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Network Urogenital Diseases (ERN eUROGEN)

ADAPTATION REPORT

ADAPTATION OF THE DUTCH QUALITY STANDARD FOR ANORECTAL MALFORMATIONS

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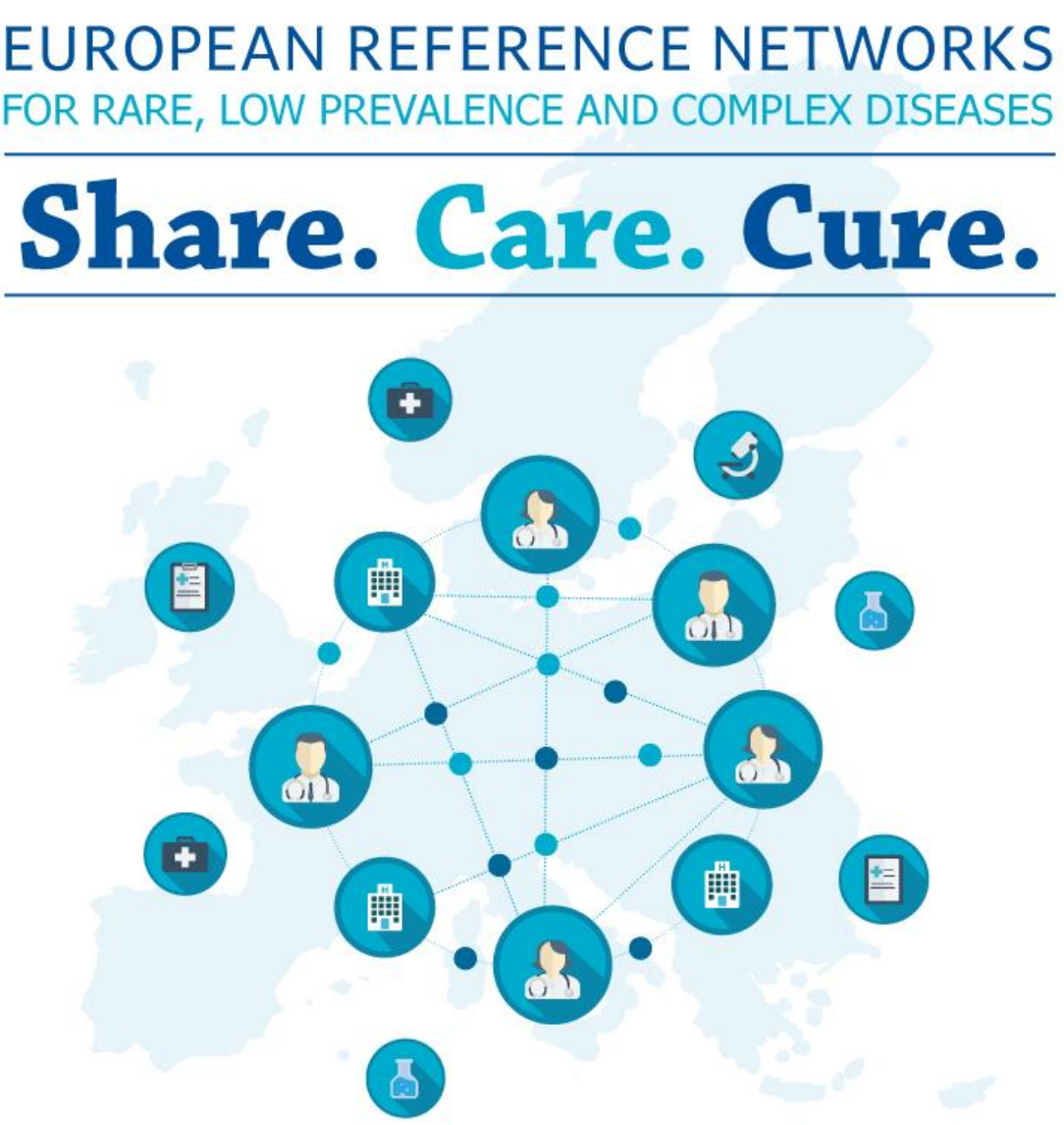

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ABSTRACT

INTRODUCTION

Anorectal malformations (ARM) are a spectrum of malformations and a rare condition. Awareness and knowledge are essential among the healthcare providers involved in the diagnosis and treatment of ARM. The complexity of ARM requires multidisciplinary and tailor-made care with expertise in this low prevalence condition.

OBJECTIVES

To adapt the Dutch Quality Standard recommendations for patients with ARM in order to enhance and protect the quality of care, specifically within the European context.

METHODS

The Adaptation Quality Standard was developed in adherence to the ADAPTE method. A Systematic literature review was conducted using Medline, Embase, and Cochrane databases to identify studies published after 2017. Systematic reviews or original studies were considered for inclusion if they provided evidence that could potentially change the course of care outlined in the Dutch Quality Standard for ARM and if they were focused in one of the following 4 thematic modules: 1) Lifelong follow-up and integrated care, 2) Organisation of care, 3) Collaboration, referral and communication between care providers and 4) Transition care.

RESULTS

After a thorough consensus meeting between ERN eUROGEN and supporting members, a number of 29 recommendations were adapted, 14 were adopted and 8 were developed de novo (new) for the modules 1) Lifelong follow-up and Integrated care, 2) Organisation of care, 3) Collaboration, referral and communication between the care providers and 4) Transition of care for patients with ARM. If applicable, in order to enhance the rigor of development, newly created recommendations were accompanied with a Grade of Recommendation according to the Level of Evidence of the respective linked study/studies. Justifications were reported, describing the areas of consensus, the development process of the recommendations and the discrepancies with the Dutch Quality Standard.

CONCLUSION

Based on current evidence and according to the European context, a set of recommendations was developed for the 1) Lifelong follow-up and Integrated care, 2) Organisation of care, 3) Collaboration, referral and communication between the care providers and 4) Transition of care of patients with ARM. However, further research is required to increase the certainty of evidence in areas of ambiguity and to develop more evidence-based recommendations within these components.

KEYWORDS

Anorectal Malformation, ARM, Quality standard, Adaptation.

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ACRONYMS AND ABBREVIATIONS

ARM	Anorectal Malformation
ERN	European Reference Network
AAWG	Adoption and Adaption Working Group (expert panel)
GP	General Practitioner
HCPs	Healthcare Providers
SP	Supervising Physician
ICP	Individual Care Plan
MDT	Multidisciplinary team
EC	Expertise Center
TC	Treatment Center
CRC	Colorectal Cancer
SCD	Spinal Cord Dysraphism
AGREE II	Appraisal of Guidelines for Research and Evaluation II

1. INTRODUCTION

1.1. GENERAL

Anorectal malformations (ARM) are rare conditions. The incidence of ARM varies between 1:3500 to 1:5000 live births.[1] There is a great need for more awareness and knowledge among all healthcare providers involved in the care of patients with an ARM in order to reduce the sometimes occurring diagnostic delay, improve diagnostics and treatment, and prevent complications and comorbidities in the short and long term as much as possible and improve the quality of life for patients. The care for patients with ARM is complex, where multidisciplinary, tailor-made care by healthcare providers with expertise is essential. An extensive introduction of ARM and associated malformations can be found in the Dutch Quality Standard.[2]

1.2. SCOPE AND OBJECTIVES

The scope of this adaptation report follows the scope of the Dutch Quality Standard. This includes the care pathway from the prenatal detection until lifelong follow-up care and transition care for patients with all types of ARM. A full overview of all types of ARM and associations is to be found in the Dutch Quality Standard.[2] The primary objectives of this adaptation are:

1. To assess currency and validity of the Dutch Quality Standard recommendations to improve and safeguard the quality of care for patients with ARM
2. To assess applicability and acceptability of the Dutch Quality Standard recommendations for ARM to fit the European context.

2. METHODS

2.1. TARGET POPULATION

The target population for this Quality Standard Adaptation is patients with ARM. Additionally, the primary target audience of the Quality Standard Adaptation consists of all members of the professional groups involved in the care of patients with an ARM, such as paediatric surgeons, paediatricians, nurse practitioners, nurses, (paediatric) gastroenterologists, paediatric anaesthesiologists, physiotherapists, dieticians, psychologists, clinical geneticists, nephrologists, general practitioners, (paediatric) urologists, (paediatric) neurosurgeons, (paediatric) gynaecologists, sexologists and (paediatric) orthopaedic surgeons.

2.2. ADAPTE

For the Adaptation of the Quality Standard the ADAPTE method was used.[3] First, the existing Quality Standard was appraised for its methodological quality with the AGREE II tool. To assess if the recommendations were current, or if new literature could update the advises in the Dutch Quality Standard, a literature review was performed. Applicability and acceptability were assessed during meetings between the ERN eUROGEN Adoption and Adaptation Working Group (AAWG) and team members from Qualicura.

Once the assessment has been completed, the AAWG considered the results of the assessment to obtain a conclusion for each recommendation. The existing recommendations from the Dutch Quality Standard were adopted as a whole or modified in order to be adapted. New recommendations emerged based on the new evidence found. In this document, the recommendations have been referred as either adopted, adapted or new. The definitions of adapted, adopted and new recommendations are displayed in Table 1. The decision for each recommendation was reached by consensus based on the methods of a consensus development conference.[4]

Adopted	Recommendation was not changed from the Dutch Quality Standard
Adapted	Recommendation was modified and adapted to the needs of the ERN
New	Recommendation was created from the new evidence found

Table 1. *Definition of adopted, adapted and new recommendations.*

A decision-making algorithm is displayed below to show the process towards a conclusion about the recommendations to create an adopted or adapted guideline (see Figure A).

