

Minutes
Hirschsprung Disease Research Collaborative (HDRC) Conference Call
March 24th, 3:00pm EST

(Attendees: Aravinda Chakravarti and Courtney Berrios, Johns Hopkins University; Jack Langer, Hospital for Sick Children; Caroline Loner, Cincinnati Children's Hospital Medical Center; Lisa Iamicelli, University of California Los Angeles; Kara Kronemeyer, Phoenix Children's Hospital; Hector Monforte, All Children's Hospital; Phil Frykman, Cedars Sinai Medical Center)

- I. Welcome – Courtney Berrios welcomed everyone to the call and thanked them for joining.
- II. HDRC Update (see attached HDRC Membership table)
 - a. Membership – Ms. Berrios reported that there have been two new IRB approvals over the last quarter; the University of Tennessee Health Science Center with PI Ankush Gosain and Penn State Hershey Children's Hospital with PI Dorothy Rocourt. In addition, UC Davis with PI Jacob Stephenson has submitted their application and it's in IRB review.
 - b. Samples – Ms. Berrios also reported that samples have been received from a variety of sites over the past quarter. Texas Children's has enrolled 2 families, Cincinnati Children's 2 families, Connecticut Children's 2 families, Emory University 2 families, Hospital for Sick Children 1 family, University of Utah/Primary Children's 5 families, Seattle Children's 5 families, University of Alabama Birmingham 2 families, Universitas Gadjah Mada 11 families. This brings the total HDRC enrollments to 322 affected individuals and 448 of their unaffected family members.
Aravinda Chakravarti thanked everyone for the work in enrollment and noted that the HDRC now has adequate numbers to begin some analyses, which will be discussed later in the call.
- III. Advisory Board – Ms. Berrios reviewed that in the previous quarterly call it was reported that Belinda Dickie has agreed to chair the HDRC Advisory Board. Dr. Dickie has now begun building the board, based on suggested names by the Steering Committee, HDRC members and Dr. Dickie, and so far has agreements to participate from Richard Wood, Sam Nurko, Nikhil Thapar, Hector Monforte. Dr. Chakravarti suggested that it may be possible for the Advisory Board to meet during the upcoming APSA Annual meeting.
- IV. Analyses
 - a. Publication – Ms. Berrios reminded the group that in a previous call Raj Kapur had mentioned the acceptance for publication of small, retrospective study of pathology findings with genetic variants carried out in the early stages of the HDRC to demonstrate feasibility of collaborative projects for the group. The paper is now published and the citation is below. A PDF will be distributed with the minutes.
Swaminathan et al. Intestinal Neuronal Dysplasia-like submucosal ganglion cell hyperplasia at the proximal margins of Hirschsprung disease resections. *Ped Devel Pathol*, Nov-Dec 2015.
 - b. Data and sample requests – Dr. Chakravarti reported that the Steering Committee has received its first inquiry about use of the samples and data by an enrolling site.

Cincinnati Children's has inquired about the process and the Steering Committee is beginning to evolve procedures to request use of samples and data for projects.

- c. Genetic analyses – Dr. Chakravarti also discussed that with the number of samples now available he can begin to use his existing genetics grant to complete genetic analyses. The lab is beginning to pursue these now. All data from these analyses will be returned to the HDRC to be available to other HDRC members.
 - d. Clinical data – Phil Frykman discussed that the Steering Committee has begun to look at some of the clinical data collected in hopes of identifying some pilot data for future grant proposals. Some concerns of data completeness and quality emerged (ie. missing pathology reports), which we need to discuss and address. Ms. Berrios added that as some initial analyses are completed the Steering Committee can share with all so that on the next call the group can discuss strategies for improving data quality. Hector Monforte-Munoz requested that this review include the number of pathologists involved at sites and reports of what is missing, including numbers of pathology reports.
- V. Grant planning – Dr. Chakravarti reported that in the most recently Steering Committee call the members discussed splitting the previous HDRC grant proposal into two proposals; one heavily focused on outcomes and one focused on genetics. Given the challenge of being assigned to a study section with expertise and interest in both and the comments in previous reviews, it may be more successful to target two separate grants to reviewers who share that focus. The Steering Committee will continue to explore this option as they plan further grant submissions.
- VI. HDRC Meeting at APSA – The HDRC will meet in conjunction with the Hirschsprung Disease Interest Group at the APSA Annual Meeting in San Diego. The meeting will be held on Sunday, May 15th at 6:30 am. The location will be sent closer to the date.

The next HDRC quarterly call will be June 22nd 1:00pm ET / 10:00am PT.