EURORDIS
RARE DISEASES EUROPE

Living with Uncertainty & Impact of Trauma

2nd 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

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Welcome & Opening Remarks

Daniel, Williams syndrome

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

President

GAMIAN Europe

Kym Winter

Founder & Clinical Director

Rare Minds

Kirsten Johnson

Fragile X International

EURORDIS Board of Director

Lucy McKay

Chief Executive Officer

Medics 4 Rare Diseases

Dorica Dan

Romanian Prader Willi Association

Vice-President of EURORDIS

Rosanne Smit

Psychologist & Researcher

Radboud University Medical Centre

EURORDIS.ORG

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: - Understanding of the root cause of the uncertainties and trauma associated with the rare disease pathway. - Identifying coping strategies to deal with uncertainty of the rare disease journey. - Identifying training needs for professionals to reduce the stressors associated with a rare disease.	Chair: Matt Bolz-Johnson Panel: • Kym Winter, Rare Minds • Lucy McKay, Medics 4 Rare Diseases • Dorica Dan, Romanian Prader Willi Association
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

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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? ☺

„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.

People these diagnosis types **need real support** and help in **coping 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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Living with uncertainty and impact of trauma

Panel Discussion

Panellist

Matt Bolz-Johnson

Mental Wellbeing Lead

Chair

Kym Winter

Rare Minds

Panellist - Psychologist

Lucy McKay

Medics 4 RD

Panellist - Medic

Dorica Dan

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.

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Questions & Answers

Panel Discussion

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Rare Disease
Research Partners

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

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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**

European Reference Networks
EJP RD
Amalia kinderziekenhuis
Radboudumc
Funded by the European Union GA n°825575

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.*

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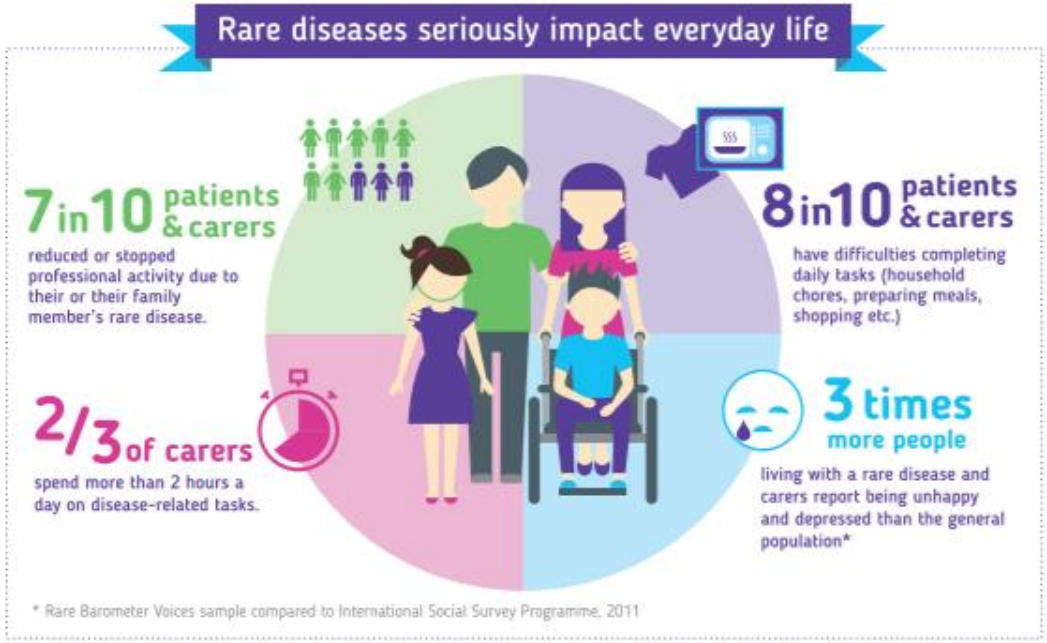
RARE 2030 RECOMMENDATIONS

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.



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European Reference Networks

RESEARCH LINE

Common needs in uncommon conditions

Amalia kinderziekenhuis

Radboudumc

EJP RD

Funded by the European Union
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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

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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)

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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 diseas were reviewed.
- **Conclusion** Several **overarching concepts** were highlighted

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)**

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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 managemen t/coping

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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

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Rare diseases:
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Aangeboren Urogenitale Aandoeningen

