

**Minutes**  
**Hirschsprung Disease Research Collaborative (HDRC) Conference Call**  
**June 22nd, 1:00pm EST**

(Attendees: Aravinda Chakravarti and Courtney Berrios, Johns Hopkins University; James Dunn, UCLA; Phil Frykman, Cedars-Sinai Medical Center; Raj Kapur, Seattle Children's; Ann Mehringer and Jana Creps, University of Michigan; Kara Kronemeyer, Phoenix Children's; Misty Troutt, Cincinnati Children's; Jen Hall and Andrea Bischoff, Children's Hospital Colorado; Stephanie Falwell, UC Davis; Ankush Gosain, University of Tennessee Health Science Center)

- I. Welcome
  - a. Dr. Aravinda Chakravarti welcomed everyone to the call and thanked them for joining.
  - b. Dr. Chakravarti also announced that the study coordinator at the HDRC Coordinating Center, Courtney Berrios, will be leaving for a new position in the near future. He thanked Ms. Berrios for her work in getting the HDRC started and its continued growth.
- II. HDRC Update (see attached HDRC Membership table)
  - a. Membership – Ms. Berrios reported that UC Davis has received IRB approval from the university and is currently working through the MTA and hospital IRB approval. In addition, Children's Hospital Colorado with Andrea Bishcoff as PI and Jen Hall as coordinator (both previously started the HDRC at Cincinnati Children's) are beginning the process to join the HDRC for Colorado Children's.
  - b. Samples - Ms. Berrios reported that many HDRC sites have continued to enroll families for the biorepository. Over the past quarter, Valley Children's Hospital enrolled 2 families, Cincinnati Children's Hospital Medical Center 3, Connecticut Children's Medical Center 1, Emory University/Children's Healthcare of Atlanta 2, Hospital for Sick Children 2, Medical University of South Carolina 2, University of Utah/Primary Children's Hospital 4, Seattle Children's Hospital 4, University of California Los Angeles 1, University of Tennessee Health Science Center 1 (their first). This brings HDRC total enrollments to 345 affected individuals and 475 of their family members. Ms. Berrios thanked everyone for their continued efforts to enroll participants for the biorepository.
  - c. Changing Coordinating Center Contacts – Ms. Berrios discussed that with her departure there will be some changes in the contacts for the HDRC Coordinating Center. Ms. Berrios' last day physically at JHU will be July 1<sup>st</sup>, but she will be working remotely in coordination with other lab members through July as others take over the coordination roles. For now, existing lab members will be helping out until a new full-time coordinator is hired. Day to day contacts about samples arriving, kits needed, questions should be directed to the Coordinating Center's senior research technicians, Dallas Auer [dallas@jhmi.edu](mailto:dallas@jhmi.edu) and Maria Sosa [msosa2@jhmi.edu](mailto:msosa2@jhmi.edu). Dr. Chakravarti, [aravinda@jhmi.edu](mailto:aravinda@jhmi.edu), will handle inquiries about joining the HDRC. Ms. Berrios will send a separate email about these contact changes out to the full mailing list.
- III. Advisory Board – Ms. Berrios reported that the members of the Advisory Board have been finalized. The members are: Belinda Dickie, chair, Michael Rollins, Richard Wood, Samuel Nurko, Hector Monforte, Nikhil Thapar, Stan Lyonnet, and a parent advocate. It's been a

challenge to find a time that all can meet, but they are working on scheduling the first meeting.

IV. Analyses and Data Sharing

- a. Dr. Chakravarti and Ms. Berrios discussed that the Steering Committee has been working on the process for member sites to request data and samples for use in projects. An application form has been developed that requests basic information about the project, including a project description of 2 pages or less and information about what data will be returned to the HDRC. Acknowledgement of the HDRC in some way is required, but it has not been determined whether this should be in the acknowledgements or with the HDRC on the author list, and this may vary by project. Andrea Bischoff suggested that this could be decided by the Advisory Board or a vote of the HDRC members. Ms. Berrios will distribute the application form with the minutes for this call.
- b. Three projects have been approved for data use. Dr. Chakravarti's group is using the DNA samples to assess regulatory variants, Dr. Kapur from Seattle Children's will review pathology reports, and Dr. Helmrath from Cincinnati Children's is looking at some genetic variants in relation to outcomes. Preparing data and samples for these projects will be an experiment in the processes of the HDRC and help develop the best ways to do so for future requests.
- c. Dr. Kapur and Dr. Chakravarti also discussed that at some point the HDRC must decide if there will be some contribution or small charge for obtaining samples to offset the work to pull data and samples together. This may need to be included in grants. This could also be something for the Advisory Board to weigh in on.

V. APSA Meeting – Raj Kapur reported that he attended the Hirschsprung Disease Interest Group meeting at the APSA Annual Meeting and was pleased that 15-20 people came out for the early morning meeting. Much of the discussion centered around the interest and work of various groups (HDRC, REACH, Nationwide Children's, University of Utah) to develop a database for Hirschsprung disease research and ways in which these groups could potentially work together to avoid duplication.

- a. Andrea Bischoff mentioned that The Hendren Project also has a funded database system that is currently being used for other conditions and could perhaps be adapted for Hirschsprung Disease.
- b. Richard Wood mentioned the importance of several groups not working in parallel and duplicating efforts.
- c. Ash Gosain also discussed that important discussions about the granularity of data collected will need to be made and that while one database system or interoperability would be useful across all project/sites, there may need to be different types of data and levels of granularity collected at differing sites due to the level of work needed to enter data and ability or not to pull data from the EMR.
- d. The HDRC will continue to explore database options and work with other groups who share this goal, while also pursuing funding for this effort.

- VI. Grant Planning – Dr. Chakravarti reviewed that the HDRC has submitted a grant to the NIDDK twice and although it was reviewed and at last submission scored at the 30<sup>th</sup> percentile, this was not good enough to be funded. The Steering Committee is taking pause before a 3<sup>rd</sup> submission and looking at separating the work into 2 grants, one that focuses on genetics and one that focuses on outcomes research. This may allow a more targeted review with experts in each field. Once the Steering Committee has consensus on aims they will pass them around to the group for feedback.