RAISING AWARENESS: THE TEDDY KIDS' WAY

Understanding Rare Diseases: A Young Person's Guide

Dr Suja Somanadhan | Associate Professor and Associate Dean
University College Dublin, Ireland

19-09-2026, Bari, Italy

OVERVIEW

What We'll Explore Together

1

What is a rare disease?

A quick, clear explanation of what “rare” really means.

2

What everyday life can look like

Hospital visits, school, friendships — and how young people cope.

3

How families are affected

The emotional and financial ripple effects on the people around you.

4

How you can help shape the future

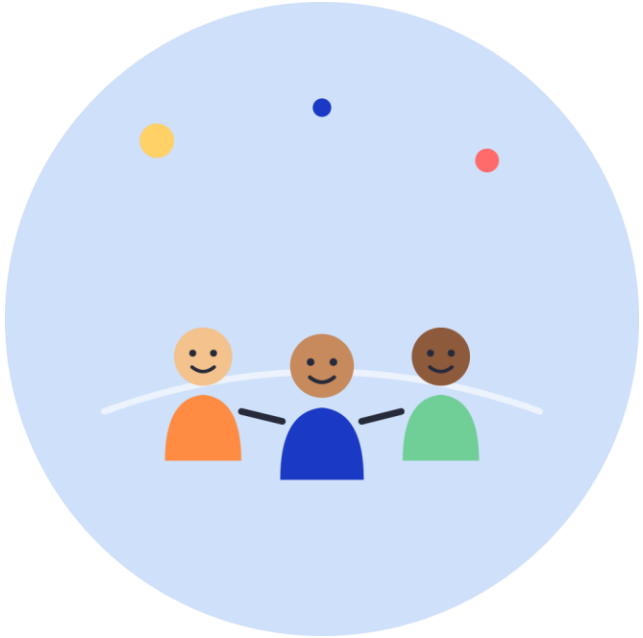
Why researchers want to hear directly from young people like you.

THE BASICS

What Is a “Rare” Disease?

8,000+
known rare diseases

1 in 2,000
EU definition of “rare”



Most rare diseases are genetic, and roughly half of everyone living with one is a child. Because each condition affects so few people, doctors and researchers don't always have much information to go on — which is exactly where research (and young people like you) can help.

Heterogeneity of Rare Diseases

Why no two transitions look alike

8000+

distinct conditions fall under Rare Renal Disorders (RRDs) in Europe, spanning nearly all paediatric kidney diagnoses

"Walking on a knife-edge" — how one participant described life with an RRD

1

Clinical variability

Age of onset, severity, and disease trajectory differ widely even within the same diagnosis — many AYAs describe their condition as unpredictable and ever-changing.

2

Multisystem complexity

RRDs frequently involve more than the kidneys — physical, cognitive, and psychosocial systems are affected together, demanding input from multiple specialties at once.

3

Diverse treatment pathways

Dialysis, transplantation, and alternative therapies each carry distinct burdens and timelines, so care and transition planning cannot follow a single template.

4

Rarity → thin adult expertise

Because each condition is individually rare, adult specialists with relevant experience are few and concentrated in a handful of tertiary centres.

Source: Kinch M, Kroll T, Buckle N, Somanadhan S. Interventions to support young adults and families with the healthcare transition between paediatric and adult nephrology health services: A systematic scoping review. J Pediatr Nurs. 2024;78:e346–e363.

Why Are Rare Diseases Different?

The Diagnostic Odyssey

Because each condition is so rare, it can take years and many doctors' visits before you get an accurate diagnosis.

