



## **COMBINEDBrain Biorepository Roadshow**

*Currently enrolling participants*

Principal investigator: CureSHANK, in partnership with CombinedBrain

### **About the study**

COMBINEDBrain is a non-profit consortium of over 60 patient advocacy groups for rare, genetic neurodevelopmental disorders, including CureSHANK. They recognize the need to collect patient samples for researchers to identify biomarkers to be used to treat /develop treatments for our children. They are on a mission to collect ~500 samples from our member organizations over the next 6-8 months and the CureSHANK is one of them. Samples collected from our community will be stored and available to researchers across the world.

**Are you interested in participating in this exciting project for the Phelan-McDermid syndrome community?**

## **HERE ARE THE BASICS:**

### **Who can participate?**

Any participant diagnosed with Phelan-McDermid syndrome (SHANK3 related) and their unaffected siblings.

### **What does participation involve?**

COMBINEDBrain will be collecting urine samples and blood samples (processed for plasma and a finger stick) to be stored in the COMBINEDBrain Biorepository and available for select biomarker projects as well as other interested researchers. They will also collect several online surveys to be completed by caregivers.

### **Where can you participate?**

COMBINEDBrain is collecting samples all across the United States this year during conferences being held by member organizations. Please see the list below of locations to find the closest biorepository collection to you. You may attend ANY of these meetings for a quick visit to provide the samples.

**Sample size:** COMBINEDBrain's goal is to collect 20 samples from each patient group, including Phelan-McDermid syndrome.

## How to enroll

If you can get to one of the conferences listed below, please send an email to [connect@cureshank.org](mailto:connect@cureshank.org) to confirm eligibility and complete the enrollment process. *COMBINEDBrain requests that those interested try to make contact at least one month in advance of the collection event you intend to attend.*

## Use of Clinical Research ID (CRID)

This study uses a CRID, or Clinical Research ID. This is a unique identification number generated and known only to the participant. This ID number allows researchers to merge data across research projects without any personally identifying information from the participant. *Please obtain a CRID prior to enrolling in this study, which is a simple online process that takes 2-3 minutes. Thank you for helping us improve data sharing in Phelan-McDermid syndrome research!*

## BIOREPOSITORY COLLECTION LOCATIONS:

### U.S. Northeast

#### **Madison, NJ**

June 22-24  
CTNNB1 Foundation

#### **Bethesda, MD**

June 23-25  
DYRK1A Foundation

#### **Queens, NY**

August 3-6  
KAND

#### **Washington, DC**

October 16  
COMBINEDBrain Meeting

### U.S. Midwest

#### **Cincinnati, OH**

September 22-23  
IRF2BPL Foundation

#### **Chicago, IL**

September 29-30  
KCNQ2 Cure Alliance

#### **Rogers, MN**

Oct 1  
Rory-Belle Foundation

### U.S. South

#### **Orlando, FL**

July 19-21  
KDVS Foundation

#### **Jacksonville, FL**

July 30-August 2  
Yellow Brick Road Project

#### **San Antonio, TX**

October 12-13  
TBRS Foundation

#### **Miami, FL** (not yet confirmed)

Nov 10-11  
FAST Global Science Summit

#### **Orlando, FL**

December 1  
SYNGAP1 Research Fund

#### **Orlando, FL**

December 1-3  
SLC6A1 Connect

### U.S. West

#### **Westminster, CO**

July 21-23  
STXBP1 Foundation

#### **Denver, CO**

October 6-7  
USP7/Prader-Willi

#### **Mercer Island, WA**

Oct 29  
FAM177A1

#### **Los Angeles, CA**

October 30-November 1  
ADNP Kids Research Foundation