Curriculum Vitae

Atsushi Takata, M.D., Ph.D.

Team Leader (Laboratory Head)

Laboratory for Molecular Pathology of Psychiatric Disorders

RIKEN Center for Brain Science

Hirosawa 2-1, Wako, Saitama, 351-0198, Japan

Phone: +81-48-467-9703

E-mail: atsushi.takata [at] riken.jp

Education

2004	M.D., Faculty of Medicine, Kyushu University
2011	Ph.D., Graduate School of Medical Sciences, Kyushu University, Neuropsychiatry

Academic Appointments

2020-present	Team Leader, Laboratory for Molecular Pathology of Psychiatric Disorders, RIKEN Center for Brain Science, Saitama, Japan
2020-present	Visiting Associate Professor, Department of Human Genetics, Yokohama City University Graduate School of Medicine, Kanagawa, Japan
2018-2020	Senior Visiting Scientist, Laboratory for Molecular Dynamics of Mental Disorders, RIKEN Center for Brain Science, Saitama, Japan
2016-2020	Assistant Professor (Lecturer), Department of Human Genetics, Yokohama City University Graduate School of Medicine, Kanagawa, Japan
2016-2018	Senior Visiting Scientist, Laboratory for Molecular Dynamics of Mental Disorders y, RIKEN Brain Science Institute, Saitama, Japan
2016	Visiting Associate Professor, Department of Psychiatry, Graduate School of Biomedical Sciences, Hiroshima University, Hiroshima, Japan
2015-2016	Tenure-track Research Scientist, Laboratory for Molecular Dynamics of Mental

Disorders, RIKEN Brain Science Institute, Saitama, Japan

2013-2015 Postdoctoral Fellow (JSPS Overseas Research Fellow), Department of Psychiatry, Columbia University Medical Center, USA

2011-2012 Visiting Lecturer, Department of Psychiatry, Fujita Health University School of Medicine, Aichi, Japan

2011-2015 Visiting Scientist, Laboratory for Molecular Psychiatry, RIKEN Brain Science Institute, Saitama, Japan

2011-2015 Visiting Scientist, Laboratory for Molecular Dynamics of Mental Disorders, RIKEN Brain Science Institute, Saitama, Japan

Hospital Appointments

2015-present Clinical Psychiatrist, Japan Depression Center Rokubancho Mental Clinic, Tokyo, Japan

2011-2013 Clinical Psychiatrist, Yowa Hospital, Tokyo, Japan

2006-2007 Senior Resident in Psychiatry, Dazaifu Hospital Psychiatric Center, Fukuoka, Japan

2005-2006 Junior Resident, Kyushu University Hospital, Fukuoka, Japan

2004-2005 Junior Resident, National Hospital Organization Kyushu Medical Center, Fukuoka, Japan

