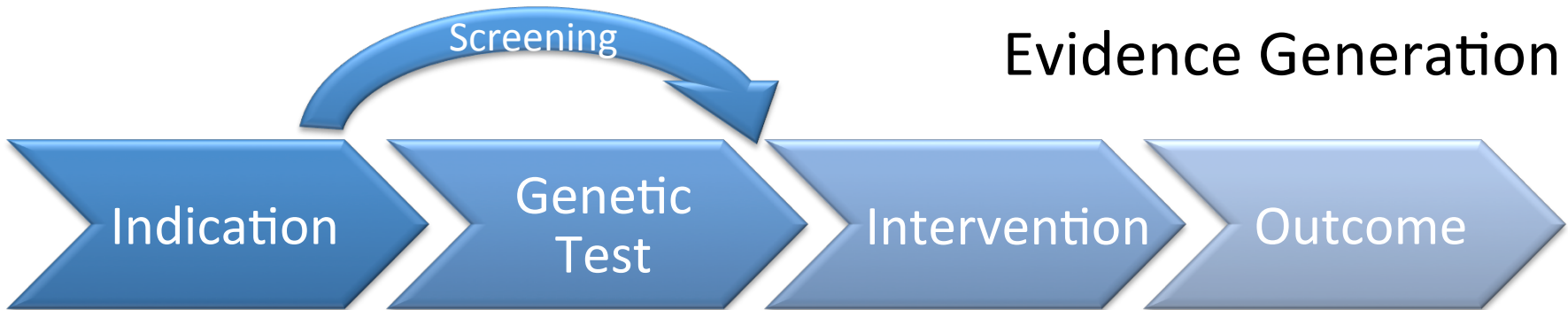


# Evidence Generation



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