

Patient Journey for Anorectal Malformation

A Lifelong Journey Taken One Step at a Time

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1. INTRODUCTION

1.1. Prevalence-Incidence

Anorectal malformations (ARM) are rare and complex birth defects. The incidence of ARM varies between 1:3500 and 1:5000 live births (*Reference 1, page 44*).

1.2. What It Means To Be Born With An Anorectal Malformation: The Clinical Perspective

ARM are birth defects occurring in the very early stages of pregnancy. Babies born with this rare condition need urgent medical attention.

Children with ARM are born without an anal opening, or the opening is in the wrong position. It is important to know that ARM represents a spectrum of abnormalities which affect the lower end of the digestive tract and interfere with the normal passage of stool. It ranges from minor (imperforate anus) to more complex malformations. In some babies, other body parts may be affected, including the bladder, heart, limbs and/or the sacrum.

Assessment and investigations are essential to ensure a correct diagnosis is made prior to surgery. Parents should feel able to discuss their child's diagnosis with the surgical team and ask any questions they need to reassure themselves.

Babies with a less complex ARM type can sometimes be treated with a single operation without forming a stoma. Most of these babies/groups of patients have a better prognosis regarding stool and urine continence.

The decision to repair the ARM in a single operation should be taken by an experienced surgeon.

Infants with an ARM where the connection is higher are classified as complex and require more than one operation. Repair/correction for these babies often means three operations: the creation of a stoma, repair surgery (pull-through), and finally, closure of the stoma. An operation to create a stoma is needed in the first days of life to remove waste from the bowel.

Infants and children must always receive care from a skilled surgeon working with an experienced multi-disciplinary team in a specialised centre where examinations, assessments, and investigations are possible to ensure an accurate diagnosis and provide the best long-term outcomes. An incorrect diagnosis and incorrect operation can have a detrimental impact, affecting the child throughout their lives.

1.3. What It Means To Be Born With An Anorectal Malformation: The Emotional Perspective

Becoming a parent is a life-changing experience, with new parents often experiencing a rollercoaster of emotions, from relief and joy to concern. When a baby is born with problems, the parents' feelings may be mixed; happiness and fear exist alongside each other. **There will be many questions, sometimes with no clear answers.**

Hearing that your baby has an ARM can be frightening; for most parents, the diagnosis comes without warning, a profound shock which may, understandably, heighten anxiety. Parents worry they have caused the condition and are sad their baby needs surgery.

The bond between a baby and its parents is a strong emotional and physical connection. It shapes the baby's development and helps them grow, learn, manage emotions, and develop a sense of identity. However, if an infant needs immediate medical treatment, the parent-baby bonding can be significantly affected.

Healthcare professionals understand these difficulties; they know it is especially important to support parents, explain what is happening, and keep the family at the centre of decision-making.

Family-integrated care helps parents remain connected to their baby, supports attachment, and improves parental confidence. This family-centred approach, involving parents at each stage, prepares the parents to meet the baby's care needs when the family eventually goes home.

Being born with an ARM means more than just needing a surgical procedure to repair the anomalies. This rare and complex diagnosis involves a lifelong journey for the patient, their parents and other family members.

This document provides an overview of important stages throughout the patient's life.

From the first signs and symptoms to diagnosis, surgery, and aftercare in the early years, from school age to adolescence, and to issues sometimes faced in adulthood.

2. METHODOLOGY

This ARM Patient Journey has been developed by the representatives of three patients and parents organisations:

- **SoMA e.V.:** German patients and parents organisation for those born with ARM, Hirschsprung's Disease or Cloacal Exstrophy
- **AIMAR:** Associazione Italiana Malformazioni Anorettali – Italian patients and parents association for ARM
- **EAT:** Esophageal ATresia Global Support Groups

The authors have also received guidance and support from:

- **Johanna B.,** Patient born with ARM, social worker, certified hand in hand instructor, member of SoMA e.V.
- **Michel Haanen,** Patient Representative in the Netherlands, and board member of SoMA e.V.
- **Annette Lemli,** Chairwomen SoMA e.V.
- **Mazeena Mohideen,** Chairwoman SoMA Austria

Our community includes over 2,500 members from more than five countries.

Our contributors include board members from national organisations. Many have worked in this specialist field for over 20 years, gathering patient experiences through interviews, questionnaires, workshops, and seminars. The authors have collaborated with individual patients and parents, listening to and valuing their lived experiences, to provide an informative and user-friendly patient journey.

We started this project in the autumn of 2022. During this time, we have met face-to-face only once. The remainder of our contacts have been via emails and many Zoom meetings, where communication can be challenging, as the subtleties of body language are sometimes misinterpreted.

Having lived with and alongside this rare and complex condition, we realise that although there is a great deal written about ARM, very little of the information takes the reader on a lifelong journey where parents, adolescents, adults, and professionals can dip into the text and find valuable information.

Each of us brings a different perspective, as parents and patients, but also as advocates of parents and patients: sharing very personal, sensitive experiences around separation from your newborn infant, putting your trust in strangers, continence, sexual health, fertility, coping, communication and self advocacy.

The data presented here has been obtained from listening to personal stories from parents of infants, adolescents, and adults born with ARM. We have reviewed medical articles and connected with professionals in different European countries who are currently working in child development and paediatric surgery.

In addition, we have worked alongside surgeons who are involved in the current new guidelines for caring for those born with ARM.

We also sought the expertise and advice of patient organisations to help us reflect all the stages of this lifelong journey. In this Patient Journey, we have emphasised these essential points:

- Parents face a huge emotional challenge when handing their precious infant to strangers.
- The anxieties parents have for their child's future; those fears never go away.
- Children face challenges when they feel different from their friends, rejected, bullied, alone and frightened.
- The struggle many adolescents and adults face to become both "socially continent" and socially accepted.

The information presented here has been provided anonymously.

3. PATIENT JOURNEY: TIMELINE

A Lifelong Journey Taken One Step At A Time

Stages of the Journey	Comments		
Prenatal diagnosis	<p>Most types of ARM cannot be diagnosed prenatally, although other anomalies may indicate the presence of an ARM.</p> <p>An ultrasound from the second trimester onwards can detect abnormalities within the urinary tract and lower pelvis, which may lead to a diagnosis of cloacal malformations.</p> <p>Persistent cloaca malformation is the most severe type of ARM with decisions concerning treatments and care being taken in the first hours of life.</p> <p>Currently, in most patients, any genetic predisposition remains unclear.</p>		
In the first days of life	<p>A diagnosis of ARM should ideally be made in the first hours of life. There may be delay in confirming the diagnosis if the baby is passing meconium (first infant stool) via a fistula to the bladder or vagina.</p> <p>Attention to other signs is essential: swollen tummy, vomiting, distress. These must be reported to the healthcare team immediately.</p>		
<p>In the first days of life after diagnosis</p> <p>The experienced surgeon will decide whether the baby needs an operation to create a colostomy (stoma)</p>	<p>Some infants may need one operation to correct the ARM; others, with more complex anomalies, will need three operations. In these more complex anomalies, staged surgery is necessary.</p> <p>Each child requires a degree of intervention which is unique to them.</p> <table border="1"> <tr> <td> <p>Group 1: A single repair procedure.</p> <p>These babies do not need a stoma. Parents can take their baby home with instructions on how to proceed and who to contact if concerns arise.</p> <p>Arrangements to perform this surgery may take a little longer. This is an elective (non-emergency) procedure.</p> </td> <td> <p>Group 2: Formation of a stoma.</p> <p>In these infants, a stoma is created soon after birth. Parents are trained in their baby's care and supported to go home prior to the main procedure.</p> </td> </tr> </table>	<p>Group 1: A single repair procedure.</p> <p>These babies do not need a stoma. Parents can take their baby home with instructions on how to proceed and who to contact if concerns arise.</p> <p>Arrangements to perform this surgery may take a little longer. This is an elective (non-emergency) procedure.</p>	<p>Group 2: Formation of a stoma.</p> <p>In these infants, a stoma is created soon after birth. Parents are trained in their baby's care and supported to go home prior to the main procedure.</p>
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<p>Usually from 2-6 months of age</p> <p>- Corrective surgery</p> <p>- Post-surgical care</p>	<p>The timing of corrective surgery followed by closure of the stoma will vary from surgeon to surgeon, hospital to hospital and country to country.</p> <p>If the baby has additional health concerns these may take priority over the corrective anorectal surgery. Healthcare teams will explain to parents the need to prioritise investigations and other surgeries.</p> <p>During this time parents are instructed in the care of their child in preparation for discharge home.</p> <p>The postoperative recovery time of any infant is individual. Some infants recover quicker than others; the majority should be home within 2 weeks. A further operation to close the stoma necessitates further advice and training for the parents.</p>		
<p>1 - 5 years</p> <p>Infancy and early years including kindergarten</p>	<p>This stage of the ARM journey will vary across different countries.</p> <p>This is a period of further adjusting to the health needs of the baby as they grow and develop, working collaboratively with the healthcare team and starting the process of sharing with other professionals in early years education.</p> <p>This should be supported by community services who can explain the ARM to non-medical staff and educate them to be vigilant in the care of the child in new surroundings.</p>		

