Why is this study being done?

Previous research has shown that Hirschsprung disease occurs when one or more genes involved in the formation and migration of nerve cells in the gut are altered. While we, and others, have identified several Hirschsprung disease genes, we have much more work to do in clarifying how genetic variants lead to disease and may affect the course and treatment of disease. In addition, the data and samples collected will build a valuable resource for use in the HDRC’s future research about Hirschsprung disease.

What if I have questions or concerns?

Many people have additional questions that they want to discuss with the study team before deciding whether or not to enroll in the study. The study team wants to be sure you have all the information you need before you decide whether to participate or not.

Our study coordinator would be happy to speak with you to answer any questions you may have. Courtney Berrios can be reached at the phone number or email address below.

Hirschsprung Disease Research Collaborative

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Principal Investigator: Aravinda Chakravarti
What is the Hirschsprung Disease Research Collaborative?

The Hirschsprung Disease Research Collaborative (HDRC) is a research study that collects samples and detailed medical information from people with Hirschsprung disease and their family members. The samples and information collected are used to study the genetics of Hirschsprung disease and are stored for future use in studies of various aspects of Hirschsprung disease. Our expectation is that the study will lead to better understanding of the genetics of Hirschsprung disease and, further down the road, improved diagnosis, treatment, and genetic counseling.

Who is doing the study?

This study is being led by Dr. Aravinda Chakravarti and members of his laboratory at Johns Hopkins University. However, the HDRC is a multi-site study, meaning that researchers at multiple hospitals and universities are involved in collecting samples and may use the data for research about Hirschsprung disease. Data is only shared between sites after identifying information has been removed.

What will I be asked to do if I choose to participate in the study?

• Complete a medical/family history questionnaire;
• Submit blood samples from the individual(s) with Hirschsprung disease and, if available, his/her parents;
• Provide access to medical records for the individual(s) with Hirschsprung disease.

How are samples and information used?

We extract DNA from the blood samples and use a variety of methods to study an individual’s DNA sequence. The information from the questionnaire and medical records is used to help us interpret the meaning of any genetic variation we find in the sample.

How do I enroll?

If you are interested in participating in the study, please contact Courtney Berrios, the study coordinator, by phone at 410-502-7541 or by email at hirschsprung@jhmi.edu. She will talk with you about the study in detail and answer any questions that you may have. If you decide to participate, a study kit can then be sent to you that contains all the materials necessary for participation. All activities for study participation can be completed local to you. Study paperwork and samples can be shipped to the study center at no cost to you.