

**Minutes**  
**Hirschsprung Disease Research Collaborative (HDRC) Conference Call**  
**January 6<sup>th</sup>, 2017, 3:00pm EST**

Attendees:

Aravinda Chakravarti, Dallas Auer, Juli Bollinger, Courtney Berrios, and Jia Yan (Johns Hopkins University School of Medicine); Kara Kronemeyer, Julie Robbins (Phoenix Children's Hospital); Misty Troutt (Cincinnati Children's Hospital Medical Center); James Dunn (University of California, Los Angeles); Belinda Carlisle (Boston Children's Hospital); Gilford Cage (missed institution); Phil Frykman (Cedars-Sinai Medical Center); Raj Kapur (Seattle Children's Hospital), Lynda Painting and Laura Cronin (University of California, Davis); Ling Fan (Nationwide Children's Hospital); JoAnn DeRosa (Johns Hopkins All Children's Hospital)

- I. Welcome
  - a. Dr. Aravinda Chakravarti welcomed everyone to the call and thanked them for joining.
  - b. Introduction of new HDRC coordinator, Dr. Jia Yan. Dr. Chakravarti introduced Dr. Yan, whose background and training is in research and genetic counseling, and who joined the lab on October 31<sup>st</sup> of 2016. Her contact information is as follows: [jyan13@jhmi.edu](mailto:jyan13@jhmi.edu), 410-502-7541. Please don't hesitate to contact her with any questions about the HDRC.
- II. HDRC Update
  - a. Membership
    - i. The University of California, Davis has recently begun active enrollment.
    - ii. Please see attached HDRC Membership table.
  - b. Samples
    - i. Many HDRC sites have continued to enroll families for the biorepository over the past quarter. As of Dec. 1st, 2016: All Children's Hospital 2 families, Texas Children's Hospital 2 families, Valley Children's 1 family, Cincinnati Children's 4 families, Penn State Hershey Children's Hospital 2 families, University of Utah/Primary Children's Hospital 2 families, Seattle Children's Hospital 2 families, University of Tennessee Health Science Center 1 family. The current enrollment count includes 380 affected individuals and 540 of their family members. We sincerely thank everyone for their continuing efforts to enroll participants for the biorepository.
- III. HDRC Advisory Board Updates
  - a. Dr. Chakravarti acknowledged that Dr. Belinda Carlisle, who is the head of the HDRC Advisory Board, is on this call and invited her to give an update. Dr. Carlisle said that the HDRC Advisory Board has had 2-3 calls. They have reviewed the HDRC grant and critique of the grant to gather information for recommendations for the HDRC. They felt that the grant is currently a bit ambitious and suggested that it may be helpful to focus on 1-2 questions for the grant.
- IV. HDRC and the PCPLC Discussion – tabled for discussion at future HDRC call

- a. Dr. Carlisle and Dr. Chakravarti also discussed possible duplicated efforts between the HDRC and the colorectal consortium, the PCPLC (Pediatric Colorectal and Pelvic Learning Consortium), which includes patients with Hirschsprung disease in addition to patients with other pelvic anomalies. There may not be as large of a duplication of effort between the two as it appears, and the information gathered within the PCPLC needs to be clarified. A discussion of possible data sharing between the HDRC and PCPLC will be deferred to a later meeting once the HDRC Program Steering Committee acquires additional information and has a resolution about data sharing with entities outside of HDRC members, which is not as straightforward as data sharing within members. Some of the areas of concern include IRB and legal issues, such as details in the Materials Transfer Agreements and Informed Consent Documents.

V. Database

- a. Update on recent discussions
  - i. The HDRC has been investigating a database in which all members can log in in a secure way to access subject data, as approved by the IRB. Dr. Chakravarti reported that HDRC Program Steering Committee has explored the use of a RedCap database. They have been in discussion with Dr. Michael Rollins as well as the Hendren Project. Both groups use RedCap. There is a need for electronic data entry, with paper records retained for data validation. Additionally, Dr. Raj Kapur is collecting information for a pathology repository. Discussion of the details of the repository will take place at the next meeting.
- b. Member comments/feedback
  - i. Jia Yan said that information and feedback about individual needs of member institutions, such as type of EHR used, will be helpful for development of a shared database that can accommodate a variety of data uses and analyses. A survey will be circulated to aid this assessment.

VI. Sample and data quality

- a. Ms. Courtney Berrios has previously made assessments of clinical data quality and there is missing phenotypic data for which there are ongoing follow-up efforts. In regard to sample quality, Ms. Dallas Auer discussed that in cases in which the submitted blood volume is low, the DNA quality and yield is low. The lab is working on improving methods for samples with low blood volumes.

VII. Sample and data sharing: group discussion

- a. Dr. Chakravarti invited discussion and questions proposed by members. Ms. Misty Troutt discussed that it would be beneficial to have a greater source of DNA, as DNA samples are limited. Because this is a limited resource, the group needs to determine the most equitable way of sharing data. For example, when two groups want to do the same project, a process needs to be in place to figure out the most equitable way to accommodate the groups. Dr. Chakravarti raised the question of how to defray costs of transporting samples and managing sample sharing, such as a potential fee to help with these costs. Timely return of samples is also essential and a timeframe needs to be agreed upon.

- b. Dr. Phil Frykman said that he would like to define sharing better and to do this in the most cost efficient manner, and asked whether a nominal fee would potentially be different depending on number of samples sent. Dr. Chakravarti said that depending on samples requested, costs may not necessarily correlate with number of samples; at times, it may be even more costly to send fewer samples compared with hundreds of samples.
- c. Dr. Raj Kapur agreed with Dr. Frykman and raised a question of whether costs should be determined by individual institution's contributions.
- d. Dr. Belinda Carlisle said that implementing a fee may deter investigators from contributing to the HDRC. Obtaining funding for the HDRC would help with this aspect and continued discussion is warranted.

Dr. Chakravarti thanked attendees and adjourned the call.

**Next Meeting Date: Friday, April 28, 2017 from 3:30 pm – 4:30pm Eastern Standard Time**