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500 Newton Road, 2182 ML
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FaceBase Biorepository

Protocol for Sample Access

Who Can Access FaceBase Biorepository Specimens and/or Data

Access to the FaceBase Biorepository specimens and associated data is intended only for scientific investigators, who are pursuing research questions that are related to understanding craniofacial birth defects and identifying genetic factors responsible for causing such diseases and disorders.

How to Request Specimens/Data

Completion of the FaceBase Biorepository Proposal for Use of Specimens and Data Form is required. This form consists of the Names/affiliations of the PI and team members, funding sources, IRB approval number, Federalwide Assurance Number (FWA #), a brief background/significance section, methods proposed including a power analysis and specific samples types/numbers/DNA quantity/phenotype data requested. Investigators are also required to provide a copy of their IRB approval memo or a determination from their local IRB their project does not fit the guidelines of human subjects research. Proposals will be reviewed within one month of submission and may require modification depending on the data access committee review. We welcome inquiries concerning sample/data options and these can be addressed to either Nichole Nidey, nichole-nidey@uiowa.edu or (319) 353-4365, or Jeff Murray, jeff-murray@uiowa.edu.

FaceBase Biorepository Specimens and data

The biorepository is comprised of DNA specimens from individuals affected with a craniofacial condition, including individuals with Syndromic and Isolated orofacial clefts and their unaffected family members. DNA specimens have been derived from Saliva, Cheek Swab, Blood and Tissue samples. In addition to DNA, information related to demographics, phenotypes, family history, medical history, clinical features and relevant genotyping data along with 3D facial images may be available. The biorepository also includes individuals with normal phenotypes with 3D image and saliva samples available. Investigators may request DNA specimens along with associated data from the biorepository and corresponding 3D images from the FaceBase Hub at the University of Pittsburgh. Inquiries concerning 3D images can be addressed to dac@facebase.org .

FaceBase Data Access Committee

The FaceBase Biorepository Data Access Committee (DAC) will review proposals from investigators who wish to utilize samples. The DAC consists of members of the greater research community (UI, FaceBase Consortium, and others) who have expertise in clinical work, research, and/or ethics. The DAC may utilize ad hoc members when either specific expertise is required to evaluate a proposal or an permanent member has a conflict of interest (as defined by NIH's rules for conflict in peer review: http://grants1.nih.gov/grants/peer/peer_coi.htm) with a given proposal. Current membership consists of the following individuals:

• Dr. Wendy Chung is the director of clinical genetics at Columbia University and has been active in both clinical care and applying genetic tecnnologies in studies of cardiovascular disease. She has background and experience in phenotyping, study design and molecular analysis.
• Dr. Michael Cunningham is a pediatrician trained in the diagnosis and management of children with cleft lip/palate and other craniofacial disorders. He has been a member of the Seattle Children's Craniofacial Center since 1993 and has served as the medical director for over 10 years. He has experience in the clinical care of orofacial clefting, measurements of treatment outcomes, and research into its pathogenesis.
• Dr. David FitzPatrick is a highly skilled clinician and a human molecular geneticist who has contributed greatly to our knowledge of craniofacial disorders. In addition, he has also overseen the EMAGE project generating 3-D images using mouse embryos and genes expressed in craniofacial development over the last five years.

Basis for Approval or Denials for Specimen/Data Access

Approval will be based on the DACs determination that the work proposed is relevant to FaceBase use and scientifically valid. Approvals will be rated yes/no and rejected investigators will be given the opportunity to resubmit and respond to short written critiques provided by the DAC. Investigators may be denied access to specimens and data in the biorepository if their proposed research questions or aims are not consistent with the informed consent research subjects signed when enrolling into the study, as mentioned above the primary goal of this project is to improve the understanding of craniofacial birth defects and genetic factors. If investigators request more than 10% of the total sample available the DAC may offer the investigator 10% of available samples or the DAC may consider a request of a larger amount. Samples will be provided to independent investigators/projects on a first in/first out basis with no priority assigned beyond DAC overall approval. Once 50% of the total sample is gone additional requests will also include notification of the DAC that sample use will now exceed 50% and they will determine if requests warrant additional diminution of the supply.

