

Moderated Discussions

December 1-2, 2011

Perspectives on Existing genetic Variation Resources

- What works? What are the gaps and challenges with existing databases?
- What else is needed to identify and label clinically actionable variants?

Current Approaches for Identifying Genetic Variants for Clinical Use

- What can we learn from these existing approaches? Are there common aspects across them?
- What key information is needed to classify variants and make decisions about implementation?

Developing Consensus for Binning Variants for Clinical Use

- What evidence is needed to classify variants into specific categories?
- How do we capture patient preferences in this process?
- How do we enhance the process for coming to consensus?
- What framework is most appropriate for our purpose?
- What groups will assess the evidence and make recommendations? How do we capture professional society guidelines in this process?

Creating a Translational Loop for Genomic Medicine

- How should actionable variants and suggested actions be disseminated to clinicians?
- What data fields and formats are needed for integration into CDS tools and EHRs?
- What barriers exist and how can they be addressed?
- How do we get information from clinical trials back to the researcher?

Translating Actionable Variants into Evidence-Based Practice

- What data fields and formats are needed for measuring outcomes and using findings to enhance resources?
- Variant interpretation will continue to evolve as new data become available; how best can this evolution be represented in databases and tools?
- What are the barriers to implementation and how can they be addressed?
- What is the difference between having genomic information in hand and having to acquire it?
- What methods are most useful for moving information into practice and what role can resources of this type play?