（報告様式４）

【16ek0109040h0003】

平成 29 年 5 月 31 日

平 成 28 年 度　委 託 研 究 開 発 成 果 報 告 書

I. 基本情報

事　業　名：（日本語）難治性疾患実用化研究事業
（英　語）Practical Research Project for Rare / Intractable Diseases

研究開発課題名：（日本語）ゲノム構造異常によって発症した自閉症・発達障害の疾患特異的 iPS 細胞を用いた病態解明と治療法開発
（英　語）Elucidation of pathology and development of therapeutic method using disease-specific iPS cells derived from patients with developmental disorders caused by abnormal genomic structure

研究開発担当者　（日本語）東京女子医科大学・准教授・山本俊至
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実 施 期 間：平成 26 年　4 月　1 日　～　平成 29 年　3 月　31 日

II. 成果の概要（総括研究報告）

・ 研究開発代表者による報告の場合

山本俊至准教授（東京女子医科大学）、山形崇倫教授（自治医科大学）、岡本伸彦部長（大阪母子医療センター）らのグループにより、自閉症スペクトラムを含む発達障害患者の発症においては、多様なゲノム構造異常や疾患遺伝子変異が関わっていることを明らかにした。それらの遺伝子は、マウス脳において機能不全にさせると遊走障害を来すなどの病態を示すことを永田浩一部長（愛知県コロニー発達障害研究所）らのグループが明らかにした。発達障害の原因となるゲノム変異が明らかになった患者から、齋藤潤准教授（京都大学 iPS 細胞研究所）らや北畠康司助教（大阪大学）らが iPS 細胞を樹立し、病態解析に供した。疾患 iPS 細胞から分化誘導した神経細胞では、シナプス形成が不良であることを、山本俊至准教授（東京女子医科大学）、永田浩一部長（愛知県コロニー発達障害研究所）、小坂仁教授（自治医科大学）らのグループが明らかにし、矢田俊彦教授（自治医科大学）らのグループはその所見をパッチクランプによる電気生理学的に証明した。

The group of Associate Professor Toshiyuki Yamamoto (Tokyo Women's Medical University), Professor Takanori Yamagata (Jichi Medical School) and Department Manager Nobuhiko Okamoto (Osaka Women's and Children's Medical Center) revealed various genomic structural abnormalities and disease gene mutations in patients with developmental disorders including autism spectrum disorder. The group of Department Manager Koichi Nagata (Aichi Prefectural Colony Developmental Disorders Research Institute) clarified that those genes show pathological conditions such as migration abnormality when those were knocked-down in the mouse brain. By the group consisted with Associate Professor Megumu Saito (iPS Cell Research Institute, Kyoto University) and Assistant Professor Koji Kitahata (Osaka University) established iPS cells from patients with genomic abnormalities, and used them for pathological analysis. In the neurons differentiated from disease-specific iPS cells, synapse dysfunction was confirmed by Associate Professor Toshiyuki Yamamoto (Tokyo Women's Medical University), Koichi Nagata (Aichi Prefectural Colony Developmental Disorders Research Institute), and Professor Hitoshi Osaka (Jichi Medical School). The group of Professor Toshihiko Yada (Jichi Medical School) revealed synaptic dysfunction by use of electrophysiological analysis using patch clamp.

III. 成果の外部への発表

（1）学会誌・雑誌等における論文一覧（国内誌　4 件、国際誌　82 件）

山本俊至

1. Shimojima K, Okamoto N, Yamamoto T. A 10q21.3q22.2 microdeletion identified in a patient with severe developmental delay and multiple congenital anomalies including congenital heart defects. Congenit Anom. 2017 (in press)
2. Okamoto N, Shimojima K, Yamamoto T. Neurological Manifestations of 2q31 Microdeletion Syndrome. Congenit Anom. 2017 (in press)

3. Shimojima K, Okamoto N, Yamamoto T. Possible genes responsible for developmental delay observed in patients with rare 2q23q24 microdeletion syndrome: literature review and description of an additional patient. Congenit Anom. 2017 (in press)
4. Alber M, Kalscheuer VM, Marco E, Sherr EH, Lesca G, Till M, Gradek G, Wiesener A, Korenke CG, Mecier S, Becker F, Yamamoto T, Scherer SW, Marshall C, Walker S, Dutta U, Dalal A, Suckow V, Jamali P, Kahrizi K, Najmabadi H, Minassian BA. The ARHGEF9 Disease: Phenotype Clarification and Genotype-Phenotype Correlation. Neurol Genet 2017 (in press)
5. Iwama K, Mizuguchi T, Takanashi J, Shibayama H, Shichiji M, Ito S, Oguni H, Yamamoto T, Sekine A, Nagamine S, Ikeda Y, Nishida H, Kumada S, Yoshida T, Awaya T, Tanaka R, Chikuchi R, Niwa H, Oka Y, Miyatake S, Nakashima M, Takata A, Miyake N, Ito S, Saitsu H, Matsumoto N. Identification of novel SNORD118 mutations in seven patients with leukoencephalopathy with brain calcifications and cysts. Clin Genet 2017 (in press).
6. Lu YP, Chong P-F, Kira R, Seto T, Ondo Y, Shimojima K, Yamamoto T. Mutations in NSD1 and NFIX in three patients with clinical features of Sotos syndrome and Malan syndrome. J Pediatr Genet. 2017 (in press).
7. Yamamoto T, Shimojima K, Ondo Y, Shimakawa S, Okamoto N. MED13L haploinsufficiency syndrome: a de novo frameshift and recurrent intragenic deletions due to parental mosaicism. Am J Med genet A. 2017, 173, 1264-9.
8. Shirai K, Higashi Y, Shimojima K, Yamamoto T. An Xq22.1q22.2 nullisomy in a male patient with severe neurological impairment. Am J Med Genet A 2007, 173A, 1124-27.
9. Sangu N, Shimojima K, Takahashi Y, Ohashi T, Tohyama J, Yamamoto T. A 7q31.33q32.1 microdeletion including LRRC4 and GRM8 is associated with severe intellectual disability and characteristics of autism. Hum Genome Var. 2017, 4, 17001.
10. Murakoshi M, Takasawa K, Nishioka M, Asakawa M, Kashimada K, Yoshimoto T, Yamamoto T, Takekoshi K, Ogawa Y, Shimohira M. Abdominal paraganglioma in a young woman with 1p36 deletion syndrome. Am J Med Genet A 2017, 173A, 495-500.
11. Matsuo M, Yamauchi A, Ito Y, Sakauchi M, Yamamoto T, Okamoto N, Tsurusaki Y, Miyake N, Matsumoto N, Saito K. Mandibulofacial dysostosis with microcephaly: A case presenting with seizures. Brain Dev 2017, 39, 177-81.
12. Shimojima K, Ondo Y, Matsufuji M, Sano N, Tsuru H, Oyoshi T, Higa N, Tokimura H, Arita K, Yamamoto T. Concurrent occurrence of an inherited 16p13.11 microduplication and a de novo 19p13.3 microdeletion involving MAP2K2 in a patient with developmental delay, distinctive facial features, and lambdoid synostosis. Eur J Med Genet 2016, 59, 559-63.
13. Yamamoto T, Shimojima K, Matsufuji M, Mashima R, Sakai E, Okuyama T. Aspartylglucosaminuria caused by a novel homozygous mutation in the AGA gene was identified by an exome-first approach in a patient from Japan. Brain Dev 2017, 39, 422–5.
14. Lu Y, Shimojima K, Sakuma T, Nakaoka S, Yamamoto T. A novel PLP1 mutation F240L identified in a patient with the connatal type of Pelizaeus-Merzbacher disease. Hum Genome Var. 2017, 4, 16044.

