Patient Workshop

Thursday 12 December 2024

PM - PART 2 Patient Journey

Hosted by Pr. Maria Puiu, Dorica Dan, Tanja Zdolšek Draksler

European Commission

EUROPEAN REFERENCE NETWORKS

Helping patients with rare or low-prevalence complex diseases

ERN ITHACA Anne Hugon

European Reference Networks ITHACA

Mental health and wellbeing and rare conditions

Kristen Johnson

European Commission

EUROPEAN REFERENCE NETWORKS

Helping patients with rare or low-prevalence complex diseases

European Reference Networks

ITHACA

Mental Health and Wellbeing in Rare Conditions

Presented by Dr Kirsten Johnson

- President, Fragile X International
- Board member, EURORDIS
- Chair of Council, Rare Diseases International

Email: kirsten.johnson@eurordis.org

Mental Health and Wellbeing Initiative

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

Impact of Rare Diseases on Mental Health & Wellbeing

54% people living with a rare disease declare that the rare disease caused or amplified isolation from friends and family.

(Juggling life and care Survey, 2019)

Emotional Wellbeing

>90% of PLWRD surveyed felt worried, anxious, stressed and/or depressed.

19% had suicidal thoughts.

(Spencer-Tansley 2022)

Access Support, Faster Diagnosis

Average time to diagnosis of 3.8 Yrs.

vs 5.1 Yrs.

PLWRD whose needs for psychological support were met accessed a diagnosis faster.

EUR. RB Survey on Diagnosis, 2022

Financial Independence

7/10 PLWRD have reduced or stopped professional activity due to their own or their family members' disease.

(Courbier et al. 2017)

Holistic Care

50-85% of PLWRD were not offered or received sufficient psychological support in an Expert Centre.

(Courbier et al. 2017; Nunn et al. 2017)

Rare Barometer

A EURORDIS INITIATIVE

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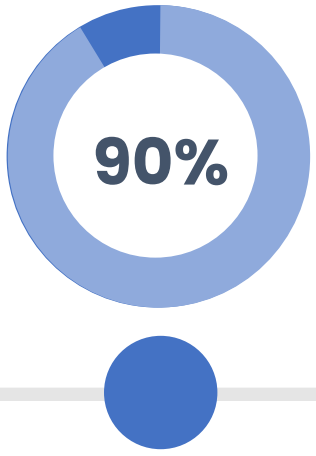
Evidence-base



>90% of PLWRD surveyed

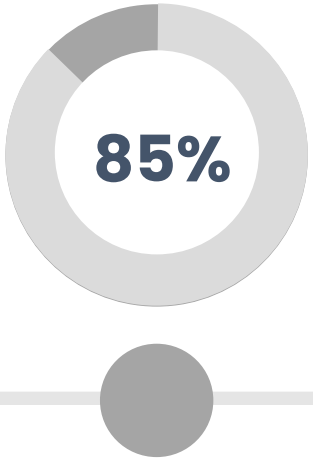
felt worried, anxious, stressed and/or depressed and 19% had suicidal thoughts.

(Spencer-Tansley 2022)

>1/3 of parents of a child

of a child with an undiagnosed disease meet clinical criteria for mild to moderate depression or anxiety.

(McConkie-Rosell 2018)



85% PLWRD

declared that the rare disease impacts upon several aspects of their physical and mental health and everyday life.

(Courbier et al. 2017)

PLWRD & their families

PLWRD and their families report being x3 times more unhappy and depressed compared with the general population.

(Courbier et al. 2017)

7 out of 10 patients & carers

7 out of 10 patients and carers must reduce or stop their professional activity due to the rare disease and associated burden.

(Courbier et al. 2017)

The diagnosis of a rare disease has a significant psychological impact upon the whole family, not just the affected individual (Kenny 2022)

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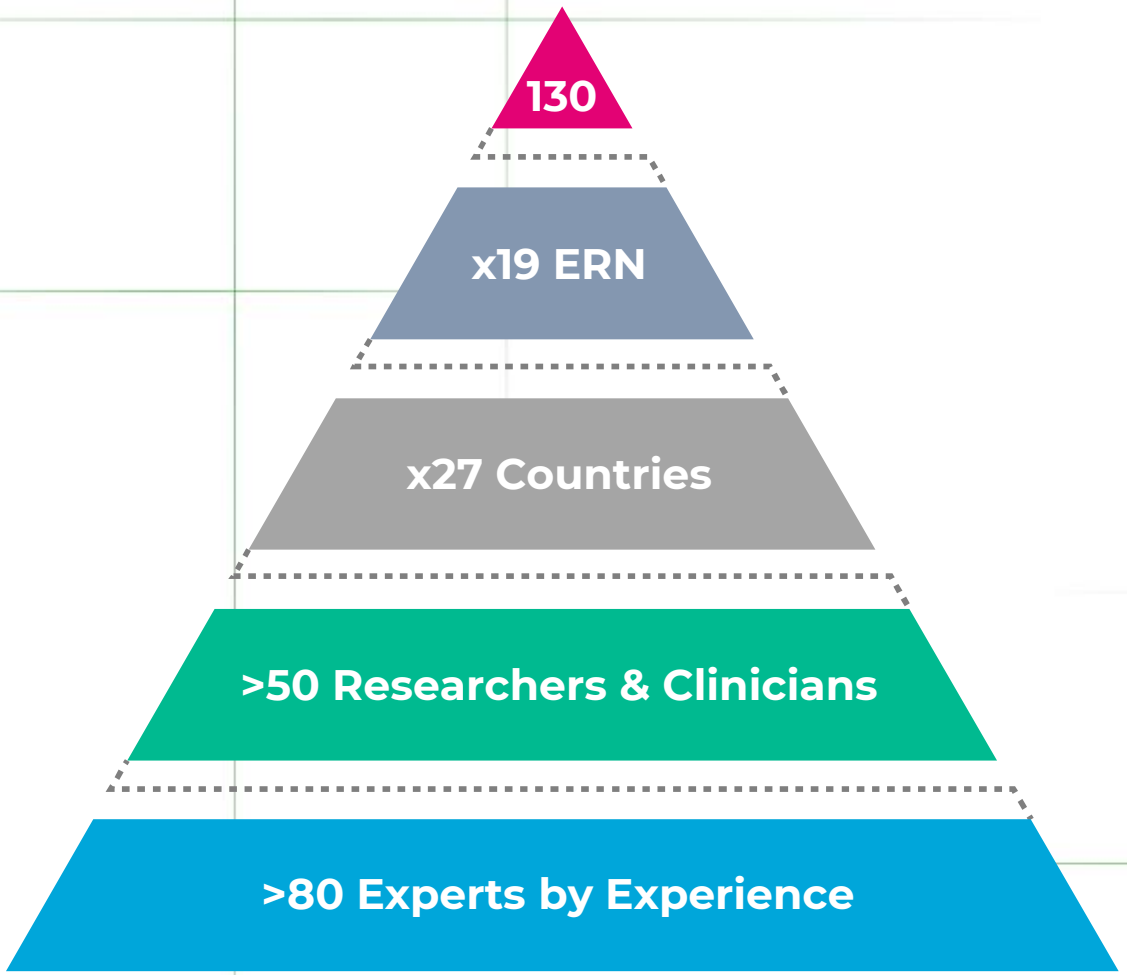
RARE DISEASES EUROPE

EURORDIS Mental Wellbeing Partnership Network

Kasper from Poland is living with Kawasaki disease

Membership



Total No. of Network Members: 130

European Reference Networks: 19 ERNs

EndoERN, ERKNet, ERN BOND, ERN CRANIO, ERN EpiCARE, ERN EURACAN, ERN eUROGEN, ERN GENTURIS, ERN Rare Liver, ERN ReCONNET, ERN EuroBloodNet, ERN EYE, ERN ITHACA, ERN LUNG, ERN-RDN, ERN Skin, ERNICA, MetabERN, VASCERN

Total No. of Member States Covered: 27 Countries

18 EU Member States incl. Austria, Belgium, Bulgaria, Croatia, Finland, France, Germany, Greece, Hungary, Ireland, Italy, Latvia, Malta, Netherlands, Portugal, Romania, Spain and Sweden

9 Non-EU MS incl. Australia, Canada, Ethiopia, Georgia, Nepal, Serbia, Switzerland, UK and USA.

Researchers, Psychologists & Medical Experts: >50

Total No. of Experts by Experience: >80

Factors can increase or decrease our likelihood of experiencing mental health problems

Positive norms

Opportunities to pursue interests

Social relationships, community and networks

Physical health

Physical and psychological safety

Good support system

Poverty, cost of living and ability to work

Bullying and experiencing prejudice

Injury, infections, health issues

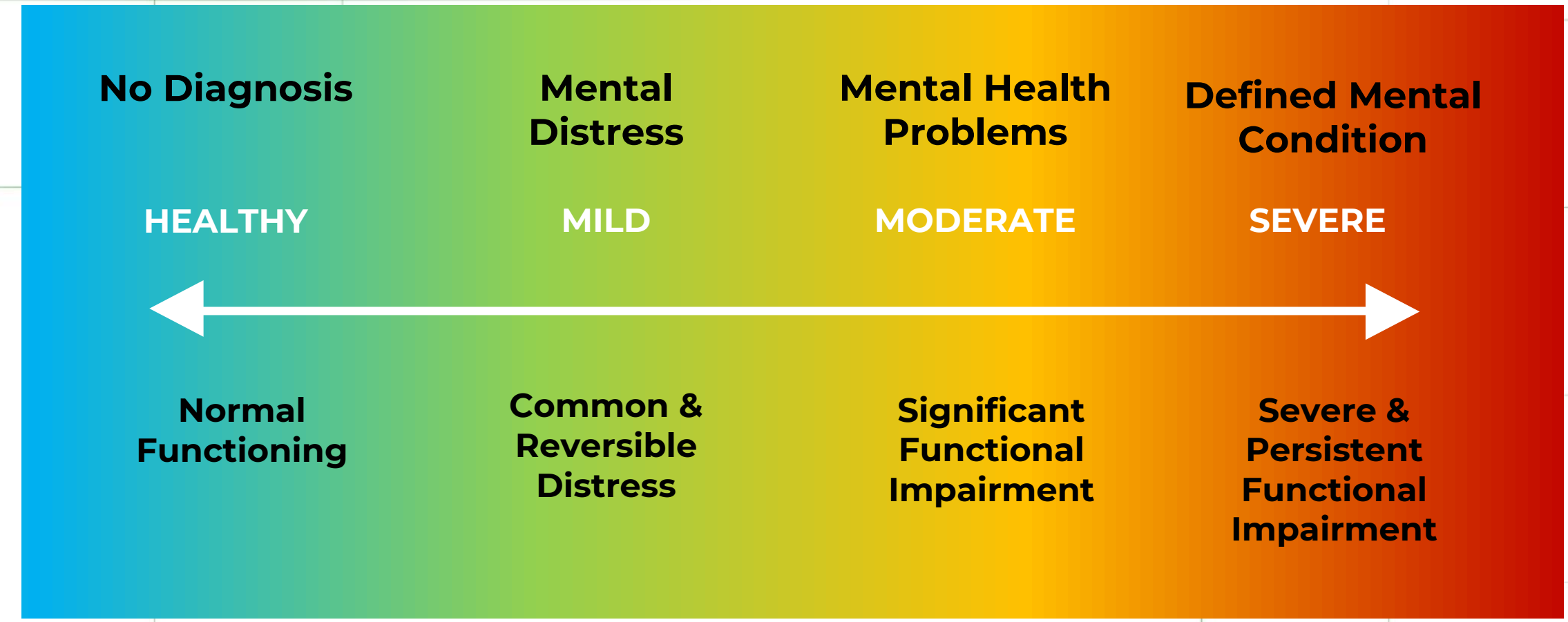
Traumatic Events, violence and adverse life events

Substance abuse

Force displacement and humanitarian emergencies

Lack of social support

When does an experience become a symptom?



Stressors Associated with Rarity

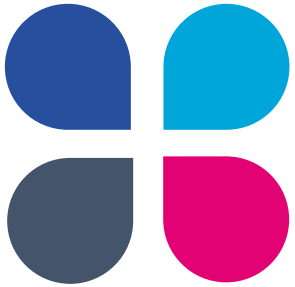
Progressive, degenerative, disabling and frequently life-threatening conditions.

Diagnostic Odyssey

- Lengthy/traumatic diagnosis odyssey (consult x5 doctors, av. 5 years).
- History of misdiagnosis (av. x3).
- Poor communication of diagnosis > Reduced trust in HCP's.

Complex interacting emotional and physical symptoms

- High impact of mental health aspects of a rare condition.
- High logistical burden of frequent hospital appointments.
- Extremely intensive tests and treatment. Medical trauma and impact on physical, social and development



Living with uncertainty

- Multiple uncertainties – prognosis and future increases anxiety.
- Low disease awareness among professionals / public, increases isolation and frustration.

Stress & Strain on Relationships

- Stress and strain on family life / couple relationships
- Impact of genetic inheritance on identity, and life choices
- Increased grief and loss

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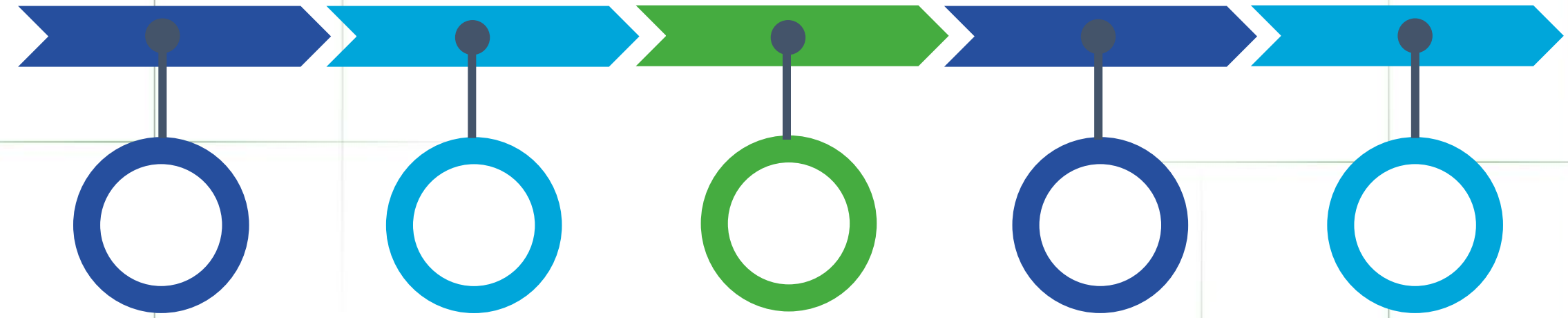
raremınds
Mental Health for the Rare Disease Community

Accumulative Impact on Wellbeing



Unseen & Neglected Needs

Unseen and neglected unmet need that is frequently overshadowed by the other medical complexities

Increase Risk Factors

Increased exposure to social inequalities and discrimination

Increase risk factors for poor mental health.

Individual Level

PLWRD can have associated mental health co-morbidities

Population Level

Community live with the increased psychological impact associated with the rare diseases journey across all stages of life

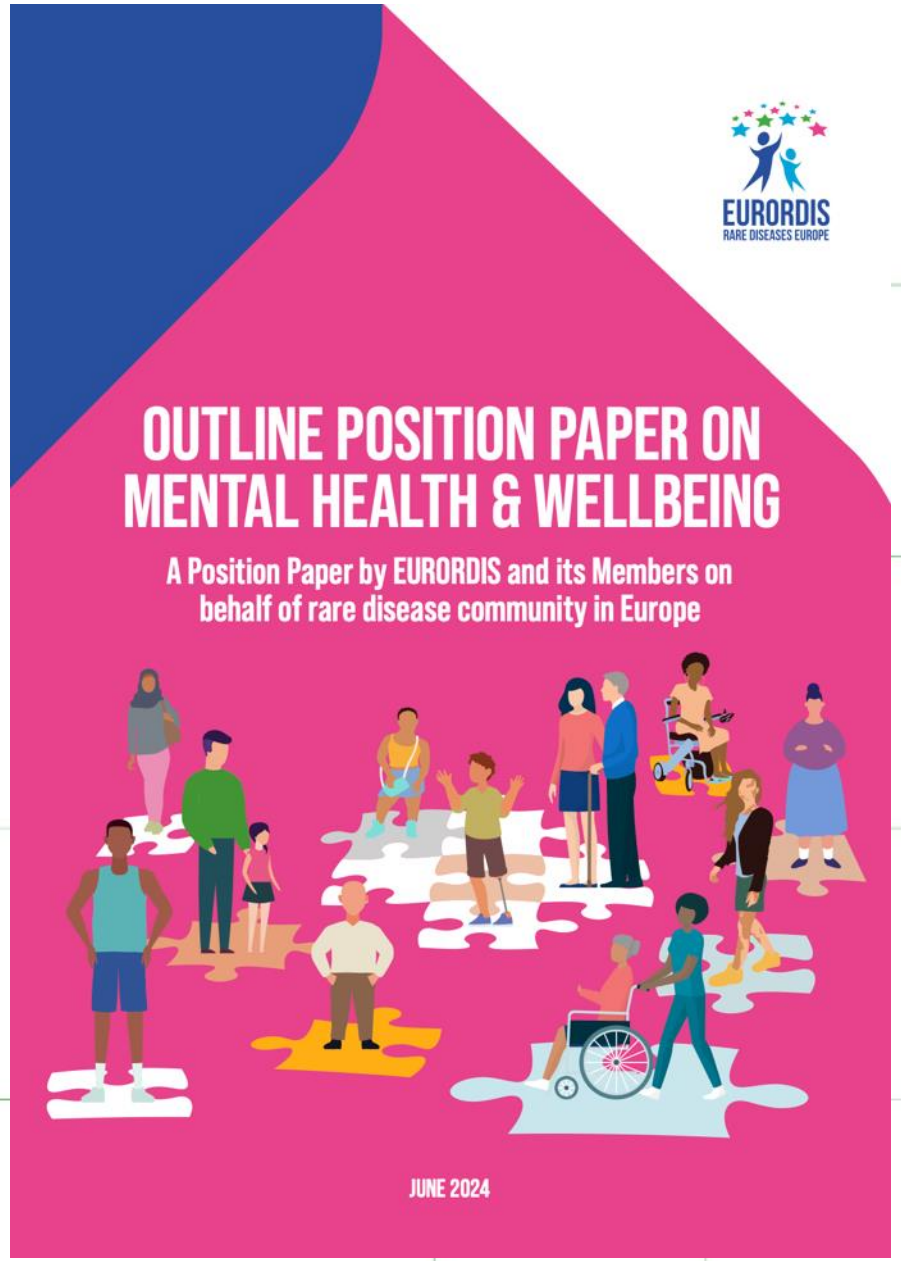
Priority Area of Needs

Look beyond the physiological symptoms of a rare disease and take action to address the psychological impact associated with rare conditions.

Outline Position Paper on Mental Health & Wellbeing

Published: 28 June 2024

https://www.eurordis.org/publications/position-paper-on-mental-health-and-wellbeing/



Development Process

Q1-2 2023

- Gathering Evidence
- Submitted Response for the Call for Evidence on RD & MH
- Submitted a Poster of a Concrete Action
- EMM2023 Satellite Workshop on Mental Wellbeing

Q3-4 2023

- Published Full Response on the new Communication
- Contribution to MEP Own Initiative (INI) Report
- EP Policy Event on People in Vulnerable Situations

Q1 2024

Draft Outline Position Paper

Feedback from:

- EURORDIS Mental Health Partnership Network
- Council of National Alliances
- EURORDIS Board of Directors & Team

Q2 2024

- "What psychosocial care looks like for PLWRD" Webinar
- WG1 Advocacy Sub-Group co-created the "blueprint for psychosocial care for PLWRD"
- Final version completed.

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Structure of Outline Position Paper

1. Introduction: Rare disease impact on mental health and wellbeing
2. Policy Context
    - MH as an EU Public Health Priority under Commission Communication
    - World Health Organization (WHO) recognises “people with an existing health condition” as one of the main vulnerable groups
    - UN GA Resolution A/RES/76/132 (2021) urges Member States to implement psychosocial support programmes for PLWRD
3. European Recommendations: visibility of rare diseases in mental health policy:
    - Call for a more inclusive approach of all populations in vulnerable situations
    - Dedicated actions with adequate financial support for all vulnerable groups
4. National Recommendations: visibility of mental health in rare disease national plans and strategies, specifically to establish psychosocial support programmes through “psychologically informed medical care”

Set out what “psychologically informed medical care” would look like addressing eight common areas of needs, specifically:

(1) family-focused care
(2) coping with uncertainty
(3) empathic communication
(4) practical support
(5) need for information
(6) psychological support
(7) interdisciplinary care
(8) social support

Policy Context

UN General Assembly Resolution on Addressing the challenges of persons living with a rare disease and their families:

10. *Urges* Member States to implement effective programmes to promote mental health and psychosocial support for persons living with a rare disease, and to promote policies and programmes that enhance the well-being of their families and caregivers. A/RES/76/132

World Health Organisation, 2022:

The World Health Organization (WHO) recognized "people with an existing health condition" as one of the main vulnerable groups who were more likely to develop symptoms of mental disorders following the pandemic, along with young people and women (WHO 2022).