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chronische nierziekte

1. QoL
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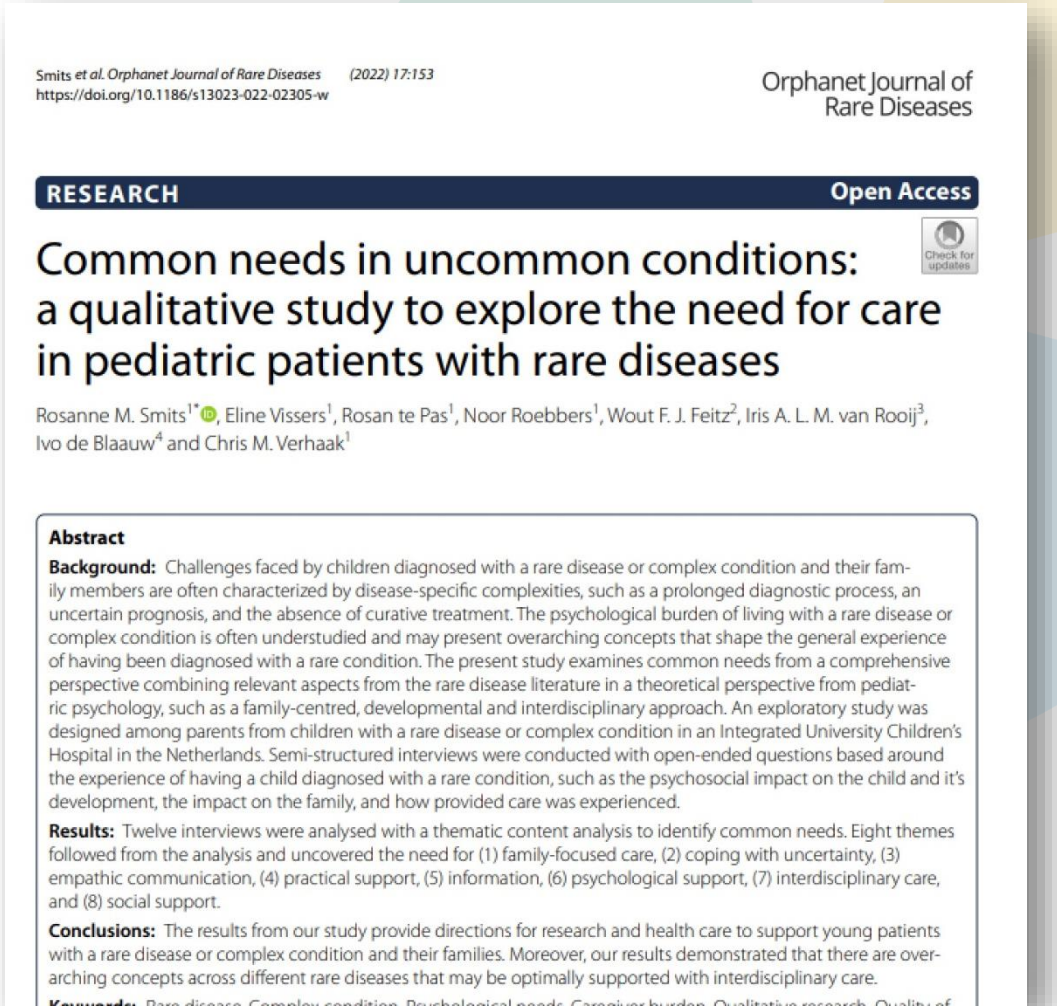
Rare diseases: Qualitative research



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[1*], Eline Vissers[1], Rosan te Pas[1], Noor Roebbers[1], Wout F. J. Feitz[2], Iris A. L. M. van Rooij[3], Ivo de Blaauw[4] and Chris M. Verhaak[1]

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 it's 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.

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)**

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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**.

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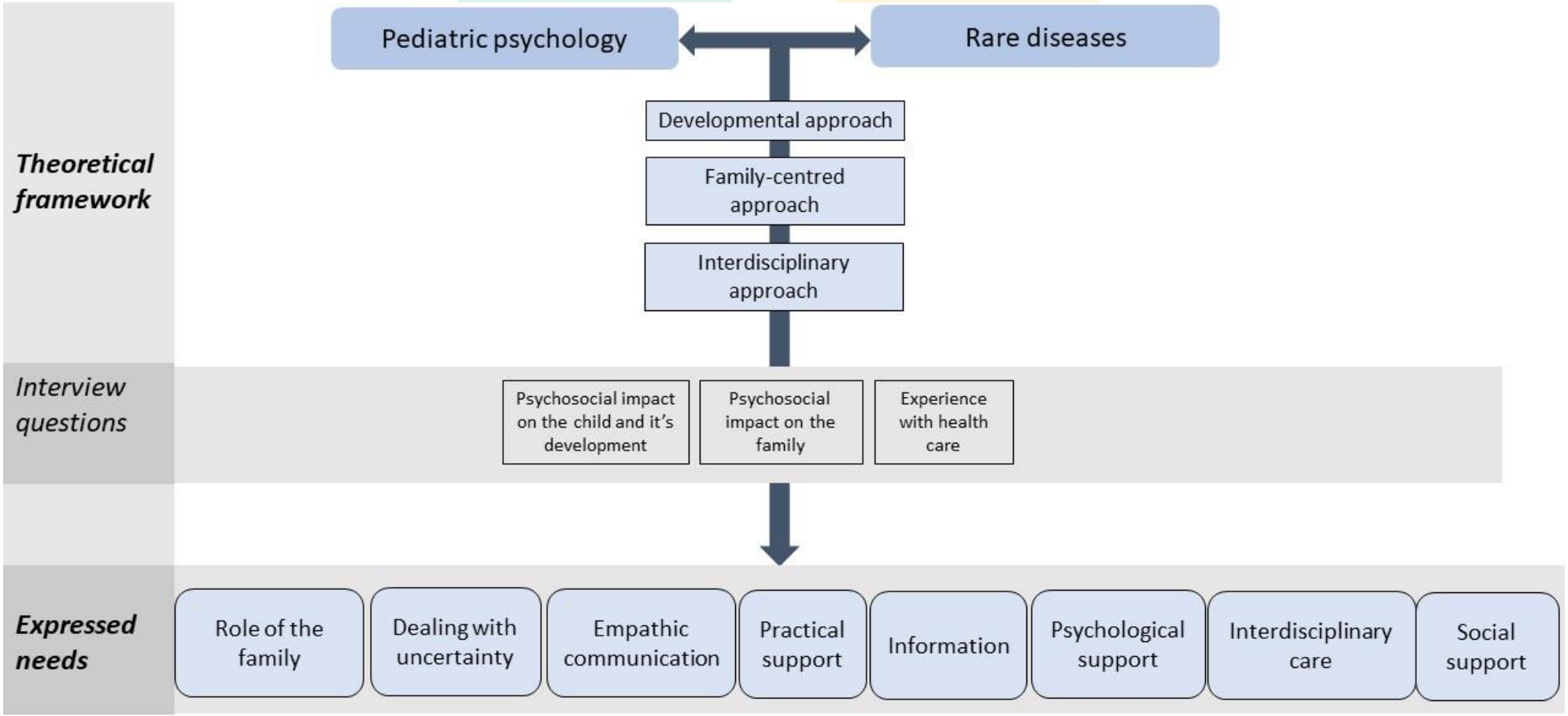
Rare diseases:
Qualitative research