Stages of the Journey	Comments
5 - 11 years School aged children	<p>This stage will vary across different countries and different education systems.</p> <p>Care must be coordinated, planned, and incorporate a “whole school” approach to meet the child’s emotional, social, medical, and educational needs.</p>
11 - 18 years Adolescence to adulthood (Transition)	<p>This new and rapidly evolving time can be challenging for any young person, but for those born with an ARM it can be even more daunting as they learn to make decisions and take responsibility for the management of their condition. Learning the skills needed to self-advocate should begin at an early stage in adolescence.</p>
From 18 years to the end of their lives	<p>It is important to recognise that not all adults born with ARM will experience health difficulties. However, some adults may need lifelong care.</p> <p>It is important to know that care does not necessarily mean further surgery but may involve other experts to address urological, gynaecological issues as well as professionals who can support the adult’s emotional wellbeing.</p> <p>There are many factors which contribute to an adult seeking advice and support. These include the type of ARM, the surgical procedures needed in childhood and health problems associated with the ARM that may arise as they age.</p> <p>Many adults struggle to explain themselves, becoming anxious and distressed.</p> <p>Most healthcare providers have not heard of ARM.</p> <p>When adults ask for help and advice, health professionals are sometimes unable to see the connection between the patients’ current symptoms and their birth condition.</p> <p>Healthcare providers should understand the need for lifelong care.</p> <p>A review of bowel management or gynaecological, andrological and/or psychological issues may be needed.</p> <p>Lifelong collaborative care should be available for those adults who request it.</p>

4. STAGE 1: PRENATALLY AND BIRTH

4.1. The Need for Family-Centred Care

Parents plan for their baby's birth. They discuss what those first minutes and hours will be like, how they will react to seeing and holding the baby, and how they will get to know their precious newborn. However, when a baby is born with an ARM, the world they had envisaged is turned upside down, emotions rapidly change from joy to fear, and parents are in crisis.

To ensure optimum care for the baby, parents must hand over a lot of control and put their trust in the medical team; this can lead to feelings of hopelessness and emotional instability. Staff should understand that what is often routine for them can be highly challenging and frightening for parents. These emotionally vulnerable parents need kindness and empathy, alongside the reassurance their baby is in the best hands to achieve optimum outcomes.

They need staff who value and support them to be effective partners in caring for their baby; this helps them cope with the often rapidly changing situation and helps them bond with their baby. Parents need staff who actively listen to their questions and respect their concerns. Staff who understand that parental irritability and agitation are normal responses to this frightening situation and respond sensitively to the emotions parents may present. This gentle, supportive approach reaffirms the parents' importance, that their questions are valid, and helps with the development of parental coping strategies.

At this time, parents need information about ARM, both verbal and written, in a clear format they can understand.

It is essential that they are included as a valued part of the care team discussions and the decision-making process, working together in the best interests of their baby.

Parents may find it challenging to maintain the role of parent in a critical care environment; they can feel disempowered and detached from what is happening around them. To overcome these additional stresses, parents need to understand and participate fully in caring for their baby. In preparation for going home, the parents need training to manage their baby's stoma and to identify symptoms which may indicate the baby needs medical assessment and further care. Additional training may be required if the baby has other anomalies.

At this stage of the patient journey, parents need to meet the staff who will be involved after the operation and the team that provides home care support. They also need to be signposted to the appropriate patient organisations.

This information provides reassurance that they are not alone and will be supported for many years ahead.

Hospitals caring for babies with ARM should have a policy to provide or organise lifelong care and support.

4.2. First Information and Questions

What are the causes? Why didn't we know before?

At present, ARM cannot be accurately diagnosed prenatally. Antenatal scans may identify other anomalies, such as heart, kidney/bladder, or spinal problems associated with this condition, which may lead to a suspicion of the presence of an ARM.

There will be so many questions...

After your baby is born you will spend time in a newborn baby unit where you will be told about the diagnosis. Some people like to have an overview to begin with, then ask the clinical team for more detail over the next few days. Your doctors and nurses will be used to this. Make sure you ask the questions you need to and understand what is happening. You will hear that the abnormality occurs when the anus, rectum, and sometimes other organs, do not develop properly during pregnancy, which can affect the baby's ability to pass stools.

What is meant by "ARM is a spectrum from mild to more complex conditions"?

A proper physical examination of the perineum (the area where the bottom usually develops) will allow vital clues to the type of ARM. This includes the absence or presence of an anal opening, whether it is big enough, the presence of a vaginal and urethral opening in girls, and the presence and location of any fistula. The position of the rectum and anus and the possible involvement of other organs determines the degree of severity of the ARM and can influence the number of operations needed.

Importance of an accurate diagnosis

Examination, assessment, investigations, pre- and post-operative care is important for your baby. At all times, patients must receive care from a skilled surgeon and an experienced multi-disciplinary team with access to appropriate examinations and investigations to ensure an accurate diagnosis is made. (Link of classification- Reference 2)

Surgical procedures – "Is my child in the best hands for managing his/her condition?"

Most infants will need surgery; this is usually arranged in the first year of life. Some infants may need one operation to correct the ARM; others, with more complex anomalies, will require three operations or more. Staged surgery gives the parents time to gather further information about their baby's ARM, about the corrective surgery they need, and if necessary, to obtain a second opinion about corrective surgery.

1. Firstly, a small section of the bowel is opened onto the abdomen (tummy). This artificial opening is called a colostomy or stoma. The stoma allows faeces (stool) to exit the bowel into a disposable bag. This allows the bowel to function normally and keeps the lower bowel free from stool in preparation for the corrective surgery.
2. The corrective surgery (anorectoplasty) is performed around 3-6 months.
3. Closure of the stoma/colostomy is performed later.

Parents are trained in their baby's care and supported to go home prior to the main procedure. An individualised plan of care should be compiled detailing the type of ARM, the presence of associated anomalies, and information about the child's overall health. The family must have assistance from experienced healthcare staff. It is especially important to recognise that not all children will face the same health difficulties.


Each infant is unique, with their own pathway determined by the type of ARM they were born with, and the treatment(s) needed. And it is essential that no one feels alone or unsupported.

Lifelong holistic care for the best possible outcomes

The overall aim is to get the best possible outcome for your baby so that their childhood is healthy and relatively uneventful, and they become independent adults with a good quality of life. To achieve this, families and clinical teams must work well together.

5. STAGE 2: THE FIRST YEAR

5.1. Step 1: Investigations, Diagnosis, and Treatment Plan

In the first days of life	What you can expect to happen	Best practice
<p>The baby may appear well, and the abnormality may be missed if stool is passed via the vagina, via a tiny perineal orifice or, very rarely, via the urethra in males.</p> <p>The baby may be discharged home without a diagnosis.</p>	<p>The newborn assessment should include examination of the perineum and genitals to observe them for abnormalities.</p> 	<p>Excellent communication is essential among doctors, nurses, and parents.</p> <p>Providing an individualised holistic care plan, with a timeline for treatments and, if necessary, operations.</p> <p>Meeting the family support worker and psychologist.</p> <p>Providing support to siblings and immediate family.</p>
<p>Be vigilant:</p> <p>A baby's temperature used to be checked using a rectal thermometer, this enabled the checking of the perineum, including noting the presence or absence of a bottom hole. This practice has now been discontinued.</p> <p>Some infants fail to pass meconium (first stool) or pass ribbon stools. They may have constipation, a swollen tummy, and/or colic, be unsettled, reluctant, and/or refuse to feed.</p> <p>An antenatal ultrasound scan may diagnose a collection of fluid in the vagina of female infants (so-called hydrocolpos) and can indicate a single opening involving the urethra, vagina, and rectum. This channel is called a cloacal malformation.</p>	<p>Absence of a bottom hole may be missed.</p> <p>Babies first stool:</p> <p>Check that meconium (first stool) is passed from the correct orifice (bottom hole).</p> <p>This is particularly important as the baby's temperature is no longer taken rectally.</p> <p>The absence of a bottom hole should be investigated further.</p> <p>Early detection and management of a hydrocolpos is essential for preserving renal function.</p> <p>A drained hydrocolpos must always be followed-up by an ultrasound.</p>	<p>Information and advice:</p> <p>Referral to the appropriate patient organisation(s).</p> <p>If this is not available, then provide contact with parents who have a child with a similar condition.</p> <p>Ensuring the parents remain central to decision-making, that there are members of the team around the child.</p> <p>Providing information about the reasons for further investigations, treatments, and where necessary operations.</p> <p>Collaborative working between the hospital and home care teams.</p> <p>Parents are notified of local support systems and know who to contact if problems arise.</p>
<p>Important to note:</p> <p>Associated anomalies may be present.</p> <p>These can be remembered using the VACTERL acronym (see Section 9).</p> <p>At this stage, finding out if your baby has other anomalies, helps with planning and treatment</p>	<p>Further tests may be needed:</p> <p>Abdominal X-ray to look for other bowel anomalies (e.g., duodenal atresia).</p> <p>Ultrasound scan of the kidneys.</p> <p>Ultrasound scan, or sometimes an MRI of the lower pelvis to check the spinal channel (e.g., for spinal dysraphism).</p>	<p>Parents should always know who, how, and when to contact the healthcare team.</p>