Guidelines for Using Biological Specimens from the FaceBase Biorepository

After notification of approval of access, the investigator will be required to complete the FaceBase Biorepository Usage Agreement.

- Please note the terms of usage for the FaceBase Biorepository:
    - The conditions for use of this research material are governed by the University of Iowa Institutional Review Board (IRB) in accordance with Department of Health and Human Services regulations at 45 CFR 46.
        - If the proposal is approved, the recipient investigator agrees to comply fully with all such conditions and to report promptly to the FaceBase Biorepository

Principal Investigator any proposed changes in the recipient's research project and any unanticipated problems involving risks to subjects or others. The recipient remains subject to applicable State or local laws or regulations and University of Iowa policies that provide additional protections for human subjects.

- The research material provided to the recipient may be utilized only in accordance with the conditions stipulated in this Usage Agreement, as approved by the UI IRB, as follows:
    - The recipient will abide by the human subjects regulations.
    - The recipient agrees not to attempt to identify the subject from whom the specimen was obtained.
    - The recipient will receive no information that could identify the subject.
    - If the recipient requests identifying information, the personnel of the FaceBase Biorepository will not provide it.
    - The recipient may not contact individuals who are collecting the material to obtain any identifying information.
    - All material is identified by a code number that is assigned by the FaceBase Biorepository for tracking purposes.
    - In addition to the research material itself, at the recipient's request, the FaceBase Biorepository may provide the recipient with the following information about the subject/material: sex, age, race, ethnicity, diagnosis, clinical features, medical history information, family history, and relevant genotyping data.
    - The recipient will **NOT** distribute specimens to third parties. Any requests to share specimens with other individuals must be approved by the FaceBase Biorepository Data Access Committee.

- Proposals which use materials within the guidelines and conditions outlined in the Usage Agreement do not require separate approval by the University of Iowa IRB.
    - However, separate IRB approval may be required by your institution.
- If your proposal would lead to the use of materials **BEYOND** the Usage Agreement, then prior review and approval by the University of Iowa IRB and, where appropriate, by an IRB at the recipient site (which must be convened under an application Office of Human Research Protections approved Federalwide Assurance) is required. In addition review by the DAC for use outside the original proposal will also be required.

Shipment of Specimens

After the biorepository receives all required documents for specimen & data access the study coordinators will initiate the shipments of specimens and transfer of data. The investigator requesting the specimens and data will be responsible for any associated shipping costs.

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FaceBase Biorepository Proposal for Use of Specimens and Data Form
www.facebase.org

Directions: Please complete this form and send to either Nichole Nidey, nichole-nidey@uiowa.edu or (319) 353-4365, or Jeff Murray, jeff-murray@uiowa.edu.

Investigator Submitting Proposal: **Please put Principle Investigator's Name here**

Investigator's Affiliation: **Please put name of institution here**

Research Team Members: **Please put names of research team members here**

Funding Sources: **Name of Funding Source and Grant #**

Federalwide Assurance Number (FWA #) for Affiliated Institution: **Please put FWA # here**

*The FWA # can be looked up at http://ohrp.cit.nih.gov/search/. Proposals will **NOT** be reviewed without an active FWA #. If you have questions about finding your institution's FWA # or about applying for a FWA #, please contact the FaceBase Biorepository Study Coordinator.*

Local/ Institutional IRB Approval Number:

Investigator's Contact Information:

Email Address:

Phone Number:

Fax Number:

Mailing Address:

Title of Proposed Project:

Aims/Research Questions your study is addressing:

Please provide a brief description

Background/Significance:

Please provide a brief description (one page or less)

Proposed Methods/Power Analysis:

Please provide a brief description (One page or less including plans for genotyping and/or sequencing)