Board Certification

2012 Designated Psychiatrist by Japanese Ministry of Health, Labour and Welfare

Professional Organizations and Societies

2006-present Japanese Society of Psychiatry and Neurology

2010-present Japanese Society of Biological Psychiatry

Awards

2013 Academic Prize, Japanese Society for Biological Psychiatry

Publications

1. Hara T, Owada Y, Takata A. Genetics of bipolar disorder: insights into its complex architecture and biology from common and rare variants. ***J Hum Genet***. 2022.
2. Hamanaka K, Miyake N, Mizuguchi T, Miyatake S, Uchiyama Y, Tsuchida N, Sekiguchi F, Mitsuhashi S, Tsurusaki Y, Nakashima M, Saitsu H, Yamada K, Sakamoto M, Fukuda H, Ohori S, Saida K, Itai T, Azuma Y, Koshimizu E, Fujita A, Erturk B, Hiraki Y, Ch'ng GS, Kato M, Okamoto N, Takata A, Matsumoto N. Large-scale discovery of novel neurodevelopmental disorder-related genes through a unified analysis of single-nucleotide and copy number variants. ***Genome Med***. 2022;14(1):40.
3. Mizuguchi T, Toyota T, Miyatake S, Mitsuhashi S, Doi H, Kudo Y, Kishida H, Hayashi N, Tsuburaya RS, Kinoshita M, Fukuyama T, Fukuda H, Koshimizu E, Tsuchida N, Uchiyama Y, Fujita A, Takata A, Miyake N, Kato M, Tanaka F, Adachi H, Matsumoto N. Complete sequencing of expanded SAMD12 repeats by long-read sequencing and Cas9-mediated enrichment. ***Brain***. 2021;144(4):1103-17.
4. Aoi H, Mizuguchi T, Suzuki T, Makino S, Yamamoto Y, Takeda J, Maruyama Y, Seyama R, Takeuchi S, Uchiyama Y, Azuma Y, Hamanaka K, Fujita A, Koshimizu E, Miyatake S, Mitsuhashi S, Takata A, Miyake N, Takeda S, Itakura A, Matsumoto N. Whole exome sequencing of fetal structural anomalies detected by ultrasonography. ***J Hum Genet***. 2021;66(5):499-507.
5. Miyatake S, Kato M, Kumamoto T, Hirose T, Koshimizu E, Matsui T, Takeuchi H, Doi H, Hamada K, Nakashima M, Sasaki K, Yamashita A, Takata A, Hamanaka K, Satoh M, Miyama T, Sonoda Y, Sasazuki M, Torisu H, Hara T, Sakai Y, Noguchi Y, Miura M, Nishimura Y, Nakamura K, Asai H, Hinokuma N, Miya F, Tsunoda T, Togawa M, Ikeda Y, Kimura N, Amemiya K, Horino A, Fukuoka M, Ikeda H, Merhav G, Ekhilevitch N, Miura M, Mizuguchi T, Miyake N, Suzuki A, Ohga S, Saitsu H, Takahashi H, Tanaka F, Ogata K, Ohtaka-Maruyama C, Matsumoto N. De novo ATP1A3 variants cause polymicrogyria. ***Sci Adv***. 2021;7(13):eabd2368.
6. Nishioka M, Kazuno AA, Nakamura T, Sakai N, Hayama T, Fujii K, Matsuo K, Komori A, Ishiwata M, Watanabe Y, Oka T, Matoba N, Kataoka M, Alkanaq AN, Hamanaka K, Tsuboi T, Sengoku T, Ogata K, Iwata N, Ikeda M, Matsumoto N, Kato T, Takata A. Systematic analysis of exonic germline and postzygotic de novo mutations in bipolar disorder. ***Nat Commun***. 2021;12(1):3750.
7. Itai T, Hamanaka K, Sasaki K, Wagner M, Kotzaeridou U, Brosse I, Ries M, Kobayashi Y, Tohyama J, Kato M, Ong WP, Chew HB, Rethanavelu K, Ranza E, Blanc X, Uchiyama Y, Tsuchida N, Fujita A, Azuma Y, Koshimizu E, Mizuguchi T, Takata A, Miyake N, Takahashi H, Miyagi E, Tsurusaki Y, Doi H, Taguri M, Antonarakis SE, Nakashima M, Saitsu H, Miyatake S, Matsumoto N. De novo variants in CELF2 that disrupt the nuclear localization signal cause developmental and epileptic encephalopathy. ***Hum Mutat***. 2021;42(1):66-76.

8. Uchiyama Y, Yamaguchi D, Iwama K, Miyatake S, Hamanaka K, Tsuchida N, Aoi H, Azuma Y, Itai T, Saida K, Fukuda H, Sekiguchi F, Sakaguchi T, Lei M, Ohori S, Sakamoto M, Kato M, Koike T, Takahashi Y, Tanda K, Hyodo Y, Honjo RS, Bertola DR, Kim CA, Goto M, Okazaki T, Yamada H, Maegaki Y, Osaka H, Ngu LH, Siew CG, Teik KW, Akasaka M, Doi H, Tanaka F, Goto T, Guo L, Ikegawa S, Haginoya K, Haniffa M, Hiraishi N, Hiraki Y, Ikemoto S, Daida A, Hamano SI, Miura M, Ishiyama A, Kawano O, Kondo A, Matsumoto H, Okamoto N, Okanishi T, Oyoshi Y, Takeshita E, Suzuki T, Ogawa Y, Handa H, Miyazono Y, Koshimizu E, Fujita A, Takata A, Miyake N, Mizuguchi T, Matsumoto N. Efficient detection of copy-number variations using exome data: Batch- and sex-based analyses. ***Hum Mutat***. 2021;42(1):50-65.

