

Melissa B. Ramocki, MD

Neurologist with additional fellowship training in Otoneurology.

Education & Medical Training

Jan 2014-Dec 2014 Fellow, Otoneurology, Harvard Medical School (MEEI)
July 2005-June 2008 Resident, Pediatric Neurology, Baylor College of Medicine
July 2004-June 2005 Resident Adult Neurology, Baylor College of Medicine
June 2003-June 2004 Intern, Department of Pediatrics, Baylor College of Medicine
Sept 1999-June 2003 Medical Doctor,
The University of Chicago Pritzker School of Medicine; Awarded the Peter Huttenlocher and Douglas Buchanan Prize in
Neurology for best medical student clinician researcher
Aug 1994-May 1999 Doctor of Philosophy, Department of Biological Sciences, Purdue University
Jan 1991-Dec 1993 Bachelor of Science, Summa Cum Laude, Phi Beta Kappa, University of Rhode Island

Hospital Privileges

Rhode Island Hospital: Consulting

Board Certification

2009-present Diplomate of the American Board of Psychiatry and Neurology with Special Qualification in Child Neurology

Professional Affiliations

Child Neurology Society
American Academy of Neurology
Rhode Island Medical Society

Publications

1. Ramocki, M. B., Johnson, S. E., White, M. A., Ashendel, C. L., Konieczny, S. F., and Taparowsky, E. J. (1997). Signaling through mitogen-activated protein kinase and Rac/Rho does not duplicate the effects of activated Ras on skeletal myogenesis. *Molecular and Cellular Biology*. 17:3547-3555.
2. Weyman, C. M., Ramocki, M. B., Taparowsky, E. J., and Wolfman, A. (1997). Distinct signaling pathways regulate transformation and inhibition of skeletal muscle differentiation by oncogenic Ras. *Oncogene*. 14:697-704.
3. Ramocki, M. B., White, M. A., Konieczny, S. F., and Taparowsky, E. J. (1998). A role for RalGDS and a novel Ras effector in the Ras-mediated inhibition of skeletal myogenesis. *Journal of Biological Chemistry*. 273:17696-17701.
4. Cardoso, C., Leventer, R. J., Matsumoto, N., Kuc, J. A., Ramocki, M. B., Mewborn, S. K., Dudliceck, L. K., May, L. F., Mills, P. L., Das, S., Pilz, D. T., Dobyns, W. B., and Ledbetter, D. H. (2000). The location and type of mutation predict malformation severity in isolated lissencephaly caused by abnormalities within the LIS1 gene. *Human Molecular Genetics*. 9(20):3019-3028.
5. Mitin, N., Ramocki, M. B., Konieczny, S. F., and Taparowsky, E. J. (2001). Ras Regulation of Skeletal Muscle Differentiation and Gene Expression. *Methods in Enzymology*. 333:232-247.
6. Matsumoto, N., Leventer, R. J., Kuc, J. A., Mewborn, S. K., Dudliceck, L. L., Ramocki, M. B., Pilz, D. T., Mills, P. L., Das, S., Ross, M. E., Ledbetter, D. H., and Dobyns, W. B. (2001). Mutation

analysis of the DCX gene and genotype/phenotype correlation in subcortical band heterotopia. *European Journal of Human Genetics*. 9:5-12.