15. Baba S, Sugawara Y, Moriyama K, Inaji M, Maehara T, Yamamoto T, Morio T. Amelioration of intractable epilepsy by adjunct vagus nerve stimulation therapy in a girl with a CDKL5 mutation. Brain Dev. 2017, 39, 341-4.
16. Seto T, Yamamoto T, Shimojima K, Shintaku H. A novel COL1A1 mutation in a family with osteogenesis imperfecta associated with phenotypic variabilities. Hum Genome Var. 2017, 4, 17007.
17. Kobayashi Y, Hanaoka Y, Akiayma T, Ohmori I, Ouchida M, Yamamoto T, Oka M, Yoshinaga H, Kobayashi K. A case of Dravet syndrome with cortical myoclonus indicated by jerk-locked back-averaging of electroencephalogram data. Brain Dev. 2017, 39, 75-9.
18. 四家達彦, 高橋幸利, 木村暢佑, 今井克美, 山下行雄, 山本俊至, 高橋孝雄. 治療戦略の変更により ADL を改善し得た CDKL5 異常症による難治性てんかんの女児例. 脳と発達 2017, 49, 28-31.
19. Shimojima K, Ondo Y, Matsufuji M, Sano N, Tsuru H, Oyoshi T, Higa N, Tokimura H, Arita K, Yamamoto T. Concurrent occurrence of an inherited 16p13.11 microduplication and a de novo 19p13.3 microdeletion involving MAP2K2 in a patient with developmental delay, distinctive facial features, and lambdoid synostosis. Eur J Med Genet. 2016, 59, 559-63.
20. Yamamoto T, Shimojima K, Yamazaki S, Ikeno K, Tohyama J. A 16q12.2q21 deletion identified in a patient with developmental delay, epilepsy, short stature, and distinctive features. Congenit Anom (Kyoto) 2016, 56, 253-5.
21. Hamatani M, Jingami N, Tsurusaki Y, Shimada S, Shimojima K, Asada-Utsugi M, Yoshinaga K, Uemura N, Yamashita H, Uemura K, Takahashi R, Matsumoto N, Yamamoto T. The first Japanese case of leukodystrophy with ovarian failure arising from novel compound heterozygous AARS2 mutations. J Hum Genet 2016, 61, 899-902.
22. Shimojima K, Narai S, Togawa M, Doumoto T, Sangu N, Vanakkere OM, De Paepee A, Edwards M, Whitehall J, Brescianini S, Petit F, Andrieux J, Yamamoto T. 7p22.1 microdeletions involving ACTB associated with developmental delay, short stature, and microcephaly. Eur J Med Genet 2016, 59, 502-6.
23. Shimojima K, Maruyama K, Kikuchi M, Imai A, Inoue K, Yamamoto T. Novel SLC16A2 mutations in patients with Allan-Herndon-Dudley syndrome. Intractable Rare Dis Res 2016, 5; 214-7.
24. Shimojima K, Ondo Y, Nishi E, Mizuno S, Ito M, Ioi A, Shimizu M, Sato M, Inoue M, Okamoto N, Yamamoto T. Loss-of-function mutations and global rearrangements in GPC3 in patients with Simpson-Golabi-Behmel syndrome. Hum Genome Var 2016, 3, 16033.
25. Morisada N, Ioroi T, Taniguchi-Ikeda M, Ye MJ, Okamoto N, Yamamoto T, Iijima K. A 12p13 GRIN2B deletion is associated with developmental delay and macrocephaly. Hum Genome Var 2016, 3, 16029.
26. Yamamoto T, Shimojima K, Ondo Y, Imai K, Chong P-F, Kira R, Amemiya M, Saito A, Okamoto N. Challenges in detecting genomic copy number aberrations using next-generation sequencing data and the eXome Hidden Markov Model: a clinical exome-first diagnostic approach. Hum Genome Var 2016, 3, 16025.

