TRANSITIONAL CARE IN PATIENTS WITH HIRSCHSPRUNG DISEASE: THOSE LEFT BEHIND

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Abstract

BACKGROUND:

The long-term effects of Hirschsprung disease are clinically variable. An improved understanding of challenges patients may face as adults can help inform transitional care management.

OBJECTIVE:

To explore the outcomes and transitional care experiences in adult patients with Hirschsprung.

DESIGN:

Cohort study.

SETTING:

Single center.

PATIENTS:

All patients treated for Hirschsprung between 1977 and 2001 (aged older than 18 years at the time of survey distribution in July 2018–2019). Eligible patients were sent validated multidomain surveys and qualitative questions regarding their transitional care.

MAIN OUTCOME MEASURES:

Status of transitional care, bowel function, and quality-of-life assessment. Qualitative analysis of transitional care experience.

RESULTS:

Of 139 patients, 20 had received transition care (10 had at least 1 visit but had been discharged and 10 were receiving ongoing follow-up). These patients had inferior bowel function and quality-of-life scores at follow-up. Twenty-three patients (17%) had issues with soiling at the time of discharge, and 7 patients received transitional care. Of these 23 patients, 9 (39%) had a normal Bowel Function Score (17 or more), 5 (22%) had a poor score (less than 12), and 1 had since had a stoma formation. Eighteen patients (13%) had active moderate–severe issues related to bowel function, only 5 had been transitioned, and just 2 remained under ongoing care. Importantly, when these patients were discharged from our pediatric center, at a median age of 14 (interquartile range, 12–16) years, 10 of 17 patients had no perceptible bowel issues, suggesting a worsening of function after discharge.

LIMITATIONS:

The retrospective design and reliance on clinical notes to gather information on discharge status as well as patient recall of events.

CONCLUSIONS:

There remains a small but significant proportion of Hirschsprung patients for whom bowel function either remains or becomes a major burden. These results support a need to better stratify patients requiring transitional care and ensure a clear route to care if their status changes after discharge.

Introduction

In recent years, the outcomes for patients with Hirschsprung disease (HSCR) have been increasingly studied; a significant minority of patients experience soiling and fecal incontinence with an associated reduction in quality of life (QoL).[1–4] In addition, there remains a risk of Hirschsprung-associated enterocolitis, with reported lifetime incidence ranging from 11% to 44%[5,6]; an emerging link with the development of IBD is now recognized.[7]

Studies during the initial childhood years after corrective pull-through surgery show that most children do not appear to demonstrate improvements in their functional status.[2,8] Adult patients have demonstrably better function, although 10% to 20% of patients have a poor outcome, with fecal incontinence, soiling, and poor psychosocial function compared to age-matched controls.[1,5,8–11] More recently, impacts in other functional domains have been noted, including urological and sexual function; fertility outcomes in female patients may also be impacted.[12–14]

Recognition of the negative impact on long-term health and well-being beyond childhood highlights the need for continued care in these patients and for transitioning pediatric patients with chronic health conditions into adult services.[15,16] Regrettably, clinical practice often falls short despite our understanding of the increased need for transition.[15,17–20] There is no standardized pathway for transitioning patients with congenital GI surgical pathology in the United Kingdom. Little is known about what happens to the patients who are not transitioned, and understanding the path patients take after discharge from pediatric services helps inform how transitional care should be managed.

This was a cross-sectional study of a single-center cohort with 2 aims. First, we performed an analysis of cross-sectional outcomes, comparing adult patients who received transitional care with those who did not. Second, we performed a thematic analysis of the written experiences of patients who were transitioned to gain insight into this process. We hypothesized that patients who have been offered transition care in the past might have worse functional and QoL outcomes, whereas those without may have improved bowel symptoms and psychosocial functioning.

Methods

This was a cross-sectional cohort study using mixed methods of all patients treated as children between 1977 and 2001 for HSCR at a single regional center. Historically, the department was one of the busiest in the United Kingdom, serving most of North London with a considerable catchment area extending to the north. There has also been a considerable number of referrals for revision surgery from across the country. Ethics approval was obtained from the National Health Service Research and Ethics Committee (No. 17/LO/1692). Patients were identified through clinical coding (International Classification of Diseases-9 751.3 and International Classification of Diseases-10 Q43.1), and case notes were reviewed to confirm a diagnosis. Patients with histologically confirmed HSCR under the care of the surgery team for any treatment were eligible for inclusion.

All patients, meeting these inclusion criteria and currently living within the United Kingdom were invited to take part in a comprehensive follow-up questionnaire (see Appendix 1 and Appendix 2), which was either emailed or mailed to them at home, and responses were collected from July 2018 to 2019. All included patients were adults at the time of survey distribution. The questionnaire included validated, multidomain surveys including the Bowel Function Score (BFS), Gastrointestinal Quality of Life Index (GIQLI), and Short Form 36 (SF-36).[12,21] Results from this cohort have been published previously.[3] The current study reports novel data relating to patients' experience of care after discharge from pediatric surgery and how the transition process may relate to their functional outcomes.

Recruitment was based on the Strengthening the Reporting of Observational Studies in Epidemiology statement. The lead investigator (J.R.D.) was not involved in the clinical care of any patients. Written consent was obtained from all patients.

Exclusion Criteria

Eligible adult patients were excluded from this particular analysis if they had died, moved abroad, or were unable to be contacted (no contact information or inappropriate to contact). Exclusions were also applied to those who were unable to complete the questionnaire for language reasons or had a learning disability related to a recognized syndrome or neurodevelopmental disability, which would preclude consent or completing the survey without an advocate.

Study Setting

Before 2001, there was no formal standardized transition practice at our center and patients (including the present study's patients who were treated between 1977 and 2001) would be referred to adult services at the pediatric surgeon's discretion. There were a total of 9 consultant pediatric surgeons caring for HSCR patients in the department during this period. For the purposes of this study, transitional care has been retrospectively defined as at least 1 appointment within the gastroenterology or colorectal surgery team immediately after discharge from pediatric surgery, which was arranged and coordinated at the discretion of the lead surgeon responsible for the patient's care at our institution.

Definitions

Based on individual recall and narrative, patients were defined as either having no transition service (NTS) or being referred to transition service (TS). TS patients were further divided into 3 categories: 1) single transition clinic appointment and discharged, 2) more than 1 visit but since discharged, or 3) ongoing care in adult services.

Details regarding the patients' functional status at discharge from pediatric services were retrieved retrospectively from clinical case notes at our institution, usually the final clinic letter. Binary outcomes were defined thus: social continence, need for rectal enemas, and requirement for a stoma or antegrade colonic enema (ACE). Functional status at the time of the study was assessed on the basis of the questionnaire responses.

Statistical Analysis (Quantitative)

Statistical analyses were performed using Prism version 9.0 (GraphPad) and SPSS version 26.0 (IBM) to delineate function at discharge compared to present day and to compare scores of both BFS and GIQLI between TS and NTS groups as well as the component scores of the SF-36—calculated by Z-score transformation from normal population data.[22,23] Data are presented as median (interquartile range [IQR]) or mean (SD). Distributions were compared with appropriate parametric and nonparametric tests, with Bonferroni correction for 2 comparisons when comparing component scores of the SF-36. A *p* value of <0.05 was considered statistically significant.

Qualitative Analysis

In addition to specific bowel function and QoL data, patients were asked open-ended questions relating to their transitional care and long-term follow-up (full questionnaires are included in Appendix 1 [male patients] and in Appendix 2 [female patients]). Qualitative analysis was performed on the basis of a grounded theory of data collection and evaluation.[24] Answers with insufficient content for analysis were excluded. Any responses to blank space questions that were answered with sufficient depth were reviewed using an inductive approach, independently reviewed by 2 authors with some experience in qualitative data handling (D.S.T. and J.R.D.; both male, pediatric surgical trainees), and categorized as positive, negative, or neutral. Discrepancies were blindly reviewed by a third independent reviewer with more extensive experience in qualitative research (K.F.; female, pediatric surgeon). Findings were reviewed by supervisors with qualitative research experience (J.C. and S.B.: male, pediatric surgeons; and S.E.: male, scientist). Further thematic analysis was performed to define common aspects of positive and negative experiences. Ultimately, themes were used to generate a proposed model of positive transition.

Results

After exclusions were applied, 340 patients were approached to take part in the survey, of whom 216 responded (64%); 139 of these patients were adults without a learning disability and therefore eligible for analysis (median age 29 [IQR, 25–34] years; see Fig. S1. Of these, 98 (71%) were men and the majority (104; 75%) had rectosigmoid disease (Table 1). Twenty patients were transitioned from the pediatric surgery clinic into adult services. Of those who were transitioned, only 10 were still receiving ongoing follow-up at the time of the survey. Patients reported seeing both gastroenterology (ie, medical GI) and general/colorectal surgery specialists, as well as 1 female patient who saw a pelvic floor specialist after childbirth.

