

# *Living with Uncertainty & Impact of Trauma*

2<sup>nd</sup> Webinar, 06.09.2023



# Welcome!

We will be starting shortly...



**Mute your microphones** when not speaking



**Turn on your camera**, if possible – **it's nice to see your face!**



Use a **headset or earphones** to **minimize echo and background noise**



**Add your name** –in the 'Participants' list, on your name click and 'Rename'



**Raise your hand** if you wish to speak & use **chat box** for Q&A and comments



# Welcome & Opening Remarks

Webinar aims **to understand the root causes of the uncertain and trauma associated** with the rare disease pathway and **identify coping strategies** to reduce impact on PLWRD and their families.

Also to **increase our understanding of the biopsychosocial continuum** and **addressing intersectional needs**.

Finally, to **take a deep drive into the impact of living with a congenital malformation** on the mental wellbeing of the family.





# Webinar Speakers



**Peter Kéri**

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**President**  
**GAMIAN Europe**



**Kym Winter**

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**Founder & Clinical  
Director**  
**Rare Minds**



**Kirsten Johnson**

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**Fragile X  
International**  
**EURORDIS Board of  
Director**



**Lucy McKay**

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**Chief Executive  
Officer**  
**Medics 4 Rare  
Diseases**



**Dorica Dan**

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**Romanian Prader  
Willi Association**  
**Vice-President of  
EURORDIS**



**Rosanne Smit**

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**Psychologist &  
Researcher**  
**Radboud University  
Medical Centre**

# Agenda

Time	Topic	Speaker
15.00 – 15.03	Welcome & Housekeeping	Concha Mayo, EURORDIS
15.03 – 15.10	Opening Remarks	Kirsten Johnson, Fragile X International & EURORDIS
15.10 – 15.30	Understanding the biopsychosocial continuum and addressing intersectional needs.	Peter Kéri, GAMIAN Europe
15.30 – 16.00	Panel discussion: <ul style="list-style-type: none"> <li>- Understanding of the root cause of the uncertainties and trauma associated with the rare disease pathway.</li> <li>- Identifying coping strategies to deal with uncertainty of the rare disease journey.</li> <li>- Identifying training needs for professionals to reduce the stressors associated with a rare disease.</li> </ul>	Chair: Matt Bolz-Johnson  Panel: <ul style="list-style-type: none"> <li>• Kym Winter, Rare Minds</li> <li>• Lucy McKay, Medics 4 Rare Diseases</li> <li>• Dorica Dan, Romanian Prader Willi Association</li> </ul>
16.00 – 16.10	Questions & Answers	All
16.10 – 16.25	Impact of living with a congenital malformations on mental wellbeing of the family.	Rosanne Smit, Radboud University Medical Centre
16.25 – 16.30	Closing Remarks & Next Steps	Kirsten Johnson, Fragile X International & EURORDIS

# Understanding the biopsychosocial continuum and addressing intersectional needs

Peter Kéri, GAMIAN Europe






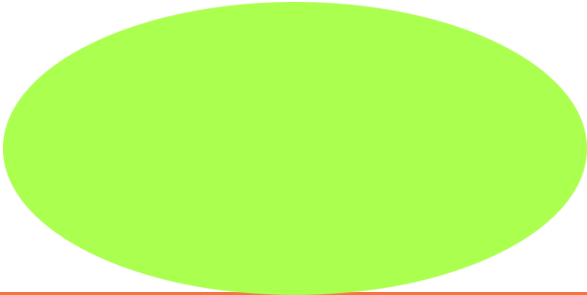
Bio?

Psycho?

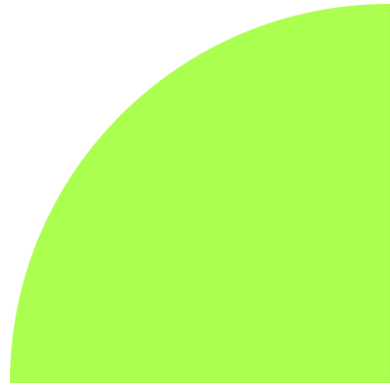
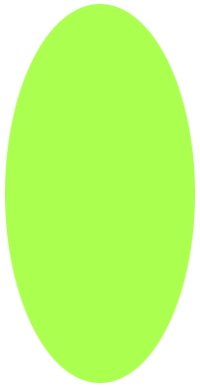
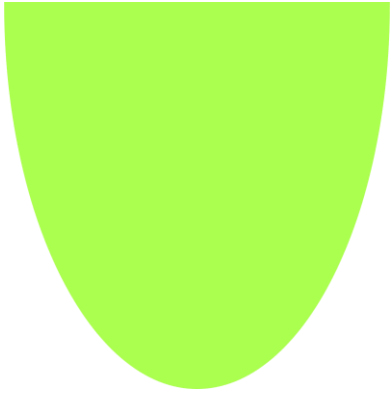
Social?