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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 communiction
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

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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)

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Epilepsy (n=1)

1. Uncertainty
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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

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Rare disease:
Common need: Need for information

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Rare diseases:
Summary literature and qualitative research

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

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THANK YOU FOR LISTENING!

Join us on social media
@RareTogether
@RareTogether

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Kacper from Poland is living with Kawasaki disease

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.

Daniel, Williams syndrome

EURORDIS Mental Wellbeing Partnership Network

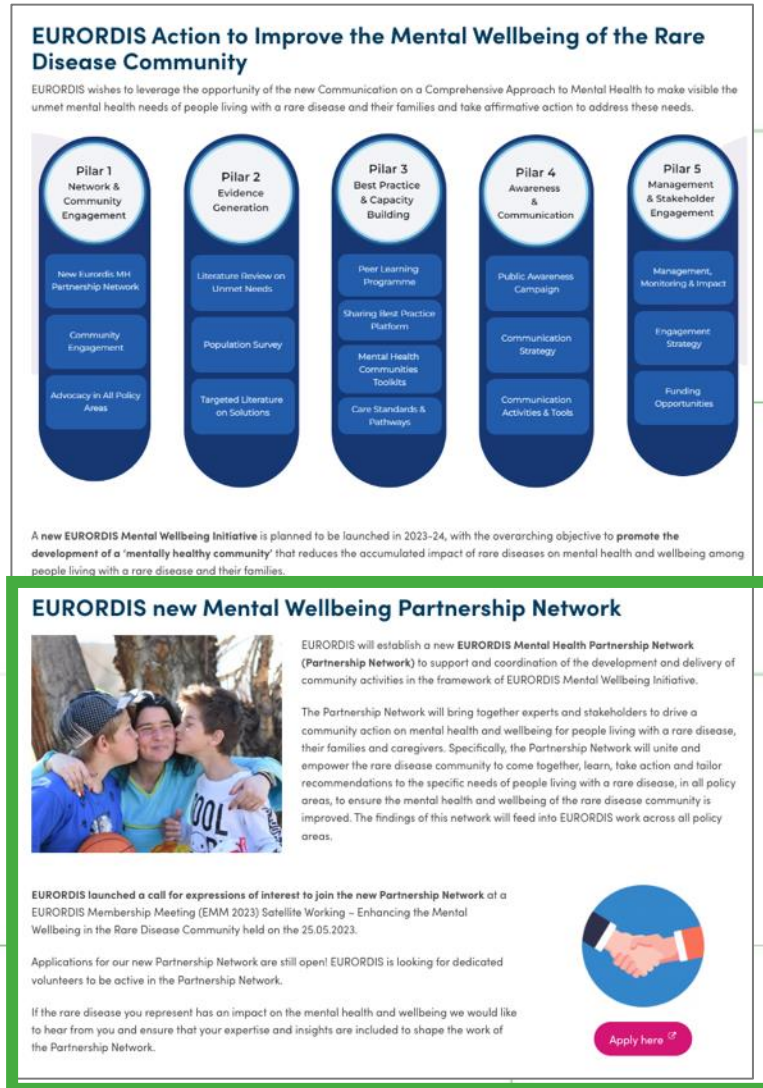
Call to join the Partnership Network …

… for patient representatives, medical expertise and researchers.

Email: Concha Mayo on concha.mayo@eurordis.org

EURORDIS.ORG

EURORDIS Mental Wellbeing Web Page



Register to join Partnership Network

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https://www.eurordis.org/mental-wellbeing/

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Thank You