European Union, 2023:

- Mental health is recognized as a public health priority in the European Union, notably in Commission President von der Leyen's State of the Union speech at the European Parliament on the 14 September 2022.
- The new Commission Communication on a Comprehensive Approach to Mental Health was published on 7 June 2023.
- The European Parliament Resolution on Mental Health, passed 12 December 2023

European Commission Communication on a Comprehensive Approach to Mental Health

x20 Flagship Initiative with a budget: EUR **1.23** billion

x6 priorities:

1. Integrating mental health across policies
2. Promoting good mental health, prevention and early intervention for mental health problems
3. Boosting the mental health of children and young people
4. **Helping those most in need**
5. Tackling psychosocial risks at work
6. Reinforcing mental health systems and improving access to treatment and care

EURORDIS recognizes that the new Communication is an important first step in a new comprehensive approach to address mental health, coordinating action across all policy areas, but needs to be inclusive of all populations in vulnerable situations.

Helping those most in need

People in Vulnerable Situations

- **Children & Young People**
- Older People
- **Cancer patients and survivors**
- **People living with disabilities**
- Migrant and refugee populations
- Ethnic minorities
- People from lower socio-economic backgrounds
- LGBTIQ+ people
- **Women**
- Victims of gender-based violence
- Victims of trafficking in human beings
- Victims of crimes
- Roma
- People living in rural or remote areas

People living with a rare disease have multiple 'intersectional' needs including:

- Children & Young People
- Women
- Older People
- Living with a disability
- Chronic Conditions & Multi-comorbidities
- Cancer

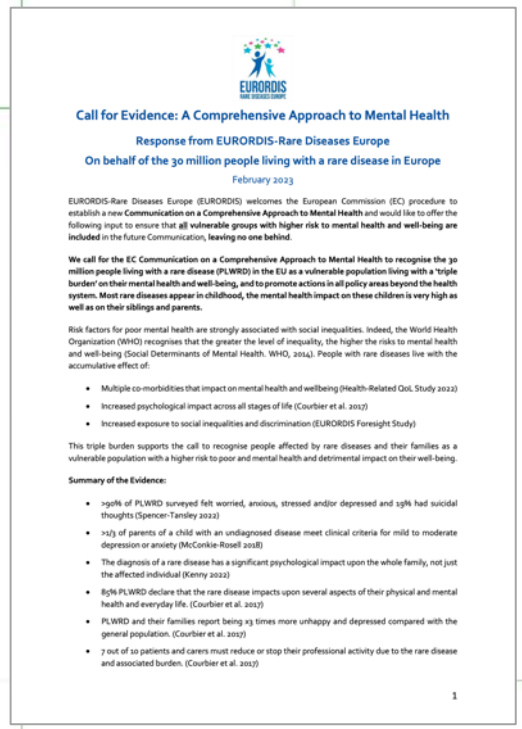
Shift in definition since the publication of Communication

Two Flagship Initiatives:

- Protecting Victims of Crime (11)
- Cancer Mission: Platform for Young Cancer Survivors (12)

Build on Evidence-base & Advocacy

Our key ask is that people living with a rare diseases are recognised as a population living in vulnerable situations, due to the multiple intersectional needs, requiring specific attention in the new Communication.



Call for Evidence: A Comprehensive Approach to Mental Health

Response from EURORDIS-Rare Diseases Europe

On behalf of the 30 million people living with a rare disease in Europe

February 2023

EURORDIS-Rare Diseases Europe (EURORDIS) welcomes the European Commission (EC) procedure to establish a new **Communication on a Comprehensive Approach to Mental Health** and would like to offer the following input to ensure that **all vulnerable groups with higher risk to mental health and well-being are included** in the future Communication, **leaving no one behind**.

We call for the EC Communication on a Comprehensive Approach to Mental Health to recognise the 30 million people living with a rare disease (PLWRD) in the EU as a vulnerable population living with a 'triple burden' on their mental health and well-being, and to promote actions in all policy areas beyond the health system. Most rare diseases appear in childhood, the mental health impact on these children is very high as well as on their siblings and parents.

Risk factors for poor mental health are strongly associated with social inequalities. Indeed, the World Health Organization (WHO) recognises that the greater the level of inequality, the higher the risks to mental health and well-being (Social Determinants of Mental Health. WHO, 2014). People with rare diseases live with the accumulative effect of:

- Multiple co-morbidities that impact on mental health and wellbeing (Health-Related QoL Study 2022)
- Increased psychological impact across all stages of life (Courbier et al. 2017)
- Increased exposure to social inequalities and discrimination (EURORDIS Foresight Study)

This triple burden supports the call to recognise people affected by rare diseases and their families as a vulnerable population with a higher risk to poor and mental health and detrimental impact on their well-being.

Summary of the Evidence:

- >90% of PLWRD surveyed felt worried, anxious, stressed and/or depressed and 19% had suicidal thoughts (Spencer-Tansley 2022)
- >2/3 of parents of a child with an undiagnosed disease meet clinical criteria for mild to moderate depression or anxiety (McConkie-Rosell 2018)
- The diagnosis of a rare disease has a significant psychological impact upon the whole family, not just the affected individual (Kenny 2022)
- 85% PLWRD declare that the rare disease impacts upon several aspects of their physical and mental health and everyday life. (Courbier et al. 2017)
- PLWRD and their families report being x3 times more unhappy and depressed compared with the general population. (Courbier et al. 2017)
- 7 out of 10 patients and carers must reduce or stop their professional activity due to the rare disease and associated burden. (Courbier et al. 2017)

1

EURORDIS Response to the EC Call for Evidence, 2023

A COMPREHENSIVE APPROACH TO MENTAL HEALTH

#MentalHealth #HealthUnion

BACKGROUND

Increased mental health risks are a reality for a range of vulnerable populations including youth, elderly, women, people with pre-existing health conditions[1] (such as chronic conditions and rare diseases) or disabilities, migrants, ethnic minorities, LGBTIQA+. Without mental health promotion, prevention and support, the psychological burden and mental health co-morbidities detrimentally impact on vulnerable populations' participation in society. The Comprehensive Approach on Mental Health needs to include all vulnerable populations and detail actions that are holistic, flexible and can be tailored to the specific needs of each population, across all policy areas.

Evidence demonstrates the link between physical health and mental wellbeing, with a correlation between the severity and complexity of a physical medical condition and the impact on mental health and wellbeing[2]. This is true for chronic conditions and rare diseases, with an increased psycho-social vulnerability in these populations. Rare diseases are complex, chronic, and often debilitating disorders with multiple intersectional unmet needs. Frequently affecting children, rare diseases lead to social exclusion and discrimination, impacting access to education and employment. The accumulative impact of living with increased co-morbidities, psychological stressors, and social inequalities results in a higher prevalence of depression and anxiety disorders.

CONCRETE ACTION

How will this help to strengthen action on mental health at EU level and enhance the mental health of European citizens?

Idea: To strengthen the capacities and empowerment of vulnerable groups to become mentally healthy communities, by developing a Mental Health & Wellbeing Toolkit for Vulnerable Populations. This toolkit can be generic, identifying best practices and evidence-based tools that can be tailored by each vulnerable population to address their specific needs.

EU added value and potential benefits: Addressing the needs of vulnerable populations present an area of added value for the European Union where EU community actions help to strengthen national measures. It is reported that each vulnerable population comprise a significant sized community – young people (67 million), older people (94 million), chronic diseases (50 million) and rare diseases (30 million).

There is a large discrepancy in health and social services available across EU countries to support these vulnerable populations. The development of a Mental Health & Wellbeing Toolkit for Vulnerable Populations would help address these gaps by strengthening the capacity of patient organisations and Civil Society Organisations (CSO) with EU-shared best practices and tools to support their respective communities, reducing avoidable downstream health, social and economic costs.

A Mental Health & Wellbeing Toolkit for Vulnerable Populations could be used by patient groups, healthcare professionals and social service providers to build knowledge and understanding of the needs of each vulnerable population and the associated impact on mental health, allowing to better target simple effective psycho-social interventions for different populations.

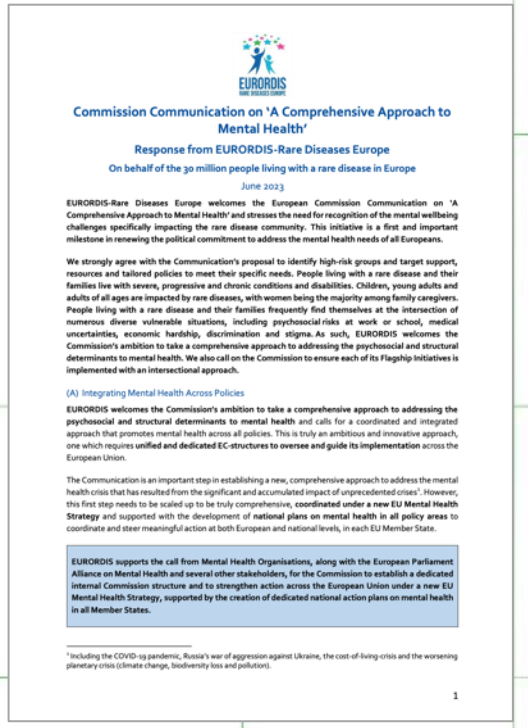
Increasing resilience and capacities among the civil society organisations and patient groups, can support and empower them to better alleviate the risk factors and drivers of poor mental health, preventing mental health co-morbidities and addressing the high levels of unmet needs of these vulnerable populations.

Specifically:

- Reducing the impact on the lives of the affected individuals and their family members, increasing social inclusion and supporting access to education, employment and independent living.

[1] World Health Organization - COVID-19 pandemic triggers 25% increase in prevalence of anxiety and depression worldwide, 2022
[2] The King's fund. Long term conditions and mental health: the cost of co-morbidities, 2012

EURORDIS Poster of a Proposal for a Concrete Action, 2023



Commission Communication on 'A Comprehensive Approach to Mental Health'

Response from EURORDIS-Rare Diseases Europe

On behalf of the 30 million people living with a rare disease in Europe

June 2023

EURORDIS-Rare Diseases Europe welcomes the European Commission Communication on 'A Comprehensive Approach to Mental Health' and stresses the need for recognition of the mental wellbeing challenges specifically impacting the rare disease community. This initiative is a first and important milestone in renewing the political commitment to address the mental health needs of all Europeans.

We strongly agree with the Communication's proposal to identify high-risk groups and target support, resources and tailored policies to meet their specific needs. People living with a rare disease and their families live with severe, progressive and chronic conditions and disabilities. Children, young adults and adults of all ages are impacted by rare diseases, with women being the majority among family caregivers. People living with a rare disease and their families frequently find themselves at the intersection of numerous diverse vulnerable situations, including psychosocial risks at work or school, medical uncertainties, economic hardship, discrimination and stigma. As such, EURORDIS welcomes the Commission's ambition to take a comprehensive approach to addressing the psychosocial and structural determinants to mental health. We also call on the Commission to ensure each of its Flagship Initiatives is implemented with an intersectional approach.

(A) Integrating Mental Health Across Policies

EURORDIS welcomes the Commission's ambition to take a comprehensive approach to addressing the psychosocial and structural determinants to mental health and calls for a coordinated and integrated approach that promotes mental health across all policies. This is truly an ambitious and innovative approach, one which requires **unified and dedicated EC-structures to oversee and guide its implementation** across the European Union.

The Communication is an important step in establishing a new, comprehensive approach to address the mental health crisis that has resulted from the significant and accumulated impact of unprecedented crises[1]. However, this first step needs to be scaled up to be truly comprehensive, **coordinated under a new EU Mental Health Strategy** and supported with the development of **national plans on mental health in all policy areas** to coordinate and steer meaningful action at both European and national levels, in each EU Member State.

EURORDIS supports the call from Mental Health Organisations, along with the European Parliament Alliance on Mental Health and several other stakeholders, for the Commission to establish a dedicated internal Commission structure and to strengthen action across the European Union under a new EU Mental Health Strategy, supported by the creation of dedicated national action plans on mental health in all Member States.

[1] Including the COVID-19 pandemic, Russia's war of aggression against Ukraine, the cost-of-living-crisis and the worsening planetary-crisis (climate change, biodiversity loss and pollution).

1

EURORDIS Response to the EC Commission Communication on the Comprehensive Approach to Mental Health, June 2023

The European Parliament Report on Mental Health

Amendment 122

(new) Motion for a resolution Amendment.

Whereas people living with a rare disease are more likely to experience symptoms of a mental health condition (such as low mood, anxiety, emotional exhaustion, and may also at times lead to suicidal thoughts or intention), than the general population.

European Parliament
2019-2024

Subcommittee on Public Health

2023/2074(INI)

28.7.2023

DRAFT REPORT

on mental health
(2023/2074(INI))

Subcommittee on

Rapporteur: Sara

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EUROPEAN PARLIAMENT

Committee on the Environment, Public Health and Food Safety

07/09/2023

Sara Cerdas
Mental health

Motion for a resolution PE751.899 - 2023/2074(INI)

Amendment 33
Susana Solís Pérez, Catherine Amalric, Frédérique Ries

Motion for a resolution
Paragraph 2 b (new)

Motion for a resolution	*Amendment*
	2 b. Underlines the heightened vulnerability to mental health problems in specific groups, namely: children and youth, the elderly, those experiencing dual disorders, people with physical or mental disabilities, individuals experiencing significant loneliness due to insufficient social connections, people living with HIV, displaced individuals including refugees and migrants, LGBTI people, victims of violence, incarcerated individuals, individuals with chronic conditions, rare diseases and those residing in rural or isolated areas. Their increased vulnerability is due to unique challenges faced by each group, which can exacerbate mental health issues;

Or. en

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Amendment 524

52. Stresses that people living with chronic NCDs, which are often characterised by permanent pain or disability, are particularly vulnerable as regards developing mental health conditions; **welcomes the UN's call for the development of effective programmes to promote mental health and psychosocial support for persons living with a rare disease;** calls on the Commission and the Member States to adequately address the impact of NCDs and other chronic diseases and disabilities in policies and programmes on mental health and suicide prevention;

European Recommendations

Call on the European Commission to better prevent and protect mental health problems and reduce further exclusion and marginalisation of the most vulnerable members of society, by recognising the 30M persons living with a rare condition in the European Union as a population living in vulnerable situations and taking action to address these high unmet needs.

SPECIFICALLY

- ✓ Call on the Commission to better protect and prevent mental health problems and reduce further exclusion and marginalisation of the most vulnerable members of society, by recognising the 30M people living with a rare condition in the European Union as a population living in vulnerable situations and taking action to address these severe unmet needs.
- ✓ Tackle stigma and discrimination! Be inclusive of all populations in vulnerable situations (including rare conditions) in the actions to implement the Comprehensive Approach to Mental Health.
- ✓ Translate the implementation of the Comprehensive Approach into a dedicated Flagship Initiative with supporting actions and adequate financial support for all vulnerable groups, including rare conditions and improve access to psychological support.

National Recommendations

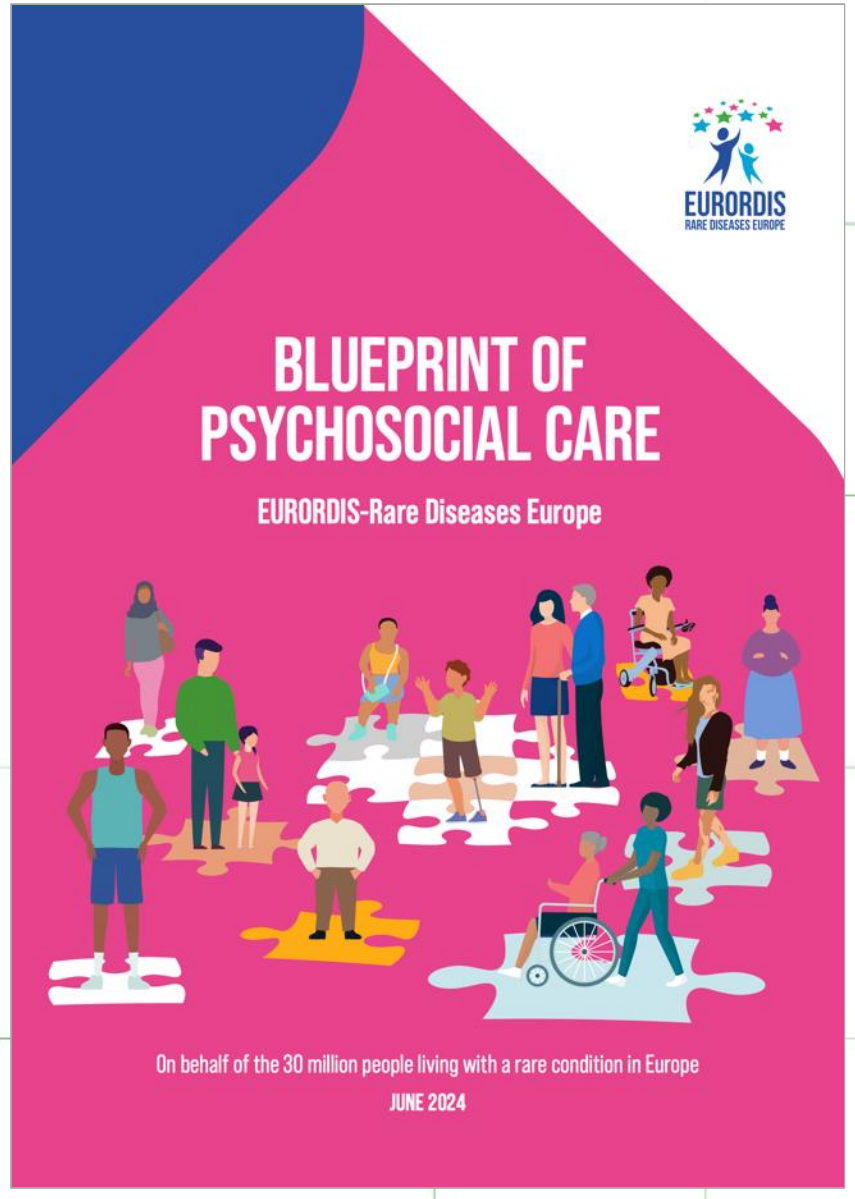
Call on EU Member States to honour the United Nations General Assembly Resolution's (A/RES/76/132) call for the development of effective programmes and national strategies to promote mental health and psychosocial support for persons living with a rare condition, and to coordinate EU action to develop and promote policies and programmes that enhance the wellbeing of their families and caregivers.

SPECIFICALLY

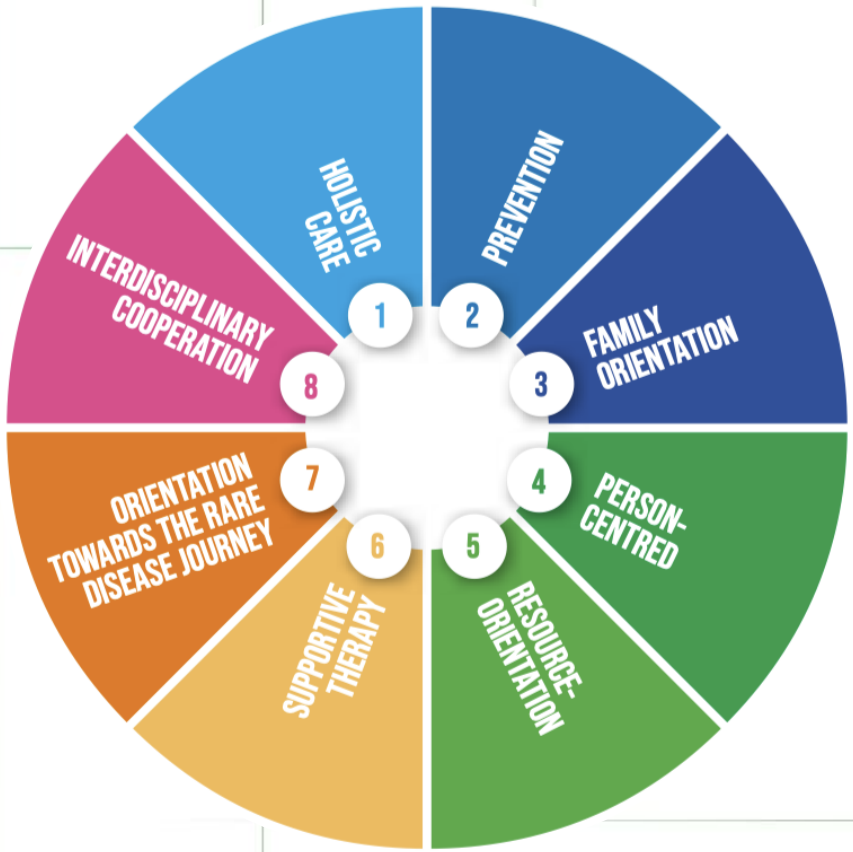
- ✓ Increase and include the visibility of mental health in the revision of Rare Disease National Plans & Strategies and commit to action to address the unmet mental health needs of PLWRD and their families.
- ✓ Integrate psychological support as an integral standard of holistic care, by incorporating psychosocial personnel as core members of the medical team, and through enhancing medical care to be psychologically informed.
- ✓ Recognise and support patient organisations to provide community and peer support, and access to trusted information, as the foundation of psychosocial care, enabling earlier detection and access to preventative support.
- ✓ Support new and targeted medical training to strengthen cross-specialty training, for all professionals to better understand the relationship between physical and mental health, and specifically for mental health practitioners to become more 'rare aware' and medical and nursing care practitioners to become 'mental health' aware.

