

## 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
井上 健	「メンデルの法則」「遺伝率」「集団遺伝」	一般社団法人日本人類遺伝学会	「コアカリ準拠臨床遺伝学テキスト-ゲノム医療に必要な考え方を身につける-」	診断と治療社	東京	2018	20-24
高梨潤一	主に小児にみられる拡散強調画像高信号の鑑別	青木茂樹、大場洋	頭部の鑑別診断のポイント	秀潤社	東京	2018	48-52
Kubota M.	Cockayne Syndrome: Clinical Aspects.	Nishiqori C., Sugasawa K.	DNA Repair Disorders	Springer	Singapore	2019	115-132
山本俊至	11p13欠失症候群 (WAGR症候群)		内分泌症候群(3版) IV-その他の内分泌疾患を含めて-	株日本臨床社	大阪	in press	2019

### 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
藤田瑞穂, 下山恭平, 大塚直哉, 前田泰宏, 林北見, 才津浩智, 松本直通, 高梨潤一	先天性片麻痺を呈したCOL4A1関連症候群の父子例	脳と発達	50	424-428	2018
黒田友紀子, 黒澤健司	序論: シンポジウム2 遺伝学的検査に振り回されない小児神経診療: 適応から結果解釈・説明まで	脳と発達	50	181-182	2018
黒澤健司	希少難病における診断・治療の進歩	こども医療センター医学誌	47	76-78	2018
前田憲多郎, 吉田誠克, 他	首下がりを中心としたアレキサンダー病の1例	臨床神経	58	198-201	2018
山本俊至	遺伝性腫瘍症候群とその対応	小児科診療 Up-to-Date	33	9-12	2018
山本俊至, 山本圭子	マイクロアレイ染色体検査の実際	遺伝子医学	9(1)	122-127	2019
吉田誠克	アレキサンダー病の臨床と病態	京都府立医科大学雑誌	128	1-8	2019

Akaboshi K, Yamamoto T	Interstitial deletion within 7q31.1q31.3 in a woman with mild intellectual disability and schizophrenia	Neuropsychiatric Disease and Treatment	14	1773-1778	2018
Akizawa Y, Yamamoto T, Tamura K, Kanno T, Takahashi N, Ohki T, Omori T, Tokushige K, Yamamoto M, Saito K.	A novel MLH1 mutation in a Japanese family with Lynch syndrome associated with small bowel cancer	Hum Genome Var	5	13	2018
Akutsu Y, Shirai K, Takei A, Goto Y, Aoyama T, Watanabe A, Imamura M, Enokizono T, Oto T, Horii T, Suzuki K, Hayashi M, Masumoto K, Inoue K.	A patient with peripheral demyelinating neuropathy, central dysmyelinating leukodystrophy, Waardenburg syndrome, and severe hypoganglionosis associated with a novel <i>SOX10</i> mutation.	Am J Med Genet Part A	176(5)	1195-1199	2018
Babaya N, Noso S, Hiromine Y, Ito H, Taketomo Y, Yamamoto T, Kawabata Y, Ikegami H	Early-Onset Diabetes Mellitus in a Patient With a Chromosome 13q34qter Microdeletion Including IRS2	J Endocr Soc	2(10)	1207-1213	2018
Belal H, Nakashima M, Matsumoto H, Yokochi K, Taniguchi-Ikeda M, Aoto K, Amin MB, Maruyama A, Nagase H, Mizuguchi T, Miyatake S, Miyake N, Iijima K, Nonoyama S, Matsumoto N, Saito H.	De novo variants in <i>RHOBTB2</i> , an atypical Rho GTPase gene, cause epileptic encephalopathy.	Hum Mutat	39(8)	1070-1075.	2018
Calmels N, Botta E, Jia N, Fawcett H, Nardone T, Nakazawa Y, Lanzafame M, Moriwaki S, Sugita K, Kubota M, O'Bringer C, Spitz MA, Stefanini M, Laugel V, Orioli D, Ogi T, Lehmann AR.	Functional and clinical relevance of novel mutations in a large cohort of patients with Cockayne syndrome.	J Med Genet	55	329- 343	2018
Chong PF, Saito H, Sakai Y, Imagi T, Nakamura R, Matsukura M, Matsumoto N, Kira R.	Deletions of <i>SCN2A</i> and <i>SCN3A</i> genes in a patient with West syndrome and autistic spectrum disorder.	Seizure.	60	91-93	2018
Fassio A, Esposito A, Kato M, Saito H, Mei D, Marini C, Conti V, Nakashima M, Okamoto N, Olmez Turker A, Albuz B, Semerci Gündüz CN, Yanagihara K, Belmonte E, Maragliano L, Ramsey K, Balak C, Siniard A, Narayanan V; C4RCD Research Group, Ohba C, Shiina M, Ogata K, Matsumoto N, Benfenati F, Guerrini R.	De novo mutations of the <i>ATP6V1A</i> gene cause developmental encephalopathy with epilepsy.	Brain.	1;141(6)	1703-1718.	2018

