Study Questionnaire

This questionnaire is designed to obtain information about the patient with Hirschsprung disease (HSCR). If you are not the patient please give all the responses on their behalf. We are interested in information on blood relatives only. **If you are unable to answer a question, please leave it unanswered and complete the others.** The study coordinator may contact you at a later date to clarify answers. *Thank you very much for your help and participation!*

A. PATIENT INFORMATION:

Patient's Name:_____

Please indicate the person completing this form (circle one):

Patient Mother of patient Father of patient Medical professional

Other_____

Patient's date of birth: \ \ (mm\dd\yyyy)

Patient's gender (circle one) Male Female

Ethnicity: Hispanic/Latino? ____ yes ____ no

Race (circle all that apply):
American Indian/Alaska Native Asian Black/African American

Native Hawaiian/Other Pacific Islander White Unknown

B. CONTACT INFORMATION:

Name of parent(s) or guardian(s) if patient is a minor or an adult with a Legally Authorized Representative:_____

Patient's Current Address: _____

_____

Telephone Number: (home)_____(mobile)_____

(work)_____who works at this number_____

Email address:_____

C. PATIENT'S HIRSCHSPRUNG DISEASE DIAGNOSIS

1. Age of patient when Hirschsprung disease was diagnosed:

2. What were the major symptoms at the time of diagnosis (newborn bowel blockage, distension and difficulty feeding, chronic constipation, enterocolitis (diarrhea, fever, infection in the bowel, other)?

3. How was the diagnosis made? (rectal biopsy, barium enema, other?)

4. Where was the transition zone (area where diseased colon ended and healthy colon with nerve cells began)? Was it in the rectum, rectosigmoid region, sigmoid colon, descending colon, transverse colon, ascending colon, distal small bowel, proximal small bowel?

5. How many inches or centimeters of the colon were affected?

6. Has the patient ever had episodes in which 5 or more of the following symptoms were present?

Diarrhea with explosive stool
Diarrhea with foul smelling stool
Diarrhea with bloody stool
Explosive discharge of gas and stool when the doctor did a rectal exam
Distended abdomen
Patient was very pale
Patient was very tired, lethargic or listless
Fever

If yes,

How many episodes occurred before any surgery for Hirschsprung disease?_____

How many occurred after one or more surgeries for Hirschsprung disease?_____

7. Has the patient ever been admitted to the hospital with severely deteriorated general condition, fever, or low blood pressure when they had diarrhea and abdominal distention?
____ yes ____ no

If yes,

How many times did this occur before any surgery for Hirschsprung disease?_____

How many times did this occur after one or more surgeries for Hirschsprung disease?___

8. Has the patient ever been told by a doctor that they had any kind of enterocolitis?
____ yes ____ no

If the patient has not yet had surgery, please skip to section D. *Below are questions about possible complications occurring after surgery. Many of these are uncommon, but it is helpful to our research to know about any complications. Some questions may not apply if the patient did not have a pull-through. Please answer any that apply to the patient.*

9. What operation did the patient have (Swenson, Transanal Swenson-type, Soave, Transanal Soave-type, Duhamel, Martin, Kimura, or other)?
Was the surgery done by:
Laparotomy (large abdominal incision)? _____yes _____no
Laparoscopy (small, keyhole incisions)? _____yes _____no

10. Did the patient have a colostomy or ileostomy? ____ yes ____ no

If yes, do they still have it_____ was it before a pull-through operation_____

11. What was the patient's age at the time of surgery/surgeries?

12. Has the patient had incontinence or difficulty controlling stools after surgery?
____ yes ____ no

If yes, How many times per week did this occur?_____

Has it improved _____ stayed the same _____ or resolved_____over time?

13. Has the patient had significant constipation requiring laxatives or enemas after surgery?
____ yes ____ no

If yes, Has it improved _____ stayed the same _____or resolved_____over time?

14. Has the patient required a repeat pull-through operation?
____ yes ____ no

If yes, What was the reason for it? A narrowing of the pull-through_____ Still had no ganglion (nerve) cells_____ A technical problem with the first operation_____

If yes, What type of surgery was the repeat pull-through (Swenson, Transanal Swenson-type, Soave, Transanal Soave-type, Duhamel, Martin, Kimura, or other)?
Was the surgery done by:
Laparotomy (large abdominal incision)? _____yes _____no
Laparoscopy (small, keyhole incisions)? _____yes _____no

15. Has the patient ever had a myectomy (cutting of the sphincter muscle)? ____ yes ____ no

If yes, Did it improve the patient's symptoms?