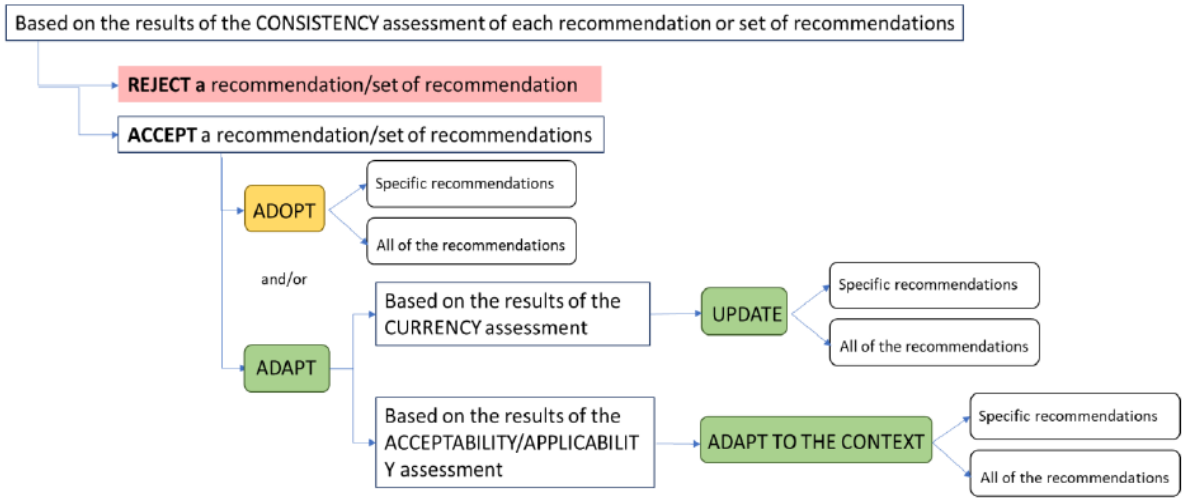

Figure A. *Decision making algorithm for the acceptance of a recommendation or set of recommendations.*

2.3. SEARCH STRATEGY

The literature searches were conducted by a professional information specialist in the following databases, (Ovid/Medline (PubMed), Cochrane CENTRAL, Embase) to identify any new relevant studies published between 2017 and March 2023. Databases were searched using relevant medical subject headings and free-text terms. The search strategy was conducted in the electronic databases in March 2023 and the searches were restricted to retrieve articles in English language. To ensure that the report contains current and relevant evidence, studies published prior to 2017 were excluded. The literature searches yielded a total number of 2248 studies (after removal of duplicates) and were subsequently screened for title and abstract (see Appendix Table 1). The screening was conducted by two methodologists (WI) and (KM). Both screened results based on title and abstract utilising the Systematic review app Rayyan,[5] and identified useful publications according to the inclusion/exclusion criteria displayed in Table 2.

Literature Selection Criteria

Inclusion
• Research covering Anorectal Malformation, Congenital AND (lifelong follow-up), (organisation of care), (collaboration care), (transition care) within the scope of clinical questions in the original quality statements
• Evidence that possibly changes the course of care as described in the Quality Standard
• Systematic review or study reporting original data.
• English language
Exclusion
• Studies with < 10 patients
• Acquired malformations/fistulas (i.e., Crohn's related)
• Published before 2017

Table 2. *Inclusion and exclusion criteria of publications on Diagnosis and Treatment of children with ARM*

After completing the screening of all titles and abstracts decision discrepancies were discussed. After the first screening round, two methodologists (WI, KM) and three pediatric surgeons (JG, ML, IB) screened the included publications and categorised them in four topics: 1) Lifelong follow-up and Integrated care, 2) Organisation of care, 3) Collaboration, referral and communication between care providers and 4) Transition care. Any results that did not match with one of these preselected topics were excluded in the second screening round. Seven new studies for the lifelong follow-up module, two new studies for the organisation of care, one new study for the transition care module and no new studies for the collaboration module were identified, adding value in the adaptation of the Quality Standard. A PRISMA flow diagram with the literature selection is available in Appendix Figure A.

3. RESULTS

3.1. GUIDELINE METHODOLOGIC REVIEW

3.1.1 APPRAISAL OF THE DUTCH QUALITY STANDARD FOR ANORECTAL MALFORMATIONS

In order to make a decision about the adoption or adaptation of the Dutch "Quality Standard for Anorectal Malformations", assessment of its quality and reporting was sought.

The Appraisal of Guidelines for Research and Evaluation (AGREE II) instrument was used to independently appraise the Dutch Quality Standard on anorectal malformations.[6] Four reviewers carried out the appraisal (two reviewers from the Aragon Institute of Health Sciences (IACS) and two from the ERN eUROGEN expert panel).

Domain scores were calculated by summing up all the scores of the individual items in a domain and by scaling the total as a percentage of the maximum possible score for that domain. The overall standard

domain score given by the reviewers was 38%. For additional information about the appraisal of the Dutch Quality Standard and the domain scores see Appendix: Dutch Quality Standard Appraisal.

The AAWG and methodologists decided that the recommendations from the Dutch Quality Standard could be adapted, however some improvements were desirable based on the consistency assessment.

3.1.2 Levels of evidence and Grades of recommendation

To improve the rigor of development the AAWG and methodologists suggested to accompany all new recommendations with a Grade of recommendation if applicable. Therefore, the working group chose to use an appraisal method that can be applied on study level. The possible levels of evidence were I, II, III and IV (OCEBM, 2011) or expert opinion and this was paired with a Grade of recommendation (Adapted from OCEBM, 2009[7]) (see Table 3).

A	Based on consistent level 1 studies
B	Based on consistent level 2 or 3 studies or extrapolations from level 1 studies
C	Based on level 4 studies or extrapolations from level 2 or 3 studies
D	Based on expert opinion **or** inconclusive/inconsistent studies of any level.

Table 3. *Correspondence between grades of recommendations and study level of evidence.*

To improve the reporting of of the adapted recommendations with more clarity, transparency and explicitness, the AAWG and methodologists will provide the adaptation report following the RIGHT-Ad@pt preferred reporting items.[8]

3.2. Currency, Acceptability, Applicability and Clarity

3.2.1 Lifelong follow-up and integrated care

Key question 1

What is the optimal follow-up for people with an anorectal malformation (ARM) in an expertise center / pediatric surgery center?

The key question consists of the following sub-questions:

a) What is the (minimum) frequency of follow-up appointments?
b) Which points of attention / questions should be addressed during a follow-up appointment at different ages?
c) Which actions / investigations must be performed during a follow-up appointment at different ages?

Recommendations for the lifelong follow-up and integrated care

Seven (7) new studies were included and considered by the panel for the lifelong follow-up and integrated care module. Twelve (12) recommendations were adapted, none were adopted and three (3) were developed de novo (new). All recommendations are reported in Table 4. A summary of all newly included studies for this sub-question is available in Appendix Table 2.

LIFELONG FOLLOW-UP AND INTEGRATED CARE	Grade of Recommendation

1	It is recommended to see all children and young adults in the expertise center for ARM / pediatric surgery center at crucial times of age and an appointment/visit should be offered in an organised follow-up programme.. **[adapted]**	-
2	After the ARM has been surgically repaired, the child is guided through a structured lifelong follow-up program. See: Table 3.1. in the Dutch Quality Standard[2] **[adapted]**	-
3	The organised follow-up program should be carried out by a pediatric surgeon (supervising physician). If needed the patient should be offered a multidisciplinary team. **[adapted]**	-
4	Throughout the follow-up, the function of the digestive tract and the urogenital tract will be monitored. **[adapted]**	B[9]
5	It is recommended to monitor quality of life during follow-up with standardised general or disease-specific questionnaires. **[adapted]**	D[10]
6	Scar morbidity and symptoms should be addressed during follow-up. If necessary, a plastic surgeon can be consulted. **[new]**	C[11]
7	Patients should be given access to information on quality of life and mental health, for example through: https://eurogen-ern.eu/ **[new]**	D
8	During puberty, adolescence and adulthood, attention is paid to the possible psychosocial and sexual problems: information material with regard to sexual function/issues should be provided, for example: https://sexuality-arm-hd.com/ , or through patient support groups or experienced professionals. **[adapted]**	C[12]
9	The parents of the child with ARM receive information immediately after birth but also during follow-up. Referral to a patient organisation is recommended as these organisations can provide additional information and support that can contribute to improving quality of life. **[adapted]**	D[13]
10	If patients associations are available, refer patients with ARM to patient associations for support and education about self-management of the malformation. **[adapted]**	C[14]
11	An experienced nurse should be involved in the post-operative care. **[adapted]**	-
12	Nutrition growth and neuromuscular development are important considerations during all of the follow-up. **[adapted]**	-
13	(Neuro) psychological follow-up is recommended on indication in children with ARM (poor academic performance or potential shorter attention span, possibly due to physical problems). **[adapted]**	C[15]
14	Gynaecological follow-up is recommended on indication in females with ARM (especially in females with a cloacal malformation or with another known	-

	malformation of the gynaecological tract), to detect abnormalities / clinical symptoms on time, or to follow up on known anomalies. In case of complaints, an ultrasound of the lower abdomen must be performed during adolescence around the time of the first menarche (on average more than a year after the thelarche / breast development). This is to evaluate whether blood is accumulating in the vagina or uterus because it cannot be drained. **[adapted]**	
15	Urodynamic studies should be performed in every patient with spinal cord dysraphism (SCD) as well as in every child with high suspicion of a neurogenic bladder to estimate the risk for the upper urinary tract and to evaluate the function of the detrusor and the sphincter. **[new]**	D[16]

Table 4. *Recommendations for lifelong follow-up and integrated care for patients with ARM.*

Justification the lifelong follow-up and integrated care

The AAWG considered the favourable benefit-risk balance, the acceptability and applicability in order to adapt or to form new recommendations. Recommendations have been made in favour of the accessibility of the ARM patients in an expertise center for ARM and of a structured lifelong follow-up program involving a multidisciplinary team.

It was recommended by the panel group that all children and young adults with ARM should be examined in the expertise center for ARM / pediatric surgery center at crucial times of age (i.e. entering kindergarten, school, puberty, transition to adulthood) and an appointment/visit should be offered in an organised follow-up programme.

The group referred to studies from van der Steeg et. al. and Hartman et al., highlighting the importance of monitoring both the digestive and urogenital tract functions during follow-up, along with measuring the quality of life using standardised general or disease-specific questionnaires.[9, 10] According to a study by Tofft et al.,[11] the healthcare providers are recommended to track and report the scar morbidity and symptoms during post-treatment follow-up. The Patient and Observer Scar Assessment Scale (POSAS) is a suggested tool for this purpose.[17] A plastic surgeon may be consulted if necessary.

