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Transitional Care for Patients with Congenital Colorectal Diseases: An EUPSA Network Office, ERNICA, and eUROGEN Joint Venture☆

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ABSTRACT

Background: Transition of care (TOC; from childhood into adulthood) of patients with anorectal malformations (ARM) and Hirschsprung disease (HD) ensures continuation of care for these patients. The aim of this international study was to assess the current status of TOC and adult care (AC) programs for patients with ARM and HD.
Methods: A survey was developed by members of EUPSA, ERN eUROGEN, and ERNICA, including patient representatives (ePAGs), comprising of four domains: general information, general questions about transition to adulthood, and disease-specific questions regarding TOC and AC programs. Recruitment of centres was done by the ERNs and EUPSA, using mailing lists and social media accounts. Only descriptive statistics were reported.
Results: In total, 82 centres from 21 different countries entered the survey. Approximately half of them were ERN network members. Seventy-two centres (87.8%) had a self-reported area of expertise for both ARM and HD. Specific TOC programs were installed in 44% of the centres and AC programs in 31% of these centres. When comparing centres, wide variation was observed in the content of the programs.
Conclusion: Despite the awareness of the importance of TOC and AC programs, these programs were installed in less than 50% of the participating centres. Various transition and AC programs were applied, with considerable heterogeneity in implementation, content and responsible caregivers involved.

Abbreviations: AC, adult care; ARM, anorectal malformation; HD, Hirschsprung disease; TOC, transition of care.

☆ Members of the Transition of Care study group are co-authors of this study and are listed under the heading Collaborators.

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Sharing best practice examples and taking into account local and National Health Care Programs might lead to a better continuation of care in the future.
Level of Evidence: III.

1. Introduction

Anorectal malformations (ARM) and Hirschsprung's disease (HD) are rare congenital colorectal diseases and occur in approximately 1 of 5000 live born neonates annually [1,2]. Both ARM and HD require treatment and follow-up in a tertiary setting (expertise centre) due to the rare nature, complexity and multidisciplinary character of both diseases. Despite the surgical correction in childhood, almost one third of all patients develop disease-specific disorders such as constipation and/or soiling, urinary problems, and sexual development disorders in the long-term [3–6]. Furthermore, associated anomalies, both rare and common (e.g., spinal cord, urogenital tract), can be present in ARM patients, which potentially worsening the fecal, urinary, or sexual outcomes [7,8]. Concerning HD, these disease-specific problems often occur due to iatrogenic injury and/or complications of the surgical reconstruction [8]. In some cases, intensive and regular follow-up is important in adulthood, as both conditions are associated with disease-specific problems in childhood and young adulthood, which can affect health related-quality of life (hr-QOL) and psychosocial functioning [8–11].

In current practice, most studies are performed to investigate short or medium-term postoperative outcomes after pediatric surgical intervention [12]. However, problems in patients with ARM or HD might develop years after surgery (e.g., additional anatomical anomalies, constipation, incontinence), or only show medical and psychological problems occurring at later age (e.g., menstrual problems in patients with uterus didelphys, gait disorders in patients with tethered spinal cord) and therefore require long-term follow-up [10]. Some centres have implemented standardized follow-up during childhood (pediatric care programs), but follow-up is not continued when a patient becomes a young adult (i.e. transition of care (TOC) program and/or adult care (AC) program) [13]. Propositions for uniform TOC for patients with ARM or HD entering (young) adulthood have been suggested, but nonetheless, no standardization has yet been reached [14,15].

Smooth transition to AC and guidance of disease-specific problems is thought to be an important aspect of quality of life, for both patients with ARM and/or HD and their parents or caregivers, as these programs might ensure continuous care availability for these patients in order to early identify disease-specific problems [16]. The importance of such programs has been acknowledged in the past by medical societies, networks of multidisciplinary experts, and patient advocacy groups, also addressing the importance of psychosexual well-being in adolescence [14,17,18]. However, only limited information is currently available about the actual implementation of such programs and their content. In order to create a template for TOC and AC programs that can be applied in the follow-up for ARM and HD patients, more clarity in current transition of care practices should be available. Therefore, the aim of this international survey study was to assess the current status of TOC and AC programs for patients with congenital colorectal diseases in multiple expert centres in different countries.

2. Methods

2.1. Study design

An international survey study was conducted to assess the current status of TOC and AC for patients with congenital colorectal diseases (ARM and HD). The study was overseen by an international steering group that developed the study protocol (DA/IB/CC/JD/ND/WF/JF/CG/CM/NS/GS/TS/CV/RW/PDC/RG). This study was endorsed by the European Pediatric Surgeons' Association (EUPSA, which assisted in the recruitment of participating hospitals through the EUPSA Network Office), ERN for rare urogenital diseases and complex conditions in both children and adults (ERN eUROGEN), and ERN on rare Inherited and Congenital Anomalies (ERNICA), who both also assisted in the recruitment of participating centres. Expert centres of eUROGEN and ERNICA went through an assessment by the ethical committee and by their own national authorities.

2.2. Survey

Through multiple online meetings, the TOC questionnaire (Supplementary Material S1) was developed by a team of medical specialists (e.g., pediatric surgeons, pediatric urologists, and psychologists), and patient representatives from European Patient Advocacy Groups (ePAGs), from different centres affiliated to EUPSA, ERN eUROGEN, and/or ERN ERNICA. The TOC questionnaire comprised of four main domains. In the first domain, general information regarding the centre was collected (e.g., location, type of centre, presence of ARM and/or HD expertise), whereas the second domain comprised of general questions on TOC pathways to adulthood. The third and fourth domains were more specifically for congenital colorectal diseases (i.e. ARM and HD), and comprised of disease-specific questions regarding transition to adulthood (domain 3), and AC programs (domain 4).

2.3. Definitions

TOC was defined as stated in the ARM-Net Consortium paper: "*Purposeful planned movement of adolescents and young adults with chronic physical and medical conditions from child-centred to adult-oriented health care systems*". In addition, AC was defined as: "*The care for adult patients (>18 years old) with a past medical history of congenital malformations (anorectal malformation (ARM), Hirschsprung disease (HD)*" [14].

2.4. Participants

Recruitment of potential participants was done by the EUPSA Network Office, ERN eUROGEN, and ERNICA dissemination channels using mailing lists and social media accounts. Centres eligible for inclusion were any centre with self-proclaimed expertise for ARM and/or HD, independent from ERN membership or affiliations. Centres without area of expertise for ARM or HD were excluded from analysis. If potential participants responded with interest in participation, an invitational email was sent by the coordinating

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researchers (CdB and RG) asking them to register their centre. Only one questionnaire could be filled out per centre. If multiple potential participants from one centre expressed interest, they were asked to choose one representative to fill out the questionnaire. After the first invitation, monthly reminders for participation and completion of the survey were sent up until 6 months. After registration of a centre, the online survey was sent through Castor EDC (Electronic Data Capture) software [19]. In case participants were unable to fill out the survey through Castor EDC, a PDF version of the survey was sent through e-mail. The survey closed on October 1st, 2022.

2.5. Statistical analysis

Data was extracted from Castor EDC, and IBM SPSS Statistics for Windows, Version 28 (IBM Corp., Armonk, N.Y., USA) was used to perform statistical analysis. Descriptive statistics were used for analysis of baseline characteristics and the survey questions. These were reported as proportions and percentages for binary or categorical variables, and as mean with standard deviation (SD) or as median with interquartile range (IQR) for continuous variables as appropriate. To test differences in categorical data, the Chi-square test was used. Univariable logistic regression analyses were performed to investigate relations between the number of new patients with ARM and HD, treated yearly, and the presence of a TOC and AC program. A p-value of <0.05 was considered statistically significant. In case centres did not fully complete the survey, available entered data was used for analysis, and missing data were described. After initial analysis, an online meeting was performed with the steering group to discuss the results and explore further research areas.

3. Results

3.1. Domain 1: general characteristics of participating centres

The TOC questionnaire was distributed through EUPSA, ERN eUROGEN and ERNICA. Ninety-seven centres answered to be interested in participating to the survey, of which 15 (academic (n = 8), pediatric (n = 5), and general hospitals (n = 2)) did not fill out the survey for unknown reason. In total, 82 centres from 21 different countries (Table S2) entered the survey, of which 77 completed it fully, and 5 partially. Participating centres were located in European Union (EU) (n = 61, 74.4%) and non-EU (n = 21, 25.6%) countries. Regarding centres located in an EU country, 37 centres (60.7%) were members of an ERN network, whereas 24 centres (39.3%) were not. In total, 35 centres (42.7%) were academic hospitals with a pediatric department, 24 centres (29.3%) were a free standing children's hospital, 13 centres (15.9%) a combination of an academic hospital with a pediatric department and specific children's hospital organization, and 10 centres (12.1%) were general hospitals with a pediatric department. Furthermore, 76 centres (92.7%) declared ARM and/or HD expertise; both ARM and HD (n = 72), solely ARM (n = 3), and solely HD (n = 1). Six centres reported a lack of expertise for ARM or HD, and these were therefore excluded from further analysis. An overview of baseline characteristics can be found in Table 1.

3.2. Domain 2: general questions on transition of care and adult care programs from expert centres (n = 76)

Only 38 centres (50.0%) reported an established TOC and/or AC program, of which 23 provided both TOC and AC, 13 solely TOC, and 2 solely AC. An overview of the number of patients treated in the included centres can be found in Table 2. A relation was identified between centres treating >10 new patients with HD yearly, compared to centres treating <5 new patients, regarding the existence of TOC and AC programs (OR 4.2, 95% CI 1.14–15.3, p = 0.032, and OR 5.0, 95% CI 1.16–21.5, p = 0.031, for TOC and AC respectively). However, no relations could be demonstrated between the existence of TOC and AC programs and the number of new patients with ARM treated in the centres yearly. In total, 45 participants (59.2%) deemed a transition clinic with a multidisciplinary team involving 1 or more appointments (team composed of both pediatric and AC providers), in combination with a transition program

Table 1
Baseline characteristics.

Country	Survey fully completed n (%)	ERN member n (%)	Area of expertise ARM and/or HD n (%)
EU			
Austria, n = 3	3 (100.0)	2 (66.7)	3 (100.0)
Belgium, n = 3	3 (100.0)	2 (66.7)	3 (100.0)
Denmark, n = 1	1 (100.0)	1 (100.0)	1 (100.0)
Finland, n = 1	1 (100.0)	1 (100.0)	1 (100.0)
France, n = 10	9 (90.0)	7 (70.0)	9 (90.0)
Germany, n = 11	10 (90.9)	6 (54.5)	9 (81.8)
Italy, n = 12	12 (100.0)	4 (33.3)	11 (91.7)
Latvia, n = 1	1 (100.0)	1 (100.0)	1 (100.0)
Lithuania, n = 1	1 (100.0)	1 (100.0)	1 (100.0)
Poland, n = 2	2 (100.0)	2 (100.0)	2 (100.0)
Spain, n = 11	7 (63.6)	3 (27.3)	9 (81.8)
Sweden, n = 3	3 (100.0)	3 (100.0)	2 (66.7)
The Netherlands, n = 4	4 (100.0)	4 (100.0)	4 (100.0)
Total, n = 61 (74.4%)	57 (93.4)	37 (60.7)	56 (91.8)
Non-EU			
Canada, n = 1	1 (100.0)	NA	1 (100.0)
Israel, n = 1	1 (100.0)	NA	1 (100.0)
North Macedonia, n = 1	1 (100.0)	NA	1 (100.0)
Serbia, n = 2	2 (100.0)	NA	2 (100.0)
South Africa, n = 1	1 (100.0)	NA	1 (100.0)
Switzerland, n = 1	1 (100.0)	NA	1 (100.0)
Turkey, n = 9	8 (88.9)	NA	8 (88.9)
United Kingdom, n = 5	5 (100.0)	NA	5 (100.0)
Total, n = 21 (25.6%)	20 (90.7)	NA	20 (90.7)

EU= European Union. ERN= European Reference Network. ARM = anorectal malformation. HD= Hirschsprung disease. n = number.

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Table 2
Number of ARM and/or HD patients treated in included centres.

	New patients n	n of centres	Yearly patients n	n of centres
ARM	<5	12 (15.8)	<25	26 (34.2)
	5 to 10	31 (40.8)	25 to 100	34 (44.7)
	>10	32 (42.1)	>100	12 (15.8)
Missing		*1 (1.3)*		*4 (5.3)*
HD	<5	18 (23.7)	<25	29 (38.2)
	5 to 10	31 (40.8)	25 to 100	33 (43.4)
	>10	26 (34.2)	>100	9 (11.8)
Missing		*1 (1.3)*		*5 (6.6)*

ARM = anorectal malformation. HD= Hirschsprung disease. n = number.

with a facilitator (a facilitator is a physician or nurse or an administrative officer organizing the appointments) the best transition model for patient care in general. The adult surgeon was most often the preferred caregiver for reference when ARM and/or HD patients enter adulthood (71.0%, and 61.8%, respectively) (Table 3).

The pediatric surgeons (n = 61) were most often deemed responsible for TOC in ARM and/or HD patients. Additionally, adult gastrointestinal surgeons (n = 26), and pediatric gastroenterologists (n = 21) were mentioned as responsible caregivers for TOC in ARM and/or HD patients. Regarding which specialist facilitate TOC for ARM and/or HD patients, a large variety of answers was provided, including pediatric and adult (gastrointestinal) surgeons (n = 72, n = 65, respectively), adult gastroenterologists (n = 54), adult and pediatric urologists (n = 58, n = 47, respectively), and adult and pediatric gynecologists (n = 54, n = 32, respectively). Other specialists were also reported to be part of this process, like a clinical nurse specialist (n = 52), psychologist (n = 46), dietician (n = 36), sexologist (n = 31) and physiotherapist (n = 28) (Table S3A (EU versus non-EU) and S3B (academic versus non-academic)).

3.3. Patients in transition of care programs in expert centres (n = 76)

In 20 centres (26.3%), no patients were entered into a TOC program; whereas all pediatric ARM and/or HD patients were entered into the TOC program in 21 centres (27.6%), and 27 centres (35.5%) selected patients based on the presence of active problems. Data was missing for 8 centres (10.5%).

3.4. How patients that are not willing to participate in transition of care programs are handled in expert centres (n = 76)

If patients were not willing to participate in a TOC program, or no transition program was in place, patients were provided with an extensive letter of discharge including contact details in case of problems, with the possibility to contact the centre directly in case of problems (n = 35), consistently invited on yearly basis (n = 14), discharged and referred back to their general practitioner/coordinating physician (n = 15), or other strategies were applied (n = 6). In 6 centres (7.9%), data was missing on their approach towards these patients.

3.5. Domain 3: disease-specific: transition of care (general)

Only 36 centres (47.4%) reported to have disease-specific TOC programs in place (EU n = 31 (86.1%) versus non-EU n = 5 (13.9%), p = 0.020. Academic hospital n = 20 (55.6%) versus non-academic n = 16 (44.4%), p = 0.864), of which 32 centres had TOC programs for ARM and HD, 3 centres solely for HD, and 1 solely for ARM. Continuity of these programs was warranted by hospital policy in 22 centres (61.1%), whereas it was not or unknown in 6 centres, respectively. In 40 centres (52.6%), no disease-specific TOC programs for ARM and/or HD were in place.

3.6. Disease-specific: transition of care program characteristics (n = 36)

In 22 (academic (n = 13); non-academic (n = 9)) of 36 centres (61.1%) with a TOC program, a protocol was available describing the TOC program for children with ARM and/or HD. These protocols were available for both ARM and HD (n = 19), only for ARM (n = 2), or only for HD (n = 1). TOC was well structured in 10 centres (27.8%). Care pathways, frequency of visits and standardized follow-up were defined internally (n = 9), or with external adult specialists (general hospitals) (n = 1). One centre did not fill out this question. In 11 centres, TOC was not structured, and patients had appointments at both pediatric and adult outpatient clinics (n = 8), or at solely adult outpatient clinics (n = 3). Parents (n = 1), patients support groups (n = 5), or both (n = 6) were involved in the transition of care design in 12 centres. Furthermore, in 12 centres (33.3%) data was entered in an (inter)national registry. Data on TOC

Table 3
An overview of reported preferred caregivers for reference of adult patients with ARM and/or HD according to EU and non-EU countries.

ARM			
EU		Non-EU	
Preferred caregiver	n (%)	Preferred caregiver	n (%)
Adult surgeon	39 (51.3)	Adult surgeon	15 (19.7)
Pediatric surgeon	28 (36.8)	Pediatric surgeon	10 (13.2)
Adult gastroenterologist	24 (31.6)	Adult gastroenterologist	10 (13.2)
Pediatric gastroenterologist	7 (9.2)	Pediatric gastroenterologist	4 (5.3)
Other	7 (9.2)		
HD			
EU		Non-EU	
Preferred caregiver	n (%)	Preferred caregiver	n (%)
Adult surgeon	33 (43.4)	Adult surgeon	14 (18.4)
Pediatric surgeon	24 (31.6)	Pediatric surgeon	12 (15.8)
Adult gastroenterologist	33 (43.4)	Adult gastroenterologist	10 (13.2)
Pediatric gastroenterologist	6 (7.8)	Pediatric gastroenterologist	4 (5.3)
Other	1 (1.3)		

EU= European Union. ARM = anorectal malformation. HD= Hirschsprung disease. n = number.

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structure, the involvement of parents and/or patient support groups, and data registry was missing for 15 centres (41.7%).

Commencement of TOC ranged from ≤14 to ≥18 years, with most centres (n = 8, 22.2%) starting TOC at 14 or 15 years of age. In both academic and non-academic centres, patients were most often followed up once a year (n = 6, 16.7%, and n = 5, 13.9%, respectively), but differences in follow-up timings were described (e.g., once every 2 or 5 years, only when problems arise). TOC of ARM patients was most often to adult gastrointestinal surgeons (n = 13), pediatric surgeons (n = 9), adult gynecologists and urologists, and clinical nurse specialists (n = 7, respectively), whereas TOC of HD patients was most often to adult gastrointestinal surgeons (n = 13), adult gastroenterologists (n = 10), and pediatric surgeons (n = 8).

3.7. Domain 4: disease-specific: adult care (general)

Only 25 centres (32.9%) reported the presence of AC programs (EU n = 23 (92.0%) versus non-EU n = 2 (8.0%), p = 0.011. Academic hospital n = 16 (64.0%) versus non-academic n = 9 (36.0%), p = 0.361), of which 21 centres had programs for both ARM and HD patients, 3 solely for ARM, and 1 solely for HD. Continuity of these programs was warranted by hospital policy in 20 centres (80.0%), whereas it was not in 1, and unknown in 4 centres. However, no AC programs were available in 44 centres (57.9%), and data was missing in 7 centres (9.2%).

3.8. Disease-specific: adult care program characteristics (n = 25)

Of those centres that reported the existence of an AC program, 7 (28.0%) provided open access for adult patients in the case of disease-specific problems, and non-regular/structured follow-up was present in 7 centres (28.0%). Regular/structured follow-up of all adult patients with a past medical history of ARM/HD was present at predefined times in only 3 centres (12.0%). Data on AC organization was missing for 8 centres (32.0%). AC programs were most often coordinated by adult and pediatric surgeons (n = 20, n = 19, respectively). Parents (n = 3), patients support groups (n = 4), or both (n = 3) were involved in the AC program design in 10 centres, and data was entered in an (inter)national registry in 7 centres (33.3%). Patients in AC programs were most often seen once a year (n = 5, 20.0%), but differences in follow-up timing were described (e.g., once every 2 or 5 years, only when problems arise). Most centres reported less than 10% or between 10 and 49% (n = 4 and n = 6, respectively) of their adult patients with a past medical history of ARM and/or HD visiting the AC program.

4. Discussion

This international survey shows that uniform definitions and structured transitional care are currently missing, and further development of these uniform definitions and templates for TOC and AC programs could be useful for patients with ARM and HD in need. Large variability in implementation, content and involved responsible caregivers was reported between centres that did have transition and/or AC programs in place. Despite increased awareness in the recent years, the majority of centres do not have established a clear or well-defined transition or AC program.

This study showed that only 47% of participating centres (n = 36) had a TOC program in place, and 22 participants (28.9%) reported that TOC was clearly protocolled. Although the importance of TOC programs for patients with ARM and/or HD have been emphasized by previous studies from expert networks (including patient representatives), the majority of the expert centres do not yet have structured TOC and/or AC programs installed [4,10,17,20]. Importance of these programs are illustrated by previously published studies on the long-term outcomes of patients with ARM or HD disease [3,21–24]. Delayed implementation could contribute to the relatively low number of expert centres with TOC and/or AC programs currently in place. Besides current improvement actions, it would also be of interest to repeat this study in the near future to evaluate the progression of implementation of the programs in centres worldwide [15,17].

Another important finding from this study is the fact that large heterogeneity was identified regarding the content or protocol of TOC programs. Different structures were applied with differences in optimal age to start transition, frequency of outpatient visits, the use of parents and/or patient support groups, and timing of follow-up. Furthermore, due to the design of the survey, it was unclear how parents and/or patient support groups were involved in the development of TOC programs in the included centres. This highlights the current lack of uniformity regarding the optimal TOC programs. In current practice, transition programs have been successfully implemented in pediatric patients with congenital heart disease [25,26]. However, in contrast to patients with isolated cardiac anomalies, patients with ARM may have multiple organ systems affected and require multidisciplinary long-term follow-up and transition of care. Previous studies from the ARM-Net consortium by Giuliani et al. (2017), and Violani et al. (2022) already address the importance of TOC programs, including multidisciplinary follow-up for associated problems (e.g., fecal and urinary, gynecological, sexual and reproductive, and psychological), but nonetheless even for ARM patients, no uniformity has yet been reached [14,17]. Even though both ARM and HD are congenital colorectal diseases, their influence and potential long-term sequelae differ, and therefore disease-specific TOC programs should be developed for both conditions, potentially by a Delphi consensus meeting. In addition, it could be discussed what further role the ERN's could fulfil to improve development and eventually implementation of these programs in relation with the patient journeys, specific guidelines, disease-specific registries, national integration programs, as well as training adult specialists regarding these congenital colorectal diseases.

