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# Rare Diseases Project Group

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National Center for Advancing Translational Sciences

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# Working Session Agenda

- Rare Disease Day at NIH
- Workshop Development
  - Review Resources/Databases
- Asks of ISCC-PEG Members

# Rare Disease Day at NIH

- March 1 from 10:30am-5:30pm
- Sponsored by NCATS and the NIH Clinical Center
- ISCC-PEG Presentation
  - Title: Partnership for Educating Healthcare Providers on Rare Diseases
- Virtual exhibit staffed by ISCC-PEG members



# Workshop Development

- Accepted for 2021 Pediatric Academic Societies virtual meeting
- Title: “Training Next Generation Pediatricians in Genomics: A Case-Study Approach”
- Interactive team-based learning (TBL) model
- Objectives:
  - Assess which patients are appropriate candidates for genetic evaluation and testing and when to collaborate with genetic specialists.
  - Describe the process for selecting the appropriate genetic test for a patient.
  - Identify ethical and legal considerations around informed consent and key aspects of pre/post test counseling.
  - Describe key aspects of genetic test result interpretation.
  - Develop skills in using online resources to assist in the care of your patients with genetic disorders.

# Workshop Exercises

1. Test Selection
2. Diagnostic Result
3. Incidental Finding
4. Explaining Results to the Patient



# Resources/Databases



- Genetic Testing Registry
- ClinGen
- OMIM
- ClinVar
- GeneReviews
- MedlinePlus Genetics
- PharmGKB
- ClinicalTrials.gov
- GARD

# Asks of ISCC-PEG Membership



- Register for Rare Disease Day at NIH:  
<https://ncats.nih.gov/news/events/rdd>