Blueprint for Psychosocial Care

https://www.eurordis.org/publications/blueprint-for-psychosocial-care/



Domains for Psychosocial Care



Pillar 1: Holistic Care

Pillar 2: Prevention

Pillar 3: Family Orientation

Pillar 4: Person-Centred

Pillar 5: Resource Orientation

Pillar 6: Supportive Therapy

Pillar 7: Orientation towards the Rare Disease Journey

Pillar 8: Interdisciplinary Cooperation

Standards for Psychosocial Care (I)

Pillar 1: Holistic Care

1. The primary aim of psychosocial care is to identify the psychosocial implications of living with a rare disease, which are often complex, chronic, multisystemic conditions, and the impact of treatment (or lack of treatment), which can result in acute and chronic psychological stress and mental health issues for patients, family members and caregivers.

2. A case manager should be appointed to coordinate care and mitigate the stress factors associated with uncoordinated care, consultations and treatments, and provide timely access to psychosocial support.

Pillar 2: Prevention

3. Early preventative support should be aimed at reducing psychosocial risk factors and safeguarding the mental health and wellbeing of the person living with a rare disease and their family. Such support should aim to prevent or reduce social isolation, financial hardship, and the overall uncertainty, stress and anxiety associated with the rare disease journey.

4. Psychosocial care should have a low threshold for accessing psychosocial assessment support and be tailored to meet the needs of the people living with a rare or undiagnosed condition and their families

5. Medical care should include regular and routine assessment and early detection for psychological stressors and include measures to prevent the deterioration of mental health of the person affected by a rare disease as well as all family members.

Standards for Psychosocial Care (II)

Pillar 3: Family Orientation

6. Families of PLWRD are the primary source of support for coping with the rare disease. Assessing the ability of parents and/or the caregiver and support system to cope with the demands and uncertainties of living with a rare disease and providing robust psychosocial support can optimise the resources of the family to provide emotional support, security and protection.

7. Psychosocial care should encompass the provision of timely information, advice and interventions aimed at optimising resilience, coping strategies and stress management as well as reducing the uncertainties experienced by the family.

Pillar 4: Person-Centred

8. Psychosocial support and guidance should be tailored to the specific needs and circumstances of an individual, as well as their family members, informed by the specifications and course of the disease and treatment. At each consultation or intervention, members of the medical team should allow time to enquire how the individual and family are coping with the rare or undiagnosed condition and treatment.

9. Information regarding the disease and treatment should be offered both during and after disclosure of the diagnosis, complementing the information and advice provided by the medical team and signposting for support when needed.

Standards for Psychosocial Care (III)

Pillar 5: Resource Orientation

10. Psychosocial care aims to promote functional coping mechanisms in PLWRD and families, strengthening resistance to psychological stress factors associated with rare or undiagnosed conditions and associated treatments.

11. Psychosocial care should leverage the individual abilities and skills of the individual and their family, promoting personal resources such as self-esteem, self-efficacy, optimism, hope and resilience, to help PWLRD and their families cope with living with a rare or undiagnosed disease.

12. Healthcare professionals should share information with the family on the local support groups and/or referrals to appropriate patient organisations that can provide peer and community support as well as access to trusted information.

Pillar 6: Supportive Therapy

13. Psychosocial care should provide supportive care, oriented towards the course of the disease through the diagnostic process and treatment. Monitoring of psychosocial needs should be a standard part of every consultation and provide advice and support throughout the whole course of the disease.

14. Psychosocial care requires sufficient time during consultations to build trusting relationships with the PLWRD and their family and to optimise support and resources.

Standards for Psychosocial Care (IV)

Pillar 7: Orientation towards the Rare Disease Journey

15. Psychosocial care should start when a rare disease is first suspected, as this can reduce the time to diagnosis; and should continue throughout the entire course of the disease. Wider-family counselling should be offered in cases with a genetic diagnosis.
16. Accurate diagnosis empowers families to strengthen their resources by connecting with a patient community and accessing peer support.
17. Healthcare professionals should be trained to ensure that the presentation of significant, life-changing news is conducted in an appropriate and sensitive manner. Follow-up consultations should be scheduled within a month of a confirmed clinical and/or genetic rare condition diagnosis, with regular follow-up consultations scheduled annually thereafter.

Pillar 8: Interdisciplinary Cooperation

18. Psychosocial care is an integrated component of medical care, requiring close interdisciplinary cooperation with the medical, nursing and social team members.
19. The psychosocial team should be led by a psychologist or clinical psychologist, nurse-specialist, with an educator and social worker, both with an additional psychotherapy qualification, and be supported by a genetic counsellor, music and art therapist, a psychotherapist and a psychiatrist.
20. The medical team members should be trained to provide psychologically informed medical care to detect underlying unmet needs early and to communicate in a sensitive manner; conversely, the psychosocial care team should be trained to be 'rare aware' to understand the common challenges rare diseases pose for affected individuals and their families.

EURORDIS
RARE DISEASES EUROPE

Rare Disease
Research Partners

Daniel, Williams syndrome

Current work

ECRD 2024 No Health Without Mental Health! Let's Co-create a Mentally Healthy Toolkit

Workshop 1: Topic Domains Agreement

6/June

Workshop 3: Community Group Support

31/July

Workshop 5: Advocacy Tools and Approaches

4/September

Jun | **Jul** | **Aug** | **Sep** | **Oct**

17/July

Workshop 2: Self-Care and Support

28/August

Workshop 4: Peer Support

15/October

Workshop 6: Health and Social Care Professional Tools and Approaches

Currently being written, with launch in 2025

RARE BAROMETER SURVEY ON MENTAL HEALTH AND WELLBEING OF PEOPLE LIVING WITH A RARE DISEASE

The survey aims to:

- **Understand the specificities and the unmet needs** of people living with a rare disease regarding mental health and wellbeing.
- Highlight the **increased psychosocial vulnerability of people living with a rare disease** compared to the general population and people living with other chronic diseases.
- **Identify facilitators and barriers** to improve mental health through the rare disease care pathway

WHAT WE KNOW

Rare Barometer & ERNs H-CARE pilot
2020; 3900 respondents

- Emotional and psychological support as part of healthcare experience: one of the lowest scores

- Score was lower for carers than for patients.

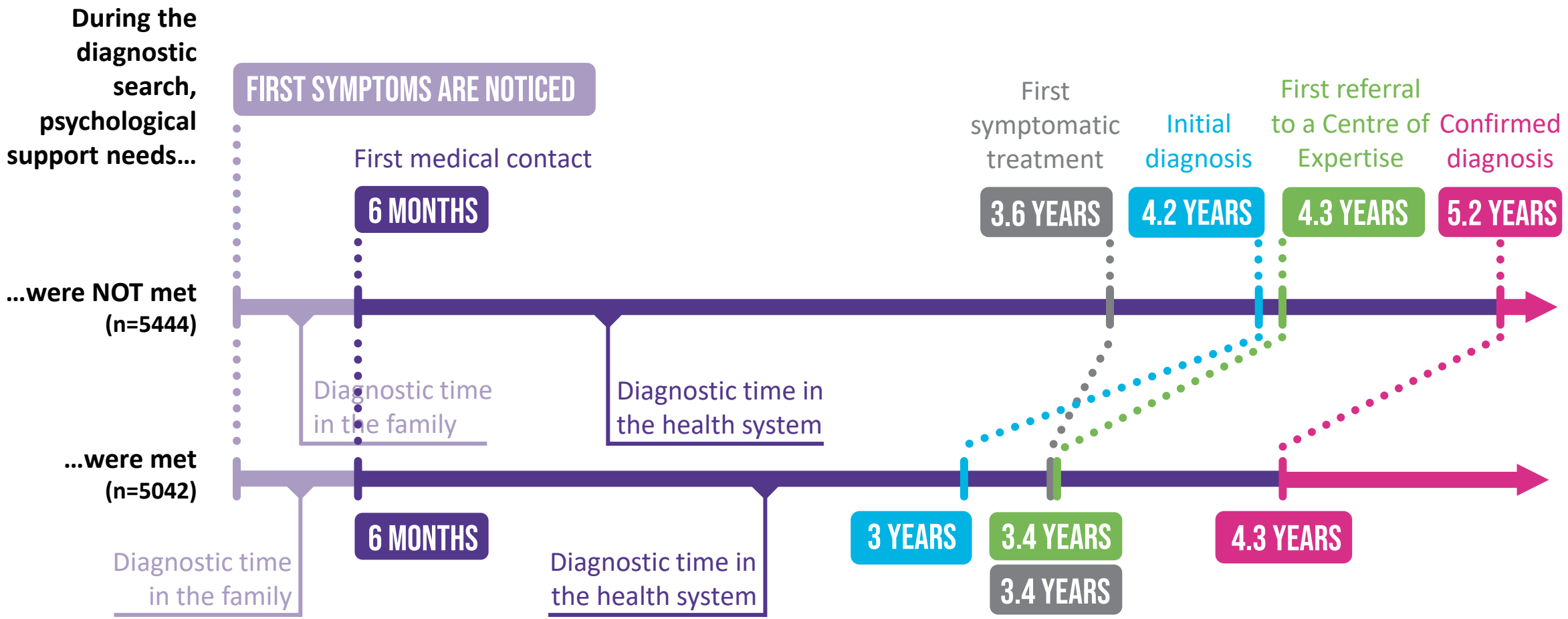
Over the past 6 months, when I/the person I care for received medical care for my/his/her rare or complex disease:			Average score on a scale from 1 (never) to 5 (always)
Questions of the adapted PACIC questionnaire	1	*[I was]* Given choices about treatments to think about	2.8
	2	*[I was]* Satisfied that my / the patient's care was well organized	3.5
	3	*[I was]* Helped to set specific goals to improve my / the patient's eating or exercise	2.6
	4	*[I was]* Given a copy of my / the patient's treatment plan[6]	2.8
	5	*[I was]* Encouraged to go to a specific group or class to help me cope with my / the patient's rare or complex disease	2.1
	6	*[I was]* Asked questions, either directly or on a survey, about my / the patient's health habits	2.8
	7	*[I was]* Helped to make a treatment plan[6] that I / the patient could do in my / his / her daily life	2.5
	8	*[I was]* Helped to plan ahead so I could take care of my / the patient's rare or complex disease even in hard time	2.5
	9	*[I was]* Asked how my / the patient's rare or complex disease affects my / the patient's life	2.8
	9C[7]	*[I was]* Asked how the patient's rare or complex disease affects my *[the carer's]* life	2.2
	10	*[I was]* Contacted after a visit to see how things were going	2.0
	11	*[I was]* Told how my visits with other specialists, like a geneticist or cardiologist, helped my / the patient's treatment	2.5
Additional questions on psychological support	12	*[Healthcare professionals]* Helped me / the patient deal with emotions related to my / his / her health status	2.3
	12C[7]	*[Healthcare professionals]* Helped me *[the carer]* deal with emotions related to the patient's health status	2.2

[5] http://www.eurordis.org/guidelines_hcaresurvey
[6] A treatment plan is a list, made with your care team, of what needs to be done to take care of your health.
[3] Questions 9C and 12C were only asked to carers.

eurordis.org/publications/results-hcare

WHAT WE KNOW

Rare Barometer survey on diagnosis
2022; 10,500 respondents in Europe



nature.com/articles/s41431-024-01604-z

Literature Review on psychological support and psychosocial need in rare conditions

Q1 2024
- Identify Topic(s)
- Map importance & effort

Q2 2024
- Draft Scope
- Conduct review (screening, full text review and data extract)

Q3 2024
- Main findings
- Draft recommendations
- Draft report & feedback

Q4 2024
- Finalise Report

Report due in 2025:

The Importance of Psychological Support for Living Well with a Rare Disease

Podcast

Key Insights

Psychosocial Care

Start where you are, the fact Patient Groups exist is of fundamental importance.

01 Self-care and supporting strategies

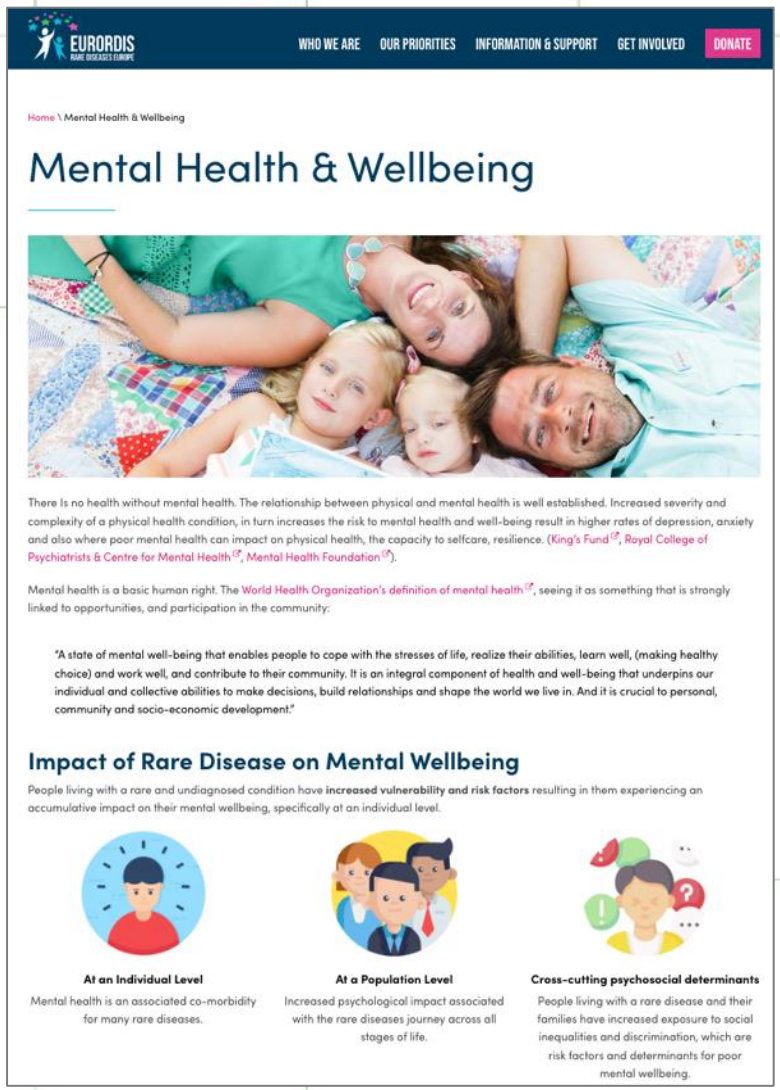
02 Community support & “belonging”

03 Peer support programmes

04 Enhanced medical to psychologically informed rare, aware care.

EURORDIS.ORG

Mental Wellbeing Communication


EURORDIS RARE DISEASES EUROPE

WHO WE ARE | OUR PRIORITIES | INFORMATION & SUPPORT | GET INVOLVED | DONATE

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 selfcare, resilience. (King's Fund, Royal College of Psychiatrists & Centre for Mental Health, Mental Health Foundation).

Mental health is a basic human right. The World Health Organization's definition of mental health, 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 Mental Health & Wellbeing Factsheet(s)


THE SOCIAL IMPACT OF RARE DISEASES ON MENTAL HEALTH & WELL-BEING

August 2016 - February 2017 | 3,071 respondents in Europe | 806 diseases represented | 42 countries

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.

3 times more people living with a rare disease reported being unhappy and depressed than the general population.*

47% people living with a rare disease said that their needs for psychological support were not met.

54% people living with a rare disease declare that the rare disease caused or amplified isolation from friends and family.

7/10 reduced or stopped professional activity due to their own or their family members' disease.

HEAR IT FROM THE RESPONDENTS THEMSELVES

"Simply getting a diagnosis was a fight. The disease has a huge impact on our mobility, our physical health and our mental health."
PATIENT & PARENT OF CHILD WITH RARE DISEASE, UK

"Our child's illness gives no respite. We feel a total loss of carefreeness, constant apprehension of the future and anxiety. We have very little psychological support and it feels like the illness is seperating us from our loved ones. They do not realise the extent of our daily difficulties."
PARENT OF CHILD WITH RARE DISEASE, FRANCE

* Rare Barometer sample compared to International Survey Programme, 2011.

30 million people are living with a rare disease in Europe and 300 million worldwide | No cure for the vast majority of diseases and few treatments available | Thank you to everyone who participated in the survey, and to the Rare Barometer partners!

For more information visit eurordis.org/rare-barometer or email rare.barometer@eurordis.org

Factsheet 1: RBV Juggling Care & Life – Mental Health & Wellbeing

Factsheet 2: Defining the Psychosocial Determinants that affect our Mental Health & Wellbeing


Factsheet #1: Defining the Psychosocial Determinants that affect our Mental Health & Wellbeing

Mental health is a fundamental part of being human. It is expressed in our mood, emotions, thoughts, cognition, control and behaviours. Our mental health is in a constant state of flux, influenced by a range of psychosocial and environmental factors as well as health determinants.

WHO Definition:

Mental health is a state of mental well-being that enables people to cope with the stresses of life, realize their abilities, learn well 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. Mental health is a basic human right.

Mental health is not the presence or absence of mental disorders. It exists on a complex continuum, which is experienced differently from one person to the next, with varying degrees of wellbeing, difficulty and distress, and potentially very different social and clinical outcomes.

Mental health issues include mental disorders and psychosocial disabilities as well as other mental states associated with significant distress and impairment in functioning. People with mental health conditions are more likely to experience lower levels of mental well-being, but this is not always or necessarily the case.

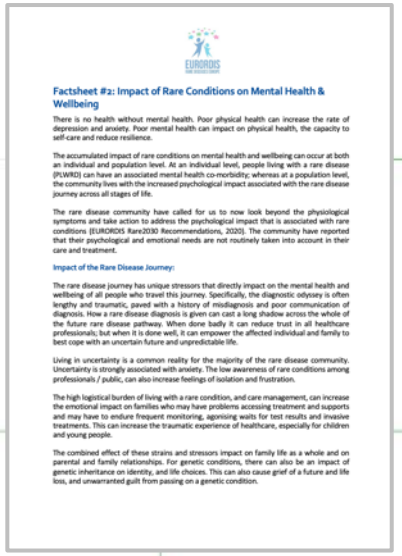
Psychosocial Determinants:

Health, both physical and mental health, can be influenced and affected by different factors, known as 'determinants of health', including economic and social status, education, physical environment, social support networks, gender, genetics and access to and use of health services. These 'psychosocial' factors are both protective in nature and can also pose a risk to poorer health.

Example of psychosocial factors:

Protection Factors	Risk Factors
- Positive norms - Opportunities to pursue interests - Social relationships, community and networks - Good physical health - Physical and psychological safety - Good support system	- Poverty, cost of living and ability to work - Bullying and experiencing prejudice - Lack of supportive relationships, family breakdown, domestic abuse - Injury, infections and health issues - Substance abuse - Lack of social support - Forced displacement and humanitarian emergencies

Factsheet 3: Impact of Rare Conditions on Mental Health & Wellbeing


Factsheet #2: Impact of Rare Conditions on Mental Health & Wellbeing

There is no health without mental health. Poor physical health can increase the rate of depression and anxiety. Poor mental health can impact on physical health, the capacity to self-care and reduce resilience.

The accumulated impact of rare conditions on mental health and wellbeing can occur at both an individual and population level. At an individual level, people living with a rare disease (PLWRD) can have an associated mental health co-morbidity; whereas at a population level, the community lives with the increased psychological impact associated with the rare disease journey across all stages of life.

The rare disease community have called for us to now look beyond the physiological symptoms and take action to address the psychological impact that is associated with rare conditions (EURORDIS Rare2030 Recommendations, 2020). The community have reported that their psychological and emotional needs are not routinely taken into account in their care and treatment.

Impact of the Rare Disease Journey:

The rare disease journey has unique stressors that directly impact on the mental health and wellbeing of all people who travel this journey. Specifically, the diagnostic odyssey is often lengthy and traumatic, paved with a history of misdiagnosis and poor communication of diagnosis. How a rare disease diagnosis is given can cast a long shadow across the whole of the future rare disease pathway. When done badly it can reduce trust in all healthcare professionals; but when it is done well, it can empower the affected individual and family to best cope with an uncertain future and unpredictable life.

Living in uncertainty is a common reality for the majority of the rare disease community. Uncertainty is strongly associated with anxiety. The low awareness of rare conditions among professionals / public, can also increase feelings of isolation and frustration.

The high logistical burden of living with a rare condition, and care management, can increase the emotional impact on families who may have problems accessing treatment and supports and may have to endure frequent monitoring, agonising waits for test results and invasive treatments. This can increase the traumatic experience of healthcare, especially for children and young people.

The combined effect of these strains and stressors impact on family life as a whole and on parental and family relationships. For genetic conditions, there can also be an impact of genetic inheritance on identity, and life choices. This can also cause grief of a future and life loss, and unwarranted guilt from passing on a genetic condition.

Factsheet 4: The accumulated impact of living with multiple intersectional needs.


Factsheet #3: The accumulated impact of living with multiple intersectional needs.

