東京大学医学部附属病院 小児科 小児神経チーム

業績

＜原著論文＞

1. 巻和佳奈、森貴幸、柿本優、竹中暁、葛西真梨子、下田木の実、佐藤敦志、岡明、佐久間啓、水口雅．多彩な高次脳機能障害をきたした重症マイコプラズマ脳炎．日本小児科学会雑誌，2020, 124(1), 55-62.
2. Suzuki S, Kita S, Morisaki M, Kikuchi R, Sato I, Iwasaki M, Otomo E, Sekiguchi H, Hirata Y, Sato A, Sugiyama M, Kamibeppu K. Nurses' perceptions regarding transitional care for adolescents and young adults with childhood-onset chronic diseases. Jpn J Nurs Sci. 2020 Jan 13: e12323.
3. Shibata A, Kasai M, Hoshino A, Miyagawa T, Matsumoto H, Yamanaka G, Kikuchi K, Kuki I, Kumakura A, Hara S, Shiihara T, Yamazaki S, Ohta M, Yamagata T, Takanashi JI, Kubota M, Oka A, Mizuguchi M. Thermolabile polymorphism of carnitine palmitoyltransferase 2: A genetic risk factor of overall acute encephalopathy. Brain Dev. 2019 Nov; 41(10): 862-869.
4. 奥野安由，山中純子，吉本優里，大熊喜彰，兼重昌夫，田中瑞恵，瓜生英子，水上愛弓，五石圭司，佐藤典子，佐藤敦志，七野浩之．大腸菌性脳室炎の 2 新生児例．日本小児科学会雑誌、2019, 123(6), 1023-1031.
5. 岩﨑美和、佐藤敦志、中村真由美、鈴木征吾、小林明日香、キタ幸子、佐藤伊織、上別府圭子、平田陽一郎．当院における移行期支援外来の取り組みと課題～他疾患と比較した循環器疾患患者の特徴に焦点を当てて～．日本成人先天性心疾患学会雑誌，2019, 8(2), 33-41.
6. Uchino S, Iida A, Sato A, Ishikawa K, Mimaki M, Nishino I, Goto YI. A novel compound heterozygous variant of ECHS1 identified in a Japanese patient with Leigh syndrome. Hum Genome Var. 2019 Apr 19; 6: 19.
7. Nguyen TQN, Doan NMT, Trinh HT, Mizuguchi M. Novel mutation in EFCAB7 alters expression and interaction of Ellis-van Creveld ciliary proteins. Congenit Anom (Kyoto). 2019 Mar; 59(2): 49-50.
8. Kotajima-Murakami H, Kobayashi T, Kashii H, Sato A, Hagino Y, Tanaka M, Nishito Y, Takamatsu Y, Uchino S, Ikeda K. Effects of rapamycin on social interaction deficits and gene expression in mice exposed to valproic acid in utero. Mol Brain. 2019 Jan 8; 12(1): 3.
9. Okubo M, Iida A, Hayashi S, Mori-Yoshimura M, Oya Y, Watanabe A, Arahata H, El Sherif R, Noguchi S, Nishino I. Three novel recessive DYSF mutations identified in three patients with muscular dystrophy, limb-girdle, type 2B. J Neurol Sci. 2018 Dec 15; 395:169-171.

