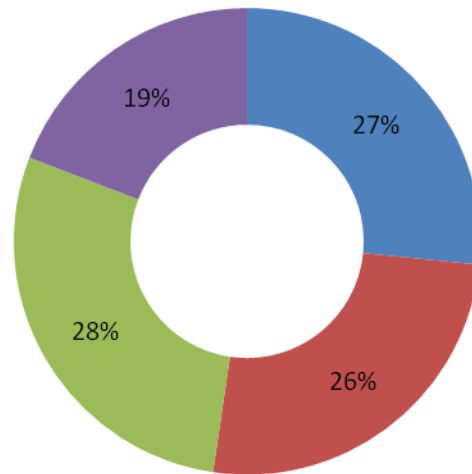


## Diagnoses Enrolled in Repository Core

■ Brain Malformation ■ Neurogenetics ■ Neuroimmunology ■ Other



### Two Approaches to Research

Topic	<i>Targeted Approach (PI specific)</i>	<i>General Approach</i>
<b>Focus</b>	Direct focus on specific disorders for specific PIs	Broader enrollment of multiple disorders
<b>Resources</b>	PI must obtain own resources for protocol development/management, enrollment, samples & data collection Samples and data will be stored within the Core	Use of some Core resources, i.e. approved protocol, study coordinator, samples & data collection, and data management Samples and data will be stored within the Core
<b>IRB</b>	PI must obtain project-specific IRB approval Templates available for protocol development	General IRB protocol approved and in place
<b>Sample Access</b>	Samples stored for PI use and can be shared only with approval of or collaboration with PI	Any investigator can access samples following approval by the Core Steering Committee

## **Repository Core Structure:**

The Neurology Core Steering Committee oversees all aspects of the Core, including IRB and HIPAA compliance, allocation of resources, and quality control of data collection, database entry, and specimen management. The Steering Committee develops and oversees policies to manage requests and prioritize use of potentially limited resources. The Core Steering Committee has, as a goal, to respect and support IRB-approved research interests of CHB faculty. The Core Operating Committee (OC) serves as a first level decision Committee; this Committee has the ability to go up to the Steering Committee to resolve any problems that may arise.

## **The Biorepository Core:**

This Core provides a neurological disease database and bio-specimen bank for translational research. The core includes a collection of various samples types (DNA, RNA, cerebral spinal fluid, brain tissue, fibroblasts) and clinical information from patients with neurodevelopmental disorders for outcome studies and trials. This core will collaborate with other existing collections of biological samples, including the hospital-wide Biobank, the Pathology Department collection, the Manton Center for Orphan Disease Research, the Gene Partnership Program and patient-specific stem cell lines from the Harvard Stem Cell Institute.

## **Sample and Data Collection:**

Examples of bio-specimens that can be obtained include DNA, RNA, blood, saliva, urine, hair, nail clippings, discarded CSF, fibroblasts, and discarded tissue samples or cell lines. Phenotypic data collection includes medical and family history obtained through the medical record, including age, ethnic background, health status, medical history, developmental history, and biological relationships between individuals. Other data obtained from medical records with the participant's consent include clinical evaluations, laboratory tests and their results, and any other necessary information retrieved from the participant's medical record as the PI sees fit.

## **Sample and Data Storage:**

All data obtained from the patient's medical record will be uploaded to REDCap (Research Electronic Data Capture), which is a secure web application for building and managing online surveys and databases. All coded data that are part of the Core will be stored in locked file cabinets in a secure place within Children's Hospital Boston or in a location stipulated by the Hospital. Bio-specimens will be stored using unique identification numbers within locked freezers at Children's Hospital Boston or in a location stipulated by the Hospital. DNA and other samples maintained at -20oC may be stored in Thermo Matrix 1.4 ml 2D barcoded screw-top tubes in a locked freezer with robotic access and processing, allowing maximum security and ease of accessibility. The Core also has -80 and -150 storage for biological samples that require this temperature for optimal storage.

## **The Core Steering Committee Members includes:**

Mustafa Sahin, MD, PhD (Neurology, Translational Neuroscience Center); Elizabeth Engle, MD (Neurology, Ophthalmology, and Medicine/Genetics, Programs in Genomics and Neuroscience); Mark Fleming, MD (Pathology); David T. Miller, MD, PhD (Medicine/Genetics; Laboratory Medicine); Charles Nelson, MD (Medicine/Developmental Medicine); Ellis Neufeld, MD (Medicine/Hematology-Oncology); Edward Smith, MD (Neurosurgery); Richard L. Robertson, MD (Neuroradiology); and Ingrid Holm, MD (Medicine/Endocrinology/Genetics, and Program in Genomics).

## **The Core Operating Committee (OC) includes:**

Mustafa Sahin, MD, PhD; Kira Dies, ScM; Mark Gorman, MD; Scott Pomeroy, MD, PhD; Annapurna Poduri, MD, MPH; and Elizabeth Engle, MD.

## **Contact Us**

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