

Criteria for Supporting New Research Opportunities in Population Genomics, Particularly at
Other NIH Institutes and Centers

- I. Meets one or more scientific goals of NHGRI population genomics program
 - A. Establishes resources for genotype-phenotype research, such as large case-control GWA collections in GAIN or GEI
 - B. Improves analytic strategies or supports novel analyses for genotype-phenotype research, such as GEI sequence data analysis RFA, replication working group
 - C. Builds successful collaborations across multiple disease domains and disciplines, such as genotyping of NHANES subjects, multi-IC symposia
 - D. Conducts novel studies to identify genetic factors related to health and disease, such as pharmacogenomic-based treatment (coumadin trial), molecular classification of disease (rheumatologic diseases, response to inflammation or injury), or variants predicting good health and function
 - E. Enhances research training and involvement of epidemiologists in genetics and geneticists in epidemiology, such as workshop on analysis and interpretation of GWA data for epidemiologists or basic principles of case-control studies for geneticists
- II. Meets one or more scientific goals of NHGRI as a whole*
 - A. Develops genomic resources and technology, such as databases for GWA studies (dbGaP) and procedures for accessing them (GWAS Data Repository Implementation Group)
 - B. Investigates genome on broad scale rather than one or a few candidate genes, such as GWA genotyping or characterization of major histocompatibility complex in well-phenotyped individuals
 - C. Expands research portfolio to approach more closely direct applications for improving health and preventing disease, such as assessing population risk of diabetes in individuals carrying TCF7L2 variants
- III. Catalyzes or facilitates genomic research
 - A. Sets paradigm for class of studies, such as
 - 1. Risk/benefit comparison of genotypic vs. phenotypic screening for hemochromatosis (HEIRS)

2. Impact of pharmacogenomic-based dose adjustments in warfarin therapy on frequency of adverse effects of treatment and compliance with therapy (NHLBI coumadin trial)
 3. Impact of pharmacogenomic-based screening for abacavir hypersensitivity (genotyping HLA-B*5701 retrospectively in persons receiving abacavir and correlating with recorded side effects; or prospectively for use in treatment selection to assess impact on frequency of hypersensitivity and compliance with treatment)
- B. Provides resource not available or supportable by single or few ICs, such as database for genome-wide association studies of multiple diseases and traits (dbGaP, “orphan databases” and Affy or Illumina controls database)
 - C. Develops research tools or resources that would not have been developed in the same fashion without NHGRI’s involvement (coumadin trial would not have examined treatment response discordant with genotype-predicted response; NHANES would/will not have made data widely available)

IV. Meets key policy or administrative goals

- A. Is cost-effective and feasible with available resources, such as programs that leverage other ICs’ existing investments in population-based studies, include co-funding for proposed population genomic effort, and/or permit rapid supplementation of existing awards
- B. Addresses priorities in minority health and health disparities, such as population studies that include or even focus on U.S. minorities or their populations of origin, or on U.S. groups with low levels of access to health care

* As defined in NHGRI Mission Statement (at <http://www.genome.gov/10001022>)

“...Now, the NHGRI’s mission has expanded to encompass a broad range of studies aimed at understanding the structure and function of the human genome and its role in health and disease. To that end NHGRI supports the development of resources and technology that will accelerate genome research and its application to human health. A critical part of the NHGRI mission continues to be the study of the ethical, legal and social implications (ELSI) of genome research. NHGRI also supports the training of investigators and the dissemination of genome information to the public and to health professionals.”