27. Iwasaki N, Tsurumi M, Asai K, Shimuzu W, Watanabe A, Ogata M, Takizawa M, Ide R, Yamamoto T, Saito K. Pancreatic developmental defect evaluated by celiac artery angiography in a patient with MODY5. Human Genome Var 2016, 3, 16022.
28. Banno K, Omori S, Hirata K, Nawa N, Nakagawa N, Nishimura K, Ohtaka M, Nakanishi M, Sakuma T, Yamamoto T, Toki T, Ito E, Yamamoto T, Kokubu C, Takeda J, Taniguchi H, Arahori H, Wada K, Kitabatake Y, OzonoK. Systematic cellular disease models reveal synergistic interaction of trisomy 21 and GATA1 mutations in hematopoietic abnormalities. Cell Reports 2016, 15, 1228-41.
29. Itakura A, Saito Y, Nishimura Y, Okazaki T, Ohno K, Sejima H, Yamamoto T, Maegaki Y. Successful treatment of migrating partial seizures in Wolf–Hirschhorn syndrome with bromide. Brain Dev 2016, 38, 658-62.
30. 下島圭子, 三宮範子, 島田姿野, 影山優子, 沼部博直, 山本俊至. 非医療系大学生のダウン症候群および出生前診断についての理解と意識の分析. 日本遺伝カウンセリング学会誌 2016, 37, 39-43.
31. Yamamoto T, Yoshioka S, Tsurusaki Y, Shino S, Shimojima K, Shigematsu Y, Takeuchi Y, Matsumoto N. White matter abnormalities in an adult patient with l-2-hydroxyglutaric aciduria. Brain Dev. 2016, 38, 142-4.
32. Oka M, Shimojima K, Yamamoto T, Hanaoka Y, Sato S, Yasuhara T, Yoshinaga H, Kobayashi K. A novel HYLS1 homozygous mutation in living siblings with Joubert syndrome. Clin Genet 2016, 89, 739-43.
33. Sangu N, Okamoto N, Shimojima K, Ondo Y, Nishikawa M, Yamamoto T. A de novo microdeletion in a patient with inner ear abnormalities suggests the existence of the responsible gene in 10q26.13. Human Genome Var 2016, 3, 16008.
34. Shimojima K, Okamoto N, Yamamoto T. A novel TUBB3 mutation in a sporadic patient with asymmetric cortical dysplasia. Am J Med Genet A 2016, 170A, 1076-9.
35. Watanabe S, Shimizu K, Ohashi H, Kosaki R, Okamoto N, Shimojima K, Yamamoto T, Chinen Y, Mizuno S, Dowa Y, Shiomi N, Toda Y, Tashiro K, Shichijo K, Minatozaki K, Aso S, Minagawa K, Hiraki Y, Shimokawa O, Matsumoto T, Fukuda M, Moriuchi H, Yoshiura K, Kondoh T. Detailed analysis of 26 cases of 1q partial duplication/triplication 1 syndrome. Am J Med Genet A 2016, 170A, 908-17.
36. Yamamoto T, Igarashi N, Shimojima K, Sangu N, Sakamoto Y, Shimoji K, Niijima S. Use of targeted next-generation sequencing for molecular diagnosis of craniosynostosis: identification of a novel de novo mutation of EFNB1. Congenit Anom (Kyoto) 2016, 56, 91-3.
37. Yamamoto T, Shimojima K, Yano T, Ueda Y, Takayama R, Ikeda H, Imai K. Loss-of-function mutations of STXBP1 in patients with epileptic encephalopathy. Brain Dev 2016, 38, 280-4.
38. Igarashi A, Okumura A, Shimojima K, Abe S, Ikeno M, Shimizu T, Yamamoto T. Focal seizures and epileptic spasms in a child with Down syndrome from a family with a PRRT2 mutation. Brain Dev. 2016, 38, 597-600.
39. Okamoto N, Toribe Y, Shimojima K, Yamamoto T. Tatton-Brown-Rahman syndrome due to 2p23 microdeletion. Am J Med Genet A. 2016, 170A, 1339-42.

40. Ishikawa N, Kobayashi Y, Fujii Y, Yamamoto T, Kobayashi M. Late-onset epileptic spasms in a patient with 22q13.3 deletion syndrome. Brain Dev. 2016, 38, 109-12.
41. Sumida K, Inoue K, Takanashi J, Sasaki M, Watanabe K, Suzuki M, Kurahashi H, Omata T, Tanaka M, Yokochi K, Iio J, Iyoda K, Kurokawa T, Matsuo M, Sato T, Iwaki A, Osaka H, Kurosawa K, Yamamoto T, Matsumoto N, Maikusa N, Matsuda H, Sato N. The magnetic resonance imaging spectrum of Pelizaeus-Merzbacher disease: A multicenter study of 19 patients. Brain Dev. 2016, 38, 571-80.

山形嵩倫

1. Goto M, Mizuno M, Matsumoto A, Yang Z, Jimbo EF, Tabata H, Yamagata T, Nagata KI. Role of a circadian-relevant gene NR1D1 in brain development: possible involvement in the pathophysiology of autism spectrum disorders. Sci Rep. 2017, 6, 43945.
2. Takayanagi Y, Yoshida M, Takashima A, Takanami K, Yoshida S, Nishimori K, Nishijima I, Sakamoto H, Yamagata T, Onaka T. Activation of Supraoptic Oxytocin Neurons by Secretin Facilitates Social Recognition. Biol Psychiatry. 2017,81, 243-51.
3. Stickley A, Tachibana Y, Hashimoto K, Haraguchi H, Miyake A, Morokuma S, Nitta H, Oda M, Ohya Y, Senju A, Takahashi H, Yamagata T, Kamio Y. Assessment of Autistic Traits in Children Aged 2 to 4½ Years With the Preschool Version of the Social Responsiveness Scale (SRS-P): Findings from Japan. Autism Res. 2017 [Epub ahead of print]
4. Yang Z, Matsumoto A, Nakayama K, Jimbo EF, Kojima K, Nagata KI, Iwamoto S, Yamagata T. Circadian-relevant genes are highly polymorphic in autism spectrum disorder patients. Brain Dev. 2016, 38, 91-9.
5. Inaguma Y, Matsumoto A, Noda M, Tabata H, Maeda A, Goto M, Usui D, Jimbo EF, Kikkawa K, Ohtsuki M, Momoi MY, Osaka H, Yamagata T, Nagata KI. Role of Class III phosphoinositide 3-kinase in the brain development: possible involvement in specific learning disorders. J Neurochem. 2016, 139, 245-55.
6. Saitsu H, Fukai R, Ben-Zeev B, Sakai Y, Mimaki M, Okamoto N, Suzuki Y, Monden Y, Saito H, Tziperman B, Torio M, Akamine S, Takahashi N, Osaka H, Yamagata T, Nakamura K, Tsurusaki Y, Nakashima M, Miyake N, Shiina M, Ogata K, Matsumoto N. Phenotypic spectrum of GNAO1 variants: epileptic encephalopathy to involuntary movements with severe developmental delay. Eur J Hum Genet. 2016, 24, 129-34.

小坂仁

1. Kondo T, Funayama M, Miyake M, Tsukita K, Era T, Osaka H, Ayaki T, Takahashi R, Inoue H. Modeling Alexander disease with patient iPSCs reveals cellular and molecular pathology of astrocytes. Acta Neuropathol Commun. 2016; 4, 69.

矢田俊彦

1. Santoso P, Nakata M, Ueta Y, Yada T. Suprachiasmatic Vasopressin to Paraventricular Oxytocin Neurocircuit in the Hypothalamus Relays Light Reception to Inhibition of Feeding

Behavior. Am J Physiol Endocrinol Metab. 2017. [Epub ahead of print]

2. Nakata M, Gantulga D, Santoso P, Zhang B, Masuda C, Mori M, Okada T, Yada T. Paraventricular NUCB2/nesfatin-1 supports oxytocin and vasopressin neurons to control feeding behavior and fluid balance in male mice. Endocrinology 2016, 157, 2322-32.

