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

ERN eUROGEN Transition Care for Individuals with Rare Uro-Recto-Genital Conditions

A Lifelong, Patient-Centered Approach

Published: 28 July 2026

Authors: The ERN eUROGEN Transition & Lifelong Care Working Group; Raimund Stein (Chair), Dalia Aminoff, Felix Campos Juanatey, Celia Cretolle, Noemi Deanesi, Anne Ebert, Margit Fisch, Magdalena Fossum, Ramon Gorter, Michel Haanen, Lieke Janssen, Kolja Kvist, Marc Miserez, Giovanni Mosiello, Matthieu Peycelon, Rebecca Pulvirenti, Katharina Rall, Alexander Stillebroer, Kate Tyler, Luisa Weil, Gjertrud Wennevik, Miriam Wilms, Luc Zwiep

Table of Contents

1. INTRODUCTION

Rare uro-recto-genital conditions require lifelong healthcare that extends beyond childhood and adolescence. Advances in medical and surgical management have significantly improved survival and long-term outcomes, resulting in a growing population of adolescents and adults living with complex congenital conditions. Consequently, healthcare systems must ensure continuity of specialised care throughout the lifespan.

Individuals with rare uro-recto-genital conditions often face evolving healthcare needs related to urinary and bowel function, kidney health, sexual health, fertility, psychosocial wellbeing, education, employment, body image, and independent living. Many have undergone multiple interventions during childhood and require lifelong surveillance and support. While healthcare professionals and clinical environments may change over time, the patient and family remain the constant element throughout the healthcare journey. This highlights the need for a coordinated, patient- and family-centred approach that ensures continuity, clear communication, and sustained support across all stages of life.

Transition should therefore be regarded as a structured and purposeful process rather than a single transfer event. It aims to prepare young people and their families for adult-oriented healthcare, promote self-management and autonomy, and ensure uninterrupted access to appropriate specialist care. Despite increasing recognition of its importance, transition care remains inconsistently implemented across Europe (1, 2). The absence of structured pathways may result in fragmented care, loss to follow-up, preventable complications, and reduced quality of life.

2. PRINCIPLES AND ORGANIZATION OF TRANSITION CARE

All individuals with rare uro-recto-genital conditions should be offered a structured transition pathway as part of lifelong follow-up. Transition planning should begin before adolescence and be adapted according to the individual's medical condition, developmental maturity, psychosocial circumstances, and personal goals. Transition should be recognised as a continuous process that spans the transition period, rather than a single transfer event based on age alone.

Transition care should be accessible and inclusive for all patients and responsive to individual needs, including neurodiversity, learning disabilities, and mental health considerations. It should promote shared decision-making, foster greater autonomy, and actively involve patients and families in developing individualised transition plans. The ultimate goal is for young adults to understand their condition and prior treatments, recognise their future healthcare needs, and navigate the healthcare system independently, knowing where and how to access expert support.

Every patient should have an individualised transition plan developed jointly with the patient, family, and healthcare team. The plan should be reviewed regularly and adapted according to changing healthcare needs and life circumstances. Transfer to adult services should occur only after an appropriate preparation process and identification of the healthcare professionals responsible for ongoing adult care.

A dedicated transition coordinator should be available to support patients and families throughout the process. This individual serves as the primary point of contact, facilitates communication between services, coordinates care, and helps prevent loss to follow-up. The transition coordinator may be a healthcare professional or allied professional integrated within the care pathway and should be easily accessible to patients and families.

At the time of transfer, every patient should receive a comprehensive transition summary detailing the diagnosis, clinical history, surgical and treatment history, current clinical status, ongoing surveillance requirements, and anticipated future healthcare needs. The summary should clearly identify the healthcare professionals responsible for ongoing adult care and be accompanied by a patient-friendly version. Whenever possible, direct communication between pediatric and adult healthcare teams should take place to ensure continuity and a shared understanding of the patient's long-term care plan.

3. MODELS OF CARE

Two principal models of transition care are currently used in Europe.