Péter Kéri  
President  
GAMIAN-Europe



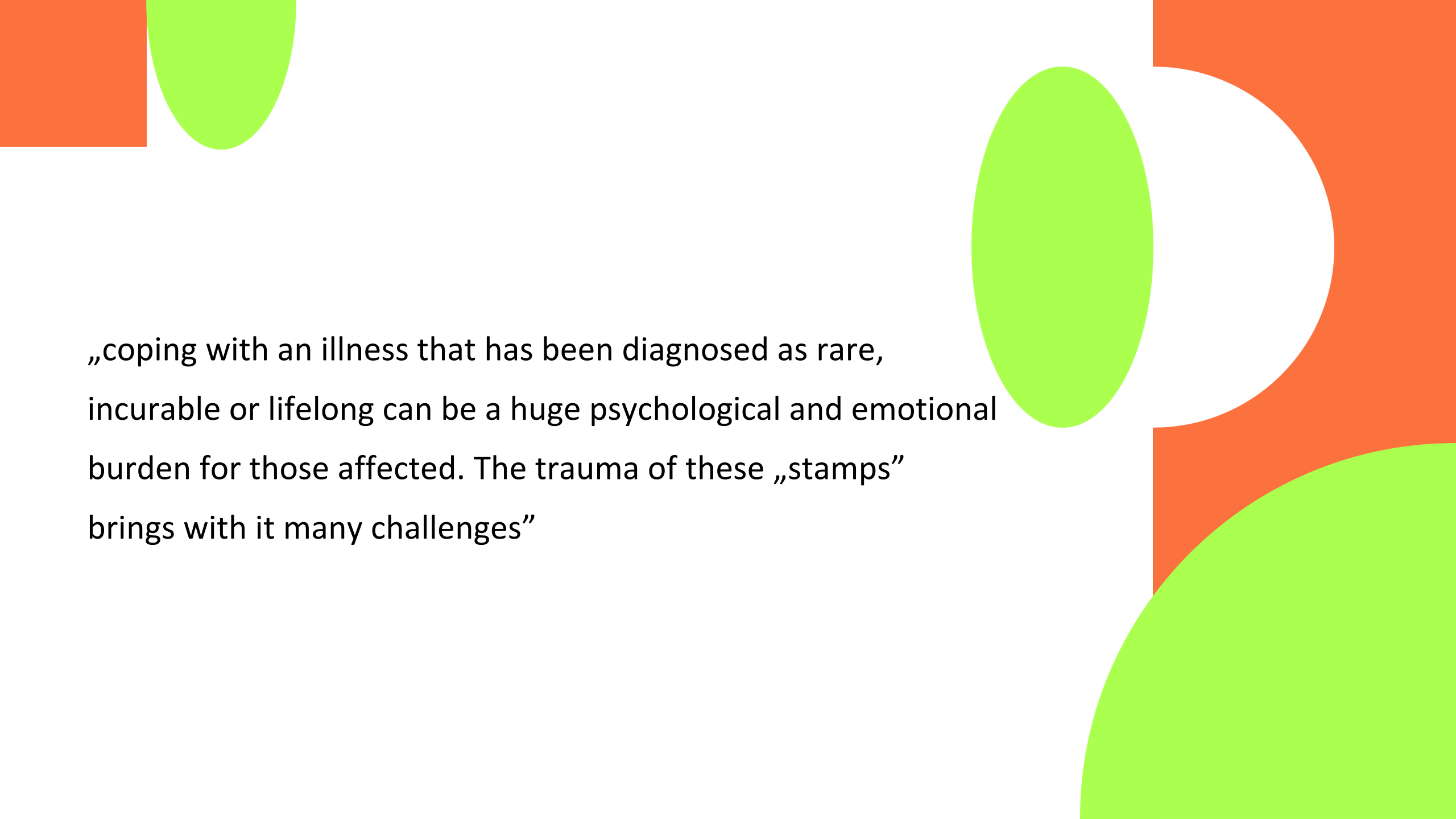




**Rare souls'r'us!**

**Having a special physical, social or mental condition should not separate anyone, it truly connects us.**

**Why is a mental health patient talking? 😊**

The background features abstract geometric shapes in orange and lime green. In the top left, there is a solid orange rectangle and a lime green oval. On the right side, there is a large white shape resembling a stylized letter 'C' or a partial circle, with a lime green oval overlapping its left edge. At the bottom right, there is a large lime green semi-circle. The text is positioned on the left side of the page, centered vertically relative to the white 'C' shape.

„coping with an illness that has been diagnosed as rare, incurable or lifelong can be a huge psychological and emotional burden for those affected. The trauma of these „stamps“ brings with it many challenges“

# What makes it hard?

Hope: after diagnosis, many people lose hope of a full recovery or cure. This can be a huge emotional burden.