Hamada N, Ogaya S, Nakashima M, Nishijo T, Sugawara Y, Iwamoto I, Ito H, Maki Y, Shirai K, Baba S, Maruyama K, Saitsu H, Kato M, Matsumoto N, Momiyama T, Nagata KI.	<i>De novo PHACTR1</i> mutations in West syndrome and their pathophysiological effects.	Brain	1;141(11)	3098-3114.	2018
Hamanaka K, Miyatake S, Zerem A, Lev D, Blumkin L, Yokochi K, Fujita A, Imagawa E, Iwama K, Nakashima M, Mitsuhashi S, Mizuguchi T, Takata A, Miyake N, Saitsu H, van der Knaap MS, Lerman-Sagie T, Matsumoto N.	Expanding the phenotype of IBA57 mutations: related leukodystrophy can remain asymptomatic.	J Hum Genet	63(12)	1223-1229	2018
Hamilton EMC, van der Lei HDW, Vermeulen G, Gerver JAM, Lourenço CM, Naidu S, Mierzevska H, Gemke RJJ, de Vet HCW, Uitdehaag BMJ, Lissenberg-Witte BI, VWM Research Group (Matsui M et al.), van der Knaap MS.	Natural History of Vanishing White Matter.	Ann Neurol	84	274-288	2018
Hayakawa I, Kubota M.	Ictal pouting: kabuki visage or chapeau de gendarme?	Pract Neurol	18(5)	410-412	2018
Imaizumi T, Kumakura A, Yamamoto-Shimajima K, Ondo Y, Yamamoto T	Identification of a rare homozygous SZT2 variant due to uniparental disomy in a patient with a neurodevelopmental disorder	Intractable & Rare Diseases Research	7(4)	245-250	2018
Ishiyama A, Muramatsu K, Uchino S, Sakai C, Matsushima Y, Makioka N, Ogata T, Suzuki E, Komaki H, Sasaki M, Mimaki M, Goto YI, Nishino I.	<i>NDUFAF3</i> Variants that Disrupt Mitochondrial Complex I Assembly may associate with Cavitating Leukoencephalopathy.	Clin Genet	93	1103-6	2018
Kunii M, Doi H, Ishii Y, Ohba C, Tanaka K, Tada M, Fukai R, Higuchi S, Kishida H, Ueda N, Kudo Y, Kugimoto C, Nakano T, Uda N, Miyatake S, Miyake N, Saitsu H, Ito Y, Takahashi K, Nakamura H, Tomita-Katsumoto A, Takeuchi H, Koyano S, Matsumoto N, Tanaka F.	Genetic analysis of adult leukoencephalopathy patients using a custom-designed gene panel.	Clin Genet.	94(2)	232-238	2018