Every Case Looks Different

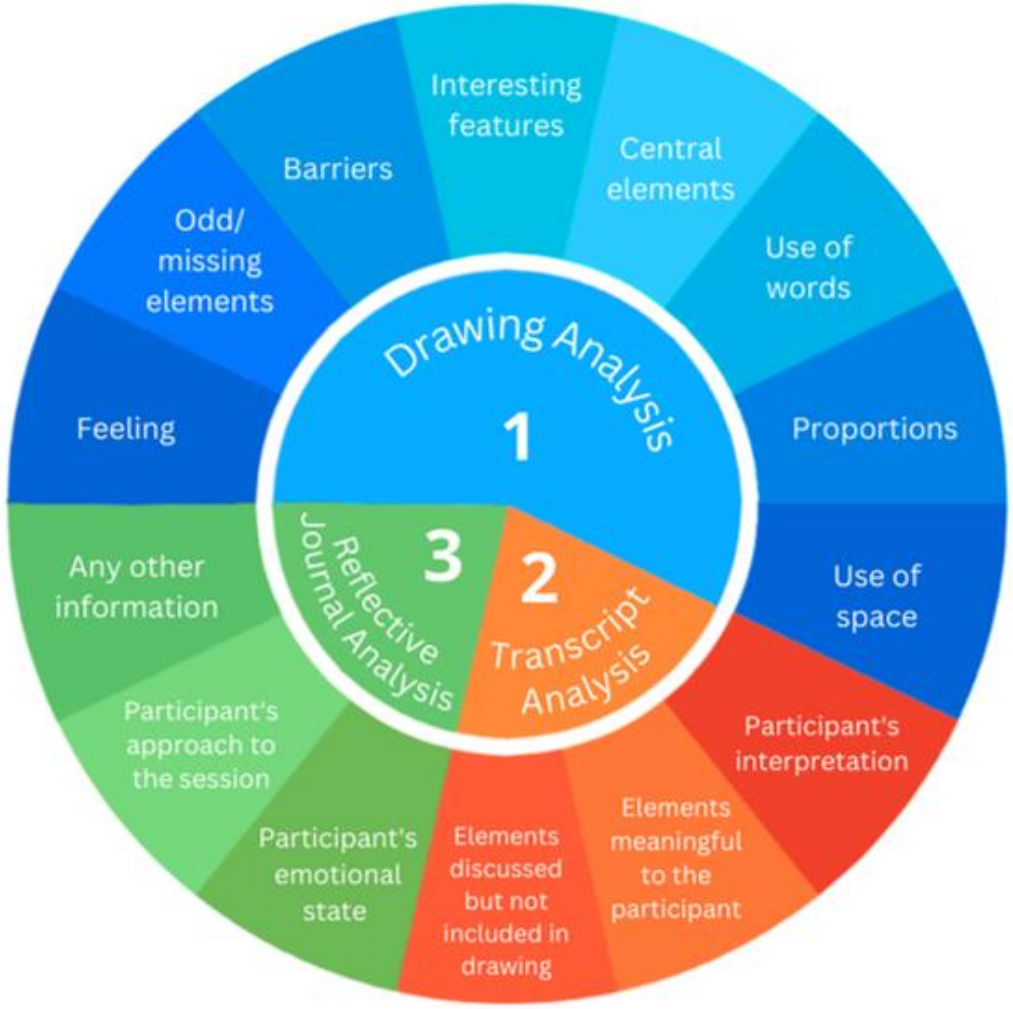
Even people who share the exact same diagnosis can have very different symptoms, needs, and day-to-day experiences.



Children's Art to Understand their Experiences of Living with Rare Diseases

- This study was part of the SAMPI Project (Children's Health Ireland at Temple Street Children's University Hospital, RPAC17-05).
- Seven children with an RD aged between 7 and 13 years participated in the Art sessions, facilitated by an Art Therapist.
- Participants were asked to draw their experience of living with their RD, then explain and title their drawing.
- As drawing is an 'open' visual medium [1], a framework influenced by Furth [2] was developed to thematically analyse the drawings.

Framework for Analyzing Children's Drawings for Health Research



Source: Buckle, O'Neill, Sweeney, Sinnott, Karpaviciute, Kroll & Somanadhan (2024). Therapeutic Advances in Rare Disease, 5.