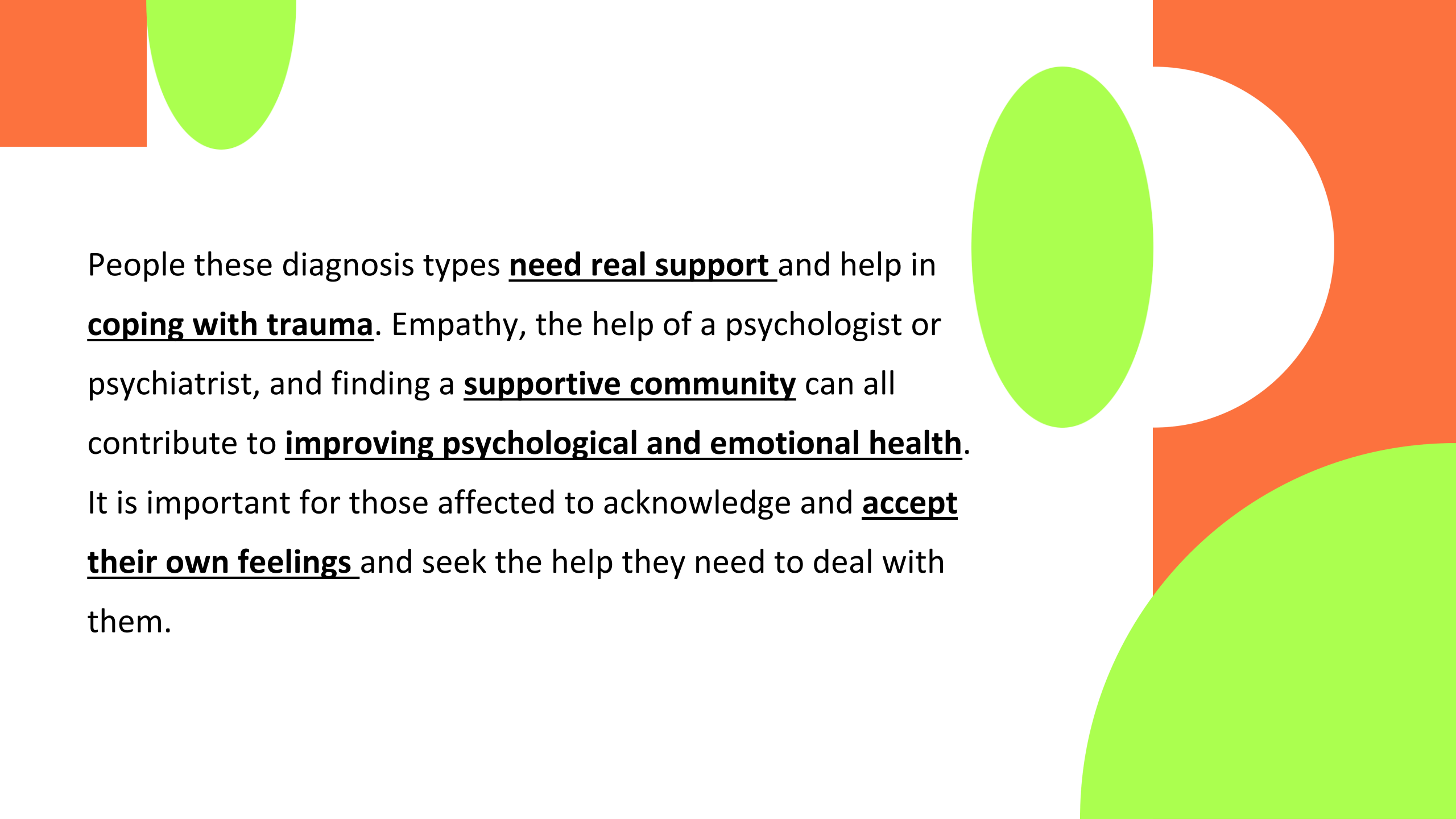
Grief and loss: people often face losses associated with their illness, such as loss of physical abilities, disruption of plans and dreams, or a reduced quality of life.

Mental health: people with rare, lifelong illnesses may be more prone to anxiety and depression because of the stress and uncertainty their condition causes.

Social isolation: people sometimes view people with rare, incurable conditions differently and people may feel isolated or excluded.

Identity and self-image changes: people affected may need to reassess their identity and self-image when they receive such a diagnosis.

Need for support: people with these conditions often need professional help and loving support to manage emotional distress.

The background features abstract geometric shapes in orange and lime green. In the top left, there is a solid orange square and a lime green oval. On the right side, there is a large white circle partially overlapping an orange shape, and a large lime green circle at the bottom right.

People these diagnosis types need real support and help in copng with trauma. Empathy, the help of a psychologist or psychiatrist, and finding a supportive community can all contribute to improving psychological and emotional health. It is important for those affected to acknowledge and accept their own feelings and seek the help they need to deal with them.