Data Request

Specimen
What types of samples are needed?
☐ DNA, from Saliva ☐ DNA, from Blood ☐ DNA, from Tissue ☐ RNA

How many samples are requested? ☐

Minumum number of samples needed to complete the study? ☐

What is the DNA quantity needed? ☐

Please check the additional data needed for your study (*The following information about subjects may be requested without requiring additional review by the University of Iowa IRB. Please note this data may not be available for all subjects in the Biorepository*):

Sex ☐ Age ☐

Race ☐ Ethnicity ☐

Diagnosis ☐ Clinical Features ☐

Medical History ☐ Family History ☐

Genotyping Data ☐

Are you requesting additional information about subjects that would require additional review by the University of Iowa IRB? Yes ☐ No ☐

If so, why are you requesting the additional information? ☐

3D Images
3D Images and Saliva have been collected from a portion of our subjects. Investigators may request DNA samples from the Biorepository and 3D images from the FaceBase Hub, at the University of Pittsburgh.

Are you requesting 3D image from the FaceBase Hub? Yes ☐

No ☐

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FaceBaseBiorepository
UI IRB # for Repository (200912764)

USAGE AGREEMENT

The recipient acknowledges that the conditions for use of this research material are governed by the University of Iowa Institutional Review Board (IRB) in accordance with Department of Health and Human Services regulations at 45 CFR 46. The recipient agrees to comply fully with all such conditions and to report promptly to the FaceBaseBiorepository Principal Investigator any proposed changes in the recipient's research project and any unanticipated problems involving risks to subjects or others. The recipient remains subject to applicable State or local laws or regulations and University of Iowa policies that provide additional protections for human subjects.

The research material provided to the recipient may be utilized only in accordance with the conditions stipulated in this Usage Agreement, as approved by the UI IRB, as follows:

- The recipient will abide by the human subjects regulations.
- The recipient agrees not to attempt to identify the subject from whom the specimen was obtained.
- The recipient will receive no information that could identify the subject.
- If the recipient requests identifying information, the personnel of the FaceBaseBiorepository will not provide it.
- The recipient may not contact individuals who are collecting the material to obtain any identifying information.

- All material is identified by a code number that is assigned by the FaceBaseBiorepository for tracking purposes.

- Subject information will be kept confidential in locked file cabinets that can be accessed only be authorized study personnel or in password-protected computer files in a secure, non-public area and can only be accessed by authorized study personnel.

- In addition to the research material itself, at the recipient's request, the FaceBaseBiorepository may provide the recipient with the following information about the subject/material: sex, age, race, ethnicity, diagnosis, clinical features, medical history information, family history, and relevant genotyping data.

- The recipient will not distribute specimens to third parties. Any requests to share specimens with other individuals must be approved by the FaceBaseBiorepository Data Access Committee and will usually entail an independent request to the DAC.

- The recipient will acknowledge the FaceBaseBiorepository and the funding organizations involved in supporting the FaceBaseBiorepository in publications resulting from the analysis of the data.

- The recipient will provide the FaceBaseBiorepository Study Coordinator with copies of publications that arise from use of these specimens.

- Results of genotyping will be returned to the FaceBase Biorepository and placed into dbGaP or other publically accessible databases at the time of any publications reporting those results have been accepted for publication.

Any use of this material beyond the terms of this agreement requires prior review and approval by the University of Iowa IRB and, where appropriate, by an IRB at the recipient site, which must be convened under an application Office of Human Research Protections approved Federalwide Assurance.

If the recipient's use of this material is within the above guidelines and conditions, University of Iowa IRB review of the recipient's research project is not required.

_____ _____

Jeffrey C. Murray, Principal Investigator
FaceBaseBiorepository

Date (Month-Day-Year)

_____ _____

Recipient Investigator

Date (Month-Day-Year)

Recipient Investigator's Project Title:

_____

_____

_____ _____

Institutional Signing Official Date (Month-Day-Year)