Stanford

maura ruzhnikov

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.

ACADEMIC APPOINTMENTS

- Member, Maternal & Child Health Research Institute (MCHRI)

PROFESSIONAL EDUCATION

- Board Certification, Clinical Genetics and Genomics , American Board of Genetics and Genomics (2019)

LINKS

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

Research & Scholarship

CLINICAL TRIALS

- A Study to Evaluate the Safety and Efficacy of ION373 in Patients With Alexander Disease (AxD), Recruiting

Publications

PUBLICATIONS

- **SUCCESSFUL CARDIAC TRANSPLANTATION AND LONG-TERM FOLLOW-UP IN DNAJC19-ASSOCIATED DILATED CARDIOMYOPATHY WITH ATAXIA**
  Tahata, S., Tise, C., Floyd, B., Cusmano-Ozog, K., Ruzhnikov, M., Enns, G.
  ACADEMIC PRESS INC ELSEVIER SCIENCE.2022: 302

- **Accelerated identification of disease-causing variants with ultra-rapid nanopore genome sequencing.** *Nature biotechnology*
  Goenka, S. D., Gorzynski, J. E., Shafin, K., Fisk, D. G., Pesout, T., Jensen, T. D., Monlong, J., Chang, P. C., Baid, G., Bernstein, J. A., Christle, J. W., Dalton, K. P., Garalde, et al
  2022

- **Ultra-Rapid Nanopore Whole Genome Genetic Diagnosis of Dilated Cardiomyopathy in an Adolescent With Cardiogenic Shock.** *Circulation. Genomic and precision medicine*
  Gorzynski, J. E., Goenka, S. D., Shafin, K., Jensen, T. D., Fisk, D. G., Grove, M. E., Spiteri, E., Pesout, T., Monlong, J., Bernstein, J. A., Ceresnak, S., Chang, P., Christle, et al
  2022: CIRCGEN121003591

- **Ultrarapid Nanopore Genome Sequencing in a Critical Care Setting.** *The New England journal of medicine*

Gorzynski, J. E., Goenka, S. D., Shafin, K., Jensen, T. D., Fisk, D. G., Grove, M. E., Spiteri, E., Pesout, T., Monlong, J., Baid, G., Bernstein, J. A., Ceresnak, S., Chang, et al

2022

- **Biallelic CACNA2D1 loss-of-function variants cause early-onset developmental epileptic encephalopathy.** *Brain : a journal of neurology*

  Dahimene, S., von Elsner, L., Holling, T., Mattas, L. S., Pickard, J., Lessel, D., Pilch, K. S., Kadurin, I., Pratt, W. S., Zhulin, I. B., Dai, H., Hempel, M., Ruzhnikov, et al

  2022

- **Delineating the Epilepsy Phenotype of NGLY1 Deficiency.** *Journal of inherited metabolic disease*

  Levy, R. J., Frater, C. H., Gallentine, W. B., Phillips, J. M., Ruzhnikov, M. R.

  2022

- **UNCOMMON NEUROIMAGING FINDINGS IN INBORN ERRORS OF METABOLISM**

  Morales, J. A., Velez-Bartolomei, F. P., Ruzhnikov, M., Enns, G. M.

  BMJ PUBLISHING GROUP.2022: 194

- **Biochemical Studies in Fibroblasts to Interpret Variants of Unknown Significance in the ABCD1 Gene.** *Genes*

  van de Stadt, S. I., Mooyer, P. A., Dijkstra, I. M., Dekker, C. J., Vats, D., Vera, M., Ruzhnikov, M. R., van Haren, K., Tang, N., Koop, K., Willemsen, M. A., Hui, J., Vaz, et al

  1800; 12 (12)

- **Variable clinical severity in TANGO2 deficiency: Case series and literature review.** *American journal of medical genetics. Part A*

  Schymick, J., Leahy, P., Cowan, T., Ruzhnikov, M. R., Gates, R., Fernandez, L., Pramanik, G., Undiagnosed Diseases Network, Yarlagadda, V., Wheeler, M., Bernstein, J. A., Enns, G. M., Lee, C.

  2021

- **Corrigendum to eP296-The yield of thorough record review in the Undiagnosed Diseases Network, Volume 132, Supplement 1, April 2021, Page S187, https://doi.org/10.1016/S1096-7192(21)00378-4.** *Molecular genetics and metabolism*

  Findley, L., Mulvihill, J. J., Bentley, A., Bernstein, J. A., Bican, A., Botto, L., Briere, L., Butte, M. J., Cope, H., Fogel, B. L., Hom, J., Kravets, E., Mak, et al

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- **A Prospective Study of Epilepsy in NGLY1 Deficiency**

  Levy, R., Frater, C., Gallentine, W., Ruzhnikov, M.

  WILEY.2021: S67-S68

- **Genetic counselor roles in the undiagnosed diseases network research study: Clinical care, collaboration, and curation.** *Journal of genetic counseling*

  Kohler, J. N., Kelley, E. G., Boyd, B. M., Sillari, C. H., Marwaha, S., Undiagnosed Diseases Network, Wheeler, M. T., Acosta, M. T., Adam, M., Adams, D. R., Agrawal, P. B., Alejandro, M. E., Alvey, J., et al

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- **Delineating the molecular and phenotypic spectrum of the SETD1B-related syndrome.** *Genetics in medicine : official journal of the American College of Medical Genetics*

  Weerts, M. J., Lanko, K., Guzman-Vega, F. J., Jackson, A., Ramakrishnan, R., Cardona-Londono, K. J., Pena-Guerra, K. A., van Bever, Y., van Paassen, B. W., Kievit, A., van Slegtenhorst, M., Allen, N. M., Kehoe, et al