9. Itai T, Miyatake S, Taguri M, Nozaki F, Ohta M, Osaka H, Morimoto M, Tandou T, Nohara F, Takami Y, Yoshioka F, Shimokawa S, Okuno-Yuguchi J, Motobayashi M, Takei Y, Fukuyama T, Kumada S, Miyata Y, Ogawa C, Maki Y, Togashi N, Ishikura T, Kinoshita M, Mitani Y, Kanemura Y, Omi T, Ando N, Hattori A, Saitoh S, Kitai Y, Hirai S, Arai H, Ishida F, Taniguchi H, Kitabatake Y, Ozono K, Nabatame S, Smigiel R, Kato M, Tanda K, Saito Y, Ishiyama A, Noguchi Y, Miura M, Nakano T, Hirano K, Honda R, Kuki I, Takanashi JI, Takeuchi A, Fukasawa T, Seiwa C, Harada A, Yachi Y, Higashiyama H, Terashima H, Kumagai T, Hada S, Abe Y, Miyagi E, Uchiyama Y, Fujita A, Imagawa E, Azuma Y, Hamanaka K, Koshimizu E, Mitsuhashi S, Mizuguchi T, Takata A, Miyake N, Tsurusaki Y, Doi H, Nakashima M, Saitsu H, Matsumoto N. Prenatal clinical manifestations in individuals with COL4A1/2 variants. ***J Med Genet***. 2021;58(8):505-13.

10. Aoto K, Kato M, Akita T, Nakashima M, Mutoh H, Akasaka N, Tohyama J, Nomura Y, Hoshino K, Ago Y, Tanaka R, Epstein O, Ben-Haim R, Heyman E, Miyazaki T, Belal H, Takabayashi S, Ohba C, Takata A, Mizuguchi T, Miyatake S, Miyake N, Fukuda A, Matsumoto N, Saitsu H. ATP6V0A1 encoding the a1-subunit of the V0 domain of vacuolar H+-ATPases is essential for brain development in humans and mice. ***Nat Commun***. 2021;12(1):1-17.

11. Miyake N, Heydari S, Garshasbi M, Saitoh S, Nasiri J, Hamanaka K, Takata A, Matsumoto N, Beheshti FH, Chaleshtori ARS. The identification of two pathogenic variants in a family with mild and severe forms of developmental delay. ***J Hum Genet***. 2021;66(4):445-8.

12. Sakamoto M, Iwama K, Sekiguchi F, Mashimo H, Kumada S, Ishigaki K, Okamoto N, Behnam M, Ghadami M, Koshimizu E, Miyatake S, Mitsuhashi S, Mizuguchi T, Takata A, Saitsu H, Miyake N, Matsumoto N. Novel EXOSC9 variants cause pontocerebellar hypoplasia type 1D with spinal motor neuronopathy and cerebellar atrophy. ***J Hum Genet***. 2021;66(4):401-7.

13. Takata A, Hamanaka K, Matsumoto N. Refinement of the clinical variant interpretation framework by statistical evidence and machine learning. ***Med***. 2021;2(5):611-32. e9.

14. Miyake N, Takahashi H, Nakamura K, Isidor B, Hiraki Y, Koshimizu E, Shiina M, Sasaki K, Suzuki H, Abe R, Kimura Y, Akiyama T, Tomizawa SI, Hirose T, Hamanaka K, Miyatake S,

Mitsuhashi S, Mizuguchi T, Takata A, Obo K, Kato M, Ogata K, Matsumoto N. Gain-of-Function MN1 Truncation Variants Cause a Recognizable Syndrome with Craniofacial and Brain Abnormalities. ***Am J Hum Genet***. 2020;106(1):13-25.

