

Illumina Genotyping Control Database Purpose Document

Introduction

This document provides a description of the Illumina Genotyping Control Database (the "Database"). It also explains the processes and limitations of submitting data to or downloading data from the Database, and will help researchers better understand the purpose of the Database.

Background

Genetic case control association studies are becoming more widespread in the scientific community. As part of an ongoing initiative at Illumina, we provide access to genotypic and phenotypic data from individuals that can be used as controls in case control association studies, wherein risk factors of individuals with a certain disease (cases) are compared to individuals without the disease (controls). Illumina provides an online data repository that is accessible to the scientific community to further scientific progress using these resources, and is subject to appropriate terms and conditions.

Access and Data Security Overview

Access and interaction with the Database is restricted to individuals who are registered in iCom, Illumina's e-commerce system. All activity of Illumina-provided web services, including access to and use of the Database, are monitored by iCom login ID for usage patterns that are outside the policies of the respective resources. To access the Database, a user must have an active iCom account, and enter their secure iCom login ID and associated password.

To enroll a new iCom account

1. Go to <https://icom.illumina.com/>
2. Follow the online instructions for New Customer registration
3. An iCom login ID will be assigned
4. An email confirmation will be generated, and a reply is required to successfully complete the registration process

iCom login IDs are assigned a scope of resource access that is reviewed periodically by an internal committee responsible for maintaining the security and health of Illumina's web resources. iCom login IDs that have been inactive for six months are periodically verified using the email and organization information that is associated with each iCom login ID.

Workflow

Submitting Data to the Database

1. Login to the Database using iCom ID and password
2. Accept and adhere to the terms and conditions of the Illumina Genotype Control Database Submission Agreement (the "Submission Agreement").
3. Submit approval documentation confirming necessary approvals, consents, and authorizations to submit the data to the Database, as applicable to the genotypic and phenotypic data and as required by federal, state and institutional laws, regulations and/or policies. (See "Phenotype Data" and "Genotype Data" tables at the end of this document). Approval documentation may consist of local IRB approval confirming that an appropriate informed consent corresponds to the data to be submitted and authorizes the data to be submitted to the Database for the Designated Purposes, as well as a representative sample of the informed consent. The approval documentation is stored in digital files that once submitted, will be controlled under 21CFR part 11.
4. The submission event (submitter identifier, acceptance of the Submission Agreement, approval documentation, and genotype/phenotype data) is logged within a transactional database for tracking purposes.
 - a. The genotype and phenotype data and the approval documentation are linked upon submission and uploaded into a staging area within the database that is accessible only to Illumina employees that are involved in the verification and validation of the data and the authorizations.

Potential Future Application

Consistent with Illumina's intent to provide this Database to facilitate scientific progress, Illumina recognizes that this effort may be advanced by cooperating with others working on similar projects. Specifically, at some point in the future Illumina expects to formalize an arrangement to share data from the Database to dbGaP (the database of Genotype and Phenotype), a database designed to archive and make available data from genome wide association studies (GWAS). dbGaP was developed and is managed by the National Center for Biotechnology Information (the "NCBI"), which is a division of the National Library of Medicine, part of the National Institutes of Health.

This is a potential future application and not a current Designated Purpose. As such, for any dataset submitted to the Database before submission to dbGaP is identified as a Designated Purpose, Illumina will not provide any such dataset for submission to dbGaP without receiving approval from the submitter that such dataset can be submitted to dbGaP. Once submission to dbGaP is identified as a Designated Purpose, submitters can evaluate at the time of submission whether they can confirm that their dataset can be submitted to dbGaP.

Phenotype Minimum Information Required

Gender (M/F/U)

Ethnicity

Age (number, queried by range)*

Optional Phenotype Data

Weight (number – queried by range)

Height (number – queried by range)

Blood type

Blood type (+/-)

Product name and revision

Phenotype(s) positive

Phenotype(s) negative

*Ages >89 years are represented as "89 years and older"

Genotype Data Captured in the Database

Beadpool versions

Cluster file name

Beadpool manifest name

GenomeStudio version

Normalization transformations

Submitter of genotype data*

Representative informed consent documents*

Genotype Data Captured for each SNP

Genotype in the top strand ("-" if no call)

Genotype in forward strand ("-" if no call)

GenCall score

Raw intensities in x and y (GRN and RED)

Normalization transformation index

Genotype Data Provided for Downloading (for each SNP)

Genotype in the Top Strand ("-" if no call)

Genotype in Forward Strand ("-" if no call)

*Not provided to users

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