Table 1. Demographics and outcomes

		Overall (n=139)	TS (n=20)	NTS (n=119)
	Sex M (%)	98 (71)	11 (55)	87 (73)
	Rectosigmoid (%)	104 (75)	12 (60)	92 (77)
	Re-do operation (%)	27 (19)	6 (30)	21 (18)
Last clinic information	Age at last clinic	14 [11-16]	16 [10-17]	14 [11-16]
	Stoma ACE Soiling Rectal enemas	3 3 24 3	1 0 7 1	2 3 17 2
	Redo Pullthrough	27	6	21
Information at Survey	Age at survey	29 [25-34]	27 [22-33]	29 [26-34]
	Current Stoma Current ACE Weekly/Daily Soiling Weekly/Daily Accid. Severe Psychological Issues	4 1 14 5 8	2 0 3 1 3	2 1 11 4 5

Data presented as n unless otherwise indicated. ACE = antegrade colonic enema (ie, by appendicostomy or caecostomy); IQR = interquartile range; NTS = no transition services; TS = transition services.

Status at discharge

Median age at discharge was similar for the TS and NTS groups (median 16 [IQR, 10–17] vs 14 [IQR, 11–16] years), and the median year of discharge was 2002 for both. At the time of discharge from the pediatric surgery department, 3 patients had a stoma (2%; 1 TS), 3 had an ACE (2%; no TS), and 3 were using rectal enemas (2%; 1 TS). Twenty-four patients (17%) had ongoing issues with soiling and social continence at the time of discharge, of whom 22 were managing their symptoms with oral medications or diet and 2 with rectal enemas. Only 7 of the 24 patients (30%) with soiling and social continence issues had received transitional care (Table 1).

Status at survey

The median age at the time of the survey was 29 years (NTS vs TS: 29 [25–34] years vs 26 [22–33.75] years, p = 0.13). One TS patient had a stoma formed as an adult. Overall, BFS was found to be higher in the NTS group compared to the TS group (17 [15–19] vs 15.5 [12.25–16], p = 0.0006; BFS scores were not applied to patients with ACE or stoma). Similarly, the GIQLI scores were higher in the NTS group (122 [108–131] vs 108 [86.5–118], p = 0.0004; Fig. 1). There was no difference in either physical (52.0 [47.1–54.8] vs 48.4 [36.0–53.1], p = 0.17) or mental (49.18 [37.6–56.5] vs 46.1 [34.9–55.4], p = 0.47) component scores (where 50 represents the mean of a matched healthy population) of the SF-36 surveys (Fig 1.).

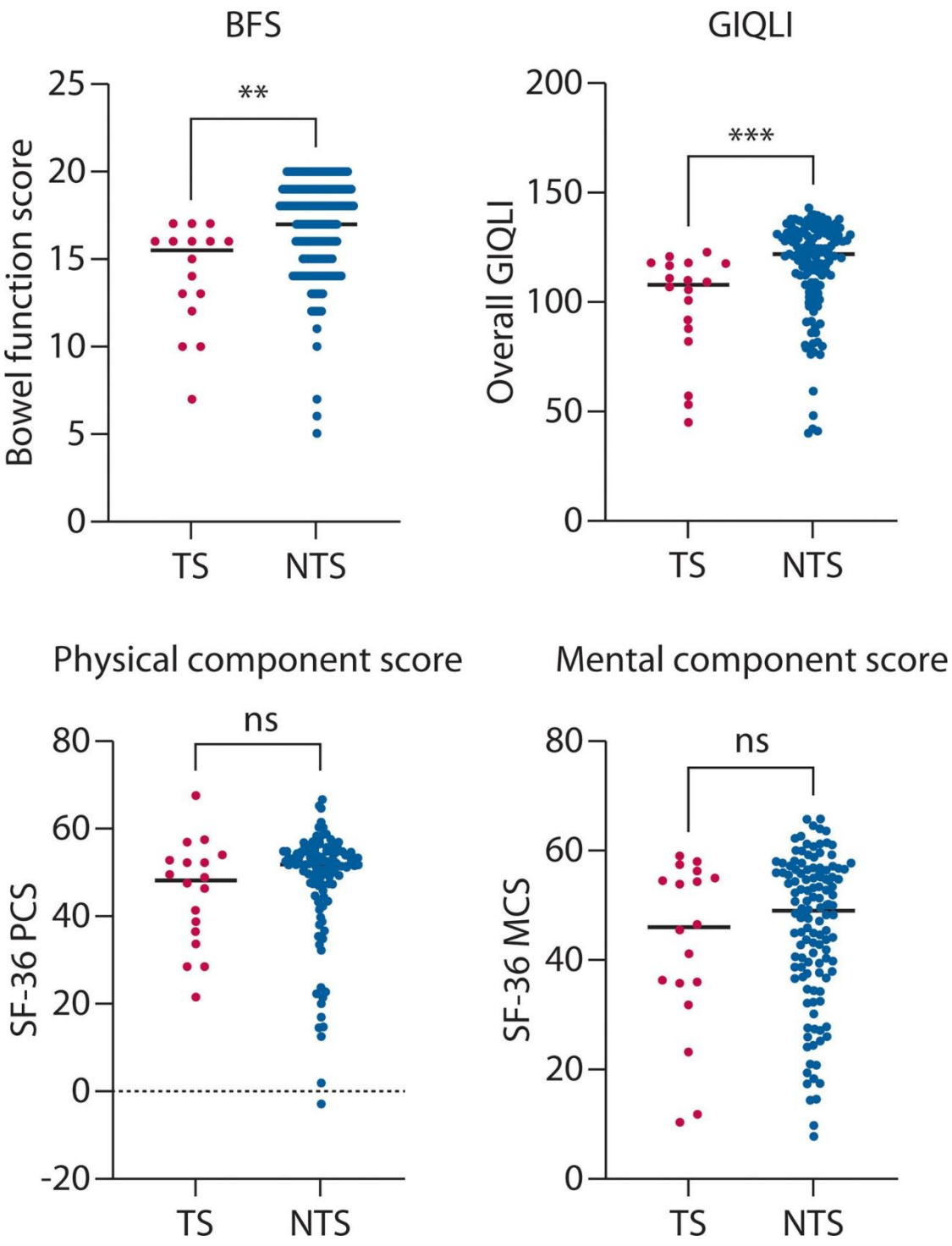

FIGURE 1.
Comparison of functional and quality-of-life outcomes. BFS = Bowel Function Score; GIQLI = Gastrointestinal Quality-of-Life Index; MCS = mental component score; ns = not significant; NTS = no transition services; PCS = physical component score; SF-36 = Short Form 36 (general quality of life questionnaire); TS = transition services. $^{**}p < 0.001$; $^{***}p < 0.0005$.

Of the 3 NTS patients with an ACE at discharge, 2 were later closed and 1 remained. An additional 3 patients had new ACEs formed but subsequently closed under the adult team. There were no ACE patients in the TS group. Three patients underwent major abdominal surgery in adulthood for adhesional obstruction and were all part of the NTS. One TS patient had a new stoma formed.

Of the 24 patients with soiling or social continence issues at discharge, 9 (38%) had BFSs defined as "normal" (BFS 17 or more) and 5 (21%) had poor bowel function (BFS less than 12 or active ACE or stoma). Of the 17 NTS patients with soiling issues at discharge, 5 (30%) reported ongoing soiling issues and 12 (71%) improved. However, 6 NTS patients who were previously well reported new soiling issues emerging in adulthood.

Daily or weekly soiling issues were actively experienced by 14 patients: 3 TS (15%) and 11 NTS (9%). A total of 8 patients reported severe psychological issues related to bowel function: 3 TS (15%) and 5 NTS (4%). Overall, 18 patients (12.9%) had frequent soiling or severe psychological issues related to bowel function. Five of these 18 patients had been transitioned, and only 2 remained under ongoing care. Importantly, when these patients with ongoing issues were last seen in the pediatric clinic, 10 of 18 (55.6%) reported no continence or bowel function issues, suggesting a deterioration after discharge.

Qualitative data

Qualitative data were extracted from the blank space questions from all transitioned patients with 100% sufficient for content analysis (20/20). When commenting on the transition process, 7 patients (35%) reflected positively. Seven patients had negative reflections (35%) and 6 responses were deemed neutral (30%).

Quotations from which categories and themes were extracted are shown in Table 2. Data saturation was reached for some but not all of the categories, specifically "challenges of a new environment," "loss of an important relationship," and "living with a chronic condition." Categories could be grouped into themes: "prior planning," "preparedness," and "new life beyond pediatric care"—with some categories such as "health literacy" cutting across these.

TABLE 2. - Extraction of themes and thematic analysis

Meaning unit	***Category***	***Theme***
"It was very smooth" "Well-orchestrated and carefully managed" "Smooth and straightforward, there wasn't long waiting times" "Transition was seamless" "I haven't had any consistent care"	Structure and organization	Prior planning
"Did not feel rushed" "Slowly introduce myself to the new team" "At no point did I feel like I was being 'kicked out' "	Slow and deliberate process	Prior planning
"It was discussed before what would happen" "It was hard to leave but they assured me that	Active communication before transitioning	Patient preparedness

Meaning unit	*Category*	*Theme*
everything will carry on… this made the transition a lot more comfortable”		
“I felt very prepared” “I have more control over my health” “I do feel I’m on my own with the disease with no real knowledge of how things have moved on”	Patient health literacy + empowerment	Patient preparedness
“Once transferred, they were fully aware of my medical history and it was just like picking up where I left off” “I had an initial appointment with the consultant just to clarify all the information provided was correct and up to date” “My GP was not really sure what was involved. I always had to recap” “They had a very fixed idea of how things would improve, however this did not help” “I found the experiences [unencouraging] and stressful”	Receiving clinicians prepared	Health services preparedness
“I was referred to a consultant. In all honesty, he had no knowledge of Hirschsprung” “Didn’t feel they understood my problems”	Adult clinicians with expertise	Health services preparedness
“I found it hard to recover in adult wards…. I was only 18 [years old]”	Challenges of a new environment	Adjusting to adulthood
“I felt sad to leave GOSH and my current surgeon as I felt I could trust him and he knows me so well” “Sure you can pass on notes but it’s not the same as long-standing history”	Loss of important relationship	Adjusting to adulthood
“I wanted to try and live with my condition and put it all behind me” “I had lots of investigations and then decided that was about as far as I could go”	Living with a chronic condition	Adjusting to adulthood

Comments from positive responses in green, negative in red, and neutral in dark gray.
GOSH = Great Ormond Street Hospital; GP = general practitioner.

Prior Planning

The process of transition was a frequent theme. When this was felt to be structured, this was associated with positive experiences: “it was very smooth,” “well-orchestrated and carefully managed,” and “the transition was seamless.” However, negative experiences included a lack of “consistent care.”

A slow speed of the process was also associated with positive experiences, with patients reporting they “did not feel rushed” and “at no point feeling they were being ‘kicked out.'”

Patient and Health Services Preparedness

Preparedness and understanding appeared most frequently as a theme across all answers and related to both expertise and understanding in the patient as well as in their treating clinical teams.

Patient preparedness for the transition process came in the form of active and consistent communication and assurances from their pediatric team and empowerment from the perspective of patient education and enabling access to patient records (ie, through an online patient portal). Experiences with Health Services were positive when information and understanding was actively sought: “they were fully aware of my medical history and it was just like picking up where I left off,” “I had a meeting with the consultant to clarify all the information was correct and up to date.” There were examples of poor preparedness among Health Services, with a lack of disease-specific knowledge and understanding being a prominent feature of several responses, as well as “fixed ideas” of treatment that might have been performed from other conditions.

Adjusting to Adulthood

This theme was undersaturated and tended to feature only negative and neutral experiences. One patient commented on the difficulties of recovering in an adult ward: “I found it hard to recover in adult wards ... I was only 18 [years old].” The loss of a trusted clinician and the difficulties of meeting a new team was also a strong theme: “After I received the most incredible care… my transition was never going to be easy.” “I felt I could trust him [pediatric surgery consultant] and he knows me so well.” Two patients described their fatigue with health care management after transition: “I wanted to live with my condition and put it behind me” and “that was about as far as I could go.”

Discussion

n the present article, we describe patients' transitional care experiences and outcomes during a 35-year period for our institution. At the point of discharge, 17% (n = 24) of our patients had issues with soiling and continence, but at the point of survey, 39% (n = 9) of these patients now had normal BFS. Indicating a continued functional improvement through adulthood. At the point of the survey, 13% (n = 18) had frequent soiling or severe psychological issues related to bowel function. Twenty patients received some form of transitional care; these patients had poorer BFS and GIQLI scores than nontransitioned patients in the survey. Several patients had not received transitional care and had clearly developed new functional issues since their discharge. Qualitative analysis of patients’ transitional pathway experiences revealed prominent themes of planning the process and preparing both the patient and their receiving team. The theme of adjusting to life outside of pediatric services was also noted.

Patients who received transitional care appointments tended to present with lower functional and QoL scores at follow-up. This suggests that the more symptomatic individuals were offered transitional care services, and these more complex patients continued to experience functional issues into adulthood, with poorer long-term outcomes and a higher symptom burden. However, we did identify that some adolescent patients with social continence issues were not offered transitional care but may have benefited from it. Furthermore, the clinical status of patients with HSCR may change after discharge: from the 102 NTS without soiling issues, 6% reported new issues in the survey. Published data from

our own cohort and others suggest that adults tend to have better outcomes than children after surgery, indicating a general improvement in function. However, longitudinal assessments are rarely reported.8,25

Prominent themes that emerged from qualitative analysis surrounded the planning and preparation of the transitional process, which has been echoed by patients elsewhere with congenital cardiac disease.26 Patients also highlighted a lack of experience with Hirschsprung disease among adult services. Adult surgeons or GI specialists with a dedicated interest in these patients would certainly be of great value, although delivering this may be challenging considering the move from centralized pediatric surgery services to more dispersed adult general surgical care. This could drive adult care of congenital GI conditions toward more centralized specialist services, as is seen with congenital cardiology in the United Kingdom (where surgeons and cardiologists continue to see the same patients throughout life). Structured patient and parent education during adolescence and instilling personal ownership of their condition could prevent patients from feeling isolated and empower them to seek help or referral where needed.

Combining objective outcomes and patient-reported experience adds insight into the patient journey. Modern services advocate a more active transition process for lifelong conditions,15,27 and this has been well demonstrated in other pediatric disciplines such as the treatment of IBD,28 congenital cardiac disease,29 and rheumatological conditions.30 Historically, consensus differed, and this is reflected in our cohort, where just 14% of patients were transitioned at the lead surgeon's discretion. The timing of transition has been highly debated and is likely to vary depending on geographical region and local services. In the United Kingdom, it has been suggested that the transition process should begin around the age of 14 years, with no definitive end point.31 Many pediatric centers have the ability to continue to see select patients until the age of 25 years, allowing for flexibility within the system.

Transition is an ever-evolving area of health care, and our findings highlight the need to reconsider how we handle these patients. We suggest a formal criterion-based assessment of patients during a transition period that will vary depending on clinical status and suggest a categorized transition accordingly:

Those with high symptom burden (ie, ongoing soiling, use of long-term stoma, or antegrade or retrograde enemas) are referred for routine ongoing follow-up to an adult specialist service, ideally one with an interest in and exposure to this patient population, with the aim of long-term follow-up. This may include a period of combined pediatric and adult cover or a specific adolescent clinic with both adult and pediatric surgeon involvement. The final adult team may be a gastroenterologist or colorectal surgeon, but the important factor is clinical expertise and exposure to this specific patient population.
Those with moderate symptom burden are referred for a single follow-up with the most appropriate regional adult care team as they approach the upper limit of pediatric age range of 16 to 18 years in the United Kingdom.
Those with the least symptom burden are referred to the care of their general practitioner, with sign-posting to relevant adult services provided to the patient and general practitioner and a care summary document provided to both.

We suggest that multidomain, objective screening tools such as BFS and GIQLI should be used to assess both functional status and QoL and to identify those who require referral to adult services. This would formalize a full and systematic review at the initiation of transition and potentially identify individuals with subtle functional concerns. It would also provide a touchstone for referring and accepting clinical teams. Because of the part-retrospective and cross-sectional nature of our review, we are unable to recommend a specific cutoff for the instruments used, but this would be interesting for further work.

It is important to reemphasize that a significant minority of patients in group 3, who are well at discharge, will deteriorate and require referral to adult services later in life. Increasing age has previously been identified as an independent risk factor for poor bowel function in HSCR patients in the only study to report objective outcomes of patients into their fifth and sixth decade of life.4 It is essential that all HSCR patients are made aware of the potential for deterioration that may be directly attributable to their condition and that they are empowered to seek referral if needed in the future. Education within the patient community has been the focus of some work by the UK–based Tracheoesophageal Fistula Support Charity (TOFS), a patient-directed charity that publishes patient information aimed at enabling adult patients with tracheoesophageal fistula and VACTRL association to recognize new symptoms as part of their condition with the aim to "encourage self-advocacy, promote wellness, and improve health outcomes."32 Similarly empowered and educated patients with HSCR would be better positioned to recognize and flag changes in their health related to HSCR and seek medical help. This may require input from the pediatric team long after discharge to ensure the appropriate transfer of relevant information to the treating adult specialist. This highlights the ongoing duty of pediatric surgeons to care for their patients, even after discharge.

Limitations of this study include its retrospective nature and reliance on clinic notes to gather information on discharge status, which is open to omission and reporting bias. Furthermore, patient recall was relied on for information about events surrounding their care at discharge or transition, which can be unreliable. Although our survey gave the opportunity for patients to express their opinions and experience of care after discharge from the pediatric service, this lacked a formal qualitative interview. More information would be gleaned from formal interviews and focus groups.15

CONCLUSION

This study provides an opportunity for a longitudinal assessment of patients with HSCR disease and a review of their transitional care. A small but significant number of patients were discharged with active soiling issues, and although some patients will improve after discharge, some will inevitably deteriorate. We identified that bowel function can be a major burden affecting the QoL of patients well beyond their time under pediatric services. This highlights the need for formal and more widespread transitional care.

Although this study focuses on Hirschsprung disease, it suggests that many patients with congenital anomalies require lifelong support. It is therefore important to establish a clear transition protocol, to identify which patients will benefit, and to safety-net others to ensure a clear route to care if their status changes after discharge. This will require strengthening links to adult specialties to provide care for these complex patient cohorts.

We believe that transitional care is a process and not a single moment in time. As pediatric surgeons, our duty of care does not end when the patient becomes an adult, and maintaining clear communication between adult and pediatric services is essential in the ongoing care of these patients.

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Long-Term Outcomes in Hirschsprung's Disease (GOSH)
Questionnaire for adult patients
V1.1 18/09/2017

Open questionnaire

Could you tell us about your transition from care at Great Ormond Street to adult services?

Where do you currently see a specialist for follow up of your Hirschsprung's Disease?

Have you had any further operations since you transitioned into adult care, including a stoma formation or Botox injections?

Do you currently have a stoma (colostomy or ileostomy)?

Yes ☐

No ☐

Do you currently take any medication for constipation, such as Movicol or Lactulose?

Do you take any other medication for any other reason?

Do you have any food allergies or intolerances?

What is your current height and weight?

Height? cm Weight? kg

Do you know of any family members who have Hirschsprung's Disease?

Long-Term Outcomes in Hirschsprung's Disease (GOSH)
Questionnaire for adult patients
V1.1 18/09/2017

Bowel function - Rintala score

1. You feel when you need to pass stool	Always	Most of the time	Uncertain	Never	
	☐	☐	☐	☐	
2. You are able to hold back passing stool	Always	Problems less than once a week	Weekly problems	No voluntary control	
	☐	☐	☐	☐	
3. You need to pass stool:	Less than once every 2 days	Once every 2 days	Once a day	Twice a day	More than twice a day
	☐	☐	☐	☐	☐
4. You have problems with faecal soiling (staining of the underwear):	Never	Less than once a week, change of underwear needed only rarely	More than once a week, change of underwear often needed	Daily, requiring protective aids (i.e. pads or diapers)	
	☐	☐	☐	☐	
5. You have accidents involving stool:	Never	Less than once a week	Weekly, requiring protective aids	Daily, requiring protective aids day and night	
	☐	☐	☐	☐	
6. Regarding constipation	No constipation	Constipation you manage with diet alone	Constipation you manage with medication	Constipation you manage with enemas	
	☐	☐	☐	☐	
7. Do you suffer from involuntary gas leakage (passing wind without realising)?	Never	Less than once a week	More than once a week	Daily / constantly	
	☐	☐	☐	☐	
8. Social impact of bowel function	No impact	Some impact (i.e bad smells sometimes)	Problems restricting social activities	Major social or psychological problems as a result of bowel function	
	☐	☐	☐	☐	

Please continue on the next page

9. Have you ever had enterocolitis (bowel inflammation) related to Hirschsprung's Disease?

Yes ☐/ No ☐

10. Have you had an episode of enterocolitis in the past year?

Yes ☐/ No ☐

11. Have you ever had recurrent enterocolitis (<u>more than 4 episodes in one year</u>?)

Yes ☐/ No ☐

Is there anything further you would wish to tell us about?

Long-Term Outcomes in Hirschsprung's Disease (GOSH)
Questionnaire for adult patients
V1.1 18/09/2017

Urinary Function Questionnaire

1. Have you ever had a urinary tract infection (UTI)?	No	Yes	If yes, how many UTI in the past year?	
	☐	☐		
2. How many times do you pass urine each day?	1 – 3 times	4 – 8 times	More than 8 times	
	☐	☐	☐	
3. Do you ever need to strain to start/continue urination?	Never	Rarely	Often	Always
	☐	☐	☐	☐
4. Does you ever get a sudden urge to pass urine?	Never	Rarely	Often	Always
	☐	☐	☐	☐
5. Is the urge so strong that urine escapes before reaching the toilet?	Never	Rarely	Often	Always
	☐	☐	☐	☐
6. Does urine ever leak upon straining (e.g.laughing, sneezing or coughing)?	Never	Rarely	Often	Always
	☐	☐	☐	☐
7. Does urine ever leak without physical activity or apparent need to urinate?	Never	Rarely	Often	Always
	☐	☐	☐	☐
8. Do you ever have night-time wetting (bedwetting)?	Never	Less than once a week	More often than once a week	Every night
	☐	☐	☐	☐
9. Do you consider yourself to have social problems <u>due to urinary incontinence</u>?	No	Yes, due to daytime urinary incontinence only	Yes, due to night-time urinary incontinence only	Yes, due to daytime and night-time urinary incontinence
	☐	☐	☐	☐

Gastrointestinal Quality of Life Index (GIQLI) – please answer in reference only to the time period in the question

1. How often in the past 2 weeks have you experienced pain in the abdomen	All of the time	Most of the time	Some of the time	A little of the time	Never
	☐	☐	☐	☐	☐
2. How often in the past 2 weeks have you experienced a fullness in the upper abdomen	All of the time	Most of the time	Some of the time	A little of the time	Never
	☐	☐	☐	☐	☐
3. How often in the past 2 weeks have you experienced bloating / gassy	All of the time	Most of the time	Some of the time	A little of the time	Never
	☐	☐	☐	☐	☐
4. How often in the past 2 weeks have you been troubled by excessive wind/flatulence/farting	All of the time	Most of the time	Some of the time	A little of the time	Never
	☐	☐	☐	☐	☐
5. How often in the past 2 weeks have you been troubled by strong burping / belching?	All of the time	Most of the time	Some of the time	A little of the time	Never
	☐	☐	☐	☐	☐
6. How often in the past 2 weeks have you been troubled by gurgling noises in the abdomen/tummy	All of the time	Most of the time	Some of the time	A little of the time	Never
	☐	☐	☐	☐	☐
7. How often in the past 2 weeks have you been troubled by frequent bowel movements?	All of the time	Most of the time	Some of the time	A little of the time	Never
	☐	☐	☐	☐	☐
8. How often in the past 2 weeks have you found eating to be a pleasure?	Never	A little of the time	Some of the time	Most of the time	All of the time
	☐	☐	☐	☐	☐
9. Because of your illness, to what extent have you restricted the kinds of foods you eat?	Very much	Much	Somewhat	A little	Not at all
	☐	☐	☐	☐	☐
10. During the past 2 weeks, how well have you been able to cope with everyday streses?	Extremely poorly	Poorly	Moderately	Well	Extremely well
	☐	☐	☐	☐	☐

Please continue on the next page

11. How often during the past 2 weeks have you been sad about being ill?	All of the time	Most of the time	Some of the time	A little of the time	Never
	☐	☐	☐	☐	☐
12. How often during the past 2 weeks have you been nervous or anxious about your illness?	All of the time	Most of the time	Some of the time	A little of the time	Never
	☐	☐	☐	☐	☐
13. How often during the past 2 weeks have you been happy about life in general?	Never	A little of the time	Some of the time	Most of the time	All of the time
	☐	☐	☐	☐	☐
14. How often during the past 2 weeks have you been frustrated about your illness?	All of the time	Most of the time	Some of the time	A little of the time	Never
	☐	☐	☐	☐	☐
15. How often during the past 2 weeks have you been tired or fatigued?	All of the time	Most of the time	Some of the time	A little of the time	Never
	☐	☐	☐	☐	☐
16. How often during the past 2 weeks have you felt unwell?	All of the time	Most of the time	Some of the time	A little of the time	Never
	☐	☐	☐	☐	☐
17. Over the past week, have you woken up in the night?	Every night	5-6 nights	3-4 nights	1-2 nights	Never
	☐	☐	☐	☐	☐
18. Have you been troubled by changes in your appearance because of your illness?	A great deal	A moderate amount	Somewhat	A little bit	Not at all
	☐	☐	☐	☐	☐
19. How much physical strength do you feel you have lost because of your illness?	A great deal	A moderate amount	Somewhat	A little bit	Not at all
	☐	☐	☐	☐	☐
20. How much endurance have you lost because of your illness?	A great deal	A moderate amount	Somewhat	A little bit	Not at all
	☐	☐	☐	☐	☐
21. Because of your illness, to what extent do you feel unfit?	Extremely unfit	Moderately unfit	Somewhat unfit	A little unfit	Fit
	☐	☐	☐	☐	☐

Please continue on the next page

22. During the past 2 weeks, how often have you been able to complete your normal daily activities (school, work, household)?	All of the time	Most of the time	Some of the time	A little of the time	Never
	☐	☐	☐	☐	☐
23. During the past 2 weeks, how often have you been able to take part in your usual patterns of leisure or recreational activities?	All of the time	Most of the time	Some of the time	A little of the time	Never
	☐	☐	☐	☐	☐
24. During the past 2 weeks, how much have you been troubled by the medical treatment of your illness?	Very much	Much	Somewhat	A little	Not at all
	☐	☐	☐	☐	☐
25. To what extent have your personal relations with people close to you (family and friends) worsened because of your illness?	Very much	Much	Somewhat	A little	Not at all
	☐	☐	☐	☐	☐
26. To what extent has your sexual life been impaired because of your illness?	Very much	Much	Somewhat	A little	Not at all
	☐	☐	☐	☐	☐
27. How often in the past 2 weeks have you been troubled by liquid or food coming up into your mouth (regurgitation)?	All of the time	Most of the time	Some of the time	A little of the time	Never
	☐	☐	☐	☐	☐
28. How often during the past 2 weeks have you felt uncomfortable because of a slow speed of eating?	All of the time	Most of the time	Some of the time	A little of the time	Never
	☐	☐	☐	☐	☐
29. How of then during the past 2 weeks have you had trouble swallowing your food?	All of the time	Most of the time	Some of the time	A little of the time	Never
	☐	☐	☐	☐	☐
30. How often during the past 2 weeks have you been troubled by urgent bowel movements?	All of the time	Most of the time	Some of the time	A little of the time	Never
	☐	☐	☐	☐	☐
31. How often during the past 2 weeks have you been troubled by diarrhoea (loose bowel movements)?	All of the time	Most of the time	Some of the time	A little of the time	Never
	☐	☐	☐	☐	☐

Please continue on the next page

32. How often during the past 2 weeks have you been troubled by constipation (hard stools)?	All of the time	Most of the time	Some of the time	A little of the time	Never
	☐	☐	☐	☐	☐
33. How often during the past 2 weeks have you been troubled by nausea (feeling sick)?	All of the time	Most of the time	Some of the time	A little of the time	Never
	☐	☐	☐	☐	☐
34. How often during the past 2 weeks have you been troubled by blood in the stool?	All of the time	Most of the time	Some of the time	A little of the time	Never
	☐	☐	☐	☐	☐
35.. How often during the past 2 weeks have you been troubled by heartburn?	All of the time	Most of the time	Some of the time	A little of the time	Never
	☐	☐	☐	☐	☐
36. How often during the past 2 weeks have you been troubled by uncontrolled stools (incontinence)?	All of the time	Most of the time	Some of the time	A little of the time	Never
	☐	☐	☐	☐	☐

Is there anything further you would wish to tell us about?

Long-Term Outcomes in Hirschsprung's Disease (GOSH)
Questionnaire for adult patients
V1.1 18/09/2017

SF-36 Survey: This survey asks for your views about your health. This information will help keep track of how you feel and how well you are able to do your usual activities. For each of the following questions, please select the number that best describes your answer.

1. In general, would you say your health is:	Excellent	Very Good	Good	Fair	Poor
	☐ 1	☐ 2	☐ 3	☐ 4	☐ 5
2. Compared to one year ago, your health is:	Much better than one year ago	Somewhat better than one year ago	About the same	Somewhat worse than one year ago	Much worse now than one year ago
	☐ 1	☐ 2	☐ 3	☐ 4	☐ 5

3. The following items are about activities you might do during a typical day. <u>Does your health now limit you in these activities? If so, how much?</u>			
a. Vigorous activities, such as running, lifting heavy objects, participating in strenuous sports	Yes, limited a lot	Yes, limited a little	No, Not limited at all
	☐ 1	☐ 2	☐ 3
b. Moderate activities, such as moving a table, pushing a vacuum cleaner, bowling, or playing golf	Yes, limited a lot	Yes, limited a little	No, Not limited at all
	☐ 1	☐ 2	☐ 3
c. Lifting or carrying groceries	Yes, limited a lot	Yes, limited a little	No, Not limited at all
	☐ 1	☐ 2	☐ 3
d. Climbing several flights of stairs	Yes, limited a lot	Yes, limited a little	No, Not limited at all
	☐ 1	☐ 2	☐ 3
e. Climbing one flight of stairs	Yes, limited a lot	Yes, limited a little	No, Not limited at all
	☐ 1	☐ 2	☐ 3
f. Bending, kneeling, or stooping	Yes, limited a lot	Yes, limited a little	No, Not limited at all
	☐ 1	☐ 2	☐ 3
g. Walking more than a mile	Yes, limited a lot	Yes, limited a little	No, Not limited at all
	☐ 1	☐ 2	☐ 3
h. Walking several blocks	Yes, limited a lot	Yes, limited a little	No, Not limited at all
	☐ 1	☐ 2	☐ 3

Please continue on the next page

i. Walking one block	Yes, limited a lot	Yes, limited a little	No, Not limited at all
	☐ 1	☐ 2	☐ 3
j. Bathing or dressing yourself	Yes, limited a lot	Yes, limited a little	No, Not limited at all
	☐ 1	☐ 2	☐ 3

4. During the past 4 weeks, have you had any of the following problems with your work or other regular daily activities as a result of your physical health?		
a. Cut down the amount of time you spent on work or other activities	☐ Yes	☐ No
b. Accomplished less than you would like	☐ Yes	☐ No
c. Were limited in the kind of work or other activities	☐ Yes	☐ No
d. Had difficulty performing the work or other activities (for example, it took extra effort)	☐ Yes	☐ No
5. During the past 4 weeks, have you had any of the following problems with your work or other regular daily activities as a result of any emotional problems (such as feeling depressed or anxious)?		
a. Cut down the amount of time you spent on work or other activities	☐ Yes	☐ No
b. Accomplished less than you would like	☐ Yes	☐ No
c. Didn't do work or other activities as carefully as usual	☐ Yes	☐ No

6. During the past 4 weeks, to what extent has your physical health or emotional problems interfered with your normal social activities with family, friends, neighbors, or groups?	Not at all	Slightly	Moderately	Quite a bit	Extremely
	☐ 1	☐ 2	☐ 3	☐ 4	☐ 5

7. How much bodily pain have you had during the past 4 weeks?	None	Very mild	Mild	Moderate	Severe	Very severe
	☐ 1	☐ 2	☐ 3	☐ 4	☐ 5	☐ 6

Please continue on the next page

8. During the past 4 weeks, how much did pain interfere with your normal work (including both work outside the home and housework)?	Not at all	A little bit	Moderately	Quite a bit	Extremely
	☐ 1	☐ 2	☐ 3	☐ 4	☐ 5

These questions are about how you feel and how things have been with you during the past 4 weeks. For each question, please give the one answer that comes closest to the way you have been feeling. (Select One Number on Each Line)

9. How much of the time during the past 4 weeks . . .						
a. Did you feel full of energy?	All of the Time	Most of the Time	A Good Bit of the Time	Some of the Time	A Little of the Time	None of the Time
	☐ 1	☐ 2	☐ 3	☐ 4	☐ 5	☐ 6
b. Have you been a very nervous person?	All of the Time	Most of the Time	A Good Bit of the Time	Some of the Time	A Little of the Time	None of the Time
	☐ 1	☐ 2	☐ 3	☐ 4	☐ 5	☐ 6
c. Have you felt so down in the dumps that nothing could cheer you up?	All of the Time	Most of the Time	A Good Bit of the Time	Some of the Time	A Little of the Time	None of the Time
	☐ 1	☐ 2	☐ 3	☐ 4	☐ 5	☐ 6
d. Have you felt calm and peaceful?	All of the Time	Most of the Time	A Good Bit of the Time	Some of the Time	A Little of the Time	None of the Time
	☐ 1	☐ 2	☐ 3	☐ 4	☐ 5	☐ 6
e. Did you have a lot of energy?	All of the Time	Most of the Time	A Good Bit of the Time	Some of the Time	A Little of the Time	None of the Time
	☐ 1	☐ 2	☐ 3	☐ 4	☐ 5	☐ 6
f. Have you felt downhearted and blue?	All of the Time	Most of the Time	A Good Bit of the Time	Some of the Time	A Little of the Time	None of the Time
	☐ 1	☐ 2	☐ 3	☐ 4	☐ 5	☐ 6
g. Did you feel worn out?	All of the Time	Most of the Time	A Good Bit of the Time	Some of the Time	A Little of the Time	None of the Time
	☐ 1	☐ 2	☐ 3	☐ 4	☐ 5	☐ 6

Please continue on the next page

h. Have you been a happy person?	All of the Time	Most of the Time	A Good Bit of the Time	Some of the Time	A Little of the Time	None of the Time
	☐ 1	☐ 2	☐ 3	☐ 4	☐ 5	☐ 6
i. Did you feel tired?	All of the Time	Most of the Time	A Good Bit of the Time	Some of the Time	A Little of the Time	None of the Time
	☐ 1	☐ 2	☐ 3	☐ 4	☐ 5	☐ 6

10. During the past 4 weeks, how much of the time has your physical health or emotional problems interfered with your social activities (like visiting with friends, relatives, etc.)?	All of the time	Most of the time	Some of the time	A little of the time	None of the time
	☐ 1	☐ 2	☐ 3	☐ 4	☐ 5

11. How TRUE or FALSE is each of the following statements for you.					
a. I seem to get sick a little easier than other people	Definitely True	Mostly True	Don't Know	Mostly False	Definitely False
	☐ 1	☐ 2	☐ 3	☐ 4	☐ 5
b. I am as healthy as anybody I know	Definitely True	Mostly True	Don't Know	Mostly False	Definitely False
	☐ 1	☐ 2	☐ 3	☐ 4	☐ 5
c. I expect my health to get worse	Definitely True	Mostly True	Don't Know	Mostly False	Definitely False
	☐ 1	☐ 2	☐ 3	☐ 4	☐ 5
d. My health is excellent	Definitely True	Mostly True	Don't Know	Mostly False	Definitely False
	☐ 1	☐ 2	☐ 3	☐ 4	☐ 5

Is there anything further you would wish to tell us about?

Long-Term Outcomes in Hirschsprung's Disease (GOSH)
Questionnaire for adult patients
V1.1 18/09/2017

Male sexual function questions – Please remember your responses will be treated with the utmost confidentiality.

1. Are you currently in a stable relationship?

☐ Yes / ☐ No

2. Have you had sexual intercourse? (if not please continue to question 6.)

☐ Yes / ☐ No

If yes, first at age years

3. Have you tried to have children with your partner?

☐ No
☐ Yes, but I have not yet conceived (fallen pregnant)
☐ Yes, we have fallen pregnant times

4. Do you have biological children?

☐ Yes, children
☐ No

Additional information (e.g. if you have biological children with other than your current spouse/partner):

5. Have you needed any fertility investigations or treatment?

☐ No, we have not needed them (no problems conceiving)
☐ No, I have not tried to have a baby
☐ No, but we are considering going for investigations
☐ Yes

Please give details:

6. Are you able to climax (have an orgasm) from sexual stimulation?

☐ Never
☐ Rarely
☐ Sometimes
☐ Usually/Always

6. Do you ejaculate when you orgasm?

☐ Yes
☐ No – absent or dry ejaculations

7. EHS (Erectile Hardness Score

How would you rate your erections/erectile function (tick box that applies)

☐ Penis is completely hard and fully rigid
☐ Penis is hard enough for penetration, but not completely hard
☐ Penis is hard, but not hard enough for penetration.
☐ Penis is larger, but not hard
☐ Penis does not enlarge.

8. Male sexual function index (MSFI)

Please circle the most appropriate answer:

	completely agree	**moderately agree**	**slightly agree**	**slightly disagree**	**moderately disagree**	**completely disagree**
1. When I think about my sexual life, I feel frustrated	☐	☐	☐	☐	☐	☐
2. When I think about my sexual life, I feel depressed	☐	☐	☐	☐	☐	☐
3. When I think about my sexual life, I feel like less of a man	☐	☐	☐	☐	☐	☐
4. I have lost confidence in myself as a sexual partner	☐	☐	☐	☐	☐	☐
5. When I think about my sexual life, I feel anxious	☐	☐	☐	☐	☐	☐
6. When I think about my sexual life, I feel angry	☐	☐	☐	☐	☐	☐
7. I worry about the future of my sexual life	☐	☐	☐	☐	☐	☐
8. When I think about my sexual life, I am embarrassed	☐	☐	☐	☐	☐	☐
9. When I think about my sexual life, I feel guilty	☐	☐	☐	☐	☐	☐
10. When I think about my sexual life, I worry that my partner feels hurt or rejected	☐	☐	☐	☐	☐	☐
11. When I think about my sexual life, I feel like I have lost something	☐	☐	☐	☐	☐	☐

Is there anything further you would wish to tell us about?

That is the end of the questionnaire, thank you for taking part.

Long-Term Outcomes in Hirschsprung's Disease (GOSH)
Questionnaire for adult patients
V1.1 18/09/2017

Open questionnaire

Could you tell us about your transition from care at Great Ormond Street to adult services?

Where do you currently see a specialist for follow up of your Hirschsprung's Disease?

Have you had any further operations since you transitioned into adult care, including a stoma formation or Botox injections?

Do you currently have a stoma (colostomy or ileostomy)?

Yes ☐

No ☐

Do you currently take any medication for constipation, such as Movicol or Lactulose?

Do you take any other medication for any other reason?

Do you have any food allergies or intolerances?

What is your current height and weight?

Height? cm Weight? kg

Do you know of any family members who have Hirschsprung's Disease?

Bowel function - Rintala score

1. You feel when you need to pass stool	Always	Most of the time	Uncertain	Never	
	☐	☐	☐	☐	
2. You are able to hold back passing stool	Always	Problems less than once a week	Weekly problems	No voluntary control	
	☐	☐	☐	☐	
3. You need to pass stool:	Less than once every 2 days	Once every 2 days	Once a day	Twice a day	More than twice a day
	☐	☐	☐	☐	☐
4. You have problems with faecal soiling (staining of the underwear):	Never	Less than once a week, change of underwear needed only rarely	More than once a week, change of underwear often needed	Daily, requiring protective aids (i.e. pads or diapers)	
	☐	☐	☐	☐	
5. You have accidents involving stool:	Never	Less than once a week	Weekly, requiring protective aids	Daily, requiring protective aids day and night	
	☐	☐	☐	☐	
6. Regarding constipation	No constipation	Constipation you manage with diet alone	Constipation you manage with medication	Constipation you manage with enemas	
	☐	☐	☐	☐	
7. Do you suffer from involuntary gas leakage (passing wind without realising)?	Never	Less than once a week	More than once a week	Daily / constantly	
	☐	☐	☐	☐	
8. Social impact of bowel function	No impact	Some impact (i.e bad smells sometimes)	Problems restricting social activities	Major social or psychological problems as a result of bowel function	
	☐	☐	☐	☐	

Please continue on the next page

9. Have you ever had enterocolitis (bowel inflammation) related to Hirschsprung's Disease?

Yes ☐/ No ☐

10. Have you had an episode of enterocolitis in the past year?

Yes ☐/ No ☐

11. Have you ever had recurrent enterocolitis (<u>more than 4 episodes in one year</u>?)

Yes ☐/ No ☐

Is there anything further you would wish to tell us about?

Urinary Function Questionnaire

1. Have you ever had a urinary tract infection (UTI)?	No	Yes	If yes, how many UTI in the past year?	
	☐	☐		
2. How many times do you pass urine each day?	1 – 3 times	4 – 8 times	More than 8 times	
	☐	☐	☐	
3. Do you ever need to strain to start/continue urination?	Never	Rarely	Often	Always
	☐	☐	☐	☐
4. Does you ever get a sudden urge to pass urine?	Never	Rarely	Often	Always
	☐	☐	☐	☐
5. Is the urge so strong that urine escapes before reaching the toilet?	Never	Rarely	Often	Always
	☐	☐	☐	☐
6. Does urine ever leak upon straining (e.g.laughing, sneezing or coughing)?	Never	Rarely	Often	Always
	☐	☐	☐	☐
7. Does urine ever leak without physical activity or apparent need to urinate?	Never	Rarely	Often	Always
	☐	☐	☐	☐
8. Do you ever have night-time wetting (bedwetting)?	Never	Less than once a week	More often than once a week	Every night
	☐	☐	☐	☐
9. Do you consider yourself to have social problems <u>due to urinary incontinence</u>?	No	Yes, due to daytime urinary incontinence only	Yes, due to night-time urinary incontinence only	Yes, due to daytime and night-time urinary incontinence
	☐	☐	☐	☐

Gastrointestinal Quality of Life Index (GIQLI) – please answer in reference only to the time period in the question

1. How often in the past 2 weeks have you experienced pain in the abdomen	All of the time	Most of the time	Some of the time	A little of the time	Never
	☐	☐	☐	☐	☐
2. How often in the past 2 weeks have you experienced a fullness in the upper abdomen	All of the time	Most of the time	Some of the time	A little of the time	Never
	☐	☐	☐	☐	☐
3. How often in the past 2 weeks have you experienced bloating / gassy	All of the time	Most of the time	Some of the time	A little of the time	Never
	☐	☐	☐	☐	☐
4. How often in the past 2 weeks have you been troubled by excessive wind/flatulence/farting	All of the time	Most of the time	Some of the time	A little of the time	Never
	☐	☐	☐	☐	☐
5. How often in the past 2 weeks have you been troubled by strong burping / belching?	All of the time	Most of the time	Some of the time	A little of the time	Never
	☐	☐	☐	☐	☐
6. How often in the past 2 weeks have you been troubled by gurgling noises in the abdomen/tummy	All of the time	Most of the time	Some of the time	A little of the time	Never
	☐	☐	☐	☐	☐
7. How often in the past 2 weeks have you been troubled by frequent bowel movements?	All of the time	Most of the time	Some of the time	A little of the time	Never
	☐	☐	☐	☐	☐
8. How often in the past 2 weeks have you found eating to be a pleasure?	Never	A little of the time	Some of the time	Most of the time	All of the time
	☐	☐	☐	☐	☐
9. Because of your illness, to what extent have you restricted the kinds of foods you eat?	Very much	Much	Somewhat	A little	Not at all
	☐	☐	☐	☐	☐
10. During the past 2 weeks, how well have you been able to cope with everyday streses?	Extremely poorly	Poorly	Moderately	Well	Extremely well
	☐	☐	☐	☐	☐

Please continue on the next page

11. How often during the past 2 weeks have you been sad about being ill?	All of the time	Most of the time	Some of the time	A little of the time	Never
	☐	☐	☐	☐	☐
12. How often during the past 2 weeks have you been nervous or anxious about your illness?	All of the time	Most of the time	Some of the time	A little of the time	Never
	☐	☐	☐	☐	☐
13. How often during the past 2 weeks have you been happy about life in general?	Never	A little of the time	Some of the time	Most of the time	All of the time
	☐	☐	☐	☐	☐
14. How often during the past 2 weeks have you been frustrated about your illness?	All of the time	Most of the time	Some of the time	A little of the time	Never
	☐	☐	☐	☐	☐
15. How often during the past 2 weeks have you been tired or fatigued?	All of the time	Most of the time	Some of the time	A little of the time	Never
	☐	☐	☐	☐	☐
16. How often during the past 2 weeks have you felt unwell?	All of the time	Most of the time	Some of the time	A little of the time	Never
	☐	☐	☐	☐	☐
17. Over the past week, have you woken up in the night?	Every night	5-6 nights	3-4 nights	1-2 nights	Never
	☐	☐	☐	☐	☐
18. Have you been troubled by changes in your appearance because of your illness?	A great deal	A moderate amount	Somewhat	A little bit	Not at all
	☐	☐	☐	☐	☐
19. How much physical strength do you feel you have lost because of your illness?	A great deal	A moderate amount	Somewhat	A little bit	Not at all
	☐	☐	☐	☐	☐
20. How much endurance have you lost because of your illness?	A great deal	A moderate amount	Somewhat	A little bit	Not at all
	☐	☐	☐	☐	☐
21. Because of your illness, to what extent do you feel unfit?	Extremely unfit	Moderately unfit	Somewhat unfit	A little unfit	Fit
	☐	☐	☐	☐	☐