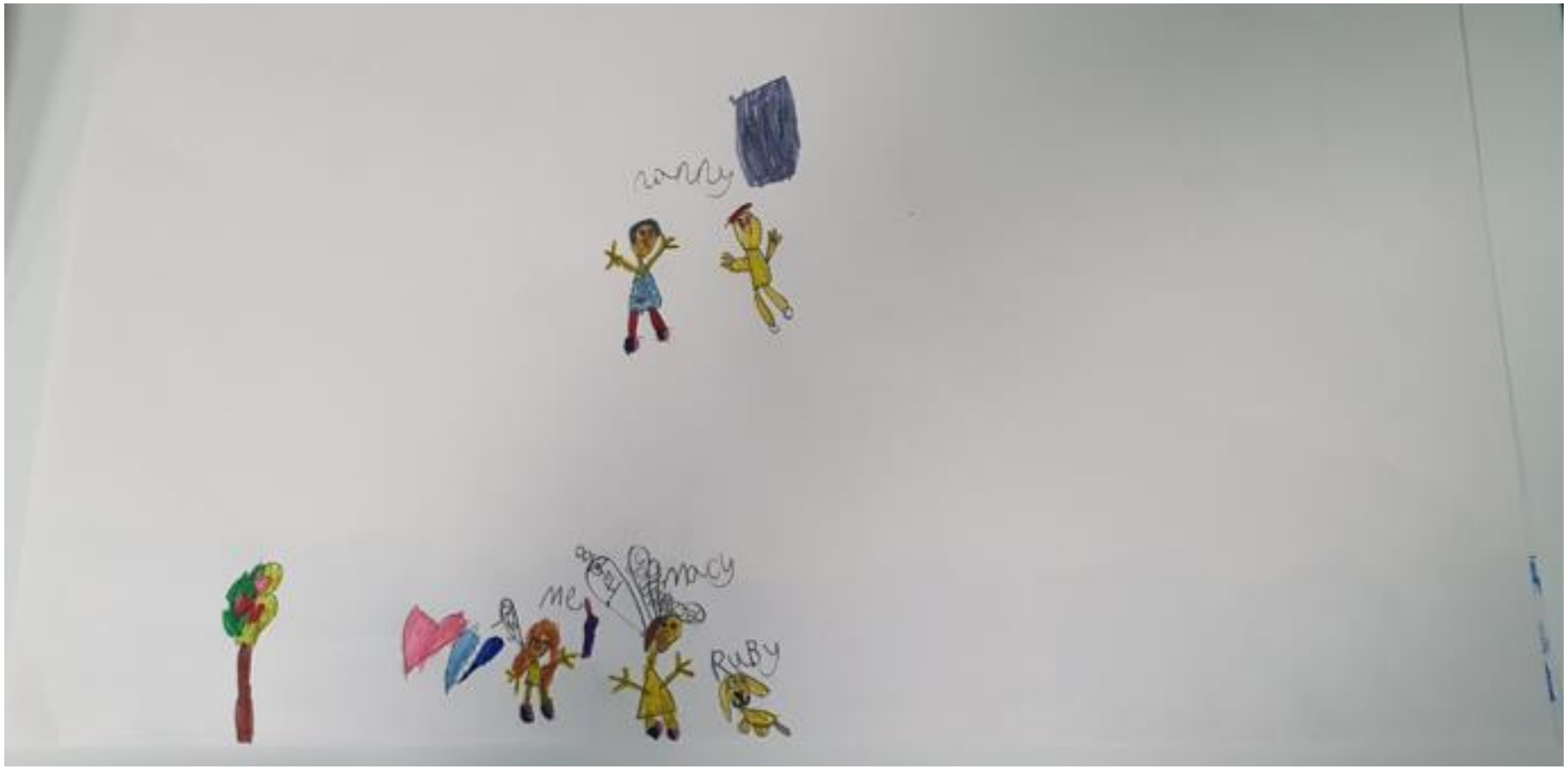
The themes that emerged from the drawings were

- ‘Normal vs Different’,
- ‘Family: Present vs Hidden ’ and ‘Friends’.

- These themes highlight the contradictory experience of living with a RD and the role of family and friends in influencing the participants’ experiences.
- This study concludes that art can help health researchers understand children’s lived experiences of RDs.

Source: Buckle, O'Neill, Sweeney, Sinnott, Karpaviciute, Kroll & Somanadhan (2024). Therapeutic Advances in Rare Disease, 5.

Participant 2’s drawing, entitled ‘The Home That I Love’.



