Principles and procedures for phenotype working-group consortium

Cohorts for Heart and Aging Research in Genomic Epidemiology (CHARGE) Consortium
(Formally approved by all participating cohort studies; Feb 1, 2008)

Participating Cohorts and contacts:

Age, Gene/Environment, Susceptibility--Reykjavik (AGES)
Contact: Tamara Harris (harrist@gw.nia.nih.gov)
Vilmundur Gudnason (v.gudnason@hjarta.is)

Atherosclerosis Risk in Communities Study (ARIC)
Contact: Eric Boerwinkle (eric.boerwinkle@uth.tmc.edu)

Cardiovascular Health Study (CHS)
Contact: Bruce M. Psaty (psaty@u.washington.edu)

Framingham Heart Study (FHS)
Contact: Christopher O'Donnell (codonnell@nih.gov)

Rotterdam Study
Contact: Bert Hofman (a.hofman@erasmusmc.nl)
Andre Uitterlinden (a.g.uitterlinden@erasmusmc.nl)

1. Purpose: collaboration to improve reliability and validity of genome-wide association findings.

2. Origin: In some instances, the Research Collaboration Committee of the consortium of population-based cohort studies may have identified a coordinator to initiate conference calls among working-group (WG) members. Available resources likely include an Analysis Committee and perhaps others as well. Responsibilities of the RCC are described in the document “Principles and procedures for consortium of population-based studies.”

3. Working Group Organization and Responsibilities: WG Coordinator convenes the WG.

   Each Working Group Steering Committee (WGSC) consists of cohorts participating in the consortium of population-based cohorts (one or two members per study). It is the responsibility of the WGSC to initiate or prepare proposals and submit them to all WG members for discussion and revision.

   Each WGSC selects a Coordinator, who may or may not be the WG Coordinator originally identified by the RCC, to run meetings and calls for practical management.

   Each WG may add non-member studies, such as case-control studies or other consortia to the WG.

   Whether other studies may join the WG will be decided by the WGSC based on consensus, and will depend on the availability of GWA data, size of the study, quality of the phenotype data and timing.

   The WGSC for each phenotype may decide whether and when a new study may become a member of the WGSC.

   The WGSC decides on a date for data freeze after which studies can no longer contribute their data to the active wave of papers.

   WG aims for transparency, timely communication, effective coordination, and a sharing of responsibilities, challenges, and opportunities among study members. WG encourages collaboration among members

   WG needs to be flexible to accommodate pre-existing agreements about collaboration and pre-existing WGs.

   WG members may opt out of a WG, but this decision to opt out of a working group must take place before WGA genotype-phenotype data are shared.

   Collaborating groups with confirmation samples for new genotyping are eligible to become members of the WG.

   WG members agree not to share whole-genome genotype-phenotype findings from other studies in the WG with any outside groups without permission of the WG members who generated the data.

   If WG members have access to other unshared data, the results that emerge from a combined analysis of shared and unshared data will be treated as if they arose from the WG shared data.
WG members may agree to work with other studies and serve as a replication study for top hits for other GWA studies or candidate gene studies. This plan should be disclosed to other WG members and should be considered acceptable as long as the number of replicated SNPs is not so large that much of the data of the participating study is made public outside of the consortium. If studies wish to share their data in other ways, these plans too should be disclosed to other WG members.

WG may encourage joint ancillary-study grant applications to accomplish new scientific aims.

WG agrees to seek opportunities to promote careers of junior investigators and fellows.

4. Working Groups Efforts: Primary work takes place in WG, often with the assistance of the Analysis Committee.

WG shares among members detailed information about phenotypes, sample size, genotyping platforms, data cleaning methods, analysis methods, imputation methods, and high priority sub-phenotypes within its WG domain.

WG standardizes phenotypes across studies, evaluate results, write papers, decide on publications, on authorship, and on the need for additional follow-up studies in confirmation samples.

**Data sharing:** results for all successfully genotyped markers in the primary GWA scan; these shared data may not be used for other purposes than the WG aims without permission from other cohort members.

**Data to be shared:** rs#, allele designation, genotype counts, model type (additive), estimated effect sizes, 95% CIs, p-values for association tests, data summaries, quality control information (duplicates, HW); plus the same sort of data for imputed SNPs to facilitate sharing across platforms.

5. Publication plans: Initial publication plan will be developed at the time of WG formation.

Plans may be modified by WG agreement based on the shape of the results.

For parallel publications, one month is a tolerable wait time; three months may not be tolerable.

WG needs to balance rapid publication, strength of findings from replication, and equal partnership.

WG members assure that parent-study disclaimers, reviews, and approvals take place as required. These approvals should occur within one month of the completion of the final draft.

Abstracts should follow the same rules as manuscripts for publications. Authorship for abstracts may be less due to space constraints, but the name of the consortium must be included.

6. Authorship: The goal is fair scientific representation from study members participating in the WG.

Multiple first and multiple senior authors may be designated as such.

First and senior authors, including the overall first author and the overall last author, should generally come from different cohorts.

Long lists of authors are permitted if all authors meet standards for authorship (such as those required by major journals, for instance the criteria used by JAMA). Contributing scientists from outside the working group may be coauthors.

Authorship position in first papers will be determined in advance by a group consensus by the researchers actively participating in the proposed pooled study. Some of the factors that may be considered include effort in WG, contributions, by size of the GWA, phenotype data quality, and the contribution of the cohort’s data to the results of the pooled study. In later papers, successful efforts to obtain additional funding for new scientific work is another criterion.

The number of authors per study should generally reflect the contribution of the study to the paper.

Rapid publication from a WG should generally include authors from all member cohorts contributing data.

Consortium authorship for the writing group is another possible model that may be considered as an alternative model if there is no group consensus on named authorship.

7. Involvement of Other Collaborating Studies: Other collaborating studies may be proposed for inclusion in individual working group analyses. If the working group is part of this proposed consortium, then the other collaborating studies will be asked to abide by all of the principles set forth by the population-based studies consortium for the purpose of the proposed working group project.

8. Communication: WG members agree to communicate when an action is taken (ordering primers for genotyping) on the basis of shared data; when an analysis is completed for follow-up genotyping or when there is a proposal to modify publication plans.
9. **Data posting:** Upon publication in a scientific journal, shared results including results from joint analyses will be posted on a public website.

10. **Intellectual property:** According to the NIH GWAS policy (Federal Register; 72 (166) 28 Aug 2007: 49290-7), “it is the hope of the NIH that genotype-phenotype associations identified through NIH-supported and NIH-maintained GWAS datasets and their obvious implications will remain available to all investigators, unencumbered by intellectual property claims.” If in case intellectual property related to the GWAS’s covered under these principles is pursued by a participating investigator, their institution, or the study itself, then the consortium needs to be made aware of the pursuit of such claims. An abstract of the claim(s) needs to be made available to the consortium at the time of submission to the relevant governing body (e.g. US patent office).

In developing these guidelines, the cohorts benefited from guidelines developed by other genetics working groups, including the “International Lung Cancer Consortium Data Sharing and Access Policy” and the “Draft guidelines for sharing genome-wide association results for quantitative traits” (drafted by Gonçalo Abecasis, Michael Boehnke, Joel Hirschhorn, Karen Mohlke, David Schlessinger).

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