Patient Journey for Anorectal Malformation

A LIFELONG JOURNEY TAKEN ONE STEP AT A TIME

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

SOMA

AIMAR
Associazione Italiana Malformazioni Ano-Rettali

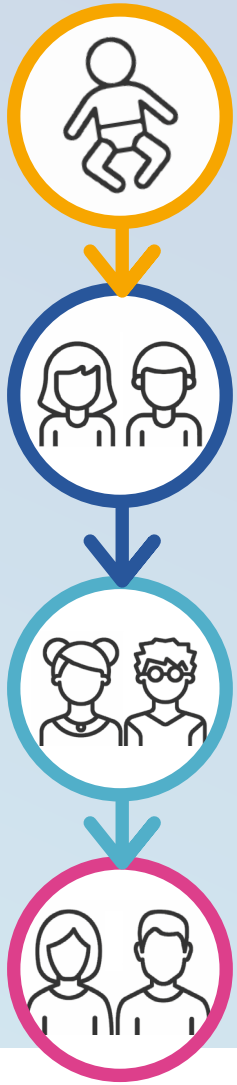
EAT
Esophageal ATresia Global Support Groups

Introduction

What is Anorectal Malformation?

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 patients, their parents and other family members.

- Anorectal malformation (ARM) is a rare and complex congenital disorder in which babies are born without an anal opening or the opening is in the wrong position.
- ARM represents a spectrum of abnormalities which affect the lower end of the digestive tract and interfere with the normal passage of stool.
- The condition ranges from minor to more complex malformations.
- In almost 50% of infants born with ARM other parts of the body may be affected, including the bladder, heart, limbs, and/or the sacrum.



BIRTH ▸ ONE YEAR

Facts about ARM

- Each infant is unique, their own pathway determined by the type of ARM and the treatment needed.
- Some infants will need one operation, whilst those with more complex anomalies will need three operations or more.

Aim » Best possible outcomes: A healthy, relatively uneventful childhood. Independent adults with a good quality of life.

Incidence of Anorectal Malformation: between 1:3500 to 1:5000 live births.
This means that throughout Europe there are many thousands of people living with the daily challenges of this condition.

Most babies are not be diagnosed prenatally.

A second trimester ultrasound may detect abnormalities within the urinary tract and lower pelvis.

! Babies need urgent medical attention to relieve bowel obstruction.

Pre-operative assessment, diagnosis and surgery are challenging.

Expertise and experience is essential to provide best long-term outcomes.

BIRTH ▸ ONE YEAR

Diagnosis and first symptoms

- A diagnosis of Anorectal Malformation should ideally be made in the first hours of life.
- Some babies may appear well, and the abnormality may be missed, particularly if stool is passed via the vagina, via a tiny perineal orifice or, very rarely, via the urethra in males.
- In some cases the baby may be discharged home without a diagnosis.

Indications of ARM

- A swollen tummy/colic
- Delayed or non passing of first stool
- Passing of ribbon stool
- Vomiting and dehydration
- Reluctance or refusal to take a feed
- Unsettled, distressed

! Urgent referral to healthcare team

BIRTH ▸ ONE YEAR

Confirming the diagnosis

What you can expect to happen:

- Newborn assessment
- Examination of the perineum and genitals
- A check if first stool (meconium) is passed from the correct bottom hole

! The absence of a bottom hole should be investigated further.

Associated anomalies may be present, further tests may be needed:

- Abdominal X-ray
- Ultrasound scan /MRI lower pelvis to check the spinal channel
- Ultrasound scan kidneys
- Echocardiogram – heart anomalies

Best practice:

- Parents are central to decision-making
- Healthcare team ensure excellent communication at all times
- Detailed care plan, including a timeline for treatments, necessary operations
- Family support worker and psychologist
- Support to siblings, immediate family
- Collaboration between hospital and home care teams
- Referral to appropriate patient organisation(s)
- Contact with parents who have a child with a similar condition
- Local support systems: who to contact if problems arise

BIRTH ▸ ONE YEAR

Formation of a temporary stoma

What you can expect to happen:

- A small section of bowel opens onto the tummy, bowel can now function normally
- Lower bowel is free from stool in preparation for surgery
- Baby can feed and thrive before further operations

Introduction of a specialist nurse:

- Parents are trained to care for their baby's stoma >>
- Healthcare professional is named contact for care at home

No one should feel alone or unsupported!

Best practice:

- Explanation/written information about the type of ARM and the operation required
- Parent/family accommodation should be available
- Travel costs may be available
- Maintain appropriate midwifery/ obstetric care for the mother

Before the main procedure/repair assessment/investigations may be needed:

- Distal Colostogram: >>
  X-ray – detailed image of ARM
- Voiding Cystourethrogram: >>
  Detect problems with urinary flow

BIRTH ▸ ONE YEAR

Corrective surgery

What you can expect to happen:

- Timing of corrective surgery depends on surgeons preference
- Rectum is positioned within the anal sphincter muscles
- A catheter may be placed to drain urine from the bladder
- Hospital stay of about one week
- Anal dilatations after surgery >>
  (may not be necessary)

- Parents feel comfortable, confident providing care
- Child is happy to play and interact with family
- Bowel is functioning
- Child is eating, drinking

Patients need a skilled surgeon working with a multi-disciplinary team.