15. Hiromoto Y, Azuma Y, Suzuki Y, Hoshina M, Uchiyama Y, Mitsuhashi S, Miyatake S, Mizuguchi T, Takata A, Miyake N, Kato M, Matsumoto N. Hemizygous FLNA variant in West syndrome without periventricular nodular heterotopia. ***Hum Genome Var***. 2020;7(1):43.
16. Hamanaka K, Imagawa E, Koshimizu E, Miyatake S, Tohyama J, Yamagata T, Miyauchi A, Ekhilevitch N, Nakamura F, Kawashima T, Goshima Y, Mohamed AR, Ch'ng GS, Fujita A, Azuma Y, Yasuda K, Imamura S, Nakashima M, Saitsu H, Mitsuhashi S, Mizuguchi T, Takata A, Miyake N, Matsumoto N. De Novo Truncating Variants in the Last Exon of SEMA6B Cause Progressive Myoclonic Epilepsy. ***Am J Hum Genet***. 2020;106(4):549-58.
17. Saida K, Kim CA, Ceroni JRM, Bertola DR, Honjo RS, Mitsuhashi S, Takata A, Mizuguchi T, Miyatake S, Miyake N, Matsumoto N. Hemorrhagic stroke and renovascular hypertension with Grange syndrome arising from a novel pathogenic variant in YY1AP1. ***J Hum Genet***. 2019;64(9):885-90.
18. Uchiyama Y, Kim CA, Pastorino AC, Ceroni J, Lima PP, de Barros Dorna M, Honjo RS, Bertola D, Hamanaka K, Fujita A, Mitsuhashi S, Miyatake S, Takata A, Miyake N, Mizuguchi T, Matsumoto N. Primary immunodeficiency with chronic enteropathy and developmental delay in a boy arising from a novel homozygous RIPK1 variant. ***J Hum Genet***. 2019;64(9):955-60.
19. Den K, Kudo Y, Kato M, Watanabe K, Doi H, Tanaka F, Oguni H, Miyatake S, Mizuguchi T, Takata A, Miyake N, Mitsuhashi S, Matsumoto N. Recurrent NUS1 canonical splice donor site mutation in two unrelated individuals with epilepsy, myoclonus, ataxia and scoliosis - a case report. ***Bmc Neurol***. 2019;19(1):1-7.
20. Aoi H, Mizuguchi T, Ceroni JR, Kim VEH, Furquim I, Honjo RS, Iwaki T, Suzuki T, Sekiguchi F, Uchiyama Y, Azuma Y, Hamanaka K, Koshimizu E, Miyatake S, Mitsuhashi S, Takata A, Miyake N, Takeda S, Itakura A, Bertola DR, Kim CA, Matsumoto N. Comprehensive genetic analysis of 57 families with clinically suspected Cornelia de Lange syndrome. ***J Hum Genet***. 2019;64(10):967-78.
21. Mukai J, Cannavo E, Crabtree GW, Sun Z, Diamantopoulou A, Thakur P, Chang CY, Cai Y, Lomvardas S, Takata A, Xu B, Gogos JA. Recapitulation and Reversal of Schizophrenia-Related Phenotypes in Setd1a-Deficient Mice. ***Neuron***. 2019;104(3):471-87 e12.
22. Alkanaq AN, Hamanaka K, Sekiguchi F, Taguri M, Takata A, Miyake N, Miyatake S, Mizuguchi T, Matsumoto N. Comparison of mitochondrial DNA variants detection using short- and long-read sequencing. ***J Hum Genet***. 2019;64(11):1107-16.
23. Okamoto N, Takata A, Miyake N, Matsumoto N. RALA mutation in a patient with autism spectrum disorder and Noonan syndrome-like phenotype. ***Congenit Anom (Kyoto)***. 2019;59(6):195-6.

24. Hamanaka K, Sugawara Y, Shimoji T, Nordtveit TI, Kato M, Nakashima M, Saitsu H, Suzuki T, Yamakawa K, Aukrust I, Houge G, Mitsuhashi S, Takata A, Iwama K, Alkanaq A, Fujita A, Imagawa E, Mizuguchi T, Miyake N, Miyatake S, Matsumoto N. De novo truncating variants in PHF21A cause intellectual disability and craniofacial anomalies. ***Eur J Hum Genet***. 2019;27(3):378-83.

25. Saida K, Silva S, Solar B, Fujita A, Hamanaka K, Mitsuhashi S, Koshimizu E, Mizuguchi T, Miyatake S, Takata A, Miyake N, Matsumoto N. SOFT syndrome in a patient from Chile. ***Am J Med Genet A***. 2019;179(3):338-40.

26. Takata A, Nakashima M, Saitsu H, Mizuguchi T, Mitsuhashi S, Takahashi Y, Okamoto N, Osaka H, Nakamura K, Tohyama J, Haginoya K, Takeshita S, Kuki I, Okanishi T, Goto T, Sasaki M, Sakai Y, Miyake N, Miyatake S, Tsuchida N, Iwama K, Minase G, Sekiguchi F, Fujita A, Imagawa E, Koshimizu E, Uchiyama Y, Hamanaka K, Ohba C, Itai T, Aoi H, Saida K, Sakaguchi T, Den K, Takahashi R, Ikeda H, Yamaguchi T, Tsukamoto K, Yoshitomi S, Oboshi T, Imai K, Kimizu T, Kobayashi Y, Kubota M, Kashii H, Baba S, Iai M, Kira R, Hara M, Ohta M, Miyata Y, Miyata R, Takanashi JI, Matsui J, Yokochi K, Shimono M, Amamoto M, Takayama R, Hirabayashi S, Aiba K, Matsumoto H, Nabatame S, Shiihara T, Kato M, Matsumoto N. Comprehensive analysis of coding variants highlights genetic complexity in developmental and epileptic encephalopathy. ***Nat Commun***. 2019;10(1):2506.