A discussion was made on the quality of life and mental health of the ARM patients and the access they have to this information. The group indicated that the respective recommendation should be part of the lifelong follow-up module instead of the treatment module, as per the Dutch Quality Standard.[2] It was considered crucial that the involved healthcare professionals should refer the patients with ARM and their parents to patient organisations, while providing them with the required guidance in order to promote their awareness and self-advocacy. More Information on patients and parents organisations can be found at eurogen-ern.eu/who-is-involved/patients. The group concluded that at the time of diagnosis support should also be offered to parents.[14]

The group unanimously agreed to incorporate and adapt the recommendations (10, 11) which were initially found in the Dutch Quality Standard under the module *'Treatment for a child with ARM'*, in this section aiming at forming a more comprehensive module with less overlap of recommendations in the chapters.[2]

A recommendation was formed regarding the significance of nutrition, growth and neuromuscular development during all of the follow-up. The original quality standard references the STRONGkids score list (screening instrument for identifying the risk of malnutrition during admission to a hospital);[2] however, the group determined that this tool may not be suitable or feasible in every EU nation and therefore this specific tool was removed from the recommendation.

The AAWG considered a study by Mert et. al.,[18] that describes the possible role of subjective scoring systems for bowel function such as Holschneider's Questionnaire and Rintala's Questionnaire. The panel considered the study but is not convinced it is of additional value as these questionnaires are not validated for the purpose of bowel follow up. The panel agreed that probably a majority of patients has psychosocial issues due to incontinence, something that is not picked up on by these questionnaires. Manometry on the other hand, can give additional information for research purposes but can add

significant anxiety in patients[19]. The panel therefore agrees that the possible benefits for clinical decision do not outweigh the burden for patients which reflects in the fact that manometry is not commonly practised amongst panel members.

Moreover, the AAWG discussed the examinations that should be carried out throughout the lifelong follow-up of a patient with ARM, whereas some being recommended after certain indications (i.e. examinations for (neuro) psychological and gynaecological follow-up). According to a case-control cohort study by Miyake et al.,[15] it is suggested that children with ARM should undergo (neuro) psychological follow-up as warranted, such as if they experience challenges in academics or display a shorter attention span, potentially stemming from physical issues. The AAWG recommended a gynaecological follow-up in females with ARM under indication, respectively; particularly those with a cloacal malformation or other known gynaecological issues and therefore adapted most recommendations on this from the Dutch Quality Standard [2]. This is important to detect abnormalities and address any clinical symptoms in a timely manner. If any complaints arise during adolescence, an ultrasound of the lower abdomen should be done around the time of menarche. Following this, an abdominal ultrasound should also be performed to assess for any blood accumulation in the vagina or uterus as a result of drainage issues due to anatomical malformations. The latter recommendation was originally found in the diagnosis module of the Dutch Quality Standard.[2] However, upon further evaluation, the panel determined that it would be better suited for inclusion in the lifelong follow-up and Integrated care module.

Additionally, the group referring to the EAU guidelines on urological infections, recommended urodynamic assessments for all patients with spinal cord dysraphism and children who show signs of a neurogenic bladder.[16] These tests can determine the potential risk to the upper urinary tract and assess the function of the detrusor and sphincter muscles. Certain ARM cases may experience urological complications, such as epididymo-orchitis (EO), or erectile dysfunction in males. The group addressed that EO can manifest more severely in ARM patients and thus require special attention.[20]

3.2.2 ORGANISATION OF CARE

Key question 2

Which care provider(s) is responsible for good care, coordination, and managing of medical, psychological, and social care for patients with anorectal malformation in the different phases of care?

The key question consists of the following sub-questions:

a) Which healthcare providers are involved in the follow-up of people with anorectal malformations (ARM) in a (pediatric surgical) care center?
b) Which disciplines must be able to be consulted on indication?
c) What is the role of the expertise center, the specialised surgical treatment center, and the treatment team for ARM patients?
d) Who is the supervising physician and what are the tasks of the supervising physician?
e) What measures are taken so that there is guidance / support on a social level (such as possible problems at school, work, insurance, applying for benefits)?
f) What measures are taken so that there is psychological / psychiatric / sexual treatment or guidance, so that attention is paid to the emotional aspect of patients with anorectal malformation?

Recommendations for the organisation of care

For the organisation of care module, two (2) new studies were included. Five (5) recommendations were adapted, four (4) were adopted and one (1) was developed de novo (new). All recommendations are reported in Table 5. A summary of all newly included studies for these sub-questions is available in Appendix Table 3.

ORGANISATION OF CARE		Grade of Recommendation
1	Hospitals treating patients with ARM should have easy access to the following healthcare providers: • Pediatric surgeon	B[21]

	• Pediatrician • Nurse practitioner / case manager pediatric surgery • (pediatric) Anesthesiologist • (pediatric) Cardiologist • Pediatric urologist • (pediatric) Neurologist • (pediatric) Neurosurgeon • Nurse practitioner in urinary incontinence, materials, ICC and stoma care • Dietitian • General practitioner • Physiotherapist • (pediatric) Gastroenterologist • (pediatric) nephrologist • Clinical geneticist • Social worker • Neonatologist • (pediatric/youth) Psychologist • (pediatric) Radiologist • Colorectal Surgeon • (pediatric) Orthopedic Surgeon • (pediatric) Cardiac surgeon • (pediatric) Gynaecologist • High risk pregnancy specialist • Urologist / andrologist • Sexologist or other provider that can help with sexual issues. • School counselor A MDT has to be tailored to the needs of the patients and local care agreements. **[adapted]**	
2	It is recommended that expertise centers for ARM should manage ≥ 10 (reconstruction) cases per year / center with multidisciplinary teams. **[new]**	B[22]
3	The examinations, decisions, and conversations with the parents of children with ARM take place in a pediatric surgical center with expertise in ARM. **[adapted]**	-
4	Within the expertise center / pediatric surgery center, attention should also be paid to the transition of care for adolescents. **[adopted]**	-
5	In cases of shared care, pediatric surgical center with expertise in ARM remains responsible for care coordination**.** The specialist center / pediatric surgery team examines which care is needed and whether this can best be offered in the expertise center / pediatric surgery team or in the own region. **[adapted]**	-
6	The expertise center/ pediatric surgery, advises and supports the other healthcare providers in the care chain and is responsible for information provision, guidelines, and evaluation of care. Information exchange, periodic reporting collaboration between expert centers and regional pediatric (surgical) departments are important to ensure accessible care close to home for patients. **[adapted]**	-
7	A supervising physician and a care coordinator (case manager) are appointed for the entire long-term multidisciplinary care of the patient with an ARM. The supervising physician in childhood directs during childhood and the transition phase. **[adopted]**	-

8	The supervising physician is a medical specialist who is aware of the recent scientific developments and treatment methods of ARM, is in charge of the total long-term multidisciplinary care (including follow-up and shared care) and is the point of contact for the (parents of the) patient regarding healthcare questions and for healthcare providers within the multidisciplinary ARM team. **[adapted]**	-
9	The care coordinator (case manager) is the first point of contact for the (parents of the) patient and care providers from outside the multidisciplinary ARM team and / or outside the center. **[adopted]**	-
10	The parents of the child with an ARM are informed who the supervising physician and care coordinator / case manager are and how they can be reached. **[adopted]**	-

Table 5. *Recommendations for organisation of care for ARM.*

Justification for the organisation of care

The AAWG considered the favourable benefit-risk balance, the acceptability and applicability in order to adopt, adapt or to form new recommendations.

The panel agreed that in order for a healthcare center to be recognised as an ARM expert facility, it should offer easy access to a wide range of medical specialties within a multidisciplinary team and perform at least 10 or more ARM reconstructions annually.[21, 22] Although the AAWG pointed out that a number of 10 (reconstruction) cases per year might not be feasible for every expertise center across EU members, this recommendation is made because the panel agrees that centralisation can be a crucial step towards improving the quality of ARM care.

In shared care, the Dutch Quality Standard recommends that the expertise center in ARM, along with the patient's relatives/caregivers, have control over the integrated care chain.[23] However, the AAWG advised that the expertise center with ARM expertise should have the exclusive responsibility for care coordination, by assessing the required care and determining whether it should be provided at the expertise center or in the care recipient's own region.

Furthermore, the group addressed the importance of centralisation, bilateral exchange of information and collaboration in shared care between the ARM expertise center and other Healthcare Providers (HCPs), facilitating also the patient's access in care close to their region.

3.2.3 COLLABORATION, REFERRAL AND COMMUNICATION BETWEEN CARE PROVIDERS

Key questions 3 and 4

3. What measures / steps (issues to be addressed) are being taken to ensure a good exchange / provision of information between patient (family) and care providers, to properly inform the patient and family and to provide good quality care?

4. Which measures / steps must be taken to achieve good cooperation between care providers and referral to care providers (from primary, secondary, tertiary care) who are involved in the care of the patient with ARM, with the aim of providing good quality care?

The key questions consist of the following sub-questions:

a) What measures / steps are being taken to ensure a good exchange of information between care providers within the same institution and with the care providers involved outside the institution?
b) What measures / steps are taken in the case of shared care for the patient (family) to achieve a good exchange of information between the care providers involved?
c) What measures / steps are taken so that the healthcare provider refers the patient with ARM to the right healthcare provider at the right time?

Recommendations for collaboration, referral and communication between care providers

For this module, no new studies were included. Seven (7) recommendations were adapted, three (3) were adopted and one (1) was developed de novo (new). All recommendations are reported in Table 6.

COLLABORATION, REFERRAL AND COMMUNICATION BETWEEN CARE PROVIDERS		Grade of Recommendation
1	The supervising physician (SP) and / or the care coordinator starts providing information to parents of a child with ARM immediately after the diagnosis and continues throughout the care process. **[adopted]**	-
2	The SP and / or the care coordinator communicates in language understandable to the parents, checks whether repetition of the information is necessary and repeats the information if necessary. **[adopted]**	-
3	The SP has the task of providing information, but can also partly delegate this task to another care provider, for example to a junior doctor or- specialised nurse in ARM. However, the SP remains ultimately responsible**. [adapted]**	-
4	The SP ensures that the specialists involved primarily and secondarily within the multidisciplinary team are present at a multidisciplinary meeting. **[adapted]**	-
5	In any case, the SP and / or care coordinator, the pediatric surgeon, the pediatrician and the specialised nurse in ARM and / or nurse practitioner are present during the multidisciplinary consultation. The SP is often a pediatric surgeon**. [adapted]**	-
6	The SP ensures the communication/cooperation (internal and external) between the involved healthcare providers and also between the patients and the parents. **[adapted]**	-
7	The SP informs the general practitioner (GP) and - if involved - the pediatrician in the regional hospital about the ARM condition, including the possible complications and in which situations the SP or the care coordinator of the pediatric surgery center should be contacted. The GP or the pediatrician at the regional hospital contacts the SP of the pediatric surgical center for consultation and referral if necessary, in the following situations: • When ARM-associated complications arise in a child with ARM. • When a child with ARM is admitted to a general hospital. Agreements regarding cooperation and involvement with the child with ARM are recorded between the partners. **[adapted]**	-
8	Within an institution, care providers must effectively coordinate and communicate about care, treatments, and changes in the Individual Care Plan (ICP). **[adapted]**	-
9	Transfer of tasks and responsibilities takes place explicitly. When designing transfer moments, it is important to take into account frequently occurring risks (during transfer) and any specific characteristics of the patient's situation. [**adopted]**	-
10	Children with ARM will increasingly receive 'shared care'. This means that a child is undergoing treatment at the Expertise Center (EC) / surgical treatment center, but also in a hospital or other treatment center (TC) close to home. The local	-

	hospital / local healthcare provider works together with the EC / surgical treatment center. **[adapted]**	
11	The countries with a patient organisation, should organise annual meetings for patients. In case there is no patient organisation the EC should coordinate a yearly meeting. **[new]**	D

Table 6. *Recommendations for collaboration, referral and communication between care providers.*

Justification for the collaboration, referral and communication between care providers

The AAWG considered the favourable benefit-risk balance, the acceptability and applicability in order to adopt, adapt or to form new recommendations. Recommendations have been made in favour of efficient collaboration, referral and communication between care providers involving with patients with ARM.

The group unanimously agreed to incorporate and adopt the recommendations (1, 2, 3), which were initially found in the Dutch Quality Standard under the module *'Communication and information exchange with parents*', in this section aiming at forming a more comprehensive module.[2]

During the panel discussion, emphasis was given in promoting an effective communication/cooperation between the involved healthcare providers and other care providers within the expertise center and outside. The SP is liable for coordinating this collaboration as well as ensuring an efficient communication between the patients and parents.

Furthermore, the AAWG addressed that hospital exchanges of patients with ARM, may imply risks. Therefore, the allocation of the tasks and responsibilities have to be well-structured and coordinated. In order to avoid potential complications, such as health risk, data loss, etc., the frequently occurring risks (during transfer) and any specific characteristics of the patient's situation should be carefully documented.

Additionally, attention was put on the patient support organisations and the importance of meetings with patients and their families. The recommendation was made for patient organisations to host and actively participate in annual meetings targeting ARM patients. The AAWG suggested that in cases where there is no local patient organisation, the ARM Expertise Center should organise an annual meeting for families, so that peer support can be established.

3.2.4 TRANSITION CARE

Key question 5

What measures / steps need to be taken to ensure a smooth transition of children with ARM from child to adult care?

The key question consists of the following sub-questions:

a) Which care provider is responsible for a smooth transition to adult care?
b) At what point will the adult surgeon / gastroenterologist (if necessary) / urologist / gynaecologist be involved in the transition process?

Recommendations for the transition care

For the transition of care module, one (1) new study was included by the panel. Six (6) recommendations were adapted, six (6) were adopted and three (3) were developed de novo (new). All recommendations are reported in Table 7. A summary of all newly included studies for this sub-question is available in Appendix table 4.

TRANSITION CARE		Grade of Recommendation
1	The nurse practitioner (case manager) and pediatric surgeon ensure a smooth transition from childhood to adulthood by preparing young people and their parents. The pediatric team is responsible for this process. If necessary, other specialists will be involved such as the psychologist etc. **[adapted]**	-
2	During the transition process the patient should be involved in all decisions. **[new]**	D
3	Treatment centers involved with the care for ARM patients are encouraged to install a structured transitional program. **[new]**	D
4	A personalised transition plan should be handed out to all ARM patients at the beginning of transitional process. **[new]**	D
5	The transition planning typically starts at the age of 12-14 years but can differ individually. The actual transition to adult care takes place depending on the local regulations for adulthood in healthcare. **[adapted]**	-
6	The care-providing expertise centers / pediatric surgery centers must take their responsibility for the transition of patients with ARM from care through the pediatric specialists to the adult equivalent (surgeon, urologist, etc.). The ideal transition seems to consist of the involvement of both the pediatric specialist and an involved adult specialist, resulting in a collaboration between the two in which easy contact and consultation is possible. **[adopted]**	C[24]
7	During the transition phase, all involved healthcare providers should pay extensive attention to the establishment of a positive transfer to a new doctor in charge of adult care. **[adapted]**	D[25]
8	Good cooperation and communication between the gastroenterologist / surgeon / urologist / gynecologist where the adult patient is being treated, and the pediatric surgeon / pediatric gastroenterologist / pediatric urologist who knows the history, is important. **[adopted]**	-
9	Follow-up for adults is not yet well organised everywhere, and many adolescents do not come to the clinic. The healthcare professionals and patients should be encouraged to provide/attend follow-up appointments/visits within an organised and structured setting. **[adapted]**	-
10	Personal attention, compassion and support are the most important factors that create a familiar environment that results in the desire of patients with ARM to continue to visit the clinic. **[adopted]**	-
11	It is recommended to create a tailored transition plan for each individual patient, considering their condition (s), intelligence and capabilities. **[adopted]**	-
12	In addition to guidance in the field of ARM and the (possible) medical consequences of the condition, there is also attention for: problems that puberty can bring, the future, relationships, sexuality, and fertility. Attention is also paid to the role of the parents during the consultations, as well as the patient's expectations and preferences regarding the transition. **[adopted]**	-
13	In patients with ARM who have questions or concerns about having children, the threshold for referral to preconception care and consultation should be low. **[adapted]**	-

14	Good guidance, a transition coordinator, a transition brochure and combined transition consultations with a child and adult care provider are part of a good protocol for structured preparation for the transition. **[adopted]**	-
15	The patients must have open access to their data , either through an overview provided by their supervising physician or by requesting a copy of their own status. This is to prevent loss of data due to relocation or changes in patient files in the hospital. **[adapted]**	-

Table 7. *Recommendations for transition care for patients with ARM.*

Justification for the transition care

The AAWG considered the favourable benefit-risk balance, the acceptability and applicability in order to adopt, adapt or to form new recommendations. Recommendations have been made in favour of efficient collaboration, referral and communication between care providers involved with patients with ARM.

The panel deliberated the importance of actively involving patients in decision-making throughout the transition period. This shared decision-making approach values the patient's input and not only enhances their overall care but also promotes a sense of ownership and responsibility for their own health.

Furthermore, in order to ensure continuity of care, the panel suggested the implementation of a structured transitional program in treatment centers providing care for ARM patients. By doing so, a structured transitional program enables the development of tailored care plans that consider the individual needs and preferences of each patient.

Due to the complexity of ARM, it is crucial to develop a personalised treatment plan tailored to the specific needs of each patient. Accordingly, the AAWG recommended providing all ARM patients with an individualised transition plan at the start of their transitional process.

The transition planning was suggested to start at the age of 12-14 years and can differ individually. The panel was not able to determine a certain age for the actual transition to adult care due to the various conditions that apply in each EU country. However, they emphasised that the transition should be completed before the age of 18.

Evidence from a retrospective single center study by Acker et al. supported the recommendation that care-providing expertise centers and pediatric surgery centers have a responsibility to ensure a seamless transition for patients with ARM as they move from pediatric care to adult care. This process should involve collaboration between both the pediatric specialist and an adult specialist in order to promote effective communication and consultation.[24]

Throughout the transition phase, there will be a smooth transfer to a new adult care physician. Special focus will be given to this aspect in order to ensure a positive transition. This recommendation was originally found in the organisation of care module of the Dutch Quality Standard.[2] However, upon further evaluation, the panel determined that it would be better suited for inclusion in the transition care module.[25]

The Follow-up for adults is not yet well organised everywhere, and many adolescents do not come to the clinic. The AAWG brought attention to the fact that follow-up for adults is lacking in organisation in several areas, resulting in a significant number of adolescents not attending their follow-up appointments. It is crucial for both healthcare providers and patients to be motivated to provide/attend these follow-ups within a structured and organised framework. The healthcare professionals and patients should be encouraged to provide/attend follow-up appointments/visits within an organised and structured setting. Additionally, the group mentioned the potential for disseminating information through conferences on transition of care and establish connection between pediatric and adult healthcare providers on various levels through national or European associations.

The AAWG highlighted that maintaining a low threshold for collaboration with preconception care specialists is essential when supporting patients with ARM who have questions and concerns about

having children. By keeping the threshold of collaboration low, healthcare professionals are still in the line of information and ensure that patients with ARM have easy access to preconception.

3.3. Implementation, Validity and Evaluation

3.3.1 Implementation

In the different phases of adapting the Quality Standard, the implementation of the Quality Standard and the practical feasibility of the recommendations have been taken into account. In doing so, explicit attention was paid to factors that could promote or hinder the implementation of the Quality Standard in practice.

The panel highlighted that varying resources and priorities in different European countries, limited investments in specialised facilities, equipment, or trained personnel, and the absence of established mechanisms for interdisciplinary collaboration may hinder the implementation of the ARM Quality Standard. Furthermore, variations in regulatory frameworks, legal systems, and knowledge gaps among healthcare professionals, policymakers, and the general public also pose barriers for the implementation of a Quality Standard across Europe.

In addition to the challenges mentioned, the panel suggested several manners to promote a widespread adoption of the European Quality Standard for ARM. As eUROGEN is a network consisting of healthcare providers across Europe, panel members can actively contribute by sharing the Quality Standard in their respective countries and advocating for its implementation. Local webinars or workshops can be organised ensuring that the knowledge spreads effectively to healthcare providers who may have limited access to educational resources about the Quality Standard. Furthermore, the AAWG aims to publish topic-specific articles in relevant journals to disseminate information about the Quality Standard. Moreover, eUROGEN organises European webinars on a regular basis, part of which are dedicated to proact the awareness of ARM among healthcare providers. Additionally, translating the Quality Standard into different languages could facilitate its implementation from healthcare professionals who may not be fluent in English.

3.3.2 Validity

This Quality Standard is valid from the moment of publication, until the publication of its update. In general, a guideline will undergo evaluation every 5 years. Consequently, the board of the eUROGEN will install a new workgroup in 2028 to evaluate which modules are in need of an update and if there is a need to develop new modules based on the latest insights.