7. Ramocki, M. B., Dowling, J., Grinberg, I., Kimonis, V. E., Cardoso, C., Gross, A., Chung, J., Martin, C. L., Ledbetter, D. H., Dobyns, W. B., and Millen, K. J. (2003). Reciprocal fusion transcripts of two novel Zn-finger genes in a female with absence of the corpus callosum, ocular colobomas and a balanced translocation between chromosomes 2p24 and 9q32. *European Journal of Human Genetics*. 11(7):527-534.
8. Mitin, N. Y., Ramocki, M. B., Zullo, A. J., Konieczny, S. F., and Taparowsky, E. J. (2004). Identification and characterization of Rain, a novel Ras-interacting protein with a unique subcellular localization. *Journal of Biological Chemistry*. 279(21):22353-22361.
9. Ramocki, M. B., Chapieski, L., McDonald, R. O., Fernandez, F., and Malphrus, A. D. (2008). SCA2 presenting with cognitive regression in childhood. *Journal of Child Neurology*. 23(9):999-1001.
10. Ramocki, M. B. and Zoghbi, H. Y. (2008). Failure of neuronal homeostasis results in common neuropsychiatric phenotypes. *Nature*. 455(7215):912-918.
11. Carvalho, C. M. B., Zhang, F., Liu, P., Patel, A., Sahoo, T., Bacino, C., Shaw, C., Peacock, S., Pursley, A., Tavyev, Y. J., Ramocki, M. B., Nawara, M., Obersztyn, E., Vianna-Morgante, A. M., Stankiewicz, P., Zoghbi, H. Y., Cheung, S. W., and Lupski, J. R. (2009). Complex rearrangements in patients with duplications of MECP2 can occur by fork stalling and template switching. *Human Molecular Genetics*. 18(12):2188-2203.
12. Ramocki, M. B., Peters, S. U., Tavyev, Y. J., Zhang, F., Carvalho, C. M. B., Schaaf, C. P., Richman, R., Fang, P., Glaze, D. G., Lupski, J. R., and Zoghbi, H. Y. (2009). Autism and other neuropsychiatric symptoms are prevalent in individuals with the MECP2 duplication syndrome. *Annals of Neurology*. 66(6):771-782.
13. Ramocki, M. B., Tavyev, Y. J., and Peters, S. U. (2010). The MECP2 duplication syndrome. *American Journal of Medical Genetics Part A*. 152A:1079-1088.
14. El-Hakam, L. M., Ramocki, M. B., Riviello, J, and Illner, A. (2010). Hyperperfusion on MR Imaging in Acute Chemotherapy-Related Leukoencephalopathy. *Journal of Child Neurology*. 25(6):776-779.
15. Ramocki, M.B., Bartnik, M., Szafranski, P., Kolodziejska, K.E., Xia, Z., Bravo, J., Miller, G.S., Rodriguez, D.L., Williams, C.A., Bader, P.I., Szczepanik, E., Mazurczak, T., Antczak-Marach, D., Coldwell, J.G., Akman, C.I., McAlmon, K., Cohen, M.P., McGrath, J., Roeder, E., Mueller, J., Kang, S-H.L., Bacino, C.A., Patel, A., Bocian, E., Shaw, C., Cheung, S.W., Mazurczak, T., and Stankiewicz, P. (2010). Recurrent Distal 7q11.23 Deletion Including HIP1 and YWHAG Identified in Patients with Intellectual Disabilities, Epilepsy, and Neurobehavioral Problems. *American Journal of Human Genetics*. 10;87(6):857-865.
16. Breman, A.M., Ramocki, M.B., Kang, S.H., Williams, M., Freedenberg, D., Patel, A., Bader, P.I., and Cheung, S.W. (2011). MECP2 duplications in six patients with complex sex chromosome rearrangements. *European Journal of Human Genetics*. 19(4):409-415.
17. Ansari, S.A., El-Hakam, L.M., Clark, G.D., Hunter, J.V., and Ramocki, M.B. (2011). Bilateral in utero Cerebellar Infarction. *Journal of Child Neurology*. 26(7):895-899.
18. Campbell, I.M., Kolodziejska, K.E., Quach, M., Wolf, V.L., Cheung, S.W., Lalani, S.R., Ramocki, M.B., and Stankiewicz, P. (2011). TGFBR2 deletion in a 20 month-old female with developmental delay and microcephaly. *American Journal of Medical Genetics Part A*. 155A(6):1442-1447.

