

Agenda

Hirschsprung Disease Research Collaborative (HDRC) Conference Call

September 18th, 2:00pm EST

(Attendees: Rebecca Ragar, Phoenix Children's; Raj Kapur, Seattle Children's; Courtney Berrios, Johns Hopkins; Lindsay Stephens, Texas Children's; James Dunn, UCLA; Ann Mehringer, University of Michigan; Michael Rollins, University of Utah; Ankush Gosain, University of Tennessee Health Science Center; Richard Wood, Nationwide Children's; Michelle Shroyer, University of Alabama Birmingham; Misty Troutt, Cincinnati Children's; Megan Durham, Children's Healthcare of Atlanta)

- I. Welcome – Courtney Berrios thanked everyone for taking time out of their day to join the call.
- II. HDRC Update (see attached HDRC Membership table)
 - a. Membership – Ms. Berrios reported that two new sites have obtained IRB approval; the University of Alabama Birmingham with Robert Russell as PI and University Gadjah Mada in Indonesia with Dr. Gunadi as PI.
 - b. Samples – Ms. Berrios also reported the sample received over the last quarter from existing sites. Sample submissions include: All Children's Hospital, 1 family; Cincinnati Children's, 7 families; Connecticut Children's, 1 Family (their first); University of Michigan, 2 families (their first); Nationwide Children's 1 family; Seattle Children's, 5 families; Hospital for Sick Children, 4 families; Texas Children's, 1 family; University of California Los Angeles, 2 families; and University of Utah, 9 families. Thanks so much to everyone and their study teams for work on enrollments!
 - c. Analyses
 - i. Ms. Berrios reported that the Johns Hopkins site has received approval from the Steering Committee to begin analyses of common genetic variants in the HDRC samples. These include polymorphisms that have been associated with risk for Hirschsprung disease, including enhancers of the RET gene.
 - ii. Raj Kapur reported that he was recently informed that a paper using the HDRC network has been accepted for publication. The paper involved a small subset of HDRC institutions (5 planned, but 3 ended up participating), and was a retrospective study involving separate IRB approval for collection of archived pathology samples with associated clinical data. The study involved histopathological analyses of the transition zone (done at Seattle Children's) and genotyping of common polymorphisms (completed at Johns Hopkins). Tests for correlations of pathology findings with common polymorphisms were thus far negative. This is the first paper published using the HDRC network and Dr. Kapur hopes it can be a proof of principle of an HDRC collaboration. A link to the manuscript will be provided through the HDRC website, once a pdf is available.
- Q. Michael Rollins asked how the interest of HDRC sites in participating in such projects is sought. Raj Kapur and Ms. Berrios responded that any HDRC member who has a project for which they are looking for collaborators may use the HDRC network to reach out to other

members for interest in participation. If the proposed study would use data and/or samples collected for the HDRC biorepository, a request for access to the materials must be made through the Steering Committee. There is no formal process set up yet for making a request to access materials from the HDRC biorepository, but the Steering Committee will be developing a process.

- III. Grant resubmission – Ms. Berrios reported that the revised HDRC grant application was submitted to the NIDDK in July 2015. This is a revision the application submitted in October 2014 that was not funded. The primary authors of the grants had concerns that the initial grant did not receive an adequate review and that the study section, KNOD (Kidney, Nutrition, Obesity and Diabetes) to which it was assigned did not have the appropriate expertise. After discussions with the Program Officer they were advised to request review by the CIMG (Clinical, Integrative, and Molecular Gastroenterology) study section in the cover letter, which was done. However, the application was again assigned to KNOD. We contacted the Program Officer for advice. KNOD has added some geneticists in this cycle that should help with that aspect of the review and the Program Officer and colleagues will try to suggest a reviewer with expertise in Hirschsprung disease. We hope this will lead to adequate review and we will let the group know when we hear about the review.

Q. Dr. Rollins asked when we would know whether or not the grant is funded. Ms. Berrios responded that it is scheduled for review in October, so we will likely know the score and percentage by late October or early November. While that may give a sense of the likelihood of funding, we won't know for certain whether or not it is funded until early next year. Ankush Gosain added that the funding decision also depends upon congressional budgeting of funds to the NIH.

- IV. Advisory Board – Ms. Berrios reported that the Steering Committee has revisited the need for an HDRC Advisory Board for which we obtained suggestions of potential members in the past. Steering Committee members are in the process of reaching out to a potential leader who will then have a role in selecting other Advisory Board members. We will update the group with Advisory Board members once it is formed.
- V. Sharing Contact Information – Ms. Berrios thanked everyone who responded to the survey recently sent out about sharing contact their contact information with other HDRC members and outside the HDRC. Most were open to sharing their contact information in some form and the Coordinating Center will follow those wishes. Regarding the earlier question, using this mechanism, we can facilitate contacts between HDRC members for those who wish to find collaborators for projects.

The next quarterly call will be held on December 1st at 2:00pm, ET.