Stanford

maura ruzhnikov

- Clinical Assistant Professor, Neurology & Neurological Sciences
- Clinical Assistant Professor, Pediatrics - Medical Genetics

CLINICAL OFFICES

- **Child Neurology**

  730 Welch Rd

  2nd Fl

  Palo Alto, CA 94304

  Tel (650) 723-0993 **Fax** (650) 721-6350

Bio

BIO

Child neurologist and medical geneticist focusing on the diagnosis and management of rare neurologic disorders. Specific interests are in genetic epilepsy syndromes, childhood neurodegenerative and neurometabolic diseases and undiagnosed suspected genetic conditions.

CLINICAL FOCUS

- Neurology - Child Neurology
- Neurogenetics
- Medical Genetics

ACADEMIC APPOINTMENTS

- Clinical Assistant Professor, Neurology & Neurological Sciences
- Clinical Assistant Professor, Pediatrics - Medical Genetics
- Member, Maternal & Child Health Research Institute (MCHRI)

PROFESSIONAL EDUCATION

- Board Certification: Clinical Genetics, American Board of Medical Genetics and Genomics (2019)
- Board Certification, Clinical Genetics and Genomics , American Board of Genetics and Genomics (2019)
- Fellowship: Stanford University School of Medicine Registrar (2017) CA
- Board Certification: Neurology - Child Neurology, American Board of Psychiatry and Neurology (2016)
- Fellowship: University of California - San Francisco (2015) CA
- Residency: University of California - San Francisco (2012) CA
- Medical Education: New York University School of Medicine (2010) NY

LINKS

- Get a Second Opinion: https://stanfordhealthcare.org/second-opinion/overview.html

Research & Scholarship

CLINICAL TRIALS

- NGLY1 Deficiency: A Prospective Natural History Study, Not Recruiting

Publications

PUBLICATIONS

- **Metabolic Disorders Presenting with Seizures in the Neonatal Period.** *Seminars in neurology*

  Brimble, E., Ruzhnikov, M. R.

  2020

- **Expanding the Molecular and Clinical Phenotypes of FUT8-CDG.** *Journal of inherited metabolic disease*

  Ng, B. G., Dastsooz, H., Silawi, M., Habibzadeh, P., Jahan, S. B., Fard, M. A., Halliday, B. J., Raymond, K., Ruzhnikov, M. R., Tabatabaei, Z., Taghipour-Sheshdeh, A., Brimble, E., Robertson, et al

  2020

- **De novo EIF2AK1 and EIF2AK2 Variants Are Associated with Developmental Delay, Leukoencephalopathy, and Neurologic Decompensation.** *American journal of human genetics*

  Mao, D., Reuter, C. M., Ruzhnikov, M. R., Beck, A. E., Farrow, E. G., Emrick, L. T., Rosenfeld, J. A., Mackenzie, K. M., Robak, L., Wheeler, M. T., Burrage, L. C., Jain, M., Liu, et al

  2020

- **LOCALIZING NEUROLOGIC FEATURES AT PRESENTATION OF VLCAD DEFICIENCY**

  Leahy, P., Matalon, D., Ruzhnikov, M., Cowan, T., Enns, G.

  ACADEMIC PRESS INC ELSEVIER SCIENCE.2019: 282

- **Extracutaneous manifestations in phacomatosis cesioflammea and cesiomarmorata: Case series and literature review** *AMERICAN JOURNAL OF MEDICAL GENETICS PART A*

  Kumar, A., Zastrow, D. B., Kravets, E. J., Beleford, D., Ruzhnikov, M. Z., Grove, M. E., Dries, A. M., Kohler, J. N., Waggott, D. M., Yang, Y., Huang, Y., Mackenzie, K. M., Eng, et al

  2019; 179 (6): 966–77

- **Consensus Guidelines: MRI surveillance of Children with Presymptomatic Adrenoleukodystrophy**

  Turk, B., Mallack, E., Adang, L., Becker, C., Eichler, F., Van Haren, K., Hollandsworth, K., Kurtzberg, J., Kwon, J., Lund, T., Maegawa, G., Moser, A., Orchard, et al

  LIPPINCOTT WILLIAMS & WILKINS.2019

- **De Novo Missense Substitutions in the Gene Encoding CDK8, a Regulator of the Mediator Complex, Cause a Syndromic Developmental Disorder** *AMERICAN JOURNAL OF HUMAN GENETICS*

  Calpena, E., Hervieu, A., Kaserer, T., Swagemakers, S. A., Goos, J. C., Popoola, O., Ortiz-Ruiz, M., Barbaro-Dieber, T., Bownass, L., Brilstra, E. H., Brimble, E., Foulds, N., Grebe, et al

  2019; 104 (4): 709–20

- **Extracutaneous manifestations in phacomatosis cesioflammea and cesiomarmorata: Case series and literature review.** *American journal of medical genetics. Part A*

  Kumar, A., Zastrow, D. B., Kravets, E. J., Beleford, D., Ruzhnikov, M. R., Grove, M. E., Dries, A. M., Kohler, J. N., Waggott, D. M., Yang, Y., Huang, Y., Undiagnosed Diseases Network, Mackenzie, K. M., et al

  2019

- **De Novo Missense Substitutions in the Gene Encoding CDK8, a Regulator of the Mediator Complex, Cause a Syndromic Developmental Disorder.** *American journal of human genetics*

  Calpena, E., Hervieu, A., Kaserer, T., Swagemakers, S. M., Goos, J. A., Popoola, O., Ortiz-Ruiz, M. J., Barbaro-Dieber, T., Bownass, L., Brilstra, E. H., Brimble, E., Foulds, N., Grebe, et al

  2019

- **LOCALIZING NEUROLOGIC FEATURES AT PRESENTATION OF VLCAD DEFICIENCY**

  Leahy, P., Matalon, D., Ruzhnikov, M., Cowan, T., Enns, G.

