European Reference Network | ERN eUROGEN
Rare Uro-Recto-Genital Diseases & Complex Conditions

ERN eUROGEN Clinical Practice Guideline on Anorectal Malformations (ARM)

Adapted from the Dutch Quality Standard for Anorectal Malformations

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ABSTRACT

Introduction

Anorectal Malformations (ARM) are rare congenital anomalies that require multidisciplinary, lifelong care. Early diagnosis, effective treatment, structured follow-up, and coordinated transitions of care are all essential to achieving improved long-term outcomes. Given the complexity of ARM and variability in care across Europe, the European Reference Network for Rare Urogenital Diseases (ERN eUROGEN) adapted the Dutch Quality Standard to provide harmonised, evidence-informed guidance suitable for the European context.

Objectives

To adapt and expand the Dutch Quality Standard for ARM to reflect current evidence and address the full care pathway, covering diagnosis, treatment, lifelong follow-up, organisation and coordination of care, collaboration between healthcare providers, and transition to adult care, specifically within the European context.

Methods

Using the ADAPTE methodology and systematic literature reviews, the ERN eUROGEN Adoption and Adaptation Working Group (AAWG) evaluated existing recommendations from the Dutch Quality Standard. The process was informed by a systematic literature review using the Medline, Embase, and Cochrane databases (2017–March 2023) and structured consensus meetings. Recommendations were adopted, adapted, or developed de novo, and, where possible, were supported by graded evidence and justifications.

Results

Across two adaptation phases, a total of 64 recommendations were adapted, 25 adopted, and 23 developed de novo. The phase completed in November 2023 focused on diagnostics and treatment of ARM, while the phase completed in January 2024 expanded to lifelong follow-up, organisation of care, healthcare provider collaboration, and transition care. Key updates included stronger guidance on prenatal and postnatal diagnostics, structured lifelong follow-up programmes, the role of multi-disciplinary teams, and the importance of early and supported transition into adult care.

Conclusion

This comprehensive European adaptation of the Dutch Quality Standard provides an updated, context-sensitive framework for ARM care. It spans the entire patient journey and is designed to support consistency, continuity, and quality of care across European healthcare systems. Further research is needed to strengthen the evidence base in areas of uncertainty and to support ongoing updates to recommendations.

Keywords

Anorectal Malformation, ARM, Quality Standard, Adaptation, Diagnosis, Treatment, Integrated Care, Transition of Care.

ACRONYMS AND ABBREVIATIONS

AAWG	Adoption and Adaptation Working Group (expert panel)
AGREE II	Appraisal of Guidelines for Research and Evaluation II
ARM	Anorectal Malformation
ATS	Anal Target Sign
CPG	Clinical Practice Guideline
CRC	Colorectal Cancer
EO	Epididymo-Orchitis
ERN	European Reference Network
GP	General Practitioner
HCP	Healthcare Provider
HMA	Heineke-Mikulicz anoplasty
MRI	Magnetic Resonance Imaging
OCEBM	Oxford Centre for Evidence-Based Medicine
SCD	Spinal Cord Dysraphism
SD	Spinal Dysraphia
SP	Supervising Physician
TC	Tethered cord
TCS	Tethered Cord Syndrome
US	Ultrasound
(V)UD	(Video)-Urodynamics

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1. INTRODUCTION

1.1. General

Anorectal malformations (ARM) are rare conditions. The incidence of ARM varies between 1 in 3,500 and 1 in 5,000 live births.[1] There is a significant need to increase awareness and knowledge among all healthcare providers involved in caring for patients with ARM to reduce diagnostic delays, improve diagnostics and treatment, and prevent complications and comorbidities in both the short and long term, thereby enhancing patients' quality of life. Caring for patients with ARM is complex, and multi-disciplinary and customised care by healthcare providers with specialised expertise is essential. An extensive introduction to ARM and associated malformations can be found in the Dutch Quality Standard on Anorectal Malformations.[2]

1.2. Scope and objectives

The scope of this adapted clinical practice guideline (CPG) follows the scope of the Dutch Quality Standard. This includes the care pathway from prenatal detection through to lifelong follow-up care and transition care for patients with all types of ARM. A comprehensive overview of all types of ARM and their associations can be found in the Dutch Quality Standard.[2]

The primary objectives of this adaptation are:

1. To assess the currency and validity of the Dutch Quality Standard recommendations to improve and safeguard the quality of care for patients with ARM.
2. To assess the 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 adapted clinical practice guideline (CPG) is patients with anorectal malformations (ARM). Additionally, the primary target audience of the CPG 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 Dutch Quality Standard[2] was appraised for its methodological quality with the AGREE II tool (see **Appendix 1**). To assess if the recommendations were current or if new literature could update the advice in the Quality Standard, a literature review was performed (see **Appendix 2**). 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 was completed, the AAWG considered the results to draw a conclusion for each recommendation. The existing recommendations from the original Quality Standard were adopted in their entirety, modified in order to be adapted, or newly developed based on the new evidence found. In this document, the recommendations have been referred to as either adopted, adapted, or new. The definitions of these categories 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 presented below to illustrate the process leading to a conclusion about the recommendations for creating an adopted or adapted guideline (see **Figure A**).

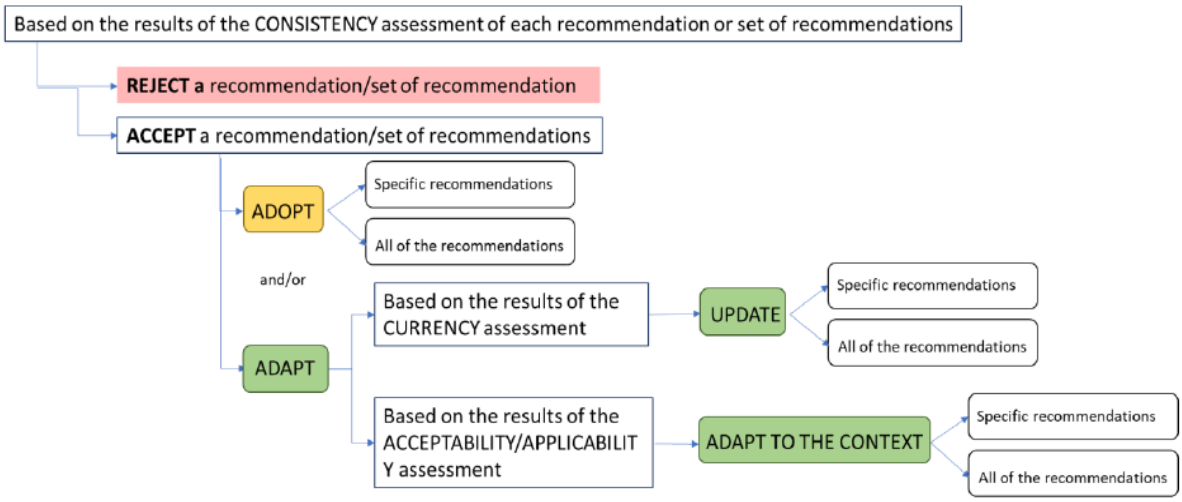

Figure A. *Decision making algorithm for the acceptance of a recommendation or set of recommendations.*

2.3. Guideline methodologic review

Appraisal of the Dutch Quality Standard for Anorectal Malformations

To inform a decision about the adoption or adaptation of the Dutch Quality Standard for Anorectal Malformations[2], an assessment of its quality and reporting was conducted.

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

Domain scores were calculated by summing the scores of all individual items within a domain and then 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 1**.

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.

Levels of Evidence and Grades of Recommendation

To improve the rigour of development, the AAWG and methodologists suggested that all new recommendations be accompanied by a Grade of Recommendation if applicable. Therefore, the working group chose to use an appraisal method that can be applied at the 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 2**).

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 2. *Correspondence between grades of recommendations and study level of evidence.*

To improve the reporting of the adapted recommendations with greater clarity, transparency, and explicitness, the AAWG and methodologists have provided this CPG in accordance with the RIGHT-Adapt preferred reporting items.[8]

2.4. Search strategy

The literature searches were conducted by a professional information specialist in the following databases: Ovid/Medline (PubMed), Cochrane CENTRAL, and Embase, to identify any new relevant studies published between 2017 and March 2023 (see **Appendix 2: Search strategy**). 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 the English language. To ensure that the report contained current and relevant evidence, studies published prior to 2017 were excluded.

The literature searches yielded a total number of 2,248 studies (after removal of duplicates), which were subsequently screened for title and abstract (see **Appendix 2: Results**). The screening was conducted by two methodologists (Willemijn Irvine and Konstantinos Mantzios). 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 3**.

Literature Selection Criteria

Inclusion
• Research covering Anorectal Malformations, Congenital AND (diagnosis), (treatment), (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 3. *Inclusion and exclusion criteria of publications*

After completing the screening of all titles and abstracts, decision discrepancies were discussed. In the first adaptation phase, two methodologists (Willemijn Irvine and Konstantinos Mantzios) and three paediatric surgeons (Jan-Hendrik Gosemann, Martin Lacher, and Ivo de Blaauw) screened and categorised the included publications into two topics: 1) Diagnosis and 2) Treatment. In the second phase, the included studies were categorised into four additional topics: 1) Lifelong follow-up and Integrated care, 2) Organisation of care, 3) Collaboration, referral and communication between care providers, 4) Transition care. Any results that did not match one of these predefined topics were excluded during the second screening round.

In total, 14 new studies were identified for the diagnosis and treatment modules (seven new studies for the diagnosis module and seven new studies for the treatment module), and 10 new studies were identified across the four follow-up and care coordination modules (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) adding value to the adaptation of the Quality Standard. A PRISMA flow diagram showing the literature selection process is available in **Appendix 2**.

3. CURRENCY, ACCEPTABILITY, APPLICABILITY AND CLARITY

3.1. Diagnostics

Key question 1

1. In patients with ARM, which diagnostic examinations are important for making the diagnosis, for evaluating the postoperative phase, and for evaluation during follow-up, and at what time should which examination be performed?

The key question consists of the following sub-questions:

a) Which diagnostics do you need to make a diagnosis of ARM and to determine which type of ARM is involved, and which diagnostics do you need to determine additional comorbidities?
b) Which diagnostic examinations are performed during and after the various surgeries, and which diagnostics are important during follow-up?
c) Which urological examination should be performed and when?
d) Which diagnostic exams of the reproductive system should be performed and when?
e) What are the indications for additional neurological examination?

Recommendations for sub-question 1a

For this sub-question, the panel included and considered seven new studies.[9-15] Six (6) recommendations were adapted, two (2) were adopted, and seven (7) 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 3.

PRENATAL		Grade of Recommendation
1	It is recommended to increase the awareness of ultrasound technicians/sonographers about the possible diagnosis of ARM before birth with prenatal ultrasound examination. **[adapted]**	B[9]
2	A dilated bowel with absence of the anal target sign (ATS) might be indicative of high ARM type. **[new]**	D
3	In case of an increased risk of abnormalities, it is recommended to perform an advanced ultrasound examination between 18 and 22 weeks of pregnancy. **[adapted]**	-
4	Sonographers must be aware of dilation of the last part of the colon/rectum. ARM may be suspected if the distal bowel is dilated on the first trimester ultrasound or if intraluminal echogenic foci are seen on the second trimester ultrasound. **[adopted]**	-
5	The visualization of the anal target sign is feasible by prenatal ultrasound and may aid in the detection of less severe ARM sub-types. **[new]**	B[10]
6	Identification of a peri-anal muscular complex during prenatal ultrasound (US) evaluation is useful for prenatal diagnosis but its sensitivity and specificity for ARM is low and a positive finding doesn't exclude low type ARM. **[new]**	C[11]
7	The possibility of a present cloaca should be suspected antenatally in case of a cystic mass in the pelvis that matches a hydrocolpos, and, in some cases, also by the presence of an expanded intestinal loop in the pelvis. Later in the pregnancy (more than 24 weeks) it is possible to visualize and assess the anal target sign. **[adapted]**	-

8	It is recommended to follow-up early (1st trimester) a cystic abdominal mass while maintaining a high index of suspicion for ARM even if the cystic mass disappears on follow-up scans, especially in isolated cases with normal genetics. **[new]**	D[12]
9	Foetal MRI may be considered in case of ultrasound identified hydrocolpos and suspected cloacal malformation. **[new]**	C[13]
NEONATAL		
1	In the acute phase, the first 24-48 hours after birth, ARM must be diagnosed by history and physical examination. **[adopted]**	-
2	In the diagnostic phase, the type of ARM (according to the Krickenbeck classification) and any additional anomalies should be carefully mapped out prior to any attempts to repair the ARM. Different diagnostic methods are available to map the phenotype but there is no evidence of superiority of one over the other. **[adapted]**	-
3	To qualify the condition, the digestive tract, the urogenital tract, the neuromuscular tract, the circulatory tract, and the respiratory tract must be or should be examined. In particular, disorders of the VACTERL association are also looked for: V = Vertebrae, anomalies of the vertebrae A = Anus, an anorectal malformation C = Cardiac, anomalies of the heart TE = Tracheoesophageal abnormality, occlusion or narrowing of the esophagus with possibly a connection to the trachea R = Renal, abnormality of the kidney, urinary tracts, or bladder L = Limb, limb abnormalities **[adapted]**	-
4	Spinal and renal ultrasound (US) in the first weeks of life are recommended to screen for spinal cord and urological anomalies. **[new]**	B[14]
5	In case of ARM with at least one other anomaly, consultation with a clinical geneticist is strongly recommended. **[new]**	B[15]
6	At the end of the diagnostic phase, the patient-specific abnormality must be clearly documented and saved throughout the life of the patient. Any digital data (diagnostic data) should also be saved. **[adapted]**	-

Table 4. *Recommendations for sub-question 1a.*

Justification for sub-question 1a

Prenatal

Recommendations have been made in favour of evaluating different signs and symptoms of ARM prenatally through ultrasound (US). The AAWG addressed that the probability of advanced US examination to diagnose ARM is enhanced when it is performed by highly skilled ultrasound specialists and at an academic centre.[2] Evidence from cohort studies and case series suggests that ARM may be suspected if the distal bowel is dilated or if intraluminal echogenic foci are seen on prenatal sonography.[9-12] US technicians/sonographers should be aware of these sonographic signs that may indicate a possible diagnosis of ARM. However, because the condition is rare, it will not often be diagnosed prenatally. Awareness among sonographers could be improved by

encouraging them to assess the ATS when checking the fetal sex.[9, 10] However, the AAWG referred to a study by Bischoff [10] concluding that the anal target sign (ATS) has the highest detection rate between 28-33 weeks, which should be taken into account since gender screening is predominantly done at an earlier term. The combination of a dilated bowel and the absence of the ATS may be indicative of a high ARM type; however, due to scarce evidence, its specificity has not yet been determined. In one of the newly included studies,[11] the authors reported that detection of the fetal anal canal and anal muscular complex may aid in the detection of anal atresia. The AAWG recognises this but wants to emphasise that the diagnostic accuracy appears to be low, and the presence of a normal anal muscular complex on US cannot exclude ARM. The AAWG members agreed that a cystic abdominal mass finding should raise suspicion, as it may be a cloaca or other ARM types in the differential diagnosis.[12] Sonographers should search for cysts in the foetal pelvis during the first and early second trimester. After considering a study on prenatal MRI,[13] the panel agreed that in cases of US findings that suggest ARM, a complementary MRI may be useful without being necessarily superior to US. Although the qualification of the screening technician may play a role in the quality of the examination, the AAWG supports that there is insufficient evidence to prove that US is inferior to MRI. MRI may add value in the diagnosis of ARM, but the consequences must be weighed in each case. Additionally, gynaecologists must be aware of the importance of diagnosing ARM during prenatal consultations.

The AAWG considered the favourable benefit-risk balance, the acceptability and applicability in order to adopt, adapt or to form new recommendations. Antenatal diagnosis of ARM seems to be accurate and does not lead to overdiagnosis and unnecessary concerns. In return, it could lead to improved counselling of parents and better planning of postnatal care. Prenatal counselling of parents and timely transfer (before birth to expert centres) could lead to parental and physician stress reduction and more adequate care for the newborn.

Neonatal

In the Dutch Quality Standard, this section is referred to as postnatal diagnosis.[2] However, the AAWG acknowledged that the included recommendations covered only the neonatal period; thus, the section was renamed accordingly. Recommendations have been made on the best methods of diagnosing ARM neonatally. The panel considered most recommendations on neonatal diagnosis to be acceptable and applicable, and only slightly modified the wording of the adapted recommendations to improve their clarity. More information regarding the diagnostic modalities for ARM can be found in the Dutch Quality Standard.[2]

Evidence from one (1) newly included study (diagnostic test accuracy) confirms that spinal US is an accurate method for helping in the diagnosis of spinal cord anomalies,[14] and the panel agreed that this is beneficial because it is a minimally invasive test that can provide important information. The clinicians pointed out that the provision of care in areas without specialisation would increase the risk of misdiagnosis, but the expertise exists within ERN eUROGEN.

The ARM type (according to the Krickenbeck classification) and any additional anomalies must be explicitly specified and documented. The AAWG considered mapping the phenotype an important part of neonatal diagnostics and recognises that there are multiple modalities to do so, including modern 3D measures, but that there is no evidence to recommend one modality over the other. It is preferable to offer screening by a clinical geneticist in case of ARM and coexisting anomalies.[15] The AAWG discussed the value of measuring the pouch-perineum distance in neonates with and without a fistula, and whether this can prevent an incorrect surgical procedure based on misdiagnosis of the type of ARM.[16] The AAWG pointed out that measuring the distance can be controversial, as there is no clear evidence that contributes to mapping the phenotype or planning the surgery. The group concluded that the pouch-perineum distance is not the most important marker; therefore, no recommendations are made on this measure.

3.1.1 Diagnostic examinations performed during and after surgeries and during follow-up

Sub-questions 1b and 1c

b) Which diagnostic examinations are performed during and after the various surgeries, and which diagnostics are important during follow-up?

c) Which urological examination should be performed and when?

Recommendations for sub-questions 1b and 1c

For these sub-questions, no new studies were included and considered. Two (2) recommendations were adapted, none were adopted or 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 4**.

	DURING and AFTER SURGERIES and DURING FOLLOW-UP	Grade of Recommendation
1	After reconstructive surgery the patient should be monitored closely (within a 3 week period). A diagnostic examination must be conducted on the function and integrity of the perineal reconstruction of the digestive tract and the urogenital tract. In case of stoma creation, proper stoma care is recommended. **[adapted]**	-
2	A possible follow-up (V)UD study will take place no earlier than 6 months after the reconstructive surgery, and only on indication (including persistent large residues or absence of spontaneous micturition postoperatively, dilation of the urinary tract postoperatively, follow-up of bladder dysfunction or vesicoureteral reflux established in previous (V)UD study, or tethered cord syndrome (TCS) follow-up). The indication for this must be determined during urological follow-up. **[adapted]**	-

Table 5. *Recommendations for sub-questions 1b and 1c.*

Justification for sub-questions 1b and 1c

Recommendations have been made on diagnostic examinations that should be performed both before and after surgery. The AAWG considered the favourable benefit-risk balance, as well as the acceptability and applicability, in order to adopt, adapt, or form new recommendations.

In the Dutch Quality Standard,[2] a diagnostic examination of the digestive tract and the urogenital tract to assess function and integrity was considered optional after reconstruction surgery or stoma creation. The AAWG proposed that this diagnostic examination and assessment should be mandatory within a 3-week period after the reconstructive surgery and within a 6-week period after a stoma creation.

Additionally, it is recommended that a possible follow-up (V)UD study be conducted no earlier than 6 months after reconstructive surgery, and only when indicated. The AAWG suggested that the indications must be determined by any means during urological follow-up.

3.1.2 Diagnostic exams of the reproductive systems

Sub-question 1d

d) Which diagnostic exams of the reproductive systems should be performed and when?

Recommendations for sub-question 1d

For this sub-question, no new studies were included and considered. Three (3) recommendations were adapted, four (4) were adopted, and none were developed de novo (new). All recommendations are reported in Table 6.

	REPRODUCTIVE SYSTEMS	Grade of Recommendation
1	Proper evaluation and documentation of the external and, if possible, internal genitalia of patients with ARM is required prior to, or at the time of reconstructive surgery. **[adopted]**	-
2	To optimise the results for the patient during a possible surgical intervention, in the case of abnormalities in the genitourinary area of patients with ARM, it is recommended to always consult a multidisciplinary team and document well their findings. Prior to the onset of puberty, a (paediatric) gynaecologist (preferably with expertise in the field of malformations) should assess and document the anatomical situation to optimize follow-up treatment in menarche/adolescence. **[adopted]**	-

3	A hydrocolpos (occlusion of the vagina/uterus with mucus build-up) may appear as a cystic swelling in the lower abdomen, especially in females with a cloaca, but can also be seen with an imperforate hymen. The filled vagina can compress the urethra and ureters and thus obstruct the flow of urine and cause hydronephrosis. In case of no or poor urination, careful examination must be done. A diagnosed hydrocolpos must be surgically relieved. Any hydronephrosis should be monitored closely in the context of a hydrocolpos in at least the first months of life. **[adopted]**	-
4	Attention is also recommended for the prevention of gynaecological anomalies in females with ARM as this can prevent (distal) vaginal stenosis, which can hinder tampon use and sexual contact. **[adopted]**	-
5	At the beginning of adolescence, females and males with ARM must be provided with information about the possibilities of problems in sexuality, fertility and reproduction. **[adapted]**	-
6	Females with ARM must be supervised and examined by a gynaecologist (preferably with experience in ARM and have access to a paediatric surgeon experienced in the field of ARM) during pregnancy. **[adapted]**	-
7	In females with a history of ARM and an abnormal perineum, it is recommended that the perineum be properly examined both pre-conceptually and during the entire pregnancy. An individual recommendation must be made on how the patient can give birth. When in doubt, the choice of Caesarean section should be strongly considered. **[adapted]**	-

Table 6. *Recommendations for sub-question 1d.*

Justification for sub-question 1d

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

In the original Quality Standard, recommendations have been made on the gynaecological examinations that should be performed for patients with ARM.[2] As suggested by the AAWG, the sub-question was reshaped, and the section has been renamed to 'Diagnostic exams of the reproductive systems'. By broadening the scope to encompass both male and female reproductive health, the group aimed for a more holistic approach to care, ensuring that patients receive thorough examinations and appropriate interventions tailored to their unique anatomical variations.

Additionally, the AAWG highlighted that it is of high importance to raise ARM patients' awareness of potential problems in sexuality, fertility and reproduction at the beginning of their adolescence and not at a later stage.

3.1.3 Indications for performing additional neurological examinations

Sub-question 1e

e) What are the indications for additional neurological examination?

Recommendations for sub-question 1e

For this sub-question, no new studies were included and considered. One (1) recommendation was adapted, two (2) were adopted, and none were developed de novo (new). All recommendations are reported in Table 7.

NEUROLOGICAL EXAMINATION		Grade of Recommendation
1	Spinal dysraphia (SD) can be an additional condition in patients with ARM and it is recommended that all patients with ARM must be screened. **[adopted]**	-

2	In addition to the radiological evaluation of the vertebra and sacrum, it is recommended to perform an ultrasound (US) of the spinal canal. The spinal canal US is performed shortly after birth, but at least in the first month, because otherwise the vertebral arches may close, and US of the spinal canal is less reliable to rule out anomalies. If there are abnormalities on the US of the spinal cord, follow-up is needed for symptoms, preferably with the help of a paediatric neurologist. **[adopted]**	-
3	If indicated, in case of neurological symptoms or abnormal findings on spinal US, an MRI can be conducted before the age of 12 months. Work-up: • All children receive US of the spinal cord within 1 month of birth. • A deteriorating neurological image (sensorimotor and/or continence) during follow-up can be an indication to consult a paediatric neurologist to evaluate the presence or not of a tethered cord (TC) or any other spinal anomaly. **[adapted]**	-

Table 7. *Recommendations for sub-question 1e.*

Justification for sub-question 1e

Recommendations have been made on the indications for performing additional neurological examinations in patients with ARM. The AAWG considered the favourable benefit-risk balance, as well as the acceptability and applicability, in order to adopt, adapt, or form new recommendations.

Patients with ARM may have associated congenital conditions, including tethered cord (TC) and spinal dysraphia (SD). Tethered spinal cord or other spinal anomalies occur in 10-60% of ARM patients.[15]

The AAWG suggested that an MRI can be conducted in cases of neurological symptoms and, if indicated, at the age of 12 months, rather than at an earlier age as recommended in the Dutch Quality Standard.[2] It is also advised that an ultrasound scan (US) of the spinal cord must be conducted within the 1st month of birth and not later. The panel did not recommend using an MRI as a first-line diagnostic in cases of a deteriorating neurological image during follow-up, as suggested in the original Quality Standard. Instead, a paediatric neurologist should be consulted to evaluate whether a TC or any other spinal anomaly is present.

3.2. Treatment

Key question 2

2. What (evidence-based and expert-based) treatment options are there for patients with ARM, and when do they apply?

The key question consists of the following sub-questions:

a) Which treatments are suitable for patients with ARM? Which reconstructive operations are performed, and which treatments are still needed afterwards?
b) What instructions are given to parents for actions after the reconstructive operations? Which healthcare provider gives the instruction, and when is this instruction given?
c) What is the work-up for the treatment of urinary and faecal continence problems and any associated complications?
d) What individually adaptable protocol can be followed for toilet training, and at what age is this offered?
e) What are effective prevention and treatment options for diaper dermatitis?
f) Which aids are available (incontinence materials, rinse aids)? Which care provider can advise on this?
g) How is/are the parents of the patient with ARM supported in self-management?

Sub-question 2a

Which treatments are suitable for patients with ARM? Which recovery operations are performed, and which treatments are still needed afterwards?

Recommendations for sub-question 2a

For this sub-question, six (6) studies were included and considered.[17-22] Six (6) recommendations were adapted, none were adopted, and six (6) were developed de novo (new). All recommendations are reported in **Table 8**. A summary of all newly included studies for this sub-question is available in **Appendix 5**.

TREATMENT MODALITES		Grade of Recommendation
1	Refer patients with an ARM diagnosis to a paediatric surgery department when available. The following measures are recommended: • Patient examination in the first 24 hours on whether there is meconium discharge and which type of ARM is involved. • Exclusion of other potentially lethal pathology, especially congenital heart disorder. • Intravenous fluid management in case of bowel obstruction or enteral nutrition if defecation is possible. • Parental counselling on the condition. **[adapted]**	-
2	Patients with cloaca and cloacal exstrophy should be referred to a reference centre with experience in the multidisciplinary treatment of these patients and long-term follow-up. **[new]**	D[17]
3	In patients with insufficient or absent meconium discharge, establish a venous access and insert a nasogastric tube. Provide IV antibiotics. Inform the parents not to feed the baby or child. **[adapted]**	-
4	For forms of ARM in which there is no meconium discharge after birth, surgery is performed. Often, three surgeries are required: • Creating a colostomy • The reconstruction surgery • Reversal/closure of the stoma **[adapted]**	-
5	If there is a phenotype of ARM in which (sufficient) meconium is discharged (e.g. ARM with perineal or vestibular fistula or anal/rectal stenosis), the anus should be calibrated and/or painlessly dilated until the daily evacuation of meconium is possible. If there is no sufficient meconium evacuation, a stoma should be considered. **[adapted]**	D[18]
6	Choose the timing of reconstructive surgery (either in the neonatal period or during infancy) based on the height of the rectum and the presence of a connection to the urinary tract. **[adapted]**	-
7	Primary reconstruction and restoring bowel continuity should be done within the first year of life, unless there are contraindications for surgery such as life-threatening comorbidities (e.g., cardiac). **[new]**	C[19]
8	In certain cases of perineal fistula, non-operative treatment might be considered. **[new]**	B[20]
9	Type of colostomy: No evidence for superiority between either loop vs split. Place: Descending sigmoid stoma is preferred but, in some cases, (cloacal malformations or associated gastrointestinal anomalies) a transverse stoma might be an option. **[new]**	D

10	The chosen surgical technique depends on the abnormalities found and the surgeons' expertise. The most common surgical techniques are the application of an anoplasty, ASARP, PSARP, LAARP (ASARP: anterior sagittal anorectoplasty; PSARP: posterior sagittal anorectoplasty; LAARP: laparoscopically assisted anorectoplasty), or a combination of the different techniques. **[adapted]**	C[21]
11	In case of anal stenosis at the skin level during follow-up after anorectoplasty, anal dilatation under general anaesthesia should be considered, otherwise a Heineke-Mikulicz anoplasty (HMA). **[new]**	B[22]
12	Postoperative routine calibrations at home may be considered but are not mandatory. Painful calibrations must by all means be avoided. If a calibration programme is started, it should not last longer than 3-4 months, and intensive follow-up is needed. **[new]**	D

Table 8. *Recommendations for sub-question 2a.*

Justification for sub-question 2a

Recommendations have been made regarding the various treatment modalities for patients with ARM. The AAWG considered the favourable benefit-risk balance, as well as the acceptability and applicability, in order to adopt, adapt, or form new recommendations.

The AAWG suggested, based on evidence found in a study by Kubota et al.,[17] that patients with cloaca and cloacal exstrophy should be referred to a specialised treatment centre with expertise in multidisciplinary care and long-term follow-up.

The AAWG agreed that the treatment of ARM depends on the type of malformation, sex, weight, possible comorbidities, and time or age at diagnosis. To make particular recommendations regarding initial treatment options, the AAWG considered it necessary to assess the presence and degree of meconium discharge. When absent or insufficient, the group noted that calibration should not be started until daily evacuation is possible.[18]

Depending on the local surgical expertise and the condition of the neonate, primary reconstruction and restoration of bowel continuity are feasible within the first year of life and should be performed within this period, unless there are contraindications for surgery, such as life-threatening comorbidities.[19]

According to a study by Amerstorfer et al.,[20] evidence supports considering non-operative treatment in some cases of perineal fistula (the anal orifice surrounded by 80% of the anal sphincter and no stenosis); thus, the group formed a recommendation in favour of this treatment method.

Regarding the surgical techniques, the AAWG did not recommend single-stage over multi-stage surgery approaches, as evidence was mainly limited to females with recto-vestibular fistulae and was considered insufficient to make a general statement.[23] The group acknowledged that this is a complex issue, with an ongoing debate. The AAWG considered it essential to take into consideration the surgeons' expertise when choosing the specific method, in addition to the patient's abnormalities and/or comorbidities, as noted in the concluding statement of Lauriti et al.[21] Among the updated evidence, the group observed a lack of long-term data on functional outcomes and insufficient data on cosmetic appearance or patient satisfaction. Given this, the group could not make any specific recommendations on surgical techniques for ARM patients with sufficient confidence.

In addition, the necessity of anal calibration for prevention of anal stenosis was discussed together with the results of an RCT on routine postoperative dilatation.[22] The results of the RCT indicated that routine postoperative dilatation did not lead to fewer skill level strictures, but these results were imprecise. The AAWG considered this a very sensitive topic, where there is an ongoing debate on whether to dilatate, for which higher-level evidence is warranted. The panel unanimously agreed that the dilatations should be painless by any means, and the benefits and harms of performing this should be carefully discussed with parents. In case of painful anal calibrations the group pointed out that they should be performed exclusively under general anaesthesia as an alternative treatment.[22] The Heineke-Mikulicz anoplasty (HMA) was presented by Ahmad et al.[22] as an alternative to routine dilatations and was found to be a viable treatment option in case of stricture at the skin level; thus, it was included in the recommendations.

The AAWG emphasised that not all surgical methods for treating ARM are viable in every healthcare centre. In these cases, a nonoperative management should be considered; patients can remain with a colostomy without reconstruction and be referred to a centre specialising in ARM treatment.

Finally, the AAWG suggested that when surgeons select a surgical technique, they should consider that perineal wound infections are usually limited and do not affect long-term functional outcomes. However, in case of extended and destructive wound infection, a colostomy may be required, and the final functional outcome, in this case, may be affected.

3.2.1 Postoperative instructions to parents: which, from whom, and when

Sub-question 2b

b) What instructions are given to parents for actions after the reconstructive operations? Which healthcare provider gives the instruction, and when is this instruction given?

Recommendations for sub-question 2b

For this sub-question, no new studies were included and considered. Two (2) recommendations were adapted, one (1) was adopted, and none were developed de novo (new). All recommendations are reported in **Table 9.**

	INSTRUCTIONS TO PARENTS	Grade of Recommendation
1	Paediatric surgeons and nurses must inform parents about available ARM resources such leaflets, brochures, website, and webinars on the neonatal aspects of ARM in an understandable way (i.e., lay language) etc., as well as about patient associations. **[adapted]**	-
2	The paediatric surgeon should provide relevant information to parents whose children undergo ARM surgery about conditions, treatment, and care after surgery and possible complications. **[adopted]**	-
3	A specialised nurse in ARM should always be involved after the operations of a child receiving a stoma. **[adapted]**	-

Table 9. *Recommendations for sub-question 2b.*

Justification for sub-question 2b

Recommendations have been made regarding the instructions to be given by which healthcare provider and when to parents after reconstructive operations. The AAWG considered the favourable benefit-risk balance, as well as the acceptability and applicability, in order to adopt, adapt, or form new recommendations.

The AAWG considered the provision of accurate information among the most important things for parents with a child with ARM and therefore adapted the recommendations accordingly. The AAWG suggested that if there is no specific patient association for ARM in their own country, parents should link to other existing international patient organisations for support. Compared to the original Quality Standard, the recommendations were adapted to be more generally applicable throughout European countries.

3.2.2 Toilet training and treatment of urinary and faecal continence problems

Sub-questions 2c and 2d

c) What is the work-up for the treatment of urinary and faecal continence problems and any associated complications?

d) What individually adaptable protocol can be followed for toilet training, and at what age is this offered?

Recommendations for sub-question 2c and 2d

For these sub-questions, one new study was included and considered. Nine (9) recommendations were adapted, one (1) was adopted, and none were developed de novo (new). All recommendations are reported in **Table 10.**

TOILET TRAINING		Grade of Recommendation
1	It is crucial to strive for urinary and faecal continence. If continence cannot be achieved, the highest possible quality of life should be aimed for (e.g., by facilitating pseudo-continence with bowel management). **[adapted]**	-
2	It is important to keep in mind at all times the patient's course, the association between ARM and urological malformations, and to act accordingly, involving the relevant healthcare professionals. **[adapted]**	-
3	An individually tailored protocol according to the child's ARM sub-type and form is recommended for toilet training. This is, on the one hand, to be done in connection with the different degrees and forms of ARM and comorbidity, and on the other hand, in connection with the differences per child in maturity and development phase. **[adapted]**	-
4	In case of problems with toilet training (unsuccessful management, whether or not related to complex problems) it is recommended that the toilet training problem should be discussed and treated within a multidisciplinary team. **[adopted]**	-
5	The approach to toilet training in children with ARM, depending on complexity, comorbidity, and social context, can be initiated at the age of 1-5 years (as with all children). **[adapted]**	-
6	It is recommended during faecal toilet training to pay attention to • care of the perineal skin • toilet posture, toilet seat reducer, relaxation of the child during toilet use, using fixed times on the toilet • paediatric pelvic floor physiotherapy • optional supervision by a psychologist/remedial educationalist **[adapted]**	-
7	Patients/parents should be educated to recognise constipation and subsequent overflow incontinence. **[adapted]**	-
8	Bowel incontinence may require an enema or rectal irrigation, which could be temporary. **[adapted]**	-
9	If rectal irrigation is not working or not possible (e.g., rectal trauma, physical inability), antegrade irrigation should be considered. **[adapted]**	-
10	In case of persistent and specific problems of incontinence, re-do surgery may be considered, in coordination with an experienced surgeon or centre. If redo-surgery is not recommended, definite stoma may be considered as a last resort. **[adapted]**	C[24]

Table 10. *Recommendations for sub-question 2c and 2d.*

Justification for sub-question 2c and 2d

Recommendations have been made for toilet training. The AAWG considered the favourable benefit-risk balance and adapted all recommendations. Changes were made to the original Quality Standard to improve the clarity of the statements and adapt them to the European context.

The panel recommended implementing a personalised protocol for toilet training that takes into account the specific subtype of ARM in children. Furthermore, this protocol should also consider any associated comorbidities, as well as individual differences in maturity and developmental stages. The toilet training strategies should be flexible and adaptable, taking into consideration several aspects of ARM and the individual variations in each child's growth and development. The AAWG also considered that

the social context of a child with ARM is an additional factor that should be taken into account in the optimal approach to toilet training. If incontinence or relevant challenges persist (e.g., a failed reconstructive surgery), the group indicated that re-do surgery should be considered, in coordination with an expert centre.[24]

3.2.3 Prevention and treatment of diaper dermatitis

Sub-question 2e

e) What constitutes good prevention and treatment for diaper dermatitis?

Recommendations for sub-question 2e

For this sub-question, no new studies were included and considered. One (1) recommendation was adapted, none were adopted, and none were developed de novo (new). All recommendations are reported in Table 11.

DIAPER DERMATITIS		Grade of Recommendation
1	Wound, stoma and perianal care should be monitored by a skin and/or stoma nurse in or out of the hospital on regular basis. **[adapted]**	-

Table 11. *Recommendations for sub-question 2e.*

Justification for sub-question 2e

One recommendation has been made regarding the prevention and treatment of diaper dermatitis in patients with ARM. The AAWG recognised the need for professional medical supervision to ensure proper care and management, thereby preventing diaper dermatitis. As a result, the panel recommended involving a skin and/or stoma nurse on a regular basis for wound, stoma, and perianal care, whether in a hospital setting or not.

3.2.4 Information about available products to aid incontinence

Sub-question 2f

f) Which aids are available (incontinence materials, rinse aids)? Which care provider can advise on this?

Recommendations for sub-question 2f

For this sub-question, no new studies were included and considered. Two (2) recommendations were adapted, none were adopted, and one (1) was developed de novo (new). All recommendations are reported in Table 12.

AVAILABLE AIDS		Grade of Recommendation
1	Patients should be provided with good information about incontinence materials and rectal irrigation products, ideally in collaboration between a specialised ARM nurse and a physician or surgeon. **[adapted]**	-
2	Patients and parents should be informed of additional aids for incontinence, together with their limitations. **[adapted]**	-

3	It is recommended that parents and/or patients contact their ARM patient associations for further support. **[new]**	-

Table 12. Recommendations for sub-question 2f.

Justification for sub-question 2f

Recommendations have been made regarding the available aids, materials and products that can be used in the care of ARM patients. The AAWG considered the favourable benefit-risk balance, as well as the acceptability and applicability, in order to adopt, adapt, or form new recommendations.

There are all kinds of incontinence materials. The aids a patient with ARM needs depend, among other things, on the type of incontinence and the extent of urine and/or stool loss. Based on an intake questionnaire, the specialised nurse will inform and help parents choose the right aids.

The AAWG considered that the availability of aids, such as incontinence materials, rinse aids, and reimbursement options, may differ substantially between European countries. Therefore, the recommendations were either modified or rejected, and the respective sub-question was adjusted accordingly.

3.2.5 The support of parents with ARM patients in self-management

Sub-question 2g

g) How is/are the parents of the patient with ARM supported in self-management?

Recommendations for sub-question 2g

For this sub-question, no new studies were included and considered. Three (3) recommendations were adapted, one (1) was adopted, and one (1) was developed de novo (new). All recommendations are reported in Table 13.

AVAILABLE AIDS		Grade of Recommendation
1	Refer patients with ARM to patient associations for support and education about self-management of the disease, or to other patients for peer learning. **[adapted]**	-
2	Include in the patient information material an up-to-date point of contact or a named person to whom patients can reach out to when needed, for 24/7 information and/or support. **[adapted]**	-
3	It is recommended to work jointly with (the parents of) a patient with ARM to develop a care plan to reach personal treatment goals. **[adapted]**	-
4	Healthcare professionals should document information about agreed treatment goals in the medical records and/or individual care plans. **[adopted]**	-
5	It is recommended to review the treatment goals and care plan periodically. **[new]**	-

Table 13. *Recommendations for sub-question 2g.*

Justification for sub-question 2g

Recommendations have been made regarding the support that the parents receive in the self-management of their children with ARM. The AAWG considered the favourable benefit-risk balance, as well as the acceptability and applicability, in order to adopt, adapt, or form new recommendations.

The AAWG agreed with most recommendations and primarily made adjustments to the wording to arrive at a set of recommendations applicable to all European countries. The AAWG discussed the importance of periodically reviewing treatment goals, which is just as crucial as setting the goals; therefore, the group formed a new recommendation. Care providers are responsible for thorough documentation of the treatment plan, and the panel members agreed that the point of contact does not need to be a specific person, as long as it is clear to everyone who takes on this role. ARM patient associations can provide additional (psychological/social) support to parents and assist them in the management of their child's condition.

3.3. Lifelong follow-up and integrated care

Key question 3

3. What is the optimal follow-up for people with an anorectal malformation (ARM) in an expertise centre/paediatric surgery centre?

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 lifelong follow-up and integrated care

Seven (7) new studies were included and considered. Twelve (12) recommendations were adapted, none were adopted, and three (3) were developed de novo (new). All recommendations are reported in **Table 14**. A summary of all newly included studies for this sub-question is available in **Appendix 6.**

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/paediatric surgery centre 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 programme. See: Table 3.1. in the Dutch Quality Standard[2] **[adapted]**	-
3	The organised follow-up programme should be carried out by a paediatric 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[25]
5	It is recommended to monitor quality of life during follow-up with standardised general or disease-specific questionnaires. **[adapted]**	D[26]
6	Scar morbidity and symptoms should be addressed during follow-up. If necessary, a plastic surgeon can be consulted. **[new]**	C[27]

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[28]
9	The parents of the child with ARM should 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[29]
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[30]
11	An experienced nurse should be involved in the post-operative care. **[adapted]**	-
12	Nutrition growth and neuromuscular development are important considerations during all follow-up. **[adapted]**	-
13	(Neuro) psychological follow-up is recommended when indicated in children with ARM (poor academic performance or potential shorter attention span, possibly due to physical problems). **[adapted]**	C[31]
14	Gynaecological follow-up is recommended when indicated 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 (US) of the lower abdomen must be performed during adolescence around the time of the first menarche (on average more than a year after 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 to the upper urinary tract and to evaluate the function of the detrusor and the sphincter. **[new]**	D[32]

Table 14. *Recommendations for lifelong follow-up and integrated care for patients with ARM.*

Justification for lifelong follow-up and integrated care

The AAWG considered the favourable benefit-risk balance, as well as the acceptability and applicability, in order to adapt or form new recommendations. Recommendations have been made in favour of the accessibility of the ARM patients in an expertise centre for ARM and of a structured lifelong follow-up programme 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 centre for ARM/paediatric surgery centre 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., [25, 26] 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. According to a study by Tofft et al.,[27] healthcare providers are recommended to track and report scar morbidity and symptoms during post-treatment follow-up. The Patient and Observer Scar Assessment Scale (POSAS) is a suggested tool for this purpose.[33] A plastic surgeon may be consulted if necessary.

A discussion was held on the quality of life and mental health of ARM patients, as well as their access to this information. The group suggested that the respective recommendation should be incorporated into the lifelong follow-up module rather than the treatment module, as per the Dutch Quality Standard.[2] It was considered crucial that the involved healthcare professionals refer patients with ARM and their parents to patient organisations, while providing them with the necessary guidance to promote 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.[30]

The group unanimously agreed to incorporate and adapt the recommendations (10, 11) initially found in the Dutch Quality Standard's *'Treatment for a child with ARM'* module in this section, aiming to form a more comprehensive module with fewer overlaps in recommendations.[2]

A recommendation was formed regarding the significance of nutrition, growth and neuromuscular development during all follow-ups. 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.,[34] 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 was not convinced that they added value as these questionnaires are not validated for the purpose of bowel follow-up. The panel agreed that probably a majority of patients have 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[35]. The panel, therefore, agrees that the possible benefits for clinical decision-making do not outweigh the burden for patients, which is reflected in the fact that manometry is not commonly practised by 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 are 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.,[31] it is suggested that children with ARM should undergo (neuro) psychological follow-up as warranted, such as when they experience academic challenges or display a shorter attention span, potentially stemming from physical issues. The AAWG recommended gynaecological follow-up in females with ARM when indicated, particularly those with a cloacal malformation or other known gynaecological issues, and therefore adapted most recommendations on this from the Dutch Quality Standard. This is important for detecting abnormalities and addressing any clinical symptoms in a timely manner. If any complaints arise during adolescence, an ultrasound (US) of the lower abdomen should be done around the time of menarche. Following this, an abdominal US 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 referred to the EAU guidelines on urological infections and recommended urodynamic assessments for all patients with spinal cord dysraphism (SCD) and for children who exhibit signs of a neurogenic bladder.[32] 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 requires special attention.[36]

3.4. Organisation of care

Key question 4

4. Which care provider(s) are responsible for good care, coordination, and management 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 (paediatric surgical) care centre?
b) Which disciplines must be consulted about indications?
c) What is the role of the expertise centre, the specialised surgical treatment centre, 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 organisation of care

For the organisation of care module, two (2) new studies were included and considered. Five (5) recommendations were adapted, four (4) were adopted, and one (1) was developed de novo (new). All recommendations are reported in **Table 15**. A summary of all newly included studies for these sub-questions is available in **Appendix 7.**

	ORGANISATION OF CARE	Grade of Recommendation
1	Hospitals treating patients with ARM should have easy access to the following healthcare providers: • Paediatric surgeon • Paediatrician • Nurse practitioner / case manager pediatric surgery • (paediatric) Anesthesiologist • (paediatric) Cardiologist • Paediatric urologist • (paediatric) Neurologist • (paediatric) Neurosurgeon • Nurse practitioner in urinary incontinence, materials, ICC and stoma care • Dietitian • General practitioner • Physiotherapist • (paediatric) Gastroenterologist • (paediatric) nephrologist • Clinical geneticist • Social worker • Neonatologist • (paediatric/youth) Psychologist • (paediatric) Radiologist • Colorectal Surgeon • (paediatric) Orthopedic Surgeon • (paediatric) Cardiac surgeon • (paediatric) Gynaecologist • High risk pregnancy specialist • Urologist/andrologist • Sexologist or other provider that can help with sexual issues. • School counsellor A MDT must be tailored to the needs of the patients and local care agreements. **[adapted]**	B[37]
2	It is recommended that expertise centers for ARM should manage ≥ 10 (reconstruction) cases per year/center within multidisciplinary teams. **[new]**	B[38]
3	The examinations, decisions, and conversations with the parents of children with ARM take place in a paediatric surgical center with expertise in ARM. **[adapted]**	-
4	Within the expertise center/paediatric surgery center, attention should also be paid to the transition of care for adolescents. **[adopted]**	-
5	In cases of shared care, the paediatric surgical centre with expertise in ARM remains responsible for care coordination**.** The specialist centre/paediatric surgery team examines what care is needed and whether this can best be offered in the expertise centre/paediatric surgery team or in the patient's own region. **[adapted]**	-
6	The expertise center/paediatric surgery team 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 centres and regional	-

	paediatric (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 centre. **[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 15. *Recommendations for organisation of care for ARM.*

Justification for the organisation of care

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

The panel agreed that in order for a healthcare centre to be recognised as an ARM expert facility, it should offer easy access to a wide range of medical specialities within a multidisciplinary team and perform at least 10 ARM reconstructions annually.[37, 38] Although the AAWG pointed out that the number of 10 (reconstructive) cases per year might not be feasible for every expertise centre across EU member states, this recommendation was made because the panel agreed that centralisation is a crucial step towards improving the quality of ARM care.

In shared care, the Dutch Quality Standard recommends that the expertise centre in ARM, along with the patient's relatives/caregivers, have control over the integrated care chain.[39] However, the AAWG advised that the expertise centre 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 centre or in the care recipient's own region.

Furthermore, the group emphasised the importance of centralisation, bilateral exchange of information, and collaboration in shared care between the ARM expertise centre and other Healthcare Providers (HCPs), thereby facilitating patients' access to care in their local region.

3.5. Collaboration, referral and communication between care providers

Key questions 5 and 6

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

6. Which measures/steps must be taken to achieve good cooperation between care providers and referral to care providers (from primary, secondary, and/or 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 care provider refers the patient with ARM to the right care provider at the right time?

Recommendations for collaboration, referral and communication between care providers

For this module, no new studies were included and considered. Seven (7) recommendations were adapted, three (3) were adopted, and one (1) was developed de novo (new). All recommendations are reported in **Table 16**.