1. In the transfer model, patients move from pediatric to adult healthcare services, often involving changes in healthcare professionals and institutions. This model requires structured communication, formal handover procedures, and robust coordination to ensure continuity of care.
2. In the integrated lifelong care model, patients remain within the same institution and care network throughout life while the focus of care gradually evolves from pediatric to adult healthcare needs. This model offers a high degree of continuity and should be encouraged whenever feasible, particularly for individuals with rare and complex conditions.

Regardless of the model used, healthcare providers who cannot offer a structured transition program should establish clear referral pathways to centres with dedicated transition services. Care should be coordinated across pediatric and adult services to ensure continuity and avoid fragmentation.

4. SYSTEM-LEVEL REQUIREMENTS

Healthcare systems should support recognised transition programs and ensure that patients with rare uro-recto-genital conditions have access to coordinated lifelong care. Centres providing specialised transition care should be identifiable, accessible, and supported by appropriate workforce planning to ensure continuity of expertise over time.

National and international collaboration should be encouraged to promote harmonised standards of care, facilitate the exchange of expertise, and improve long-term outcomes for individuals with rare uro-recto-genital conditions.

5. RECOMMENDATIONS

ERN eUROGEN recommends that:

1. All individuals with rare uro-recto-genital conditions should have access to a structured transition program as part of lifelong follow-up.
2. Transition should be recognised as a continuous process rather than a single transfer event.
3. Transition planning should begin early and be reviewed regularly.
4. Every patient should have a named transition coordinator and an individualised transition plan.
5. Patients and families should be active partners in transition planning and decision-making.
6. A comprehensive transition summary and future care plan should accompany every transfer to adult services.
7. Healthcare providers unable to offer structured transition care should establish referral pathways to centres with dedicated transition programs.

6. CONCLUSION

Transition from paediatric to adult care is an essential component of lifelong management for individuals with rare uro-recto-genital conditions.

Successful transition preserves continuity of care, fosters patient autonomy, and optimises long-term health and quality-of-life outcomes. Delivering high-quality transition care requires early preparation, dedicated coordination, effective communication, multidisciplinary collaboration, and sustained access to specialist expertise across the lifespan. As healthcare providers and services change over time, patients and their families remain the only constant throughout the care journey. Placing them at the centre of transition planning, shared decision-making, and ongoing support is therefore fundamental to achieving safe, effective, and person-centred lifelong care.

Patient advocacy groups also play a pivotal role in this process by providing education, peer support, empowerment, and continuity beyond healthcare settings, helping individuals and families navigate transition, advocate for their needs, and remain connected to specialist resources and communities. Strengthening partnerships between healthcare professionals, patients, families, and advocacy organisations is essential for achieving equitable, coordinated, and lifelong care.

7. DISEASE/CONDITION-SPECIFIC MULTIDISCIPLINARY TEAM REQUIREMENTS

7.1. Differences of Sex Development (DSD) (ERN eUROGEN Expertise Area EA 1.1)

7.1.1 Conditions include

- 46, XY DSD/mosaic
- 46, XX DSD due to prenatal androgen excess/mosaic
- Turner syndrome
- Classic congenital adrenal hyperplasia/adrenogenital syndrome

7.1.2 Core specialists (definitely required)

- Endocrinologist
- Psychologist/mental health specialist
- Urologist

- Gynecologist
- Clinical geneticist

7.1.3 Additional specialists (recommended)

- Sexual medicine specialist/sex therapist
- Andrologist
- Fertility or infertility specialist

7.1.4 Optional involvement

- Primary care physician/family doctor

7.2. Bladder Exstrophy–Epispadias Complex (ERN eUROGEN Expertise Area EA 1.2)

7.2.1 Conditions include

- Bladder exstrophy
- Epispadias–exstrophy complex
- Cloacal exstrophy

7.2.2 Core specialists (definitely required)

- Urologist
- Gynecologist
- Nephrologist
- Psychologist
- Gastroenterologist
- Sexual medicine specialist

7.2.3 Additional specialists (recommended)

- Endocrinologist
- Andrologist
- Fertility specialist
- Internal medicine specialist
- General surgeon
- Plastic/reconstructive surgeon