永田浩一

1. Inoue M, Iwai R, Tabata H, Konno D, Komabayashi-Suzuki M, Watanabe C, Iwanari H, Mochizuki Y, Hamakubo T, Matsuzaki F, Nagata K, Mizutani K. Prdm16 is critical for progression of the multipolar phase during neural differentiation of the developing neocortex. Development. 2017 (in press)
2. Hamada N, Negishi Y, Mizuno M, Miya F, Hattori A, Okamoto N, Kato M, Tsunoda T, Yamasaki M, Kanemura Y, Kosaki K, Tabata H, Saitoh S, Nagata K. Role of a heterotrimeric G-protein, Gi2, in the corticogenesis: Possible involvement in periventricular nodular heterotopia and intellectual disability. J. Neurochem. 2017, 140, 82-95.
3. Goto M, Mizuno M, Matsumoto A, Yang Z, Jimbo F.E, Tabata H, Yamagata T, Nagata K. Role of a circadian-relevant gene, NR1D1, in the brain development: possible involvement in the pathophysiology of autism spectrum disorders. Sci Rep. 2017, 7, 43945.
4. Inaguma Y, Ito H, Iwamoto I, Matsumoto A, Yamagata T, Tabata H, Nagata K. Morphological characterization of Class III phosphoinositide 3-kinase during mouse brain development. Med. Mol. Morphol. 2016, 49, 28-33.
5. Yuan Q, Yang F, Xiao Y, Tan S, Husain N, Ren M, Hu Z, Martinowich K, Ng J S, Kim P J, Han W, Nagata K, Weinberger DR, H. Je S. Regulation of BDNF exocytosis and GABAergic interneuron synapse by the schizophrenia susceptibility gene dysbindin-1. Biol Psychiatry. 2016, 80, 312–22.
6. Yang Z, Matsumoto A, Nakayama K, Jimbo F E, Kojima K, Nagata K, Iwamoto S, Yamagata T. Circadian-relevant genes are highly polymorphic in autism spectrum disorder patients. Brain Dev. 2016, 38, 91-9.
7. Hamada N, Ito H, Nishijo T, Iwamoto I, Morishita R, Tabata H, Momiyama T, Nagata K. Essential role of the nuclear isoform of RBFOX1, a candidate gene for autism spectrum disorders, in the brain development. Sci Rep. 2016, 6, 30805.
8. Ito H, Morishita R, Nagata K. Schizophrenia susceptibility gene product dysbindin-1 regulates the homeostasis of cyclin D1. BBA-Molecular Basis of Disease 2016, 1862, 1383-91.
9. Inaguma Y, Matsumoto A, Noda M, Tabata H, Maeda A, Goto M, Usui D, Jimbo F.E, Kikkawa K, Ohtsuki M, Momoi YM, Osaka H, Yamagata T, Nagata K. Role of Class III phosphoinositide 3-kinase in the brain development: possible involvement in specific learning disorders. J. Neurochem. 2016, 139, 245-55.

岡本伸彦

1. Okamoto N, Miya F, TsunodaT, Kato M, Saitoh S, Yamasaki M, Kanemura Y, Kosaki K. Novel MCA/ID syndrome with ASH1L mutation. Am J Med Genet A. 2017 (in press).

2. Gordon CT, Xue S, Yigit G, Filali H, Chen K, Rosin N, Yoshiura KI, Oufadem M, Beck TJ, McGowan R, Magee AC, Altmüller J, Dion C, Thiele H, Gurzau AD, Nürnberg P, Meschede D, Mühlbauer W, Okamoto N, Varghese V, Irving R, Sigaudy S, Williams D, Ahmed SF, Bonnard C, Kong MK, Ratbi I, Fejjal N, Fikri M, Elalaoui SC, Reigstad H, Bole-Feysot C, Nitschké P, Ragge N, Lévy N, Tunçbilek G, Teo AS, Cunningham ML, Sefiani A, Kayserili H, Murphy JM, Chatdokmaiprai C, Hillmer AM, Wattanasirichaigoon D, Lyonnet S, Magdinier F, Javed A, Blewitt ME, Amiel J, Wollnik B, Reversade B. De novo mutations in SMCHD1 cause Bosma arhinia microphthalmia syndrome and abrogate nasal development. Nat Genet. 2017. [Epub ahead of print]
3. Shaw ND, Brand H, Kupchinsky ZA, Bengani H, Plummer L, Jones TI, Erdin S, Williamson KA, Rainger J, Stortchevoi A, Samocha K, Currall BB, Dunican DS, Collins RL, Willer JR, Lek A, Lek M, Nassan M, Pereira S, Kammin T, Lucente D, Silva A, Seabra CM, Chiang C, An Y, Ansari M, Rainger JK, Joss S, Smith JC, Lippincott MF, Singh SS, Patel N, Jing JW, Law JR, Ferraro N, Verloes A, Rauch A, Steindl K, Zweier M, Scheer I, Sato D, Okamoto N, Jacobsen C, Tryggestad J, Chernausek S, Schimmenti LA, Brasseur B, Cesaretti C, García-Ortiz JE, Buitrago TP, Silva OP, Hoffman JD, Mühlbauer W, Ruprecht KW, Loeys BL, Shino M, Kaindl AM, Cho CH, Morton CC, Meehan RR, van Heyningen V, Liao EC, Balasubramanian R, Hall JE, Seminara SB, Macarthur D, Moore SA, Yoshiura KI, Gusella JF, Marsh JA, Graham JM Jr, Lin AE, Katsanis N, Jones PL, Crowley WF Jr, Davis EE, FitzPatrick DR, Talkowski ME. SMCHD1 mutations associated with a rare muscular dystrophy can also cause isolated arhinia and Bosma arhinia microphthalmia syndrome. Nat Genet. 2017 [Epub ahead of print].
4. Okamoto N, Shimojima K, Yamamoto T. Neurological Manifestations of 2q31 Microdeletion Syndrome. Congenit Anom. 2017 (in press)
5. Shimojima K, Okamoto N, Yamamoto T. Possible genes responsible for developmental delay observed in patients with rare 2q23q24 microdeletion syndrome: literature review and description of an additional patient. Congenit Anom. 2017 (in press).
6. Okamoto N, Watanabe M, Naruto T, Matsuda K, Kohmoto T, Saito M, Masuda K, Imoto I. Genome-first approach diagnosed Cabezas syndrome via novel CUL4B mutation detection. Hum Genome Var. 2017, 4, 16045.
7. Negishi Y, Miya F, Hattori A, Johmura Y, Nakagawa M, Ando N, Hori I, Togawa T, Aoyama K, Ohashi K, Fukumura S, Mizuno S, Umemura A, Kishimoto Y, Okamoto N, Kato M, Tsunoda T, Yamasaki M, Kanemura Y, Kosaki K, Nakanishi M, Saitoh S. A combination of genetic and biochemical analyses for the diagnosis of PI3K-AKT-mTOR pathway-associated megalencephaly. BMC Med Genet. 2017, 18, 4.
8. Hamada N, Negishi Y, Mizuno M, Miya F, Hattori A, Okamoto N, Kato M, Tsunoda T, Yamasaki M, Kanemura Y, Kosaki K, Tabata H, Saitoh S, Nagata KI. Role of a heterotrimeric G-protein, Gi2, in the corticogenesis: Possible involvement in periventricular nodular heterotopia and intellectual disability. J Neurochem. 2017, 140, 82-95.

