Environmental Polymorphism Registry (EPR)

Instructions & Forms for Requesting Samples and Subject Identification

The NIEHS is establishing a repository of DNA samples collected as part of the Environmental Polymorphism Registry (EPR). This is an ongoing project initiated by the Office of Clinical Research to help NIEHS and other scientists screen for genetic polymorphisms in "environmentally sensitive genes". The DNA samples in the EPR are coded with unique identification numbers but these are linked to the subjects' identities, allowing investigators to recontact subjects with the "genotypes of interest" for follow-up studies.

Researchers can request DNA samples from the EPR by following the instructions below. NIEHS investigators are allowed to genotype the EPR samples under existing NIEHS IRB protocols (04-E-N053 and 02-E-N004). Outside investigators must follow the regulations of their own institution for obtaining IRB approval or IRB exemption in order to receive EPR samples for genotyping. Documentation of IRB approval or exemption will be required.

A major objective of the EPR is to facilitate studies that require ascertainment by genotype. Therefore, priority will be given to genotyping projects that are likely to lead to follow-up studies. However, follow-up studies will require their own IRB review and approval - no EPR samples will be decoded for follow-up studies until IRB documentation is provided. The instructions for requesting the identities and contact information of EPR subjects for follow-up studies are also below.

SAMPLE REQUESTS

Individual sample quantities are limited, so please request only the minimum amount required. The maximum limit is 1 μg DNA per sample. Samples can be requested by gender, race/ethnicity, age, and type of population (clinic or general population). For outside investigators who will seek IRB exemption, the key linking the samples back to the subjects' identities can be completely destroyed after they are shipped, if requested.

Investigators requesting EPR DNA samples for genotyping must do the following:

1. Complete the attached Sample Request/Genotyping Project Overview Form. The information for Item #10 should not exceed 2 pages. *All information provided in this Form will be kept confidential.*

2. If the recipient of the EPR DNA samples is a non-NIEHS investigator, he or she will be required to complete and sign the EPR Material Transfer Agreement (MTA).

Submit the Sample Request/Genotyping Project Overview Form to Michael Spencer, Clinical Program Coordinator (ext. 1-1168, spencermi@niehs.nih.gov).

Applications will be reviewed by the Core EPR Steering Committee which consists of the NIEHS EPR principal investigators (Drs. Perry Blackshear and Pat Chulada); Dr. David Resnick, Bioethicist; Dr. Paul Watkins, Director of the General Clinical Research Center, UNC; and Dr. Douglas Bell, Senior Investigator, Laboratory of Molecular Genetics (LMG). Subject matter experts might be added to the Core Steering Committee if needed.

If approved, the investigator will receive the required number and type of EPR DNA samples in coded form. Any investigator who receives EPR samples agrees to retain control of the samples at all times. The investigator further agrees not to transfer one or more EPR samples to other people not under his or her direct supervision, unless first obtaining permission from the Core EPR Steering Committee. The investigator also agrees to dispose of the EPR samples as directed by the Committee once the project is completed or after three years have elapsed. For outside investigators, these rules and responsibilities are also outlined in the mandatory EPR Material Transfer Agreement (MTA).

The Core EPR Steering Committee recommends and encourages that researchers publish all findings of new genotypes in dbSNP - http://www.ncbi.nlm.nih.gov/SNP/. Instructions for contributing SNP genotype data to dbSNP are available under the link:

SUBJECT IDENTIFICATION FOR FOLLOW-UP STUDIES

Once genotyping and data analysis are complete, if the investigator wishes to have samples decoded in order to recontact EPR participants for a follow-up study, she or he must do the following:

1. Obtain scientific approval based on the rules and regulations of his/her own laboratory, branch, and/or institution (NIEHS investigators only).

2. Apply to the Office of Clinical Research by completing the Pre-IRB Study Review Form and providing an IRB Study Protocol.

   For NIEHS investigators, information for the NIEHS IRB can be found at:
   http://dir.niehs.nih.gov/dirosd/ocr/irb/
   http://dir.niehs.nih.gov/dirosd/ocr/irb/irb_stan_format_04.doc

Submit the Pre-IRB Study Review Form and IRB Study Protocol to Michael Spencer, Clinical Program Coordinator (ext. 1-1168, spencermi@niehs.nih.gov).

Applications for follow-up studies and sample identification will be reviewed by an expanded EPR Steering Committee. For NIEHS scientists only, this can also serve as the pre-IRB scientific and statistical review required by the NIEHS for all clinical protocols. The expanded EPR Steering Committee will consist of the EPR PIs (Drs. Perry Blackshear and Pat Chulada); Dr. William Schrader, the Deputy Scientific Director; Dr.

