



- **Name:** Naomichi Matsumoto, M.D., Ph.D.
- **Current Position:**
Professor and Chair, Department of Human Genetics, Yokohama City University Graduate School of Medicine
- **Country:** Japan
- **Educational Background:**
 - 1986 Kyushu University School of Medicine, M.D.
 - 1997 Graduate School of Medical Science, Nagasaki University School of Medicine, Ph.D.
- **Professional Experiences:**
 - 1986-1993 Obstetrician & Gynecologist at Kyshu University related hospitals
 - 1993-1997 Graduate School of Nagasaki University, Department of Human Genetics
 - 1997-1998 Post Doctoral Fellow at Department of Human Genetics, The University of Chicago
 - 1998-2000.1 Research associate at Department of Human Genetics, The University of Chicago
 - 2000-2003 Associate professor at Department of Human Genetics, Nagasaki University School of Medicine
 - 2003-present Professor and Chair at Department of Human Genetics, Yokohama City University Graduate School of Medicine
- **Professional Organizations**
Member of Japan Society of Human Genetics (Councilor 2003-) (Director 2011-)
EDITORIAL BOARD
 - J Hum Genet (Editor-in-Chief 2014-)
 - Clin Genet (2005-)
 - Am J Med Genet Part A (2008-)
 - Hum Genet (2015-)



•Honors and awards:

- | | |
|------|---|
| 2003 | Japan Society of Human Genetics Award for Young Scientist |
| 2011 | Japan Society of Human Genetics Award |

•Main Scientific Publications:

- Nakashima M, Saitsu H, Tohyama J, Kato M, Shiina M, Takei N, Kitaura H, Shirozu H, Masuda H, Watanabe K, Ohba C, Tsurusaki Y, Miyake N, Takebayashi H, Ogata K, Kameyama S, Kakita A, *Matsumoto N. Somatic Mutations in the MTOR Gene Cause Focal Cortical Dysplasia Type IIb. *Ann Neurol* 78(3):375-386, 2015.
- Miyatake S, *Matsumoto N (*: correspondence). Clinical exome sequencing in neurology practice. (News & View) *Nat Rev Neurol* 10(12):676-678, 2014.
- Tsurusaki Y, Ohashi H, Phadke S, Koshimizu E, Kou I, Shiina M, Suzuki T, Okamoto N, Imamura S, Yamashita M, Watanabe S, Yoshiura K-i, Kodera H, Miyatake S, Nakashima N, Saitsu H, Ogata K, Ikegawa S, Miyake N, and *Matsumoto N. *De novo SOX11* mutations cause Coffin-Siris syndrome. *Nat Commun* 5:4011, 2014.
- *Saitsu H[#], Nishimura T[#], Muramatsu K[#] ([#] denotes equal contribution), 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, *Matsumoto N (*: co-corresponding). *De novo* mutations in the autophagy gene *WDR45* cause static encephalopathy of childhood with neurodegeneration in adulthood. *Nat Genet* 45(4): 445-449, 2013.
- Tsurusaki Y, Okamoto N, Ohashi H, Kosho T, Imai Y, Hibi-Ko Y, Kaname T, Naritomi K, Kawame H, Wakui K, Fukushima Y, Homma T, Kato M, Hiraki Y, Yamagata T, Yano S, Mizuno S, Sakazume S, Ishii T, Nagai T, Shiina M, Ogata K, Ohta T, Niikawa N, Miyatake S, Okada I, Mizuguchi T, Doi H, Saitsu H, *Miyake N, *Matsumoto N (*: co-corresponding). Mutations affecting components of the SWI/SNF complex cause Coffin-Siris syndrome. *Nat Genet* 44(4):376-378, 2012
- Ng S, Bigham A, Buckingham K, Hannibal M, McMillin M, Gildersleeve H, Beck A, Tabor H, Cooper G, Mefford H, Lee C, Turner E, Smith J, Rieder M, Yoshiura K, Matsumoto N, Ohta T, Niikawa N, Nickerson D, *Bamshad M, *Shendure J. Exome sequencing identifies



- Saitsu H, Kato M, Mizuguchi T, Hamada K, Osaka H, Tohyama J, Uruno K, Kumada S, Nishiyama K, Nishimura A, Okada I, Yoshimura Y, Hirai S-i, Kumada T, Hayasaka K, Fukuda A, Ogata K, Matsumoto N. *De novo* mutations in the gene encoding STXBP1 (MUNC18-1) cause early infantile epileptic encephalopathy. *Nat Genet* 40(6): 782-788, 2008
- Niihori T, Aoki Y, Narumi Y, Neri G, Cavé H, Alain Verloes A, Okamoto N, Hennekam RCM, Gillessen-Kaesbach G, Wieczorek D, Kavamura MI, Kurosawa K, Ohashi H, Wilson L, Heron D, Bonneau D, Corona G, Kaname T, Naritomi K, Baumann C, Matsumoto N, Kato K, Kure S, Matsubara Y. Germline *KRAS* and *BRAF* mutations in cardio-facio-cutaneous (CFC) syndrome. *Nat Genet* 38(3): 294-296, 2006
- Mizuguchi T, Collod-Beroud G, Akiyama T, Abifadel M, Harada N, Morisaki T, Allard D, Varret M, Claustres M, Morisaki H, Ihara M, Kinoshita A, Yoshiura K-i, Junien C, Kajii T, Jondeau G, Ohta T, Kishino T, Furukawa Y, Nakamura Y, Niikawa N, Boileau C, Matsumoto N. Heterozygous *TGFBR2* mutations in Marfan syndrome. *Nat Genet* 36(8): 855-860, 2004
- Yamada K, Andrews C, Chan W-M, McKeown CA, Magli A, de Berardinis T, Loewenstein A, Lazar M, O'Keefe M, Letson R, London A, Ruttum M, Matsumoto N, Saito N, Lisa Morris, Del Monte M, Johnson RH, Uyama E, Houtman WA, de Vries B, Carlow TJ, Blaine L Hart BL, Krawiecki N, Shoffner J, Vogel MC, Katowitz J, Goldstein SM, Levin AV, Sener EC, Ozturk BT, Akarsu AN, Brodsky MC, Hanisch F, Cruse RP, Zubcov AA, Robb RM, Roggenkämper P, Gottlob I, Kowal L, Battu R, Traboulsi BI, Franceschini P, Newlin A, Demer JL, Engle EC. Heterozygous mutations of the kinesin KIF21A in congenital fibrosis of the extraocular muscles type 1 (CFEOM1). *Nat Genet* 35 (4): 318-321, 2003
- Kurotaki N, Imaizumi K, Harada N, Masuno M, Kondoh T, Nagai T, Ohashi H, Naritomi K, Tsukahara M, Makita Y, Sugimoto T, Sonoda T, Hasegawa T, Chinen Y, Tomita H-A, Kinoshita A, Mizuguchi T, Yoshiura K-I, Ohta T, Kishino T, Fukushima Y, Niikawa N, Matsumoto N. Haploinsufficiency of the *NSD1* gene causes Sotos syndrome. *Nat Genet* 30 (4): 365-366, 2002