9. Matsuo M, Yamauchi A, Ito Y, Sakauchi M, Yamamoto T, Okamoto N, Tsurusaki Y, Miyake N, Matsumoto N, Saito K. Mandibulofacial dysostosis with microcephaly: A case presenting with seizures. Brain Dev. 2017, 39, 177-81.
10. Morisada N, Ioroi T, Taniguchi-Ikeda M, Juan Ye M, Okamoto N, Yamamoto T, Iijima K. A 12p13 GRIN2B deletion is associated with developmental delay and macrocephaly. Hum Genome Var. 2017, 3, 16029.
11. Miyake N, Tsurusaki Y, Koshimizu E, Okamoto N, Kosho T, Jane Brown N, Yang Tan T, Jia Jiunn Yap P, Suzumura H, Tanaka T, Nagai T, Nakashima M, Saitsu H, Niikawa N, Matsumoto N. Delineation of clinical features in Wiedemann-Steiner syndrome caused by KMT2A mutations. Clin Genet. 2016, 89, 115-9.
12. Shimojima K, Ondo Y, Nishi E, Mizuno S, Ito M, Ioi A, Shimizu M, Sato M, Inoue M, Okamoto N, Yamamoto T. Loss-of-function mutations and global rearrangements in GPC3 in patients with Simpson-Golabi-Behmel syndrome. Hum Genome Var. 2016, 3, 16033.
13. Miyake N, Fukai R, Ohba C, Chihara T, Miura M, Shimizu H, Kakita A, Imagawa E, Shiina M, Ogata K, Okuno-Yuguchi J, Fueki N, Ogiso Y, Suzumura H, Watabe Y, Imataka G, Leong HY, Fattal-Valevski A, Kramer U, Miyatake S, Kato M, Okamoto N, Sato Y, Mitsuhashi S, Nishino I, Kaneko N, Nishiyama A, Tamura T, Mizuguchi T, Nakashima M, Tanaka F, Saitsu H, Matsumoto N. Biallelic TBCD Mutations Cause Early-Onset Neurodegenerative Encephalopathy. Am J Hum Genet. 2016, 99, 950-61.
14. Tsutsumi M, Yokoi S, Miya F, Miyata M, Kato M, Okamoto N, Tsunoda T, Yamasaki M, Kanemura Y, Kosaki K, Saitoh S, Kurahashi H. Novel compound heterozygous variants in PLK4 identified in a patient with autosomal recessive microcephaly and chorioretinopathy. Eur J Hum Genet. 2016, 24, 1702-6.
15. Yamamoto T, Shimojima K, Ondo Y, Imai K, Chong PF, Kira R, Amemiya M, Saito A, Okamoto N. Challenges in detecting genomic copy number aberrations using next-generation sequencing data and the eXome Hidden Markov Model: a clinical exome-first diagnostic approach. Hum Genome Var. 2016, 3, 16025.
16. 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, 90, 526-35.
17. Sangu N, Okamoto N, Shimojima K, Ondo Y, Nishikawa M, Yamamoto T. A de novo microdeletion in a patient with inner ear abnormalities suggests the existence of the responsible gene in 10q26.13. Hum Genome Var. 2016, 3, 16008.
18. Miyake N, Abdel-Salam G, Yamagata T, Eid MM, Osaka H, Okamoto N, Mohamed AM, Ikeda T, Afifi HH, Piard J, van Maldergem L, Mizuguchi T, Miyatake S, Tsurusaki Y, Matsumoto N. Clinical features of SMARCA2 duplication overlap with Coffin-Siris syndrome. Am J Med Genet A. 2016, 170, 2662-70.

