

## Group #4

- Diagnostic Testing
  - Need to know how many genetic/rare diseases? How many need clinical genetic diagnostic testing?
    - What are the available resources to answer that question?
      - Orphanet
      - GeneTests
      - NIH rare disease terminology
      - EuroGentest
      - EDDNAL
- ACTION ITEMS
  - Conference call with interested groups (Giovanna to organize)
  - Clinical utility definition, operational definition (Giovanna, Bonnie Pagon, Manuel Posada)

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- CETT (as a model)
  - Determine research labs with potential clinical tests.
    - Explore with EuroGentest the need for CETT-like model and /or identification of common goals
    - Exploration with non-US/European countries
- ACTION ITEMS:
  - Develop a plan and arrange a conference call (S. Hogarth, D. Taruscio, A. Faucett, L.Kalman)

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- Nomenclature

- How to promote standard nomenclature according to HGVS.

- ACTION ITEMS:

- Work with WHO, ICORD, OECD, professional societies, major journal editors, disease-specific groups (S. Amye, B. Pagon, M. Posada)