19. Ramocki, M.B., Scaglia, F., Stankiewicz, P., Belmont, J.W., Jones, J.Y., and Clark, G.D. (2011). Recurrent Partial Rhombencephalosynapsis and Holoprosencephaly in Siblings with Mutation of ZIC2. *American Journal of Medical Genetics Part A*. 155A(7):1574-1580.
20. Murdock, D., Clark, G.D., Bainbridge, M., Gibbs, R., and Ramocki, M.B. (2011). Whole-exome sequencing identifies compound heterozygous mutations in WDR62 in siblings with recurrent polymicrogyria. *American Journal of Medical Genetics Part A*. 155A(9):2071-2077.
21. Carvalho, C.M.B., Ramocki, M.B., Pehlivan, D., Franco, L.M., Pivnick, E., Hines-Dowell, S., Seaver, L., Friebling, L., Lee, S., Smith, R., del Gaudio, D., McCall, A., Withers, M., Liu, P., Cheung, S.W., Belmont, J.W., Hastings, P.J., Zoghbi, H.Y., and Lupski, J.R. (2011). Inverted genomic segments and complex triplication rearrangements are mediated by inverted repeats in the human genome. *Nature Genetics*. 43(11):1074-1081.
22. The Simons VIP Consortium. (2012). Simons Variation in Individuals Project (Simons VIP): A Genetics-First Approach to Studying Autism Spectrum and Related Neurodevelopmental Disorders. *Neuron*. 73(6):1063-1067. (Consortium member collaborator.)
23. Hanchard, N.A., Carvalho, C.M., Bader, P., Thome, A., Omo-Griffith, L., Del Gaudio, D., Pehlivan, D., Fang, P., Schaaf, C.P., Ramocki, M.B., Lupski, J.R., Cheung, S.W. (2012). A partial MECP2 duplication in a mildly affected adult male: a putative role for the 3' untranslated region in the MECP2 duplication phenotype. *BMC Med Genet*. 13(1):71.
24. Zufferey, F., Sherr, E.H., Beckmann, N.D., Hanson, E., Maillard, A.M., Hippolyte, L., Macé, A., Ferrari, C., Kutalik, Z., Andrieux, J., Aylward, E., Barker, M., Bernier, R., Bouquillon, S., Conus, P., Delobel, B., Faucett, W.A., Goin-Kochel, R.P., Grant, E., Harewood, L., Hunter, J.V., Lebon, S., Ledbetter, D.H., Martin, C.L., Mannik, K., Martinet, D., Mukherjee, P., Ramocki, M.B., Spence, S.J., Steinman, K., Tjernagel, J., Spiro, J.E., Reymond, A., Beckmann, J.S., Chung, W.K., Jacquemont, S. (2012). A 600 kb deletion syndrome at 16p11.2 leads to energy imbalance and neuropsychiatric disorders. *Journal of Medical Genetics*. 49(10):660-8.
25. Yang, T., Ramocki, M.B., Neul, J.L., Lu, W., Roberts, L., Knight, J., Ward, C.S., Zoghbi, H.Y., Kheradmand, F., Corry, D.B. (2012). Overexpression of methyl-CpG binding protein 2 impairs T(H)1 responses. *Sci Transl Med*. 4(163):163ra158.
26. Peters, S.U., Hundley, R.J., Wilson, A.K., Warren, Z., Vehorn, A., Carvalho, C.M., Lupski, J.R., Ramocki, M.B. (2012). The Behavioral Phenotype in MECP2 Duplication Syndrome: A Comparison with Idiopathic Autism. *Autism Res*. 6(1):42-50.
27. Peters, S.U., Hundley, R.J., Wilson, A.K., Carvalho, C.M., Lupski, J.R., Ramocki, M.B. (2013). Regression Timing and Associated Features in MECP2 Duplication Syndrome. *J Autism Dev Disord*. 43(10):2484-90.
28. Wiszniewski, W., Hunter, J.V., Hanchard, N.A., Willer, J.R., Shaw, C., Tian, Q., Illner, A., Wang, X., Cheung, S.W., Patel, A., Campbell, I.M., Hixson, P., Ester, A.R., Azamian, M.S., Potocki, L., Zapata, G., Hernandez, P.P., Ramocki, M.B., Santos-Cortez, R.L., Wang, G., York, M.K., Justice, M.J., Chu, Z.D., Bader, P.I., Omo-Griffith, L., Madduri, N.S., Scharer, G., Crawford, H.P., Yanatatsaneejit, P., Eifert, A., Kerr, J., Bacino, C.A., Franklin, A.I., Goin-Kochel, R.P., Simpson, G., Immken, L., Haque, M.E., Stosic, M., Williams, M.D., Morgan, T.M., Pruthi, S., Omary, R., Boyadjiev, S.A., Win, K.K., Thida, A., Hurles, M., Hibberd, M.L., Khor, C.C., Van Vinh Chau, N., Gallagher, T.E., Mutirangura, A., Stankiewicz, P., Beaudet, A.L., Maletic-Savatic, M., Rosenfeld, J.A., Shaffer, L.G., Davis, E.E., Belmont, J.W., Dunstan, S., Simmons, C.P., Bonnen, P.E., Leal, S.M., Katsanis, N., Lupski, J.R., Lalani, S.R. (2013). TM4SF20 Ancestral Deletion and Susceptibility to a Pediatric Disorder of Early Language Delay and Cerebral White Matter Hyperintensities. *Am J Hum Genet*. 93(2):197-210.