19. Fujita A, Isidor B, Piloquet H, Corre P, Okamoto N, Nakashima M, Tsurusaki Y, Saitsu H, Miyake N, Matsumoto N. De novo MEIS2 mutation causes syndromic developmental delay with persistent gastro-esophageal reflux. J Hum Genet. 2016, 61, 835-8.
20. Hori I, Miya F, Ohashi K, Negishi Y, Hattori A, Ando N, Okamoto N, Kato M, Tsunoda T, Yamasaki M, Kanemura Y, Kosaki K, Saitoh S. Novel splicing mutation in the ASXL3 gene causing Bainbridge-Ropers syndrome. Am J Med Genet A. 2016, 170, 1863-7.
21. Okamoto N, Toribe Y, Shimojima K, Yamamoto T. Tatton-Brown-Rahman syndrome due to 2p23 microdeletion. Am J Med Genet A. 2016, 170A, 1339-42.
22. Uehara DT, Hayashi S, Okamoto N, Mizuno S, Chinen Y, Kosaki R, Kosho T, Kurosawa K, Matsumoto H, Mitsubuchi H, Numabe H, Saitoh S, Makita Y, Hata A, Imoto I, Inazawa J. SNP array screening of cryptic genomic imbalances in 450 Japanese subjects with intellectual disability and multiple congenital anomalies previously negative for large rearrangements. J Hum Genet. 61:335-43, 2016
23. Fukai R, Saitsu H, Tsurusaki Y, Sakai Y, Haginoya K, Takahashi K, Hubshman MW, Okamoto N, Nakashima M, Tanaka F, Miyake N, Matsumoto N. De novo KCNH1 mutations in four patients with syndromic developmental delay, hypotonia and seizures. J Hum Genet. 2016, 61, 381-7.
24. Watanabe S, Shimizu K, Ohashi H, Kosaki R, Okamoto N, Shimojima K, Yamamoto T, Chinen Y, Mizuno S, Dowa Y, Shiomi N, Toda Y, Tashiro K, Shichijo K, Minatozaki K, Aso S, Minagawa K, Hiraki Y, Shimokawa O, Matsumoto T, Fukuda M, Moriuchi H, Yoshiura KI, Kondoh T. Detailed analysis of 26 cases of 1q partial duplication/triplication syndrome. Am J Med Genet A. 2016, 170A, 908-17.
25. Fukai R, Saitsu H, Okamoto N, Sakai Y, Fattal-Valevski A, Masaaki S, Kitai Y, Torio M, Kojima-Ishii K, Ihara K, Chernuha V, Nakashima M, Miyatake S, Tanaka F, Miyake N, Matsumoto N. De novo missense mutations in NALCN cause developmental and intellectual impairment with hypotonia. J Hum Genet. 2016, 61, 451-5.
26. Shimojima K, Okamoto N, Yamamoto T. A novel TUBB3 mutation in a sporadic patient with asymmetric cortical dysplasia. Am J Med Genet A. 2016, 170A, 1076-9.
27. Yaoita M, Niihori T, Mizuno S, Okamoto N, Hayashi S, Watanabe A, Yokozawa M, Suzumura H, Nakahara A, Nakano Y, Hokosaki T, Ohmori A, Sawada H, Migita O, Mima A, Lapunzina P, Santos-Simarro F, García-Miñaúr S, Ogata T, Kawame H, Kurosawa K, Ohashi H, Inoue S, Matsubara Y, Kure S, Aoki Y. Spectrum of mutations and genotype-phenotype analysis in Noonan syndrome patients with RIT1 mutations. Hum Genet. 2016, 135, 209-22.
28. Saitsu H, Fukai R, Ben-Zeev B, Sakai Y, Mimaki M, Okamoto N, Suzuki Y, Monden Y, Saito H, Tziperman B, Torio M, Akamine S, Takahashi N, Osaka H, Yamagata T, Nakamura K, Tsurusaki Y, Nakashima M, Miyake N, Shiina M, Ogata K, Matsumoto N. Phenotypic spectrum of GNAO1 variants: epileptic encephalopathy to involuntary movements with severe developmental delay. Eur J Hum Genet. 2016, 24, 129-34.

29. Takenouchi T, Okamoto N, Ida S, Uehara T, Kosaki K. Further evidence of a mutation in CDC42 as a cause of a recognizable syndromic form of thrombocytopenia. Am J Med Genet A. 2016, 170A, 852-5.
30. Nakagawa T, Taniguchi-Ikeda M, Murakami Y, Nakamura S, Motooka D, Emoto T, Satake W, Nishiyama M, Toyoshima D, Morisada N, Takada S, Tairaku S, Okamoto N, Morioka I, Kurahashi H, Toda T, Kinoshita T, Iijima K. A novel PIGN mutation and prenatal diagnosis of inherited glycosylphosphatidylinositol deficiency. Am J Med Genet A. 2016, 170A, 183-8.

齋藤潤

1. Kawasaki Y, Oda H, Ito J, Niwa A, Tanaka T, Hijikata A, Seki R, Nagahashi A, Osawa M, Asaka I, Watanabe A, Nishimata S, Shirai T, Kawashima H, Ohara O, Nakahata T, Nishikomori R, Heike T, Saito MK. Identification of a High-Frequency Somatic NLRC4 Mutation as a Cause of Autoinflammation by Pluripotent Cell-Based Phenotype Dissection. Arthritis Rheumatol. 2017, 69, 447-59.
2. Ohta R, Niwa A, Taniguchi Y, Suzuki N, Toga J, Yagi E, Saiki N, Nishinaka-Arai Y, Okada C, Watanabe A, Nakahata T, Sekiguchi K, Saito MK. Laminin-guided highly efficient endothelial commitment from human pluripotent stem cells. Scientific Reports. 2016, 6, 35680.
3. Sugimine Y, Niwa A, Matsubara H, Kobayashi K, Tabata Y, Heike T, Nakahata T, Saito MK. A portable platform for stepwise hematopoiesis from human pluripotent stem cells within PET-reinforced collagen sponges. Int J Hematol. 2016, 104, 647-60.
4. Saito MK, Niwa A. Hematological disorders. In: Fukuda K (ed), Human iPS cells in disease modeling, New York : Springer; 2016. 69-81.

北畠康司

1. Banno K, Omori S, Hirata K, Nawa N, Nakagawa N, Nishimura K, Ohtaka M, Nakanishi M, Sakuma T, Yamamoto T, Toki T, Ito E, Yamamoto T, Kokubu C, Takeda J, Taniguchi H, Arahori H, Wada K, Kitabatake Y, Ozono K. Systematic Cellular Disease Models Reveal Synergistic Interaction of Trisomy 21 and GATA1 Mutations in Hematopoietic Abnormalities. Cell Reports. 2016, 15, 1228-41.
2. 坂野公彦, 北畠康司. 創薬をめざした疾患モデル iPS 細胞の作製. All About ゲノム編集, 2016, 34, 141-145.
3. 坂野公彦, 北畠康司. ダウン症候群における GATA1 変異と造血異常. 血液内科, 2016, 74, 252-257.
4. Omori S, Tanabe H, Banno K, Tsuji A, Nawa N, Hirata K, Kawatani K, Kokubu C, Takeda J, Taniguchi H, Arahori H, Wada K, Kitabatake Y, Ozono K. A Pair of Maternal Chromosomes Derived from Meiotic Nondisjunction in Trisomy 21 Affects Nuclear Architecture and Transcriptional Regulation. Scientific Reports. 2017, 7, 764.