4. DISCUSSION

The purpose of this paper was to adapt the Dutch Quality Standard for patients with Anorectal Malformation to fit the European context. By tailoring the Dutch Quality Standard, the AAWG aimed to establish a framework that would effectively guide healthcare providers in delivering standardised, high-quality care for patients with ARM throughout Europe, and thus improving overall care In the following thematic modules: 1) Lifelong follow-up and Integrated care, 2) Organisation of care, 3) Collaboration, referral and communication between care providers and 4) Transition of care and by ensuring equitable access to care. The AAWG considered the favorable benefit-risk balance, the acceptability and applicability in order to adopt, adapt or to form new recommendations.

During the process, areas of consensus were identified and areas of ambiguity that require further research were revealed. Upon review of existing literature, the panel verified that the majority of studies lack sufficient evidence, being restricted to retrospective reviews and case series. Important areas of consensus and ambiguity and their research implications are addressed below.

Lifelong follow-up and Integrated care for ARM

Limited evidence suggests that scar morbidity is not explicitly documented during the follow-up of ARM patients.[11] The panel acknowledges the need for further research into this matter, such as through systematic reviews. Furthermore, gathering indirect evidence from other patient groups could add value in this cause.

The AAWG indicated that there is currently no fully validated and acceptable score available to assess bowel function and continence in patients with ARM. Consequently, the panel recommended against using the Krickenbeck criteria for fecal incontinence classification and suggested further research for a comprehensive scoring system.[18] The panel emphasised the need to consider the psychological burden experienced by patients and the lack of adequate assessment for psychosocial issues. Additionally, the AAWG agrees that the use of questionnaires/ clinical screening tools for assessment of bowel function may be of added value and the recommended options are: Rintala Continence Score, Bowel Function Score (BFS), Wexner Incontinence Score, Groningen Defecation and Fecal Continence Score.[26-29]

Despite the short duration of the first decade of life, the AAWG highlighted that it is imperative to maintain awareness regarding the potential risk of colorectal cancer (CRC) in young adults born with ARM. Despite this concern, there is currently no evidence supporting the implementation of a screening program for this specific population. Incidental cases of CRC have been reported In literature,[30] but comprehensive research is needed to establish the exact risk associated with CRC in individuals with ARM. Therefore, it is crucial to emphasise the urgent need for further investigation to gather substantial evidence that can guide the development of appropriate screening strategies and interventions for CRC in young adults with ARM.

Organisation of care

The AAWG highlighted the need for further research on centralisation in ARM care. When centralisation is achieved, long-term follow-up for patients is crucial. Patients who undergo ARM procedures require continued surveillance and support throughout their lives, as they may experience ongoing symptoms or complications. Establishing a robust system for long-term follow-up is crucial to ensure that patients receive appropriate care even after the initial treatment.

The AAWG unanimously agreed that an increasing number of procedures could improve quality. Considering the discrepancies across the European countries, the AAWG reached in a consensus that each ARM expertise center should be responsible for at least 10 ARM reconstructions per year involving multidisciplinary teams.[22] The establishment of minimum procedural volumes will ensure that centers have enough experience and expertise to deliver optimal care for ARM patients.

By conducting further research and carefully considering the organisational demands, countries can work towards implementing centralisation strategies that are feasible within their specific healthcare settings. This approach will help ensure that ARM patients receive optimal care while taking into account the unique circumstances of each country.

Collaboration, referral and communication between care providers

The AAWG underlined that there is a lack of evidence on collaboration, referral and communication between care providers in ARM found in current literature. The AAWG indicated that it is crucial to address these gaps and explore the barriers and facilitators that impact collaboration and communication between care providers in ARM.

In order to address this issue, additional research is necessary to gain a deeper understanding of the difficulties faced by care providers in ARM care collaboration and communication. Future studies should strive to pinpoint the obstacles that impede efficient teamwork, with the goal of developing interventions to surmount them and enhance collaboration among care providers.

Similarly, exploring the facilitators that promote efficient collaboration and communication is equally important. This could include identifying successful strategies used in other healthcare settings or specific interventions that have been effective in enhancing multidisciplinary collaboration. By understanding these facilitators, efforts can be made to replicate and implement them within ARM care, ensuring a smoother flow of information and coordination between care providers.

Transition care

The high prevalence of ongoing symptoms during the transition phase emphasises the need for effective follow-up of bowel and urinary, sexual and psychosocial function and quality of life. A well-structured transition program can play a crucial role in addressing these symptoms and ensuring a smoother transition for patients.

To achieve this, it is essential for the transition program to focus on providing adequate knowledge about the disease. Educating patients about their condition, its causes, symptoms, and treatment options can empower them to actively participate in their own care. This knowledge not only increases their understanding but also helps them make informed decisions regarding their treatment and lifestyle choices.

Additionally, considering the patient's perspective and involving them in the development and implementation of the transition program is crucial. Understanding their experiences, concerns, and preferences will allow healthcare providers to tailor the program to meet individual needs. This patient-centred approach ensures that the importance of the transition program is conveyed effectively, increasing patient engagement and adherence.

Overall, a well-structured transition program that focuses on disease knowledge and incorporates the patient's perspective can significantly benefit both patients and healthcare providers. It promotes better outcomes, increased patient satisfaction, and improved quality of life during the transition phase. Therefore, more extensive research should be conducted over the transition care of patients with ARM. Meanwhile, consulting other already established transition programmes should be encouraged.

5. CONCLUSION

ERN eUROGEN aims to form a common Quality Standard for ARM that applies in each of the European countries. A set of recommendations was developed for the following thematic modules: 1) Lifelong follow-up and integrated care, 2) Organisation of care, 3) Collaboration, referral and communication between care providers and 4) Transition care according to the European context and based on current evidence. Close attention should be paid to the dissemination and the implementation of this adapted Quality Standard across the target groups. Further research is required to enhance the certainty of evidence in areas of ambiguity and to develop more evidence-based recommendations within these components of ARM care.

ABOUT THIS DOCUMENT

DISCLOSURES

We would like to disclose that two of the authors of this adaptation report, (IDB, CEJS) were also authors of the Dutch Quality Standard. However, it is important to note that neither IDB, nor CEJS have any conflict of interest to declare and they did not advocate for any particular viewpoints or positions in relation to the content of the Dutch Quality Standard. No conflicting interests were declared from the authors.

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COLLABORATION

This work was performed in collaboration with experts involved in ERN ERNICA, EUPSA and Qualicura.

European Reference Network
for rare or low prevalence complex diseases
Network Inherited and Congenital Anomalies (ERNICA)

EUPSA European Paediatric Surgeons' Association

Qualicura

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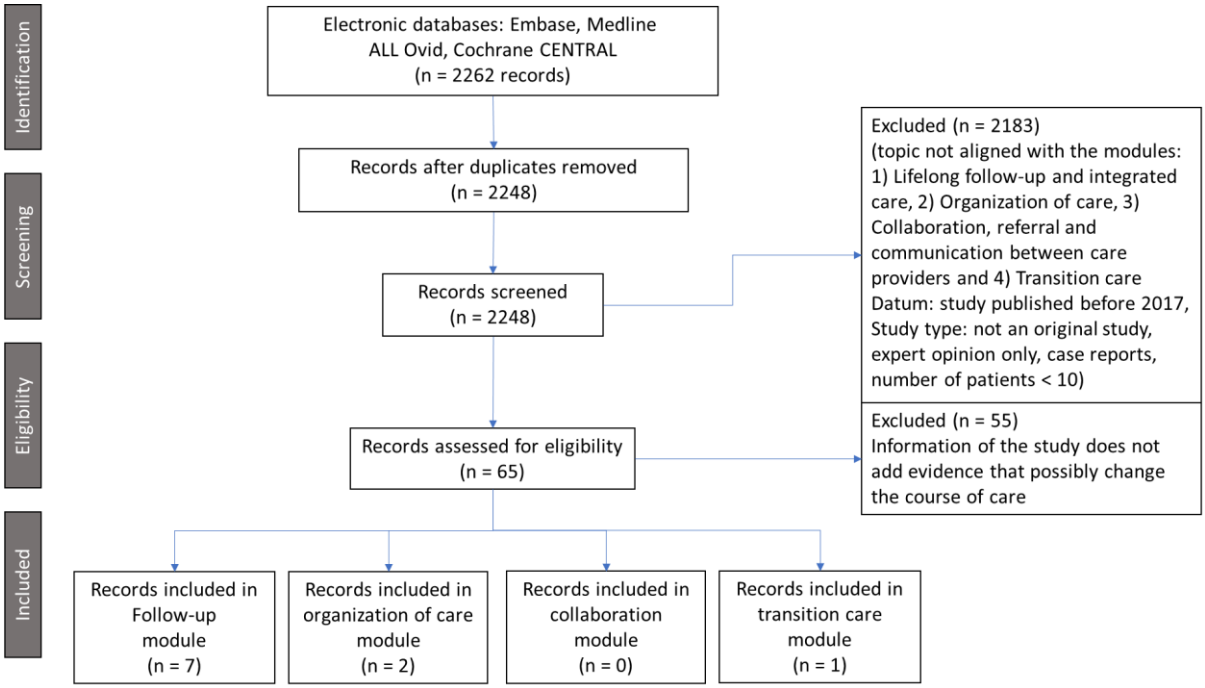
APPENDIX

SYSTEMATIC LITERATURE SEARCH

	EMBASE	OVID/MEDLINE	Cochrane CENTRAL	Deduplicated
SRs	223	135		249
RCTs	411	155		475
Observational studies	1356	884		1526
Other			58	12
Total				2262

Appendix Table 1. Systematic literature search

LITERATURE REVIEW



Appendix Figure A. PRISMA flow diagram of literature selection

Summary of the evidence for follow-up and integrated care

Seven (7) new studies, published between 2018 and 2023, were found for follow-up and Integrated care module.

FOLLOW-UP AND INTEGRATED CARE	AUTHOR
Type of study: A (prospective) multi-center cohort study	van der Steeg et al.,2022

Aim: to evaluate bowel function of RVF-patients at preschool/early childhood age and determine risk factors for poor functional outcome. Population: ARM patients with rectovestibular fistula (RVF) Results: The study included 111 RVF-patients. Median BFS was 16 (range 6-20). The 'below normal' group consisted of 61 patients (55.0%). Overall, we reported soiling, fecal accidents, and constipation in 64.9%, 35.1% and 70.3%, respectively. Bowel management was performed in 23.4% of patients. Risk factors for poor outcome were tethered cord and low sacral ratio, while sacral anomalies, low sacral ratio, prior enterostomy, post-reconstructive complications, and one-year constipation were for being on bowel management. Level of evidence: Level III study (OCEBM, 2011) Authors' conclusions: Although median BFS at 4-7 year follow-up is nearly normal, the majority of patients suffers from some degree of soiling and constipation, and almost 25% needs bowel management. Several factors were associated with poor bowel function outcome and bowel management.	
Type of study: Local non-randomized sample study Aim: to assess the physical and psychosocial significance of abdominal scarring in ARM and to propose a scar treatment approach Population: ARM patients, 13 (48%) females and 14 (52%) males with a median age of 12 (5–24) years Results: The median POSAS score of all assessed scars was 44 (15–78) and increased with age. Postoperative scarring had a negative physical impact with recurrent scar pain and/or scar pruritus occurring in 29% of participants. 37% of participants had moderate to severe scar symptoms. No differences between male/female patients were found. Level of evidence: Level IV study (OCEBM, 2011) Authors' conclusions: In conclusion, both physical- and psychosocial scar morbidity should be addressed regardless of gender in ARM follow-up programs.	Tofft et al., 2022
Type of study: Local non-randomized sample study Aim: Asses view op parents on talking about sexuality with children born with ARM Population: Parent to child with ARM not older than 21 (n=93) Results: Overall, 65.6% of parents never talked about sex with their child, 72% feels that their child should be able to talk to them about it. Children's age ranged from 4 to 21, with a mean age of 10.7 (SD 4.7) Correlational analyses showed that children's age was marginally positively correlated with occasions to discuss sexuality with their parents (r=0.202, p=0.053). Level of evidence: Level IV (OCEBM, 2011) Author conclusions: Psychologists, gynecologists/andrologists, and pediatric surgeons are seen as key resources for talking about sexuality. A great number of parents express the wish that their children had more opportunities to discuss sexual topics with pediatric surgeons.	Eleuteri et al., 2022
Type of study: qualitative systematic review (63 articles includes published between 1980 and 2019) Objective: To highlight the psychosocial, emotional, and behavioral themes that affect anorectal malformation (ARM) and Hirschsprung disease (HSCR) patients.	Svetanoff et al., 2022

Population: Patients with anorectal malformation and Hirschsprung disease. Results: In the neonatal period, parents relayed uncertainty about the future and feeling overwhelmed by lack of social support. Difficulties with anxiety, peer rejection, and behavioural problems were noted in primary grades, while adolescents experienced low self-confidence, poor body image, and depression. Young adults expressed hesitancy to engage in romantic relationships or sexual activity. Lack of long-term follow-up, an incomplete transition to adult healthcare, and lack of psychology services leave young adults without guidance to manage a chronic condition. Level of evidence: Level V study (OCEBM, 2011) Authors' conclusions: Multiple psychosocial stressors are present in the lives of ARM and HSCR patients. Provision of developmentally matched medical, psychological, and community-based supports for ARM and HSCR patients and their families can lead to improved quality of life (QoL).	
Type of study: Prospective study (1.5 years duration with 30 consecutive ARM patients) Objective: To assess the quality of life and the psychosocial burden of anorectal malformation and to compare quality and psychosocial burden among parents between staged and definitive group. Population: 30 consecutive ARM patients <12 years old. Results: The results of our study should be taken into account in policy making to provide better and more specific supports and interventions for this group of diseases. More attention should be given to parents (and in particular mothers) needs. Social support and different coping strategies should be developed to respond positively to individual v Changing needs and in buffering parents from the stress of having a child with anorectal malformation. Level of Evidence: Level IV study (OCEBM, 2011) Authors' conclusions: Most of our parents experienced greater psychosocial burden and their quality of life worsened following surgery. Among all domains of quality of life, social relationship was affected most followed by physical and psychological. Environmental domain was least affected. Our study also compares the psychosocial burden and quality of life of caregiver between staged and definitive group and study revealed that there was greater psychosocial burden and poor quality of life in staged surgery group.	Bhartiya et al., 2019
Type of study: Retrospective case-control study (96 ARM cases and 960 controls, data from 1991 to 2017). Population: Children diagnosed with ARM. Comparison: Age-matched control group. Objective: To assess real-world educational outcomes, neurodevelopmental disorders and mental health disorders in ARM patients and compare to an age-matched control group. Results: A total of 96 ARM cases and 960 controls were identified. Cases were at greater risk of failing to meet expectations on Grades 7 and 8 assessments. After entering high school, ARM patients were at no greater risk than their peers of failing to meet expectations. Cases were more likely to have a developmental or	Miyake et al., 2023

intellectual disability (OR 3.59, p < 0.001), anxiety (OR 1.86, p = 0.023), depression (OR 2.35, p = 0.022) or hyperactivity disorder (OR 2.01, p = 0.036). Level of Evidence: Level IV (OCEBM, 2011) Authors' conclusions: Our study demonstrated that ARM patients may be more likely to perform poorly in junior high school than controls and may be at greater risk of neurodevelopmental and mental health disorders. It is important for pediatric surgeons to anticipate these challenges and endorse psychosocial supports to optimize educational and mental health outcomes.	
Type of study: Guidelines Aim: To increase the quality of care for children with urological conditions	Radmayr et al., 2023

Appendix Table 2. Summary of evidence for follow-up and Integrated care studies.

Summary of the evidence for organisation of care

Two (2) new study published between 2020 and 2022 were found for the organization of care module.

ORGANIZATION OF CARE FOR ARM PATIENTS	AUTHOR
Type of study: Retrospective review (n=354 patients included, data from 2005–2017). Hypothesis: The development of a colorectal center at our children's hospital decreased readmissions in our colorectal surgery population. Population: anorectal malformation (ARM) and Hirschsprung disease (HD) patients Results: A total of 354 patients were identified. 178 patients (113 ARM, 65 HD) were treated prior to and 176 patients (110 ARM, 66 HD) were treated after the development of the colorectal center. Forty-five (25.3%) patients underwent neonatal repair prior to development of the center compared to 15 (8.5%) after. 111 (62.4%) patients underwent colostomy prior to the colorectal center comparted to 95 (54%) after. The rate of readmission within 120 days of discharge in the early group was 63% compared to 52% in those managed in the multidisciplinary colorectal center (p = 0.04). Conversely, the rate of emergency room visits increased from 8.4% to 27.3% (p = 0.01). The decrease in readmission rates was more pronounced in the ARM group, while the HD cohort had similar readmission rates before and after the establishment of the center. Multivariate logistic regression revealed an odds ratio of 0.59 (95% CI 0.37–0.92) for readmission following the development of the multidisciplinary colorectal center. Level of Evidence: Level III, retrospective comparative study. Authors' conclusions: The development of a multidisciplinary colorectal center at our institution was associated with decreased hospital readmissions, but an increase in emergency department resource utilization. These findings suggest improved efficiency in patient care with the implementation of a multispecialty, patient centered approach while also identifying areas of focus for future quality improvement initiatives.	Kastenberg et al., 2020
Type of study: Retrospective review (n=2162 newborns, data from 2015-2017). Objective: To assess the operative volume of the most relevant congenital malformations at German academic pediatric surgical institutions over the past years.	Lacher et al., 2022

Population: Newborns underwent surgery for congenital malformations and neonatal abdominal emergencies at German academic medical centers Results: From 2015 through 2017, a total 2,162 newborns underwent surgery for congenital malformations and neonatal abdominal emergencies at German academic medical centers, representing 51% of all expected newborn cases nationwide. The median of cases per center within the study period was 101 (range 18-258). Four institutions (21%) were classified as "high volume" centers, four (21%) as "medium volume" centers, and 11 (58%) as "low volume" centers. The proportion of patients operated on in high-volume centers varied per disease category: esophageal atresia/tracheoesophageal fistula: 40%, duodenal atresia: 40%, small and large bowel atresia: 39%, anorectal malformations: 40%, congenital diaphragmatic hernia: 56%, gastroschisis: 39%, omphalocele: 41%, Hirschsprung disease: 45%, posterior urethral valves: 39%, and necrotizing enterocolitis (NEC)/focal intestinal perforation (FIP)/gastric perforation (GP): 45%. Level of Evidence: Level III Authors' conclusions: This study provides a national benchmark for neonatal surgery performed in German university hospitals. The rarity of these cases highlights the difficulties for individual pediatric surgeons to gain adequate clinical and surgical experience and research capabilities. Therefore, a discussion on the centralization of care for these rare entities is necessary.	

Appendix Table 3. Summary of evidence for organisation of care studies.

Summary of the evidence for transition care

One (1) new study published In 2019 was found for the transition care module.

TRANSITION CARE	AUTHOR
Type of study: retrospective review (88 cases with colorectal anomalies of which 55 are ARM from 1983 until 2017) Population: Patients with colorectal anomalies (88 cases in total. 51 patients had ARM, 18 cloacas, 9 presacral masses, 3 HD, 2 spina bifida and 5 with other diagnoses (3 vaginal anomalies, 1 cloacal exstrophy, 1 obstructed seminal vesical). Objective: To describe some of the most common problems experienced by adult patients with congenital colorectal malformations. Results: The specific problems addressed were: complications from previous operations (41), rectal prolapse (25), fecal incontinence (11), gynecologic concerns (12), urologic concerns (6), and recurrent recto urogenital fistula (3). We performed 83 surgical interventions, including 13 rectal prolapse repair, 13 continent appendicostomies, 44 PSARP or redo PSARP, 11 resections of presacral masses, 11 vaginoplasties, 2 examinations under anesthesia, and 2 Mitrofanoff procedures. Five patients were treated medically (bowel management program, obstetric, urologic evaluation). Level of evidence: Level III, cohort study Authors' conclusions: There is a growing need to better prepare adult providers to assume the care of patients born with congenital colorectal disease as these patients transition to adulthood. A collaboration between specialized pediatric referral centers with adult colorectal surgeons, urologists and gynecologists is a potential pathway for the adequate transition of care.	Shannon Acker et al., 2019

Appendix Table 4. Summary of evidence for transition care studies.