16. Has the patient ever had an injection of botox? ____ yes ____ no

If yes, Did it improve the patient's symptoms?

17. Did the patient have problems with a severe rash on the skin around the anus after the surgery? ____ yes ____ no

18. Did the patient have any other complications during or after the pull-through surgery? ____ yes ____ no

If yes,

Bleeding requiring a transfusion? ____ yes ____ no

Bowel blockage or obstruction? ____ yes ____ no

Hernia in the incision? ____ yes ____ no

Infection in the incision? ____ yes ____ no

Infection in the abdominal cavity? ____ yes ____ no

Leakage from a suture line or staple line? ____ yes ____ no

Scarring or stricture of a suture line or staple line resulting in a blockage? ____ yes ____ no

Other?_____

Survey continues on next page…

D. PATIENT'S CHILDREN

***Complete this section ONLY if the patient has children**

Does the individual with Hirschsprung disease (the patient) have any children? yes no

(If yes, please complete the following chart. DOB – date of birth in mm/dd/yyyy; Living? – answer yes or no, if deceased, please indicate age and cause of death; HSCR – Hirschsprung disease, answer yes or no; CSC – chronic severe constipation, answer yes or no and if yes, indicate bowel movements per week)

Name	Sex	DOB	Living?	HSCR	CSC

Do all of the children listed above share the same mother and father? yes no
(If not, please indicate if they are related through the mother or the father.
_____)

Please answer the following questions on behalf of the above listed children's biological parent who is **not** the patient with Hirschsprung disease on whose behalf the questionnaire is being completed.

Parent's name_____

Parent's date of birth_____

Does the parent have Hirschsprung disease or chronic constipation? yes no
(If yes, please explain_____
_____)

E. PATIENT'S PARENTS:

Please complete the chart below about the patient's parents.

(DOB – date of birth in mm/dd/yyyy; Living? – answer yes or no, if deceased, please indicate age and cause of death; HSCR – Hirschsprung disease, answer yes or no; CSC – chronic severe constipation, answer yes or no and if yes, indicate bowel movements per week)

Name	DOB	Living?	HSCR	CSC
Father				
Mother				

F. PATIENT'S BROTHERS OR SISTERS

Does the patient have brothers or sisters? yes no

(If yes, please complete the following chart. DOB – date of birth in mm/dd/yyyy; Living? – answer yes or no, if deceased, please indicate age and cause of death; HSCR – Hirschsprung disease, answer yes or no; CSC – chronic severe constipation, answer yes or no and if yes, indicate bowel movements per week)

Name	Sex	DOB	Living?	HSCR	CSC

Do all of the above individuals share the same mother and father? yes no
(If not, please indicate in the margin for each if they share a mother, father or both with the patient.)

G. PATIENT'S GRANDPARENTS:
Please fill in as much information as you know.

FATHER'S SIDE:

(DOB – date of birth in mm/dd/yyyy; Living? – answer yes or no, if deceased, please indicate age and cause of death; HSCR – Hirschsprung disease, answer yes or no; CSC – chronic severe constipation, answer yes or no and if yes, indicate bowel movements per week)

Name	DOB	Living?	HSCR	CSC	Ancestry
Grandfather					
Grandmother					

MOTHER'S SIDE:

(DOB – date of birth in mm/dd/yyyy; Living? – answer yes or no, if deceased, please indicate age and cause of death; HSCR – Hirschsprung disease, answer yes or no; CSC – chronic severe constipation, answer yes or no and if yes, indicate bowel movements per week)

Name	DOB	Living?	HSCR	CSC	Ancestry
Grandfather					
Grandmother					

H. PATIENT'S AUNTS AND UNCLES:
Please fill in as much information as you know.

Please list all the brothers/sisters of the **patient's father**. (DOB – date of birth in mm/dd/yyyy; Living? – answer yes or no, if deceased, please indicate age and cause of death; HSCR – Hirschsprung disease, answer yes or no; CSC – chronic severe constipation, answer yes or no and if yes, indicate bowel movements per week)

Name	Sex	DOB	Living?	HSCR	CSC

Please list all the brothers/sisters of the **patient's mother:**

Name	Sex	DOB	Living?	HSCR	CSC

Is there any additional family history that you feel may be helpful to us?