Best practice:

Careful postoperative management:

- Good pain management
- Not sitting for too long
- Diaper change – no position which keeps legs widely separated
- Loose-fitting clothing
- Keep the repair site clean and dry
- Stitches are usually dissolvable

After discharge from hospital:

- Home-Specialist nurse
- Post-operative review
- Regular appointments

BIRTH ▸ ONE YEAR

Closure of the stoma

What you can expect to happen (1/2):

The surgeon reconnects the intestine, providing bowel continuity, allowing normal passing of stool via the newly formed anus.

In the initial postoperative period:

- The baby may not be allowed to take fluids/food for a couple of days after the operation
- Intravenous fluids during this time to maintain hydration
- Stool passing within two to three days

Best practice:

- **Monitoring by the childs health care team:**
  Wound healing, bowel sounds, passing gas, type of and frequency of stool
- Advice about skin cleansing, wipes, creams, lotions individual for each child

BIRTH ▸ ONE YEAR

Closure of the stoma

What you can expect to happen (2/2):

After discharge from the hospital:

- Bowel function can be unpredictable and challenging
- Initially stool is watery/loose and may cause skin irritation
- Later constipation/overflow of faeces may be mistaken for normal bowel action

Signs and symptoms that may be an indication of bowel obstruction:

- Severe colic or upset tummy
- Green vomiting
- Refusing to take feeds

Serious condition known as an ileus requires urgent medical attention. Contact clinical team if any of these happen.

Best practice:

- Post operative surgical review after two weeks
- Regular follow up – surgical team
- Specialist nurse – home care

Dietary advice:

- Laxative foods:
  Stool hard/difficult to pass
- Constipating foods:
  Stool is very loose

- Appointments:
  Paediatric Urologist,
  Neurologist as necessary

INFANCY ▸ SCHOOL AGE

Bowel continence is a complex process
needing motility, sensation, and a functioning sphincter.

Despite a successful operation, continence can be a challenge, the child may be slower to gain bowel control.

A safe, supportive environment
for discussions and the employment of a specialist practitioner are essential.

Coordinating care between home and school, monitoring growth and development, identifying and resolving problems as they arise

A multi-disciplinary approach
to promote the child's wellbeing is essential; to include a dietician, physical therapist, psychologist, and play therapist.

! It is important to involve the child in sharing of their personal information.

The children need help to understand their ARM,
giving them a sense of ownership and control of interventions and treatments. They need to be provided with choices instead of instructions.

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.

INFANCY ▸ SCHOOL AGE

What may happen:

Best practice:

What may happen:	Best practice:
• Poor, reduced appetite, food intolerances, sensitivities • Child may struggle to keep clean • Constipation, overflow, soiling • Stool withholding, stool avoidance • Need for diapers/protective clothing	• Nurture cooperation between child, parent, professionals, carers • Bowel management programme • Medications, enemas, bowel washouts • Seating positions – encourage pelvic floor relaxation • Core strengthening, pelvic floor exercises
• Urinary incontinence, infection	• Bladder catheterisation may be necessary – training and ongoing support is provided
• Hospital appointments, admission Illness – isolated from friends	• Ensure Kindergarten, preschool staff understand ARM • Provide verbal, written explanations support introductions to school
• School refusal – low self esteem, withdrawn, anxious, unhappy	• Finding the right school for your child. Sharing diagnosis and care needs with identified school staff can prevent problems escalating

ADOLESCENCE / all genders

Adolescence is a unique stage of human development, an important time for laying down the foundations of good health.

Learning that today's self care/ treatments can improve quality of life and longevity.

Collaboration between paediatric and adult teams and parents should be in place to enable the adolescent to develop emotionally healthy ways of living with and managing their chronic illness.

What may happen:

- Anxiety about body image
- Fear of intimacy, discomfort, pain
- Soiling, faeces marking clothing
- Smell of stool/urine
- Scars
- Genitalia different
- Loneliness
- Reluctant to form friendships
- Low self-esteem

Best practice:

Empowerment/Self-Advocacy

- Young person at the centre of decision making
- Make informed choices about their health
- In depth understanding ARM – impact adulthood

Parents/Professionals:

- Listen, encourage self-confidence
- Self-reliance, promote independence
- Patient associations – contact with other young people
- Transition – single point of contact/care coordinator
- Signposting – multidisciplinary professionals

ADOLESCENCE / female

What may happen:

- Abnormalities in vagina/uterus – obstruction to menstrual flow
- Urinary tract infections

Best practice:

- Ensure menstrual flow drains completely
- Monitoring/treatment of infections

Relationships: Female Adolescent Intimacy

Not all adolescents face these difficulties.