27. Tsuchida N, Kirino Y, Soejima Y, Onodera M, Arai K, Tamura E, Ishikawa T, Kawai T, Uchiyama T, Nomura S, Kobayashi D, Taguri M, Mitsuhashi S, Mizuguchi T, Takata A, Miyake N, Nakajima H, Miyatake S, Matsumoto N. Haploinsufficiency of A20 caused by a novel nonsense variant or entire deletion of TNFAIP3 is clinically distinct from Behcet's disease. ***Arthritis Res Ther***. 2019;21(1):1-12.

28. Iwama K, Mizuguchi T, Takeshita E, Nakagawa E, Okazaki T, Nomura Y, Iijima Y, Kajiura 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, Saitsu H, Matsuishi T, Goto YI, Matsumoto N. Genetic landscape of Rett syndrome-like phenotypes revealed by whole exome sequencing. ***J Med Genet***. 2019;56(6):396-407.

29. Fujita A, Higashijima T, Shirozu H, Masuda H, Sonoda M, Tohyama J, Kato M, Nakashima M, Tsurusaki Y, Mitsuhashi S, Mizuguchi T, Takata A, Miyatake S, Miyake N, Fukuda M, Kameyama S, Saitsu H, Matsumoto N. Pathogenic variants of DYNC2H1, KIAA0556, and PTPN11 associated with hypothalamic hamartoma. ***Neurology***. 2019;93(3):e237-e51.

30. Hamanaka K, Takata A, Uchiyama Y, Miyatake S, Miyake N, Mitsuhashi S, Iwama K, Fujita A, Imagawa E, Alkanaq AN, Koshimizu E, Azuma Y, Nakashima M, Mizuguchi T, Saitsu H, Wada Y, Minami S, Katoh-Fukui Y, Masunaga Y, Fukami M, Hasegawa T, Ogata T, Matsumoto

N. MYRF haploinsufficiency causes 46,XY and 46,XX disorders of sex development: bioinformatics consideration. ***Hum Mol Genet***. 2019;28(14):2319-29.