	COLLABORATION, REFERRAL AND COMMUNICATION BETWEEN CARE PROVIDERS	Grade of Recommendation
1	The supervising physician (SP) and/or the care coordinator starts providing information to the 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 paediatric surgeon, the paediatrician and the specialised nurse in ARM and/or nurse practitioner are present during the multidisciplinary consultation. The SP is often a paediatric surgeon**. [adapted]**	-
6	The SP ensures the communication/cooperation (internal and external) between the involved care providers and with the patient (and the parents). **[adapted]**	-
7	The SP informs the general practitioner (GP) and (if involved) the paediatrician in the regional hospital about the ARM condition, including the possible complications and in which situations the SP or the care coordinator of the peadiatric surgery center should be contacted. The GP or the paediatrician at the regional hospital contacts the SP of the paediatric surgical centre 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 Centre/surgical treatment center, but also in a hospital or other treatment center close to home. The local hospital/local care provider works together with the EC/surgical treatment center. **[adapted]**	-
11	Countries with a patient organisation should organise annual meetings for patients. In case there is no patient organization, the Expertise Centre should coordinate a yearly meeting. **[new]**	D

Table 16. *Recommendations for collaboration, referral and communication between care providers.*

Justification for collaboration, referral and communication between care providers

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

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

During the panel discussion, emphasis was placed on promoting effective communication and cooperation between the involved healthcare providers (HCPs) and other care providers within the expertise centre and outside. The Supervising Physician (SP) is responsible for coordinating this collaboration and ensuring efficient communication between patients and their parents.

Furthermore, the AAWG addressed that hospital exchanges of patients with ARM may pose risks. Therefore, the allocation of tasks and responsibilities has 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 given to 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 Centre should organise an annual meeting for families, allowing peer support to be established.

3.6. Transition care

Key question 7

7. 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 transition care

For the transition of care module, one (1) new study was included and considered. Six (6) recommendations were adapted, six (6) were adopted, and three (3) were developed de novo (new). All recommendations are reported in **Table 17**. A summary of all newly included studies for this sub-question is available in **Appendix 8.**

TRANSITION CARE		**Grade of Recommendation**
1	The nurse practitioner (case manager) and paediatric surgeon ensure a smooth transition from childhood to adulthood by preparing young people and their parents. The paediatric 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 centres 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 the transition process. **[new]**	D
5	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 centres/paediatric surgery centres must take responsibility for the transition of patients with ARM from care by the paediatric specialists to the adult equivalent (surgeon, urologist, etc.). The ideal transition seems to consist of the involvement of both a paediatric specialist and an adult specialist, resulting in a collaboration between the two in which easy contact and consultation is possible. **[adopted]**	C[40]
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[41]
8	Good cooperation and communication between the gastroenterologist / surgeon / urologist / gynaecologist where the adult patient is being treated, and the paediatric surgeon / paediatric gastroenterologist / paediatric 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 in creating a familiar environment that results in patients with ARM wanting 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, attention is also paid to: the 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 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 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 17. *Recommendations for transition care for patients with ARM.*

Justification for transition care

The AAWG considered the favourable benefit-risk balance, as well as the acceptability and applicability, in order to adopt, adapt, or 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, not only enhancing their overall care but also promoting a sense of ownership and responsibility for their own health.

Furthermore, to ensure continuity of care, the panel suggested implementing a structured transitional programme in treatment centres that provide care for ARM patients. By doing so, a structured transitional programme 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.

Transition planning is recommended to begin at the age of 12-14 years, but can vary individually. The panel was unable to determine a specific age for the transition to adult care due to the varying conditions that apply in each European Union (EU) member state. However, they emphasised that the transition should be completed before the age of 18.

Evidence from a retrospective single-centre study by Acker et al. [40] supported the recommendation that care-providing expertise centres and paediatric surgery centres have a responsibility to ensure a seamless transition for patients with ARM as they move from paediatric care to adult care. This process should involve collaboration between the paediatric specialist and an adult specialist to promote effective communication and consultation.

Throughout the transition phase, a smooth transfer to a new adult care physician will occur. Special attention should be given to this aspect to ensure the smooth 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.[41]

Follow-up for adults is not yet well-organised everywhere, and many adolescents do not attend 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. 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 on the transition of care through conferences and establishing connections between paediatric and adult healthcare providers at 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 remain informed and ensure that patients with ARM have easy access to preconception care.

4. IMPLEMENTATION, VALIDITY AND EVALUATION

4.1. Implementation

During the various phases of adapting the Dutch Quality Standard[2], the implementation of the Clinical Practice Guideline (CPG) and the practical feasibility of the recommendations have been considered. In doing so, explicit attention was paid to factors that could promote or hinder the implementation of the CPG in practice.

The panel highlighted that varying resources and priorities in different EU/EEA 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 CPG. Furthermore, variations in regulatory frameworks, legal systems, and knowledge gaps among healthcare professionals, policymakers, and the general public also pose barriers to implementing a CPG across Europe.

In addition to the challenges mentioned, the panel suggested several ways to promote widespread adoption of the CPG. Because ERN eUROGEN is a network consisting of healthcare providers across the EU/EEA, panel members can actively contribute by sharing the CPG in their respective countries and advocating for its implementation. Local webinars or workshops can be organised to ensure that knowledge spreads effectively to healthcare providers who may have limited access to educational resources about the CPG. Furthermore, the AAWG has published topic-specific articles in the European Journal of Pediatric Surgery to disseminate information about the CPG.[47-51] Moreover, ERN eUROGEN organises European webinars on a regular basis, some of which are dedicated to promoting awareness of ARM among healthcare providers.[52] Additionally, translating the CPG into different languages could facilitate its implementation by healthcare professionals who may not be fluent in English.

4.2. Validity

This CPG is valid from the moment of publication until the publication of its update. Generally, guidelines are evaluated every five years. Consequently, the Strategic Board of ERN eUROGEN will establish a new working group in 2028 to assess which modules require updates and whether new modules should be developed based on the latest insights.

5. DISCUSSION

The purpose of this project was to adapt the Dutch Quality Standard for patients with Anorectal Malformation[2] into a Clinical Practice Guideline (CPG) tailored to the European context. By doing this, the AAWG aimed to establish a framework that would effectively guide healthcare providers (HCPs) in delivering standardised, high-quality care for patients with ARM throughout Europe, and thus improving overall diagnostic, treatment, and long-term care outcomes by ensuring equitable access to care.

The CPG covers the following thematic modules: 1) Diagnosis, 2) Treatment, 3) Lifelong follow-up and Integrated care, 4) Organisation of care, 5) Collaboration, referral and communication between care providers, and 6) Transition of care. The AAWG considered the favourable benefit-risk balance, as well as the acceptability and applicability, in order to adopt, adapt, or develop new recommendations.

During the process, areas of consensus were identified, as well as areas of ambiguity that require further research. Upon reviewing the existing literature, the AAWG confirmed that the majority of studies lack sufficient evidence, being limited to retrospective reviews and case series. Important areas of consensus and ambiguity, along with their research implications, are addressed below.

5.1. Diagnostics

The panel did reach consensus on the benefits of improving prenatal ultrasound (US) diagnostics. Additionally, the panel emphasised that a complementary MRI may add value in the prenatal diagnosis of ARM,[13] but the consequences have to be assessed in each case, and further research is required to address the benefit-risk balance of the prenatal MRI. The AAWG emphasised that sonographers' awareness should be improved by assessing the anal target sign (ATS), among other factors, especially during the 28-33 weeks of the prenatal phase.[9, 10] The AAWG was able to form a recommendation, suggesting that a combination of a dilated bowel and the absence of the ATS might be indicative of a high ARM type. However, it was claimed that further research should be conducted to enhance the quality of evidence. Furthermore, sonographers are encouraged to search for cysts in the foetal pelvis during the first and early second trimester, since a cystic abdominal mass finding could raise suspicion for a cloaca or other ARM types.[12]

The panel strongly agreed that neonatal ultrasound (US) screening examinations for the urinary tract, spinal cord (SC), and heart within the first month of life should be mandatory and comprise the gold standard. Screening for malformations within the VACTERL association, as well as genital and gastrointestinal malformations, should be performed and documented in both the medical chart and the individual patient's information. In cases where at least one other concomitant malformation is present, a referral to a clinical geneticist or genetic screening should be considered.

The panel also reached consensus on broadening the section consisting of gynaecological diagnostic examinations by renaming it '*diagnostic exams of the reproductive systems*'. By doing so, the group aimed to broaden the scope, ensuring that patients receive thorough examinations tailored to their unique anatomical variations, and also include males. Future research should focus on providing insights for the diagnostic examinations of all genders and their respective reproductive systems.

5.2. Treatment

The panel did not reach consensus on a preferred surgical technique for patients with ARM. There are multiple surgical modalities used for treating ARM, depending on the abnormalities and comorbidities found in each individual patient or the surgeon's expertise. Further investigation is necessary through clinical trials in ARM expertise centres to assess the effectiveness of various treatment modalities and make specific recommendations for (subgroups of) patients.

There was consensus on the optimal timing of primary reconstruction and restoring bowel continuity. Unless there are contraindications such as life-threatening comorbidities, the panel agreed that the surgical procedure should be conducted within the first year of life.[19] Importantly, a careful preoperative diagnostic work-up should be prioritised in rare and complicated ARM subtypes.

Furthermore, the AAWG unanimously agreed that certain questions from the original Quality Standard, related to the treatment module, had to be modified or rejected. These questions were deemed unsuitable for inclusion in a European Clinical Practice Guideline (CPG) due to heterogeneity in healthcare practices and policies across Europe. For these reasons, no appropriate recommendations could be formed for the management of frequent urinary tract infections. If recommendations are updated in the future, the evaluation of best practices could be more thorough if specific PICO questions and the GRADE methodology are applied.