- Sexual relationships can present difficulties for oral sex and intercourse
- Physical limitations during sex
- Uncomfortable, painful intercourse
- Anatomy of genitalia can be different
- Vaginal dryness
- Reduced blood supply in clitoral area
- Vaginal narrowing
- Relaxation may result in faecal leakage
- Adolescents with a stoma, scars, continence issues may find intimacy difficult
- Shame/fear of rejection

- Sharing concerns with your partner
- Connecting with others with the same difficulties

Family doctor referrals:

- Colorectal specialist
- Bowel management programme
- Urologist
- Gynaecologist experienced in Anorectal Malformation
- Sexologist (sex therapist) >>
- Psychologist
- Stoma therapy advice

ADOLESCENCE / male

What may happen:

- Urinary tract infections
- Urinary tract obstruction
- Epididymo-orchitis

Best practice:

- Urodynamics and cystoscopy to assess urinary system
- Monitoring/treatment of infections

Relationships: Male Adolescent Intimacy

Not all adolescents face these difficulties.

- Sexual relationships can present difficulties for oral sex and intercourse
- Dysfunction of erection and ejaculation
- Painful/premature ejaculation
- Semen may travel backwards into bladder
- Relaxation may result in faecal leakage
- Adolescents with a stoma, scars, continence issues may find intimacy difficult
- Shame/fear of rejection

- Sharing concerns with your partner
- Connecting with others with the same difficulties

Family doctor referrals:

- Urologist investigations/ treatment
- Colorectal specialist
- Bowel management programme
- Urologist
- Sexologist (sex therapist) >>
- Psychologist
- Stoma therapy advice

ADULTHOOD / all genders

Structured lifelong holistic care is needed.
Provided by a multidisciplinary team, skilled in the care of patients born with Anorectal Malformation. Attention to physical and psychosocial needs from empathetic healthcare providers is essential.

Adults born with ARM must have their lived experience valued and respected.
A multi-disciplinary, holistic approach to care can contribute to improved quality of life and empower adults to live a life unlimited.

Good physical and emotional health outcomes are possible. However for some adults continence issues can significantly reduce education and career opportunities.

Sexual health concerns and infertility can also have a major impact on quality of life.

Be reassured that you are not alone – advice and support can be found from many sources!

ADULTHOOD / female

What may happen (1/2):

- Problems with menstruation notably where uterus duplication is present:
  Vaginal aplasia, double vagina, uterus aplasia

- Sexual relationships can present difficulties both for oral sex and intercourse
- Genitalia may be different
- Painful intercourse
- Vaginal dryness/narrowing, reduced blood supply in the clitoral area

- Deterioration in bowel/ bladder function
- Constipation /faecal leakage
- Rectal prolapse/rectal bleeding
- Faecal/urinary incontinence during intercourse
- Uncontrolled passing gas or farting
- Soiling/smelling of stool/urine

Best practice:

Family doctor referrals:
- Gynaecologist experienced in ARM

- Sexologist (sex therapist) >>

- Colorectal specialist
- Bowel management programme
- Physiotherapist

ADULTHOOD / female

What may happen (2/2):

Best practice:

What may happen (2/2):	Best practice:
• Adults with a stoma, scars, continence issues may find intimacy difficult	• Stoma therapy advice >>
• Urinary tract infections, particularly following sexual intercourse	• Urologist investigations/ treatment
• Shame/fear of rejection	• Psychologist
• Fertility may be affected	• Reproductive medicine specialist • Pre-conceptual care • Access to GPs, midwives, and obstetricians
• Anxiety – pregnancy/childbirth	• Vaginal delivery in females born with ARM should be avoided because of the increased risk of injury to their pelvic floor and further weakening bowel/ bladder
• Reluctance to share concerns	• Wherever possible share your concerns with your partner • Connect with patient organisation(s)

ADULTHOOD / male

What may happen (1/2):

- Urinary tract infections
- Epididymo-orchitis
- Erectile/ejaculation dysfunction
- Premature ejaculation
- Semen may travel backwards into bladder

Best practice:

Family doctor referrals:

- Urologist investigations/ treatment
- Recurrence of epididymo-orchitis protection of sperm may be necessary

- Sexual relationships can present difficulties both for oral sex and intercourse
- Genitalia may be different

- Sexologist (sex therapist) >>

- Deterioration in bowel/ bladder function.
- Constipation/faecal leakage
- Rectal prolapse/rectal bleeding
- Faecal/urinary incontinence during intercourse
- Uncontrolled passing gas or farting
- Soiling/smelling of stool/urine

- Colorectal specialist
- Bowel management programme
- Physiotherapist
- Urologist

ADULTHOOD / male

What may happen (2/2):

Best practice:

What may happen (2/2):	Best practice:
• Adults with a stoma, scars, continence issues may find intimacy difficult	• Stoma therapy advice >>
• Shame/fear of rejection	• Psychologist
• Fertility may be affected	• Reproductive medicine specialist
• Reluctance to share concerns	• Wherever possible share your concerns with your partner • Connect with patient organisation(s)

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This brochure is an overview of the Anorectal Malformation Patient Journey. A more detailed version and a separate list of International Patient Organisations can be found under "Patient Information": >>
https://eurogen-ern.eu/workstreams-and-expertise/workstream-1/urorectal-anorectal-malformations/

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