7.2.4 Optional involvement

- Primary care physician/family doctor

7.3. Rare Urological Stone and Kidney Diseases (ERN eUROGEN Expertise Area EA 1.3)

7.3.1 Conditions include

- Cystinuria
- Dent disease
- Cystinosis
- Bartter syndrome
- Homocystinuria with cystine stones
- Familial cystic kidney diseases

7.3.2 Core specialists (definitely required)

- Nephrologist

- Urologist
- Internal medicine specialist

7.3.3 Additional specialists (recommended)

- Endocrinologist
- Psychologist
- Clinical geneticist
- Dietitian / metabolic specialist

7.3.4 Optional involvement

- Primary care physician/family doctor

7.4. Neurogenic Bladder Disorders (ERN eUROGEN Expertise Area EA 1.4)

7.4.1 Examples

- Spina bifida
- Neurogenic bladder dysfunction
- Caudal regression syndrome

7.4.2 Core specialists (definitely required)

- Neuro-urologist / urologist
- Nephrologist
- Urotherapist / stoma therapist
- Gynaecologist (in female patients)
- Psychologist
- Neurosurgeon
- Orthopedic surgeon

7.4.3 Additional specialists (recommended):

- Endocrinologis
- Sexual medicine specialist
- Andrologist
- Fertility specialist
- Internal medicine specialist

7.4.4 Optional involvement

- Primary care physician/family doctor

7.5. Congenital Müllerian and Vaginal Anomalies (ERN eUROGEN Expertise Area EA 1.4)

7.5.1 Examples

- Agenesis or aplasia of the uterus
- Vaginal atresia
- Partial vaginal agenesis
- Septate vagina

7.5.2 Core specialists (definitely required)

- Gynecologist
- Psychologist

7.5.3 Additional specialists (recommended)

- Endocrinologist
- Sexual medicine specialist
- Fertility specialist

7.5.4 Optional involvement

- Primary care physician/family doctor

7.6. Primary Megaureter (ERN eUROGEN Expertise Area EA 1.4)

7.6.1 Core specialists (definitely required)

- Urologist
- Nephrologist

7.6.2 Additional specialists (recommended)

- Internal medicine specialist

7.6.3 Optional involvement

- Primary care physician/family doctor

7.7. Urachal Anomalies (ERN eUROGEN Expertise Area EA 1.4)

In most patients, surgical correction in childhood is curative, and long-term specialised follow-up is usually not required. Therefore, structured transition programs are generally not necessary unless complications or persistent symptoms occur.

7.8. Rare Penile and Genital Malformations (ERN eUROGEN Expertise Area EA 1.4)

7.8.1 Examples

- Urethral duplication
- Congenital scrotal agenesis
- Diphallia
- Penile agenesis
- Penoscrotal transposition

7.8.2 Core specialists (definitely required)

- Urologist

7.8.3 Additional specialists (recommended)

- Plastic/reconstructive surgeon
- Endocrinologist
- Sexual medicine specialist
- Psychologist

7.8.4 Optional involvement

- Primary care physician/family doctor

7.9. Congenital Bilateral Absence of Vas Deferens (CBAVD) (ERN eUROGEN Expertise Area EA 1.4)

7.9.1 Core specialists (definitely required)

- Andrologist/urologist
- Fertility specialist
- Pulmonologist or internal medicine specialist (in patients with cystic fibrosis)

7.9.2 Additional specialists (recommended)

- Endocrinologist
- Psychologist

7.9.3 Optional involvement

- Primary care physician/family doctor

7.10. Isolated Micropenis (ERN eUROGEN Expertise Area EA 1.4)

7.10.1 Core specialists (definitely required)

- Urologist
- Endocrinologist
- Psychologist

7.10.2 Additional specialists (recommended)

- Plastic surgeon

7.10.3 Optional involvement

- Primary care physician/family doctor

7.11. Posterior Urethral Valves and Congenital Lower Urinary Tract Obstruction (ERN eUROGEN Expertise Area EA 1.5)

7.11.1 Conditions include

- Posterior urethral valves
- Congenital lower urinary tract obstruction (cLUTO)

It should be noted that up to 60% of patients with fetal lower urinary tract obstruction are ultimately diagnosed with posterior urethral valves, and differentiation during fetal life or early infancy may be difficult, particularly in patients treated with vesico-amniotic shunting.