5.3. Lifelong follow-up and integrated care

Limited evidence suggests that scar morbidity is not explicitly documented during the follow-up of ARM patients.[11] The AAWG acknowledges the need for further research into this matter, including systematic reviews. Furthermore, gathering indirect evidence from other patient groups could add value to 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 faecal incontinence classification and suggested further research for a comprehensive scoring system.[18] The AAWG 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, and Groningen Defecation and Fecal Continence Score.[42-45]

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 the literature,[46] 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.

5.4. 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 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 a consensus that each ARM expertise centre should be responsible for at least 10 ARM reconstructions per year involving multidisciplinary teams.[38] The establishment of minimum procedural volumes will ensure that centres 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.

5.5. Collaboration, referral and communication between care providers

The AAWG emphasised that there is a lack of evidence in the current literature regarding collaboration, referral, and communication among ARM care providers. The AAWG emphasised the importance of addressing these gaps and exploring the barriers and facilitators that affect collaboration and communication between care providers in ARM.

To address this issue, further research is needed to gain a deeper understanding of the challenges faced by ARM care providers in care collaboration and communication. Future studies should aim to identify the obstacles that hinder efficient teamwork, with the goal of developing interventions to overcome these obstacles 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.

5.6. 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, as well as quality of life. A well-structured transition programme 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 programme 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 to 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 programme 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 programme is conveyed effectively, increasing patient engagement and adherence.

Overall, a well-structured transition programme 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 on the transition care of patients with ARM. Meanwhile, consulting other already established transition programmes should be encouraged.

6. CONCLUSION

ERN eUROGEN aimed to form a common Clinical Practice Guideline (CPG) on ARM that applies in each of the EU/EEA countries.

A set of recommendations was developed for the following thematic modules: 1) Diagnosis, 2) Treatment, 3) Lifelong follow-up and integrated care, 4) Organisation of care, 5) Collaboration, referral and communication between care providers, and 6) Transition of care, according to the European context and based on current evidence.

Close attention should be paid to the dissemination and implementation of this adapted Clinical Practice Guideline (CPG) 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 across all components of ARM care.

7. ABOUT THIS DOCUMENT

7.1. Disclosures

We would like to disclose that two of the authors of this Clinical Practice Guideline (Ivo de Blaauw and Cornelius E.J. Sloots) 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.

7.2. Acknowledgements

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7.3. Collaboration

This work was performed in collaboration with experts from ERN ERNICA (the European Reference Network for Inherited and Congenital Anomalies), the European Paediatric Surgeons' Association (EUPSA), and Qualicura.

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51. Aubert O, Irvine W, Aminoff D, de Blaauw I, Cascio S, Cretolle C, Iacobelli BD, Lacher M, Mantzios K, Midrio P, Miserez M, Sarnacki S, Schmiedeke E, Sloots C, Stenström P, Schwarzer N, Gosemann JH. ERN eUROGEN Guidelines on the Management of Anorectal Malformations, Part IV: Organization of Care and Communication between Providers. Eur J Pediatr Surg. 2024 Oct 11.
52. ERN eUROGEN & ERN ERNICA Colorectal Webinar Series – YouTube Playlist: https://youtube.com/playlist?list=PL8VXOqklv7jr1o7PRMkHE_cLyIAjSw3mk&si=R6ft6jA6Z32xfIGh
53. European Reference Network: Clinical Practice Guidelines and Clinical Decision Support Tools Programme. 7 March 2023. https://health.ec.europa.eu/publications/european-reference-network-clinical-practice-guidelines-and-clinical-decision-support-tools_en

9. APPENDICES

9.1. Appendix 1. Dutch Quality Standard appraisal

9.1.1 AGREE II domains and items

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.

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 each reviewer awarding full marks for each signalling question, resulting in a standard domain score 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 stated in the guideline that systematic methods were used and that a procedure was 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) had the lowest standard domain score, at 46%. However, there was a high level of disagreement between reviewers in this domain for all of the signalling questions.
- Domain 6 (editorial independence) had a standard domain score of 75%. The competing interests weren´t clear to some of the reviewers.

The overall standard domain score, as 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.

9.1.2 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**			
Domain 3. Rigour of Development	Item 7. Systematic methods were used to search for evidence.	7	7	7	7	28	**32**	**224**	**78%**
	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%**
	Total	5	4	7	7	**23**			
Would you recommend this guideline for use?		**Yes, with modifications**	**Yes, with modifications**	**Yes**	**Yes**				

9.1.3 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%

9.2. Appendix 2. Systematic literature search

9.2.1 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 OR prisma:ti,ab OR prospero:ti,ab OR (((systemati* OR scoping OR umbrella OR 'structured	733409

	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*' 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.))	5381225

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)

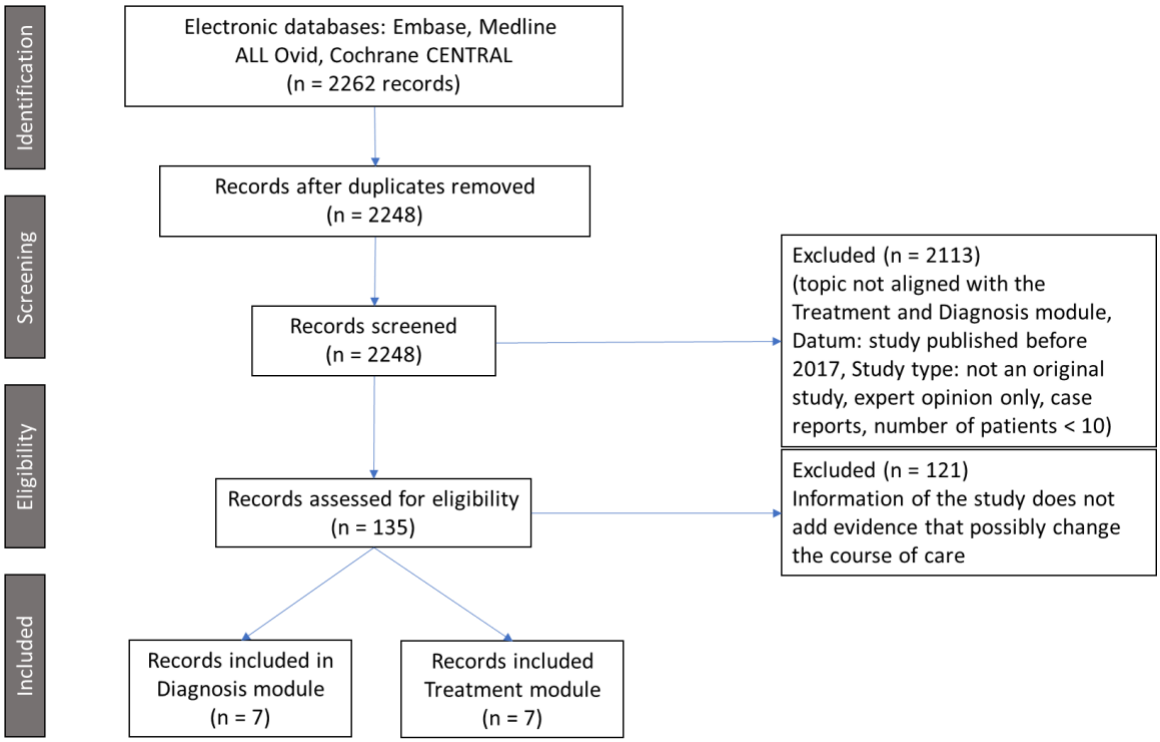
#2 MeSH descriptor: [Anorectal Malformations] explode all trees 113

#3 2017 -

9.2.2 Results

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

9.2.3 Prisma flow diagram of literature selection



9.3. Appendix 3. Summary of evidence for prenatal and neonatal diagnostics

Five (5) new studies, published between 2018 and 2022, were found for prenatal diagnosis, and one (1) new study, published in 2020, was found for neonatal diagnosis.

PRENATAL	Author
Study type: Retrospective chart review (case series) Aim: review of diagnostic approaches for diagnosing foetal imperforate anus. Population: neonates with ARM Results: Ultrasonography was performed in 19 cases of foetal imperforate anus from 2016 to 2019 at the prenatal diagnostic centre. Foetuses at 21–24 weeks and 30–34 weeks were examined for anal deformity. The ability to recognize the most typical ultrasound findings in imperforate anus can improve the detection rate. Conclusion: The absence of the 'target sign' and then the presence of the 'line sign' can assist in the diagnosis of imperforate anus. The 'line sign' can be used as a secondary assessment to determine the type of the malformation following non-visualization of the 'target sign'	Yin et al., 2022[9]
Type of study: Prospective Case series Population: 900 ultrasounds in 372 different pregnant women. Interventions: Prenatal ultrasound to visualize anal dimple Results: In the fetuses with more than one ultrasound, the anal dimple (AD) was visualized at least once in 164 (81%) cases, and not visualized in any ultrasound in 39 (19%). The optimal gestational age range for AD visualization was 28–33 weeks + 6 days, with 78.1% visualization rate ($p < 0.001$). Only one baby was found to have ARM. Conclusion: The study concludes that visualization of the anal dimple by prenatal ultrasound is feasible and may aid in the detection of less severe ARM Level of evidence: Level III study (OCEBM, 2011)	Bischoff et al. 2021[10]
Type of study: Prospective cohort Population: Between January 2008 and January 2016, 63,101 foetuses (gestational age, 20–38 weeks) were prospectively evaluated using 2-dimensional US scans. Results: Among the investigated foetuses, 28 showed evidence of anorectal atresia on US scans, and 22 of those with anorectal atresia had additional anomalies. Six cases of isolated anorectal atresia were successfully detected during the preclusive prenatal US scans. Conclusion: Identifying the abnormal appearance or absence of the foetal anal canal and rectum on preclusive US anomaly scans is useful for prenatal diagnosis or exclusion of anorectal atresia, which may help improve the detection of isolated anorectal atresia. However the diagnosic accuracy is low and detection of a muscular complex does not exclued low ARM types. Level of evidence: Level III study	Su et al., 2019[11]
Type of study: Systematic review of case reports Population: Patients where ARM was diagnosed prenatally Results: In 12 of the 14 cases reported in the literature of ARM diagnosed in the first trimester, the first sonographic sign was a hypoechoic cystic mass in the abdomen. In four cases, the hypoechoic content converted into a hyperechoic mass during the second trimester. In four other cases, the cystic mass disappeared and the bowel had a normal appearance at mid gestation. In five cases, the cystic mass remained unchanged until mid-gestation.	Liberty et al., 2018[12]