（2）学会・シンポジウム等における口頭・ポスター発表

山本俊至

1. Two cases of monosomy of 3q with cerebral MRI findings, ポスター, Dowa Y, Sameshima K, Ichinomiya K, Shiihara T, Shimojima K, Yamamoto T, The 13th International Congress of Human Genetics, April 3rd-7th 2016, Kyoto, Japan, 国内.
2. Similarities of the ectodermal dysplasia, hypohidrotic, with hypothyroidism and agenesis of the corpus callosum (OMIM 225040) and cardio-facio-cutaneous syndrome (OMIM 115150)、Moroto M, Chiyonobu T, Tokuda S, Kosaka K, Morioka S, Yamamoto T, Aoki Y, Morimoto M、The 13th International Congress of Human Genetics, April 3rd-7th 2016, Kyoto, Japan, 国内.
3. Tatton-Brown-Rahman syndrome due to 2p23 microdeletion, ポスター, Ueda K, Okamoto N, Toribe Y, Shimojima K, Yamamoto T, The 13th International Congress of Human Genetics, April 3rd-7th 2016, Kyoto, Japan, 国内.
4. Novel mutation in the COL1A1 gene causes severe scoliosis and valvular heart disease in a Japanese family with osteogenesis imperfecta, ポスター, Seto T, Yamamoto T, Shimojima K, Shintaku H, The 13th International Congress of Human Genetics, April 3rd-7th 2016, Kyoto, Japan, 国内.
5. Mutation in the gene encoding eukaryotic translation initiation factor 2B in Japanese patients with vanishing white matter disease, ポスター, Shimada S, Shimojima K, Yamamoto T, Nagata S, The 13th International Congress of Human Genetics, April 3rd-7th 2016, Kyoto, Japan, 国内.
6. A case of mandibulofacial dysostosis with microcephaly presenting with epilepsy, ポスター, Matsuo M, Sakauchi M, Yamauchi A, Ito Y, Yamamoto T, Okamoto N, Tsurusaki Y, Miyake N, Matsumoto N, Saito K, The 13th International Congress of Human Genetics, April 3rd-7th 2016, Kyoto, Japan, 国内.
7. Novel PLA2G6 mutations associated with an exonic deletion due to non-allelic homologous recombination in a patient with infantile neuroaxonal dystrophy, ポスター, Yamamoto T, Shimojima K, Shibata T, Akiyama M, Oka M, Akiyama T, Yoshinaga H, Kobayashi K, The 13th International Congress of Human Genetics, April 3rd-7th 2016, Kyoto, Japan, 国内.
8. Single nucleotide variation in CLCN6 identified in patients with benign partial epilepsies in infancy and/or febrile seizures, ポスター, Yamamoto T, Shimojima K, Komoike Y, Ishii A, Abe S, Yamashita S, Imai K, Kubota T, Fukasawa T, Okanishi T, Enoki H, Tanabe T, Saito A, Furukawa T, Shimizu T, Milligan CJ, Petrou S, Heron SE, Dibbens LM, Hirose S, Okumura A, The 13th International Congress of Human Genetics, April 3rd-7th 2016, Kyoto, Japan, 国内.
9. CHCHD2 is down-regulated in neuronal cells differentiated from iPS cells derived from patients with lissencephaly, ポスター, Shimojima K, Okumura A, Hayashi M, Kondo T, Inoue H, Yamamoto T, The 13th International Congress of Human Genetics, April 3rd-7th 2016, Kyoto, Japan, 国内.

10. 網羅的遺伝子診断, 口頭, 山本俊至, 第 58 回日本小児神経学会学術集会 2016.6.3-5, 国内.
11. 非対称性大脳皮質異形成症症例に認められた新規 TUBB3 変異, 口頭, 下島圭子, 岡本伸彦, 山本俊至, 第 58 回日本小児神経学会学術集会 2016.6.3-5, 国内.
12. Megalencephalic leukoencephalopathy with subcortical cysts の日本人 8 名における MLC1 遺伝子解析, 口頭, 島田姿野, 山本俊至, 下島圭子, 永田智, 第 58 回日本小児神経学会学術集会 2016.6.3-5, 国内.
13. 指定発言「種々の介入を要した Emanuel 症候群の乳児期の発達経過」, 口頭, 山本俊至, 第 628 回日本小児科学会東京都地方会講和会, 2016.6.11, 国内.
14. 次世代シーケンスによる SNV スクリーニングだけでは CNV が見逃される, ポスター, 山本俊至, 下島圭子, 恩藤由美子, 岡本伸彦, 第 56 回日本先天異常学会学術集会, 2016.7.29-31, 国内.
15. ラムダ縫合早期癒合を示した MAP2K2 領域の微細欠失による RASopathies 患者, ポスター, 下島圭子, 恩藤由美子, 松藤まゆみ, 佐野のぞみ, 水流尚志, 山本俊至, 第 56 回日本先天異常学会学術集会, 2016.7.29-31, 国内.

矢田俊彦

1. オキシトシン点鼻投与による血中インスリン分泌促進と耐糖能改善, ポスター, 出崎克也, 三浦李菜, 矢田俊彦, 第 37 回日本肥満学会, 2016.10.7-8, 国内.
2. Synaptic plasticity onto oxytocin neurons in the hypothalamic paraventricular nucleus is regulated by systemic energy states, 口頭, Suyama S, Hirano Kodaira M, Otgon-Uul Z, Ueta Y, Nakata M, Yada T, 第 39 回日本神経科学大会, 2016.7.20-22, 国内.

永田浩一

1. Comprehensive analyses of RBFOX1, a causative gene for neurodevelopmental and psychiatric diseases, in the brain development, Nagata K, The 14th Meeting of the Asian-Pacific Society for Neurochemistry, Symposium: Common molecular aspects of neurodevelopmental and psychiatric disorders, 2016/8/28, 国外.

岡本伸彦

1. Novel MCA/ID syndrome with ASH1L mutation, Okamoto N, Miya F, Nishioka K, Soejima H, Tsunoda T, Kato M, Saitoh S, Yamasaki M, Kanemura Y, Kosaki K, 第 13 回国際人類遺伝学会. 2016/4, 国内.
2. Novel Splicing Mutation in the ASXL3 gene causing Bainbridge-Ropers Syndrome, Hori I, Miya F, Ohashi K, Negishi Y, Hattori A, Ando N, Okamoto N, Kato M, Tsunoda T, Yamasaki M, Kanemura Y, Kosaki K, Saitoh S, 第 13 回国際人類遺伝学会, 2016/4, 国内.
3. Broadening the phenotypic spectrum of ANKRD11-related syndrome, Miyatake S, Okamoto N, Stark Z, Tsurusaki Y, Nakashima M, Saitsu H, Miyake N, Ohtake A, Matsumoto N, 第 13 回国際人類遺伝学会, 2016/4, 国内.
4. CDC42 as a new human disease causative gene, Uehara T, Okamoto N, Takenouchi T, Ida S, Kosaki K, 第 13 回国際人類遺伝学会, 2016/4, 国内.