David Resnick, Bioethicist; and Dr. Paul Watkins, Director of the UNC General Clinical Research Center. Depending on the complexity and scope of the study, other individuals might be added to the Steering Committee such as a medical and/or scientific subject matter expert, a biostatistician, and others relevant to the specific study. Once the application is approved, the investigator can then submit his or her IRB Study Protocol for IRB review. Documentation for IRB approval must be submitted back to the Clinical Program Coordinator before the EPR samples will be decoded and contact information provided.

Note - The NIEHS IRB will not review any project involving EPR subjects which has not been cleared by the EPR Steering Committee. Steering Committee clearance does not substitute for or guarantee IRB approval.

For questions concerning these applications, contact Michael Spencer (ext. 1-1168, spencermi@niehs.nih.gov). For questions concerning the EPR, contact Pat Chulada (ext. 1-7736, Chulada@niehs.nih.gov).

NIEHS Environmental Polymorphism Registry (EPR)
Sample Request/Genotyping Project Overview Form

*** Please complete this form in order to get approval from the EPR Steering Committee for genotyping. This is **not** the form needed to obtain clearance to have your project forwarded to the IRB.*
*** **If this form is not completed in full, it will be returned to the contact scientist**.*

1) **Project Title:**

2) **Principal Investigator's Name:**

3) **Contact Scientist's Name:**

4) **Phone Number:**

5) **E-mail Address:**

6) **Name of Institution and Address or NIEHS Lab and Group:**

7) **Signature of Laboratory Chief (NIEHS only) to indicate this study has been reviewed and approved (An attached email is acceptable):**

8) **List the following requirements for DNA request:**

   a) **Number of samples:**

   b) **Amount of DNA (1 µg limit) required per sample:**

   c) **Gender, ethnicity/race, and age requirements:**

   d) **Type of population required (clinic, general population or doesn't matter)**

9) **Name of gene (s) to be genotyped:**

   a) **Approximate number of SNPs:**

10) **Please answer the following questions:**

   a) **Will your project likely require recontacting EPR subjects for future follow-up studies (Yes or No) :**

b) **Could your requested samples be anonymized? (Yes or No)**
If yes, the sample key will be destroyed and sample decoding will not be possible.

10) Genotyping <u>Project Overview</u> (*Overview should not exceed 2 pages)*:

a) **Background: For each candidate gene provide a short summary, including any previous genetic studies:**

b) **For each gene listed, provide a list of specific SNPs (with dbSNP rs# if available) to be genotyped and relevance to function, clinical trait or disease (if known):**

c) **Genotype prevalence (estimation) in populations, if known:**

d) **Study hypothesis, as related to this specific genotyping request:**

e) **Any other relevant supporting data:**

Please submit form to:
Michael Spencer, spencermi@niehs.nih.gov , MD A2-05, Room A253

To be completed by EPR review personnel:

EPR Project number:

Date form received:

Approval Date:

NIEHS Environmental Polymorphism Registry (EPR)
Pre-IRB Study Review

*** This form, along with the IRB study protocol, will be reviewed by the EPR Steering Committee. If approved, the investigator can then submit the IRB protocol for review by the NIEHS or other relevant IRB. For NIEHS investigators, approval by the EPR Steering Committee can substitute for pre-IRB scientific and statistical review but cannot substitute for NIEHS IRB review. This is **not** the form needed to obtain EPR samples for genotyping.*

*** If this form is not completed in full, it will be returned to the contact scientist.*

1) **Project Title:**

2) **Principal Investigator's Name:**

3) **Contact Scientist's Name:**

4) **Phone Number:**

5) **E-mail Address:**

6) **Name of Institution and Address or NIEHS Lab and Group:**

7) **For principal investigators outside of the NIEHS only - has this study been reviewed for scientific merit? (YES or NO)**

   Write short description (two to three sentences) of how your study was reviewed for scientific merit?

8) Signature of Laboratory Chief (NIEHS PIs only) to indicate that this study has been reviewed and approved:

_____ _____

Signature of Lab Chief **Lab**

**** Attach IRB Study Protocol to form. Forms without a complete IRB Protocol will be returned.**

Please submit form to:
Michael Spencer, spencermi@niehs.nih.gov, MD A2-05, Room A253

To be completed by EPR review personnel

EPR Project Number:
Date form received:
Steering Committee approval date: