Study Survey

Below is a composite of the study survey. Variations in wording based on whether the participant was an affected adult, parent of an affected child, or affected adult who is also the parent of an affected child are indicated. Where the text indicates “your/your child’s”, “my/my child’s”, or similar statements the survey showed the appropriate possessive based on whether the participant was being asked about their own experience as an affected adult or as a parent to an affected child. Italicized items did not appear to participants in the study, but represent ways in which skip and display logic were used to display the survey contents to participants. “Next” represents page breaks.

Views and Experiences with Hirschsprung Disease

Who is doing this research study? This study is being carried out by researchers at the Johns Hopkins School of Medicine.

Who may participate in the study? If you have Hirschsprung disease (HD), have a child with HD, or both then you are invited to participate in the study.

What is this research study about? We are interested in your views about HD and how it may affect your life. We also want to know how these may be the same or different between parents of children with HD and adults who have HD themselves. The study also asks questions about your interest in genetic counseling or testing for HD.

What is involved in participating? If you choose to participate in the study we will ask you to complete a survey that will take approximately 30 minutes.

What are the benefits? There is no direct benefit to you for participating. The results may help the medical community to understand how HD affects individuals and families. We hope it will help medical providers to better address the needs of families and individuals with Hirschsprung disease.

What are the risks? There are no physical risks to the study. You may get bored or tired while answering the survey questions.

Do I have to participate in the study? No, your participation is completely voluntary.

Will I be paid? No, you will not be paid or given any gift for completing the survey.

Problems or Questions? This study has been approved by the Johns Hopkins Medical Institutions IRB as protocol # IRB00081079. If you have any problems or questions about this study, or about your rights as a participant, please contact Courtney Berrios from the study team by phone at 410-502-7541. If you have questions or concerns about Hirschsprung disease, please talk to your doctor, nurse, or other healthcare provider.

Your completion of this survey or questionnaire will serve as your consent to be in this research study. Please click next to complete the survey.

Please choose the most correct statement below.
I have Hirschsprung disease.
I have a child (or multiple children) who has Hirschsprung disease.
Both I and one or more of my children have Hirschsprung disease.
Neither me nor any of my children have Hirschsprung disease.

If one of top three…

How old are you?
Less than 18 years of age
18 years of age or older

Next

If not eligible by either question then will get message: "Thank you for your time, but you are not eligible for this study. Please close your browser window."
If eligible then get message: "You are eligible for this study. Please click the button with forward arrows below to continue on to rest of survey."

Next

Based on the participant's response to the first question, Survey Flow in Qualtrics was used to place the participant into a survey with questions and wording specific to their situation (adult with HD only, parent of a child with HD only, or adult with HD who also has a child with HD). If the survey was completed by mail only the version of the survey specific to their situation was mailed.

What is your age?
Text

What is your gender?
Male
Female
Other (please specify)_____

If the first question indiciates they have a child or children with HD…
What is the age or ages of your child/children with HD?
Text

If the first question indiciates they have a child or children with HD…
What is the gender(s) your child or children with HD?
Text

Do you have any other family members who have Hirschsprung disease?
Yes
No

Next

Participants who indicated that they have HD themselves and have a child with HD will see the text…
For the next few sets of questions we will ask you to answer the questions first while thinking about your Hirschsprung disease and second while thinking about your child's Hirschsprung disease. Because we will ask you to do this for some parts of the survey it may take you somewhat longer to take the survey than estimated.

Your Views of Your/Your Child's Hirschsprung Disease

Listed below are a number of symptoms that you/your child may or may not have experienced with your/your child's HD. Please indicate by circling Yes or No, whether you or your child have experienced any of these symptoms, and whether you believe that these symptoms are related to your/your child's HD.

Any participant who indicated that they have a child with HD will see the text…
If you have more than one child with HD, please answer in regards to the child you feel is most severely affected by HD.
For participants with children the questions with an asterisk next to them will be in two forms, first for consequences and control for the child and second for consequences and control for the parent.

Symptom	**I or my child have experienced this symptom**		**I believe this symptom is related to my my child's HD**	
Pain	Yes	No	Yes	
Constipation	Yes	No	Yes	
Nausea or Vomiting	Yes	No	Yes	
Weight Loss	Yes	No	Yes	
Fatigue	Yes	No	Yes	
Difficulty Feeding / Eating	Yes	No	Yes	
Upset Stomach	Yes	No	Yes	
Soiling	Yes	No	Yes	

We are interested in your own personal views of how you now see your/your child's Hirschsprung disease. Please indicate how much you agree or disagree with the following statements about your/your child's HD by ticking the appropriate box. Please respond based on how you CURRENTLY view your/your child's HD.

	Views about Hirschsprung disease	Strongly Disagree	Disagree	Neither Agree nor Disagree	Agree	Strongly Agree
IP1	My/my child's HD will last a short time.					
IP2	My/my child's HD is likely to be permanent rather than temporary.					
IP3	My/my child's HD will last for a long time.					
IP4	My/my child's HD will pass quickly.					
IP5	I expect I/my child have HD for the rest of our lives.					
IP6	My/my child's HD is a serious condition.					
IP7*	My/my child's HD has major consequences on my/their life.					
IP8*	My/my child's HD does not have much effect on my/their life.					
IP9*	My/my child's HD strongly affects the way others see me/them.					
IP10	My/my child's HD has serious financial consequences.					
IP11	My/my child's HD causes difficulties for those who are close to me/them.					
IP12*	There is a lot which I/my child can do to control my/their HD.					
IP13*	What I/my child do/does can determine whether my/my child's HD gets better or worse.					
IP14*	The course of my/my child's HD depends on me/them.					
IP15*	Nothing I/my child do/does will affect my child's HD.					
IP16*	I have the power to influence my/my child's HD.					
IP17*	My/My child's actions will have no affect on the outcome of my/my child's HD.					
IP18	My/my child's HD will improve in time.					
IP19	There is very little that can be done to improve my/my child's HD.					
IP20	My/My child's treatment will be effective in curing my/their HD.					
IP21	The negative effects of my/my child's HD can be prevented (avoided) by my/their treatment.					
IP22	My/My child's treatment can control my/their HD.					
IP23	There is nothing which can help my/my child's HD.					
IP24	The symptoms of my/my child's HD are puzzling to me.					
IP25	My/My child's HD is a mystery to me.					
IP26	I don't understand my/my child's HD.					
IP27	My/my child's HD doesn't make any sense to me.					

IP28	I have a clear picture or understanding of my/my child's HD.					
IP29	The symptoms of my/my child's HD change a great deal from day to day.					
IP30	My/my child's symptoms come and go in cycles.					
IP31	My/my child's HD is very unpredictable.					
IP32	I/My child go/goes through cycles in which my/their HD gets better and worse.					
IP33	I get depressed when I think about my/my child's HD.					
IP34	When I think about my/my child's HD I get upset.					
IP35	My/My child's HD makes me feel angry.					
IP36	My/My child's HD does not worry me.					
IP37	Having/My child having HD makes me feel anxious.					
IP38	My/My child's HD makes me feel afraid.					

Next

We are interested in what you consider may have been the cause of your/your child's HD. We are most interested in your own views about the factors that caused your/your child's HD rather than what others, including doctors or family, may have suggested to you. Please indicate how much you agree or disagree that each factor below was a cause for you/your child by ticking the appropriate box.

	Possible Causes	**Strongly Disagree**	**Disagree**	**Neither Agree nor Disagree**	**Agree**	**Strongly Agree**
C1	Stress or worry					
C2	Hereditary – it runs in my family					
C3	A germ or virus					
C4	Diet or eating habits					
C5	Chance or bad luck					
C6	Pollution in the environment					
C7	My own behavior					
C8	Overwork					
C9	Genetics					
C10	Alcohol					
C11	Smoking					
C12	Accident or injury					
C14	Altered immunity					

In the table below, please list in rank-order the three most important factors that you now believe caused YOUR/YOUR CHILD'S Hirschsprung disease. You may use any of the items from the box above, or you may have additional ideas of your own.

The most important causes for me/my child:-

1. _____

2. _____

3. _____

How severe do you consider Hirschsprung disease to be?
Not At All Severe Not Very Severe Somewhat Severe Very Severe

Next

In this section we are interested in how satisfied you are with the following aspects of YOUR life. ***If they have a child with Hirschsprung disease…*****Please answer from YOUR point of view, not your child's.**

<u>Part 1</u>. For each of the following, please choose the response that best describes how *<u>satisfied</u>* you are with that area of YOUR life. There are no right or wrong answers.

	HOW SATISFIED ARE YOU WITH:	**VERY DISSATISFIED**	**MODERATELY DISSATISFIED**	**SLIGHTLY DISSATISFIED**	**SLIGHTLY SATISFIED**	**MODERATELY SATISFIED**	**VERY SATISFIED**
1.	Your health?	1	2	3	4	5	6
2.	Your health care?	1	2	3	4	5	6
3.	The amount of pain that you have?	1	2	3	4	5	6
4.	The amount of energy you have for everyday activities?	1	2	3	4	5	6
5.	Your ability to take care of yourself without help?	1	2	3	4	5	6
6.	The amount of control you have over your life?	1	2	3	4	5	6
7.	Your chances of living as long as you would like?	1	2	3	4	5	6
8.	Your family's health?	1	2	3	4	5	6
9.	Your children?	1	2	3	4	5	6
10.	Your family's happiness?	1	2	3	4	5	6

11.	Your sex life?	1	2	3	4	5	6
12.	Your spouse, lover, or partner?	1	2	3	4	5	6
13.	Your friends?	1	2	3	4	5	6
14.	The emotional support you get from your family?	1	2	3	4	5	6
15.	The emotional support you get from people other than your family?	1	2	3	4	5	6
16.	Your ability to take care of family responsibilities?	1	2	3	4	5	6
17.	How useful you are to others?	1	2	3	4	5	6
18.	The amount of worries in your life?	1	2	3	4	5	6
19.	Your neighborhood?	1	2	3	4	5	6
20.	Your home, apartment, or place where you live?	1	2	3	4	5	6
21.	Your job (if employed)?	1	2	3	4	5	6
22.	Not having a job (if unemployed, retired, or disabled)?	1	2	3	4	5	6
23.	Your education?	1	2	3	4	5	6
24.	How well you can take care of your financial needs?	1	2	3	4	5	6
25.	The things you do for fun?	1	2	3	4	5	6
26.	Your chances for a happy future?	1	2	3	4	5	6
27.	Your peace of mind?	1	2	3	4	5	6
28.	Your faith in God or a higher power?	1	2	3	4	5	6
29.	Your achievement of personal goals?	1	2	3	4	5	6
30.	Your happiness in general?	1	2	3	4	5	6
31.	Your life in general?	1	2	3	4	5	6
32.	Your personal appearance?	1	2	3	4	5	6
33.	Yourself in general?	1	2	3	4	5	6

Next

PART 2. For each of the following, please choose the response that best describes how *<u>important</u>* that area of your life is to YOU. There are no right or wrong answers.

	HOW IMPORTANT TO YOU IS:	**VERY UMIMPORTANT**	**MODERATELY UMIMPORTANT**	**SLIGHTLY UNIMPORTANT**	**SLIGHTLY IMPORTANT**	**MODERATELY IMPORTANT**	**VERY IMPORTANT**
1.	Your health?	1	2	3	4	5	6
2.	Your health care?	1	2	3	4	5	6