	<p>Echocardiogram to look for heart anomalies.</p> <p>If the baby has other abnormalities affecting breathing and or heart function, emergency interventions, including surgery, may be necessary.</p>	
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5.2. Step 2: Formation Of A Stoma, Also Known As A Colostomy

In the first days of life/after colostomy	What you can expect to happen	Best practice
<p>Formation of a stoma.</p> <p>The bowel is brought onto the abdominal (tummy) wall.</p> <p>This artificial opening is called a stoma.</p> <p>The stoma allows stool to leave the bowel and be collected into a small disposable bag.</p>	<p>Explanation and written information (booklet for parents) regarding the type of ARM and the surgery required.</p> <p>Identifying any difficulties or risks which may arise immediately after the operation.</p> <p>A named healthcare professional or a specifically qualified healthcare practitioner should be the point of contact for parents.</p>	<p>Reassurance that the healthcare team will strive to achieve the optimum outcomes for the baby.</p> <p>Parent/family accommodation should be available.</p> <p>Assistance with travel costs may be available, particularly if the infant is transferred to a specialised centre away from home. If this helps you care for your baby, please ask.</p> <p>Maintain appropriate midwifery / obstetric care for the mother whilst staying within the acute hospital.</p>
<p>The stoma allows the bowel to function correctly.</p> <p>The stoma keeps the lower bowel free from stool in preparation for anorectoplasty.</p>	<p>The infant should now be able to feed and thrive before further operations are performed.</p> <p>Going home:</p> <p>Most of the time parents can take their baby home once the infant has recovered from the anaesthetic and from the operation.</p> <p>Delayed discharge sometimes occurs if complications arise, or additional health problems or malformations are identified.</p>	<p>Introduction of a specialist nurse:</p> <p>Learning to care for a baby with a stoma:</p> <p>Training parents to identify and manage possible stoma related problems.</p> <p>Maintain skin integrity, changes in size and colour of the stoma.</p> <p>It is essential to ensure the stoma is functioning correctly.</p> <p>Aim for a smooth transition to home: Parents must be fully trained and competent, and the baby must be thriving before discharge home.</p> <p>Support at home is arranged prior to discharge.</p> <p>Parents are notified of local support systems and know who to contact if problems arise.</p>

5.2.1 Care Of Your Baby's Stoma

In the first days of life/following the formation of a stoma	What you can expect to happen	Best practice
Stoma care	<p>It is not unusual to have problems with your child's stoma.</p> <p>These issues are frequently seen and are often easily resolved with help from your homecare team.</p>	<p>Parents/caregivers should always know who, how, and when to contact their child's healthcare team.</p>
	<p>Stoma bleeding. Leakage of faeces.</p> <p>Stool on the skin.</p> <p>The stoma bag may not fit correctly and may need resizing.</p>	<p>Click here for: CPOC Paediatric Stoma Care Guidelines</p>
	<p>Sore skin:</p> <p>Seek advice and support from a specialist home care stoma nurse.</p>	<p>Taking a photo of the stoma helps as you can send it to the healthcare team remotely, and seek advice</p>
Maintain optimum health.	<p>Stoma is discoloured:</p> <p>Referral to stoma nurse, or the hospital as appropriate</p> <p>Explanation and emphasis on the importance of regular reviews with the specialist team.</p>	<p>Parents/caregivers should always know who, how, and when to contact their child's healthcare team.</p> <p>Specialist centre follow-up with the surgeon and other multi-disciplinary team members is arranged – frequently in the first weeks, with eventual annual review as the child grows.</p>

5.3. Step 3: Before The Main Procedure/Repair

Assessments and Investigations:

Further Assessment and investigations may be needed before the surgical repair construction is performed:

- Distal Colostogram:
 - It is usually performed a few weeks after the stoma operation.
 - This special type of X-ray, with some contrast material, shows a detailed image of your child's ARM. From this image, the surgical team can more precisely diagnose your child's specific type of ARM.
 - It will show exactly where the end of the rectum is situated and its relationship with surrounding structures.
 - The diagnosis will help the team plan the surgery and determine the prognosis for your child's future bowel control and other treatments.

Another investigation sometimes accompanies this contrast study:

- Voiding Cystourethrogram, also known as a (VCUG).
 - Urodynamics and cystoscopy are used to detect underlying anatomical or functional causes.
 - This test enables the bladder and urethra to be seen clearly. It can detect vesicoureteral reflux (VUR), where urine flows backwards from the bladder to one or both ureters and sometimes to the kidneys.
 - In some types of ARM, bladder catheterisation may be needed to preserve the function of the kidneys.

In All Male Infants:

Prior to pull-through surgery, the parents, caregivers, and nurses should look and record whether a male with ARM has spontaneous erections when changing diapers. This information is important when the boy reaches adolescence.

In males born with a rectourethral fistula, a condition known as epididymo-orchitis can present at any age. It is an inflammation of the testis and the epididymis (spermatic cord). It occurs when part of the testis, the epididymis, becomes swollen and painful. It can be caused by ascending urinary tract infections or reflux of sterile urine into the seminal ducts. If epididymo-orchitis recurs despite the correct treatment, clipping of the vas to protect the (future) production of sperm cells may be necessary. Fortunately, this only happens sometimes.

Meeting Patient Needs:

The overall aim is to reach the best possible outcome for your baby so that their childhood is healthy and relatively "uneventful" and they become independent adults with a good quality of life. This means assessing the infant's immediate and long-term needs and planning appropriate evidence-based care.

To achieve this, families and clinical teams must work well together. The parents must be included in these discussions and share in decision-making.

Necessary Actions:

- Determine the type of ARM.
- Decide the procedure needed for the repair.
- Referral to other specialist teams if additional anomalies are diagnosed.
- Carry out investigations as necessary.

Best Practice:

The main repair procedure is an elective operation, and there is time to collect all the necessary information prior to the surgery.

Careful planning is essential to achieve the best possible outcomes.

Gathering evidence from tests, such as a colostogram, voiding cystourethrogram, and scans, will assist the surgeon in deciding on the most appropriate procedure.

Collaboration across many disciplines improves care and long-term outcomes.

The primary aim is to achieve, wherever possible, socially acceptable faecal continence.

5.4. Step 4: Corrective Surgery

This kind of surgery is called posterior sagittal anorectoplasty (PSARP) or laparoscopically assisted anorectoplasty (LAARP) and usually takes place in the first year of life, around 3 to 12 months of age.

Patients must always receive care from a skilled surgeon working with an experienced multi-disciplinary team in a specialist centre.

Parents can ask: “Is my child in the best hands for managing his/her condition?” and can seek a second opinion before proceeding to surgery (see Support Tool in Section 12).

Corrective Surgery	What you can expect to happen	Best practice
<p>This procedure is performed when the child is a few months old, has gained weight, and is thriving.</p>	<p>With good pain management, the child should be comfortable and pain free.</p> <p>An inpatient stay of about one week is necessary to check that the child has recovered from the operation and to ensure the repair site is healing satisfactorily.</p>	<p>The goal is to make the parents feel comfortable and confident caring for their child after these procedures.</p> <p>The child’s pain is well-managed.</p> <p>Ideally, the child should be comfortable and happy to play and interact with the family.</p> <p>The child can start eating and drinking when the surgeon decides this is possible.</p>
<p>Surgery is performed under a general anaesthetic:</p> <p>The child’s rectum is moved away from the urinary tract or vagina and positioned within the anal sphincter muscles.</p> <p>The child’s repair site should be carefully managed.</p>	<p>Caring for your child:</p> <p>Keeping the child from sitting for too long.</p> <p>Ideally not putting pressure on the healing area.</p> <p>Be careful and gentle when cleaning their bottom and changing diapers, particularly when opening their legs; do not over-stretch this area.</p> <p>Do not place your child in any position which keeps their legs widely separated (e.g., rocking horses, infant bouncing toys, infant bikes, car seats, front and back infant carriers).</p> <p>Dress your child in loose-fitting clothing particularly around their bottom. If possible, expose the child’s bottom to the air.</p> <p>Keep the repair site clean and dry; the stitches are usually dissolvable.</p> <p>Do not rub the wound side-to-side as that might break the stitches. It is better to dab gently when the area needs cleaning.</p>	<p>Specialist nurse in hospital and at home:</p> <p>Parents should feel supported in all aspects of their child’s care and feel confident to return home.</p> <p>The nurses will help you with this, please make sure you ask.</p>
<p>Catheter care:</p> <p>A catheter may be in place to drain urine from the bladder.</p>	<p>This can usually be removed within one to two weeks of the surgery.</p>	<p>A specialist nurse provides care and support at home.</p> <p>After discharge home, a post-operative review with the surgical team should occur within two weeks after the surgery, followed by regular appointments to review progress.</p>

5.5. Step 5: Anal Dilations

These are not always necessary. The benefit of this procedure is currently being researched.