# 01.

## Difficulties in diagnosis

Professionals have little information about the disease and symptoms often resemble those of other, more common diseases, making it difficult to make a correct diagnosis

„I have consulted so many professionals. Many have diagnosed me differently. I feel like I've wasted a lot of time before I know approximately what I have”

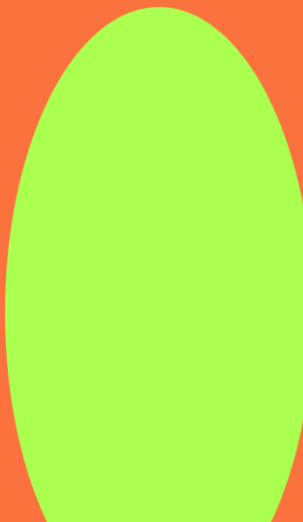
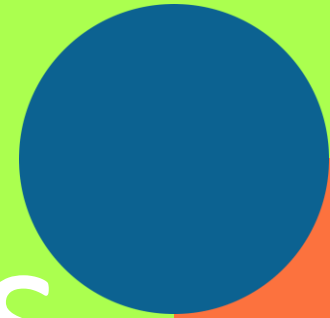


# 02.

# Treatment options

Fewer resources are available for research and development to treat these illnesses. Therefore, people affected may find it more difficult to access effective treatments.

„I never thought a condition I am experiencing could raise as many questions about treatment as mine did. It's disappointing.”



# 03.

# Psychosocial burden

We can often experience psychosocial distress and isolation, as we may receive less support and understanding from those around them.

„I realised that my condition was not only a huge difficulty in my life, but also a very big problem for my loved ones. I did not want to cause such problems”

# 04.

# Financing problems

We usually face high medical costs and drug prices, as treatments and medicines are often specific and more expensive. Our well-being costs more for us, and for all around us.

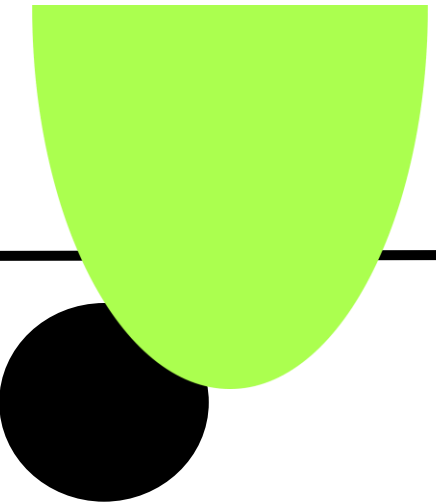
„achieving health is a huge financial challenge for me. I cannot always afford not only the right medical treatments, but also a healthy lifestyle”

# 05.

## Positive outcome

In the less common conditions, it is more difficult to find a certain positive outcome. The helping environment is much smaller and more dispersed.

„I often feel that the maintenance of my condition itself is the positive goal in life, and I forget what kind of life I dreamed for myself, for us”




## Stigma and prejudice

People often react negatively or distantly when they don't understand or are afraid of something. Such reactions may be the result of a lack of knowledge or misconceptions about the illness or mental health condition.




## Coping mechanisms

People many times try to distance themselves because they find it difficult to empathise or help.



## Cultural or social factors



In some cultures or social settings, there is a stronger stigma even blame towards people with „special” diagnoses.



# TOGETHER!

- These conditions as **unknown, alien** diseases in society limits empathy and understanding.
- **Ignorance and distance** often leads to prejudice and stigma.
- **Joining forces and collective efforts**, can help people to better understand and accept both people with mental health problems and those living with rare diseases.
- **Sharing awareness and empathy** can help society to integrate these groups and create a more supportive environment for us.
- **The opportunity to learn from each other and share our own experiences and contribute to social change.**





# Thank you!

WithU!

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mail2peterkeri@gmail.com



# Living with uncertainty and impact of trauma

Panel Discussion



# Panellist



Matt Bolz-Johnson

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**Mental Wellbeing  
Lead**

**Chair**



Kym Winter

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**Rare Minds**

**Panellist -  
Psychologist**



Lucy McKay

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**Medics 4 RD**

**Panellist - Medic**



Dorica Dan

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**Romanian Prader  
Willi Association**

**Panellist - Patient  
Representative**

# Question 1

## Macro uncertainties:

- Timing of onset of symptoms
- Searching for and getting a diagnosis
- Disease progression over time
- Unclear prognosis
- Mortality
- Inherent unpredictability compromising future life-planning
- Searching for an effective treatment and cure
- Responsiveness to treatment and/or symptom control (initially and over time)
- Transition from paediatric and adult services or adult to premature aging challenges
- Willingness and capacity of carers/educationalists/employers to learn and adapt to individual needs (at systemic and individual level)



## Question 2

### **Micro uncertainties:**

- Rollercoaster of monitoring i.e.: tests/observations, results/decision-making, further 'watch and wait' or treatment
- Lack of communication around waiting times for appointment dates/results
- Variable intensity of symptoms day-to-day i.e.: 'good and bad days'
- How to understand certain behaviours, pains or mood i.e.: is this condition related, or something else...?
- Capacity of others to understand a rare disease and its impact i.e.: across statutory services, workplace, and also in personal relationships
- Lack of support to coordinate care and delays in referral
- How to integrate the child into the community and daily life e.g.: finding accessible leisure facilities and places in the community; family holidays.
- Accessing social support and respite care; will my child be safe.

# Questions & Answers

Panel Discussion





# Impact of living with a congenital malformations on mental wellbeing of the family.



Rosanne Smit, Radboud University Medical Centre

RESEARCH LINE

# Common needs in uncommon conditions

Amalia kinderziekenhu  
**Radboudumc**

# Background

🌈 Rare disease = 1 in 2,000 (EU) / 1 in 1,600 (USA)

- ⚙️ Ultra rare = 1 in 50,000
- ⚙️ Undiagnosed

🌈 Challenges in research and clinical care:

- ⚙️ Studies with small samples
- ⚙️ Heterogeneity of phenotypes
- ⚙️ Limited insights in prognosis / unpredictable disease course
- ⚙️ No treatment available in many cases
- ⚙️ Limited insights in **quality of life / psychosocial outcomes**





# Rare Diseases

- 🌈 “There are between 6000 and 8000 rare diseases which means that although each disease is rare, it is not rare to have a rare disease.”
- 🌈 Rare diseases affect 30 million European Union citizens.
- 🌈 80% of rare diseases are of genetic origin, often start in childhood, are chronic and life-threatening.

# RARE 2030 RECOMMENDATIONS

RARE 2030 GOAL

4



*Reduce the level of psychological, social and economic vulnerability of people with a rare disease and their families by one third.*

Rare diseases seriously impact everyday life

**7 in 10** patients & carers

reduced or stopped professional activity due to their or their family member's rare disease.



**8 in 10** patients & carers

have difficulties completing daily tasks (household chores, preparing meals, shopping etc.)



**2/3** of carers

spend more than 2 hours a day on disease-related tasks.



**3 times** more people

living with a rare disease and carers report being unhappy and depressed than the general population\*



\* Rare Barometer Voices sample compared to International Social Survey Programme, 2011

RESEARCH LINE

# Common needs in uncommon conditions



Amalia kinderziekenhuis

**Radboudumc**

# Common challenges in uncommon conditions: Aims of the research line

## **Goal 1: Explore**

Explore the (psychosocial) need for care for children and families with rare disease

## **Goal 2: Network**

Create visibility within rare diseases networks; more representation for psychosocial needs

## **Goal 3: Develop**

Develop tools/interventions together with patient networks, suitable for many rare diseases with 'add-on' modules for disease specific needs



# Rare diseases

## Goal 1: Explore common needs



Identify psychosocial needs in rare diseases

1

Literature overview

Articles (n=56)

Mitochondrial disease (n=13)

Prader Willi Syndrome (n=13)

DSD (n=9)

Urogenital disease (n=9)

ARM (n=9)

Rare kidney disease (n=5)

2

Qualitative data  
(TRANSIT interviews)

Interviews (n=12)

ARM (n=3)

Urogenital disease (n=4)

Epilepsy (n=1)






No diagnosis (n=1)

Turner (n=1)

Metabolic disease (n=1)

FAS (n=1)

# Rare diseases: Literature study overview

-  **Background** Psychosocial determinants and Quality of Life are under researched in (pediatric) rare disease
-  **Aim** To explore common needs for six of the Rare Diseases specialized in the Radboudumc/Amalia Children's Hospital
-  **Method** Literature study, search string "mitochondrial disease", "Prader-Willy" "PWS" "Differences in sexual development" "DSD" "Urogenital disease", "Anorectal malformations" "rare kidney disease" AND "Quality of Life" "psych\*" "psychosoc\*" "burden" "Mental well-being"
-  **Results** 56 articles were selected and needs for each condition/rare disease were reviewed.
-  **Conclusion** Several **overarching concepts** were highlighted

1

Literature overview

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Urogenital disease (n=9)

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Rare kidney disease (n=5)



# Rare diseases: Summary (Literature overview)



## MITO

1. Need for information
2. Coping/communication
3. Practical support health care
4. Concerns about inheritance



## PWS

1. Coping: Hyperphagia
2. Coping: Sleep
3. Well-being/QoL
4. Role of caregiver



## DSD

1. Need for information Sex/fertility
2. Non-binary information
3. Holistic approach
4. Follow-up and transition to adult care
5. Role of caregiver
6. QoL
7. Information disease management



## ARM

1. Sex /fertility
2. Holistic approach
3. transition to adult care
4. QoL
5. Role of caregiver
6. Information about disease and management/coping



