Rare Diseases Project Group

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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