Anal dilatations	What you can expect to happen	Best practice
Postoperative anal dilatation.	The appropriateness of this practice is currently being researched and discussed. Some surgeons do not recommend this procedure.	If anal dilatation is necessary, the standards below should be in place: Parents have been given training from the child's surgeon or the specialist nurses.
	Calibration and dilatation are different procedures. It is important parents understand this.	Parents are competent and confident to perform the dilatations.
	Decisions relating to calibration and dilation of the operated anus can be assessed at the outpatient clinic at follow up.	Anal dilatations should ideally not continue for longer than six months.
Difficulty performing the dilatation.	The procedure should not be painful or traumatic for the child.	Dilations are performed with minimal holding of the child.
Bleeding associated with dilatations.		Seek advice from your child's nurse specialist or medical team if these occur.

5.6. Step 6: Closure Of The Stoma

Usually carried out within a few months following the anorectal malformation repair.

Closure of the Stoma	What you can expect to happen	Best Practice
<p>The surgeon reconnects the intestine, providing bowel continuity.</p> <p>This allows the normal passing of stool via the newly formed anus.</p> <p>Be vigilant of the following and let your clinical team know if any of these happen:</p> <p>Severe colic or upset tummy.</p> <p>Green vomiting.</p> <p>Refusing to take feeds.</p>	<p>In the initial post-operative period:</p> <p>With good pain management, the child should be comfortable and not in pain.</p> <p>After the stoma is closed, your child will not be able to eat or drink for a few hours or days. This allows the connection between the two parts of the bowel to heal.</p> <p>They will be given intravenous fluids during this time to maintain hydration.</p> <p>Children usually start passing stool within two to three days after surgery to close the stoma.</p>	<p>In situations where the child is irritable the source of the pain must be identified, and appropriate pain management must be provided.</p> <p>Keep the child comfortable and happy to play with toys and interact with family.</p> <p>Advice and support to parents / caregivers is essential.</p>
	<p>Bowel function following stoma closure can be unpredictable and challenging to manage.</p> <p>It can be difficult to separate a regular childhood illness from complications related to the child's previous surgery. These signs and symptoms may be an indication of bowel obstruction.</p> <p>This serious condition is also known as an ileus and requires urgent medical attention.</p> <p>Most children go home shortly after they begin passing stool.</p> <p>Stool may be frequent, watery, and loose. The stool may cause skin irritation. "Nappy rash" (contact dermatitis) may result in very sore skin which can be painful and distressing to the child.</p> <p>Medications: If needed, these will be provided by the hospital.</p>	<p>Monitoring in hospital:</p> <p>Your child's health care team will be monitoring: wound healing, bowel sounds (heard with a stethoscope on the tummy), passing gas, and the type of and frequency of stool.</p> <p>Hospital and Home Care:</p> <p>Parents and caregivers should be trained to recognise stool consistency and be confident the bowel has emptied sufficiently.</p> <p>The specialist nurse or doctor should review the child to rule out anything related to the surgery. In some instances, the child will need to be admitted to hospital for further investigations.</p> <p>Caution must always be exercised for infants who have undergone this surgery. Constipation and overflow of faeces are often seen in these children and mistaken for normal bowel action.</p> <p>Parents are trained to monitor bowel function and to work towards optimum skin health prior to discharge home.</p> <p>Advice about skin cleansing, wipes, creams, and lotions are individual to each child.</p> <p>The specialist nurse can assess and recommend appropriate products to ease any discomfort.</p> <p>Care at home by specialist nurse and postoperative review with surgical team within two weeks of surgery followed by regular appointments.</p>

	<p>Dietary advice can be useful.</p> <p>Laxative foods: When the stool is hard and difficult to pass.</p> <p>Constipating foods: When the stool is very loose and may be needed in the early days after the stoma is closed.</p>	<p>Referral to a Dietician may be helpful.</p>
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5.7. Associated Malformations: Be Mindful

These occur in almost 50% of infants with an ARM. To exclude these defects, all infants must be assessed at birth.

Urological problems	What you can expect to happen	Best practice
Approximately 50 percent of infants born with an ARM will also have anomalies in the urinary tract and/or the genitals.	At the time of an ARM diagnosis additional investigations may be necessary.	In these infants, urinary tract infections are more prevalent.
These anomalies may occur at any point along the urinary system. In the upper urinary tract, the problems are renal dysplasia, vesicoureteral reflux, hydronephrosis, renal duplications, and renal malposition or single kidney.	The surgical team will explain this in more detail and arrange the appropriate tests where necessary.	Parents and caregivers are trained to identify early signs of infection. If an infection is suspected, it is important to inform the child's medical team as soon as possible.
In the genital tract in girls the most common anomalies are double vagina and/or uterus and missing vagina and/or uterus. However, both are uncommon. In boys the most frequently occurring anomalies are undescended testis and hypospadias.		The homecare team and hospital specialist provide training and ongoing monitoring.
For further information see the glossary at the end of the document.		

Spinal Anomalies

Other malformations of the bones of the spine, especially those leading to scoliosis, should be diagnosed early enough so that the optimal timing for orthopaedic treatment will not be missed.

Tethered Spinal Cord

Children with ARM may have problems with the spinal cord and the nerves within it, including a tethered cord (TC). This happens when the spinal cord remains fixed at the base of the spine and becomes abnormally stretched as the child grows.

The stretching on the TC can limit the movement of the spinal cord within the spinal column and may result in progressive signs and symptoms involving motor-sensory, orthopaedic, urinary, and bowel functions, leading to TC syndrome.

All infants should be assessed and subsequently monitored for early indication of TC.

An ultrasound soon after birth, usually in the first weeks, can provide essential information.

Any decisions about treatment should be taken by experienced neurosurgeons (see webinar link below):

[ERN eUROGEN/ERN ERNICA Webinar: Tethered Cord in the Context of Urogenital Malformations](#)

6. STAGE 3: INFANCY - EARLY YEARS AND SCHOOL AGE

6.1. Continence: A Complex Process

Bowel continence is a complex process that requires three factors to succeed: motility, sensation, and a functioning sphincter. Some children born with anorectal malformation miss at least two of these elements. The type of anorectal malformation will impact the child's ability to achieve continence. Despite a successful operation, continence will remain a challenge for some children, their families, and other carers.

Children born with anorectal malformation should have regular follow-up appointments with their paediatric team, including the surgeon(s) who carried out the operation. The employment of a specialist practitioner is essential throughout childhood to coordinate the child's care and monitor growth and development. Identifying problems as they arise and providing a multi-disciplinary approach to address these issues is vital.

Possible Problems	What you can expect to happen	Best practice
<p>Delayed toilet training.</p> <p>Soiling, hard stool, constipation</p> <p>Urinary tract infections.</p> <p>Urinary incontinence.</p>	<p>A child born with an ARM may be slower to gain bowel control.</p> <p>This is due to the initial altered anatomy and subsequent operations.</p> <p>Stool may be frequent and loose and can sometimes be mistaken for diarrhoea when, in fact, the child is constipated with a hard stool present and liquid stool leaking around it. This type of liquid stool, due to constipation, is called overflow.</p> <p>Urinary tract infections are common, the child may need urotherapy, including clean intermittent catheterisation (CIC).</p>	<p>It is important that kindergarten and pre-school staff understand the reasons behind the delayed continence and do not push the child into toileting.</p> <p>Care plans should be in place to manage soiling or accidents with emphasis on maintaining the child's privacy and dignity.</p> <p>The child is likely to wear diapers or protective pads later than their friends. Staff must handle this with sensitivity and discuss any concerns with the child's parents.</p> <p>Sharing sensitive information with other parents and children must be led by the child and their parents</p>
<p>Food intolerances.</p> <p>Food sensitivities.</p>	<p>Some foods may increase loose stools, others cause constipation; every child is different in their responses to food.</p>	<p>Referral to a dietician may be helpful.</p> <p>Wherever possible choose foods that help the formation of bulky and soft stool instead of hard stool (constipated) or stool too loose to control.</p> <p>With the help of a nutritionist (maybe starting in the hospital) build a healthy digestive system that is individualised to each child.</p>

6.2. School-Aged Children: Building Self-Confidence

Working In Partnership with Families: A Good Start To School Life Can Help Self Confidence

At this time children are learning about the similarities and the differences in the world around them. These may be between places, people and things. The child’s ability to focus on tasks is developing, their memory is increasing. This is a time to help children understand and respect the differences of their friends.

