Evidence Generation

Outcomes dependent on phenotyping
- Need to develop standards for phenotyping and tools to aid in collection
  -ICHPT*
  -GA4GH
  -ClinGen
  -EHR Evolution
  -Others...

Outcomes dependent on test content and interpretation
- Need method standards
  -ACMG, CAP, CDC, NIST
  -CMGS, EBMG, EuroGenTest
- Need guidelines for the evidence-based assessment of genes and variants
  -ACMG
  -ClinGen
  -Others..
- Need experts to evaluate genes and variants for medical use
  -ClinGen

Health Improvements +/- Economic Savings
- Randomized Control Trials
- Comparative Effectiveness Research
- Sound Arguments
- Rarity of phenotypes and variants may require aggregation of studies by gene, disease, category of disease
- Need EHR tools to collect outcomes

*International Consortium for Human Phenotype Terminologies (HPO, PhenoDB, Orphanet, Elements of Morphology, SNOMed-CT)
Evidence Generation

State the priorities for this area that will enable genomic medicine implementation

1. Catalog evidence generating projects – IGNITE?
   - Availability of data and specimens

2. Standards for a test (method, content, interpretation, risk prediction)

3. Definition of evidence (gene, variant, test, treatment)
   - Standards should not be higher than other areas of medicine

4. Encourage adoption of genomic medicine applications with existing evidence
   - Intersociety coordinating committee – competencies in residency training
   - Making a case for economic benefit
   - Engage physicians in identifying areas of genomic implementation
   - Society practice guidelines

State the opportunities for action as individual nations or as a collective group

1. Identify countries/systems willing to enable access to patient data
   - Sharing evidence generated in those systems

2. Need systems to capture evidence - Facilitate a federated network and standardized APIs to share data – GA4GH

What are the next steps toward a plan to facilitate implementation in this area

1. Discuss areas of overlap with activities of other organizations