Please continue on the next page

22. During the past 2 weeks, how often have you been able to complete your normal daily activities (school, work, household)?	All of the time	Most of the time	Some of the time	A little of the time	Never
	☐	☐	☐	☐	☐
23. During the past 2 weeks, how often have you been able to take part in your usual patterns of leisure or recreational activities?	All of the time	Most of the time	Some of the time	A little of the time	Never
	☐	☐	☐	☐	☐
24. During the past 2 weeks, how much have you been troubled by the medical treatment of your illness?	Very much	Much	Somewhat	A little	Not at all
	☐	☐	☐	☐	☐
25. To what extent have your personal relations with people close to you (family and friends) worsened because of your illness?	Very much	Much	Somewhat	A little	Not at all
	☐	☐	☐	☐	☐
26. To what extent has your sexual life been impaired because of your illness?	Very much	Much	Somewhat	A little	Not at all
	☐	☐	☐	☐	☐
27. How often in the past 2 weeks have you been troubled by liquid or food coming up into your mouth (regurgitation)?	All of the time	Most of the time	Some of the time	A little of the time	Never
	☐	☐	☐	☐	☐
28. How often during the past 2 weeks have you felt uncomfortable because of a slow speed of eating?	All of the time	Most of the time	Some of the time	A little of the time	Never
	☐	☐	☐	☐	☐
29. How of then during the past 2 weeks have you had trouble swallowing your food?	All of the time	Most of the time	Some of the time	A little of the time	Never
	☐	☐	☐	☐	☐
30. How often during the past 2 weeks have you been troubled by urgent bowel movements?	All of the time	Most of the time	Some of the time	A little of the time	Never
	☐	☐	☐	☐	☐
31. How often during the past 2 weeks have you been troubled by diarrhoea (loose bowel movements)?	All of the time	Most of the time	Some of the time	A little of the time	Never
	☐	☐	☐	☐	☐

Please continue on the next page

32. How often during the past 2 weeks have you been troubled by constipation (hard stools)?	All of the time	Most of the time	Some of the time	A little of the time	Never
	☐	☐	☐	☐	☐
33. How often during the past 2 weeks have you been troubled by nausea (feeling sick)?	All of the time	Most of the time	Some of the time	A little of the time	Never
	☐	☐	☐	☐	☐
34. How often during the past 2 weeks have you been troubled by blood in the stool?	All of the time	Most of the time	Some of the time	A little of the time	Never
	☐	☐	☐	☐	☐
35.. How often during the past 2 weeks have you been troubled by heartburn?	All of the time	Most of the time	Some of the time	A little of the time	Never
	☐	☐	☐	☐	☐
36. How often during the past 2 weeks have you been troubled by uncontrolled stools (incontinence)?	All of the time	Most of the time	Some of the time	A little of the time	Never
	☐	☐	☐	☐	☐

Is there anything further you would wish to tell us about?

Long-Term Outcomes in Hirschsprung's Disease (GOSH)
Questionnaire for adult patients
V1.1 18/09/2017

SF-36 Survey: This survey asks for your views about your health. This information will help keep track of how you feel and how well you are able to do your usual activities. For each of the following questions, please select the number that best describes your answer.

1. In general, would you say your health is:	Excellent	Very Good	Good	Fair	Poor
	☐ 1	☐ 2	☐ 3	☐ 4	☐ 5
2. Compared to one year ago, your health is:	Much better than one year ago	Somewhat better than one year ago	About the same	Somewhat worse than one year ago	Much worse now than one year ago
	☐ 1	☐ 2	☐ 3	☐ 4	☐ 5

3. The following items are about activities you might do during a typical day. <u>Does your health now limit you in these activities? If so, how much?</u>			
a. Vigorous activities, such as running, lifting heavy objects, participating in strenuous sports	Yes, limited a lot	Yes, limited a little	No, Not limited at all
	☐ 1	☐ 2	☐ 3
b. Moderate activities, such as moving a table, pushing a vacuum cleaner, bowling, or playing golf	Yes, limited a lot	Yes, limited a little	No, Not limited at all
	☐ 1	☐ 2	☐ 3
c. Lifting or carrying groceries	Yes, limited a lot	Yes, limited a little	No, Not limited at all
	☐ 1	☐ 2	☐ 3
d. Climbing several flights of stairs	Yes, limited a lot	Yes, limited a little	No, Not limited at all
	☐ 1	☐ 2	☐ 3
e. Climbing one flight of stairs	Yes, limited a lot	Yes, limited a little	No, Not limited at all
	☐ 1	☐ 2	☐ 3
f. Bending, kneeling, or stooping	Yes, limited a lot	Yes, limited a little	No, Not limited at all
	☐ 1	☐ 2	☐ 3
g. Walking more than a mile	Yes, limited a lot	Yes, limited a little	No, Not limited at all
	☐ 1	☐ 2	☐ 3
h. Walking several blocks	Yes, limited a lot	Yes, limited a little	No, Not limited at all
	☐ 1	☐ 2	☐ 3

Please continue on the next page

i. Walking one block	Yes, limited a lot	Yes, limited a little	No, Not limited at all
	☐ 1	☐ 2	☐ 3
j. Bathing or dressing yourself	Yes, limited a lot	Yes, limited a little	No, Not limited at all
	☐ 1	☐ 2	☐ 3

4. During the past 4 weeks, have you had any of the following problems with your work or other regular daily activities as a result of your physical health?		
a. Cut down the amount of time you spent on work or other activities	☐ Yes	☐ No
b. Accomplished less than you would like	☐ Yes	☐ No
c. Were limited in the kind of work or other activities	☐ Yes	☐ No
d. Had difficulty performing the work or other activities (for example, it took extra effort)	☐ Yes	☐ No
5. During the past 4 weeks, have you had any of the following problems with your work or other regular daily activities as a result of any emotional problems (such as feeling depressed or anxious)?		
a. Cut down the amount of time you spent on work or other activities	☐ Yes	☐ No
b. Accomplished less than you would like	☐ Yes	☐ No
c. Didn't do work or other activities as carefully as usual	☐ Yes	☐ No

6. During the past 4 weeks, to what extent has your physical health or emotional problems interfered with your normal social activities with family, friends, neighbors, or groups?	Not at all	Slightly	Moderately	Quite a bit	Extremely
	☐ 1	☐ 2	☐ 3	☐ 4	☐ 5

7. How much bodily pain have you had during the past 4 weeks?	None	Very mild	Mild	Moderate	Severe	Very severe
	☐ 1	☐ 2	☐ 3	☐ 4	☐ 5	☐ 6

Please continue on the next page

8. During the past 4 weeks, how much did pain interfere with your normal work (including both work outside the home and housework)?	Not at all	A little bit	Moderately	Quite a bit	Extremely
	☐ 1	☐ 2	☐ 3	☐ 4	☐ 5

These questions are about how you feel and how things have been with you during the past 4 weeks. For each question, please give the one answer that comes closest to the way you have been feeling. (Select One Number on Each Line)

9. How much of the time during the past 4 weeks . . .						
a. Did you feel full of energy?	All of the Time	Most of the Time	A Good Bit of the Time	Some of the Time	A Little of the Time	None of the Time
	☐ 1	☐ 2	☐ 3	☐ 4	☐ 5	☐ 6
b. Have you been a very nervous person?	All of the Time	Most of the Time	A Good Bit of the Time	Some of the Time	A Little of the Time	None of the Time
	☐ 1	☐ 2	☐ 3	☐ 4	☐ 5	☐ 6
c. Have you felt so down in the dumps that nothing could cheer you up?	All of the Time	Most of the Time	A Good Bit of the Time	Some of the Time	A Little of the Time	None of the Time
	☐ 1	☐ 2	☐ 3	☐ 4	☐ 5	☐ 6
d. Have you felt calm and peaceful?	All of the Time	Most of the Time	A Good Bit of the Time	Some of the Time	A Little of the Time	None of the Time
	☐ 1	☐ 2	☐ 3	☐ 4	☐ 5	☐ 6
e. Did you have a lot of energy?	All of the Time	Most of the Time	A Good Bit of the Time	Some of the Time	A Little of the Time	None of the Time
	☐ 1	☐ 2	☐ 3	☐ 4	☐ 5	☐ 6
f. Have you felt downhearted and blue?	All of the Time	Most of the Time	A Good Bit of the Time	Some of the Time	A Little of the Time	None of the Time
	☐ 1	☐ 2	☐ 3	☐ 4	☐ 5	☐ 6
g. Did you feel worn out?	All of the Time	Most of the Time	A Good Bit of the Time	Some of the Time	A Little of the Time	None of the Time
	☐ 1	☐ 2	☐ 3	☐ 4	☐ 5	☐ 6