People living with a rare disease and their families live with severe, progressive and chronic conditions and disabilities. Children, young adults and adults of all ages are impacted by rare diseases, with women being the majority among family caregivers. People living with a rare disease and their families frequently find themselves at the intersection of numerous diverse vulnerable situations, including psychosocial risks at work or school, medical uncertainties, economic hardship, discrimination and stigma.

EURORDIS-Rare Barometer Voices Survey on the impact of the COVID-19 pandemic, 2020, showed that:

- 2/3 of respondents suffered from depression and/or a feeling of not being able to overcome their problems since the beginning of the pandemic.
- 6 in 10 found the interruptions to care detrimental to their health or the health of the person they care for. 3 in 10 perceive that these interruptions to care could even probably (21%) or definitely (9%) be life-threatening.

Quote: "The first weeks of COVID I had fears. Suddenly there was no longer much help or contact. Some operations that should have been done have been postponed. I'm really waiting for that. I'm in a lot of pain right now. In addition, I have the feeling that I am on my own." Person living with a rare disease.

People in vulnerable situations

The World Health Organization (WHO) confirmed EURORDIS RBV survey results and also recognized "people with an existing health condition" as one of the main vulnerable groups who were more likely to develop symptoms of mental disorders following the pandemic, along with young people and women (WHO 2022).

The European Commission also highlights the following groups to have higher level of risks to good mental health.

- Children & Young People
- Older People
- Cancer patients and survivors
- People living with disabilities
- Chronic conditions & multi-comorbidities
- Migrant and refugee populations
- Ethnic minorities
- People from lower socio-economic backgrounds
- LGBTIQ+ people
- Women
- Victims of gender-based violence
- Victims of trafficking in human beings
- Victims of crimes
- Roma
- People living in rural or remote areas

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

THANK YOU

Role of genetic counseling in managing the impact of a diagnosis 1.

Sofia Douzgou Houge

European Commission

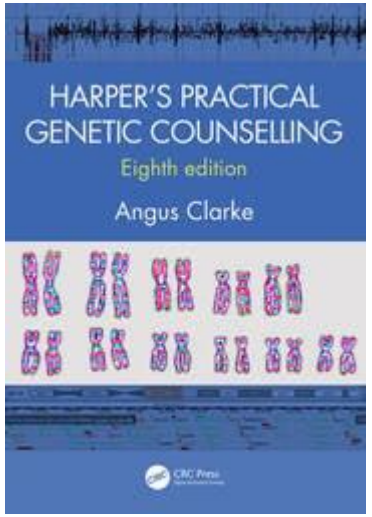
EUROPEAN REFERENCE NETWORKS

Helping patients with rare or low-prevalence complex diseases

European Reference Networks ITHACA

What do we mean by ‘genetic counselling’?

The elements of genetic counselling as practiced are:



i. The initial listening to the questions and concerns of the patient or family and establishing a relationship with appropriate empathy,

ii. Addressing the diagnostic and clinical aspects, including the gathering of information from the patient and family and the checking and documentation of important clinical information about the patient, and also about family members. This may occur in the process of a single consultation or it may become a process that takes many months,

iii. Recognition of the inheritance pattern and risk estimation (when relevant),

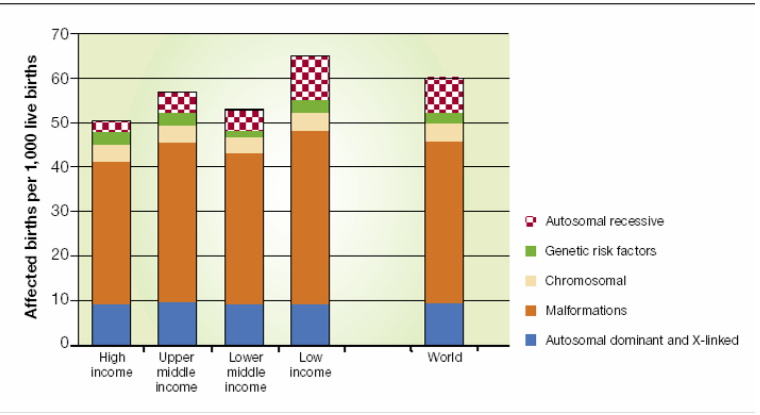
iv. Communication with those being seen and counselled, attempting to answer their questions in light of the facts,

v. Support for the patient and family to understand their situation and adjust to it,

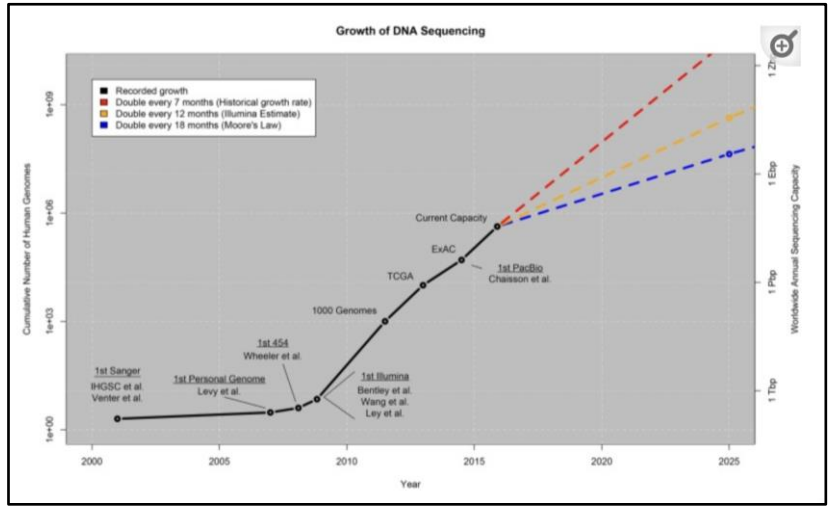
vi. Providing information on available options and further measures, for pursuing the diagnosis (if that remains unclear), for managing the medical aspects of the condition and for questions of reproduction,

vii. Support for the making of decisions and for implementing decisions already made.

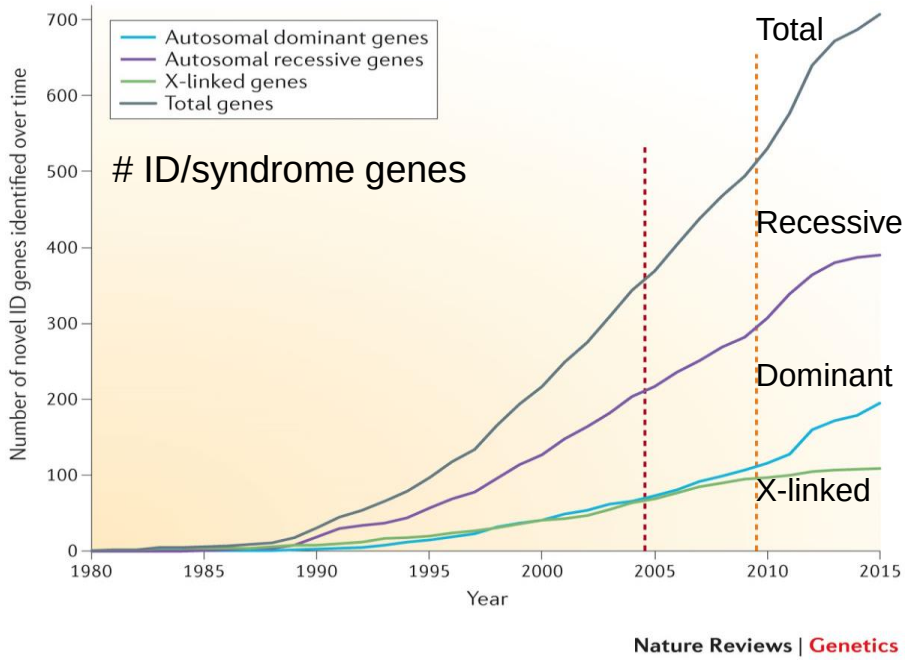
The genotype-first approach that informs medical genetics healthcare services across EU



March of Dimes Global Report on birth defects, 2006



Stephens ZD et al, PLOS Biology 2015

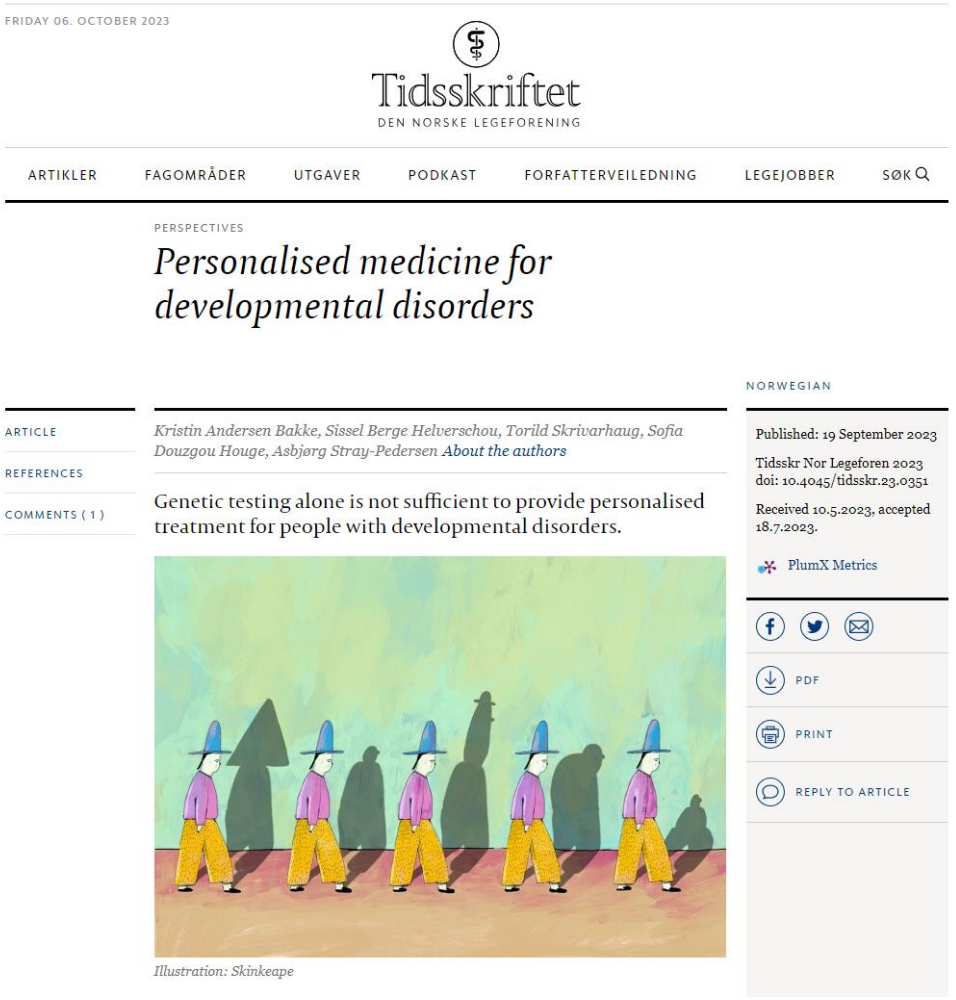


Vissers ELM L, Gilissen G and Veltman JA, 2016

European Reference Networks ITHACA

Impact of genetic testing

- a unifying diagnosis for a patient's features
- a better understanding of prognosis and associated symptoms
- screening, where relevant
- access to condition-specific patient support groups for many rare conditior
- personalised letters for education, allowing access to funding/tailored support
- recommended specific treatments for a small but growing number of genetic conditions
- guidance of symptomatic treatments in a wider number of conditions
- access to participation in research opportunities
- information regarding implications for future pregnancies, including the possibility of reproductive 'options', such as natural pregnancy, adoption, donor gametes, invasive or non-invasive testing in pregnancy, or pre-implantation genetic diagnosis
- access to predictive testing for relatives and an understanding of their chance of being affected
- possibility of future treatments



FRIDAY 06. OCTOBER 2023

Tidsskriftet
DEN NORSKE LEGEFORENING

ARTIKLER FAGOMRÅDER UTGAVER PODKAST FORFATTERVEILEDNING LEGEJOBBER SØK

PERSPECTIVES

Personalised medicine for developmental disorders

NORWEGIAN

ARTICLE
REFERENCES
COMMENTS (1)

Kristin Andersen Bakke, Sissel Berge Helverschou, Torild Skrivarhaug, Sofia Douzgou Houge, Asbjørg Stray-Pedersen About the authors

Genetic testing alone is not sufficient to provide personalised treatment for people with developmental disorders.

Illustration: Skinkeape

Published: 19 September 2023

Tidsskr Nor Legeforen 2023 doi: 10.4045/tidsskr.23.0351

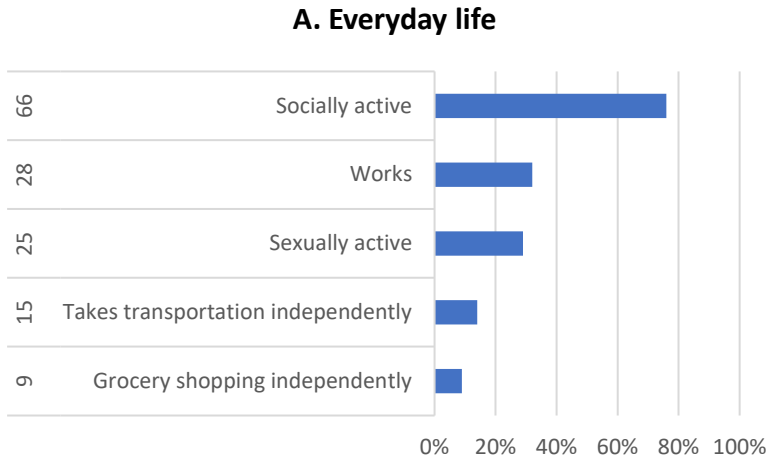
Received 10.5.2023, accepted 18.7.2023.

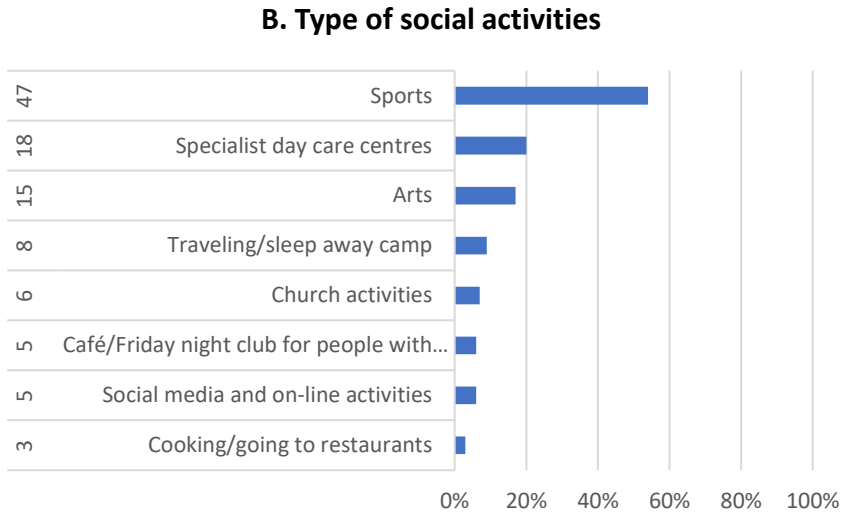
PlumX Metrics

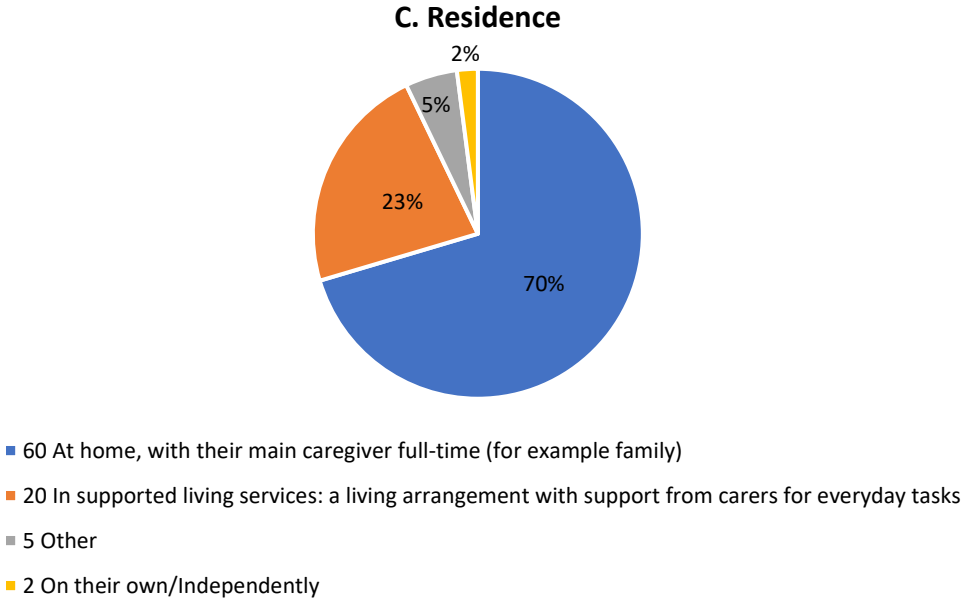
PDF
PRINT
REPLY TO ARTICLE

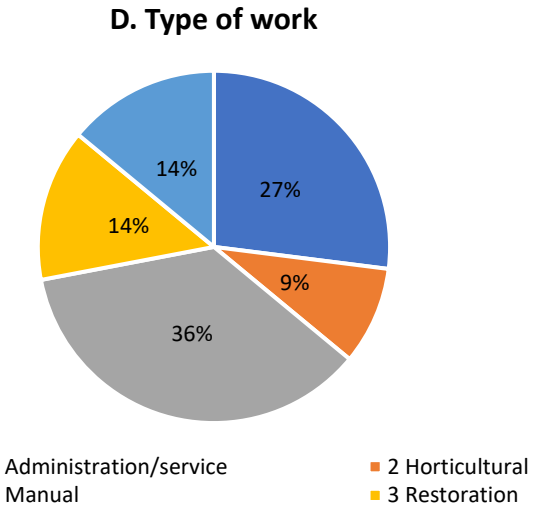
European Reference Networks ITHACA

How to interact with the families ? – rare and diagnosed









The natural history of adults with Rubinstein-Taybi syndrome: a families-reported experience

Douzgou et al., 2022

How to interact with the families ? – rare and diagnosed

Literature curation

Database
- PubMed databases
- Meetings abstracts
- Book chapters

Chronological range
- years 2012-2018
- follow-up, years 2019-2021

Search terms (at least 2)
- MCAP
- Megalencephaly Capillary Malformations
- PIK3CA
- PROS
- CLOVES
- CLAPO
- Macrodactyly
- Klippel-Trenaunay syndrome
- prenatal
- neonatal
- growth
- malformation
- endocrine
- heart/cardiac
- vascular
- bone/limb/joints/musculoskeletal
- skin
- neurological/neurodevelopmental
- autism
- brain
- polymicrogyria
- cognitive

Process For Agreeing and Scoring Recommendations – the AGREE II tool

Agreement Score

+++	>75% agree with the recommendation
++	50-74% agree with the recommendation
+	25-49% agree with the recommendation
-	<25% agree with recommendation

Evidence Score

A	Based on evidence +/- expert consensus
B	Based only on consensus agreement and/or best practice
C	No evidence or consensus agreement/not currently specified as best practice

Expert consensus meeting - 2 day workshop

1st day: Patient-led presentations and discussion

2nd day: Workshop for healthcare professionals

Participation
- 15 clinicians and researchers with expertise in PROS
- 3 representatives of patients, families and advocacy groups
- 6 countries: United Kingdom, Unites States, France, Spain, The Netherlands, Switzerland

Process for agreeing the consensus document

1. A clinical lead for each section presented an outline to address the objectives of the consensus meeting linked to given area of medical management

2. Preliminary recommendations for uniform clinical management guidelines were derived from literature review and prior presentations.

3. The proposed recommendations were presented and further debated and refined.

4. Where there were differences of opinion, these were debated and draft recommendations re-worded.

5. Specific onsiderations were given to:

   a. applicability of the available published evidence to the patient population

   b. how consistent the body of evidence was

   c. how feasible it would be to tailor the recommendations to health and education systems across different countries and settings

   d. the balance of the anticipated benefits and risks of the recommendations.

6. Following the expert consensus meeting, a first draft of the consensus statement was circulated to members of the expert group for review.

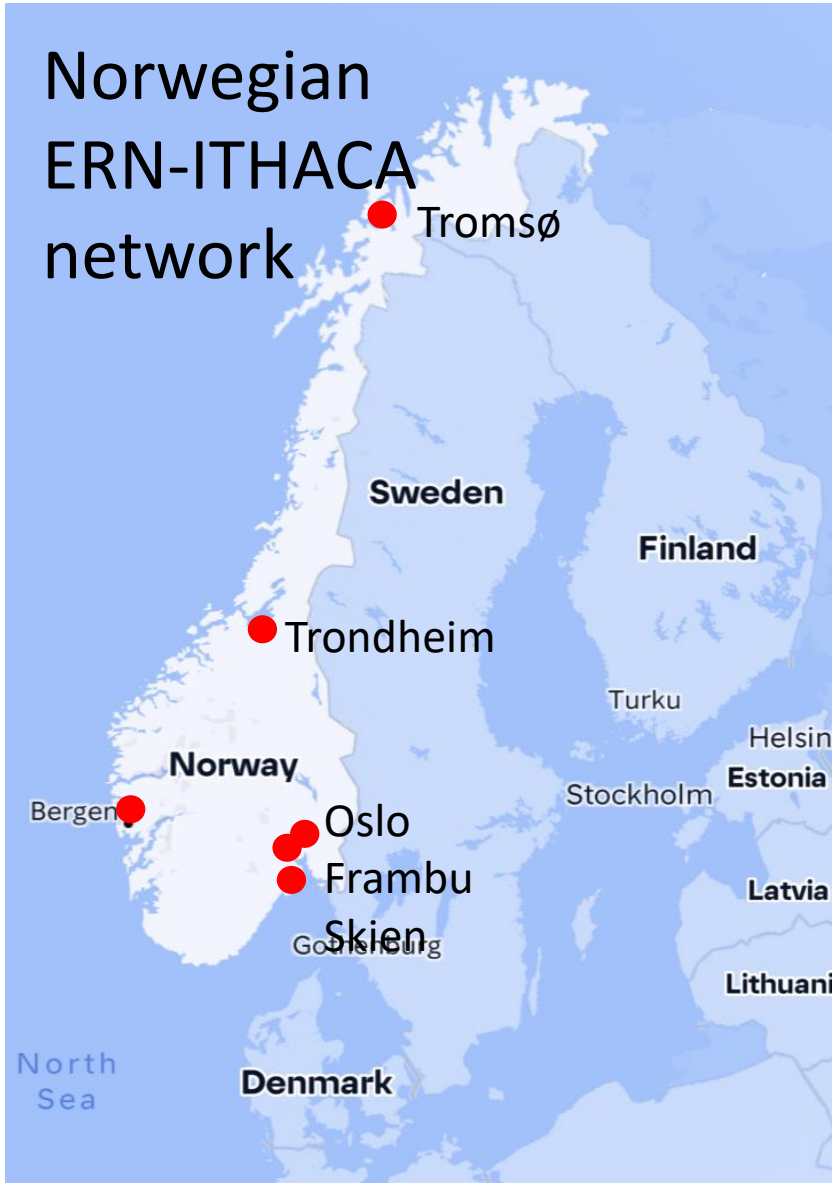
7. 2-years-long electronic correspondence among the experts which led to the agreed final version of the consensus document that is presented here

M-CM NETWORK

A standard of care for individuals with *PIK3CA*-related disorders (PROS): an international expert consensus statement

Douzgou et al., 2021

How to interact with the families ? – rare and undiagnosed



Nasjonal kompetansetjeneste for
SJELDNE DIAGNOSER

Following

NHS
North West
NHS Genomic Laboratory Hub

HELSE BERGEN
Haukeland universitetssjukehus

Norsk helsenett

Genomic technologies

Web-based technologies

Genomic
Syndromology

Healthcare resources

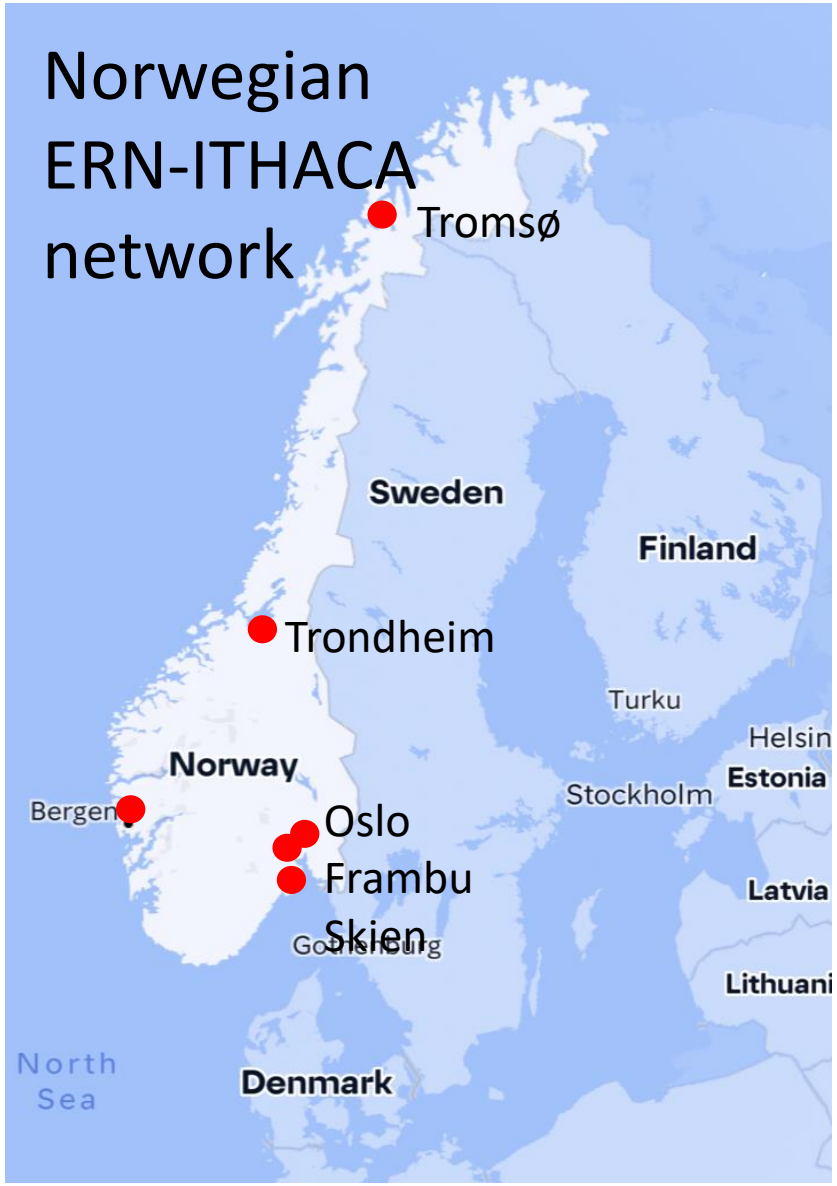
FRAMBU

NevSom

NevSom er Nasjonalt kompetansesenter for nevroutviklingsforstyrrelser og hypersomnier. NevSom har kompetanseansvar for søvnsykdommene narkolepsi, idiopatisk hypersomni og Kleine-Levin syndrom og for nevroutviklingsforstyrrelser ved sjeldne tilstander.

How to interact with the families ? – rare and undiagnosed

Norwegian ERN-ITHACA network



FRAMBU kompetansesenter for sjeldne diagnoser

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Frambu > Nyhetsarkiv > Tilbyr ny genetisk utredning til personer med utviklingshemning uten kjent årsak

Tilbyr ny genetisk utredning til personer med utviklingshemning uten kjent årsak

Publisert: 7. juli 2023

Personer som har fått diagnosen utviklingshemning, men der det ikke er funnet en genetisk årsaksdiagnose, kan nå få tilbud om ny genetisk undersøkelse for å prøve å finne svar på hva utviklingshemningen skyldes ved å bli med i prosjektet: "The post-exome clinic: improving the impact of exome sequencing for developmental disorders in Norway". Prosjektet er et samarbeid mellom Frambu og de genetiske avdelingene i Norge. Tips gjerne personer dette kan være aktuelt for.

Frambu har i mange år hatt et kurstilbud til gruppen utviklingshemning uten kjent årsak. Personer som tilhører denne gruppen tilbys nå en utvidet genetisk undersøkelse.

Er du interessert i å delta i dette prosjektet?

Ta kontakt med Heidi E. Nag på Frambu på telefon 64 85 60 62 for mer informasjon, eller send ditt navn og telefonnummer til hel@frambu.no, så tar h[illegible]

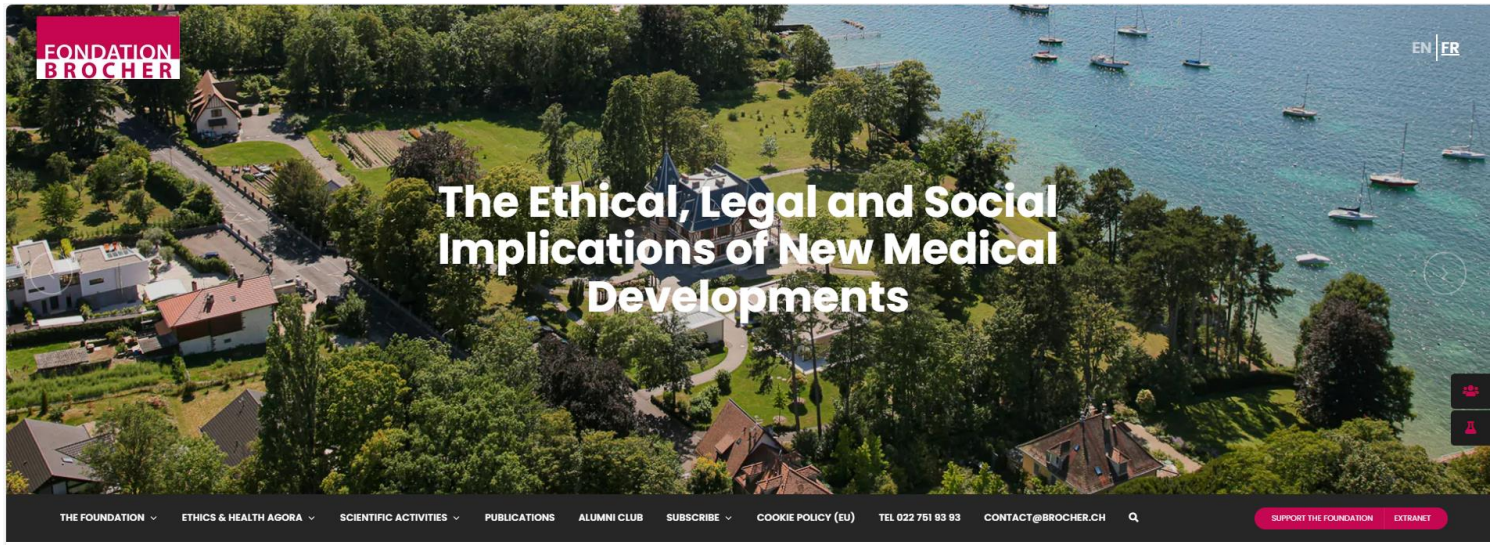
European Reference Networks · ITHACA

What sources of research funds do those who treat rare patients have (in Norway)?

- The "major diseases" are prioritized, and the fact that rare diseases can be biological spotlights for common diseases does not seem to be understood by the committees.
- Hospitals' finances are tight; the opportunity we previously had to be able to conduct "small research" on Departmental budgets has disappeared (even though research is one of the four pillars of the Hospitals Act).
- The rare field unfortunately has few alternatives:
  - no strong patient association with its own research funds behind it;
  - no ad hoc national funding research streams
  - no suitable private funds that can be applied for
  - NKSD (the Norwegian national rare center) and the EU
  - NKSD has modest R&D funds at its disposal (around 7-800 K Euros/year), but they are used well and have been the salvation for many.
  - The EU is an important source, but applying there is probably too demanding for many who are not already part of European research networks.
  - ERDERA is promising, but the problem is that the research is program-driven (in the next round, for example, towards therapy) – free innovative rare research is not supported.

Courtesy of Gunnar Douzgos Houge

How to reach consensus about approaches to common problems ?



The Brocher Foundation in the Service of Bioethics

Rare Disease Research UK.

Ethical Legal and Social Issues in Rare Conditions Research and Clinical Practice - ELSI

Workshop, January 2025 : 'Rare Diseases, Genomics and Justice'

Scientific Advisory Board

Principal Investigator: Ramona Moldovan (U Manchester)
Co Investigator: Anneke Lucassen (U Oxford)
Co Investigator: Angus Clarke (U Cardiff)

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- Thank you !

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Helping patients with rare or low-prevalence complex diseases

ERN ITHACA
Patient Workshop
Role of Genetic Counseling in Managing the Impact of a Diagnosis (2)

Adela Chirita-Emandi

Bucharest

12 December 2024

European Reference Networks ITHACA

Communication & counselling around uncertain findings

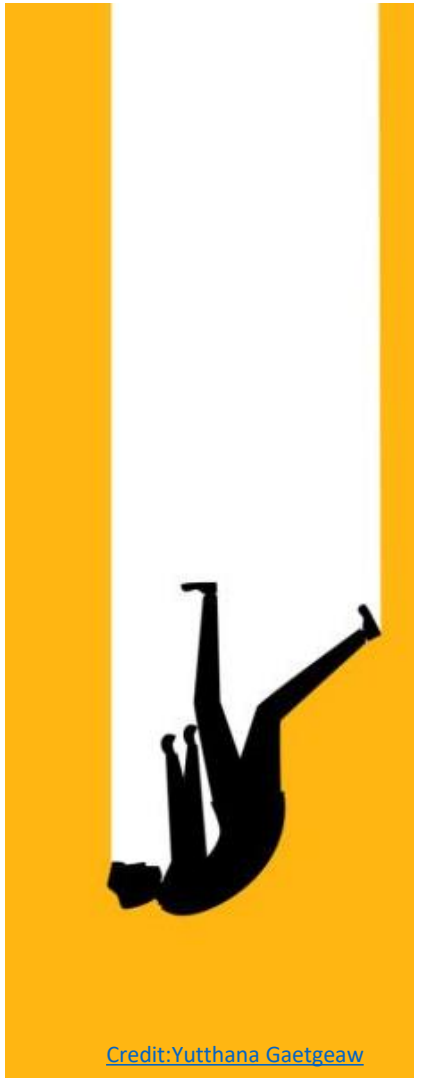
The diagnosis of a rare disease is a difficult milestone for the person and the family

➢The delivering of the diagnosis is a time of great anxiety

fear of isolation, feeling of injustice, fear of the future, culpability, fear of not being able to manage and much more…

➢AND it's also a complicated time for the physician …

➢There is no "good way" to deliver "bad news", HOWEVER a balanced message is very important to keep the faith & hope and to be able to overcome the challenges



Credit:Yutthana Gaetgeaw

Communication & counselling around uncertain findings

Challenges with delivering a diagnosis with uncertainty are many

- How does one deliver a rare diagnosis, with many uncertainties?

➢ *Different shades of rare: rare → ultra rare disease*

➢ *Unknown genotype phenotype relation*

➢ *Reduced penetrance and variable expressivity of genes*

➢ *VUS variant findings that seem to explain the phenotype*

➢ *Findings in genes not yet associated with disease*

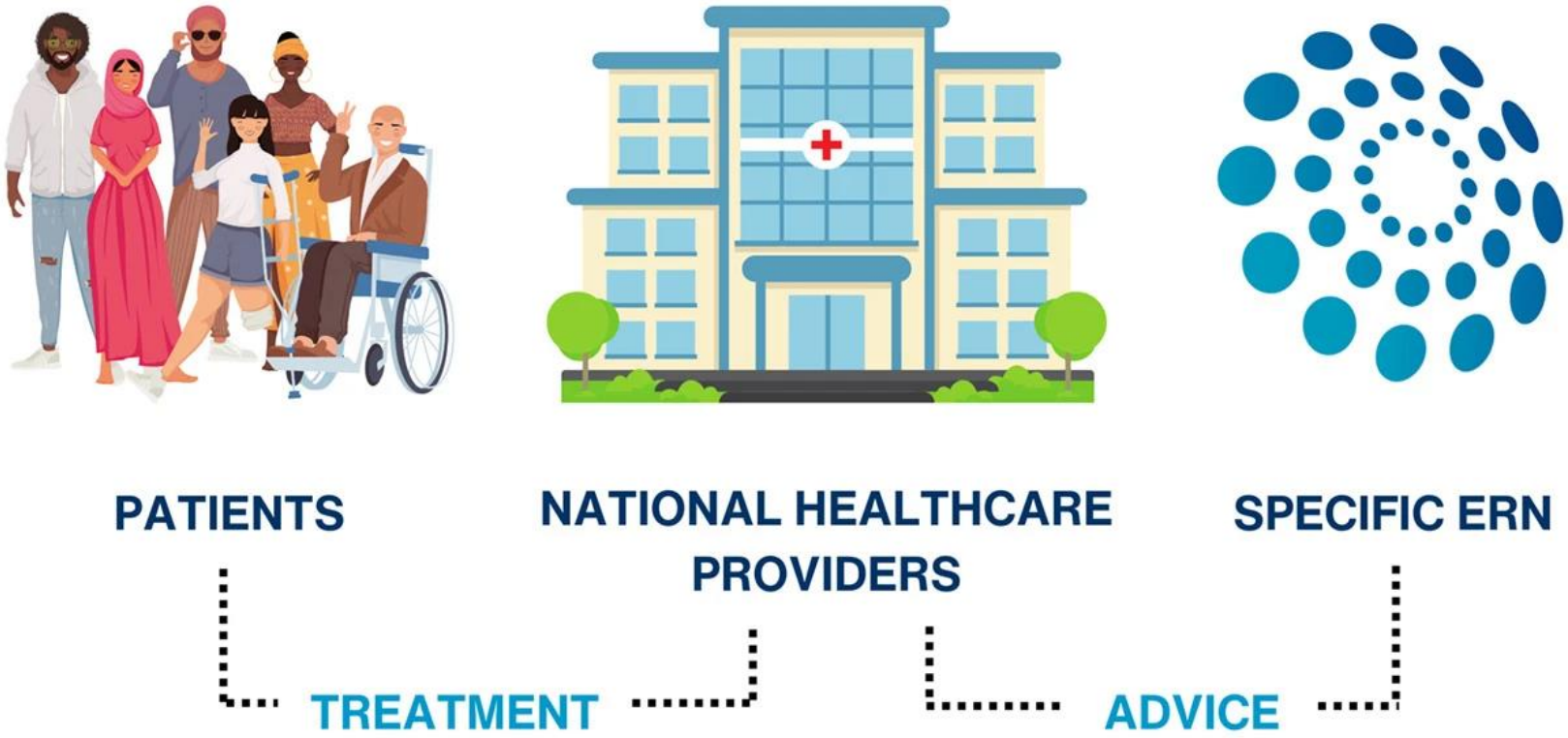
➢ *Negative results, despite suggestive phenotype*

How health professional compensate with not knowing, but still being the expert in the field?

Credit:Yutthana Gaetgeaw

Communication & counselling around uncertain findings

What helps in dealing with diagnosis & uncertainty?



Why rare diseases are posing a massive societal challenge
February 26, 2024, By Jen Tidman

Communication & counselling around uncertain findings

What helps in dealing with diagnosis & uncertainty in rare diseases?

- Diagnosis in Romania – 7 Regional Centers for Medical Genetics in hospitals
- Research in Romania includes some small projects and a big one:

- The University of Medicine and Pharmacy "Carol Davila" in Bucharest created the **Institute for Research and Development in Genomics**.
- ROGEN project has European funding of 85 million euros, through the Health Operational Program, to develop genomic research in Romania.
- Objectives sequencing the entire genome of 10,000 people, clinically healthy, but also sequencing for people with rare diseases (~ 500)
- All medical universities and other research institutions from Romania are involved in ROGEN

Credit:Yutthana Gaetgeaw

Communication & counselling around uncertain findings

What helps in dealing with diagnosis & uncertainty?

- Take responsibility for uncertainty
- Communicate uncertainty –what is known & not known
- The uncertainty gap can cause a trust deficit

Trust deficit can lead to denial of diagnosis, not accepting treatment

- Be transparent in order to fosters trust
- Genetic centers are not just “the point of diagnosis” but a continuous supporting system
- Communicating accurate information (including uncertainties) in a clear, understandable and positive manner at the time of diagnosis is crucial

Communication & counselling around uncertain findings

What helps in dealing with diagnosis & uncertainty?

- Explain how things work, why and what is expected. Have written information.
- Allow time to discuss the information with the family and to find resources. Enable 2’nd or 3’rd visit.
- Help to find the next connecting points in the system and a reference for a Patient Organization.
- Offer connection with other families, if available (language).
- Give back control to people.

A diagnosis does not define who they are!

Communication & counselling around uncertain findings

- Thank you !