3.	Having no pain?	1	2	3	4	5	6
4.	Having enough energy for everyday activities?	1	2	3	4	5	6
5.	Taking care of yourself without help?	1	2	3	4	5	6
6.	Having control over your life?	1	2	3	4	5	6
7.	Living as long as you would like?	1	2	3	4	5	6
8.	Your family's health?	1	2	3	4	5	6
9.	Your children?	1	2	3	4	5	6
10.	Your family's happiness?	1	2	3	4	5	6
11.	Your sex life?	1	2	3	4	5	6
12.	Your spouse, lover, or partner?	1	2	3	4	5	6
13.	Your friends?	1	2	3	4	5	6
14.	The emotional support you get from your family?	1	2	3	4	5	6
15.	The emotional support you get from people other than your family?	1	2	3	4	5	6
16.	Taking care of family responsibilities?	1	2	3	4	5	6
17.	Being useful to others?	1	2	3	4	5	6
18.	Having no worries?	1	2	3	4	5	6
19.	Your neighborhood?	1	2	3	4	5	6
20.	Your home, apartment, or place where you live?	1	2	3	4	5	6
21.	Your job (if employed)?	1	2	3	4	5	6
22.	Not having a job (if unemployed, retired, or disabled)?	1	2	3	4	5	6
23.	Your education?	1	2	3	4	5	6
24.	Being able to take care of your financial needs?	1	2	3	4	5	6
25.	Doing things for fun?	1	2	3	4	5	6
26.	Having a happy future?	1	2	3	4	5	6
27.	Peace of mind?	1	2	3	4	5	6
28.	Your faith in God or a higher power?	1	2	3	4	5	6
29.	Achieving your personal goals?	1	2	3	4	5	6
30.	Your happiness in general?	1	2	3	4	5	6
31.	Being satisfied with life?	1	2	3	4	5	6
32.	Your personal appearance?	1	2	3	4	5	6
33.	How important are you to yourself?	1	2	3	4	5	6

Next

Participants that indicated that they themselves have HD and the have a child with HD will see the following text…

For the next group of questions we will again ask you to answer the questions first while thinking about your HD and second while thinking about your child's HD.

Please choose the response for each item below that best fits how your/your child's HD may have impacted you.
Living with HD has….
OR
Being a caregiver of a child with HD has…
OR
Living with HD and being a caregiver of a child with HD has…

Any participant who indicated that they have a child with HD will see the text…
If you have more than one child with HD, please answer in regards to the child you feel is most severely affected by HD.

		Not At All	**A Little Bit**	**Some-what**	**Quite a Bit**	**Very Much**
1.	Helped me accept the way things work out	1	2	3	4	5
2.	Helped me learn to deal better with uncertainty	1	2	3	4	5
3.	Taught me how to adjust to things I cannot change	1	2	3	4	5
4.	Helped me take things as they come	1	2	3	4	5
5.	Helped me to look at things in a more positive way	1	2	3	4	5
6.	Helped me learn to handle difficult times	1	2	3	4	5
7.	Helped me become more comfortable with who I am	1	2	3	4	5
8.	Helped me become a stronger person	1	2	3	4	5
9.	Helped me feel better about my ability to handle problems	1	2	3	4	5
10.	Helped me become a better person	1	2	3	4	5
11.	Helped me know who I can count on in times of trouble	1	2	3	4	5
12.	Makes me more willing to help others	1	2	3	4	5
13.	Helped relationships become more meaningful	1	2	3	4	5
14.	Helped me become closer to people I care about	1	2	3	4	5
15.	Helped me become more aware of the love and support available from other people	1	2	3	4	5

16.	Helped me learn my life is more meaningful	1	2	3	4	5
17.	Given me a greater appreciation for life	1	2	3	4	5
18.	Helped me develop a deeper sense of purpose in life	1	2	3	4	5
19.	Helped me feel peaceful	1	2	3	4	5
20.	Helped me find strength in my faith or spiritual beliefs	1	2	3	4	5

Next

Please mark how frequently each of the items below has occurred to YOU. *If they have a child with Hirschsprung disease…* Please answer from your point of view, not your child's.