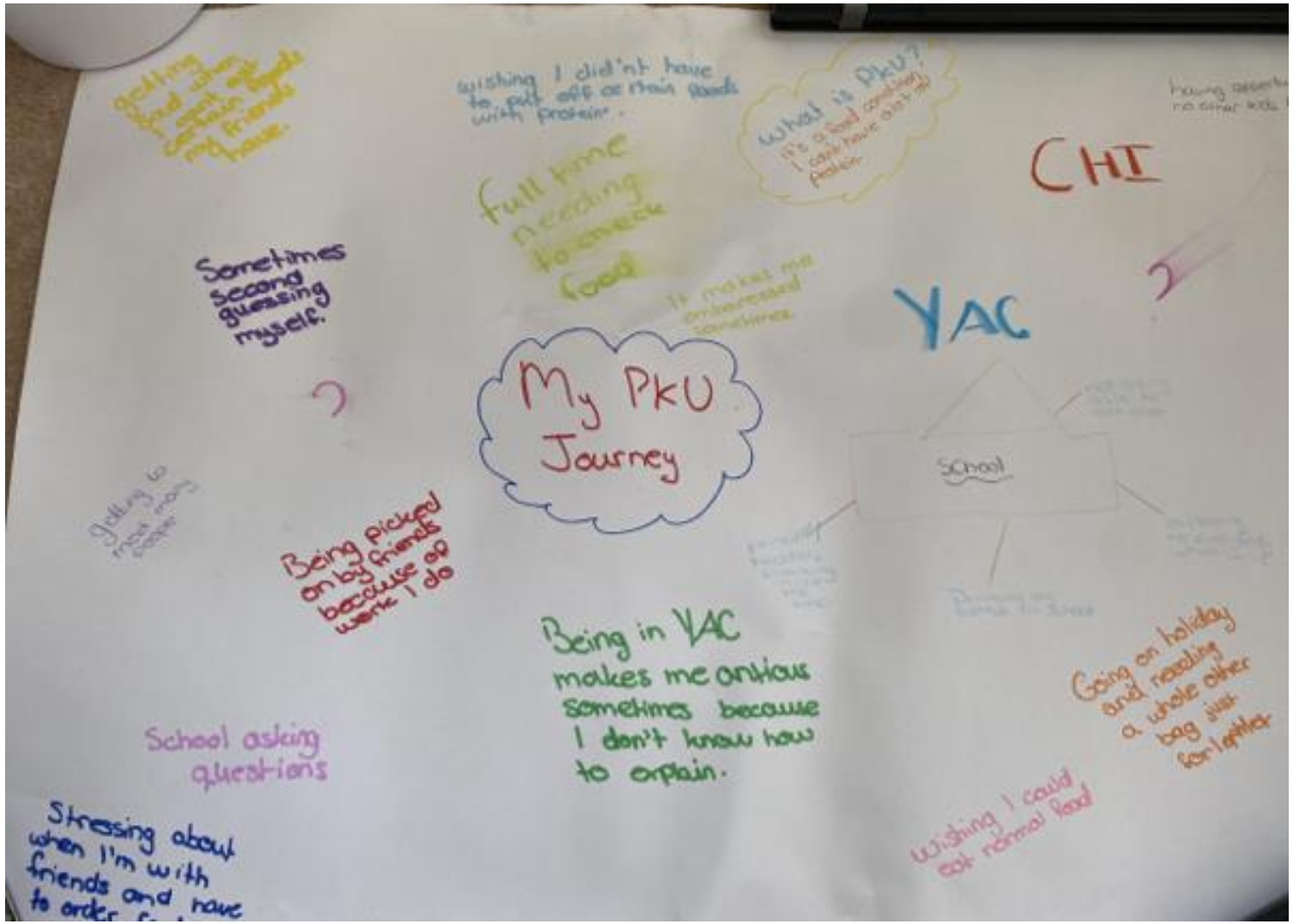
Source: Buckle, O'Neill, Sweeney, Sinnott, Karpaviciute, Kroll & Somanadhan (2024). Therapeutic Advances in Rare Disease, 5.

Participant 4's drawing, entitled '[Participant's Name]'s Art'



Source: Buckle, O'Neill, Sweeney, Sinnott, Karpaviciute, Kroll & Somanadhan (2024). Therapeutic Advances in Rare Disease, 5.