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Helping patients with rare or low-prevalence complex diseases

Communicating diagnosis: addressing the needs of patients, families and doctors

Eduardo Tizzano

European Reference Networks ITHACA

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Helping patients with rare or low-prevalence complex diseases

Diagnostic journey of a rare disease patient, a parent perspective

Erika Stariha

European Reference Networks ITHACA

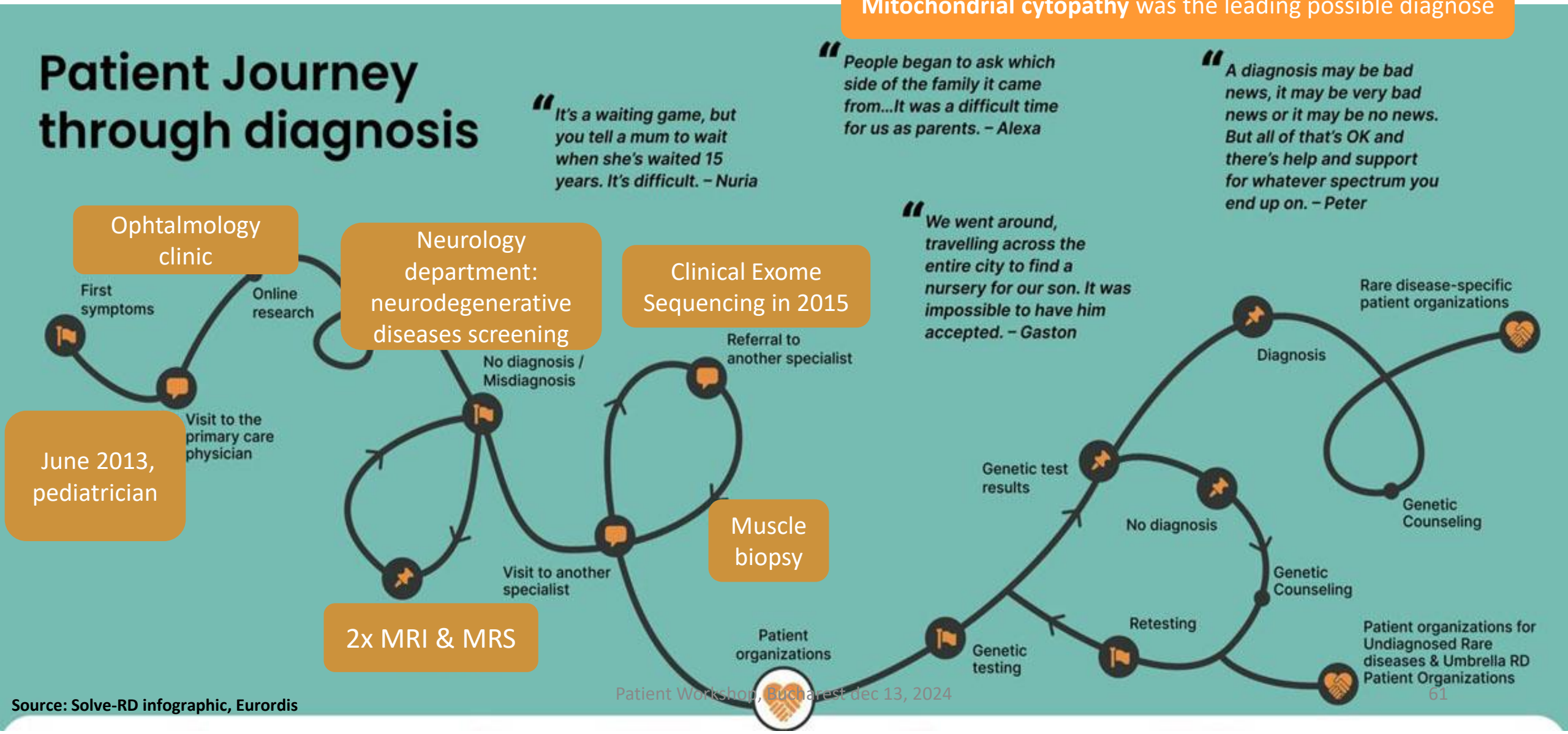
ERN ITHACA
Patient Workshop
Bucharest
12 December 2024

Diagnostic journey of a Rare Disease Patient (parent perspective)

Erika Stariha, Slovenia

Our diagnostic journey started at 3 months of age

Mitochondrial cytopathy was the leading possible diagnose



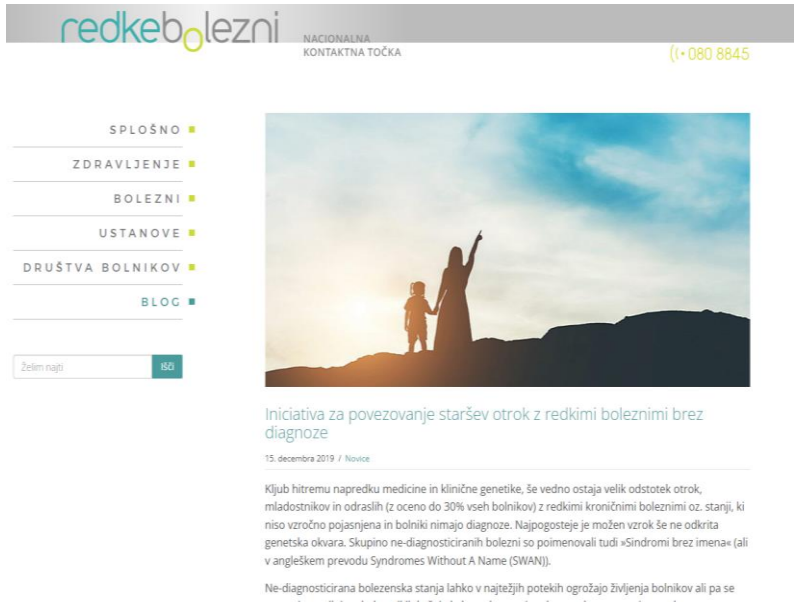
Source: Solve-RD infographic, Eurordis

BEING UNDIAGNOSED

MENTAL BURDEN

- „Why do you search for the label, it will not change anything…“
  „What is wrong with your child?“
- No psychological support
- You **don‘t belong** to any community

I have started an initiative to connect parents of children without a diagnosis in Slovenia



The CHILD and Family BURDEN:

- Guessing the right rehabilitation and medical support for the child
- Future family planning
- Vaccinations with live vaccines?
- Preventive measures cannot be taken
- Access to social and educational rights

SYSTEM GAPS

- Accessibilty to genetic testing early on (to AVOID invasive unnecessary tests)
- Are patients/parents educated on genetic testing?
- Can we implement systematical re-invitation of undiagnosed
  for genetic test? Do we know who „they“ are?

Our diagnostic journey…took 7 years

Patient Journey through diagnosis

"It's a waiting game, but you tell a mum to wait when she's waited 15 years. It's difficult. – Nuria

"People began to ask which side of the family it came from…It was a difficult time for us as parents. – Alexa

"We went around, travelling across the entire city to find a nursery for our son. It was impossible to have him accepted. – Gaston

June 2013, pediatrician

First symptoms

Online research

Visit to the primary care physician

Ophtalmology clinic

Neurology department: neurodegenerative diseases screening

No diagnosis / Misdiagnosis

2x MRI & MRS

Visit to another specialist

Referral to another specialist

Clinical Exome Sequencing (CES) in 2015

Muscle biopsy

Patient organizations

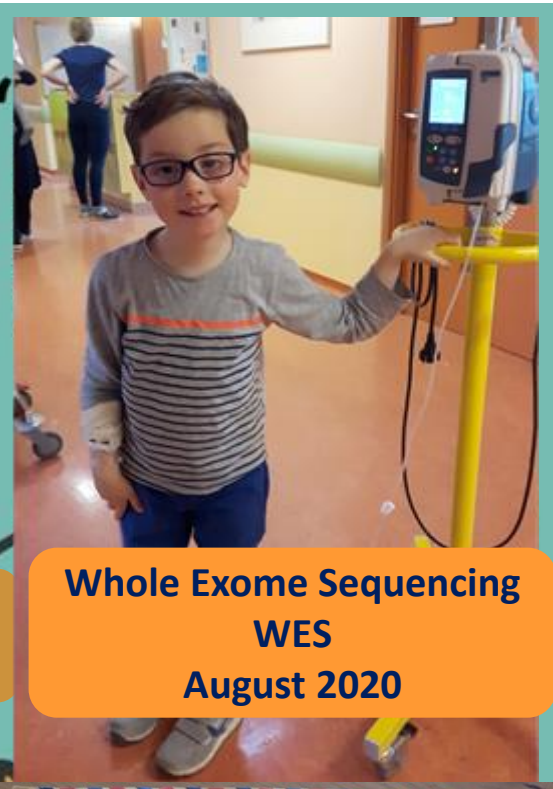
Genetic testing

Second opinion UDN

Lumbal punction

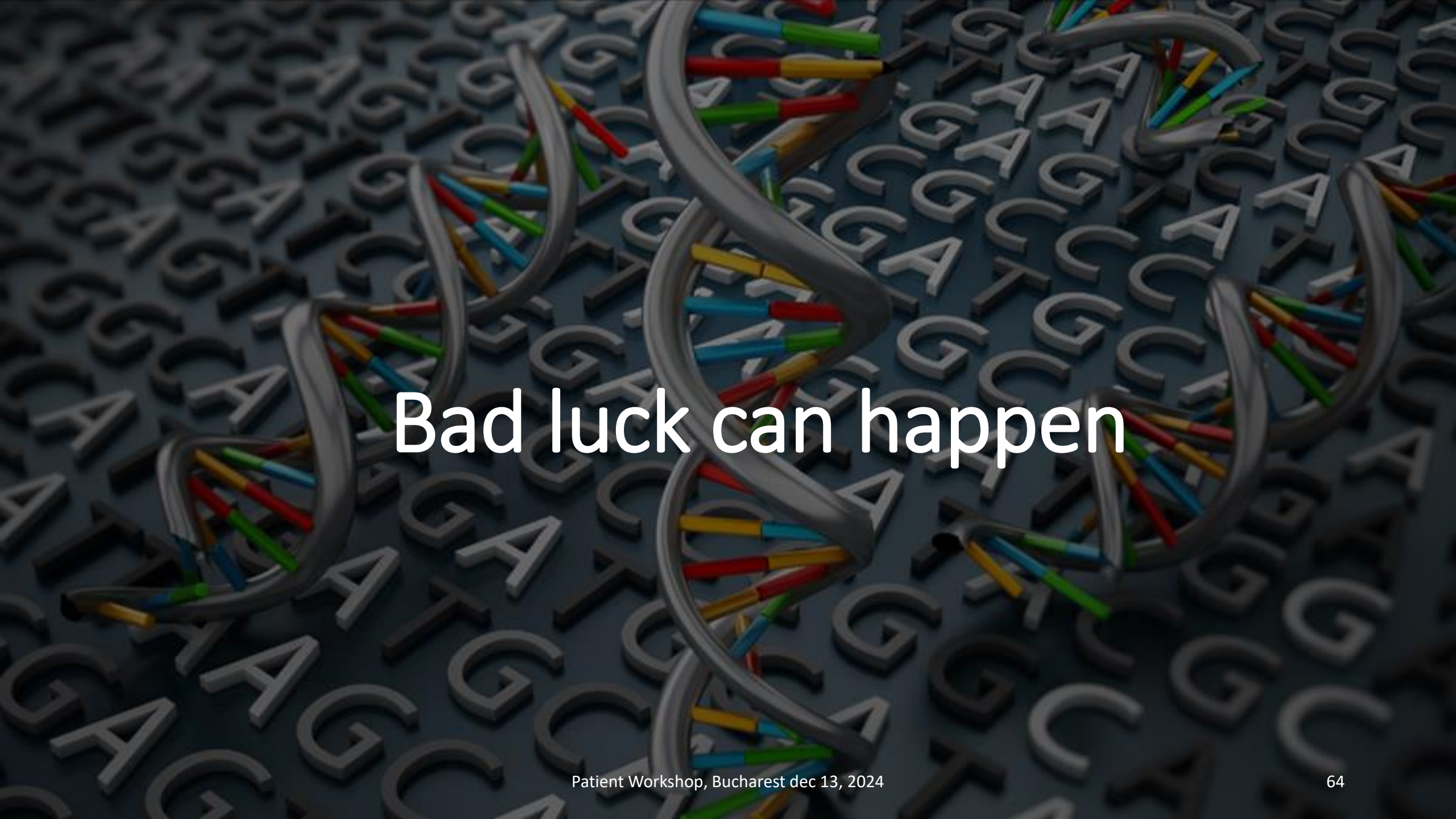
No diagn

Retestin



Whole Exome Sequencing WES August 2020

Source: Solve-RD infographic, Eurordis


Bad luck can happen

Different genetic tests –are we educated as patients/parents?

- **Cromosomal tests** (Karyotype, FISH, CMA) These tests detect **large-scale genetic abnormalities**, such as extra, missing, or rearranged chromosomes.
- **Clinical exome sequencing CES** coding region in a limited set of genes (**3-6.000 genes**)
- **WES Whole exome sequencing** coding region across entire Genome (**20.000+ genes**)
- **WGS Whole genome sequencing** coding and uncoding Genome (3 billion bp)
- **Mitochondrial DNA seq**
- **Protein and Biochemical Testing** (Gaucher disease or Fabry disease)
- **Epigenetic testing**

Useful resource at Global Genes: How to Successfully Navigate the Diagnostic Journey - Global Genes

SATB2-ASSOCIATED SYNDROME

Frequent SAS Characteristics

ABSENT OR LIMITED SPEECH

INTELLECTUAL DISSABILTIY

DEVELOPMENTAL DELAY

CLEFT PALATE

BONE ANOMALIES

BEHAVIOURAL ISSUES

AUTISTIC SPECTRUM DISORDER

UNIQUE SMILE

CONTAGIOUS LAUGHTER

SLEEPING DIFFICULTIES

DENTAL ANOMALIES

EPILEPSY

FEEDING ISSUES

JOVIAL PERSONALITY

GROSS & FINE MOTOR DIFFICULTIES

Speech

Abnormal palate

Teeth

Behaviour, Bone, Brain

2nd chromosome

Around 800 known cases in the world, half in Europe, 4 in Slovenia so far

The diagnosis brought a **new chapter** for our family!

We are creating new, easier paths for next generations!

Patient advisory board feedback on communicating diagnosis, discussion -

Ellen Koekoeckx

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Helping patients with rare or low-prevalence complex diseases

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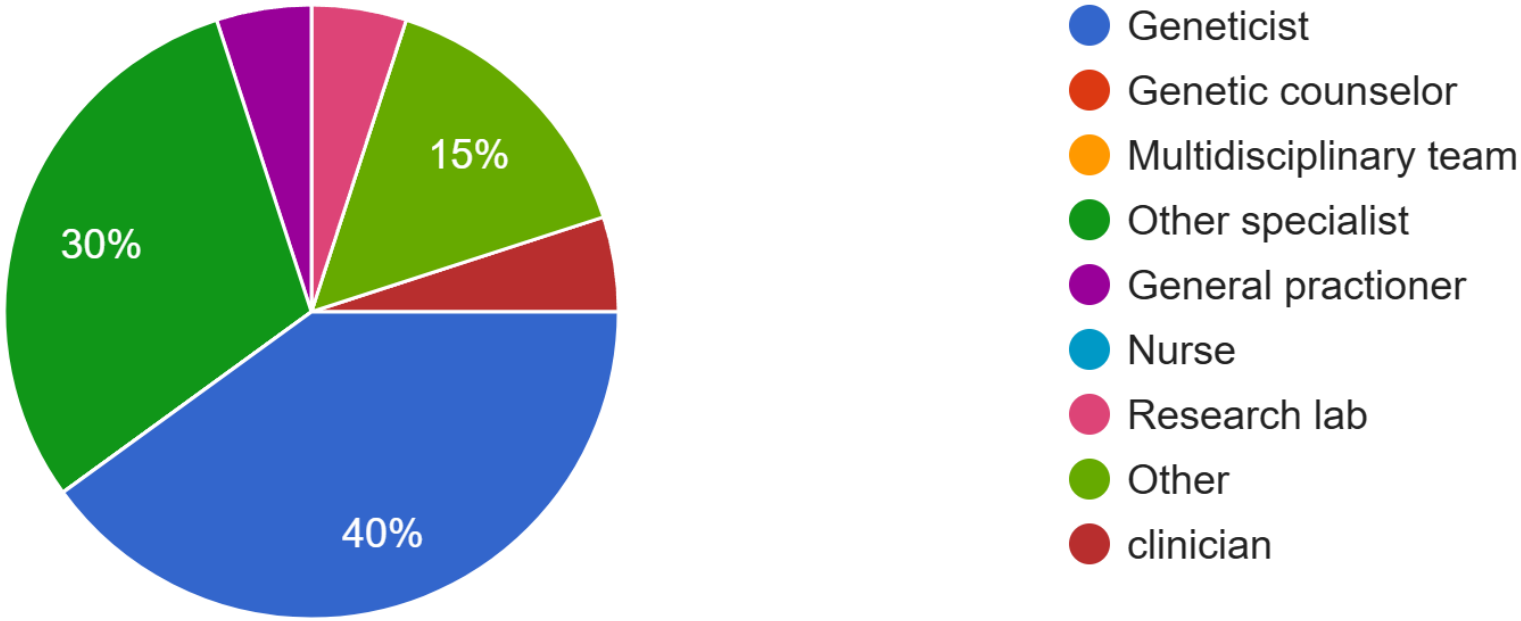
PAB Feedback on communicating diagnosis

Who delivered the diagnosis?

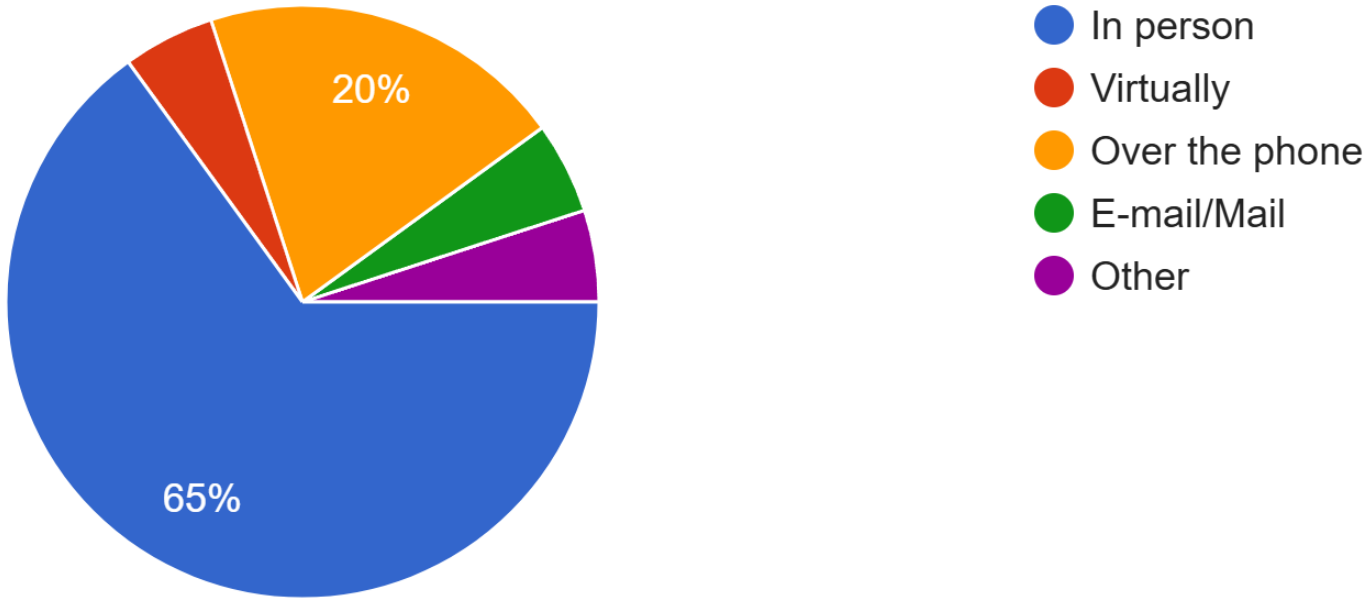
20 responses



PAB Feedback on communicating diagnosis

How was your diagnosis delivered ?

20 responses



PAB Feedback on communicating diagnosis

Was the diagnosis explained in a way that was easy for you to understand?

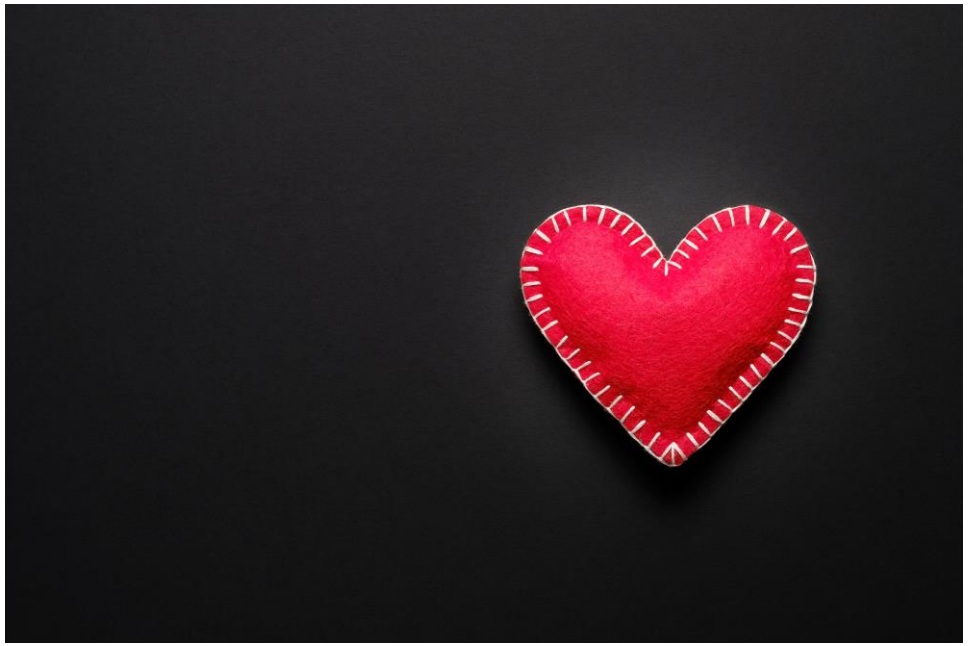
20 responses

PAB Feedback on communicating diagnosis

Did you feel you had enough time to ask all your questions and express your feelings?

20 responses

What part of the conversation made you feel supported?



- Kindness/empathy
  Contact details for follow-up
- Level of information provided
- Sufficient time

What part of the conversation made you feel alone or confused?

- No empathy
- Over the phone
- Use of difficult medical terms

- No information about the disorder (in my language)
- Lack of information on patient organizations/connect with other families
- No clear path forward –best way to support your child

- Not enough time
- Too shocked to ask questions at first visit

Did you benefit from any resources following the diagnosis?

Referral to patient organization or other families

Medical publication

What advice would you give someone delivering a diagnosis?

- Multidisciplinary team
- Psychological support
- In person !!

- Empathy
- Clear language without euphemisms
- Realistic but hopeful
- Share positive examples

- Sufficient time
- Follow-up visit

- More detailed information
- Read into minimum background info

- Connection to patient organizations, families and FB groups **nationally and internationally**

- Referrals to centers of expertise
- Referrals to therapists
- Plan preventative examinations
- Referrals to support services

- Information about ongoing research and clinical trial options

- Thank you !

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Session 2: Parallel sessions

Group 1

Identifying and mapping resources to boost family resilience

Group 2

Different faces of grief

Parallel sessions Group 1
Identifying and mapping resources to boost family resilience

Identifying & mapping resources to boost family resilience

Introduction : Ammi Andersson

- A stressful situation that can lead to physical and/or psychological illness

- Needs for special care
- Many HCP contacts
- Insufficient sleep
- Bigger responsibility and demands “Its all up to me“
- Attitudes from individuals and authorities
- Expected to take greater responsibility
- Coordinate contacts “the profession thinks they are the only contact and doesn´t understand why you sometimes have to cancel.”
- Unemployment or on sick leave

Identifying and mapping resources to boost family resilience

Family Resilience	Protecting supportive relationships?	Reducing isolation and connecting network?
What can I do to be able to cope having a family member with a rare disease?	What can I do to protect relationships?	What can I do to reduce an isolation?
What can a clinician do to make it easier to cope?		What can a HCP do to reduce isolation?
What can the community and municipality do to make it easier to cope?		What can a community or municipality do?

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Helping patients with rare or low-prevalence complex diseases

European Reference Network for rare or low prevalence complex diseases

ITHACA – Network Intellectual Disability and Congenital Malformations (ERN ITHACA)

Identifying and mapping resources to boost family resilience
- medical perspective -

IOANA STREATA, DORICA DAN, MARIA PUIU

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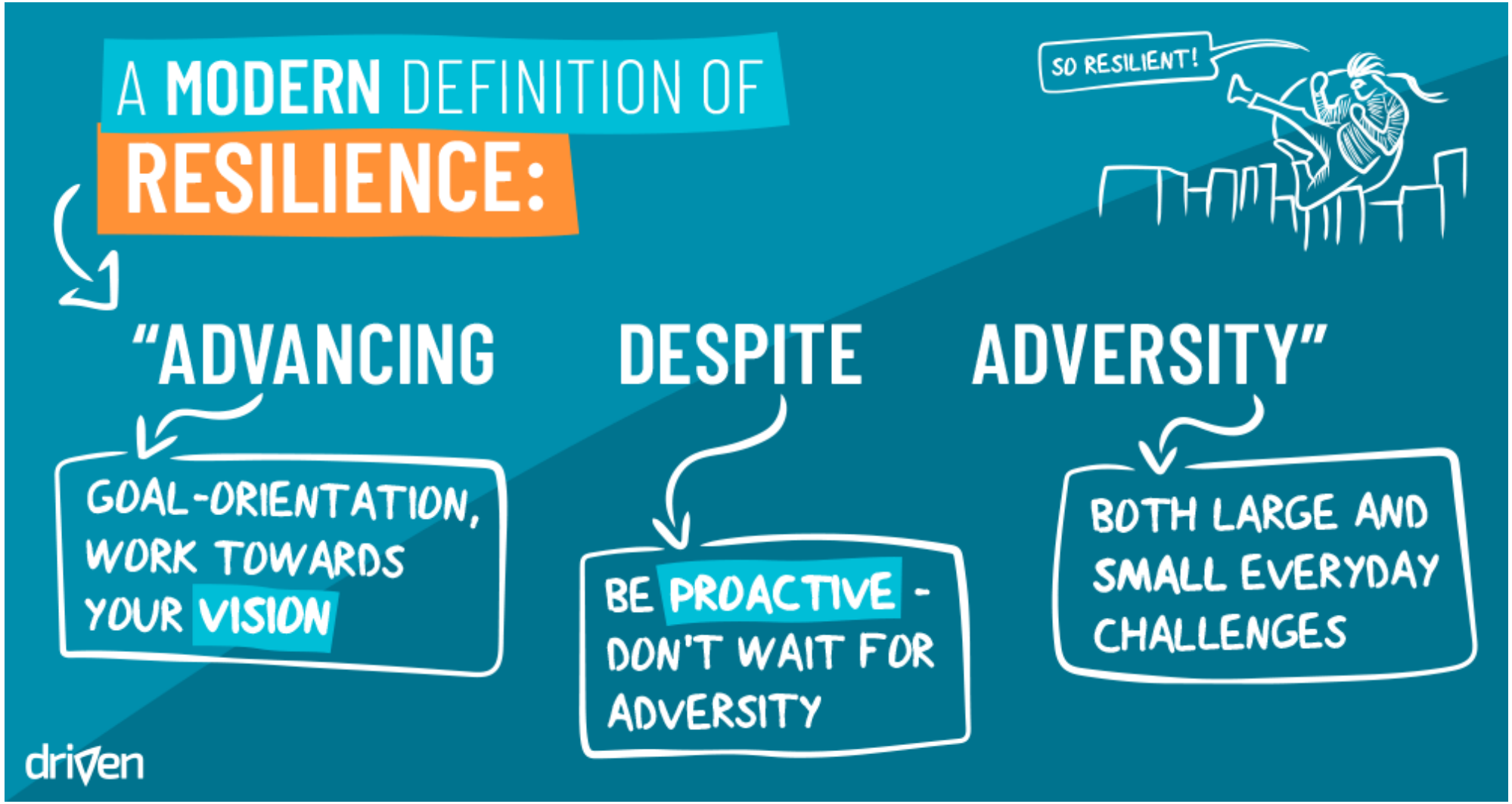
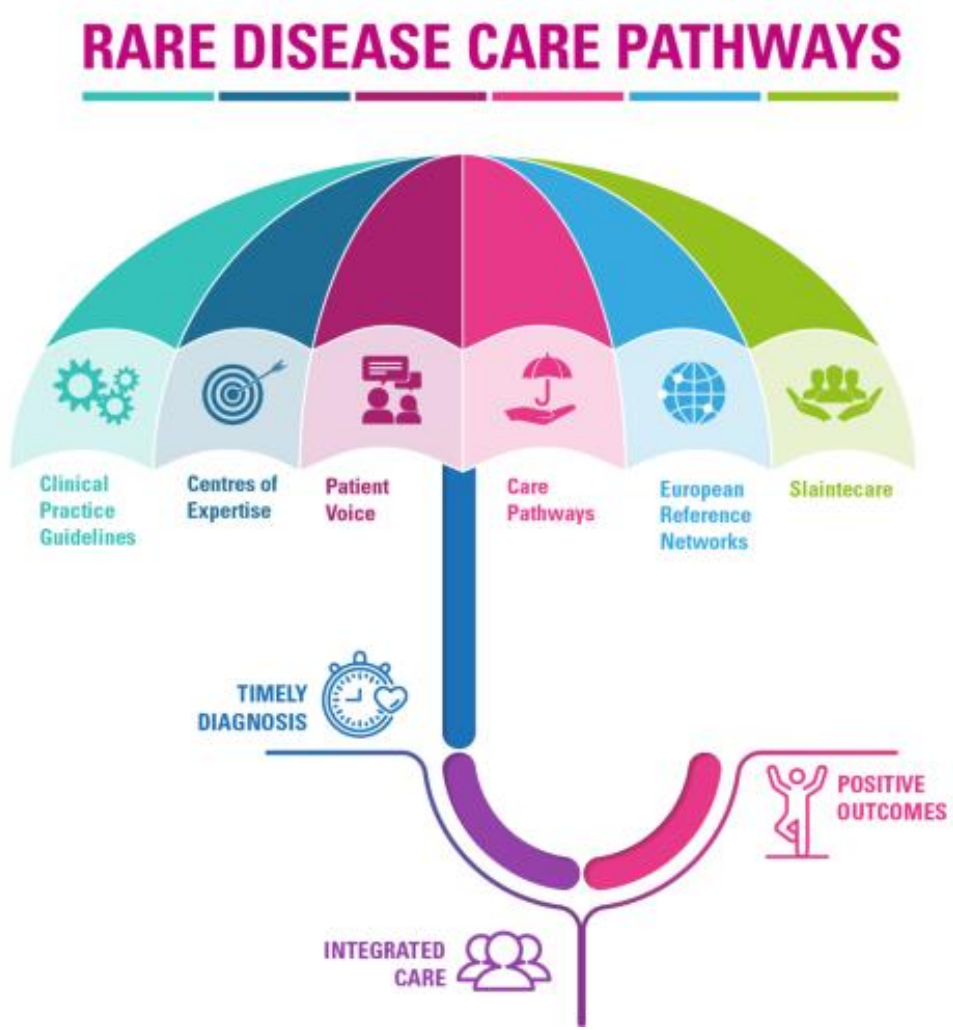
RESILIENCE DEFINITION



RESILIENCE REQUIREMENTS

RARE DISEASES

NRDO
National Rare Diseases Office



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HEALTHCARE IN RARE DISEASES

NRDO
National Rare Diseases Office

RARE DISEASE CARE PATHWAYS

TIMELY DIAGNOSIS

INTEGRATED CARE

POSITIVE OUTCOMES

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HEALTH CARE SYSTEMS RESILIENCE

Characteristics/Capabilities

Dimension 5: Whereby?
Key management functions/capabilities and individual workforce characteristics/ capabilities contribute to the generation of resilience capability at the system level (e.g., Bea and Haas 2019, Windle 2011). This is based on acknowledging the interconnectedness of the system components.

Dynamic Resilience Paths

Dimension 4: How?
Absorptive, adaptive and transformative resilience paths are characterised by phase-specific core capabilities (Conz and Magnani 2020, Blanchet et al. 2017). Continuous adjustment and implementation of these core capabilities pave the way for dynamically building resilience capability on the system level.

Intertemporal Phases

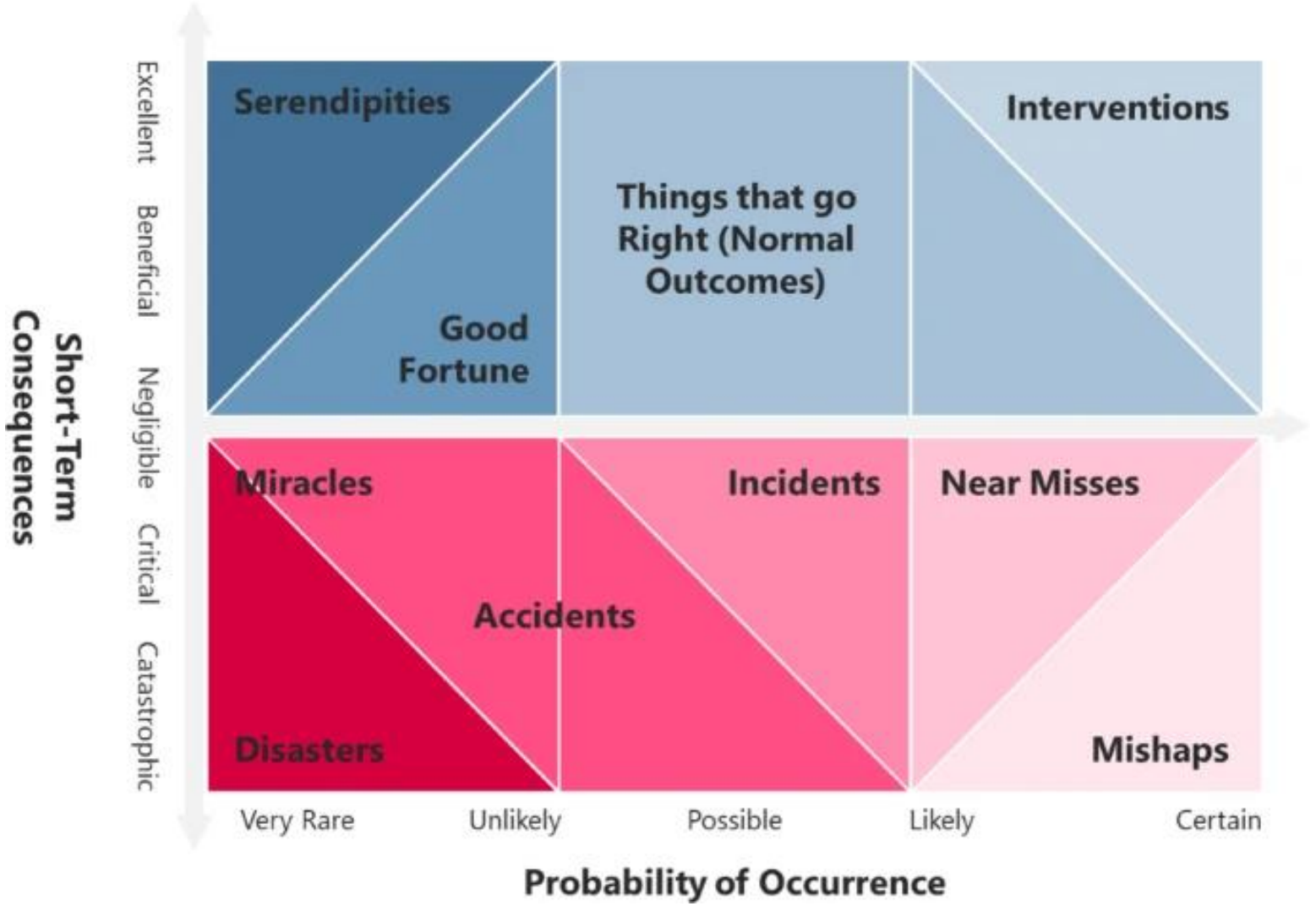
Dimension 3: When?
Resilience is discussed as a dynamic attribute of a system and refers to phases before/during/after an exogenous event (Conz and Magnani 2011).

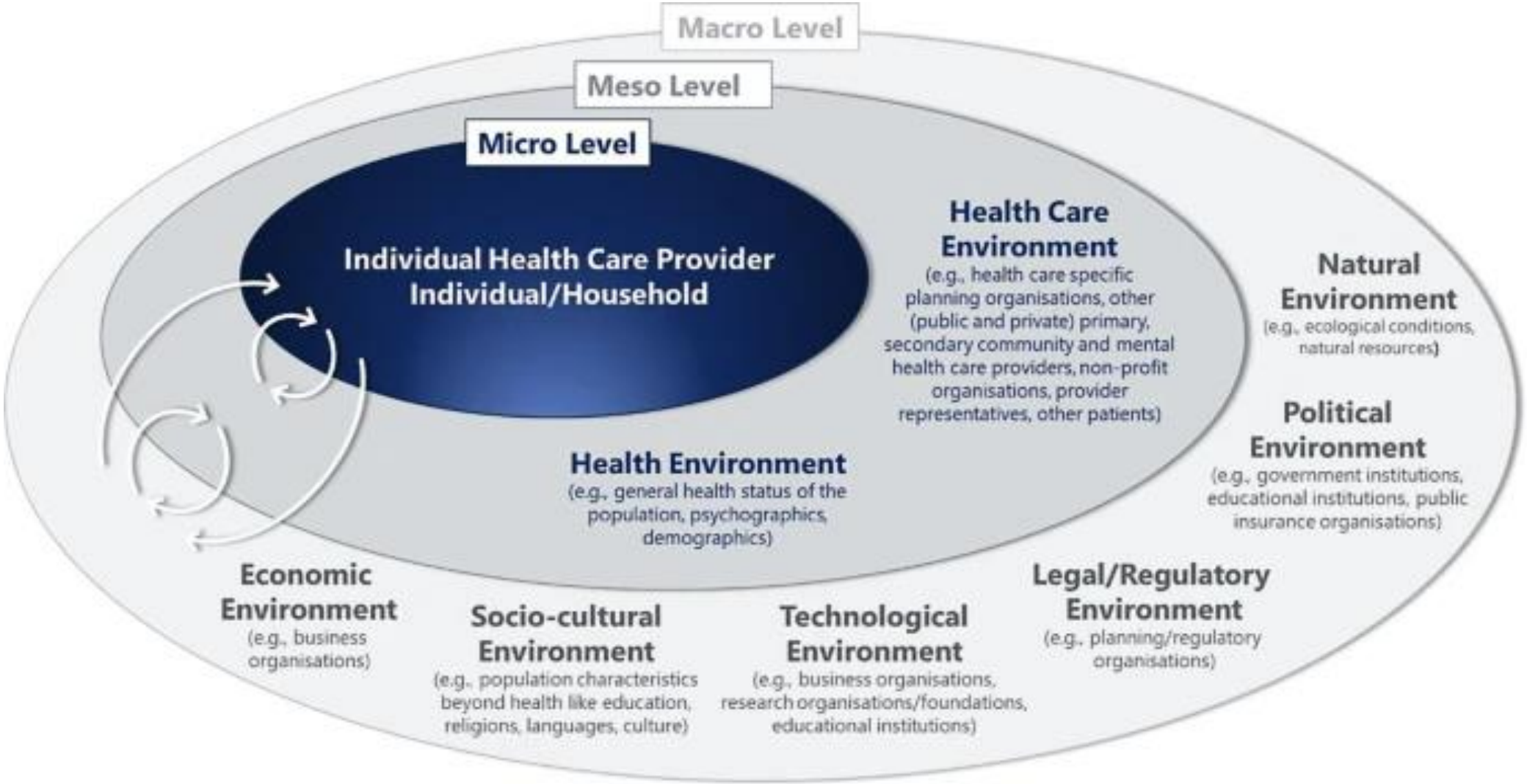
Type/Probability of Occurence/ Consequences of an Event

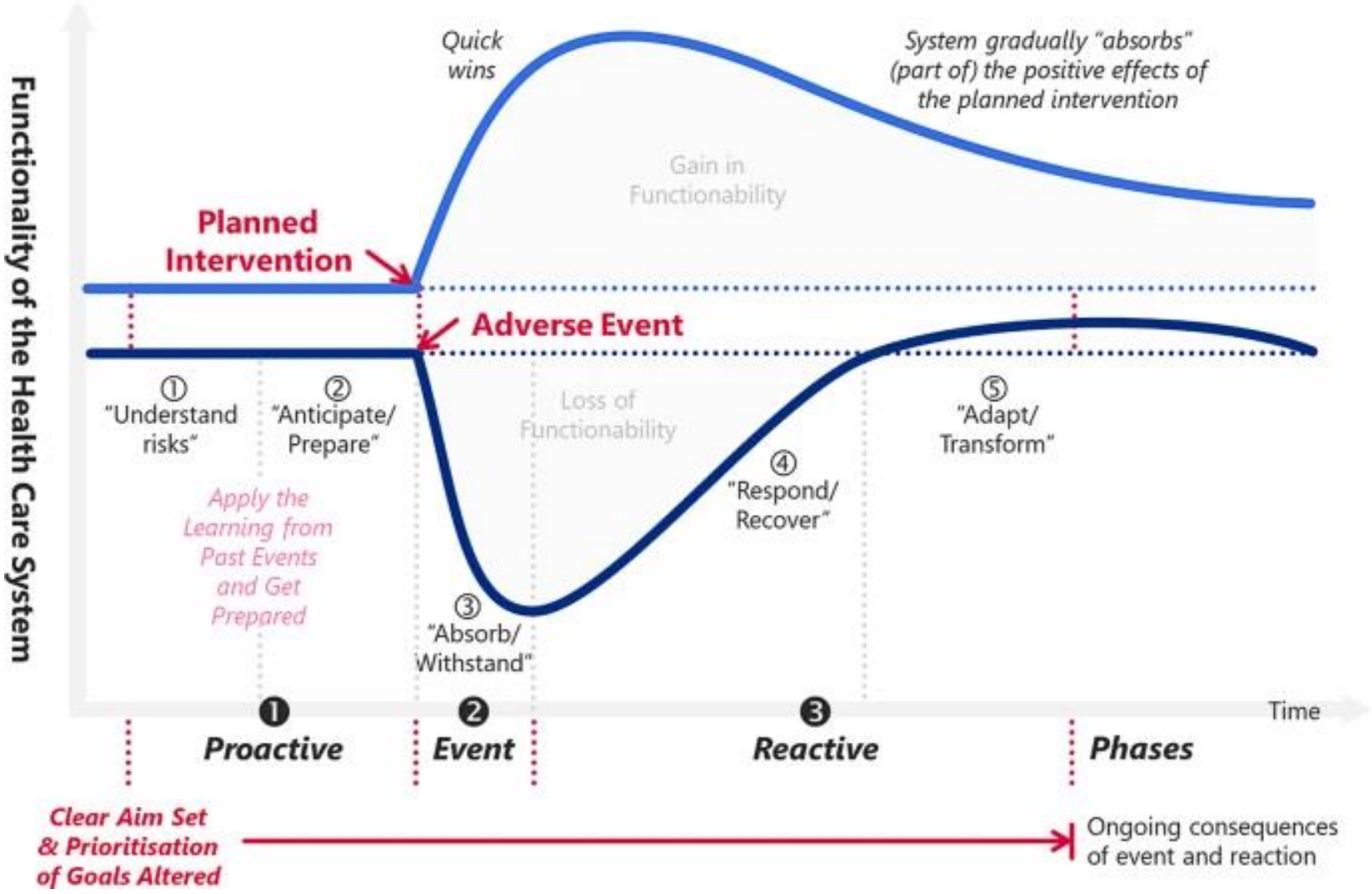
Dimension 1: What?
Exogenous events can be likely or unlikely; adverse, neutral or beneficial (Hollnagel, 2011); anything from an unforeseen shock (e.g., adverse event) to a lasting change (e.g., planned intervention). The long-run consequences characterise the system response by yielding either dysfunction, survival, return to the pre-event state or return with a growth (Carver 1998, Patterson and Kelleher 2005). Both the discussion of short- and long-term consequences contribute to the understanding of resilience.

