



Anne (Annie) Devereux Niehaus

- Affiliate, Dean's Office Operations - Dean Other
- Resident in Graduate Medical Education

Bio

BIO

Medical Genetics and Genomics resident physician

BOARDS, ADVISORY COMMITTEES, PROFESSIONAL ORGANIZATIONS

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

INTERNET LINKS

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

Publications

PUBLICATIONS

- **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.
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Niehaus, A. D., Mendelsohn, B. A., Zimmerman, B., Lee, C. U., Manning, M. A., Cusmano-Ozog, K. P., Tise, C. G.
2023
- **Phenotypic variability in RERE-related disorders and the first report of an inherited variant.** *American journal of medical genetics. Part A*
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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
- **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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- **Points to consider for sharing variant-level information from clinical genetic testing with ClinVar** *COLD SPRING HARBOR MOLECULAR CASE STUDIES*
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- **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*
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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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