Participant 7's drawing, entitled 'My PKU Journey'



Source: Buckle, O'Neill, Sweeney, Sinnott, Karpaviciute, Kroll & Somanadhan (2024). Therapeutic Advances in Rare Disease, 5.

A Young Person's Everyday Reality

H

Hospital Visits & Treatments

Regular appointments and treatments can mean time away from school and friends.

S

Staying Connected at School

Missing school or feeling “different” from classmates is common — but friendships and support help a lot.

♥

Your Feelings Are Valid

Frustration, anxiety, or pride in how you manage it all — there's no single “right” way to feel.



BEYOND THE PATIENT

The Ripple Effect on Families

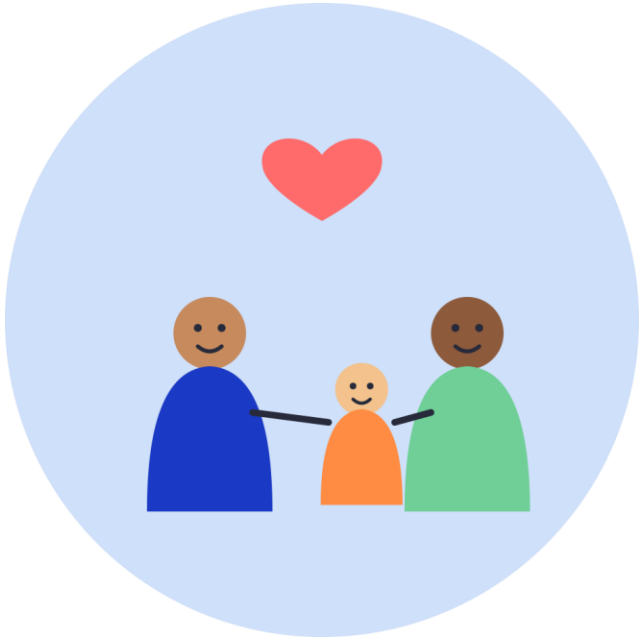
A rare disease diagnosis doesn't just affect one person — it reshapes life for the whole family.

Financial Strain

Frequent travel to specialists, therapies, and equipment can add up to significant, ongoing costs for families.

Time & Caregiving

Parents often reduce work hours or leave jobs altogether to provide care and coordinate appointments.



Source: Buckle, Doyle, Kodate, Kinch & Somanadhan (2024). Healthcare (Basel), 12(24), 2578.

TEDDY NETWORK
Coordinator and promoter of the Biotech Week in Italy
FEDERCHIMICA ASSOBIOTEC
Associazione nazionale per lo sviluppo delle biotecnologie
European Biotech Week
Innovation is in our Genes
PART OF GLOBAL BIOTECH WEEK

MOVING FORWARD

Growing Up: The Transition Journey

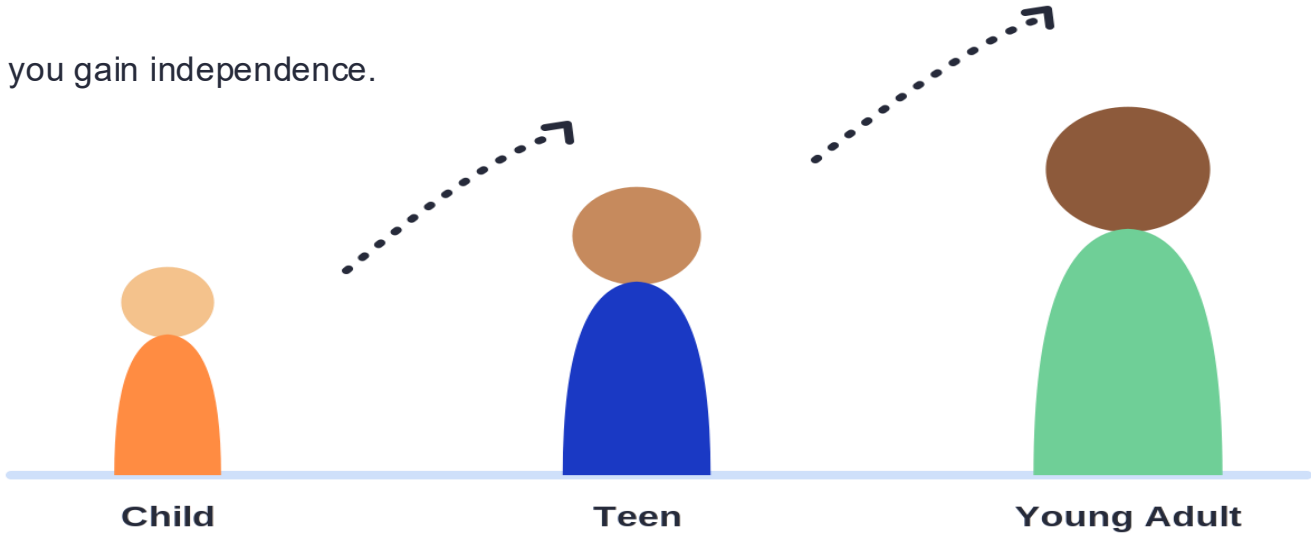
Moving from children's to adult healthcare services is a major life change for adolescents and young adults with rare diseases.

