CURRICULUM VITAE

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EDUCATION

1980 - 1984 **B.Sc.** (March 1984) Molecular Biology,
Faculty of Science, **Kyoto University**, Kyoto, Japan.

1989 - 1992 **Ph.D.** (May 1992) Medical Genetics
Faculty of Medicine, **Osaka University**, Osaka, Japan.

1994 - 1996 Postdoctoral fellowship,
Medical Genetics, Cedars-Sinai Medical Center,
UCLA School of Medicine, California, USA.

RESEARCH AND PROFESSIONAL EXPERIENCES

1984 - 1989 Researcher
Department of Biochemistry, General Institute, Toyobo Co., Ltd.,
Shiga, Japan.

1989 - 1994 Researcher
Department of Biochemistry, Cancer Institute
Tokyo, Japan.

1994 - 1996 Research fellow
Department of Medical Genetics, Cedars-Sinai Medical Center,
UCLA School of Medicine, California, USA.

1996 - 1997 Assistant professor-Adjunct, Pediatrics of UCLA School of Medicine.
Research Scientist, Medical Genetics, Cedars-Sinai Reserch Institute, California, USA.

1997 - present Laboratory Head, Laboratory for Neurogenetics,
RIKEN Brain Science Institute, Saitama, Japan

TEACHING EXPERIENCES

2008 - present Invited Professor (Concurrent position), Saitama University, saitama, Japan

MEMBERSHIP OF ACADEMIC SOCIETIES

Society for Neuroscience

American Society of Human Genetics
Japanese Society of Human Genetics
Japanese Society of Neuroscience
Japanese Society of Epilepsy
Japanese Society of Child Neurology
Infantile Seizure Society

EDITORIAL SERVICES

2007~ Editorial Advisor for the Japanese journal "Epilepsy", Medical Review, Co.Ltd.

AWARDS

2009 Research Award: The Japan Epilepsy Research Foundation

PUBLICATIONS

1. Amano K, Fujii M, Arata S, Tojima T, Ogawa M, Shimohata A, Furuichi T, Itohara S, Kamiguchi H, Korenberg J.R, Arata A, Yamakawa K (2009) DSCAM deficiency causes loss of pre-inspiratoru neuron synchroneity and perinatal death. ***The Journal of Neuroscience*** **29**:2984-2996
2. Suzuki T, Miyamoto H, Nakahari T, Inoue I, Suemoto T, Jiang B, Hirota Y, Itohara S, Saido TC, Tsumoto T, Sawamoto K, Hensch TK, Delgado-Escueta AV, and Yamakawa K. (2009) Efhc1 deficiency causes spontaneous myoclonus and increased seizure susceptibility. ***Human Molecular Genetics*** **18**: 1099-1109.
3. Fukuyo N, Haginoya K, Togashi N, Uematsu M, Kitamura T, Kakisaka Y, Ishitobi M, Wakusawa K, Iinuma K, Oguni H, Yanakawa K, Tsuchiya S. (2009) Ictal Vomiting as an Initial Symptom of Severe Myoclonic Epilepsy in Infancy: A Case Report. ***Journal of Child Neurology*** **24**:228-230.
4. Bai D, Bailey JN, Durón RM, Alonso ME, Medina MT, Martínez-Juárez IE, Suzuki T, Machado-Salas J, Ramos-Ramírez R, Tanaka M, Ortega RH, López-Ruiz M, Rasmussen A, Ochoa A, Jara-Prado A, Yamakawa K, Delgado-Escueta AV. (2008) DNA variants in coding region of EFHC1: SNPs do not associate with juvenile myoclonic epilepsy. **Epilepsia**, Sep 20. (Epub ahead of print)
5. Miyama S, Goto T, Inoue Y, Yamakawa K. (2008) Monozygotic twins with severe myoclonic epilepsy in infancy discordant for clinical features. ***Pediatric Neurology*** **39**: 120-2.
6. Medina M.T, Suzuki T, Alonso M.E, Durón R.M, Martínez-Juárez I.E, Bailey J.N, Bai D, Inoue Y, Yoshimura I, Kaneko S, Montoya M.C, Ochoa A, Jara Prado A, Tanaka M, Machado-Salas J, Fujimoto S, Ito M, Hamano S, Sugita K, Ueda Y, Osawa M, Oguni H, Rubio-Donnadieu F, Yamakawa K., Delgado-Escueta A.V (2008) Novel mutations in *Myoclonin1/EFHC1* in sporadic and familial juvenile myoclonic epilepsy. ***Neurology*** **70**: 2137-2144.
7. Suzuki T, Inoue I, Yamagata T, Morita N, Furuichi T, Yamakawa K. (2008) Sequential expression of Efhc1/myoclonin1 in choroid plexus and ependymal cell cilia. ***Biochemical and Biophysical Research Communications*** **367**:226-233.
8. Amano K, Yamada K, Iwayama Y, Detera-Wadleigh SD, Hattori E, Toyota T, Tokunaga K, Yoshikawa T, Yamakawa K. (2008) Association study between the Down syndrome cell adhesion molecule (*DSCAM*) gene and bipolar disorder. ***Psychiatric Genetics*** **18**:1-10.
9. Ogiwara I, Miyamoto H, Morita N, Atapour N, Mazaki E, Inoue I, Takeuchi T, Itohara S, Yanagawa Y, Obata K, Furuichi T, Hensch TK, Yamakawa K. (2007) $Na_v1.1$ Localizes to Axons of

Parvalbumin-Positive Inhibitory Interneurons: a Circuit Basis for Epileptic Seizures in Mice Carrying an *Scn1a* Gene Mutation. ***The Journal of Neuroscience*** **27**: 5903-5914.

