

Minutes
Hirschsprung Disease Research Collaborative (HDRC) Conference Call
January 31, 2018, 1:00 pm EST

Attendees:

Aravinda Chakravarti, Courtney Berrios, and Jia Yan (Johns Hopkins University School of Medicine); Lois Sayrs (Phoenix Children's Hospital); Ling Fan, Ovais Aziz, Richard Wood, and Cheryl Gariepy (Nationwide Children's Hospital); Jill Ketzer (Children's Hospital Colorado); Hartley Nakimuli (Cincinnati Children's Hospital Medical Center); Lynda Painting (University of California, Davis); Jack Langer (Hospital for Sick Children); JoAnn DeRosa (Johns Hopkins All Children's Hospital); Phil Frykman (Cedar Sinai Medical Center); Raj Kapur (Seattle Children's Hospital); Michael Rollins (University of Utah Primary Children's Center)

- I. Welcome
- II. HDRC Coordinating Center Move
 - a. The official start date at NYU for the Chakravarti lab will be April 2, 2018. The lab will physically move during the last week of March.
- III. Membership and samples
 - a. During the period from Tuesday, October 17, 2017 through Tuesday, January 30, 2018 the following have been enrolled: Valley Children's - 1 family, Hospital for Sick Kids – 1 family, Gadjah Mada – 12 families, Texas Children's Hospital – 1 family.
 - b. HDRC membership update: Jill Ketzer has submitted the IRB application for the HDRC's newest site, the Children's Hospital Colorado, and the application has been approved. Congratulations Jill and the team at CHCO!
 - c. The current enrollment count includes 455 probands and 645 of their family members, for a total of 1100 participants who are enrolled in the HDRC as of January, 30, 2018. We sincerely thank everyone for their continuing efforts to enroll participants for the biorepository.

Membership Summary Table

Category	Total
Proband	455
Father	250
Mother	364
Sibling	21
Other relative	10
Total	1100

- IV. HDRC Coordinating Site move to NYU
 - a. Update on MTAs
 - i. JHU has drafted an amendment to the existing MTAs regarding relinquishing samples from JHU to NYU. NYU is working on adding its own amendment language to include confirmation that NYU will serve as the coordinating site for the HDRC and that NYU now enters into the same agreement that JHU had with each HDRC site. Once this draft is complete, we will send both the JHU and NYU statements as an amendment to the existing MTAs to the legal and contracts

teams at each HDRC site. Each HDRC site can then review and either agree and sign or request any changes. Once the authorized institutional signing official at the HDRC site has signed, the respective authorized institutional signing officials at JHU and NYU will then sign.

- ii. Phil Frykman suggested that it would be helpful to have an accompanying memo summarizing the gist of the amendment – that it states NYU will enter into the same agreement that the HDRC site has with JHU. Jia Yan will write a memo to go with each amendment.
- iii. Jill Ketzer asked whether or not CHCO, as a new HDRC site, should hold off on the MTA until the amendment is complete. Jia Yan checked with JHU’s Office of Research Administration about this and they suggested that the new HDRC site move forward with the MTA process, as the amendment will not change the language of the original agreement with JHU. Once the amendment is ready, it will be signed along with the original parent MTA that JHU already had in place with the site.

V. Samples for HDRC

- a. Update on samples – last date to send samples to JHU will be March 1, 2018. Samples sent that day will be processed when JHU receives the samples on March 2, 2018. Resume date at NYU TBD once lab has physically moved to NYU.

VI. Update on database

- a. Jia Yan gave an update on the database development - a REDCap data dictionary has been created by Ray Everngam from the Hendren Project. This consists of the questions in the HDRC study questionnaire. However, the sample information including blood sample, DNA concentrations, dilutions from original samples, and family history structure that is currently represented in the PostgreSQL database that JHU has developed is not in the data dictionary. REDCap may not have the functionality to preserve this information, so some of these features will need to be removed in order to migrate all data to REDCap. An alternate option is to develop the REDCap database for the purpose of collecting clinical information and have a separate database to manage sample data.
- b. Discussion
 - i. Raj Kapur said that the option of having the clinical information in REDCap that can link to the SQL database containing sample information would be a good idea.
 - ii. Michael Rollins said that OpenClinica, which is used by the PCPLC, can allow for import of data. Jack Langer said that the thought of combining data is to link data between the PCPLC rather than importing the data. This is particularly important as HDRC is not currently gathering longitudinal data.
 - iii. Cheryl Gariepy said that it would be best not to lose functionality and perhaps it would make sense to figure out a method to link the genetic, sample and clinical data.
 - iv. Making changes to REDCap database: In regard to REDCap functionality, Michael Rollins said that in his experience a great deal of design decisions should be made at the front end before data collection, such as number of entries. Richard

Wood has found that REDCap does allow for a number of adaptations, but that the branching logic needs to be evaluated and set. REDCap is able to track changes and flag changes that may be problematic.

- v. The coordinating site has contacted the REDCap team at NYU and will work with the HDRC to complete development of the database there.
- VII. Scanning all questionnaires and medical records for all records for each site
- a. Keep family together and index by:
 - i. Sample tracking
 - ii. Questionnaire
 - iii. Medical records
 - b. May incorporate into database for use a reference document, though all data will be entered into the database.
- VIII. Member comments/feedback
- a. Jack suggested that it would be helpful to have an HDRC meeting going over the data that the HDRC has so far. Jia agreed and will schedule this meeting after pulling together detailed summaries of the phenotypic data.
- IX. Next Quarterly Call: TBD