Matsumoto, A., Tulyeu, J., Furukawa, R., Watanabe, C., Monden, Y., Nozaki, Y., Mori, M., Namekawa, M., Jimbo, E.F., Aihara, T., Yamagata, T., Ohsaka, H.	A case of severe Alexander disease with de novo c. 239T>C, p.(F80S), in GFAP.	Brain Dev.	40(7)	587-591	2018
Miyatake S, Schneeberger S, Koyama N, Yokochi K, Ohmura K, Shiina M, Mori H, Koshimizu E, Imagawa E, Uchiyama Y, Mitsuhashi S, Frith MC, Fujita A, Satoh M, Taguri M, Tomono Y, Takahashi K, Doi H, Takeuchi H, Nakashima M, Mizuguchi T, Takata A, Miyake N, Saitsu H, Tanaka F, Ogata K, Hennet T, Matsumoto N.	Biallelic COLGALT1 variants are associated with cerebral small vessel disease.	Ann Neurol.	84(6)	843-853	2018
Mizuguchi T, Nakashima M, Kato M, Okamoto N, Kurahashi H, Ekhlévitch N, Shiina M, Nishimura G, Shibata T, Matsuo M, Ikeda T, Ogata K, Tsuchida N, Mitsuhashi S, Miyake S, Takata A, Miyake N, Hata K, Kaname T, Matsubara Y, Saitsu H, Matsumoto N.	Loss-of-function and gain-of-function mutations in PPP3CA cause two distinct disorders.	Hum Mol Genet.	15:27(8)	1421-1433.	2018
Nagai K, Maekawa T, Terashima H, Kubota M, Ishiguro A.	Severe anti-GAD antibody-associated encephalitis after stem cell transplantation.	Brain Dev.	41(3)	301-304	2018
Nakayama T, Ishii A, Yoshida T, Nasu H, Shimojima K, Yamamoto T, Kure S, Hirose S.	Somatic mosaic deletions involving SCN1A cause Dravet syndrome.	Am J Med Genet A	176	657-662	2018
Saikusa T, Hara M, Iwama K, Yuge K, Ohba C, Okada JI, Hisano T, Yamashita Y, Okamoto N, Saitsu H, Matsumoto N, Matsuishi T.	De novo HDAC8 mutation causes Rett-related disorder with distinctive facial features and multiple congenital anomalies.	Brain Dev.	40(5)	406-409	2018
Saito K, Shigetomi E, Yasuda R, Sato R, Nakano M, Tashiro K, Tanaka FK, Ikenaka K, Mikoshihara K, Mizuta I, Yoshida T, Nakagawa M, Mizuno T, Koizumi S.	Aberrant astrocyte Ca <sup>2+</sup> signals "AxCa signals" exacerbate pathological alterations in an Alexander disease model.	GLIA	66	1053-1067	2018

Sato T, Kato M, Moriyama K, Haraguchi K, Saitsu H, Matsumoto N, Moriuchi H.	A case of tubulinopathy presenting with porencephaly caused by a novel missense mutation in the TUBA1A gene.	Brain Dev.	40(9)	819-823	2018
Sato, Y Shibusaki J, Aida N, Hiiragi K, Kimura Y, Akahira-Azuma M, Yumi Enomoto Y, Tsurusaki Y, Kurosawa K.	Novel <i>COL4A1</i> mutation in a fetus with early prenatal onset of schizencephaly.	Human Genome Variation	5	4	2018
Shimada S, Hirasawa K, Takeshita A, Nakatsukasa H, Yamamoto-Shimajima K, Imaizumi T, Nagata S, Yamamoto T.	Novel compound heterozygous EPG5 mutations consisted with a missense mutation and a microduplication in the exon 1 region identified in a Japanese patient with Vici syndrome.	Am J Med Genet	176(12)	2803-2807	2018
Shimada S, Hirasawa K, Takeshita A, Nakatsukasa H, Yamamoto-Shimajima K, Imaizumi T, Nagata S, Yamamoto T	Novel compound heterozygous EPG5 mutations consisted with a missense mutation and a microduplication in the exon 1 region identified in a Japanese patient with Vici syndrome	Am J Med Genet A	176(12)	2803-2807	2018
Shimada S, Oguni H, Otani Y, Nishikawa A, Ito S, Eto K, Nakazawa T, Yamamoto-Shimajima K, Takanashi J, Nagata S, Yamamoto T	An episode of acute encephalopathy with biphasic seizures and late reduced diffusion followed by hemiplegia and intractable epilepsy observed in a patient with a novel frameshift mutation in HNRNPU	Brain Dev	40(9)	813-818	2018
Shioda N*, Yabuki Y, Yamaguchi K, Onozato M, Li Y, Kurosawa K, Tanabe H, Okamoto N, Era T, Sugiyama H, Wada T* and Fukunaga K.	Targeting G-quadruplex DNA as cognitive function therapy for ATR-X syndrome.	Nature Medicine	24	802-813	2018
Shiraku H, Nakashima M, Takeshita S, Khoo CS, Haniffa M, Ch'ng GS, Takada K, Nakajima K, Ohta M, Okanishi T, Kanai S, Fujimoto A, Saitsu H, Matsumoto N, Kato M.	PLPBP mutations cause variable phenotypes of developmental and epileptic encephalopathy.	Epilepsia Open.	1;3(4)	495-502	2018
Takeguchi R, Haginoya K, Uchiyama Y, Fujita A, Nagura M, Takeshita E, Inui T, Okubo Y, Sato R, Miyabayashi T, Togashi N, Saito T, Nakagawa E, Sugai K, Nakashima M, Saitsu H, Matsumoto N, Sasaki M.	Two Japanese cases of epileptic encephalopathy associated with an FGF12 mutation.	Brain Dev.	40(8)	728-732	2018
Tojima M, Murakami G, Hikawa R, Yamakado H, Yamashita H, Takahashi R, Matsui M.	Homozygous 31 trinucleotide repeats in the SCA2 allele are pathogenic for cerebellar ataxia.	Neurol genet,	4(6)	E283	2018

