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
Welcome & Opening Remarks

Iris, Ehlers-Danlos syndrome, chronic paroxysmal hemicrania, cluster headaches
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
## Agenda

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<tr>
<th>Time</th>
<th>Topic</th>
<th>Speaker</th>
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<td>15.00 – 15.03</td>
<td>Welcome &amp; Housekeeping</td>
<td>Concha Mayo, EURORDIS</td>
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<tr>
<td>15.03 – 15.10</td>
<td>Opening Remarks</td>
<td>Kirsten Johnson, Fragile X International &amp; EURORDIS</td>
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<td>15.10 – 15.30</td>
<td>Understanding the biopsychosocial continuum and addressing intersectional needs.</td>
<td>Peter Kéri, GAMIAN Europe</td>
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<td>15.30 – 16.00</td>
<td>Panel discussion:</td>
<td>Chair: Matt Bolz-Johnson</td>
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<td></td>
<td>- Understanding of the root cause of the uncertainties and trauma associated with the rare disease pathway.</td>
<td>Panel: Kym Winter, Rare Minds, Lucy McKay, Medics 4 Rare Diseases, Dorica Dan, Romanian Prader Willi Association</td>
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<td>- Identifying coping strategies to deal with uncertainty of the rare disease journey.</td>
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<td>- Identifying training needs for professionals to reduce the stressors associated with a rare disease.</td>
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<td>16.00 – 16.10</td>
<td>Questions &amp; Answers</td>
<td>All</td>
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<td>16.10 – 16.25</td>
<td>Impact of living with a congenital malformations on mental wellbeing of the family.</td>
<td>Rosanne Smit, Radboud University Medical Centre</td>
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</table>
Understanding the biopsychosocial continuum and addressing intersectional needs

Peter Kéri, GAMIAN Europe
Bio?
Psycho?
Social?

Péter Kéri
President
GAMIAN-Europe
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?

<table>
<thead>
<tr>
<th>Hope: after diagnosis, many people lose hope of a full recovery or cure. This can be a huge emotional burden.</th>
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<tbody>
<tr>
<td>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.</td>
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<tr>
<td>Mental health: people with rare, lifelong illnesses may be more prone to anxiety and depression because of the stress and uncertainty their condition causes.</td>
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<td>Social isolation: people sometimes view people with rare, incurable conditions differently and people may feel isolated or excluded.</td>
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<tr>
<td>Identity and self-image changes: people affected may need to reassess their identity and self-image when they receive such a diagnosis.</td>
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<tr>
<td>Need for support: people with these conditions often need professional help and loving support to manage emotional distress.</td>
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</table>
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”
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.”
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”
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“
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”
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.

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

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!

president@gamian.eu
mail2peterkeri@gmail.com
Living with uncertainty and impact of trauma

Panel Discussion
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)
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
Common needs in uncommon conditions
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.
Common needs in uncommon conditions
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

<table>
<thead>
<tr>
<th>Literature overview</th>
<th>Qualitative data (TRANSIT interviews)</th>
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<tbody>
<tr>
<td>Articles (n=56)</td>
<td>Interviews (n=12)</td>
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<tr>
<td>Mitochondrial disease (n=13)</td>
<td>ARM (n=3)</td>
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<tr>
<td>Prader Willi Syndrome (n=13)</td>
<td>Urogenital disease (n=4)</td>
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<td>DSD (n=9)</td>
<td>Epilepsy (n=1)</td>
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<tr>
<td>Urogenital disease (n=9)</td>
<td>No diagnosis (n=1)</td>
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<tr>
<td>ARM (n=9)</td>
<td>Turner (n=1)</td>
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<td>Rare kidney disease (n=5)</td>
<td>Metabolic disease (n=1)</td>
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<td>FAS (n=1)</td>
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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
<table>
<thead>
<tr>
<th>Rare diseases:</th>
<th>Summary (Literature overview)</th>
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<tbody>
<tr>
<td>MITO</td>
<td>1. Need for information</td>
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<td>2. Coping/communication</td>
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<td></td>
<td>3. Practical support health care</td>
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<td></td>
<td>4. Concerns about inheritance</td>
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<tr>
<td>PWS</td>
<td>1. Coping: Hyperphagia</td>
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<td></td>
<td>2. Coping: Sleep</td>
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<td></td>
<td>3. Well-being/QoL</td>
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<td></td>
<td>4. Role of caregiver</td>
</tr>
<tr>
<td>DSD</td>
<td>1. Need for information Sex/fertility</td>
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<td>2. Non-binary information</td>
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<td>3. Holistic approach</td>
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<td></td>
<td>4. Follow-up and transition to adult care</td>
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<td></td>
<td>5. Role of caregiver</td>
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<td>6. QoL</td>
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<td>7. Information disease management</td>
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<tr>
<td>ARM</td>
<td>1. Sex/fertility</td>
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<td>5. Role of caregiver</td>
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<td>6. Information about disease and management/coping</td>
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<tr>
<td>Radboudumc Expertisecentrum Aangeboren Urogenitale Aandoeningen</td>
<td>1. QoL</td>
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<td>2. Information in sexual development</td>
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<td>3. Body image</td>
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<td>4. Transition to adult care</td>
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<td>chronicische nierziekte</td>
<td>1. QoL</td>
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<td>2. FoP</td>
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<td>3. Fear of transplantation</td>
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<td>4. Unknown future</td>
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<td>5. Desire for new treatment</td>
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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 about disease and management/coping

**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
5. Role of caregiver
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**chronische nierziekte**
1. QoL
2. FoP
3. Fear of transplantation
4. Unknown future
5. Desire for new treatment
# Rare diseases: Qualitative research

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

**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-centered, 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 analyzed 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.