10. Suganuma E, Oka A, Sakata H, Adachi N, Asanuma S, Oguma E, Yamaguchi A, Furuichi M, Uejima Y, Sato S, Takano T, Kawano Y, Tanaka R, Arai T, Oh-Ishi T. 10-year follow-up of congenital cytomegalovirus infection complicated with severe neurological findings in infancy: a case report. BMC Pediatr. 2018 Nov 23; 18(1):369.
11. Tanaka M, Sato A, Kasai S, Hagino Y, Kotajima-Murakami H, Kashii H, Takamatsu Y, Nishito Y, Inagaki M, Mizuguchi M, Hall FS, Uhl GR, Murphy D, Sora I, Ikeda K. Brain hyperserotonemia causes autism-relevant social deficits in mice. Mol Autism. 2018 Nov 26; 9: 60.
12. 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. NDUFAF3 variants that disrupt mitochondrial complex I assembly may associate with cavitating leukoencephalopathy. Clin Genet. 2018 May; 93(5):1103-1106.
13. Nakagama Y, Inuzuka R, Ichimura K, Hinata M, Takehara H, Takeda N, Kakiuchi S, Shiraga K, Asakai H, Shindo T, Hirata Y, Saitoh M, Oka A. Accelerated Cardiomyocyte Proliferation in the Heart of a Neonate with LEOPARD Syndrome-Associated Fatal Cardiomyopathy. Circ Heart Fail. 2018 Apr; 11(4): e004660.
14. Nakashima M, Kato M, Aoto K, Shiina M, Belal H, Mukaida S, Kumada S, Sato A, Zerem A, Lerman-Sagie T, Lev D, Leong HY, Tsurusaki Y, Mizuguchi T, Miyatake S, Miyake N, Ogata K, Saitsu H, Matsumoto. De Novo Hotspot Variants in CYFIP2 Cause Early-Onset Epileptic Encephalopathy. Ann Neurol. 2018 Apr; 83(4): 794-806.
15. Koyano S, Morioka I, Oka A, Moriuchi H, Asano K, Ito Y, Yoshikawa T, Yamada H, Suzutani T, Inoue N; Japanese Congenital Cytomegalovirus Study Group. Congenital cytomegalovirus in Japan: More than 2 year follow up of infected newborns. Pediatr Int. 2018 Jan; 60(1):57-62.
16. Fujii T, Oka A, Morioka I, Moriuchi H, Koyano S, Yamada H, Saito S, Sameshima H, Nagamatsu T, Tsuchida S, Inoue N; Japanese Congenital Cytomegalovirus Study Group. Newborn Congenital Cytomegalovirus Screening Based on Clinical Manifestations and Evaluation of DNA-based Assays for In Vitro Diagnostics. Pediatr Infect Dis J. 2017 Oct; 36(10):942-946.
17. Kuroda Y, Mizuno Y, Mimaki M, Oka A, Sato Y, Ogawa S, Kurosawa K. Two patients with 19p13.2 deletion (Malan syndrome) involving NFIX and CACNA1A with overgrowth, developmental delay, and epilepsy. Clin Dysmorphol. 2017 Oct; 26(4):224-227.
18. Okubo M, Goto K, Komaki H, Nakamura H, Mori-Yoshimura M, Hayashi YK, Mitsuhashi S, Noguchi S, Kimura E, Nishino I. Comprehensive analysis for genetic diagnosis of Dystrophinopathies in Japan. Orphanet J Rare Dis. 2017 Aug 31; 12(1):149.
19. Shimoda K, Mimaki M, Fujino S, Takeuchi M, Hino R, Uozaki H, Hayashi M, Oka A,

Mizuguchi M. Brain edema with clasmatodendrosis complicating ataxia telangiectasia. Brain Dev. 2017 Aug;39(7):629-632.

