

Curriculum Vitae

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

Team Leader (Laboratory Head)

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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. 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* 106:549-558, 2020
2. 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 S, 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* 106:13-25, 2020
3. 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* 64:1173-1186, 2019
4. Mukai J, Cannavo E, Crabtree GW, Sun ZY, Diamantopoulou A, Thakur P, Chang CY, Cai YF, Lomvardas S, Takata A, Xu B, Gogos JA: Recapitulation and Reversal of Schizophrenia-Related Phenotypes in Setd1a-Deficient Mice. *Neuron* 104:471-+, 2019
5. Okamoto N, Takata A, Miyake N, Matsumoto N: RALA mutation in a patient with autism spectrum disorder and Noonan syndrome-like phenotype. *Congenit Anom* 59:195-196, 2019
6. 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* 64:1107-1116, 2019

7. 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* 19:253, 2019
8. 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* 64:967-978, 2019
9. 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* 64:885-890, 2019
10. Uchiyama Y, Kim CA, Pastorino AC, Ceroni J, Lima PP, Dorna MD, 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* 64:955-960, 2019
11. 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* 64:821-827, 2019
12. 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* 93:E237-E251, 2019
13. 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* 28:2319-2329, 2019

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15. 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 J, 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* 10:2506, 2019
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18. 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* 64:347-350, 2019
19. 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* 179:338-340, 2019
20. 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* 27:378-383, 2019
21. Takata A: Estimating contribution of rare non-coding variants to neuropsychiatric disorders. *Psychiatry Clin Neurosci* 73:2-10, 2019 (review)
22. 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* 84:843-853, 2018
23. 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* 63:1223-1229, 2018
24. 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* 94:538-547, 2018
25. Uchiyama Y, Yanagisawa K, Kunishima S, Shiina M, Ogawa Y, Nakashima M, Hirato J,

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31. 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. *Br J Haematol* 181:843-847, 2018
32. 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* 63:673-676, 2018
33. 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* 27:1421-1433, 2018
34. 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* 63:487-491, 2018
35. Miyake N, Ozasa S, Mabe H, Kimura S, Shiina M, Imagawa E, Miyatake S, Nakashima M, Mizuguchi T, Takata A, Ogata K, Matsumoto N: A novel missense mutation affecting the same amino acid as the recurrent PACS1 mutation in Schuurs-Hoeijmakers syndrome. *Clin Genet* 93:929-930, 2018
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37. 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. *Clin Genet* 93:577-587, 2018
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