## Radboudumc Expertisecentrum Aangeboren Urogenitale Aandoeningen

1. QoL
2. Information in sexual development
3. Body image
4. Transition to adult care



## chronische nierziekte

1. QoL
2. FoP
3. Fear of transplantation
4. Unknown future
5. Desire for new treatment



# Rare diseases: Summary (Literature overview)



## MITO

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2. Coping/communication

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## PWS

1. Coping: Hyperphagia

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## DSD

1. Need for information Sex/fertility

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3. Holistic approach

4. Follow up and transition to adult care

5. Role of caregiver

6. QoL

7. Information disease management



## ARM

1. Sex /fertility

2. Holistic approach

3. transition to adult care

4. QoL

5. Role of caregiver

6. Information about disease and management/coping



## Radboudumc Expertisecentrum Aangeboren Urogenitale Aandoeningen

1. QoL

2. Information in sexual development

3. Body image

4. Transition to adult care



## chronische nierziekte

1. QoL

2. FoP

3. Fear of transplantation

4. Unknown future

5. Desire for new treatment

# Rare diseases: Qualitative research

Smits et al. *Orphanet Journal of Rare Diseases* (2022) 17:153  
<https://doi.org/10.1186/s13023-022-02305-w>

Orphanet Journal of  
Rare Diseases

RESEARCH

Open Access



## Common needs in uncommon conditions: a qualitative study to explore the need for care in pediatric patients with rare diseases

Rosanne M. Smits<sup>1\*</sup>, Eline Vissers<sup>1</sup>, Rosan te Pas<sup>1</sup>, Noor Roebbers<sup>1</sup>, Wout F. J. Feitz<sup>2</sup>, Iris A. L. M. van Rooij<sup>3</sup>, Ivo de Blaauw<sup>4</sup> and Chris M. Verhaak<sup>1</sup>

### Abstract

**Background:** Challenges faced by children diagnosed with a rare disease or complex condition and their family members are often characterized by disease-specific complexities, such as a prolonged diagnostic process, an uncertain prognosis, and the absence of curative treatment. The psychological burden of living with a rare disease or complex condition is often understudied and may present overarching concepts that shape the general experience of having been diagnosed with a rare condition. The present study examines common needs from a comprehensive perspective combining relevant aspects from the rare disease literature in a theoretical perspective from pediatric psychology, such as a family-centred, developmental and interdisciplinary approach. An exploratory study was designed among parents from children with a rare disease or complex condition in an Integrated University Children's Hospital in the Netherlands. Semi-structured interviews were conducted with open-ended questions based around the experience of having a child diagnosed with a rare condition, such as the psychosocial impact on the child and its development, the impact on the family, and how provided care was experienced.

**Results:** Twelve interviews were analysed with a thematic content analysis to identify common needs. Eight themes followed from the analysis and uncovered the need for (1) family-focused care, (2) coping with uncertainty, (3) empathic communication, (4) practical support, (5) information, (6) psychological support, (7) interdisciplinary care, and (8) social support.

**Conclusions:** The results from our study provide directions for research and health care to support young patients with a rare disease or complex condition and their families. Moreover, our results demonstrated that there are overarching concepts across different rare diseases that may be optimally supported with interdisciplinary care.

**Keywords:** Rare disease, Complex condition, Psychological needs, Caregiver burden, Qualitative research, Quality of

2

Qualitative data  
(TRANSIT interviews)

Interviews (n=12)

ARM (n=3)

Urogenital disease (n=4)

Epilepsy (n=1)





No diagnosis (n=1)

Turner (n=1)