Please continue on the next page

h. Have you been a happy person?	All of the Time	Most of the Time	A Good Bit of the Time	Some of the Time	A Little of the Time	None of the Time
	☐ 1	☐ 2	☐ 3	☐ 4	☐ 5	☐ 6
i. Did you feel tired?	All of the Time	Most of the Time	A Good Bit of the Time	Some of the Time	A Little of the Time	None of the Time
	☐ 1	☐ 2	☐ 3	☐ 4	☐ 5	☐ 6

10. During the past 4 weeks, how much of the time has your physical health or emotional problems interfered with your social activities (like visiting with friends, relatives, etc.)?	All of the time	Most of the time	Some of the time	A little of the time	None of the time
	☐ 1	☐ 2	☐ 3	☐ 4	☐ 5

11. How TRUE or FALSE is each of the following statements for you.					
a. I seem to get sick a little easier than other people	Definitely True	Mostly True	Don't Know	Mostly False	Definitely False
	☐ 1	☐ 2	☐ 3	☐ 4	☐ 5
b. I am as healthy as anybody I know	Definitely True	Mostly True	Don't Know	Mostly False	Definitely False
	☐ 1	☐ 2	☐ 3	☐ 4	☐ 5
c. I expect my health to get worse	Definitely True	Mostly True	Don't Know	Mostly False	Definitely False
	☐ 1	☐ 2	☐ 3	☐ 4	☐ 5
d. My health is excellent	Definitely True	Mostly True	Don't Know	Mostly False	Definitely False
	☐ 1	☐ 2	☐ 3	☐ 4	☐ 5

Is there anything further you would wish to tell us about?

Long-Term Outcomes in Hirschsprung's Disease (GOSH)
Questionnaire for adult patients
V1.1 18/09/2017

Female Sexual function questions – Please remember your responses will be treated with the utmost confidentiality.

1. Are you currently in a stable relationship?

☐ Yes / ☐ No

2. Have you started having periods?

☐ Yes / ☐ No

If yes, began at age years

3. Have you had sexual intercourse? (If No please move straight to question 8 – FSFI)

☐ Yes / ☐ No

If yes, first at age years

4. Have you tried to have children with your partner?

☐ *No*
☐ *Yes, but I have not yet conceived (fallen pregnant)*
☐ *Yes, I have fallen pregnant times*

5. Do you have biological children?

☐ *Yes, children*
☐ *No*

Additional information (e.g. if you have biological children with other than your current spouse/partner):

6. Have you needed any fertility investigations or treatment?

☐ *No, we have not needed them (no problems conceiving)*
☐ *No, I have not tried to have a baby*
☐ *No, but we are considering going for investigations*
☐ *Yes*

Please give details:

7. Do you have pain when having intercourse?

☐ *Yes (Often/Always)*
☐ *No (Not usually or not at all)*

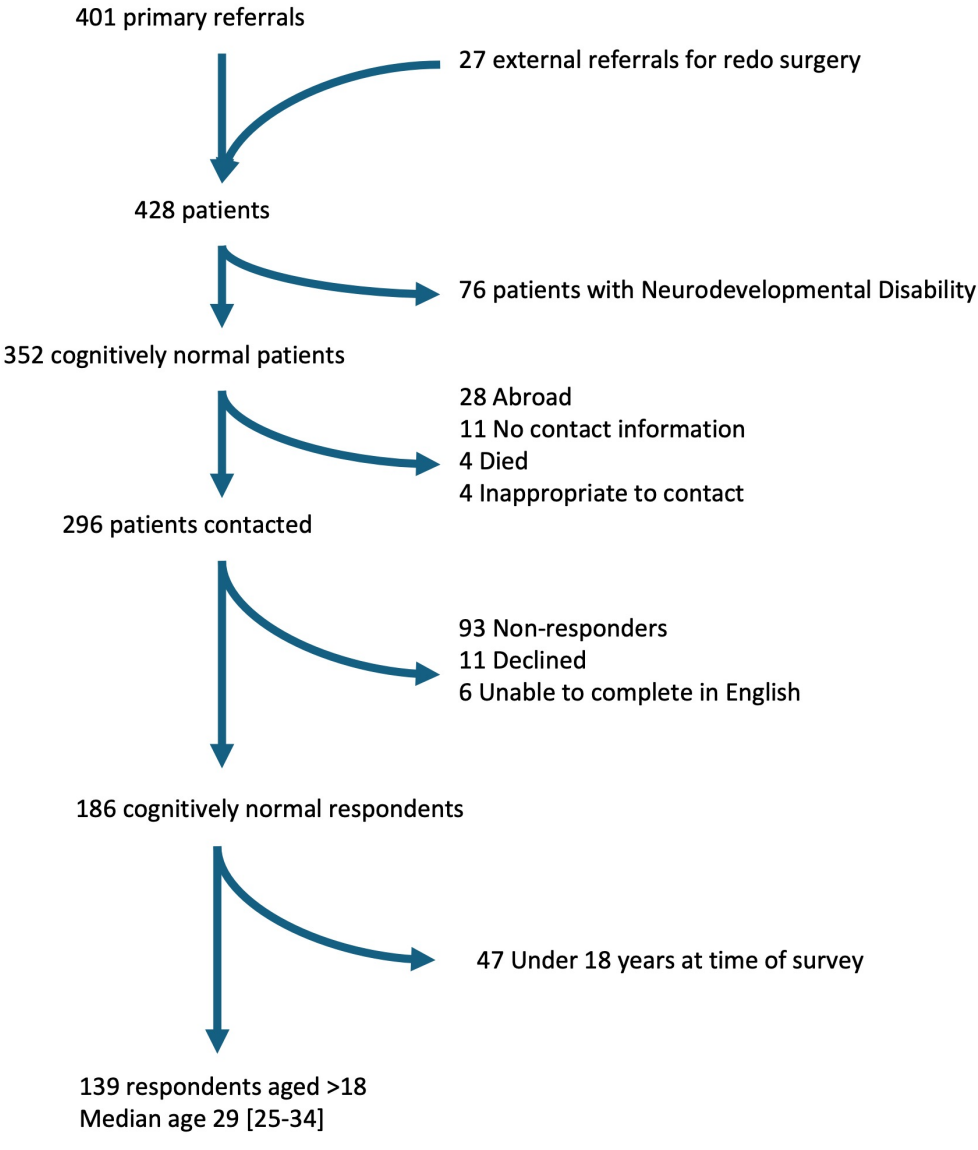
8. Female Sexual Quality of Life Questionnaire

For each question, please select the most appropriate answer:

	completely agree	**moderately agree**	**slightly agree**	**slightly disagree**	**moderately disagree**	**completely disagree**
1. When I think about my sex life, it is an overall enjoyable part of my life	☐	☐	☐	☐	☐	☐
2. When I think about my sexual life, I feel frustrated	☐	☐	☐	☐	☐	☐
3. When I think about my sexual life, I feel depressed	☐	☐	☐	☐	☐	☐
4. When I think about my sexual life, I feel like less of a woman	☐	☐	☐	☐	☐	☐
5. When I think about my sexual life, I feel good about myself	☐	☐	☐	☐	☐	☐
6. I have lost confidence in myself as a sexual partner	☐	☐	☐	☐	☐	☐
7. When I think about my sexual life, I feel anxious	☐	☐	☐	☐	☐	☐
8. When I think about my sexual life, I feel angry	☐	☐	☐	☐	☐	☐
9. When I think about my sex life, I feel close to my partner	☐	☐	☐	☐	☐	☐
10. I worry about the future of my sexual life	☐	☐	☐	☐	☐	☐
11. I have lost pleasure in sexual activity	☐	☐	☐	☐	☐	☐
12. When I think about my sexual life, I am embarrassed	☐	☐	☐	☐	☐	☐
13. When I think about my sexual life, I feel I can talk to my partner about sexual matters	☐	☐	☐	☐	☐	☐
14. I try to avoid sexual activity	☐	☐	☐	☐	☐	☐
15. When I think about my sexual life, I feel guilty	☐	☐	☐	☐	☐	☐
16. When I think about my sexual life, I worry that my partner feels hurt or rejected	☐	☐	☐	☐	☐	☐
17.When I think about my sexual life, I feel like I have lost something	☐	☐	☐	☐	☐	☐
18. When I think about my sexual life, I am satisfied with the frequency of sexual activity.	☐	☐	☐	☐	☐	☐

Is there anything further you would wish to tell us about?

That is the end of the questionnaire, thank you for taking part.


401 primary referrals
27 external referrals for redo surgery
428 patients
76 patients with Neurodevelopmental Disability
352 cognitively normal patients
28 Abroad
11 No contact information
4 Died
4 Inappropriate to contact
296 patients contacted
93 Non-responders
11 Declined
6 Unable to complete in English
186 cognitively normal respondents
47 Under 18 years at time of survey
139 respondents aged >18
Median age 29 [25-34]