20. Hori I, Otomo T, Nakashima M, Miya F, Negishi Y, Shiraishi H, Nonoda Y, Magara S, Tohyama J, Okamoto N, Kumagai T, Shimoda K, Yukitake Y, Kajikawa D, Morio T, Hattori A, Nakagawa M, Ando N, Nishino I, Kato M, Tsunoda T, Saitsu H, Kanemura Y, Yamasaki M, Kosaki K, Matsumoto N, Yoshimori T, Saitoh S Defects in autophagosome-lysosome fusion underlie Vici syndrome, a neurodevelopmental disorder with multisystem involvement. Sci Rep. 2017 Jun 14; 7(1):3552.
21. Ohta S, Isojima T, Mizuno Y, Kato M, Mimaki M, Seki M, Sato Y, Ogawa S, Takita J, Kitanaka S, Oka A. Partial monosomy of 10p and duplication of anothr chromosome in two patients. Pediatr Int. 2017 Jan; 59(1):99-102.
22. Suzuki T, Miyake N, Tsurusaki Y, Okamoto N, Alkindy A, Inaba A, Sato M, Ito S, Muramatsu K, Kimura S, Ieda D, Saitoh S, Hiyane M, Suzumura H, Yagyu K, Shiraishi H, Nakajima M, Fueki N, Habata Y, Ueda Y, Komatsu Y, Yan K, Shimoda K, Shitara Y, Mizuno S, Ichinomiya K, Sameshima K, Tsuyusaki Y, Kurosawa K, Sakai Y, Haginoya K, Kobayashi Y, Yoshizawa C, Hisano M, Nakashima M, Saitsu H, Takeda S, Matsumoto N. Molecular genetic analysis of 30 families with Joubert syndrome. Clin Genet. 2016 Dec; 90(6):526-535.
23. Nishimura N, Higuchi Y, Kimura N, Nozaki F, Kumada T, Hoshino A, Saitoh M, Mizuguchi M. Familial acute necrotizing encephalopathy without RANBP2 mutation: Poor outcome. Pediatr Int. 2016 Nov; 58(11):1215-1218.
24. Yamaguchi Y, Torisu H, Kira R, Ishizaki Y, Sakai Y, Sanefuji M, Ichiyama T, Oka A, Kishi T, Kimura S, Kubota M, Takanashi J, Takahashi Y, Tamai H, Natsume J, Hamano S, Hirabayashi S, Maegaki Y, Mizuguchi M, Minagawa K, Yoshikawa H, Kira J, Kusunoki S, Hara T. A nationwide survey of pediatric acquired demyelinating syndromes in Japan. Neurology. 2016 Nov 8; 87(19):2006-2015.
25. Hoshino A, Saitoh M, Miyagawa T, Kubota M, Takanashi JI, Miyamoto A, Tokunaga K, Oka A, Mizuguchi M. Specific HLA genotypes confer susceptibility to acute necrotizing encephalopathy. Genes Immun. 2016 Sep;17(6):367-369.
26. Sato A. mTOR, a potential target to treat autism spectrum disorder. CNS Neurol Disord Drug Targets. 2016 Jun; 15(5): 533-543.
27. Kono Y, Oka A, Tada H, Itabashi K, Matsui E, Nakamura Y. Perinatal dioxin exposure and psychosocial and behavioral development in school-aged children. Early Hum Dev. 2015 Sep; 91(9): 499-503.
28. 柴田 明子, 山本 真梨子, 渡邊 優, 寺嶋 宙, 柏井 洋文, 久保田 雅也, 師田 信人．Baclofen 持続髄注療法の機能不全の診断に RI シンチグラフィが役立った脳室