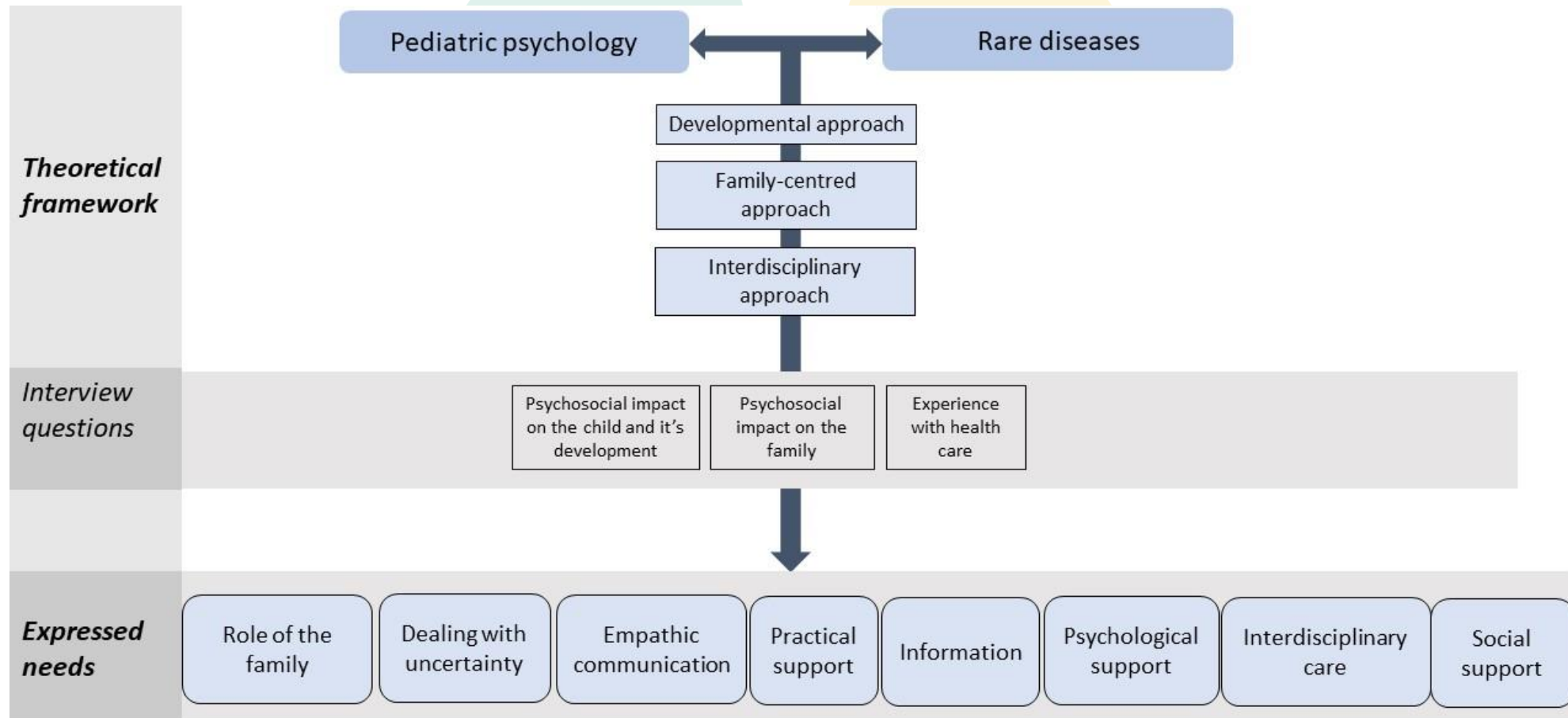
Metabolic disease (n=1)

FAS (n=1)

# Rare diseases: Qualitative research

-  **Background** The psychological burden of living with a rare disease or complex condition is often **understudied** and may present **overarching concepts** that shape the general experience of having been diagnosed with a rare condition.
-  **Aim** The present study examines common needs from a comprehensive perspective combining relevant aspects from the rare disease literature in a theoretical perspective from pediatric psychology, such as a **family-centred, developmental and interdisciplinary approach**.
-  **Method** Twelve interviews were conducted among parents from children with a rare disease or complex condition in an Integrated University Children's Hospital in the Netherlands
-  **Results** Eight themes followed from the analysis uncovered the need for **1) family-focused care, 2) coping with uncertainty, 3) empathic communication, 4) practical support, 5) information, 6) psychological support, 7) interdisciplinary care, and 8) social support**.

# Rare diseases: Qualitative research



# Rare diseases: Summary (Qualitative Research)



## ARM (n=3)

1. Uncertainty
2. Need for Information
3. Empathic communication
4. Psych support
5. Family centered approach
6. Practical support health care



## Urogenital (n=4)

1. Need for information
2. Family centered approach
3. Holistic approach
5. Empathic communication
4. Uncertainty
6. Transition to Adult Care



## Epilepsy (n=1)

1. Uncertainty
2. Empathic communication
3. Psych support
4. Holistic approach
5. Family centered approach
6. Practical support



## No Diagnosis (n=1)

1. Uncertainty
2. Empathic Communication
3. Holistic approach
4. Family centered approach



## Turner (n=1)

1. Need for information
2. Family centered approach
3. Practical support



## FAS (n=1)

1. Psych support

# Rare diseases: Summary (Qualitative Research)



## ARM (n=3)

1. Uncertainty
2. Need for Information
3. Empathic communication
4. Practical support health care
5. Family centered approach
6. Practical support health care



## Urogenital (n=4)

1. Need for information
2. Family centered approach
3. Holistic approach
4. Uncertainty
5. Empathic communication
6. Transition to Adult Care



## Epilepsy (n=1)

1. Uncertainty
2. Empathic communication
3. Psych support
4. Holistic approach
5. Family centered approach
6. Practical support



## No Diagnosis (n=1)

1. Uncertainty
2. Empathic Communication
3. Holistic approach
4. Family centered approach



## Turner (n=1)

1. Need for information
2. Family centered approach
3. Practical support



## FAS (n=1)

1. Psych support

# Rare disease: Common need: Need for information





# Rare diseases: Summary literature and qualitative research

Amalia kinderziekenhuis  
Radboudumc

## Common challenges in uncommon conditions:

1.  
Family focused  
care

2.  
Dealing with  
uncertainty

3. Empathic  
communication

4.  
Practical  
support

5.  
Need for  
information

6.  
Psychological  
support

7.  
Interdisciplinary  
care

8. Social  
support







Amalia kinderziekenhu  
**Radboudumc**

**THANK YOU FOR LISTENING!**

Join us on social media

 @RareTogether

 @RareTogether



# Closing remarks

## New EURORDIS Mental Wellbeing Partnership Network

Support and reinforce a united and empowered rare disease community affected by mental wellbeing ...