Creating an environment where everyone feels welcome and supported is particularly important for a child born with an anorectal malformation, who may have continence difficulties, may miss school due to hospital appointments, may struggle to form friendships and may feel isolated from their peers.

Families are recommended to talk to prospective schools to see which school will be the best for their child.

Preparation to start school and fixing any problems when they arise is always best done with partnership between the parents, school, and the clinical team.

Overall, in the patient's journey, faecal and urinary incontinence can be quite distressing for the child and the family. Where appropriate, especially for faecal incontinence/soiling the timely provision of trans-anal irrigation or even a stoma can be life-changing and not necessarily a negative step.

Quality of Life (QOL) is very important.

Possible problems	Possible causes	Best practice
<p>Bloated tummy.</p> <p>Faecal overflow/diarrhoea.</p> <p>Soiling.</p> <p>Refusal to sit on the toilet.</p> <p>Poor or reduced appetite.</p> <p>Urinary incontinence.</p>	<p>Chronic constipation.</p> <p>Stool withholding/stool avoidance.</p> <p>Pain/discomfort when trying to have bowels opened.</p> <p>A reluctance to push.</p> <p>The child links eating to discomfort, constipation, and/or soiling.</p>	<p>Care Combination Therapy:</p> <p>Establishing a bowel management programme with a specialist paediatric team. This may include medications (i.e., laxatives and stool softeners), enemas, and bowel washouts.</p> <p>Comfortable, absorbent underwear.</p> <p>Advice may be needed about protective underwear and swimwear.</p> <p>Seating positions that encourage the pelvic floor to relax, such as feet supported on a firm surface (e.g., a stool), leaning forwards, elbows on knees.</p> <p>Core strengthening and pelvic floor exercise with a specialist physiotherapist.</p> <p>Move away from painful, negative pelvic floor experiences to a gentler relationship of trust and cooperation between the child and adult professionals and carer(s).</p>
<p>Continence difficulties in school and settings outside the family home.</p> <p>An unpleasant smell which is noticed by others, but the child does not appear to be aware of.</p> <p>Withdrawn.</p> <p>Anxious.</p> <p>Unhappy.</p> <p>Socially isolated.</p> <p>School absences.</p>	<p>Children with faecal incontinence are often unable to smell their own stool.</p> <p>Wearing diapers/pads.</p> <p>The child may struggle to keep themselves socially clean.</p> <p>Bullying.</p> <p>Illness.</p> <p>Hospitalisation.</p> <p>Clinic appointments.</p>	<p>Providing a personal bathroom:</p> <p>This allows the child to have privacy when they need to use the toilet and manage episodes of soiling which may arise during the school day.</p> <p>Identify a specialist practitioner:</p> <p>A single person “First Point of Contact” to create optimal conditions to achieve continence at home and in school. Provide verbal and written explanations to assist introductions into school.</p> <p>Sharing the diagnosis with the family and school may be necessary to prevent problems escalating.</p>

<p>Low self-esteem. Feeling different. School refusal. Loss of identity.</p> <p>Urinary tract infections. Urinary incontinence.</p>	<p>Living with a rare disease can be very challenging and distressing.</p> <p>Urinary tract infections are common in children born with ARM.</p> <p>The child may need urotherapy including clean intermittent catheterisation (CIC).</p>	<p>Patient support groups:</p> <p>Connecting with others who have a similar condition, realising they are not alone, can be very beneficial to the child.</p> <p>Sharing information and “tips” on how to manage their condition can boost confidence and self-esteem.</p> <p>Maintain adequate fluid intake:</p> <p>Bladder catheterisation may be necessary to protect kidney function. The healthcare team will provide training and support at home and in school</p> <p>Nutrition is very individual:</p> <p>Build a healthy digestive system. Promote fruit, vegetables, and nutritious cereals as tolerated</p>
		<p>Support from healthcare professionals:</p> <p>When well-informed, going to school can be valuable and should be offered.</p> <p>Create a safe, supportive environment for discussions. Provide a multi-disciplinary approach to promote the child’s wellbeing to include a dietician, physical therapist, psychologist, and play therapist.</p> <p>Involve the child in the sharing of their personal information.</p> <p>Annual reviews to ensure best practice care is implemented and maintained. Assess bowel and bladder function, bowel management and washouts, and urological checks to review renal function.</p> <p>Regular monitoring of renal function is essential.</p>

6.3. Meeting The Needs of Patients and Parents

The needs of children born with ARM will vary from child to child and from day to day.

A partnership needs to be established between the family and the professionals involved to ensure that time spent apart from the parents is a happy and enriching experience for the child. Parents need to access a school whose policies set out how they support children with long-term conditions, considering staff training, personalised care planning, medication administration, and integration into school activities.

Care must be coordinated, planned, and incorporate a “whole school” approach to meet the child’s emotional, social, medical, and academic needs. To help the child navigate each day and to reassure their parents, a system must be in place that facilitates regular communication between all parties. The child should be part of this process because monitoring and recording their progress can help them feel valued and in control of their lives. Families and school staff can exchange information to prevent minor issues from becoming major obstacles. The child needs to feel confident that all adults are working collaboratively to support them in enjoying school time.

Some children need access to private bathroom facilities where their practical care is carried out in a private and dignified manner. The intensely intimate issues around continence must be managed sensitively and kindly; the child must feel comfortable and safe with their support staff and trust their health needs remain confidential.

Staff identified for this role must have knowledge and training before they carry out care, and wherever possible, the parents and their child should be involved in the recruitment process of these staff.

The child will likely need time off school when unwell at home, in hospital, or attending appointments with their healthcare team. They need explanations as to why their bodies differ from their friends and why professionals need to monitor their growth and development, discuss their bowel management, and perform any necessary investigations. Providing the child with information in a format they can understand helps them cope with the challenges an anorectal malformation presents, including why, on occasions, they have to miss out on the activities their friends are enjoying.

As children grow, they need to be helped to understand their ARM, giving them a sense of ownership and control of interventions and treatments. Whenever possible, they need to be provided with choices instead of instructions. To equip themselves for this role, parents should be provided with copies of medical records and test results so that when their child asks about their ARM journey, the parents can answer appropriately. Children can help produce a journal of their lives, including photographs of themselves and their healthcare team, identity bracelets, and other items personal to them.

7. STAGE 4: ADOLESCENCE TO ADULTHOOD

“Empowerment: becoming able to deal with adult specialists with a self-determined attitude”

Adolescence is the phase of life between childhood and adulthood, from ages 10 to 19 (as defined by the World Health Organisation).

It is a unique stage of human development and an important time for laying the foundations of good health.

7.1. Transition

This new and rapidly evolving time can be challenging for any young person, but for those born with an ARM, it can be even more daunting as they learn to make decisions and take responsibility for managing their condition. To do this, they need to be equipped with an understanding of how their body functions differently and why it is vital to manage their personal care correctly.

Access to early-life hospital records is essential. This enables the family to explain what occurred in the early weeks and months of their child’s life and gives the adolescent a starting point from which to build on knowledge and understanding of their condition.

Transition should not be rushed or delayed but paced according to the young person’s ability to cope with being different and needing lifelong care. Information detailing the type of ARM and the number and type of operations required in early life will help the young person understand their scars and continence difficulties.

The young person needs to be at the centre of all decision-making to help them develop a more in-depth understanding of their condition and how their health needs may affect their quality of life into adulthood. Teenagers often disengage with healthcare providers, alter their care plans, and omit medications and treatments. Although this approach is a normal part of adolescence, in the longer term, non-compliance can have a detrimental effect on health outcomes. Adolescents need support to understand that today’s treatments can improve their quality of life and longevity.

We know sometimes others can be cruel, even brutal, with hurtful and disrespectful comments. To build self-esteem, adolescents need to know they are in control of their lives and that their condition does not need to be kept a dark secret. Parents and professionals can help by equipping their teenagers with the tools they need to navigate awkward, personal questions from their peers and maintain privacy if that is their preference.

Teenagers can feel overwhelmed with emotional issues, feeling self-conscious, being different from their friends, and struggling with relationships and intimacy. This rollercoaster of emotions can present as anxiety, depression, frustration, and anger. Parents and professionals must help teens develop emotionally healthy ways of living with and managing their chronic illness, encouraging them to ask questions, share ideas and concerns, and support and praise them rather than scolding their sometimes-risky behaviours.