## Interviews (n=12)

<table>
<thead>
<tr>
<th>Disease</th>
<th>Number</th>
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<tr>
<td>ARM</td>
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<tr>
<td>Urogenital disease</td>
<td>4</td>
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<tr>
<td>Epilepsy</td>
<td>1</td>
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<tr>
<td>No diagnosis</td>
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<tr>
<td>Turner</td>
<td>1</td>
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<td>Metabolic disease</td>
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<td>FAS</td>
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Funded by the European Union

[GA n° 825575](https://doi.org/10.1186/s13232-022-02305-w)
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

Theoretical framework:
- Pediatric psychology
- Rare diseases
  - Developmental approach
  - Family-centred approach
  - Interdisciplinary approach

Interview questions:
- Psychosocial impact on the child and its development
- Psychosocial impact on the family
- Experience with health care

Expressed needs:
- Role of the family
- Dealing with uncertainty
- Empathic communication
- Practical support
- Information
- Psychological support
- Interdisciplinary care
- Social support
# Rare diseases: Summary (Qualitative Research)

<table>
<thead>
<tr>
<th>Condition</th>
<th>ARM (n=3)</th>
<th>Urogenital (n=4)</th>
<th>Epilepsy (n=1)</th>
<th>No Diagnosis (n=1)</th>
<th>Turner (n=1)</th>
<th>FAS (n=1)</th>
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<td>5. Family centered approach</td>
<td>5. Empathic communication</td>
<td>5. Family centered approach</td>
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<td>6. Practical support health care</td>
<td>6. Uncertainty</td>
<td>6. Transition to Adult Care</td>
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# Rare diseases: Summary (Qualitative Research)

<table>
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<tr>
<th>Rare Disease</th>
<th>ARM (n=3)</th>
<th>Urogenital (n=4)</th>
<th>Epilepsy (n=1)</th>
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**1. Need for Information**
- ARM: 1.
- Urogenital: 1.
- Epilepsy: 1.
- No Diagnosis: 1.
- Turner: 1.
- FAS: 1.

**2. Family centered approach**
- ARM: 2.
- Urogenital: 2.
- Epilepsy: 2.
- No Diagnosis: 2.
- Turner: 2.
- FAS: 2.

**3. Holistic approach**
- ARM: 3.
- Urogenital: 3.
- Epilepsy: 3.
- No Diagnosis: 3.
- Turner: 3.
- FAS: 3.

**4. Empathic communication**
- ARM: 4.
- Urogenital: 4.
- Epilepsy: 4.
- No Diagnosis: 4.
- Turner: 4.
- FAS: 4.

**5. Empathic communication**
- ARM: 5.
- Urogenital: 5.
- Epilepsy: 5.
- No Diagnosis: 5.
- Turner: 5.
- FAS: 5.

**6. Practical support health care**
- Urogenital: 6.
- Epilepsy: 6.
- No Diagnosis: 6.

**6. Transition to Adult Care**
- Urogenital: 6.
- Epilepsy: 6.
- No Diagnosis: 6.
Rare disease:
Common need: Need for information

1. Need for information
   - Information about coping w/ hyperphagia
2. Information in sex development
3. Non-binary information
4. Unknown future
5. Information about disease and management/coping
6. Information about disease and management/coping
7. Information about disease and management/coping

Uncertainty
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
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.

Email: Concha Mayo on concha.mayo@eurordis.org
EURODIS Mental Wellbeing Web Page

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 can raise the risk of mental health problems. Studies also show that poor mental health can impact on physical health, the capacity to work, resilience, well-being and quality of life. Mental Health is a basic human right.

“Health is a state of mental well-being that enables people to cope with the stresses of life, to perform their daily activities, to work and socialise, and to 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 in which 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 disease and their families have increased exposure to real inequities and discrimination, which are risk factors and determinants for their mental wellbeing.

EURODIS Action to Improve the Mental Wellbeing of the Rare Disease Community

EURODIS will be launching the new EURODIS Mental Health Partnership Network (Partnership Network) to support and coordinate the development and delivery of community activities aligned to the framework of EURODIS 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 carers. Specifically, the Partnership Network will unite and empower the rare disease community to come together, make an impact action and better research and treatment options for 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 work will feed into EURODIS work across all policy areas.

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

Register to join Partnership Network
Thank You

Iris, Ehlers-Danlos syndrome, chronic paroxysmal hemicrania, cluster headaches