Tsuchida N, Hamada K, Shiina M, Kato M, Kobayashi Y, Tohyama J, Kimura K, Hoshino K, Ganesan V, Teik KW, Nakashima M, Mitsuhashi S, Mizuguchi T, Takata A, Miyake N, Saito H, Ogata K, Miyatake S, Matsumoto N.	GRIN2D variants in three cases of developmental and epileptic encephalopathy.	Clin Genet.	94(6)	538-547	2018
Tulyeu, J., Tamaura, M., Jimbo, E., Shimbo, H., Takano, K., Iai, M., Yamashita, S., Goto, T., Aida, N., Tokuhiko, E., Yamagata, T., Osaka, H.	Aggregate formation analysis of GFAP(R416W) found in one case of Alexander disease.	Brain Dev.	41(2)	195-200	2018
Ueda, A., Shimbo, H., Yada, Y., Koike, Y., Yamagata, T., Osaka, H.	Pelizaeus-Merzbacher disease can be a differential diagnosis in males presenting with severe neonatal respiratory distress and hypotonia.	Hum Genome Var	29:5	18013	2018
Yamamoto T, Lu Y, Nakamura R, Shimojima K, Kira R.	Novel A178P mutation in SLC16A2 in a patient with Allan-Herndon-Dudley syndrome	Congenit Anom	58	143-144	2018
Yamamoto T, Yamamoto-Shimojima K, Ueda Y, Imai K, Takahashi Y, Imagawa E, Miyake N, Matsumoto N	Independent occurrence of de novo HSPD1 and HIP1 variants in brothers with different neurological disorders - leukodystrophy and autism	Hum Genome Var	5	18	2018
Yamashita K, Seto T, Fukushima S, Fujita K, Hikita N, Yamamoto T, Shintaku H.	Evaluation of the relationship between the serum immunoglobulin G2 level and repeated infectious diseases in children.	Osaka City Med J	64	19-30	2018
Yanagishita T, Yamamoto-Shimojima K, Nakanishi S, Sasaki T, Shigematsu H, Imai K, Yamamoto T	Phenotypic features of 1q41q42 microdeletion including WDR26 and FBXO28 are clinically recognizable: The first case from Japan	Brain Dev	41(5)	452-455	2018
Yokota K, Sano S, Murofushi Y, Yoshimaru D, Takanashi J.	Neurochemistry evaluated by MR spectroscopy in a patient with xeroderma pigmentosum group A.	Brain Dev	40	931-933	2018
Hayakawa I, Kubota M.	Digital Amputation by Congenital Insensitivity to Pain with Anhidrosis.	The Journal of Pediatrics	208	290	2019
Iida A, Takeshita E, Kosugi S, Kamatani Y, Momozawa Y, Kubo M, Nakagawa E, Kurosawa K, Inoue K, Goto YI.	A novel intragenic deletion in OPHN1 in a Japanese patient with Dandy-Walker malformation.	Hum Genome Var	6	1	2019
Imaizumi T, Mogami Y, Okamoto N, Yamamoto-Shimojima K, Yamamoto T	A de novo 1p35.2 microdeletion including PUM1 identified in a patient with sporadic West syndrome	Congenit Anom	in press		2019