Transitioning to adult services can be very scary; adolescents need referrals to adult healthcare teams who understand and have experience with ARM. A directory of specialists with knowledge and experience of ARM would be beneficial. These would be professionals the teenager can trust and doctors who will support them in the future and understand their early life surgery.

At this time, questions about their future may arise. For some adolescents, sexual health concerns and infertility have a major impact on their quality of life. Friends start discussing menstruation and pregnancy as the norm; however, periods do not happen for some women with complex urogenital conditions. Questions may arise: how do they manage intimate relationships? Will they be able to have a family of their own? Are they the only ones born with an ARM? They need reassurance and love to help them through these challenging years so they do not enter adulthood feeling alone and vulnerable.

Moving Towards Independence

Past Medical History:

Transition includes all involved professionals providing information about the original congenital malformation.

It means providing a properly sorted file with all medical reports, investigations and results, operations, and nursing protocols. This should be headed by a brief overview, to portray the often very complex medical history.

Current Care:

A review of current care and treatments should be completed in a centre of transitional medicine by a multi-disciplinary team. The team should include paediatric and adult surgeons, proctologists, gastroenterologists, sexologists, urologists, gynaecologists, plastic surgeons, stoma therapists, and psychologists.

This collaborative approach will provide the general practitioner with the correct information about the original disease, the actual problems, and the hidden challenges.

It will also help the patient understand their own condition.

Through a patient association, you can contact people your age who are struggling with the same questions. See the glossary below for links to helpful organisations.

7.2. Adolescent Health Needs

Possible problems	Possible causes	Best practice
<p>In males:</p> <p>Urinary tract infections or urinary tract obstruction in males born with rectourethral fistula.</p>	<p>Epididymo-orchitis can be caused by ascending urinary tract infections or reflux of sterile urine into the seminal ducts.</p> <p>This can be present at any age.</p>	<p>Voiding cystourethrogram (VCUG).</p> <p>Urodynamics and cystoscopy are used to detect underlying anatomical or functional causes.</p> <p>If epididymo-orchitis recurs, despite correct treatment, a procedure may be necessary to protect the production of sperm cells. Monitoring and treatment of infections is an essential part of care to protect renal function</p>
<p>In females:</p> <p>Problems with menstruation.</p> <p>In girls, the menarche marks the middle of puberty.</p> <p>Severe lower abdominal pain monthly, especially if breasts and other features of maturity have appeared but periods have not.</p> <p>Severe abdominal pain from menstruation.</p> <p>Females can be susceptible to urinary tract infections.</p>	<p>Care should be taken to ensure that the menstrual flow after menarche drains completely.</p> <p>Blockages in the vagina or uterus can lead to obstructed menstruation and painful bleeding into the abdominal cavity.</p>	<p>Attention to this is important, particularly where uterus duplication is present.</p> <p>A septate “double” vagina (one that is divided into two parts) should be treated.</p> <p>Monitoring and treatment of infections is an essential part of care to protect renal function.</p>
<p>In both males and females:</p> <p>Anxiety about body image.</p> <p>Not knowing who to trust with sensitive information about their bodies.</p>	<p>Fear of intimacy, discomfort, pain.</p> <p>Uncontrollable gas.</p> <p>Soiling. Faeces marking clothing.</p> <p>The smell of stool/urine.</p> <p>Scars may be present.</p> <p>Genitalia may be laid out differently in those born with ARM.</p>	<p>Multi-disciplinary team:</p> <p>Supporting the young person to understand intimate relationships, and to explore their sexuality. When sensitive issues like relationships and intimacy are troubling the young person, a professional can provide support and guidance.</p> <p>Empowerment/Self-Advocacy</p> <p>Providing information to enable the young person to make informed choices about their future health.</p>

7.3. Emotional Wellbeing

The young person must be at the centre of decision-making.

Possible problems	Possible causes	Best practice
<p>Reluctance to gain independence.</p> <p>An overreliance on adults to provide their personal care.</p> <p>Loneliness.</p> <p>Wanting to fit in and have friends.</p> <p>Reluctant to form friendships and/or join clubs.</p>	<p>Parents have been caregivers throughout the young person’s formative years.</p> <p>Parents have previously advocated for them, had discussions with professionals on their behalf.</p> <p>Parents have made decisions about their child’s treatments and healthcare.</p> <p>Extended hospital stays and frequent clinic appointments.</p> <p>Time away from home and school has resulted in reduced opportunities for developing friendships.</p> <p>Feeling different to their peers.</p> <p>They have a rare disease, involving bowel and bladder function and this is often embarrassing to talk about.</p>	<p>The young person should be encouraged and supported to take on the responsibilities of their health needs.</p> <p>This new and evolving role can be challenging for both the adolescent and their family.</p> <p>This can be a time of rapid change for the young person.</p> <p>A balance must be struck to enable them to become independent whilst ideally following their personalised care plan to maintain optimal health.</p> <p>Transition should not be delayed nor rushed but paced alongside the young person’s abilities to manage their personal situation.</p> <p>Parents, family, and professionals can help them with this process.</p>
<p>Sadness, irritability, overwhelm.</p> <p>Social isolation; excluded from friendship circles.</p> <p>Loss of identity.</p>	<p>Discussing intimate issues with others is mainly avoided, sometimes for years.</p>	<p>Parents:</p> <p>Should keep the young person at the centre of decision-making and help them to develop a more in-depth understanding of their condition and how this may impact their adulthood.</p>
<p>Poor self-esteem.</p>	<p>Some adolescents struggle with this change and may fear taking full responsibility for their care.</p> <p>Some adolescents enjoy the prospect of independence and actively push parents away to achieve this.</p>	<p>Parents and Professionals:</p> <p>Should actively listen and encourage self-confidence, self-reliance and promote independence.</p> <p>Change can be overwhelming and patient associations can provide contact with other young people facing these changes.</p>
		<p>Professionals:</p> <p>Should provide a single point of contact, a care coordinator, to assist with transition.</p> <p>Someone available to answer sensitive questions, who can empower the adolescent to have control over their bowel management programme.</p> <p>It is essential to support a smooth transition to specialist adult services.</p> <p>With growing independence, knowing how to contact their care coordinator can empower the young person.</p>

7.4. Relationships: Male Adolescent Intimacy

Please be reassured that not all adolescents face these difficulties.

Advice and support can be found from many sources.

Possible problems	Possible causes	Best practice
<p>Sexual relationships can present difficulties both for oral sex and intercourse.</p> <p>Some young men may experience dysfunction of erection and ejaculation.</p> <p>Producing and maintaining an erection may be difficult.</p> <p>Ejaculation can be painful.</p> <p>Premature ejaculation - this is when an orgasm comes faster than preferred.</p>	<p>These problems may result from spinal and/or bladder complications related to the ARM and/or surgical repair.</p> <p>Semen may not exit through the penis but travel backwards into the bladder. (known as retrograde ejaculation)</p> <p>Buttocks, anus, and genitalia can be different for those born with anorectal malformation and impact the sexual experience.</p>	<p>Your family doctor can help by listening and where appropriate arranging a referral for investigations and treatment with a urologist experienced in congenital anorectal malformation.</p> <p>Referral to a sexologist also known as a sex therapist. This is a professional trained in understanding emotional and physical difficulties associated with anorectal malformations. and the impact on sexual health</p>
<p>Some adolescents with a stoma, scars, or continence problems, may find intimacy very difficult and avoid relationships.</p> <p>Genitalia may be laid out differently in those born with an ARM.</p> <p>Physical limitations during sex.</p> <p>Lowered libido - decreased desire to have sexual relationships.</p> <p>Some men experience episodes of faecal or urinary incontinence during sexual intercourse.</p> <p>Intimate touch may trigger memories of invasive procedures in childhood.</p>	<p>Scars can cause fear of rejection and feelings of shame.</p> <p>Pelvic floor structures are weaker than in most males.</p> <p>Relaxation of abdominal muscles may result in faecal leakage.</p> <p>This can be related to childhood treatments involving rectal examinations, investigations, bowel, and colon irrigations.</p>	<p>What may help:</p> <p>Being able to share your concerns with your partner.</p> <p>Connecting with others with the same difficulties can help you overcome some of these barriers.</p> <p>Faecal incontinence may be improved by following a bowel management programme and advice from a colorectal specialist can be invaluable.</p> <p>Click here for Sexual Support: Anorectal Malformations & Hirschsprung's Disease</p>

7.5. Relationships: Female Adolescent Intimacy

Please be reassured that not all adolescents face these difficulties.

Advice and support can be found from many sources.

