

HUMAN GENETICS RESOURCES CORE

Under the direction of Dr. Richard Mayeux, the purpose of the Core is to facilitate genetic research in human populations campus-wide. It has three components: 1) Cell and DNA Repository for tissue culture, cell banking and DNA isolation and storage); 2) Database Management Systems for tracking of laboratory and clinical research information; 3) Data Analysis for the coordination of statistical analysis of genetic data from human studies. This Core is a resource for all departments, centers and institutes that wish to perform genetic research in human populations. Additionally, the Core provides cell banking, DNA isolation and storage for planned genetic research from the clinical (medical) genetics program. As such, it represents a key component of human genetics research at Columbia University Medical Center.

Physical Space

The current facility is located on the 19th floor of the Columbia University's Physicians & Surgeons Building. This space is split between five adjacent rooms. Four rooms are designated work rooms with bench space and freezers; the fifth room is an isolated cell culture room. The lab has 1,600 square feet and is furnished with brand new equipment which was purchased beginning in last couple of years. This equipment includes: tissue culture hoods, 6 incubators, -20 freezer, plate reader, centrifuge, 10 -80 freezers and PCR machine. The DNA and cell line storage facility will include freezers and one refrigerator. The entire lab is maintained through the Progeny LIMS and Brady bar-coding software systems.

Services Provided

I. Cell and DNA Repository (Tissue Culture and Cell Banking Facility and DNA Isolation and Storage).

This consists of a tissue culture facility with storage of transformed cell lines and a laboratory for isolation and storage of DNA. Buffy coats and lymphocytes are also catalogued and stored at the request of the investigators. The laboratory has the ability to store human tissue samples for genetic research. This serves as a campus-wide resource to support research in human genetics by all departments requesting such activity.

Marielba Zerlin, Ph.D. currently directs the day-to-day administration of the tissue culture/cell banking/DNA isolation and storage laboratory. She communicates with respective departments and investigators

At present there are 27,310 DNA samples and 7,099 lymphoblastoid cell lines stored in support of research by 36 investigators. In addition to the cell culture, DNA isolation and banking activities, this Core also provide maintenance and preparation of samples for genotyping activities.

II. Database Management Systems for Laboratory and Clinical Research Information.

Database systems are available for tracking samples throughout the Cell/DNA Repository. We have implemented a state-of-the-art database system to track samples through the collection, moving and storage of samples and laboratory data.

The demographic and clinical data is maintained in the Progeny Lab software system. This database is maintained by a fulltime programmer. This software provides a complete suite of pedigrees and genetic data management tools and provides a unique method of tracking, managing and viewing family history and genetic data. Progeny links family and clinical data, allows up-dating, provides access to multiple users, tracks contact information and blood collection and interview status. Progeny also records diagnostic information and has built-in data integrity checks and customized data checks for quality assurance. Progeny Lab is newly available and can help manage all laboratory aspects of a study from sample location through genotyping results.

A clinical genetic database system is available for data entry for information relating to the samples included in the Cell/DNA Repository. Imported and exported data for investigators would be provided and application development to generate files for linkage or association analyses and to batch information from genotyping or sequencing. Remote entry of data and manipulation of files can be provided. Maintenance capability, reporting and archiving would also be a component of the database. The Progeny Lab software system can support multiple independent investigators who wish to have access to their samples and is HIPPA compliant.

One senior programmer coordinates this activity. The programmer for the database management systems is housed in the GH Sergievsky Center/Taub Institute space.

III. Data Analysis.

This component of the Human Genetic Resource Core consists of a senior genetic data analyst who will provide support for investigators in the design and analysis of human genetic studies. This individual works with the existing group of genetic analysts on campus including Drs. Susan Hodge, David Greenberg, Joe Lee, Rong Cheng and Joseph Terwilliger. These individuals will be assisted by junior analysts to provide statistical genetics support to interested investigators on campus. The activities can range from study design for grant preparation to data analysis and interpretation for manuscript preparation. The data analyst is housed in the GH Sergievsky Center/Taub Institute space.