31. Hamanaka K, Miyatake S, Koshimizu E, Tsurusaki Y, Mitsuhashi S, Iwama K, Alkanaq AN, Fujita A, Imagawa E, Uchiyama Y, Tawara N, Ando Y, Misumi Y, Okubo M, Nakashima M, Mizuguchi T, Takata A, Miyake N, Saitsu H, Iida A, Nishino I, Matsumoto N. RNA sequencing solved the most common but unrecognized NEB pathogenic variant in Japanese nemaline myopathy. ***Genet Med***. 2019;21(7):1629-38.
32. Takata A. Estimating contribution of rare non-coding variants to neuropsychiatric disorders. ***Psychiatry Clin Neurosci***. 2019;73(1):2-10.
33. Sekiguchi F, Tsurusaki Y, Okamoto N, Teik KW, Mizuno S, Suzumura H, Isidor B, Ong WP, Haniffa M, White SM, Matsuo M, Saito K, Phadke S, Kosho T, Yap P, Goyal M, Clarke LA, Sachdev R, McGillivray G, Leventer RJ, Patel C, Yamagata T, Osaka H, Hisaeda Y, Ohashi H, Shimizu K, Nagasaki K, Hamada J, Dateki S, Sato T, Chinen Y, Awaya T, Kato T, Iwanaga K, Kawai M, Matsuoka T, Shimoji Y, Tan TY, Kapoor S, Gregersen N, Rossi M, Marie-Laure M, McGregor L, Oishi K, Mehta L, Gillies G, Lockhart PJ, Pope K, Shukla A, Girisha KM, Abdel-Salam GMH, Mowat D, Coman D, Kim OH, Cordier MP, Gibson K, Milunsky J, Liebelt J, Cox H, El Chehadeh S, Toutain A, Saida K, Aoi H, Minase G, Tsuchida N, Iwama K, Uchiyama Y, Suzuki T, Hamanaka K, Azuma Y, Fujita A, Imagawa E, Koshimizu E, Takata A, Mitsuhashi S, Miyatake S, Mizuguchi T, Miyake N, Matsumoto N. Genetic abnormalities in a large cohort of Coffin-Siris syndrome patients. ***J Hum Genet***. 2019;64(12):1173-86.
34. Den K, Kato M, Yamaguchi T, Miyatake S, Takata A, Mizuguchi T, Miyake N, Mitsuhashi S, Matsumoto N. A novel de novo frameshift variant in SETD1B causes epilepsy. ***J Hum Genet***. 2019;64(8):821-7.
35. Mizuguchi T, Nakashima M, Moey LH, Ch'ng GS, Khoo TB, Mitsuhashi S, Miyatake S, Takata A, Miyake N, Saitsu H, Matsumoto N. A novel homozygous truncating variant of NECAP1 in early infantile epileptic encephalopathy: the second case report of EIEE21. ***J Hum Genet***. 2019;64(4):347-50.
36. Imagawa E, Yamamoto Y, Mitsuhashi S, Isidor B, Fukuyama T, Kato M, Sasaki M, Tanabe S, Miyatake S, Mizuguchi T, Takata A, Miyake N, Matsumoto N. PRUNE1-related disorder: Expanding the clinical spectrum. ***Clin Genet***. 2018;94(3-4):362-7.
37. Iwama K, Osaka H, Ikeda T, Mitsuhashi S, Miyatake S, Takata A, Miyake N, Ito S, Mizuguchi T, Matsumoto N. A novel SLC9A1 mutation causes cerebellar ataxia. ***J Hum Genet***. 2018;63(10):1049-54.
38. Imagawa E, Albuquerque EVA, Isidor B, Mitsuhashi S, Mizuguchi T, Miyatake S, Takata A, Miyake N, Boguszewski MCS, Boguszewski CL, Lerario AM, Funari MA, Jorge AAL, Matsumoto N. Novel SUZ12 mutations in Weaver-like syndrome. ***Clin Genet***. 2018;94(5):461-6.
39. Fukuda H, Imagawa E, Hamanaka K, Fujita A, Mitsuhashi S, Miyatake S, Mizuguchi T, Takata

A, Miyake N, Kramer U, Matsumoto N, Fattal-Valevski A. A novel missense SNAP25b mutation in two affected siblings from an Israeli family showing seizures and cerebellar ataxia. ***J Hum Genet***. 2018;63(5):673-6.

