

Minutes
Hirschsprung Disease Research Collaborative (HDRC) Conference Call
September 30th, 2016, 3:00pm EST

Attendees:

(Aravinda Chakravarti, Dallas Auer, and Juli Bollinger, Johns Hopkins University; Andrea Bischoff, Children's Hospital Colorado; Carolyn Urish, Phoenix Children's; Caroline Loner, Cincinnati Children's; Dorothy Rocourt, Penn State Hershey Children's; Jana Creps, University of Michigan; Richard Wood, Nationwide Children's; Phil Frykman, Cedars-Sinai Medical Center; Michael Rollins, Primary Children's, University of Utah; Ankush Gosain, University of Tennessee Health Science Center; Representative (missed name), Texas Children's)

- I. Welcome
 - a. Dr. Aravinda Chakravarti welcomed everyone to the call and thanked them for joining.
 - b. Dr. Chakravarti announced that Dr. Daniel Teitelbaum (University of Michigan Mott Children's) passed away last month. Dr. Teitelbaum's enthusiasm and efforts for the HDRC will be sorely missed.
 - c. Dr. Chakravarti discussed the staff changes that are occurring in his lab. He introduced Dr. Jia Yan, a newly-hired genetic counselor and research scientist who will begin working next month; Ms. Juli Bollinger, an experienced genetic counselor who will be assisting the lab in its transition over the next few months; and Mrs. Dallas Auer, Dr. Chakravarti's lab manager and senior technician who has been assisting in sample collection and study coordination since Ms. Courtney Berrios left Johns Hopkins in July 2016.
- II. HDRC Update
 - a. Membership – Mrs. Auer reported that there have been no new developments in terms of membership in the last quarter.
 - b. Samples – Mrs. Auer reported that many HDRC sites have continued to enroll families for the biorepository. In the last quarter, Children's Hospital of Central California enrolled 2 families, Cincinnati Children's 5, Pennsylvania State Hershey Children's Hospital 7 (their first), University of Utah/Primary Children's Hospital 4, and Seattle Children's Hospital 4. The current enrollment count includes 364 affected individuals and 515 of their family members. Mrs. Auer thanked everyone for their continuing efforts to enroll participants for the biorepository.
- III. Advisory Board Meeting
 - a. Dr. Chakravarti relayed that Ms. Corinne Bridges has been selected to serve as a member of the HDRC Advisory Board as a parent advocate.
 - b. Dr. Chakravarti shared that he met with the HDRC Advisory Board on August 25th and will serve as their liaison to the HDRC. Their advice will be extremely valuable and instructive as we move forward.
- IV. Data/Sample Collection and Sharing
 - a. Dr. Chakravarti stressed the importance of improving the quality of the samples and data that the HDRC collects. Dr. Phil Frykman added that the HDRC needs to find workable policies on data and sample sharing that will benefit everyone, keeping in mind that specimens are finite but data is not. The HDRC Program Steering Committee will discuss improving the repository during their meeting next week.

- b. Dr. Chakravarti stated that his lab's investigation of the genetic factors involved in Hirschsprung Disease is temporarily postponed during the personnel transition his lab is currently experiencing, but he will provide an update during the next call.

V. Database Options/Discussions

- a. Dr. Chakravarti discussed the importance of sharing internal HDRC information, perhaps through intranet access on the HDRC website for study members. His group will work on developing this in the coming weeks.
- b. In terms of a database for HDRC patient information, it was agreed that it is important to ensure remote access for all champions, PHI protection, and the separation of clinical and genetic data. Dr. Chakravarti also emphasized the long-term difficulties that result from setting up a database too rigidly too early in its development.
- c. The Hendren Project and Michael Rollins should be consulted for their expertise as the HDRC selects a database. The Advisory Board has been consulted on this issue. Database development will be discussed at the next Program Steering Committee meeting, and they will present a plan during the next quarterly call.

VI. Grant Planning

- a. The HDRC grant has so far been submitted twice to the NIH and while it scored well, it did not score well enough to be funded. Dr. Chakravarti reported that the National Institute of Diabetes and Digestive and Kidney Diseases (NIDDK) and the National Human Genome Research Institute (NHGRI) both expressed interest in funding portions of the grant, but neither was very enthusiastic about funding it in its entirety. Perhaps a co-funding partnership between the two would be successful.
- b. Main goals for the HDRC study include the collection of longitudinal and pathological data to research quality-of-life assessments and clinical outcomes, as well as the collection of genetic data. The possibility was discussed to split the grant into two portions: clinical outcomes and genomic research. This may increase the probability of obtaining funding. The Advisory Board has been consulted for their opinion, and the Program Steering Committee will discuss their deliberations.