29. Campbell, I.M., Rao, M., Arredondo, S.D., Lalani, S.R., Xia, Z., Kang, S.H., Bi, W., Breman, A.M., Smith, J.L., Bacino, C.A., Beaudet, A.L., Patel, A., Cheung, S.W., Lupski, J.R., Stankiewicz, P., Ramocki, M.B., Shaw, C.A. (2013). Fusion of Large-Scale Genomic Knowledge and Frequency Data Computationally Prioritizes Variants in Epilepsy. *PLoS Genet.* Sep;9(9).
30. Carvalho, C.M., Pehlivan, D., Ramocki, M.B., Fang, P., Alleva, B., Franco, L.M., Belmont, J.W., Hastings, P.J., Lupski, J.R. Replicative mechanisms for CNV formation are error prone. (2013). *Nat Genet.* 45(11):1319-26.
31. Carvalho, C.M., Vasanth, S., Shinawi, M., Russell, C., Ramocki, M.B., Brown, C.W., Graakjaer, J., Skytte, A.B., Vianna-Morgante, A.M., Krepischi, A.C., Patel, G.S., Immken, L., Aleck, K., Lim, C., Cheung, S.W., Rosenberg, C., Katsanis, N., Lupski, J.R. (2014). Dosage changes of a segment at 17p13.1 lead to intellectual disability and microcephaly as a result of complex genetic interaction of multiple genes. *Am J Hum Genet.* 6;95(5):565-78.
32. D'Angelo, D., Lebon, S., Chen, Q., Martin-Brevet, S., Snyder, L.G., Hippolyte, L., Hanson, E., Maillard, A.M., Faucett, W.A., Macé, A., Pain, A., Bernier, R., Chawner, S.J., David, A., Andrieux, J., Aylward, E., Baujat, G., Caldeira, I., Conus, P., Ferrari, C., Forzano, F., Gérard, M., Goin-Kochel, R.P., Grant, E., Hunter, J.V., Isidor, B., Jacqueline, A., Jønch, A.E., Keren, B., Lacombe, D., Le Caignec, C., Martin, C.L., Männik, K., Metspalu, A., Mignot, C., Mukherjee, P., Owen, M.J., Passeggeri, M., Rooryck-Thambo, C., Rosenfeld, J.A., Spence, S.J., Steinman, K.J., Tjernagel, J., Van Haelst, M., Shen, Y., Draganski, B., Sherr, E.H., Ledbetter, D.H., van den Bree, M.B., Beckmann, J.S., Spiro, J.E., Reymond, A., Jacquemont, S., Chung, W.K.; Cardiff University Experiences of Children With Copy Number Variants (ECHO) Study; 16p11.2 European Consortium; Simons Variation in Individuals Project (VIP) Consortium. (2016). Defining the Effect of the 16p11.2 Duplication on Cognition, Behavior, and Medical Comorbidities. *JAMA Psychiatry.* Jan;73(1):20-30. (Consortium member collaborator.)
33. Nageshappa, S., Carromeu, C., Trujillo, C.A., Mesci, P., Espuny-Camacho, I., Pasciuto, E., Vanderhaeghen, P., Verfaillie, C.M., Raitano, S., Kumar, A., Carvalho, C.M., Bagni, C., Ramocki, M.B., Araujo, B.H., Torres, L.B., Lupski, J.R., Van Esch, H., Muotri, A.R. (2016). Altered neuronal network and rescue in a human MECP2 duplication model. *Mol Psychiatry.* Feb;21(2):178-88.
34. Bernier, R., Steinman, K.J., Reilly, B., Wallace, A.S., Sherr, E.H., Pojman, N., Mefford, H.C., Gerds, J., Earl, R., Hanson, E., Goin-Kochel, R.P., Berry, L., Kanne, S., Snyder, L.G., Spence, S., Ramocki, M.B., Evans, D.W., Spiro, J.E., Martin, C.L., Ledbetter, D.H., Chung, W.K. (2016). Clinical phenotype of the recurrent 1q21.1 copy-number variant. *Genet Med.* Apr;18(4):341-9.
35. Green, Snyder L., D'Angelo, D., Chen, Q., Bernier, R., Goin-Kochel, R.P., Wallace, A.S., Gerds, J., Kanne, S., Berry, L., Blaskey, L., Kuschner, E., Roberts, T., Sherr, E., Martin, C.L., Ledbetter, D.H., Spiro, J.E., Chung, W.K., Hanson, E.; Simons VIP consortium. (2016). Autism Spectrum Disorder, Developmental and Psychiatric Features in 16p11.2 Duplication. *J Autism Dev Disord.* Aug;46(8):2734-2748. (Consortium member collaborator.)
36. Steinman, K.J., Spence, S.J., Ramocki, M.B., Proud, M.B., Kessler, S.K., Marco, E.J., Green Snyder, L., D'Angelo, D., Chen, Q., Chung, W.K., Sherr, E.H.; Simons VIP Consortium. (2016). 16p11.2 deletion and duplication: Characterizing neurologic phenotypes in a large clinically ascertained cohort. *Am J Med Genet A.* Nov; 170(11):2943-2955.
37. Martin-Brevet, S., Rodríguez-Herreros, B, Nielsen, J.A., Moreau, C., Modenato, C., Maillard, A.M., Pain, A., Richetin, S., Jønch, A.E., Qureshi, A.Y., Zürcher, N.R., Conus, P.; 16p11.2 European Consortium; Simons Variation in Individuals Project (VIP) Consortium, Chung, W.K., Sherr, E.H., Spiro, J.E., Kherif, F., Beckmann, J.S., Hadjikhani, N., Reymond, A., Buckner, R.L., Draganski, B., Jacquemont, S. (2018). Quantifying the Effects of 16p11.2 Copy Number Variants