腹腔シャントを有する男児例．脳と発達，2015, 47(5), 367-371.
29. Taniguchi R, Koyano S, Suzutani T, Goishi K, Ito Y, Morioka I, Nakamura H, Yamada H, Oka A, Inoue N. A Thr72Ala polymorphism in the NKG2D gene is associated with early symptomatic congenital cytomegalovirus disease. Infection. 2015 Jun; 43(3):353-359.
30. Takamatsu Y, Hagino Y, Sato A, Takahashi T, Nagasawa SY, Kubo Y, Mizuguchi M, Uhl GR, Sora I, Ikeda K. Improvement of learning and increase in dopamine level in the frontal cortex by methylphenidate in mice lacking dopamine transporter. Curr Mol Med, 2015 Mar; 15(3): 245-252.
31. Yui K, Sato A, Imataka G. Mitochondrial dysfunction and its relationship with mTOR signaling and oxidative damage in autism spectrum disorders. Mini Rev Med Chem, 2015; 15(5): 373-389.
32. 古田島（村上）浩子，佐藤敦志，池田和隆．自閉症スペクトラム障害の病態解明と治療薬開発を目指して　自閉症スペクトラム障害の分子薬理学的研究．日本薬理学会雑誌，2015, 145(4), 193-200.
33. 佐藤敦志，高松幸雄，笠井慎也，小林敏之，樋野興夫，池田和隆，水口雅．Tsc2 haploinsufficiency is associated with more severe autism-related behavioral deficits in mouse models of tuberous sclerosis complex.　日本神経精神薬理学会雑誌，2015, 35(2) 51-52.
34. Hoshino H, Kubota M. Canavan disease: clinical features and recent advances in research. Pediatr Int. 2014 Aug; 56(4):477-483.
35. Nakashima M, Kashii H, Murakami Y, Kato M, Tsurusaki Y, Miyake N, Kubota M, Kinoshita T, Saitsu H, Matsumoto N. Novel compound heterozygous PIGT mutations caused multiple congenital anomalies-hypotonia-seizures syndrome 3. Neurogenetics. 2014 Aug; 15(3): 193-200.
36. Akamatsu T, Dai H, Mizuguchi M, Goto Y, Oka A, Itoh M. LOX-1 is a novel therapeutic target in neonatal hypoxic-ischemic encephalopathy. Am J Pathol. 2014 Jun; 184(6): 1843-1852.
37. 佐藤敦志，笠井慎也，小林敏之，高松幸雄，樋野興夫，池田和隆，水口雅．結節性硬化症モデルマウスの自閉症様行動における mTOR シグナル系の関与．日本神経精神薬理学会雑誌，2014, 34(2), 51-52.
38. Kataoka M, Aimi Y, Yanagisawa R, Ono M, Oka A, Fukuda K, Yoshino H, Satoh T, Gamou S. Alu-mediated nonallelic homologous and nonhomologous recombination in the BMPR2 gene in heritable pulmonary arterial hypertension. Genet Med. 2013 Dec; 15(12): 941-947.
39. Taniguchi R, Koyano S, Suzutani T, Goishi K, Ito Y, Morioka I, Oka A, Nakamura H, Yamada H, Igarashi T, Inoue N. Polymorphisms in TLR-2 are associated with congenital cytomegalovirus (CMV) infection but not with congenital CMV disease. Int J Infect Dis.