40. Iwama K, Takaori T, Fukushima A, Tohyama J, Ishiyama A, Ohba C, Mitsuhashi S, Miyatake S, Takata A, Miyake N, Ito S, Saitsu H, Mizuguchi T, Matsumoto N. Novel recessive mutations in MSTO1 cause cerebellar atrophy with pigmentary retinopathy. ***J Hum Genet***. 2018;63(3):263-70.
41. Tsuchida N, Nakashima M, Kato M, Heyman E, Inui T, Haginoya K, Watanabe S, Chiyonobu T, Morimoto M, Ohta M, Kumakura A, Kubota M, Kumagai Y, Hamano SI, Lourenco CM, Yahaya NA, Ch'ng GS, Ngu LH, Fattal-Valevski A, Hubshman MW, Orenstein N, Marom D, Cohen L, Goldberg-Stern H, Uchiyama Y, Imagawa E, Mizuguchi T, Takata A, Miyake N, Nakajima H, Saitsu H, Miyatake S, Matsumoto N. Detection of copy number variations in epilepsy using exome data. ***Clinical Genetics***. 2018;93(3):577-87.
42. Uchiyama Y, Ogawa Y, Kunishima S, Shiina M, Nakashima M, Yanagisawa K, Yokohama A, Imagawa E, Miyatake S, Mizuguchi T, Takata A, Miyake N, Ogata K, Handa H, Matsumoto N. A novel GFI1B mutation at the first zinc finger domain causes congenital macrothrombocytopenia. ***Brit J Haematol***. 2018;181(6):843-7.
43. Miyatake S, Kato M, Sawaishi Y, Saito T, Nakashima M, Mizuguchi T, Mitsuhashi S, Takata A, Miyake N, Saitsu H, Matsumoto N. Recurrent SCN3A p.Ile875Thr variant in patients with polymicrogyria. ***Ann Neurol***. 2018;84(1):159-61.
44. Takata A, Miyake N, Tsurusaki Y, Fukai R, Miyatake S, Koshimizu E, Kushima I, Okada T, Morikawa M, Uno Y, Ishizuka K, Nakamura K, Tsujii M, Yoshikawa T, Toyota T, Okamoto N, Hiraki Y, Hashimoto R, Yasuda Y, Saitoh S, Ohashi K, Sakai Y, Ohga S, Hara T, Kato M, Nakamura K, Ito A, Seiwa C, Shirahata E, Osaka H, Matsumoto A, Takeshita S, Tohyama J, Saikusa T, Matsuishi T, Nakamura T, Tsuboi T, Kato T, Suzuki T, Saitsu H, Nakashima M, Mizuguchi T, Tanaka F, Mori N, Ozaki N, Matsumoto N. Integrative Analyses of De Novo Mutations Provide Deeper Biological Insights into Autism Spectrum Disorder. ***Cell Rep***. 2018;22(3):734-47.
45. Iwama K, Iwata A, Shiina M, Mitsuhashi S, Miyatake S, Takata A, Miyake N, Ogata K, Ito S, Mizuguchi T, Matsumoto N. A novel mutation in SLC1A3 causes episodic ataxia. ***J Hum Genet***. 2018;63(2):207-11.
46. Tsuchida N, Nakashima M, Miyauchi A, Yoshitomi S, Kimizu T, Ganesan V, Teik KW, Ch'ng GS, Kato M, Mizuguchi T, Takata A, Miyatake S, Miyake N, Osaka H, Yamagata T, Nakajima H, Saitsu H, Matsumoto N. Novel biallelic SZT2 mutations in 3 cases of early-onset epileptic encephalopathy. ***Clin Genet***. 2018;93(2):266-74.
47. 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***. 2018;63(12):1223-9.

48. 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***. 2018;84(6):843-53.
49. 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, Saitsu H, Ogata K, Miyatake S, Matsumoto N. GRIN2D variants in three cases of developmental and epileptic encephalopathy. ***Clin Genet***. 2018;94(6):538-47.
50. Uchiyama Y, Yanagisawa K, Kunishima S, Shiina M, Ogawa Y, Nakashima M, Hirato J, Imagawa E, Fujita A, Hamanaka K, Miyatake S, Mitsuhashi S, Takata A, Miyake N, Ogata K, Handa H, Matsumoto N, Mizuguchi T. A novel CYCS mutation in the alpha-helix of the CYCS C-terminal domain causes non-syndromic thrombocytopenia. ***Clin Genet***. 2018;94(6):548-53.
51. Hamanaka K, Takahashi K, Miyatake S, Mitsuhashi S, Hamanoue H, Miyaji Y, Fukai R, Doi H, Fujita A, Imagawa E, Iwama K, Nakashima M, Mizuguchi T, Takata A, Miyake N, Takeuchi H, Tanaka F, Matsumoto N. Confirmation of SLC5A7-related distal hereditary motor neuropathy 7 in a family outside Wales. ***Clin Genet***. 2018;94(2):274-5.
52. Mizuguchi T, Nakashima M, Kato M, Okamoto N, Kurahashi H, Ekhilevitch N, Shiina M, Nishimura G, Shibata T, Matsuo M, Ikeda T, Ogata K, Tsuchida N, Mitsuhashi S, Miyatake 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***. 2018;27(8):1421-33.
53. Sekiguchi F, Nasiri J, Sedghi M, Salehi M, Hosseinzadeh M, Okamoto N, Mizuguchi T, Nakashima M, Miyatake S, Takata A, Miyake N, Matsumoto N. A novel homozygous DPH1 mutation causes intellectual disability and unique craniofacial features. ***J Hum Genet***. 2018;63(4):487-91.
54. Miyake N, Inaba M, Mizuno S, Shiina M, Imagawa E, Miyatake S, Nakashima M, Mizuguchi T, Takata A, Ogata K, Matsumoto N. A case of atypical Kabuki syndrome arising from a novel missense variant in HNRNPK. ***Clin Genet***. 2017;92(5):554-5.
55. Takata A, Matsumoto N, Kato T. Genome-wide identification of splicing QTLs in the human brain and their enrichment among schizophrenia-associated loci. ***Nat Commun***. 2017;8(1):1-11.
56. Iwama K, Mizuguchi T, Takanashi JI, 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 YI, 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;92(2):180-7.