  2021

- **A novel likely pathogenic heterozygous HECW2 missense variant in a family with variable expressivity of neurodevelopmental delay, hypotonia, and epileptiform EEG patterns** *AMERICAN JOURNAL OF MEDICAL GENETICS PART A*

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  2021

- **Delineating the genotypic and phenotypic spectrum of HECW2-related neurodevelopmental disorders.** *Journal of medical genetics*

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- **Variants in PRKAR1B cause a neurodevelopmental disorder with autism spectrum disorder, apraxia, and insensitivity to pain** *GENETICS IN MEDICINE*

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  ACADEMIC PRESS INC ELSEVIER SCIENCE.2021: S187

- **Early Signs and Symptoms of Leukodystrophies: A Case-Based Guide.** *Pediatrics in review*
  Ruzhnikov, M. R., Brimble, E., Hickey, R. E., Leukodystrophy Care Network
  2021; 42 (3): 133–46

- **Commonalities across computational workflows for uncovering explanatory variants in undiagnosed cases.** *Genetics in medicine : official journal of the American College of Medical Genetics*
  Kobren, S. N., Baldridge, D., Velinder, M., Krier, J. B., LeBlanc, K., Esteves, C., Pusey, B. N., Zuchner, S., Blue, E., Lee, H., Huang, A., Bastarache, L., Bican, et al
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- **Aicardi-Goutières syndrome may present with positive newborn screen for X-linked adrenoleukodystrophy.** *American journal of medical genetics. Part A*
  Tise, C. G., Morales, J. A., Lee, A. S., Velez-Bartolomei, F. n., Floyd, B. J., Levy, R. J., Cusmano-Ozog, K. P., Feigenbaum, A. S., Ruzhnikov, M. R., Lee, C. U., Enns, G. M.
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- **PROSPECTIVE STUDY OF EPILEPSY IN NGLY1 DEFICIENCY**
  Levy, R. J., Frater, C. H., Galentine, W. B., Ruzhnikov, M. R.
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- **Use of electronic medical record templates improves quality of care for patients with infantile spasms** *HEALTH INFORMATION MANAGEMENT JOURNAL*
  Santoro, J. D., Sandoval Karamian, A. G., Ruzhnikov, M., Brimble, E., Chadwick, W., Wusthoff, C. J.
  2021; 50 (1-2): 47–54

- **Identification of protein quality control regulators using a Drosophila model of TPI deficiency.** *Neurobiology of disease*
  Hrizo, S. L., Eicher, S. L., Myers, T. D., McGrath, I. n., Wodrich, A. P., Venkatesh, H. n., Manjooran, D. n., Swoger, S. n., Gagnon, K. n., Bruskin, M. n., Lebedev, M. V., Zheng, S. n., Vitantonio, et al
  2021: 105299

- **Metabolic Disorders Presenting with Seizures in the Neonatal Period.** *Seminars in neurology*
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- **Expanding the Molecular and Clinical Phenotypes of FUT8-CDG.** *Journal of inherited metabolic disease*
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  Mao, D. n., Reuter, C. M., Ruzhnikov, M. R., Beck, A. E., Farrow, E. G., Emrick, L. T., Rosenfeld, J. A., Mackenzie, K. M., Robak, L. n., Wheeler, M. T., Burrage, L. C., Jain, M. n., Liu, et al
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  BMJ PUBLISHING GROUP.2020: A116

- **MONOZYGOTIC TWINS DISCORDANT FOR CRANIAL DYSINNERVATION DISORDER: EVIDENCE OF VASCULAR DISRUPTION**
  Tise, C. G., DeFilippo, C., Ruzhnikov, M. R., Stevenson, D.
  BMJ PUBLISHING GROUP.2020: A147

- **Clinical sites of the Undiagnosed Diseases Network: unique contributions to genomic medicine and science.** *Genetics in medicine : official journal of the American College of Medical Genetics*
  Schoch, K. n., Esteves, C. n., Bican, A. n., Spillmann, R. n., Cope, H. n., McConkie-Rosell, A. n., Walley, N. n., Fernandez, L. n., Kohler, J. N., Bonner, D. n., Reuter, C. n., Stong, N. n., Mulvihill, et al
  2020

- **MRI Surveillance of Boys with X-linked Adrenoleukodystrophy Identified by Newborn Screening: Meta-analysis and Consensus Guidelines.** *Journal of inherited metabolic disease*

  Mallack, E. J., Turk, B. R., Yan, H. n., Price, C. n., Mlis, M. D., Moser, A. B., Becker, C. n., Hollandsworth, K. n., Adang, L. n., Vanderver, A. n., Van Haren, K. n., Ruzhnikov, M. n., Kurtzberg, et al

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- **De Novo EIF2AK1 and EIF2AK2 Variants are Associated with Developmental Delay, Movement Disorders, Cerebellar Ataxia, Leukoencephalopathy, and Neurologic Decompensation**

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- **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

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- **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

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  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

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- **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*

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

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

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- **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

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- **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

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- **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*
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- **Infantile Spasms of Unknown Cause: Predictors of Outcome and Genotype-Phenotype Correlation.** *Pediatric neurology*
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- **Clinical Transcriptome Sequencing Confirms Activation of a Cryptic Splice Site in Suspected <it><bold>SYNGAP1</it></bold>-Related Disorder** *MOLECULAR SYNDROMOLOGY*
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  Hollander, S. A., Alsaleh, N., Ruzhnikov, M., Jensen, K., Rosenthal, D. N., Stevenson, D. A., Manning, M.
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