... to come together and be seen, learn, advocate and supported each other.





# EURORDIS Mental Wellbeing Partnership Network

**Call to join the Partnership  
Network ...**

... for patient representatives,  
medical expertise and  
researchers.



# EURORDIS Mental Wellbeing Web Page

Home \ Mental Health & Wellbeing

## Mental Health & Wellbeing

There is no health without mental health. The relationship between physical and mental health is well established. Increased severity and complexity of a physical health condition, in turn increases the risk to mental health and well-being result in higher rates of depression, anxiety and also where poor mental health can impact on physical health, the capacity to self-care, resilience. (King's Fund<sup>1</sup>, Royal College of Psychiatrists & Centre for Mental Health<sup>2</sup>, Mental Health Foundation<sup>3</sup>).

Mental health is a basic human right. The World Health Organization's definition of mental health<sup>4</sup>, seeing it as something that is strongly linked to opportunities, and participation in the community:

"A state of mental well-being that enables people to cope with the stresses of life, realize their abilities, learn well, (making healthy choice) and work well, and contribute to their community. It is an integral component of health and well-being that underpins our individual and collective abilities to make decisions, build relationships and shape the world we live in. And it is crucial to personal, community and socio-economic development."

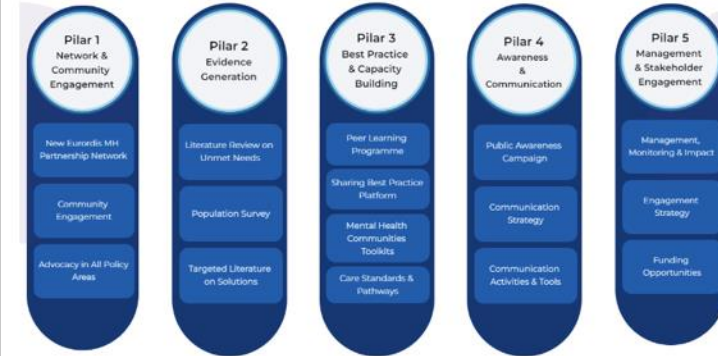
### Impact of Rare Disease on Mental Wellbeing

People living with a rare and undiagnosed condition have increased vulnerability and risk factors resulting in them experiencing an accumulative impact on their mental wellbeing, specifically at an individual level.

- At an Individual Level**  
Mental health is an associated co-morbidity for many rare diseases.
- At a Population Level**  
Increased psychological impact associated with the rare diseases journey across all stages of life.
- Cross-cutting psychosocial determinants**  
People living with a rare disease and their families have increased exposure to social inequalities and discrimination, which are risk factors and determinants for poor mental wellbeing.

## EURORDIS Action to Improve the Mental Wellbeing of the Rare Disease Community

EURORDIS wishes to leverage the opportunity of the new Communication on a Comprehensive Approach to Mental Health to make visible the unmet mental health needs of people living with a rare disease and their families and take affirmative action to address these needs.



A new EURORDIS Mental Wellbeing Initiative is planned to be launched in 2023-24, with the overarching objective to promote the development of a 'mentally healthy community' that reduces the accumulated impact of rare diseases on mental health and wellbeing among people living with a rare disease and their families.

## EURORDIS new Mental Wellbeing Partnership Network



EURORDIS will establish a new EURORDIS Mental Health Partnership Network (Partnership Network) to support and coordination of the development and delivery of community activities in the framework of EURORDIS Mental Wellbeing Initiative.

The Partnership Network will bring together experts and stakeholders to drive a community action on mental health and wellbeing for people living with a rare disease, their families and caregivers. Specifically, the Partnership Network will unite and empower the rare disease community to come together, learn, take action and tailor recommendations to the specific needs of people living with a rare disease, in all policy areas, to ensure the mental health and wellbeing of the rare disease community is improved. The findings of this network will feed into EURORDIS work across all policy areas.

EURORDIS launched a call for expressions of interest to join the new Partnership Network at a EURORDIS Membership Meeting (EMM 2023) Satellite Working – Enhancing the Mental Wellbeing in the Rare Disease Community held on the 25.05.2023.

Applications for our new Partnership Network are still open! EURORDIS is looking for dedicated volunteers to be active in the Partnership Network.

If the rare disease you represent has an impact on the mental health and wellbeing we would like to hear from you and ensure that your expertise and insights are included to shape the work of the Partnership Network.



Apply here <sup>1</sup>

Register to join Partnership Network





# Thank You