New Systems, New Independence

You'll navigate new clinical teams and take on more ownership of your own care.

Parents Are on the Journey Too

Parents often need support adjusting to a less central role as you gain independence.



Sources: Kinch, Kroll & Somanadhan (2026). PLOS ONE, 21(6), e0348445. | Kinch, Somanadhan, Walsh & Larkin (2026). Palgrave Macmillan.

What Heterogeneity Means for Transition Care

Complexity multiplies across systems, timelines, and services

1 No standard pathway

Wide variation in HCT practice across centres and countries is a well-documented consequence of RRD heterogeneity — internationally, transition support remains inconsistent rather than protocolised.

2 Age-based transfer ill-fits readiness

A fixed transfer age doesn't map onto AYAs' actual cognitive, emotional, and self-management maturity, which varies with each condition and its trajectory.

3 Care coordination gaps

Multisystem involvement means AYAs see several specialists across services; without a designated coordinator, follow-up, records, and communication routinely fall through the cracks.

4 Individualised planning required

Because no two RRD journeys are alike, effective transition demands person-centred, flexibly-timed plans — co-designed with AYAs and families — rather than one-size-fits-all protocols.

Source: Kinch M, Kroll T, Buckle N, Somanadhan S. Interventions to support young adults and families with the healthcare transition between paediatric and adult nephrology health services: A systematic scoping review. J Pediatr Nurs. 2024;78:e346–e363.

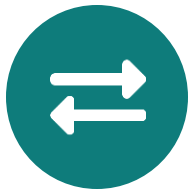
A Fragmented Journey for Young Patients

Children and young people with rare diseases face compounding coordination gaps at every stage



The Diagnostic Odyssey

Rare conditions in children often take years to diagnose. Every additional month without a diagnosis is a month without access to coordinated, multidisciplinary care.



The Transition Cliff

Childhood-onset, lifelong conditions (e.g. Neurofibromatosis Type 1, 22q11.2 deletion syndrome, Angelman syndrome) show significant gaps in adult services — leaving GPs and families to coordinate care alone.



The Communication Gap

Across Europe, 65% of rare disease families report visiting multiple health, social, and local services in a short period, and 67% say these services communicate poorly with one another.

Sources: Walton et al., Int J Integr Care 2020; Ward et al., Orphanet J Rare Dis 2022; INNOVCare / EURORDIS Rare Barometer survey data.

Coordinated Care Changes the Trajectory

What effective coordination looks like across a child's whole care journey

Family-Centred

Care is organised around the whole family's needs — medical, psychosocial, and practical — not just the diagnosis.

A Named Coordinator

A nurse specialist or case manager acts as a single point of contact, reducing repeated appointments and message fatigue.

Planned Transition

Dedicated transition coordinators

The Child at the Centre

Multidisciplinary teams (genetics, psychology, social work) work as one unit around the child, not in parallel silos.

For children and young people with rare diseases, coordination is not an add-on — it is the care.

YOUR SUPPORT SYSTEM

Who's on Your Team?

Paediatric team are often a constant, trusted presence throughout a young person's rare disease journey.

C **Caring**
Hands-on, compassionate care at every appointment and admission.

A **Advocating**
Speak up for your needs with doctors, nurses, psychologists, schools, and services.

E **Empowering**
Helping you and your family build confidence to manage your own health.



Source: Somanadhan, Roberts, Pelentsov, Canbulat Sahiner, Scott & Foster (2025). Journal of Pediatric Nursing.

TEDDY NETWORK
Coordinator and promoter of the Biotech Week in Italy
FEDERCHIMICA ASSOBIOTEC
Associazione nazionale per lo sviluppo delle biotecnologie
European Biotech Week
Innovation is in our Genes
PART OF GLOBAL BIOTECH WEEK

GET INVOLVED

Why Your Voice Matters in Research

Researchers are increasingly partnering WITH young people and families — not just studying them.

Creative Methods

Art, storytelling, and photography help researchers understand daily life in your own words and images.

Co-Design

Young people help shape research questions, study materials, and how results are shared.



If you're ever asked to take part in research — your voice, your art, your story can help change the future for the next young person diagnosed.

Source: Buckle, O'Neill, Sweeney, Sinnott, Karpaviciute, Kroll & Somanadhan (2024). Therapeutic Advances in Rare Disease, 5.

TEDDY NETWORK

Coordinator and promoter of the Biotech Week in Italy
FEDERCHIMICA ASSOBIOTEC
Associazione nazionale per lo sviluppo delle biotecnologie

European Biotech Week
Innovation is in our Genes

PART OF GLOBAL BIOTECH WEEK

Key Takeaways

- Rare alone, but not alone together — millions worldwide share this journey.
- Every experience is different — and every experience is valid.
- Your support system matters as much as your medical team.
- You have a voice — in your care, and in research.



SOURCES

References

Buckle, N., Doyle, O., Kodate, N., Kinch, M. and Somanadhan, S. (2024). Caregiver-reported economic impacts of pediatric rare diseases: A scoping review. Healthcare (Basel), 12(24), p.2578. https://doi.org/10.3390/healthcare12242578

Buckle, N., O'Neill, A., Sweeney, A., Sinnott, S., Karpaviciute, S., Kroll, T. and Somanadhan, S. (2024). The use of art as a creative research method to understand psychosocial care needs for children with rare diseases. Therapeutic Advances in Rare Disease, 5. https://doi.org/10.1177/26330040241265449

Kinch, M., Kroll, T. and Somanadhan, S. (2026). The river meeting the sea: A qualitative exploration of the healthcare transition experiences of adolescents and young adults living with rare renal disorders and their parents. PLOS ONE, 21(6), e0348445. https://doi.org/10.1371/journal.pone.0348445

Kinch, M., Somanadhan, S., Walsh, C., Larkin, P. (2026). Life Transitions of the Families of Children Living with Rare Diseases. In: Jindal-Snape, D. (eds) The Palgrave International Handbook of Multiple and Multi-Dimensional Educational and Life Transitions. Palgrave Macmillan, Cham. https://doi.org/10.1007/978-3-032-11145-6_16

Somanadhan, S., Roberts, K., Pelentsov, L., Canbulat Sahiner, N., Scott, S. D., & Foster, M. (2025). The pivotal role of pediatric nurses: Caring, empowering, and advocating for children and families with rare diseases. Journal of Pediatric Nursing. https://doi.org/10.1016/j.pedn.2025.10.015

Ward AJ, Murphy D, Marron R, McGrath V, Bolz-Johnson M, Cullen W, Daly A, Hardiman O, Lawlor A, Lynch SA, MacLachlan M, McBrien J, Ni Bhriain S, O'Byrne JJ, O'Connell SM, Turner J, Treacy EP. Designing rare disease care pathways in the Republic of Ireland: a co-operative model. Orphanet J Rare Dis. 2022 Apr 11;17(1):162. doi: 10.1186/s13023-022-02309-6. PMID: 35410222; PMCID: PMC8996209.

Walton H, Hudson E, Simpson A, Ramsay AIG, Kai J, Morris S, Sutcliffe AG, Fulop NJ. Defining Coordinated Care for People with Rare Conditions: A Scoping Review. Int J Integr Care. 2020 Jun 25;20(2):14. doi: 10.5334/ijic.5464. PMID: 32607101; PMCID: PMC7319081.