5. A combination of targeted enrichment methodologies for whole-exome sequencing reveals novel pathogenic mutations, Miya F, Kato M, Shiohama T, Okamoto N, Saitoh S, Yamasaki M, Shigemizu D, Abe T, Morizono T, Boroevich KA, Kosaki K, Kanemura Y, Tsunoda T, 第 13 回国際人類遺伝学会, 2016/4, 国内.
6. A novel COL11A1 mutation affecting splicing in a Japanese patient with Stickler syndrome, Naruto T, Kohmoto T, Kobayashi H, Watanabe M, Okamoto N, Masuda K, Okamoto N, Imoto I, 第 13 回国際人類遺伝学会, 2016/4, 国内.
7. Tatton-Brown-Rahman syndrome due to 2p23 microdeletion, Ueda K, Okamoto N, Toribe Y, Shimojima K, Yamamoto T, 第 13 回国際人類遺伝学会, 2016/4, 国内.
8. A case of mandibulofacial dysostosis with microcephaly presenting with epilepsy, Matsuo M, Sakauchi M, Yamauchi A, Ito Y, Yamamoto T, Okamoto N, Tsurusaki Y, Miyake N, Matsumoto N, Saito K, 第 13 回国際人類遺伝学会, 2016/4, 国内.
9. Novel compound heterozygous mutations in ISPD gene from two cases of Japanese Walker-Warburg syndrome identified by whole-exome sequencing, Kanemura Y, Miya F, Shofuda T, Yoshioka E, Kanematsu D, Itoh K, Fushiki S, Okinaga T, Sago H, Kosaki R, Minagawa K, Okamoto N, Tsunoda T, Kato M, Saitoh S, Kosaki K, Yamasaki M, 第 13 回国際人類遺伝学会, 2016/4, 国内.
10. Homozygous ADCY5 mutation causes movement disorder with severe intellectual disability, Okamoto N, Miya F, Tsunoda T, Kato M, Saitoh S, Yamasaki M, Kanemura Y, Kosaki K, 第 13 回国際人類遺伝学会, 2016/4, 国内.
11. Haploinsufficiency of ZNF385B and neurological manifestations in 2q31microdeletion syndrome, Okamoto N, Shimojima K, Yamamoto T, 欧州人類遺伝学会, 2016/5, 国外.
12. Craniosynostosis is a common complication in Rasopathies, Okamoto N, Ueda K, Niihori T, Aoki Y, 米国人類遺伝学会, 2016/10, 国外.

齋藤潤

1. iPS 細胞をもちいた自己炎症性疾患の病態解析, 口頭, 齋藤潤, 第 60 回日本リウマチ学会総会・学術集会, 2016/4/21, 国内.
2. Defined laminin matrices を用いた多能性幹細胞からの血管内皮細胞分化, 口頭, 齋藤潤, マトリクソーム科学（ニッピ）寄附研究部門 開設記念シンポジウム, 2016/6/2, 国内.
3. Monocytic cell lines established from patient specific iPS cells serve a versatile platform for phenotype-based compound screening, 口頭, Saito MK, ヨーロッパリウマチ学会, 2016/6/10, 国外.
4. 疾患 iPS 細胞を用いた血液・免疫難病の病態解析と創薬へ向けた研究, 口頭, 齋藤潤, 日本炎症・再生医学会, 2016/6/16, 国内.
5. 自己炎症性疾患の iPS 細胞を用いた解析, 口頭, 齋藤潤, 日本炎症・再生医学会, 2016/6/17, 国内.
6. iPS 細胞を用いた 先天性免疫疾患の解析について, 口頭, 齋藤潤, 第 14 回 iPS 細胞・再生医学研究会, 2016/7/1, 国内.

7. Decoding the pathophysiology of immunological disorders using human iPS cells, 口頭, Saito MK, JAPAN-SPAIN JOINT WORKSHOP ON NANOMEDICINE RESEARCH, 2016/12/1, 国外.
8. 疾患特異的 iPS 細胞樹立のための 基盤形成事業について, 口頭, 齋藤潤, 東京女子医科大学公開シンポジウム「自閉症・発達障害の成因解明と将来の治療に向けて」, 2017/1/7, 国内.
9. iPS 細胞の医学応用へ向けた研究の現状について, 口頭, 齋藤潤, 京都私立病院協会講演会, 2017/1/24, 国内.
10. 疾患特異的 iPS 細胞を用いた自己炎症性疾患の病態解析と創薬に向けたアプローチ, 口頭, 齋藤潤, 愛媛大学プロテオサイエンスセンターシンポジウム, 2017/2/11, 国内.

北畠康司

1. ダウン症候群の胎児期から成人期までをとらえる ―初めてでもわかりやすい iPS 細胞研究の‘いま’ー, 北畠康司, 第 6 回新生児科指導医教育セミナー, 2016/8/13, 国内.
2. Genome Editing of Human iPS Cells to Generate Systematic Disease Models of Down Syndrome -A New Concept for an Old Disease-, 北畠康司, 三重大学大学院セミナー, 2016/11/16, 国内.
3. 疾患 iPS 細胞とゲノム編集をもちいたダウン症候群における TAM の病態解析－1, 北畠康司, 坂野公彦, 大森早也佳, 平田克弥, 那波伸敏, 中川夏季, 谷口英俊, 荒堀仁美, 和田和子, 大薗恵一, 第 119 回日本小児科学会, 2016/5/13-15, 国内.
4. 疾患 iPS 細胞とゲノム編集をもちいたダウン症候群における TAM の病態解析－2, 北畠康司, 坂野公彦, 大森早也佳, 平田克弥, 那波伸敏, 中川夏季, 谷口英俊, 荒堀仁美, 和田和子, 大薗恵一, 第 61 回日本新生児成育医学会, 2016/12/1-3, 国内.
5. トリソミー患児由来の iPS 細胞の樹立とその神経分化誘導によるトリソミー特異的神経障害の病態解明, 平田克也, 北畠康司, 坂野公彦, 川谷圭司, 辻田麻友子, 荒堀仁美, 和田和子, 大薗恵一, 第 89 回日本組織培養学会大会, 2016/5/25-26, 国内.

（3）「国民との科学・技術対話社会」に対する取り組み

1. 患者さんの細胞で病気を調べる, 齋藤潤, NHK文化センター京都教室 特別講座, 2018/12/28, 国内.

（4）特許出願

なし