57. Kasahara T, Ishiwata M, Kakiuchi C, Fuke S, Iwata N, Ozaki N, Kunugi H, Minabe Y, Nakamura K, Iwata Y, Fujii K, Kanba S, Ujike H, Kusumi I, Kataoka M, Matoba N, Takata A, Iwamoto K, Yoshikawa T, Kato T. Enrichment of deleterious variants of mitochondrial DNA polymerase gene (POLG1) in bipolar disorder. ***Psychiatry Clin Neurosci***. 2017;71(8):518-29.
58. Miyake N, Ozasa S, Mabe H, Kimura S, Shiina M, Imagawa E, Miyatake S, Nakashima M, Mizuguchi T, Takata A. A novel missense mutation affecting the same amino acid as the recurrent PACS1 mutation in Schuurs-Hoeijmakers syndrome. ***Clinical genetics***. 2017;93(4):929-30.
59. Takata A, Ionita-Laza I, Gogos JA, Xu B, Karayiorgou M. De Novo Synonymous Mutations in Regulatory Elements Contribute to the Genetic Etiology of Autism and Schizophrenia. ***Neuron***. 2016;89(5):940-7.
60. Kataoka M, Matoba N, Sawada T, Kazuno AA, Ishiwata M, Fujii K, Matsuo K, Takata A, Kato T. Exome sequencing for bipolar disorder points to roles of de novo loss-of-function and protein-altering mutations. ***Mol Psychiatry***. 2016;21(7):885-93.
61. Kasahara T, Takata A, Kato TM, Kubota-Sakashita M, Sawada T, Kakita A, Mizukami H, Kaneda D, Ozawa K, Kato T. Depression-like episodes in mice harboring mtDNA deletions in paraventricular thalamus. ***Mol Psychiatry***. 2016;21(1):39-48.
62. Takata A, Xu B, Ionita-Laza I, Roos JL, Gogos JA, Karayiorgou M. Loss-of-Function Variants in Schizophrenia Risk and SETD1A as a Candidate Susceptibility Gene. ***Neuron***. 2014;82(4):773-80.
63. Takata A, Iwayama Y, Fukuo Y, Ikeda M, Okochi T, Maekawa M, Toyota T, Yamada K, Hattori E, Ohnishi T, Toyoshima M, Ujike H, Inada T, Kunugi H, Ozaki N, Nanko S, Nakamura K, Mori N, Kanba S, Iwata N, Kato T, Yoshikawa T. A population-specific uncommon variant in GRIN3A associated with schizophrenia. ***Biol Psychiatry***. 2013;73(6):532-9.
64. Takata A, Kato M, Nakamura M, Yoshikawa T, Kanba S, Sano A, Kato T. Exome sequencing identifies a novel missense variant in RRM2B associated with autosomal recessive progressive external ophthalmoplegia. ***Genome Biol***. 2011;12(9):R92.
65. Takata A, Kim SH, Ozaki N, Iwata N, Kunugi H, Inada T, Ujike H, Nakamura K, Mori N, Ahn YM, Joo EJ, Song JY, Kanba S, Yoshikawa T, Kim YS, Kato T. Association of ANK3 with bipolar disorder confirmed in East Asia. ***Am J Med Genet B Neuropsychiatr Genet***. 2011;156B(3):312-5.
66. Takata A, Kawasaki H, Iwayama Y, Yamada K, Gotoh L, Mitsuyasu H, Miura T, Kato T, Yoshikawa T, Kanba S. Nominal association between a polymorphism in DGKH and bipolar disorder detected in a meta-analysis of East Asian case-control samples. ***Psychiat Clin Neuros***. 2011;65(3):280-5.
67. Takata A, Kakiuchi C, Ishiwata M, Kanba S, Kato T. Behavioral and gene expression analyses in heterozygous XBP1 knockout mice: Possible contribution of chromosome 11qA1 locus

to prepulse inhibition. ***Neurosci Res***. 2010;68(3):250-5.

68. Gotoh L, Mitsuyasu H, Kobayashi Y, Oribe N, Takata A, Ninomiya H, Stanton VP, Jr., Springett GM, Kawasaki H, Kanba S. Association analysis of adenosine A1 receptor gene (ADORA1) polymorphisms with schizophrenia in a Japanese population. ***Psychiatr Genet***. 2009;19(6):328-35.