Over-arching Goal

- To provide expert guidance to NIH and the scientific community on the utility of sequencing large sample collections to improve the understanding and treatment of complex disease.
- Not to define the role of rare variants in the missing heritability, or to identify which cohorts to sequence.
- Need to be inclusive of population, patient and socio-demographic groups.

Two Key Scientific Objectives

- Identify key biomedical questions that can be addressed by sequencing large well-designed samples of deeply phenotyped individuals.

- Defining criteria for selecting samples to answer those questions.

Issues to Keep in Mind

- Strengths and weaknesses of prospective cohort or retrospective case-control designs, family or extremes designs
- General considerations about power and sample size
- Costs and benefits, including analytic approaches, of whole genome vs whole exome sequencing
- Consideration of different –omic data types (expression, proteomics, etc.)