Imaizumi T, Yamamoto-Shimajima K, Yamamoto H, Yamamoto T	Establishment of a simple and rapid method to detect MECP2 duplications using digital polymerase chain reaction	Congenit Anom	in press		2019
Iwama K, Mizuguchi T, Takeshita E, Nakagawa E, Okazaki T, Nomura Y, Iijima Y, Kajiu I, Sugai K, Saito T, Sasaki M, Yuge K, Saikusa T, Okamoto N, Takahashi S, Amamoto M, Tomita I, Kumada S, Anzai Y, Hoshino K, Fattal-Valevski A, Shiroma N, Ohfu M, Moroto M, Tanda K, Nakagawa T, Sakakibara T, Nabatame S, Matsuo M, Yamamoto A, Yukishita S, Inoue K, Waga C, Nakamura Y, Watanabe S, Ohba C, Sengoku T, Fujita A, Mitsuhashi S, Miyatake S, Takata A, Miyake N, Ogata K, Ito S, Saito H, Matsuishi T, Goto YI, Matsumoto N.	Genetic landscape of Rett syndrome-like phenotypes revealed by whole exome sequencing.	J Med Genet.	In press		2019
Kanamori K, Terashima H, Anzai M, Ishiguro A, Kubota M.	Prolonged mild disturbance of consciousness and acute encephalopathy.	Pediatr Int.	61(2)	175-179	2019
Kumagai T, Terashima H, Uchida H, Fukuda A, Kasahara M, Kosuga M, Okuyama T, Tsunoda T, Inui A, Fujisawa T, Narita A, Eto Y, Kubota M.	A case of Niemann-Pick disease type C with neonatal liver failure initially diagnosed as neonatal hemochromatosis.	Brain Dev.	41	460-464.	2019
Matsuo M, Yamamoto T, Saito K.	Long-term natural history of an adult patient with distal 22q11.2 deletion from low copy repeat-D to E.	Congenit Anom	59(3)	102-103	2019
Nozawa A, Ozeki M, Kawasaki R, Nakama M, Iwata H, Yamamoto T, Fukao T	Identification of homozygous somatic DICER1 mutation in pleuropulmonary blastoma	J Pediat Hematol Onc	in press		2019
Okazaki-Fukui K, Kubota M, Terashima H, Ishiguro A.	Early administration of vitamins B1 and B6 and L-carnitine prevents a second attack of acute encephalopathy with biphasic seizures and late reduced diffusion: a case control study.	Brain Dev.	In press		2019
Takashima S, Saito H, Shimozawa N.	Expanding the concept of peroxisomal diseases and efficient diagnostic system in Japan.	J Hum Genet.	64(2)	145-152	2019

Tsukada I, Shimada S, Shono T, Nishizaki N, Oda H, Suzuki K, Niizuma T, Obinata K, Yamamoto T, Shimizu T	PRRT2 mutation in a sporadic case of paroxysmal kinesigenic dyskinesia	Juntendo Medical Journal	in press		2019
Yamamoto-Shimajima K, Kouwaki M, Kawashima Y, Itomi K, Momosaki K, Ozasa S, Okamoto N, Yokochi K, Yamamoto T.	Natural histories of patients with Wolf-Hirschhorn syndrome derived from variable chromosomal abnormalities.	Congenit Anom	in press		2019
Yamamoto-Shimajima K, Okamoto N, Matsumura W, Okazaki T, Yamamoto T.	Three Japanese patients with 3p13 microdeletions involving FOXP1	Brain Dev	41(3)	257-262	2019