2013 Dec; 17(12): e1092-1097.

40. Miyake N, Koshimizu E, Okamoto N, Mizuno S, Ogata T, Nagai T, Kosho T, Ohashi H, Kato M, Sasaki G, Mabe H, Watanabe Y, Yoshino M, Matsuishi T, Takanashi J, Shotelersuk V, Tekin M, Ochi N, Kubota M, Ito N, Ihara K, Hara T, Tonoki H, Ohta T, Saito K, Matsuo M, Urano M, Enokizono T, Sato A, Tanaka H, Ogawa A, Fujita T, Hiraki Y, Kitanaka S, Matsubara Y, Makita T, Taguri M, Nakashima M, Tsurusaki Y, Saitsu H, Yoshiura K, Matsumoto N, Niikawa N. MLL2 and KDM6A mutations in patients with Kabuki syndrome. Am J Med Genet A. 2013 Sep; 161(9): 2234-2243.
41. Hachiya Y, Miyata R, Tanuma N, Hongou K, Tanaka K, Shimoda K, Kanda S, Hoshino A, Hanafusa Y, Kumada S, Kurihara E, Hayashi M. Autoimmune neurological disorders associated with group-A beta-hemolytic streptococcal infection. Brain Dev. 2013 Aug; 35(7): 670-674.
42. Saitsu H, Nishimura T, Muramatsu K, Kodera H, Kumada S, Sugai K, Kasai-Yoshida E, Sawaura N, Nishida H, Hoshino A, Ryujin F, Yoshioka S, Nishiyama K, Kondo Y, Tsurusaki Y, Nakashima M, Miyake N, Arakawa H, Kato M, Mizushima N, Matsumoto N. De novo mutations in the autophagy gene WDR45 cause static encephalopathy of childhood with neurodegeneration in adulthood. Nat Genet. 2013 Apr; 45(4): 445-449, 449e1.
43. Shinohara M, Saitoh M, Nishizawa D, Ikeda K, Hirose S, Takanashi JI, Takita J, Kikuchi K, Kubota M, Yamanaka G, Shiihara T, Kumakura A, Kikuchi M, Toyoshima M, Goto T, Yamanouchi H, Mizuguchi M. ADORA2A polymorphism predisposes children to encephalopathy with febrile status epilepticus. Neurology. 2013 Apr 23; 80(17): 1571-1576.
44. Tumurkhuu M, Saitoh M, Takita J, Mizuno Y, Mizuguchi M. A novel SOS1 mutation in Costello/CFC syndrome affects signaling in both RAS and PI3K pathways. J Recept Signal Transduct Res. 2013 Apr; 33(2): 124-128.
45. Kakiuchi S, Nonoyama S, Wakamatsu H, Kogawa K, Wang L, Kinoshita-Yamaguchi H, Takayama-Ito M, Lim CK, Inoue N, Mizuguchi M, Igarashi T, Saijo M. Neonatal herpes encephalitis caused by a virologically confirmed acyclovir-resistant herpes simplex virus 1 strain. J Clin Microbiol. 2013 Jan; 51(1): 356-359.
46. Sato A, Mizuguchi M, Ikeda K. Social interaction test: a sensitive method for examining autism-related behavioral deficits. Protocol Exchange. doi:10.1038/protex.2013.046.
47. Sato A, Kasai S, Kobayashi T, Takamatsu Y, Hino O, Ikeda K, Mizuguchi M. Rapamycin reverses impaired social interaction in mouse models of tuberous sclerosis complex. Nat Commun. 2012; 3: 1292.
48. 黒田友紀子，三牧正和，寺島宙，佐藤敦志，高橋寛，狩野博嗣，岡明，水口雅,

五十嵐隆．Guillain-Barré 症候群を合併した川崎病の１例．日本小児科学会雑誌, 2012, 116(6), 985-989.

49. Mimaki M, Wang X, McKenzie M, Thorburn DR, Ryan MT. Understanding mitochondrial complex I assembly in health and disease. Biochim Biophys Acta. 2012 Jun; 1817(6): 851-862.
50. Saitoh M, Shinohara M, Hoshino H, Kubota M, Amemiya K, Takanashi JL, Hwang SK, Hirose S, Mizuguchi M. Mutations of the SCN1A gene in acute encephalopathy. Epilepsia. 2012 Mar; 53(3): 558-564.
51. Tucker EJ, Mimaki M, Compton AG, McKenzie M, Ryan MT, Thorburn DR. Next-generation sequencing in molecular diagnosis: NUBPL mutations highlight the challenges of variant detection and interpretation. Hum Mutat. 2012 Feb; 33(2): 411-418.
52. Tonoki H, Harada N, Shimokawa O, Yosozumi A, Monzaki K, Satoh K, Kosaki R, Sato A, Matsumoto N, Iizuka S. Axenfeld–Rieger Anomaly and Axenfeld–Rieger Syndrome: Clinical, Molecular-Cytogenetic, and DNA Array Analyses of Three Patients with Chromosomal Defects at 6p25. Am J Med Genet A. 2011 Dec; 155A (12): 2925-2932.
53. Shinohara M, Saitoh M, Takanashi J, Yamanouchi H, Kubota M, Goto T, Kikuchi M, Shiihara T, Yamanaka G, Mizuguchi M. Carnitine palmitoyl transferase II polymorphism is associated with multiple syndromes of acute encephalopathy with various infectious diseases. Brain Dev. 2011 Jun; 33(6): 512-517.
54. Mizuno Y, Takahashi K, Igarashi T, Saito M, Mizuguchi M. Congenital infection-like syndrome with intracranial calcification. Brain Dev. 2011 Jun; 33(6): 530-533.
55. Takahashi K, Oka A, Mizuguchi M, Saitoh M, Takita J, Sato A, Mimaki M, Kato M, Ogawa S, Igarashi T. Interstitial deletion of 13q14.13-q32.3 presenting with Arima syndrome and bilateral retinoblastoma. Brain Dev. 2011 Apr; 33(4): 353-356.
56. Tumurkhuu M, Saitoh M, Sato A, Takahashi K, Mimaki M, Takita J, Takeshita K, Hama T, Oka A, Mizuguchi M. Comprehensive genetic analysis of overlapping syndromes of RAS/RAF/MEK/ERK pathway. Pediatr Int. 2010 Aug; 52(4): 557-562.
57. Mizuno Y, Tsuchida S, Kakiuchi S, Ishiguro A, Goishi K, Kamei Y, Kanamori Y, Yamazaki Y, Sekine T, Igarashi T. Case report: prenatal intervention for severe anterior urethral valve. Pediatr Int. 2010 Apr; 52(2): e92-95.