on Brain Structure: A Multisite Genetic-First Study. *Biol Psychiatry*. Aug 15;84(4):253-264. (Consortium member collaborator.)

38. Rodan, L.H. and Ramocki, M.B. (2020). Genetics and Metabolism in Pediatric Balance Disorders. *Manual of Pediatric Balance Disorders, Second Edition*. 185-202.
39. Modenato, C., Kumar, K., Moreau, C., Martin-Brevet, S., Huguet, G., Schramm, C., Jean-Louis, M., Martin, C.O., Younis, N., Tamer, P., Douard, E., Thébault-Dagher, F., Côté, V., Charlebois, A.R., Deguire, F., Maillard, A.M., Rodriguez-Herreros, B., Pain, A., Richetin, S.; 16p11.2 European Consortium; Simons Searchlight Consortium, Melie-Garcia, L., Kushan, L., Silva, A.I., van den Bree, M.B.M., Linden, D.E.J., Owen, M.J., Hall, J., Lippé, S., Chakravarty, M., Bzdok, D., Bearden, C.E., Draganski, B., Jacquemont, S. (2021). Effects of eight neuropsychiatric copy number variants on human brain structure. *Transl Psychiatry*. Jul 20;(1):399. (Consortium member collaborator.)
40. El Achkar, C.M., Rosen, A., Kessler, S.K., Steinman, K.J., Spence, S.J., Ramocki, M.B., Marco, E.J., Snyder, L.G., Spiro, J.E., Chung, W.K., Annapurna, P., Sherr, E.H. Clinical Characteristics of Seizures and Epilepsy in Individuals with Recurrent Deletions and Duplications in the 16p11.2 Region.

Personal

Dr. Melissa Ramocki returned home to Rhode Island and joined our practice in 2014. She and her husband live in Rhode Island with their three children and Boston terrier Fizban.