

Center for Inherited Disease Research

Application for Human Whole Genome SNP Linkage Scans

This application should be used only for Human Whole Genome SNP Linkage Scan projects. The following items should be provided to the CIDR Access Committee in a document not to exceed 10 pages (excluding appendices). Text may be single-spaced but the type size must conform to NIH guidelines, i.e., letter height no smaller than 10 point, type density no greater than 15 characters per inch, and no more than 6 lines within a vertical inch. Applications that do not conform to these guidelines will be returned.

Extramural NIH grantees supported by a participating institute require prior approval from the institute liaison before submitting an application to CIDR. Intramural NIH investigators should contact Dr. Camilla Day before preparing an application. See CIDR web site for institute liaison contact information – www.cidr.jhmi.edu.

Please address the following items in the order they are listed.

Principal Investigator Information

Provide contact information including:

- Name
- Institution and address
- Telephone, fax and e-mail
- Name and e-mail of contact person if other than PI

Project Analyst Information

Provide the name and affiliation of project analyst(s)

Co-Investigator / Collaborator Information

Provide the name and affiliation of major co-investigator(s) and collaborator(s)

Project Information

Provide information about the project including:

- Project title
- Whether the project has undergone previous peer review, and if so, by whom and the outcome of the review
- Whether this is a resubmission to CIDR, and if so, attach a 1-2 page cover letter addressing the criticisms from the previous review. Note: this cover letter does not count toward the page limit
- Current and pending funding sources relevant to this project (provide NIH grant number if applicable)
- If required, whether the project has been approved by the appropriate institute liaison

- How genotyping costs will be paid if the project is not supported by one of the thirteen NIH supporting institutes
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Sample Information

Provide information about the samples including:

- Approximate number of samples to be genotyped. Note: we strongly suggest submitting samples in multiples of 88
 - Source of DNA and extraction method used. Note: approximately 9 µg of DNA is required at a concentration of 75-150 ng/ul. A higher concentration range is recommended for whole genome amplified samples
 - When samples will be available to ship to CIDR
 - If samples will not be submitted at one time, if multiple releases are requested. Provide information on how the data sets will be merged
 - Whether the samples been previously genotyped, and if so, briefly describe. Note: previous genotyping is not required; this information is useful for internal quality control
 - The approximate number of parent-parent-child trios in samples. Note: experimental trios are not required; this information is useful for internal quality control
 - Whether the project has human subjects protection by an appropriate Institutional Review Board, and if so, attach a copy of approval; if no, whether plans been made to obtain such approval. Note: samples will *not* be accepted until IRB approval is received
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Project Details

Provide details about the project including:

- Abstract
- Specific aims
- Description of disease/trait
 - Describe the disease/trait to be mapped, which must be a complete outline of the phenotypic evaluation of the study subjects, including, where appropriate, specific inclusion and exclusion criteria. A copy of a questionnaire to collect phenotype information is not a sufficient definition of the trait to be mapped
 - Provide background information about the disease/trait to be mapped including the rationale for carrying out this particular study. Describe any unique features about the disease/trait that would single out this project for special consideration
- Evidence of genetic etiology - Provide evidence of a genetic component to the disease/trait. While this may be obvious with single gene disorders, it is not true for many complex traits. Provide summaries of twin studies, segregation analyses, estimates of relative risks, etc. to support the approach
- Data management - Describe plans to manage the large number of genotypes. Provide specific information about computing resources and software, and the training and experience of personnel charged with database management. If appropriate, provide information about how sample and family file data from separate collection sites will be coordinated for submission to CIDR
- Data analysis - Describe the analysis strategy for the resulting genotypic information. Include a discussion of the issue of power for the study, including power calculations where appropriate, and choice of analytic methods and software. If collaborations are established for analytical services with personnel at CIDR or otherwise, include letters of collaboration
 - Discuss the following points:
 - If applicable, what the largest pedigree is to be analyzed

- How map errors will be assessed since genetic maps are based on interpolations of existing wider-spaced marker maps and are likely to contain errors
 - How genotyping and Mendelian errors will be detected and what will be done to address these errors during data analysis
 - How linkage disequilibrium among the SNPs will be accounted for since many linkage software programs assume linkage equilibrium among all marker loci
- Plan for next phase - Provide the plan/strategy for the next phase of the project including the names of molecular geneticists and how they will be involved. Attach biographical sketches and letters of collaboration from these individuals

Appendix

Include the following documents in the Appendix:

- Literature cited
- CVs of key personnel in four-page, NIH format
- Letters of support/commitment from major collaborators and/or co-investigators
- Pedigree diagrams, if appropriate (indicate on the diagram if DNA samples are available for members of the pedigree)
- Essential reprints or preprints
- If required, documentation of institute liaison approval

Application Submission

Submit one original application and three complete copies, including appendices, to Dr. Camilla Day, Scientific Review Officer and Executive Director, CIDR Board of Governors, in the NHGRI Office of Scientific Review:

Camilla Day, Ph.D.
Center for Inherited Disease Research (CIDR)
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5635 Fishers Lane
National Human Genome Research Institute
National Institutes of Health
Bethesda, MD 20892-9306
(*Courier Services should use Rockville, MD 20852*)
Telephone: (301) 402-8837
FAX: (301) 435-1580
dayc@mail.nih.gov

At the time that you mail in your application, please send Dr. Day an e-mail message so that she will be looking for the application and can confirm receipt.

Last Updated 02/04/2008