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.)