

ADRC Participant Access Request

Access Request Goal

Goal - Formal request for ADRC data

Principal Investigator

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No Co-PI listed in survey

Study and Theme Details

Hypothesis

The goal of this project is to test the effects of genetic variants on the function of candidate cis-regulatory elements using massively parallel reporter assays, or MPRA. The study design uses genetic material from a pooled cohort as input which is randomly sheered and cloned into a reporter vector. The DNA and RNA is then read out from a cell of interest using sequencing, revealing regulatory activity levels. The presence of genetic variants allows for assessing their effects on regulatory activity.

Specific Aims

This aim is a part of my K99/R00, specifically Aim 3, which is:

3. Determine how non-coding variation alters A β -induced regulatory element recruitment. (R00)
 - A. Nominate common and rare non-coding variants that affect cis-regulatory element recruitment using SuRE-seq (van Arensbergen et al. 2019) with and without A β treatment in human neurons and microglia.
 - B. Further delineate function of cis-regulatory elements nominated in A. with confirmatory massively parallel reporter arrays and dCas9-KRAB repressions plus Capture-C to assess target genes.

Study related to Deep South Disparities

The goal of this project is to assess genetic variation effects, including variants that are enriched or exclusive to underrepresented populations.

Funding and IRB Details

Funding source - Already funded

Entity - NIH funded grant/application

Details - NIH R00 AG068271

IRB Contact - Yes, we have IRB approval

IRB Protocol # - This project is not human subjects research because it uses deidentified material.

Subject Sample Size and Profile

Sample size by cognitive ability

Normal Controls	50
MCI	25
Mild Dementia	25
Total N	100

Additional inclusion/exclusion details

We propose to use existing material from DNAs received to date at HudsonAlpha. NO NEW MATERIAL IS REQUESTED.

Racial minorities and other stratification

This study does NOT test hypothesis on racial disparities

Requested Resources

Banked biospecimen

Blood

DNA

Statistical support

Statistician has already been consulted - Greg Cooper and Brian Roberts