7.11.2 Core specialists (definitely required)

- Urologist
- Nephrologist
- Urotherapist / stoma therapist
- Psychologist

7.11.3 Additional specialists (recommended)

- Endocrinologist
- Andrologist
- Internal medicine specialist

7.11.4 Optional involvement

- Primary care physician/family doctor

7.12. Posterior Hypospadias (ERN eUROGEN Expertise Area EA 1.6)

7.12.1 Core specialists (definitely required)

- Urologist

7.12.2 Additional specialists (recommended)

- Endocrinologist
- Sexual medicine specialist
- Andrologist
- Psychologist

7.12.3 Optional involvement

- Internal medicine specialist
- Primary care physician/family doctor

7.13. Anorectal Malformations (ERN eUROGEN Expertise Area EA 1.4)

Anorectal malformations present with a wide spectrum of types and severity and are frequently associated with other congenital anomalies. In approximately 50% of cases, urological abnormalities are also present. For this reason, management requires a multidisciplinary team.

7.13.1 Core specialists (definitely required)

According to the ERN eUROGEN Clinical Practice Guideline on Anorectal Malformations (3), this is the multidisciplinary team required during the pediatric period, which may continue for follow-up in adulthood with adult clinicians:

- Paediatric surgeon
- Paediatrician
- Nurse practitioner/case manager pediatric surgery
- (Paediatric) Anesthesiologist
- (Paediatric) Cardiologist
- Paediatric Urologist
- (Paediatric) Neurologist
- (Paediatric) Neurosurgeon
- Nurse practitioner in urinary incontinence, materials, ICC and stoma care
- Dietitian
- General practitioner
- Physiotherapist
- Urotherapist
- (Paediatric) Gastroenterologist
- (Paediatric) nephrologist
- Clinical geneticist
- Social worker
- Neonatologist
- (Paediatric/youth) Psychologist
- (Paediatric) Radiologist
- Colorectal Surgeon
- (Paediatric) Orthopedic Surgeon
- (Paediatric) Cardiac surgeon
- (Paediatric) Gynaecologist

- High-risk pregnancy specialist
- Urologist/andrologist
- Sexologist or other provider who can help with sexual issues
- Plastic surgeon
- School counsellor

Please note that a colorectal surgeon is not necessarily the primary clinician needed when a patient is transitioned to adult services (1, 2)

8. REFERENCES

1. de Beaufort CMC, Aminoff D, de Blaauw I, Crétolle C, Dingemann J, Durkin N, et al. Transitional Care for Patients with Congenital Colorectal Diseases: An EUPSA Network Office, ERNICA, and eUROGEN Joint Venture. J Pediatr Surg. 2023;58(12):2319–26.

2. Violani C, Grano C, Fernandes M, Prato AP, Feitz WFJ, Wijnen R, et al. The Transition of Care for Patients with Anorectal Malformations and Hirschsprung Disease: A European Survey. Eur J Pediatr Surg. 2023;33(3):191–7.

3. Jan-Hendrik Gosemann WI, Konstantinos Mantzios, Ivo de Blaauw,, Cornelius E.J. Sloots DA, Nicole Schwarzer, Marc Miserez, Eberhard Schmiedeke,, Pernilla Stenström BDI, Celia Cretolle, Paola Midrio, Salvatore Cascio,, IACS group ML. ERN eUROGEN Clinical Practice Guideline on Anorectal Malformations (ARM) Adapted from the Dutch Quality Standard for Anorectal Malformations 2025. https://eurogen-ern.eu/wp-content/uploads/2025/08/ERN-eUROGEN-CPG-on-ARM-Combined-Document-FINAL-2025-08-19.pdf

Funded by the European Union

ERN eUROGEN is a European Reference Network (ERN) approved by the ERN Board of Member States (BoMS). For more information about the ERNs and the EU health strategy, please click here. The ERNs are funded by the European Union.