Possible problems	Possible causes	Best practice
<p>Sexual relationships can present difficulties both for oral sex and intercourse.</p> <p>Physical limitations during sex.</p> <p>Lowered libido - decreased desire to have sexual relationships.</p> <p>Uncomfortable, painful intercourse.</p>	<p>These problems may result from spinal and/ or bladder problems related to the anorectal malformation and or the surgical repair.</p> <p>Buttocks, anus, and genitalia can be different for those born with ARM and impact the sexual experience.</p> <p>Vaginal dryness.</p> <p>A reduced blood supply in the clitoral area.</p> <p>Vaginal narrowing from previous surgery.</p>	<p>Your family doctor can help by listening and where appropriate arranging a referral for investigations and treatment from a gynaecologist experienced in congenital ARM.</p> <p>Referral to a sexologist also known as a sex therapist. This is a professional trained in understanding emotional and physical difficulties associated with anorectal malformations and the impact on sexual health.</p>
<p>Some adolescents with a stoma, scars, or continence problems, may find intimacy very difficult and avoid relationships.</p> <p>Some females can experience episodes of faecal incontinence during sexual intercourse.</p> <p>Intimate touch may trigger memories of invasive procedures in childhood.</p>	<p>Pelvic floor structures are weaker than in most women.</p> <p>Relaxation of abdominal muscles may result in faecal leakage.</p> <p>Scars can cause fear of rejection and feelings of shame.</p> <p>This can be related to childhood treatments involving rectal examinations, investigations, bowel, and colon irrigations.</p>	<p>What may help:</p> <p>Being able to share your concerns with your partner.</p> <p>Connecting with others with the same difficulties can help you overcome some of these barriers.</p> <p>Faecal incontinence may be improved by following a bowel management programme and advice from a colorectal specialist can be invaluable.</p> <p>Click here for Sexual Support: Anorectal Malformations & Hirschsprung's Disease</p>

8. STAGE 5: ADULTHOOD

Most healthcare providers have not heard of ARM, and when adults ask professionals for help and advice, they are sometimes unable to see the connection between the patient's symptoms and their birth condition. Despite the patient trying to explain what they are experiencing, often they are dismissed and feel they have not been heard. Many adults struggle to explain themselves, becoming anxious and distressed. This is further complicated if they are affected by medical trauma from multiple interventions and treatments received in childhood.

This is a very challenging time. Previously, parents have spoken to the medical team, but now the patient is an adult and must advocate for themselves. Enabling them to discuss their health issues with healthcare providers is vital. Patients need to feel safe and have their opinions valued and respected.

Continence issues can significantly impact further education, job prospects, and career opportunities. Adults born with an ARM should not feel their wings have been clipped but instead believe that with the correct support, they can live a life unlimited. There is often a struggle to become socially clean. Hence, adults must know how to navigate their healthcare system and be signposted to the appropriate specialists to assess and treat symptoms swiftly.

For some adults, sexual health concerns and infertility have a major impact on their quality of life, causing worry and distress. Friends discuss menstruation and pregnancy as the norm, yet for some females with complex urogenital conditions, periods do not occur, and some may never conceive. Family doctors need to respect the urgency around fertility; their patients may need further investigations and surgery before they can conceive. It can feel that the biological clock is running out whilst waiting for a uro-gynaecologist and specialists in reproductive medicine to be found.

A directory of specialists with knowledge and experience of ARM would be beneficial; adults can put their trust in doctors who understand their early-life surgery. Patients need to be able to choose the gender of their specialist; having a male doctor can sometimes make it much harder for female patients to express themselves, to share problems relating to intimacy and continence, and particularly embarrassing having physical examinations.

Adults need professionals who understand the interplay of the health issues found in rare and complex malformations. They need attention to their physical and psychosocial needs from empathetic healthcare providers and supportive family members and friends who value and respect them.

8.1. Male Adults Born With Anorectal Malformations

It is important to remember that good physical and emotional health outcomes are possible. Be reassured that you are not alone. Advice and support can be found from many sources.

Possible problems	Possible causes	Best practice
Urinary tract infections in males born with rectourethral fistula.	These problems can result from spinal and/ or bladder problems related to ARM and or surgical repair.	Voiding cystourethrogram (VCUG), urodynamics, and cystoscopy are used to detect underlying anatomical or functional causes.
Epididymo-orchitis. Fertility may be affected.	Ascending urinary tract infections, reflux of sterile urine into the seminal ducts, or anomalies in the genital tract.	If epididymo-orchitis recurs despite correct treatment, a procedure may be necessary to protect the production of sperm cells. Referral to a centre specialising in reproductive medicine.
Some men may experience dysfunction of erection and ejaculation. Ejaculation can be painful. Premature ejaculation when an orgasm comes faster than preferred.	Semen may not exit through the penis but travel backwards into the bladder, (retrograde ejaculation).	Family doctors: Help by listening and arranging referrals for investigations and treatment from a urologist experienced in congenital anorectal malformation.
Incontinence. Constipation with overflow / diarrhoea. Soiling and smelling of stool/urine. Faecal staining of underwear. Uncontrolled passing gas or farting. Some men experience episodes of faecal or urinary incontinence during sexual intercourse.	Can be linked to ARM.	Referral for bowel management advice from a colorectal specialist can be invaluable. Other specialists, such as a sexologist who is qualified in sexual health, may provide advice and support. Being able to share your concerns with your partner. Connecting with other adults with the same difficulties can help you overcome some of these problems.
Adults with a stoma: Faecal leakage from the stoma bag. Soiling of clothing. Problems with adherence of appliances. Rectal bleeding, discomfort, mucus discharge. Deterioration in bowel and/or bladder function.	The skin can become sore-excoriated. Stoma bag not fitting comfortably. Rectal prolapse, related to ARM, weakened pelvic floor structures, previous surgery.	Advice from stoma therapists: Click here

8.2. Female Adults Born With Anorectal Malformations

It is important to remember that good physical and emotional health outcomes are possible. Be reassured that you are not alone. Advice and support can be found from many sources.

Possible problems	Possible causes	Best practice
Problems with periods.	Care should be taken to ensure that after menarche the menstrual flow drains completely.	Attention to this is important particularly where uterus duplication is present.
Urinary tract infections.	Can be more noticeable following sexual intercourse.	Family doctors: Help by listening and arranging referrals for investigations and treatment from a gynaecologist experienced in congenital ARM.
Vaginal aplasia or double vagina, Uterus aplasia Sexual relationships can present difficulties both for oral sex and intercourse. Uncomfortable intercourse. Painful intercourse. Intimate touch may trigger memories of invasive procedures in childhood	In those born with ARM, the genitalia may be different from most women. Vaginal dryness. A reduced blood supply in the clitoral area. Vaginal narrowing from previous surgery. Linked to childhood treatments involving rectal examinations, bowel irrigations.	Other specialists, such as a sexologist who is qualified in sexual health, may provide advice and support. Being able to share your concerns with your partner. Connecting with other adults with the same difficulties can help you overcome some of these problems.
Incontinence. Constipation with overflow / diarrhoea. Soiling and smelling of stool/urine. Faecal staining of underwear. Uncontrolled passing gas or farting. Some women experience episodes of faecal or urinary incontinence during sexual intercourse.	Pelvic floor structures are weaker than in most women.	Referral for bowel management advice from a colorectal specialist or gastroenterologist can be invaluable.
Fertility may be affected. Anxiety related to pregnancy and childbirth.		Referral to a centre which specialises in reproductive medicine. Pre-conceptual care should be available. Additional reassurance and support may be necessary. Regular access to GPs, midwives, and obstetricians to allay concerns. Vaginal delivery in females born with ARM should be avoided because of the increased risk of injury to their pelvic floor and further weakening of the bowel and bladder.

<p>Abdominal scars from surgeries.</p> <p>Rectal prolapse related to ARM.</p>	<p>Weakened pelvic floor structures.</p> <p>Previous surgery.</p>	<p>Best practice means referral to a professional with experience of ARM</p> <p>Help from other disciplines like physiotherapists.</p>
<p>Adults with a stoma:</p> <p>Faecal leakage from the stoma bag.</p> <p>Soiling of clothing,</p> <p>Problems with adherence of appliances</p> <p>Rectal bleeding, discomfort, and mucus discharge.</p> <p>Deterioration in bowel and or bladder function.</p>	<p>The skin surrounding the stoma can become sore-excoriated.</p> <p>The stoma bag is not fitting comfortably. Stool leaking from stoma onto the skin.</p>	<p>Advice from stoma therapists: Click here</p>

8.3. Lifelong Physical And Psychological Support

Lifelong physical and psychological support is needed as the adult copes with the uncertainty of their rare disease.

- Information to help them understand how ARM may impact their health as they age.
- Referral to colorectal specialists.
- Dietary advice.
- Bowel/bladder management programme.
- Pelvic floor physiotherapy.
- They may need an operation.

To be born with an ARM means living with a rare and complex disease.

Holistic care is an understanding of the person's physical, psychological, emotional, and spiritual needs.