＜受賞歴＞

2018 年

- 大久保真理子：Elsevier WMS Membership Award
  Nonsense mutation induced exon skipping in Becker muscular dystrophy.
- 佐藤敦志：第 28 回日本臨床精神神経薬理学会　第 48 回日本神経精神薬理学会合同年会　優秀プレゼンテーション賞
  mTOR 阻害剤を使用中の結節性硬化症患者における自閉症症状の変化.

2017 年

- 柏井洋文：JSNP Excellent Presentation Award for AsCNP 2017
  Rapamycin treatment of impaired social behavior in adolescent Tsc2+/- mice.
- 柏井洋文：第 47 回日本神経精神薬理学会学生優秀発表賞
  Tsc2 ヘテロ欠損マウスの自閉症様行動に対する rapamycin の継続投与の効果とその副作用の検討

2015 年

- Sato Atsushi: Best Poster Award in 13th AOCCN
  Tsc2 haploinsufficiency causes more severe autistic-like behavioral deficit than Tsc1 in mice.

2014 年

- Sato Atsushi: JSNP Excellent Presentation Award for CINP 2014 Vancouver (June 22-26)
  Tsc2 happloinsufficiency is associated with the more severe autism-related behavioral deficits in mouse models of tuberous sclerosis complex.

2013 年

- Sato Atsushi: JSNP Excellent Presentation Award for AsCNP 2013 Beijing (Sep 11-14)
  Causal role of unregulated mTOR signaling in autism-related behavioral deficits in mouse models of tuberous sclerosis complex.
- Sato Atsushi: Best Poster Award of AsCNP 2013
  Causal role of unregulated mTOR signaling in autism-related behavioral deficits in mouse models of tuberous sclerosis complex.
- 星野愛：第 55 回日本小児神経学会学術集会　優秀論文賞

2012 年

- 三牧正和：2012 JSCN Award for Excellent Abstract

- 石井礼花：平成 24 年度日本医師会医学研究奨励賞
  思春期注意欠如多動性障害へのペアレントトレーニングは愛着の神経基盤を変化させるか

2011 年

- Sato Atsushi: The 2nd Congress of Asian College of Neuropsychopharmacology, Young Investigator Fellowship Award
  Autism-like Behaviors in Mouse Models of Tuberous Sclerosis Complex and Their Recovery by Rapamycin.
- Sato Atsushi: The 2nd Congress of Asian College of Neuropsychopharmacology, New Research Award
  Autism-like Behaviors in Mouse Models of Tuberous Sclerosis Complex and Their Recovery by Rapamycin.