DUTCH QUALITY STANDARD APPRAISAL

Domain	Signaling question
Domain 1. Scope and purpose	Item 1. The overall objective(s) of the guideline is (are) specifically described.
	Item 2. The health question(s) covered by the guideline is (are) specifically described.
	Item 3. The population (patients, public, etc.) to whom the guideline is meant to apply is specifically described.
Domain 2. Stakeholder Involvement	Item 4. The guideline development group includes individuals from all relevant professional groups.
	Item 5. The views and preferences of the target population (patients, public, etc.) have been sought.
	Item 6. The target users of the guideline are clearly defined.
Domain 3. Rigour of Development	Item 7. Systematic methods were used to search for evidence.
	Item 8. The criteria for selecting the evidence are clearly described.
	Item 9. The strengths and limitations of the body of evidence are clearly described.
	Item 10. The methods for formulating the recommendations are clearly described.
	Item 11. The health benefits, side effects, and risks have been considered in formulating the recommendations.
	Item 12. There is an explicit link between the recommendations and the supporting evidence.
	Item 13. The guideline has been externally reviewed by experts prior to its publication.
	Item 14. A procedure for updating the guideline is provided.
Domain 4. Clarity of Presentation	Item 15. The recommendations are specific and unambiguous.
	Item 16. The different options for management of the condition or health issue are clearly presented.
	Item 17. Key recommendations are easily identifiable.
Domain 5. Applicability	Item 18. The guideline describes facilitators and barriers to its application.
	Item 19. The guideline provides advice and/or tools on how the recommendations can be put into practice.

	Item 20. The potential resource implications of applying the recommendations have been considered.
	Item 21. The guideline presents monitoring and/or auditing criteria.
Domain 6. Editorial Independence	Item 22. The views of the funding body have not influenced the content of the guideline.
	Item 23. Competing interests of guideline development group members have been recorded and addressed.

Appendix Table 5. AGREE II domains and items.

Results

Domain scores were calculated by summing up all the scores of the individual items in a domain and by scaling the total as a percentage of the maximum possible score for that domain.

- Domain 1 (scope and purpose) had a standard domain score of 88%. The objectives and population were clearly described in the guideline; however, the health questions covered by the guideline were only vaguely described.
- Domain 2 (stakeholder involvement) was the highest scoring domain with full marks being awarded by each reviewer for each signaling question resulting in a standard score domain of 1.
- Domain 3 (rigour of development) had a standard domain score of 78%. There was a variety of responses and disagreement in this domain between reviewers. It was clearly described in the guideline that systematic methods were used and that there is a procedure in place for updating the guideline. However, the other questions in this domain were more ambiguous throughout the methods of the guideline development.
- Domain 4 (clarity of presentation) had a standard domain score of 75%. The recommendations were evaluated as being unspecific and ambiguous and the key recommendations were not easily identifiable.
- Domain 5 (applicability) was the lowest scoring domain with a standard domain score of 46%. However, there was a high level of disagreement between reviewers in this domain for all of the signaling questions.
- Domain 6 (editorial independence) had a standard domain score of 75%. The competing interests wasn´t clear to some of the reviewers.

The overall standard domain score given by the reviewers was 38%. The guideline was recommended for use with some modifications by two reviewers and recommended for use without modifications by the other two reviewers.

AGREE II quality scores

Quality Standard for Anorectal Malformation		ES	PG	ML	JG	Total	Min	Max	Standard Domain Score
Domain 1. Scope and purpose	Item 1. The overall objective(s) of the guideline is (are) specifically described.	7	6	7	7	27	**12**	**84**	**88%**
	Item 2. The health question(s) covered by the guideline is (are) specifically described.	7	3	7	7	24			
	Item 3. The population (patients, public, etc.) to whom the guideline is meant to apply is specifically described.	4	6	7	7	24			
	Total	18	15	21	21	**75**			
Domain 2. Stakeholder Involvement	Item 4. The guideline development group includes individuals from all relevant professional groups.	7	7	7	7	28	**12**	**84**	**100%**
	Item 5. The views and preferences of the target population (patients, public, etc.) have been sought.	7	7	7	7	28			
	Item 6. The target users of the guideline are clearly defined.	7	7	7	7	28			
	Total	21	21	21	21	**84**			
	Item 7. Systematic methods were used to search for evidence.	7	7	7	7	28	**32**	**224**	**78%**

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Domain 3. Rigour of Development	Item 8. The criteria for selecting the evidence are clearly described.	5	4	7	7	23			
	Item 9. The strengths and limitations of the body of evidence are clearly described.	3	1	7	7	18			
	Item 10. The methods for formulating the recommendations are clearly described.	5	3	7	7	22			
	Item 11. The health benefits, side effects, and risks have been considered in formulating the recommendations.	5	4	7	7	23			
	Item 12. There is an explicit link between the recommendations and the supporting evidence.	1	3	7	7	18			
	Item 13. The guideline has been externally reviewed by experts prior to its publication.	7	4	7	7	25			
	Item 14. A procedure for updating the guideline is provided.	7	3	7	7	24			
	Total	40	29	56	56	**181**			
Domain 4. Clarity of Presentation	Item 15. The recommendations are specific and unambiguous.	3	4	7	7	21	**12**	**84**	**75%**
	Item 16. The different options for management of the condition or health issue are clearly presented.	7	6	7	7	27			
	Item 17. Key recommendations are easily identifiable.	3	1	7	7	18			

	Total	13	11	21	21	**66**			
Domain 5. Applicability	Item 18. The guideline describes facilitators and barriers to its application.	1	2	7	7	17	**16**	**112**	**46%**
	Item 19. The guideline provides advice and/or tools on how the recommendations can be put into practice.	4	1	5	5	15			
	Item 20. The potential resource implications of applying the recommendations have been considered.	1	1	7	7	16			
	Item 21. The guideline presents monitoring and/or auditing criteria.	1	1	5	5	12			
	Total	7	5	24	24	**60**			
Domain 6. Editorial Independence	Item 22. The views of the funding body have not influenced the content of the guideline.	7	7	7	7	28	**8**	**56**	**75%**
	Item 23. Competing interests of guideline development group members have been recorded and addressed.	7	7	1	1	16			
	Total	14	14	8	8	**44**			
Overall assessment Please, rate the overall quality of this guideline		5	4	7	7	23	**7**	**49**	**38%**

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	Total	5	4	7	7	**23**			
Would you recommend this guideline for use?		**Yes, with modifications**	**Yes, with modifications**	**Yes**	**Yes**				

Appendix Table 6. AGREE II quality scores

Standardised scores per AGREE II domain

AGREE II Instrument	Quality Standard for Anorectal Malformation
Domain 1. Scope and Purpose	88%
Domain 2. Stakeholder Involvement	100%
Domain 3. Rigour of Development	78%
Domain 4. Clarity of Presentation	75%
Domain 5. Applicability	46%
Domain 6. Editorial Independence	75%

Appendix Figure 2. Standard domain scores of each domain

SYSTEMATIC LITERATURE SEARCH STRATEGY

Embase

No.	Query	Results
#11	#9 NOT #8 NOT #7 OBS	1356
#10	#8 NOT #7 Clinical trials, RCT	411
#9	#2 AND (#5 OR #6)	1564
#8	#2 AND #4	346
#7	#2 AND #3SR	223
#6	'case control study'/de OR 'comparative study'/exp OR 'control group'/de OR 'controlled study'/de OR 'controlled clinical trial'/de OR 'crossover procedure'/de OR 'double blind procedure'/de OR 'phase 2 clinical trial'/de OR 'phase 3 clinical trial'/de OR 'phase 4 clinical trial'/de OR 'pretest posttest design'/de OR 'pretest posttest control group design'/de OR 'quasi experimental study'/de OR 'single blind procedure'/de OR 'triple blind procedure'/de OR (((control OR controlled) NEAR/6 trial):ti,ab,kw) OR (((control OR controlled) NEAR/6 (study OR studies)):ti,ab,kw) OR (((control OR controlled) NEAR/1 active):ti,ab,kw) OR 'open label*':ti,ab,kw OR (((double OR two OR three OR multi OR trial) NEAR/1 (arm OR arms)):ti,ab,kw) OR ((allocat* NEAR/10 (arm OR arms)):ti,ab,kw) OR placebo*:ti,ab,kw OR 'sham-control*':ti,ab,kw OR (((single OR double OR triple OR assessor) NEAR/1 (blind* OR masked)):ti,ab,kw) OR nonrandom*:ti,ab,kw OR 'non-random*':ti,ab,kw OR 'quasi-experiment*':ti,ab,kw OR crossover:ti,ab,kw OR 'cross over':ti,ab,kw OR 'parallel group*':ti,ab,kw OR 'factorial trial':ti,ab,kw OR ((phase NEAR/5 (study OR trial)):ti,ab,kw) OR ((case* NEAR/6 (matched OR control*)):ti,ab,kw) OR ((match* NEAR/6 (pair OR pairs OR cohort* OR control* OR group* OR healthy OR age OR sex OR gender OR patient* OR subject* OR participant*)):ti,ab,kw) OR ((propensity NEAR/6 (scor* OR match*)):ti,ab,kw) OR versus:ti OR vs:ti OR compar*:ti OR ((compar* NEAR/1 study):ti,ab,kw) OR (('major clinical study'/de OR 'clinical study'/de OR 'cohort analysis'/de OR 'observational study'/de OR 'cross-sectional study'/de OR 'multicenter study'/de OR 'correlational study'/de OR 'follow up'/de OR cohort*:ti,ab,kw OR 'follow up':ti,ab,kw OR followup:ti,ab,kw OR longitudinal*:ti,ab,kw OR prospective*:ti,ab,kw OR retrospective*:ti,ab,kw OR observational*:ti,ab,kw OR 'cross sectional*':ti,ab,kw OR cross?ectional*:ti,ab,kw OR multicent*:ti,ab,kw OR 'multi-cent*':ti,ab,kw OR consecutive*:ti,ab,kw) AND (group:ti,ab,kw OR groups:ti,ab,kw OR subgroup*:ti,ab,kw OR versus:ti,ab,kw OR vs:ti,ab,kw OR compar*:ti,ab,kw OR 'odds ratio*':ab OR 'relative odds':ab OR 'risk ratio*':ab OR 'relative risk*':ab OR 'rate ratio':ab OR aor:ab OR arr:ab OR rrr:ab OR ((('or' OR 'rr') NEAR/6 ci):ab)))	13923341
#5	'major clinical study'/de OR 'clinical study'/de OR 'case control study'/de OR 'family study'/de OR 'longitudinal study'/de OR 'retrospective study'/de OR 'prospective study'/de OR 'comparative study'/de OR 'cohort analysis'/de OR ((cohort NEAR/1 (study OR studies)):ab,ti) OR (('case control' NEAR/1 (study OR studies)):ab,ti) OR (('follow up' NEAR/1 (study OR studies)):ab,ti) OR (observational NEAR/1 (study OR studies)) OR ((epidemiologic NEAR/1 (study OR studies)):ab,ti) OR (('cross sectional' NEAR/1 (study OR studies)):ab,ti)	6767914
#4	'clinical trial'/exp OR 'randomization'/exp OR 'single blind procedure'/exp OR 'double blind procedure'/exp OR 'crossover procedure'/exp OR 'placebo'/exp OR 'prospective study'/exp OR rct:ab,ti OR random*:ab,ti OR 'single blind':ab,ti OR 'randomised controlled trial':ab,ti OR 'randomized controlled trial'/exp OR placebo*:ab,ti	3302394
#3	'meta analysis'/exp OR 'meta analysis (topic)'/exp OR metaanaly*:ti,ab OR 'meta analy*':ti,ab OR metanaly*:ti,ab OR 'systematic review'/de OR 'cochrane database of systematic reviews'/jt	733409