		Not at all or less than 1 day last week.	**One or two days last week.**	**Three or four days last week.**	**Five to seven days last week.**	**Nearly every day for two weeks.**
1.	My appetite was poor					
2.	I could not shake off the blues					
3.	I had trouble keeping my mind on what I was doing					
4.	I felt depressed					
5.	My sleep was restless					
6.	I felt sad					
7.	I could not get going					
8.	Nothing made me happy					
9.	I felt like a bad person					
10.	I lost interest in my usual activities					
11.	I slept more than usual					
12.	I felt like I was moving too slowly					
13.	I felt fidgety					
14.	I wished I were dead					
15.	I wanted to hurt myself					
16.	I was tired all the time					
17.	I did not like myself					
18.	I lost a lot of weight without trying to					
19.	I had a lot of trouble getting to sleep					
20.	I could not focus on the important things					

Next

Please respond below about your interest in each of the items below.

A genetic counseling appointment could provide you with information about the chances that a future child could have HD. If you are, or hypothetically were, pregnant or planning a pregnancy would you be interested in having a genetic counseling appointment?
Yes
No
Unsure

If a genetic variant that caused HD can be identified in a family then, in some cases, prenatal genetic testing can allow families to find out during a pregnancy additional information about how likely it is the baby will or will not be born with HD? If you are, or were, pregnant or planning a pregnancy would you be interested in prenatal genetic testing?
Yes
No
Unsure

Another technology, called Preimplantation Genetic Diagnosis, can allow some families who use InVitro Fertilization (IVF) and have a genetic variant known to cause HD to test the embryos created by IVF. The testing can show whether each embryo has inherited the genetic variant that causes HD before they decide which embryos to implant. If you are, or were, planning a pregnancy would you be interested in using Preimplantation Genetic Diagnosis?
Yes
No
Unsure

Next

Please share your thoughts about the questions below.
For parents only:
What was your greatest need when your child was diagnosed with HD?

For both parents and affected adults:
What is your current greatest need in relation to HD?

What is the greatest challenge you have faced having/having a child with HD?

What benefits have you found from having/having a child with HD?

Next

Participants that indicated that they themselves have HD and they have a child with HD will see the following text…

One last time, for the next set of questions we will again ask you to answer the questions first while thinking about your HD and second while thinking about your child's HD.

YOUR/YOUR CHILD'S Bowel Function

For affected adults…Please choose the response for each question below that best describes your current bowel function.
For those with an affected child…If the child with HD you have answered questions about above is 4 YEARS OF AGE OR OLDER, please choose the responses below that best fit YOUR CHILD'S current bowel function. If this child is younger than 4 years old, please skip this section.

<u>Ability to hold back defecation</u>
Always
Problems less than 1/week
Weekly problems
No voluntary control

<u>Feels/reports the urge to defecate</u>
Always
Most of the time
Uncertain
Absent

<u>Frequency of defecation</u>
Every other day to twice a day
More often than twice a day
Less often than every other day

<u>Soiling</u>
Never
Staining less than 1/week, no change of underwear required
Frequent staining, change of underwear often required
Daily soiling, requires protective aids

<u>Accidents</u>
Never
Fewer than 1/week
Weekly accidents; often requires protective aids
Daily, requires protective aids during day and night

Constipation
No constipation
Managed with diet
Manageable with laxatives
Manageable with enemas

Social problems
No social problems
Sometimes (foul odors)
Problems causing restrictions in social life
Severe social and/or psychological problems

Next

Below are a few final questions.

What is the highest level of education you have completed?
Less than high school
High school
Associates degree
Undergraduate degree
Graduate degree

What is your race?
American Indian / Alaska Native
Asian
Black / African American
Native Hawaiian / Other Pacific Islander
White
Unknown

Do you consider yourself to be Hispanic/Latino?
Yes
No

If you would like to provide any additional comments to the researchers, please use the space below to do so.
Text box

Finish

Thank you for taking the time to complete this survey. We truly appreciate the effort you made to share your views and experiences about Hirschsprung disease. We hope the results will help medical providers to better address the needs of individuals with HD.