_____
_____
_____
_____
_____
_____

I. PATIENT'S MEDICAL INFORMATION:

Please check a box to indicate if the patient has had any of the below listed medical evaluations. If yes, please note any known results.

Evaluation	Has Patient Had?	Results
Examination by Clinical Geneticist	☐ Yes ☐ No ☐ Unknown	
Examination by Neurologist	☐ Yes ☐ No ☐ Unknown	
Examination by Ophthalmologist (eye doctor)	☐ Yes ☐ No ☐ Unknown	
Chromosome Analysis or Genetic Testing	☐ Yes ☐ No ☐ Unknown	
Echocardiogram (ultrasound of the heart)	☐ Yes ☐ No ☐ Unknown	
Ultrasound of kidneys or other organs	☐ Yes ☐ No ☐ Unknown	
IQ or Developmental Evaluation	☐ Yes ☐ No ☐ Unknown	
CT scan or MRI of the brain	☐ Yes ☐ No ☐ Unknown	
Hearing Evaluation	☐ Yes ☐ No ☐ Unknown	
Other – please explain		

J. MEDICAL FAMILY HISTORY INFORMATION:

Please indicate (check box) if the **patient and/ or any blood relatives** [this includes brothers, sisters (full or half), parents, children, grandparents, uncles, aunts, nephews, cousins, etc.] is known to have any of the following diagnoses or conditions. If a relative is affected, please indicate how the individual(s) is (are) related to the patient in the space provided. Please give additional information about the diagnosis if requested in the box.

PAT= patient, REL= relative

Hirschsprung disease	☐ PAT	☐ REL _____
Chronic constipation	☐ PAT	☐ REL _____
Other intestinal abnormalities (malrotation, hypoganglionosis, IND etc.) Please describe type:	☐ PAT	☐ REL _____
Mental retardation	☐ PAT	☐ REL _____
Learning disability	☐ PAT	☐ REL _____
Down syndrome	☐ PAT	☐ REL _____
Other chromosome abnormality or genetic disorder Please describe:	☐ PAT	☐ REL _____
Growth retardation (short stature)	☐ PAT	☐ REL _____
Epilepsy (persistent seizures)	☐ PAT	☐ REL _____
Heart defect Please indicate type:	☐ PAT	☐ REL _____
Microcephaly (small head)	☐ PAT	☐ REL _____
Agenesis of corpus callosum (a type of brain anomaly)	☐ PAT	☐ REL _____
Color differences in eyes/skin/hair (light or dark spots on skin, white piece of hair, different colored eyes) Please describe:	☐ PAT	☐ REL _____
Cleft lip and/or palate	☐ PAT	☐ REL _____
Ptosis (drooping eyelids)	☐ PAT	☐ REL _____
Skeletal abnormalities (problems with the bones) Please describe:	☐ PAT	☐ REL _____

Kidney problems Please describe:	☐ PAT	☐ REL _____
Esophageal atresia (blockage of tube carrying food to stomach)	☐ PAT	☐ REL _____
Pyloric stenosis (blockage of outlet from stomach to intestines)	☐ PAT	☐ REL _____
Abnormality of internal or external genitals (sex organs) (including undescending testes) Please describe:	☐ PAT	☐ REL _____
Imperforate anus (closed anus at birth)	☐ PAT	☐ REL _____
Club foot	☐ PAT	☐ REL _____
Polydactyly (extra fingers or toes) Please describe:	☐ PAT	☐ REL _____
Syndactyly (fused/webbed fingers or toes) Please describe:	☐ PAT	☐ REL _____
Thyroid cancer Please indicate type (medullary, papillary, etc.):	☐ PAT	☐ REL _____
Pheochromocytoma (tumor of adrenal gland)	☐ PAT	☐ REL _____
Other tumors or cancers Please indicate type:	☐ PAT	☐ REL _____
Hypercalcemia (high blood calcium)	☐ PAT	☐ REL _____
Deafness / hearing loss Please give severity and age of onset:	☐ PAT	☐ REL _____
Waardenburg syndrome	☐ PAT	☐ REL _____
Congenital central hypoventilation/ unable to breathe when asleep / apnea	☐ PAT	☐ REL _____
Miscarriages or stillbirths Please indicate which and how many:	☐ PAT	☐ REL _____

Is there any additional information that you feel is relevant or may be helpful to the study? If you need additional space for any of your answers please feel free to write it here.

Signature of person completing form_____

Today's date: _____

Thank you very much for your help and participation.