DOI:[number]

	OR prisma:ti,ab OR prospero:ti,ab OR (((systemati* OR scoping OR umbrella OR 'structured literature') NEAR/3 (review* OR overview*)):ti,ab) OR ((systemic* NEAR/1 review*):ti,ab) OR (((systemati* OR literature OR database* OR 'data base*') NEAR/10 search*):ti,ab) OR (((structured OR comprehensive* OR systemic*) NEAR/3 search*):ti,ab) OR (((literature NEAR/3 review*):ti,ab) AND (search*ti,ab OR database*:ti,ab OR 'data base*':ti,ab)) OR (('data extraction':ti,ab OR 'data source*':ti,ab) AND 'study selection':ti,ab) OR ('search strategy':ti,ab AND 'selection criteria':ti,ab) OR ('data source*':ti,ab AND 'data synthesis':ti,ab) OR medline:ab OR pubmed:ab OR embase:ab OR cochrane:ab OR (((critical OR rapid) NEAR/2 (review* OR overview* OR synthes*)):ti) OR ((((critical* OR rapid*) NEAR/3 (review* OR overview* OR synthes*)):ab) AND (search*:ab OR database*:ab OR 'data base*':ab)) OR metasynthes*:ti,ab OR 'meta synthes*':ti,ab	
#2	#1 AND [1-1-2017]/sd NOT ('conference abstract'/it OR 'editorial'/it OR 'letter'/it OR 'note'/it) NOT (('animal'/exp OR 'animal experiment'/exp OR 'animal model'/exp OR 'nonhuman'/exp) NOT 'human'/exp)	3517
#1	'anorectal malformation'/exp OR 'cloacal malformation'/exp OR 'cloacal anomaly'/exp OR (((rectal OR anorectal OR anal OR anus OR cloaca*) NEAR/3 (atres* OR anomal* OR malformat* OR atret* OR imperforat* OR agenes*)):ti,ab,kw) OR ((currarin* NEAR/3 (syndrome* OR triad)):ti,ab,kw) OR (((lumbar OR townes OR pallister) NEAR/3 syndrome*):ti,ab,kw) OR 'hypothalam* hamartoblastom*':ti,ab,kw OR 'hydrocolpos'/exp OR 'urethra fistula'/exp OR hydrocolpos:ti,ab,kw OR (((urethra OR perineal OR rectovestibular OR rectobulbar OR 'recto bulbar' OR 'recto vestibular' OR rectourinary OR 'recto urinary' OR 'recto bladder' OR 'peri anal' OR perianal) NEAR/3 fistula):ti,ab,kw) OR 'perineal fistula'/exp OR 'rectovestibular fistula'/exp OR 'rectobulbar fistula'/exp OR 'rectal stenosis'/exp OR 'rectal stenos*':ti,ab,kw	16668

Ovid/Medline

#	Searches	Results
12	10 not 9 not 8 OBS	884
11	9 not 8 Clinical trials, RCTs	155
10	3 and (6 or 7)	1078
9	3 and 5	190
8	3 and 4 SR	135
7	Case-control Studies/ or clinical trial, phase ii/ or clinical trial, phase iii/ or clinical trial, phase iv/ or comparative study/ or control groups/ or controlled before-after studies/ or controlled clinical trial/ or double-blind method/ or historically controlled study/ or matched-pair analysis/ or single-blind method/ or (((control or controlled) adj6 (study or studies or trial)) or (compar* adj (study or studies)) or ((control or controlled) adj1 active) or "open label*" or ((double or two or three or multi or trial) adj (arm or arms)) or (allocat* adj10 (arm or arms)) or placebo* or "sham-control*" or ((single or double or triple or assessor) adj1 (blind* or masked)) or nonrandom* or "non-random*" or "quasi-experiment*" or "parallel group*" or "factorial trial" or "pretest posttest" or (phase adj5 (study or trial)) or (case* adj6 (matched or control*)) or (match* adj6 (pair or pairs or cohort* or control* or group* or healthy or age or sex or gender or patient* or subject* or participant*)) or (propensity adj6 (scor* or match*))).ti,ab,kf. or (confounding adj6 adjust*).ti,ab. or (versus or vs or compar*).ti. or ((exp cohort studies/ or epidemiologic studies/ or multicenter study/ or observational study/ or seroepidemiologic studies/ or (cohort* or 'follow up' or followup or longitudinal* or prospective* or retrospective* or observational* or multicent* or 'multi-cent*'	5381225

	or consecutive*).ti,ab,kf.) and ((group or groups or subgroup* or versus or vs or compar*).ti,ab,kf. or ('odds ratio*' or 'relative odds' or 'risk ratio*' or 'relative risk*' or aor or arr or rrr).ab. or (("OR" or "RR") adj6 CI).ab.))	
6	Epidemiologic studies/ or case control studies/ or exp cohort studies/ or Controlled Before-After Studies/ or Case control.tw. or cohort.tw. or Cohort analy$.tw. or (Follow up adj (study or studies)).tw. or (observational adj (study or studies)).tw. or Longitudinal.tw. or Retrospective*.tw. or prospective*.tw. or consecutive*.tw. or Cross sectional.tw. or Cross-sectional studies/ or historically controlled study/ or interrupted time series analysis/ [Onder exp cohort studies vallen ook longitudinale, prospectieve en retrospectieve studies]	4391876
5	exp clinical trial/ or randomized controlled trial/ or exp clinical trials as topic/ or randomized controlled trials as topic/ or Random Allocation/ or Double-Blind Method/ or Single-Blind Method/ or (clinical trial, phase i or clinical trial, phase ii or clinical trial, phase iii or clinical trial, phase iv or controlled clinical trial or randomized controlled trial or multicenter study or clinical trial).pt. or random*.ti,ab. or (clinic* adj trial*).tw. or ((singl* or doubl* or treb* or tripl*) adj (blind$3 or mask$3)).tw. or Placebos/ or placebo*.tw.	2566361
4	meta-analysis/ or meta-analysis as topic/ or (metaanaly* or meta-analy* or metanaly*).ti,ab,kf. or systematic review/ or cochrane.jw. or (prisma or prospero).ti,ab,kf. or ((systemati* or scoping or umbrella or "structured literature") adj3 (review* or overview*)).ti,ab,kf. or (systemic* adj1 review*).ti,ab,kf. or ((systemati* or literature or database* or data-base*) adj10 search*).ti,ab,kf. or ((structured or comprehensive* or systemic*) adj3 search*).ti,ab,kf. or ((literature adj3 review*) and (search* or database* or data-base*)).ti,ab,kf. or (("data extraction" or "data source*") and "study selection").ti,ab,kf. or ("search strategy" and "selection criteria").ti,ab,kf. or ("data source*" and "data synthesis").ti,ab,kf. or (medline or pubmed or embase or cochrane).ab. or ((critical or rapid) adj2 (review* or overview* or synthes*)).ti. or (((critical* or rapid*) adj3 (review* or overview* or synthes*)) and (search* or database* or data-base*)).ab. or (metasynthes* or meta-synthes*).ti,ab,kf.	655973
3	2 not ((exp animals/ or exp models, animal/) not humans/) not (letter/ or comment/ or editorial/)	2394
2	limit 1 to yr="2017 -Current"	2548
1	Anorectal Malformations/ or Anus, Imperforate/ or Hydrocolpos/ or Rectovaginal Fistula/ or ((rectal or rectum or anorectal or anal or anus or cloaca*) adj3 (atres* or anomal* or malformat* or atret* or imperforat* or agenes*)).ti,ab,kf. or (currarin* adj3 (syndrome* or triad)).ti,ab,kf. or ((lumbar or townes or pallister) adj3 syndrome*).ti,ab,kf. or hypothalam* hamartoblastom*.ti,ab,kf. or hydrocolpos.ti,ab,kf. or ((urethra or perineal or rectovestibular or rectobulbar or recto bulbar or recto vestibular or rectourinary or recto urinary or vesicorectal or vesico rectal or rectobladder or recto bladder or perianal or peri anal) adj3 fistula).ti,ab,kf. or rectal stenos*.ti,ab,kf.	11279

Cochrane CENTRAL

#1 (“anorectal malformation” OR “perineal fistula” OR “rectovestibular fistula” OR “recto-bulbar urethral fistula” OR “recto-bladder neck fistula” OR “anus atresia” OR “rectal atresia” OR “rectal stenosis” OR “hydrocolpos” OR “bulbar fistula” OR “recto-urethral fístula” OR “urethra fistula” OR “anus malformation” OR “rectum malformation” OR “urogenital tract malformation” OR “anal atresia” OR “imperforate anus”):ti,ab,kw (Word variations have been searched)

#2 MeSH descriptor: [Anorectal Malformations] explode all trees 113

#3 2017 - 58

European Reference Networks

https://ec.europa.eu/health/ern_en

European Reference Network
for rare or low prevalence complex diseases
Network Urogenital Diseases (ERN eUROGEN)

https://eurogen-ern.eu/

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