Recommendations Currently in Early Stages of Implementation

- Identify a few key phenotypes and exposures for standardization; coordinate and find best partners among ICs
- Develop analytic methods to address combinatorial effect of multiple small gene effects (GxG interaction)
- Incorporate new technologies into GWA and population (?family) studies such as structural variation; others when sufficiently mature for large-scale application
- Conduct GWA studies in minority populations, including not only those defined by race/ethnicity
- Continue current roles with dbGaP: orphan datasets, policy development esp for future data types

Recommendations Currently in Early Stages of Implementation (2)

- Methodologic research such as allele calling, population structure, imputation and when NCBI should do this as automated service
- Develop tools for looking at traits across studies
- Include more diverse populations, particularly more cosmopolitan (diverse and admixed) populations (such as NYC or LA)
- Examine ways of using AIMs and similar markers in describing populations in combination with current social lexicon
- Expand training efforts through traditional NIH training mechanisms and institutional mechanisms

Recommendations Currently in Early Stages of Implementation (3)

- Improve efforts to educate science writers and journalists
- Encourage participation of minority investigators and institutions in population genomics research

Recommendations Appropriate for Leadership by Other ICs or NIH-Wide

- Standardize phenotype definitions (trans-NIH), promote use as clinical standards, and emphasize importance of retaining primary data
- Ensure clinical trials are adequately used for genomic research– DNA collection as basic step
- Mine existing sample banks of multiple specimen types from ongoing epidemiologic, clinical studies
- Facilitate structured review of clinical significance of variant alleles: partner with disease-specific ICs
- Design intervention studies to target those at genetic risk whose environment can be modified
- Integrate statistical methodologic research projects as part of center grants– make part of RFA

Recommendations Appropriate for Leadership by Other ICs or NIH-Wide (2)

- Need to sample geographic areas of origin better given disparities in US-derived populations
- Need GWA studies of drug response and environmental exposures
- Study female advantage in survival
- Study effects of age rather than adjusting it away
- Examine "antagonistic pleotropy"—reproductive fitness may have unexpected effects later in life
- Consider consensus conference on current use of genetic markers in assessment of risk of disease

Recommendations Appropriate for Leadership by Other ICs or NIH-Wide (3)

- Survey public knowledge of genetics periodically in national surveys; identify appropriate, sustainable interventions to address knowledge gaps
- Identify compelling cases nearly ready for clinical application where new discoveries could be tested in research environment

Recommendations for NHGRI/OPG

- Improve connections with CTSAs and promote genomic infrastructure, but recognize need for NHGRI translational efforts
- Revisit implementation recommendations for possible future large cohort study
- Establish cell repositories and derived datasets for gene function in tissue samples (not just LCL)– SNP genotyping, expression, splicing, ?proteomics, ?epigenetics
- Enhance community interactions around issues of return of results and how data will be used

Recommendations for NHGRI/OPG (2)

- Develop and disseminate consensus guidelines for collection of samples for next generation of studies
- Beyond SNPs and CNVs: Plan for analysis of full/extended sequence data
- Support methodologic research to evaluate different sources of controls for GWA
- Conduct Users' Workshops for dbGaP and related resources (access and use)
- Explore genetics of good health, including modifiers of Mendelian disorders (GxG)

Recommendations for NHGRI/OPG (3)

- Have "user's" workshop on interpretation of race/ethnic categories in genomic research
- Identify and address gaps in knowledge base for current direct to consumer genetic testing
- Examine ways to train Clinical Pharmacists and other health professionals in genetics to serve as test translators
- Need clear way to identify "what's ready" for clinical application and deal with uncertainty in interim
- Enhance understanding of preventive medicine among genomics researchers