10. Osaka H, Ogiwara I, Mazaki E, Okamura N, Yamashita S, Iai M, Yamada M, Kurosawa K, Iwamoto H, Yasui-Furukori N, Kaneko S, Fujiwara T, Inoue Y, Yamakawa K. (2007) Patients with a sodium channel alpha 1 gene mutation show wide phenotypic variation ***Epilepsy Research*** **75**: 46-51.
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12. Mittal S, Dubey D, Yamakawa K, Ganesh S. (2007) Lafora disease proteins malin and laforin are recruited to aggresomes in response to proteasomal impairment. ***Human Molecular Genetics*** **16**:753-762.
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14. Singh B, Ogiwara I, Kaneda M, Tokonami N, Mazaki E, Baba K, Matsuda K, Inoue Y, Yamakawa K. (2006) A K_v4.2 truncation mutation In a patient with temporal lobe epilepsy. ***Neurobiology of Disease*** **24**:245-253.
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the pedigree of several patients with complex febrile seizures. ***Epilepsy Research*** **63**:151-156.

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associations with schizophrenia. ***American Journal of Medical Genetics*** **114**:304-309.

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43. Agarwala KL, Ganesh S, Tsutsumi U, Suzuki T, Amano K, Yamakawa K (2001) Cloning and Functional Characterization of DSCAML1, a Novel DSCAM-Like Cell Adhesion Molecule that Mediates Homophilic Intercellular Adhesion. ***Biochemical and Biophysical Research Communications*** **285**:760-772.
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48. Sugawara T, Tsurubuchi Y, Agarwala KL, Ito M, Fukuma G, Mazaki-Miyazaki E, Nagafuji H, Noda M, Imoto K, Wada K, Mitsudome A, Kaneko S, Montal M, Nagata K, Hirose S, Yamakawa K (2001) A missense mutation of the Na^+ channel □□□ subunit gene $Na_v1.2$ in a patient with febrile

and afebrile seizures causes channel dysfunction. ***Proc Natl Acad Sci USA* 98**: 6384-6389. (Erratum in: Proc Natl Acad Sci U S A 2001 Aug 28;98(18):10515.)

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50. Agarwala KL, Ganesh S, Amano K, Suzuki T, Yamakawa K (2001) DSCAM, a highly conserved gene in mammals, expressed in differentiating mouse brain. ***Biochemical and Biophysical Research Communications* 281**: 697-705.
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that maps to chromosome band 7q31. ***Genomics* 35**:456-465.

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Main Review Articles (English only - Japanese articles were excluded)

1. Yamakawa K (2009) Molecular basis of severe myoclonic epilepsy in infancy. **Brain Development** Feb 7;. [Epub ahead of print]
2. Yamakawa K (2006) Na channel gene mutations in epilepsy- The functional consequences. **Epilepsy Research 70**:218-S222.
3. Yamakawa K (2005) Epilepsy and sodium channel gene mutations: Gain or loss of function? **NeuroReport 16**:1-3.

Main Invited Lectures in International Meetings; Presentation Titles & Places

1. Yamakawa K (October 4-6, 2009 - scheduled) "Molecular and cellular basis of Dravet syndrome", International symposium for severe myoclonic epilepsy in infancy, Verona, **Italy**.
2. Yamakawa K (March 20-29, 2009) "Circuits for seizures in mice with SCN1A mutaions", Jasper's Basic Mechanisms of the Epilepsies 4th International Workshop, Yosemite National Park, California, **USA**.
3. Yamakawa K. (April 10-11, 2008) "Molecular basis of severe myoclonic epilepsy", International

Symposium for Febrile Seizures and Related Condition. Ohtsu, Shiga, **Japan**.

4. Yamakawa K. (July 6-11, 2007) How to go about it and what are potential clinical applications? the27th International Epilepsy Congress, **Singapore**.
5. Yamakawa K (August 14-16, 2006) "EFHC1 mutations and juvenile myoclonic epilepsy". 2nd Pacific Rim Brain Conference Queensland, **Australia**.
6. Yamakawa K (June 8-11, 2006) For the plenary session "Epilepsy and sodium channel gene mutations: gain or loss of function". For parallel session"Development of The Molecular Genetics of Epilepsy" Gyeongju-City, Gyeongsangbuk-Do, **Korea**.
7. Yamakawa K (November 3-6, 2005) "Mutations in EFHC1 cause juvenile myoclonic epilepsy". 7th China-India-Japan-Korea Joint Workshop on Neurobiology and Neuroinformatics (NBNI-2005) Xiamen, **China**.
8. Yamakawa K (April 29-May 1, 2005) "Sodium channel mutations- gain or loss of function?" International symposium on epileptic syndromes in infancy and early childhood. Tokyo, **Japan**.
9. Yamakawa K (November 20-23, 2003) "Voltage-gated sodium channel and epilepsy". 5th China-India-Japan-Korea Joint Workshop on Neurobiology and Neuroinformatics (NBNI-2003) Daejeon, **Korea**.
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12. Yamakawa K (September 21-26, 2002) "Mutations of sodium channel in GEFS+ and SMEI". Joint Congress of ICNA and AOCNA 2002, Beijing, **China**.
13. Yamakawa K (September 11-14, 2002) "Genetics of myoclonic epilepsies". The 4th Asian and Oceanian Epilepsy Congress organized in collaboration with The 36th Annual Congress of the Japan Epilepsy Society, Karuizawa, **Japan**.