Incontinence, which may accompany this condition, can be offensive for some members of society, and this, being outside the social norm, can leave the individual feeling ashamed and alone.

Adults benefit from supportive family members and friends who value and respect them.

Appropriate support can improve mental health and wellbeing and reduce social isolation.

Attention to physical and psychosocial needs from empathetic healthcare providers is essential.

Adults need professionals who recognize the interplay of the health issues found in rare and complex malformations.

A multi-disciplinary, holistic approach to care can contribute to improved quality of life and empower adults to live a life unlimited.

9. VACTERL ASSOCIATION

All babies with an ARM should be examined for other congenital problems to ensure other conditions are not overlooked.

Some infants may have additional congenital anomalies diagnosed as VACTERL Association. VACTERL is an acronym for **V**ertebral, **A**norectal, **C**ardiac, **T**racheo-oesophageal, **R**enal, and **L**imb. This collection of anatomical differences occurs in the early stages of foetal development and affects around 1:10,000 - 1:40,000 of the population.

Babies with anorectal malformation are diagnosed as having VACTERL Association if, in addition to an ARM, they have two further associated anomalies as listed below:

Area	Possible problems
Vertebral	Affecting the bones that make up the spine. Some bones may be misshapen, fused or missing. There may be problems with pinched nerves which can result in problems with mobility, bladder, and bowel function.
Anorectal	See the rest of this document.
Cardiac	Affecting the baby's heart. The most common presentation is a ventricular septal defect (VSD) also known as a hole in the heart. Other heart conditions can be present.
Tracheo-oesophageal fistula/oesophageal atresia (TOF/OA)	Occurs when there is an abnormal connection between the trachea and the oesophagus, and the oesophagus has not formed correctly.
Renal	Affecting the kidneys and the urinary tract. Problems can vary in severity. Investigations and monitoring are essential to preserve renal function.
Limb	Most commonly affecting the thumb or forearm. The baby may have under development or absence of bones.

Many of the VACTERL conditions are dealt with in the first year of life, with expert paediatric teams prioritising the necessary treatments, including operations, to optimise the baby's immediate condition and improve the quality of their long-term health.

This can be a time of great uncertainty, with parents' fears for their baby's future at the forefront of their minds. Healthcare providers must ensure that parents remain central to discussions and decision-making at this time.

No two children with VACTERL Association are likely to be affected in the same way.

Thanks to increased understanding of the condition, expert management and care, health outcomes have improved, although some children will have lifelong health needs.

Therefore, it is essential that healthcare practitioners provide a high-quality transition from children's services to centralised adult services. Multi-disciplinary, collaborative care pathways with targeted, holistic support should address the young adult's educational, vocational, and psychosocial needs.

Some adults have become their own "patient experts," but many still need the support of an advocate health professional. Some adults may avoid visiting the doctor when there is a problem due to prior bad experiences that deter them from seeking the care they need.

Even if well-informed, the power imbalance between the patient and medical staff, particularly when the patient is feeling unwell, may make the patient unable to speak up for themselves.

A care coordinator can be invaluable at this time, ensuring the adolescents' and adults' changing health needs are identified and, wherever possible, met, helping all to live their best lives.

10. USEFUL ARTICLES & LINKS

Articles

Postoperative complications in adults with anorectal malformation: a need for transition: Schmidt D, Jenetzky E, Zwink N, Schmiedeke E, Maerzheuser S.. German Network for Congenital Uro-Rectal Malformations (CURE-Net). *Pediatr Surg Int* 2012;28:793–5. <https://pubmed.ncbi.nlm.nih.gov/22772590/>

Transition of care in patients with anorectal malformations: Consensus by the ANORECTAL MALFORMATION-net consortium: Giuliani S, Grano C, Aminoff D, Schwarzer N, van de Vorle M, Crétolle C, et al.: *J Ped Surg* 2017;52:1866–72.). <https://pubmed.ncbi.nlm.nih.gov/28688794/>

Links

“ARM - Dilatations after surgery” (ERN eUROGEN Webinar 93): <https://www.youtube.com/watch?v=v0ibiDRyaGo>

“Caring for your child after Reconstruction surgery for IA/ARM” video (produced by the Division of Colorectal and Pelvic Reconstruction at [Children’s National Hospital](#), Washington DC, USA): <https://www.youtube.com/watch?v=wmWZsnvjtc>

Adult Stoma Care: <https://www.coloplast.co.uk/stoma/people-with-a-stoma/>

Alone We Are Rare, Together We are Strong: https://rarediseases.org/wp-content/uploads/2014/12/NRD_Newsletter_LR-2011.pdf

C Krickenbeck classification (see Reference 2)

EURORDIS Mental Health and Wellbeing: <https://www.eurordis.org/mental-wellbeing/>

I’m a teenager – do I care if my disease is rare?: <https://blogs.biomedcentral.com/on-biology/2016/02/22/teenager-care-disease-rare>

Max’s Trust UK: <https://maxtrust.org>

Sexual Support: Anorectal Malformations & Hirschsprung’s Disease: <https://www.sexuality-arm-hd.com/>

VACTERL Association UK: <https://www.vacterl-association.org.uk/>

VACTERL: Congenital Anomalies Occurring with OA/TOF Booklet: <https://tofs.org.uk/2023/07/new-booklet-released-vacterl-congenital-anomalies-occurring-with-oa-tof/>

11. GLOSSARY

Term	Definition
Anorectal malformations	Birth defects in which the lower end of the digestive tract (rectum and anus) does not develop properly.
Cloacal malformation	Birth defect where the urethra, vagina and rectum fail to separate into three distinct tubes resulting in a single channel.
Colostogram	Provides information re: location of the lower colon and how it may connect to the urinary system or genitalia.
Hydrocolpos in anorectal malformation	A collection of fluid within the vagina - can indicate a single opening.
Hydronephrosis	Kidney has become stretched due to a build-up of urine.
Renal dysplasia	Kidney has not developed properly.
Renal agenesis	The absence of one or both kidneys.
Renal duplications	The presence of two ureters (normally one) coming from a single kidney.
Spinal dysraphism	A congenital malformation resulting in an abnormal structure in the spine, including the bony structure, the spinal cord, and the nerve roots.
Tethered spinal cord	The spinal cord is pulled down and fixed to the spinal canal.
VACTERL Association	Affects many body systems: Vertebral, Anal, Cardiac, Tracheo-Oesophageal Fistula, Renal, and Limb.
Vaginal aplasia	A rare malformation in which the vagina does not develop properly.
Vesicoureteral reflux	A condition in which urine flows backwards from the bladder – sometimes to the kidneys.

12.SUPPORTING TOOL

Parents can ask: "Is my child in the best hands for managing their condition?"

This Anorectal Malformation Parents / Caregivers Checklist can help you make an informed choice.

Are you unsure about your child's future? Do you think a second opinion is needed?

Would you like a second opinion?

Facts	Yes	No
Our child is suspected to have an anorectal malformation		
Our child has additional anomalies or associated malformations		
Our child has urological problems		
The hospital has little experience in treating patients with anorectal malformation		
We are not sure if our child is receiving optimum care in this hospital		
We have not been given information about risks, possible complications, and frequent postoperative problems		
The medical approach is unclear and difficult for us to understand		
Doctors often contradict each other in their communication and conclusions		
The answers to our questions are not satisfactory		
We don't always have the same contact person(s); we often see someone different		
We need more consistent and reliable information to make the right decisions about the treatment of our child		

Patients must always receive care from a skilled surgeon and an experienced multi-disciplinary team with access to examinations and investigations to ensure an accurate diagnosis. Having a second opinion can provide the best possible treatments and long-term outcomes for your child.

If you answered YES to one or more of these statements, you are advised to seek a second opinion.

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14. REFERENCES

1. Prevalence/Incidence:

https://eu-rd-platform.jrc.ec.europa.eu/eurocat/eurocat-data/prevalence_en

<https://angeborene-fehlbildungen.com/>

Wilms M, Jenetzky E, Märzheuser S, Busse R, Nimptsch U. Treatment of anorectal malformations in German Hospitals: Analysis of national hospital discharge data from 2016 to 2021. Eur J Pediatr Surg. 2024 Feb 2. DOI: [10.1055/a-2260-5124](https://doi.org/10.1055/a-2260-5124)

2. Link classification:

Holschneider A, Hutson J, Peña A, Beket E, Chatterjee S, Coran A, et al. Preliminary report on the international conference for the development of standards for the treatment of anorectal malformations. J Pediatr Surg. 2005;40:1521–6.

DOI: [10.1016/j.jpedsurg.2005.08.002](https://doi.org/10.1016/j.jpedsurg.2005.08.002)



European Reference Networks

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



European Reference Network

for rare or low prevalence
complex diseases

 **Network**
Urogenital Diseases
(ERN eUROGEN)

<https://eurogen-ern.eu/>



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