Conclusion: The study emphasizes the importance of maintaining a high index of suspicion for ARM when a cystic mass appears in the lower abdomen in the first trimester Level of evidence: Level IV (OCEBM, 2011)	
Type of study: Retrospective comparison Population: A retrospective study included 13 US and 8 MRI exams, with 7 children having both exams. Results: All cases (n=14) had associated congenital anomalies. The detection rate of US was 31% [95% CI 0.25–0.37] (4/13; 3 anorectal malformations and 1 cloaca), and for MRI was 50% [95% CI 0.41–0.59] (4/8; 1 anorectal malformation and 3 cloacae). The study conclude that the foetal perineum should be carefully examined in prenatal US screening exams, especially in foetuses with urinary and spinal anomalies. Conclusion: Complementary MRI should be performed in foetuses with cloaca or multiple congenital anomalies at US screening Level of evidence: Level III	Rohrer et al., 2020[13]
NEONATAL	**Author**
Type of study: Diagnostic test accuracy Aim: The study aimed to correlate spinal ultrasound and magnetic resonance imaging findings in patients with ARM. Population: 193 patients with ARM. Results: Spinal ultrasound had an overall sensitivity of 91% and specificity of 75% compared to MRI for detecting spinal cord anomalies in children with ARM. Conclusion: spinal ultrasound performed in a tertiary paediatric imaging department was a good screening test for spinal cord anomalies in children with ARM. The finding of a low, borderline low or tethered cord on US mandates an MRI to confirm the findings and correlates with the need for operative correction of spinal cord tethering	Jehangir et al., 2020[14]

9.4. Appendix 4. Summary of evidence for diagnostic examinations performed during and after surgeries and during follow-up

One (1) new study published in 2018 was found.

BEFORE SURGERY	Author
Study type: Case series on diagnostic features Aim: To evaluate the difference in pouch-perineum distance (PPD) between neonates with a low-type ARM with and without an opened fistula. Populations: Retrospective analysis of 24 neonates with low-type ARM who underwent sonography before surgery. Results: The mean PPD in neonates with an opened fistula was 10.6 mm ± 3.4 mm (range, 5.6–14.0 mm), compared to 7.1 ± 1.7 mm (range 3.3–10.0 mm) without an opened fistula (p<.02). Conclusion: The study found that the pouch-perineum distance is substantially longer in neonates with an ARM with an opened fistula than in those without an opened fistula. Therefore, caution should be exercised by the sonographic examiner in evaluating pouch-perineum distance in neonates with an opened fistula to prevent an incorrect surgical procedure based on misdiagnosis of the type of ARM Level of evidence: Level III study (OCEBM, 2011)	Hosokawa et al., 2018[16]

9.5. Appendix 5. Summary of evidence for treatment

Seven (7) new studies published between 2017 and 2022 were found.

SINGLE-STAGE AND MULTI-STAGE ANORECTOPLASTY	Author
Type of study: systematic review with meta-analysis (5 retrospective studies and 1 randomized controlled trial (RCT); n = 199 female patients) Interventions: one-stage sagittal anorectoplasty (SARP) without prior colostomy vs conventional two or three-step sagittal anorectoplasty (SARP). Population: females with recto-vestibular fistula (RVF). Results: one-stage approach increases the risk of postoperative complications, such as wound infection, dehiscence and anorectal stenosis. Nevertheless, the one-stage approach was not associated with reduced faecal continence and constipation compared to the multi-stage repair. Level of evidence: Level III study (OCEBM) Authors' conclusions: choose the surgical approach based on the surgeon's experience and the patient's general conditions.	Lauriti et al, 2019[21]
Type of study: RCT Interventions: single-stage procedures vs two or three-step procedures. Population: Females with low-type ARM (85% VF), mean age Results: Wound dehiscence (superficial and deep) occurred in 13 of 33 (39.4%) children in Group I and in 6 of 31 (18.2%) children in Group II. This difference was significant (P = 0.038). On logistic regression analysis, the odds ratio of wound dehiscence associated with a staged procedure was 0.55 (95% confidence interval = 0.31-0.98; P = 0.042). Level of evidence: Level III study (OCEBM) (rated down for high risk of bias) Authors' conclusions: Females with low-type ARM treated with primary definitive procedure have a significantly higher incidence of wound dehiscence, immediate and early postoperative complications. Intraoperative faecal contamination of the surgical site significantly increased the incidence of wound dehiscence.	Gupta et al., 2017[23]
ANAL DILATIONS /CALIBRATIONS	
Type of study: randomised controlled trial. Population: 50 patients (40% females) with ARM who underwent a posterior sagittal anorectoplasty (PSARP). Interventions: postoperative dilation with Hegar protocol vs no postoperative dilations (except if a stricture was detected during follow-up). Results: no statistically significant differences in stricture formation between both groups. 13% of patients in the dilation group vs 16% of patients in the control group needed surgery to manage skin-level strictures; A redo PSARP was needed in 8% of patients of both groups to manage deep strictures. Level of evidence: Level II study (OCEBM, 2011) Authors' conclusions: parents should be given a choice whether or not to perform routine dilations at home. Close follow-up is recommended if they choose not to perform. Contemplate non-dilation as a viable alternative, with Heineke-Mikulicz anoplasty as a backup plan when a stricture develops.	Ahmad et al., 2021[22]

OTHER SURGICAL APPROACHES	
Type of study: treatment guidelines (TGs) Population: patients with persistent cloaca (PC), cloacal exstrophy (CE), or Mayer–Rokitansky–Küster–Häuser syndrome (MRKH). Results: Clinical questions (CQs) concerning treatment outcomes of the genitourinary system, pregnancy and delivery, and quality of life in adulthood were prepared as six themes for PC and CE and five themes for MRKH. We were able to publish statements on chronic renal dysfunction, hydrometrocolpos, and pregnancy, based on four CQs about PC, four about CE, and two about MRKH, respectively Level of evidence: Level IV study (OCEBM, 2011) Authors' conclusions: due to the paucity of proper manuscripts, we were unable to make conclusions about the correct timing and method of vaginoplasty for patients with PC, CE, and MRKH or the usefulness of early bladder closure for patients with CE. These TGs may help clarify the current treatments for PC, CE, and MRKH in childhood, which have been carried out on an institutional basis. To improve clinical outcomes, systematic clinical trials revealing comprehensive clinical data of the urinary and reproductive systems, especially the length of the common channel in PC, are essential.	Kubota et al., 2019[17]
Type of study: retrospective cohort study. Interventions: surgical complications and postoperative defecation function between neonates and infants undergoing anterior sagittal anorectoplasty (ASARP). Population: 35 females with AVF (17 undergoing surgery as neonates and 18 as infants). Results: Surgery duration was shorter for neonates, as well as time to restart oral intake. Postoperative complications more frequent in infants (mostly wound dehiscence). No significant differences in long-term defecation function scores, though less patients in neonatal group required every day enemas or defecation assistance during a follow-up of 4 years. Level of evidence: Level IV study (OCEBM, 2011) Authors' conclusions: single-stage ASARP for anovestibular fistula can be performed even in the neonatal period.	Shirota et al., 2018[19]
Type of study: systematic review and consensus statements Population: patients with normal Anus, Anterior anus (AA) and milder types of ARM such as congenital anal stenosis (CAS) and perineal fistula (PF). Results: a consensus on definitions, clinical characteristics, diagnostic management, and treatment modalities was established, and a diagnostic algorithm was proposed. The algorithm enables pediatricians, midwives, gynecologists, and surgeons to make a timely correct diagnosis of any abnormally looking anus and initiate further management if needed. Level of evidence: Level II study (OCEBM, 2011) Authors' conclusions: the routine physical inspection of a newborn should include the inspection of the anus and define its position, relation to the external sphincter, and caliber. A correct diagnosis and use of the presented terminology will avoid misclassifications and allow the initiation of correct management. This will provide a reliable comparison of different therapeutic management and outcomes of these patient cohorts in the future.	Amerstorfer et al, 2020[20]
Type of study: case series. Population: 153 patients (29% females) with previous ARM repair. Aims: assess the benefit of a redo posterior sagittal anorectoplasty (PSARP) in patients still suffering from faecal incontinence.	Wood et al., 2020[24]

Results: significant improvement in validated faecal incontinence and quality of life scores (Baylor and PedsQL scores) after a redo PSARP. All patients included for a redo procedure were also placed on an aggressive bowel management program with either a laxative or an enema-based regimen. Level of evidence: Level IV study (OCEBM, 2011) Authors' conclusions: a redo procedure in patients with a previously repaired ARM along with intensive bowel management can significantly improve both faecal continence, cleanliness for stool and quality of life in a majority of patients.	

9.6. Appendix 6. Summary of evidence for follow-up and integrated care

Seven (7) new studies, published between 2018 and 2023, were found.

FOLLOW-UP AND INTEGRATED CARE	AUTHOR
Type of study: A (prospective) multi-center cohort study 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.	van der Steeg et al.,2022
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. 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).	Svetanoff et al., 2022
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 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.	Miyake et al., 2023

Type of study: Guidelines Aim: To increase the quality of care for children with urological conditions	Radmayr et al., 2023

9.7. Appendix 7. Summary of evidence for organisation of care

Two (2) new studies, published between 2020 and 2022, were found.

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. 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.	Lacher et al., 2022

9.8. Appendix 8. Summary of evidence for transition care

One (1) new study, published in 2019, was found.

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

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