Stanford



Anne (Annie) Devereux Niehaus

Bio

ACADEMIC APPOINTMENTS

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

ADMINISTRATIVE APPOINTMENTS

- Associate Program Director, Pediatrics-Medical Genetics and Genomics Residency Program, Stanford, (2023- present)

BOARDS, ADVISORY COMMITTEES, PROFESSIONAL ORGANIZATIONS

- Fellow, American College of Medical Genetics and Genomics (ACMG) (2017 - present)

PROFESSIONAL EDUCATION

- BA, Pomona College , Public Policy Analysis - Chemistry (2014)

LINKS

- Linkedin: www.linkedin.com/in/anne-annie-niehaus-medicalgenetics

Teaching

GRADUATE AND FELLOWSHIP PROGRAM AFFILIATIONS

- Medical Genetics (Fellowship Program)

Publications

PUBLICATIONS

- **De Novo Variants Associated With Autosomal Recessive Conditions: Case Series and Implications for Genetic Testing and Counseling.** *American journal of medical genetics. Part A*

  Niehaus, A. D., Bonner, D. E., Carter, J., Avello, K., Jacob, N., Neu, M. B., Mendez, R., Qiao, W., Scott, S. A., Levy, R. J., Mattas, L., Schymick, J., Van Andel, et al

  2026

- **Medical Genetics and Genomics Residency Programs: Trends in Applications, Match Rates, and Matriculation from 2015 to 2024.** *Genetics in medicine : official journal of the American College of Medical Genetics*

  Niehaus, A. D., Stevenson, D. A., Blitzer, M. G.

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- **Langerhans Cell Histiocytosis in Cardiofaciocutaneous Syndrome.** *American journal of medical genetics. Part A*

  Keehan, L., Sleightholm, R., Marks, L. J., Stevenson, D. A., Niehaus, A. D.

2025: e64294

- **Characterizing and Evaluating the Structures of Combined Pediatrics and Medical Genetics and Genomics Residency Programs.** *American journal of medical genetics. Part A*
  Niehaus, A. D., Stevenson, D. A.
  2024: e63916

- **Mitochondrial HMG-CoA Synthase Deficiency: A Cyclic Vomiting Mimic Without Reliable Biochemical Markers.** *Journal of investigative medicine high impact case reports*
  Niehaus, A. D., Cooper, H., Lee, C. U.
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- **Newborn Screening for X-Linked Adrenoleukodystrophy (X-ALD): Biochemical, Molecular, and Clinical Characteristics of Other Genetic Conditions.** *Genes*
  Mares Beltran, C. F., Tise, C. G., Barrick, R., Niehaus, A. D., Sponberg, R., Chang, R., Enns, G. M., Abdenur, J. E.
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- **Clinicopathologic Features of IDEDNIK (MEDNIK) Syndrome in a Term Infant: Histopathologic Features of the Gastrointestinal Tract and Report of a Novel AP1S1 Variant.** *Pediatric and developmental pathology : the official journal of the Society for Pediatric Pathology and the Paediatric Pathology Society*
  Lu, J. G., Namjoshi, S. S., Niehaus, A. D., Tahata, S., Lee, C. U., Wang, L., McDonnell, E., Seely, M., Martin, M. G., Hazard, F. K.
  2023: 10935266231177402

- **A homozygous Gly470Ala variant in PEX6 causes severe Zellweger spectrum disorder.** *American journal of medical genetics. Part A*
  Galarreta, C. I., Wong, K., Carmichael, J., Woods, J., Tise, C. G., Niehaus, A. D., Schildt, A. J., Verscaj, C. P., Cusmano-Ozog, K. P.
  2023

- **Neonatal lupus is a novel cause of positive newborn screening for X-linked adrenoleukodystrophy.** *American journal of medical genetics. Part A*
  Niehaus, A. D., Mendelsohn, B. A., Zimmerman, B., Lee, C. U., Manning, M. A., Cusmano-Ozog, K. P., Tise, C. G.
  2023

- **BIOCHEMICAL, MOLECULAR, AND CLINICAL CHARACTERISTICS OF PEROXISOMAL DISORDERS DETECTED BY CALIFORNIA NEWBORN SCREENING (NBS) PROGRAM**
  Beltran, C., Abdenur, J., Chang, R., Barrick, R., Spongberg, R., Tise, C. G., Niehaus, A. D., Enns, G.
  ACADEMIC PRESS INC ELSEVIER SCIENCE.2023: 72

- **Phenotypic variability in RERE-related disorders and the first report of an inherited variant.** *American journal of medical genetics. Part A*
  Niehaus, A. D., Kim, J., Manning, M. A.
  2022

- **A survey of program directors for combined pediatrics and medical genetics and genomics residency programs: Perspectives when evaluating applicants.** *American journal of medical genetics. Part A*
  Niehaus, A. D., Rassbach, C. E., Stevenson, D. A.
  2022

- **VARIABLE EXPRESSIVITY IN NTRK1-CONGENITAL INSENSITIVITY TO PAIN WITH ANHIDROSIS (CIPA)**
  Niehaus, A. D., Tise, C. G., Manning, M., Stevenson, D.
  BMJ PUBLISHING GROUP.2022: 192-193

- **A survey assessing adoption of the ACMG-AMP guidelines for interpreting sequence variants and identification of areas for continued improvement** *GENETICS IN MEDICINE*
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- **Development of Clinical Domain Working Groups for the Clinical Genome Resource (ClinGen): lessons learned and plans for the future** *GENETICS IN MEDICINE*
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- **Development of a consent resource for genomic data sharing in the clinical setting** *GENETICS IN MEDICINE*
  Riggs, E., Azzariti, D. R., Niehaus, A., Goehringer, S. R., Ramos, E. M., Rodriguez, L., Knoppers, B., Rehm, H. L., Martin, C., Clinical Genome Resource Educ Work
  2019; 21 (1): 81-88

- **Points to consider for sharing variant-level information from clinical genetic testing with ClinVar** *COLD SPRING HARBOR MOLECULAR CASE STUDIES*
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  2018; 4 (1)

- **A standardized, evidence-based protocol to assess clinical actionability of genetic disorders associated with genomic variation** *GENETICS IN MEDICINE*
  Hunter, J., Irving, S. A., Biesecker, L. G., Buchanan, A., Jensen, B., Lee, K., Martin, C., Milko, L., Muessig, K., Niehaus, A. D., O'Daniel, J., Piper, M. A., Ramos, et al
  2016; 18 (12): 1258-1268

- **Using ClinVar as a Resource to Support Variant Interpretation.** *Current protocols in human genetics*
  Harrison, S. M., Riggs, E. R., Maglott, D. R., Lee, J. M., Azzariti, D. R., Niehaus, A., Ramos, E. M., Martin, C. L., Landrum, M. J., Rehm, H. L.
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- **Providing Access to Genomic Variant Knowledge in a Healthcare Setting: A Vision for the ClinGen Electronic Health Records Workgroup.** *Clinical pharmacology and therapeutics*
  Overby, C. L., Heale, B. n., Aronson, S. n., Cherry, J. M., Dwight, S. n., Milosavljevic, A. n., Nelson, T. n., Niehaus, A. n., Weaver, M. A., Ramos, E. M., Williams, M. S.
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- **After Myriad: Genetic Testing in the Wake of Recent Supreme Court Decisions about Gene Patents.** *Current genetic medicine reports*
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