Only 34% of participants (n = 25) reported to have AC programs installed in their centres (general or academic hospital with a pediatric department, n = 16; free standing children's hospitals, n = 9, p = 0.121). Within these programs, a large heterogeneity was again identified. Opinions varied on what is required in AC for patients with a past medical history of ARM and/or HD. In this study, different specialists were reported as the ideal caregiver to provide AC for ARM and/or HD patients, with adult surgeons most often mentioned for both ARM and HD patients. In our opinion, the presence of a multidisciplinary team is of great importance when patients with a past medical history of a congenital colorectal disease enter adulthood, as often the most troublesome symptoms are not from the colorectal disease, but the additional co-morbidities which can emerge and are in need of intensive guidance and follow-up [22]. For example, patients with urological additional anomalies such as a mono-kidney require follow-up from an adult urologist and/or nephrologist; patients with gynecological anomalies and/or obstetrical problems require guidance from a gynecologist; and patients with additional spinal cord anomalies require follow-up from neurologists and/or neurosurgeons [27–30]. The role of a clinical nurse specialist, psychologist and/or psychotherapist, dietitian, and physiotherapist has also been mentioned as potentially needed, directed to age-specific problems, as these may support the patient to better adhere to treatments and cope with the transition process [24,31]. Additionally, in our opinion, the pediatric surgeon should be involved and easy accessible for consultation because of experience with these patients at younger ages. The importance of a multidisciplinary team of caregivers for this

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specific patient group has been mentioned in previous studies [14,15,17,32]. However, to date, no uniform AC program and/or protocol is available that could help to form such a (patient-specific) multidisciplinary team. Furthermore, potentially not all ARM and/or HD patients require intense follow-up as in AC programs, but patients at risk for problems in the long-term should be adequately identified and informed. To achieve this, the inclusion of patients and patient representative groups is of great importance [14,33].

In this study, we found that parents and/or patient support groups were only involved in the design of TOC and/or AC programs in 33% and 40% of the centres, respectively. Nonetheless, this survey only assessed the opinions and the experiences of the representatives of the participating centres (most often pediatric surgeons), and did not involve the opinion of patient representatives and patients and/or parents. In our opinion, conversely, it would be of great interest to involve patients, to investigate and continuously evaluate their needs in transition and AC, as their experience is of great additional value in the development of TOC and AC programs [33]. Moreover, patient reported outcome measures should be used to evaluate the opinion of patients to whom TOC and AC are applicable and/or patients that underwent TOC. Additionally, identification of tailor-made patient specific follow-up may be of value. TOC and/or AC programs should be developed and available for all patients, with well-balanced and informed patient choice as to whether or not they participate in such a program (shared-decision-making) [34]. Ultimately, it is important that patients know they can always turn to an expert centre when in need of further help.

This study should be interpreted considering some strengths and limitations. First, we were able to include a large number of participating centres, located within the EU (n = 61), as well as outside EU countries (n = 21). In addition, we did not exclude centres that were not a member or affiliated to any ERN network. Second, a high percentage of participants completed the survey (94%), adding value and wider applicability to the results. However, based on the results found with this survey, no hard suggestions or a predefinition of what a transitional program should involve and require could be made. For example, it is difficult to compare various TOC programs since we did not have data regarding number of patients enrolled or missing at follow-up. In addition, most centres included were located within Europe. To obtain a stronger study the survey should have been distributed more widely. Finally, although patient organisations were involved in the Delphi rounds regarding the questionnaire, the opinions of parents and/or patients were not considered in this study. However, as stated before, these opinions are of great interest and importance to share the knowledge and improve and innovate the currently provided care.

Future research should be performed to investigate what centres understand and define as TOC or AC programs, and different available protocols should be assessed to identify best practices. In addition, it will be important to assess country-differences, and the available resources per country should be considered, as low-income countries might have different health care structures compared to high-income countries. These steps should facilitate the creation of a uniform, easily applicable TOC and AC program for patients with ARM and HD, based on best practices.

In conclusion, despite the awareness of the importance of TOC and AC programs, these programs were present in less than 50% of the participating centres. Various transition and AC programs were applied, with considerable heterogeneity in implementation, content and responsible caregivers involved. Future studies should further investigate the different transition and AC programs to create a template for these programs for patients with congenital colorectal diseases, and henceforth improve implementation of these programs on a larger scale in order to share knowledge, improve, and innovate the currently provided care.

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Conflict of interest

The authors declare that they have no competing interests.

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Appendix A. Supplementary data

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