院	東京	2022	990-992
佐々木征行	脊髄小脳変性症、脊髄小脳失調症		小児疾患診療のための病態生理	東京医学社	東京	2022	p370-4

## 雑誌

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Matsushima M, et al.	Multiple system atrophy in Hokkaido, Japan: a prospective registry study of natural history and symptom assessment scales followed for 5 years	BMJ Open	11(2)	e045100	2021
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Nakamura H et al.	Long-read sequencing identifies the pathogenic nucleotide repeat expansion in RFC1 in a Japanese case of CANVAS	J Hum Genet	65(5)	475-480	2020

Satoh S, Kondo Y, Ohara S, Yamaguchi T, Nakamura K, Yoshida K.	Intrafamilial phenotypic variation in spinocerebellar ataxia type 23	Cerebellum & Ataxias	7	7	2020
Yoshihisa Takiyama	Defining the clinical, molecular and imaging spectrum of adaptor protein complex 4-associated hereditary spastic paraplegia: a cross-sectional analysis of 156 patients	Brain	143(10)	2929-2944	2020
Yoshihisa Takiyama	A novel mutation in the <i>GBA2</i> gene in a Japanese patient with SPG46: a case report	eNeurologicalSci	19	100238	2020
Yoshihisa Takiyama	A Japanese SPG4 patient with a confirmed de novo mutation in the <i>SPAST</i> gene	Intern Med	59	2311-2315	2020
Yoshihisa Takiyama	<i>RFC1</i> repeat expansion in Japanese patients with late-onset cerebellar ataxia	J Hum Genet	65	1143-1147	2020
Yoshihisa Takiyama	A case of late onset Chediak-Higashi syndrome with progressive gait disturbance and cognitive dysfunction caused by novel variant in <i>LYST</i> gene	Neurology and Clinical Neuroscience	8	415-418	2020
Yoshihisa Takiyama	Identification of a novel mutation in <i>ATP13A2</i> associated with a complicated form of hereditary spastic paraplegia	Neurol Genet	6	E514	2020
Yoshihisa Takiyama	SPG9A with the new occurrence of an <i>ALDH18A1</i> mutation in a <i>CMT1A</i> family with PMP22 duplication: case report	BMC Neurol	21	64	2020
Yoshihisa Takiyama	Sympathetic nerve outflow to skin in a case with dentatorubral-pallidolusian atrophy	J Clin Neurosci	87	80-83	2020
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宮井一郎	ヒトにおける歩行と姿勢制御の脳内機構とリハビリテーション治療への適用	リハビリテーション医学 (J Rehabil Med)	10(57)	965-973	2020
平松佑一, 藤本宏明, 乙宗宏範, 島中めぐみ, 矢倉一, 宮井一郎	SCD・MSA に対するリハビリテーションの実際	難病と在宅ケア	26(3)	10-13	2020
平松佑一, 宮井一郎	運動失調の病態と臨床症状	作業療法ジャーナル	54(10)	1072-1077	2020
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Kurumada K, Sugiyama A, Hirano S, Yamamoto T, Yamanaka Y, Araki N, Yakiyama M, Yoshitake M, Kuwabara S.	Pareidolia in Parkinson's disease and multiple system atrophy	Parkinson's Disease	2021	2704755.	2021
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Kimura M et al.	Takotsubo Cardiomyopathy in Bickerstaff Brainstem Encephalitis Triggered by COVID-19	Front Neurol	12	822247	2021
Lipponen J, et al.	Molecular epidemiology of hereditary ataxia in Finland	BMC Neurol	21(1)	382	2021
Kubota S et al.	SGTA associates with intracellular aggregates in neurodegenerative diseases	Mol Brain	14(1)	59	2021

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高尾昌樹	脳表ヘモジデリン沈着症のオーバビュー	神経治療学	38(2)	80-82	2021
大平雅之	脳表ヘモジデリン沈着症の疫学	神経治療学	38(2)	83-85	2021
N Miyazawa et al.	Case of cortical superficial siderosis presenting with corticobasal syndrome	Clin Neurosci	10(2)	95-97	2022
藤本宏明, <u>宮井一郎</u>	脊髄小脳変性症のニューロリハビリテーション治療	Jpn J Rehabil Med	58(5)	536-543	2021

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Mitsui S, Otomo A, Sato K, Ishiyama M, Shimakura K, Okada-Yamaguchi C, Warabi E, Yanagawa T, <u>Aoki M</u> , Shang HF, Hadano S.	SQSTM1, a protective factor of SOD1-linked motor neuron disease, regulates the accumulation and distribution of ubiquitinated protein aggregates in neuron	Neurochem Int	158	105364	2022
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Kubota T, Hosaka T, Ando D, Ikeda K, Izumi R, Misu T, Warita H, <u>Aoki M.</u>	Spinal Cord Infarction in an Adolescent with Protein S Deficiency: A Case Report and Literature Review	Intern Med	(Epub ahead of print)		2023
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<u>青木正志</u>	難治性疾患(難病)を学ぶ 筋萎縮性側索硬化症(ALS)	遺伝子医学	12(3)	114-121	2022
<u>青木正志</u> , 高橋俊明	【遺伝性神経・筋疾患-診療と研究の最前線】ミオパチー, 筋ジストロフィーの病態・診断・治療法開発 Dysferlin遺伝子異常に伴う筋ジストロフィー Dysferlinopathy	医学のあゆみ	283(10)	983-987	2022
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井泉瑠美子, 鈴木直輝, <u>青木正志</u>	【炎症性筋疾患に関する最新の知見】封入体筋炎	臨床免疫・アレルギー科	78(4)	430-437	2022
金子仁彦, 浪岡靖弘, 大山綾音, 高井良樹, 檜森紀子, 中澤徹, 三須建郎, <u>青木正志</u>	再発後早期に eculizumab を導入した抗アクアポリン4抗体陽性視神経脊髄炎の2例	神経治療学	39(4)	731-735	2022
高井良樹, 三須建郎, 藤原一男, <u>青木正志</u>	多発性硬化症・視神経脊髄炎の治療トレンド最前線 Myelin oligodendrocyte glycoprotein 抗体関連疾患の治療 現状と課題	神経治療学	39(3)	282-288	2022
<u>青木正志</u> , 高橋俊明	【日本発の神経疾患-発見の歴史からのメッセージ】遺伝性疾患 三好型遠位型筋ジストロフィー 原因究明の歴史	Clinical Neuroscience	41(1)	119-121	2023



鈴木直輝, 割田仁, <u>青木正志</u>	【骨格筋のすべて-メカニズムからサルコペニアまで】筋症状を伴う疾患 筋萎縮性側索硬化症-骨格筋の症状と分子病態	Clinical Neuroscience	41(2)	256-259	2023
<u>青木正志</u> , 西山亜由美, 割田 仁	【ALS ーどこまでわかり, どこまで治るか】原因と発症機序 SOD1	Clinical Neuroscience	41(3)	334-337	2023
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Matsukawa T, et al.	Clinical and Genetic Features of Multiplex Families with Multiple System Atrophy and Parkinson's Disease	Cerebellum	(Online ahead of print)		2022
Higashiyama Y, Kuroki M, Kudo Y, Hamada T, Morihara K, Saito A, Miyaji Y, Kimura K, Joki H, Kishida H, Doi H, Ueda N, Takeuchi H, Johkura K, Tanaka F.	Reduced likelihood of the Poggendorff illusion in cerebellar strokes: A clinical and neuroimaging study	Brain Commun	5(2)	fcad053	2023
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