  ACADEMIC PRESS INC ELSEVIER SCIENCE.2019: 311

- **De Novo Mutations Affecting the Catalytic C alpha Subunit of PP2A, PPP2CA, Cause Syndromic Intellectual Disability Resembling Other PP2A-Related Neurodevelopmental Disorders** *AMERICAN JOURNAL OF HUMAN GENETICS*
  Reynhout, S., Jansen, S., Haesen, D., van Belle, S., de Munnik, S. A., Bongers, E. F., Schieving, J. H., Marcelis, C., Amiel, J., Rio, M., Mclaughlin, H., Ladda, R., Sell, et al
  2019; 104 (1): 139–56

- **Perinatal distress in 1p36 deletion syndrome can mimic hypoxic ischemic encephalopathy.** *American journal of medical genetics. Part A*
  Carter, L. B., Battaglia, A., Cherry, A., Manning, M. A., Ruzhnikov, M. R., Bird, L. M., Dowsett, L., Graham, J. M., Alkuraya, F. S., Hashem, M., Dinulos, M. B., Vallee, S., Adam, et al
  2019

- **Infantile Spasms of Unknown Cause: Predictors of Outcome and Genotype-Phenotype Correlation** *PEDIATRIC NEUROLOGY*
  Yuskaitis, C. J., Ruzhnikov, M. Z., Howell, K. B., Allen, I., Kapur, K., Dlugos, D. J., Scheffer, I. E., Poduri, A., Sherr, E. H., EPGP Investigators
  2018; 87: 48–56

- **A case report of a suspected dual diagnosis: 22q11.2 deletion syndrome and X-linked chondrodysplasia punctata** *CLINICAL DYSMORPHOLOGY*
  Brimble, E., Pacione, M., Farrelly, E., Stevenson, D. A., Ruzhnikov, M. Z.
  2018; 27 (4): 151–53

- **Use of electronic medical record templates improves quality of care for patients with infantile spasms.** *Health information management : journal of the Health Information Management Association of Australia*
  Santoro, J. D., Sandoval, A., Ruzhnikov, M., Brimble, E., Chadwick, W., Wusthoff, C. J.
  2018: 1833358318794501

- **Infantile Spasms of Unknown Cause: Predictors of Outcome and Genotype-Phenotype Correlation.** *Pediatric neurology*
  Yuskaitis, C. J., Ruzhnikov, M. R., Howell, K. B., Allen, I. E., Kapur, K., Dlugos, D. J., Scheffer, I. E., Poduri, A., Sherr, E. H.
  2018

- **Clinical Transcriptome Sequencing Confirms Activation of a Cryptic Splice Site in Suspected <it><bold>SYNGAP1</it></bold>-Related Disorder** *MOLECULAR SYNDROMOLOGY*
  Brimble, E., Lee-Messer, C., Nagy, P. L., Propst, J., Ruzhnikov, M. Z.
  2018; 9 (6): 295–99

- **Variable clinical course of identical twin neonates with Alström syndrome presenting coincidentally with dilated cardiomyopathy.** *American journal of medical genetics. Part A*
  Hollander, S. A., Alsaleh, N., Ruzhnikov, M., Jensen, K., Rosenthal, D. N., Stevenson, D. A., Manning, M.
  2017; 173 (6): 1687-1689

- **DIAGNOSTIC OUTCOMES AND RELATIVE COST OF CLINICAL WHOLE EXOME SEQUENCING**
  Ruzhnikov, M. R., Alsadah, A., Mendelsohn, B., Alhariri, A., Cilio, M. R., Wu, Y., Marco, E. J., Hsiao, E., Sullivan, J., Shieh, J., Slavotinek, A., Sherr, E. H.
  LIPPINCOTT WILLIAMS & WILKINS.2016: 247

- **De novo mutations in KIF1A cause progressive encephalopathy and brain atrophy.** *Annals of clinical and translational neurology*
  Esmaeeli Nieh, S., Madou, M. R., Sirajuddin, M., Fregeau, B., McKnight, D., Lexa, K., Strober, J., Spaeth, C., Hallinan, B. E., Smaoui, N., Pappas, J. G., Burrow, T. A., McDonald, et al
  2015; 2 (6): 623–35

- **De novo mutations in epileptic encephalopathies.** *Nature*
  Allen, A. S., Berkovic, S. F., Cossette, P., Delanty, N., Dlugos, D., Eichler, E. E., Epstein, M. P., Glauser, T., Goldstein, D. B., Han, Y., Heinzen, E. L., Hitomi, Y., Howell, et al
  2013; 501 (7466): 217–21