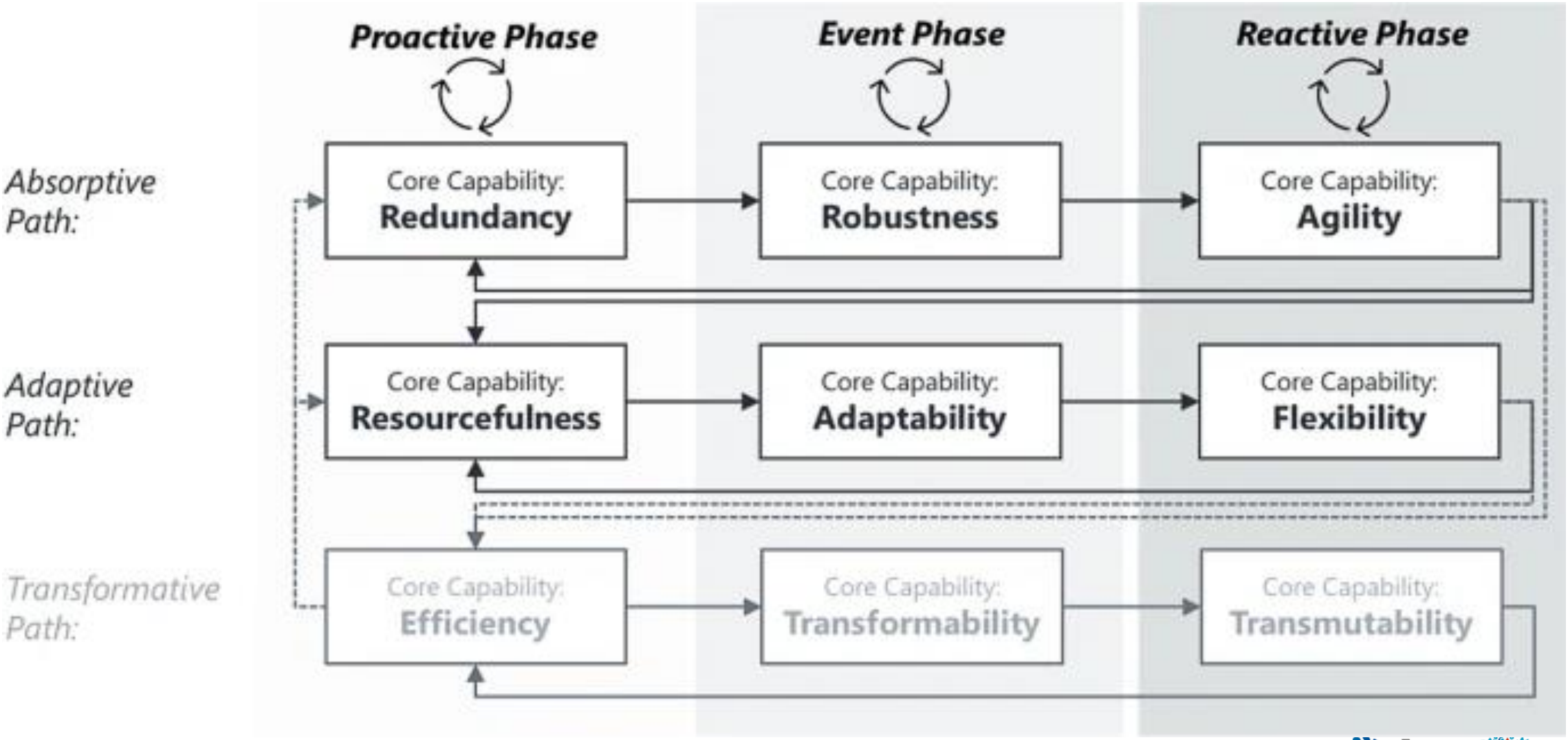
Objects/Subjects

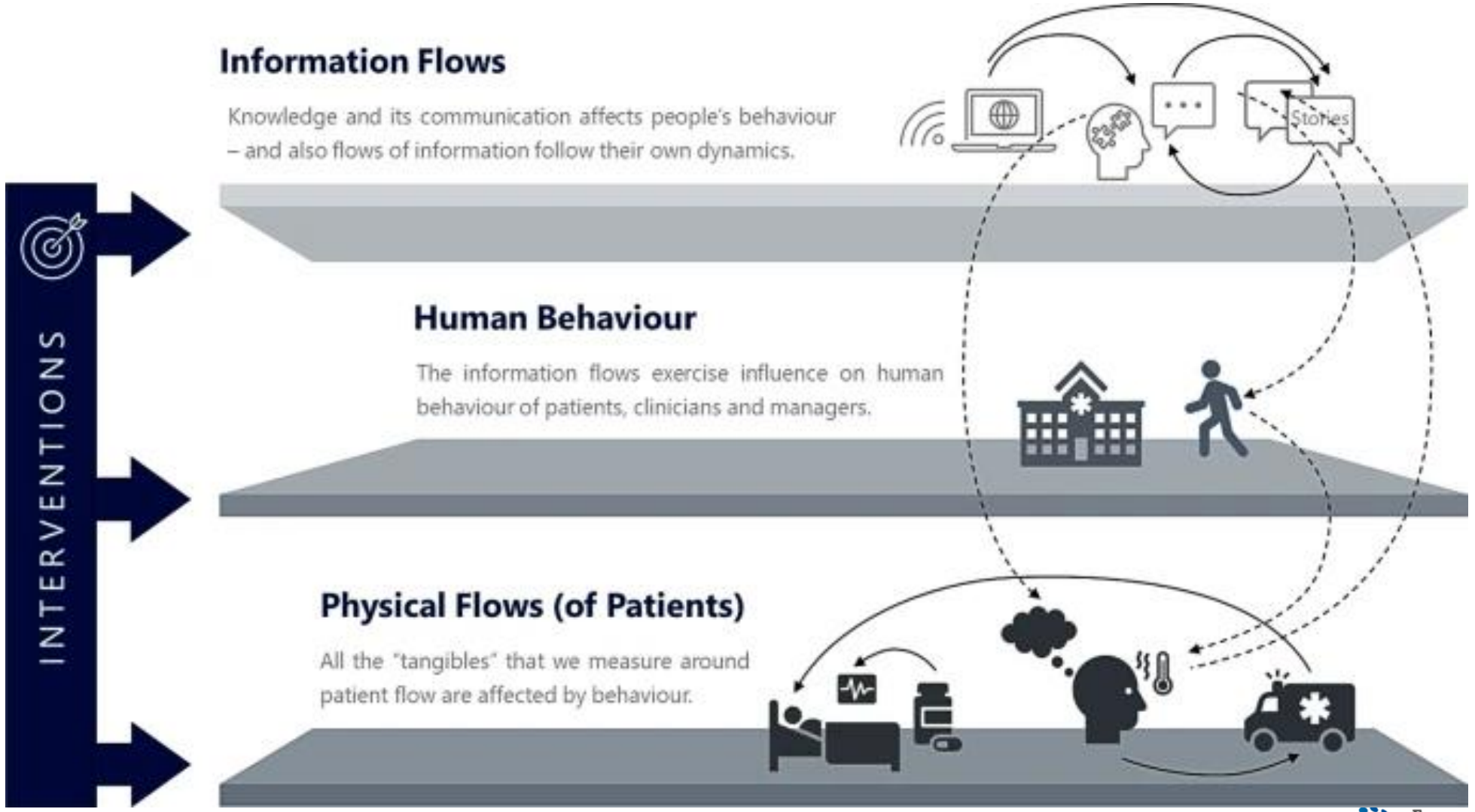
Dimension 2: Who?
Stakeholders interact in subsystems creating interconnected environments – across the boundaries of the macro, meso and micro level (e.g., Lim et al. 2020, Martin-Breen and Anderies 2011, Swayne et al. 2006).

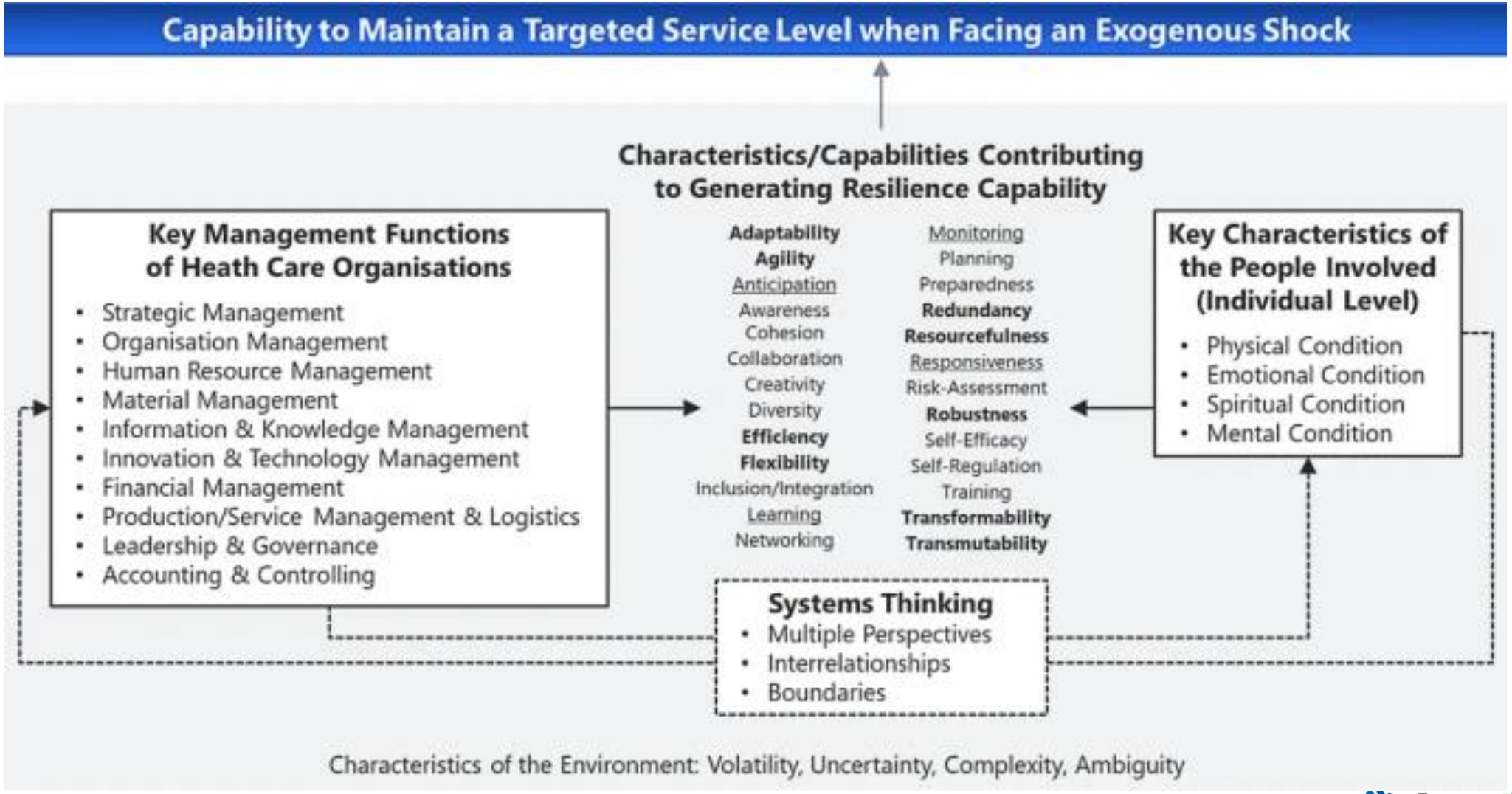

Short-Term Consequences
Excellent
Beneficial
Negligible
Critical
Catastrophic
Serendipities
Good Fortune
Things that go Right (Normal Outcomes)
Interventions
Miracles
Incidents
Near Misses
Accidents
Disasters
Mishaps
Very Rare
Unlikely
Possible
Likely
Certain
Probability of Occurrence


Macro Level
Meso Level
Micro Level
Individual Health Care Provider
Individual/Household
Health Care Environment
(e.g., health care specific planning organisations, other (public and private) primary, secondary community and mental health care providers, non-profit organisations, provider representatives, other patients)
Natural Environment
(e.g., ecological conditions, natural resources)
Political Environment
(e.g., government institutions, educational institutions, public insurance organisations)
Health Environment
(e.g., general health status of the population, psychographics, demographics)
Economic Environment
(e.g., business organisations)
Socio-cultural Environment
(e.g., population characteristics beyond health like education, religions, languages, culture)
Technological Environment
(e.g., business organisations, research organisations/foundations, educational institutions)
Legal/Regulatory Environment
(e.g., planning/regulatory organisations)

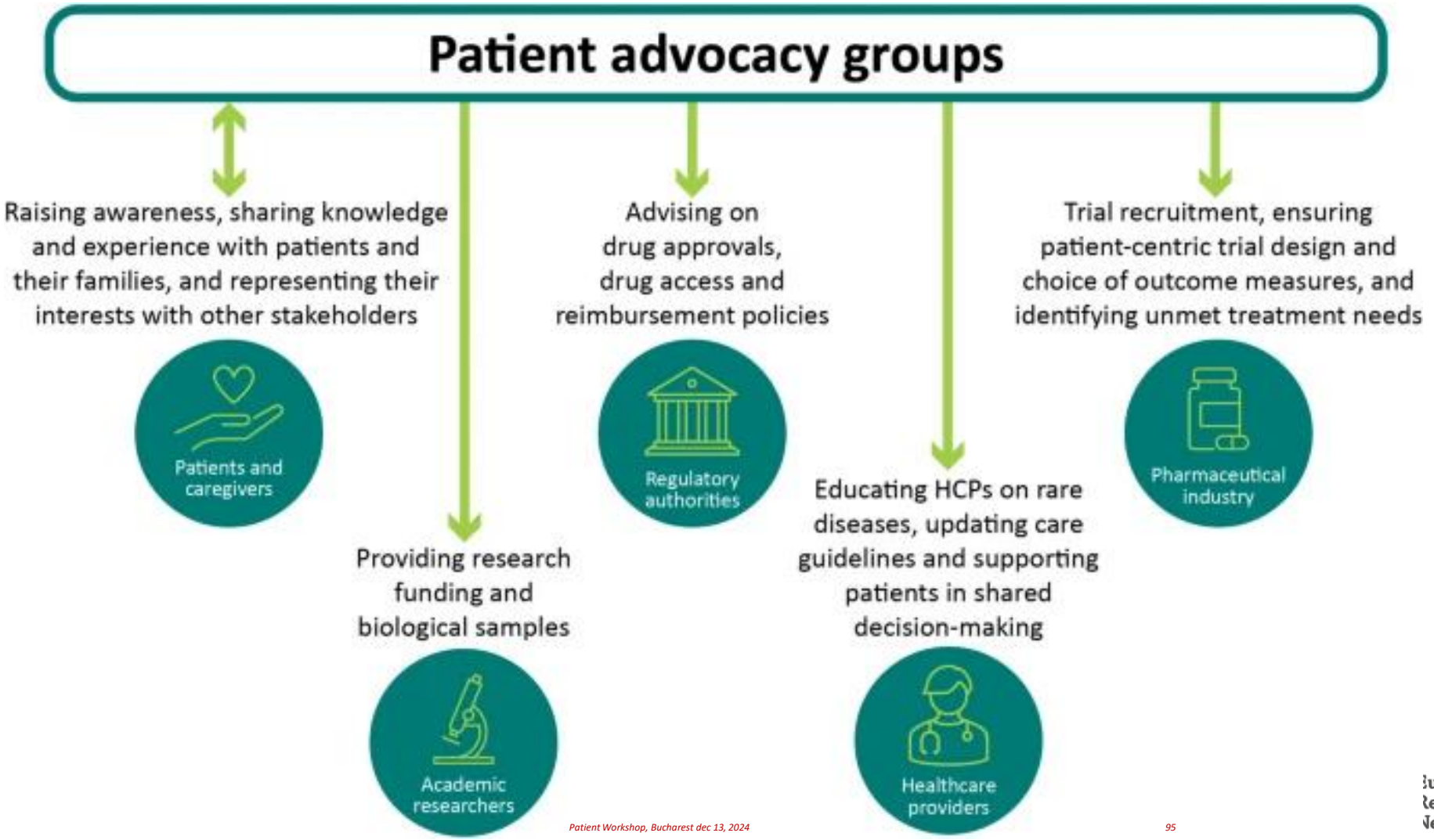

Functionality of the Health Care System
Quick wins
System gradually "absorbs" (part of) the positive effects of the planned intervention
Gain in Functionability
Planned Intervention
Adverse Event
① "Understand risks"
② "Anticipate/ Prepare"
Loss of Functionability
④ "Respond/ Recover"
⑤ "Adapt/ Transform"
Apply the Learning from Past Events and Get Prepared
③ "Absorb/ Withstand"
Time
❶ Proactive
❷ Event
❸ Reactive
Phases
Clear Aim Set & Prioritisation of Goals Altered
Ongoing consequences of event and reaction


Proactive Phase
Event Phase
Reactive Phase
Absorptive Path:
Core Capability: Redundancy
Core Capability: Robustness
Core Capability: Agility
Adaptive Path:
Core Capability: Resourcefulness
Core Capability: Adaptability
Core Capability: Flexibility
Transformative Path:
Core Capability: Efficiency
Core Capability: Transformability
Core Capability: Transmutability


Information Flows
Knowledge and its communication affects people's behaviour – and also flows of information follow their own dynamics.
Stories
INTERVENTIONS
Human Behaviour
The information flows exercise influence on human behaviour of patients, clinicians and managers.
Physical Flows (of Patients)
All the "tangibles" that we measure around patient flow are affected by behaviour.


Capability to Maintain a Targeted Service Level when Facing an Exogenous Shock
Characteristics/Capabilities Contributing to Generating Resilience Capability
Key Management Functions of Heath Care Organisations
• Strategic Management
• Organisation Management
• Human Resource Management
• Material Management
• Information & Knowledge Management
• Innovation & Technology Management
• Financial Management
• Production/Service Management & Logistics
• Leadership & Governance
• Accounting & Controlling
Adaptability
Agility
Anticipation
Awareness
Cohesion
Collaboration
Creativity
Diversity
Efficiency
Flexibility
Inclusion/Integration
Learning
Networking
Monitoring
Planning
Preparedness
Redundancy
Resourcefulness
Responsiveness
Risk-Assessment
Robustness
Self-Efficacy
Self-Regulation
Training
Transformability
Transmutability
Key Characteristics of the People Involved (Individual Level)
• Physical Condition
• Emotional Condition
• Spiritual Condition
• Mental Condition
Systems Thinking
• Multiple Perspectives
• Interrelationships
• Boundaries
Characteristics of the Environment: Volatility, Uncertainty, Complexity, Ambiguity

HEALTH CARE SYSTEMS RESILIENCE IN RARE DISEASES



YOUR OPINION

- 1.
- 2.

Thank you for your attention !

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Group 2
Different faces of grief

European Commission

EUROPEAN REFERENCE NETWORKS

Helping patients with rare or low-prevalence complex diseases

Loss and griefs from the professionals' perspective

Dr Marie Christine Rousseau

European Reference Networks ITHACA

Loss and griefs from the professionals' perspective

- Treating chronic, permanent health conditions (person with a rare disease/disability) means abandoning the medical model of healing

CARING≠CURING

- Time and onset of death cannot be anticipated/Death cannot be considered as the best thing for the persons they care for

Providing care entails a particular form of palliative care witch extends over years

A kind of taboo!
« Keep it professional! »

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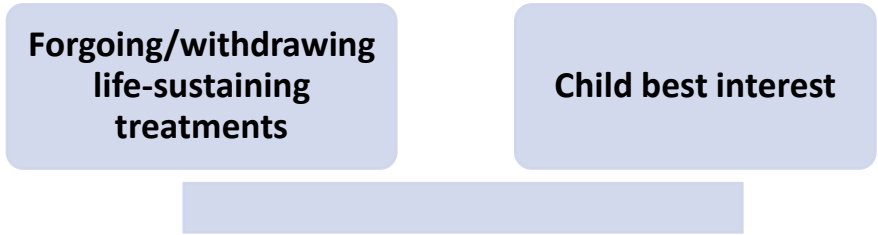
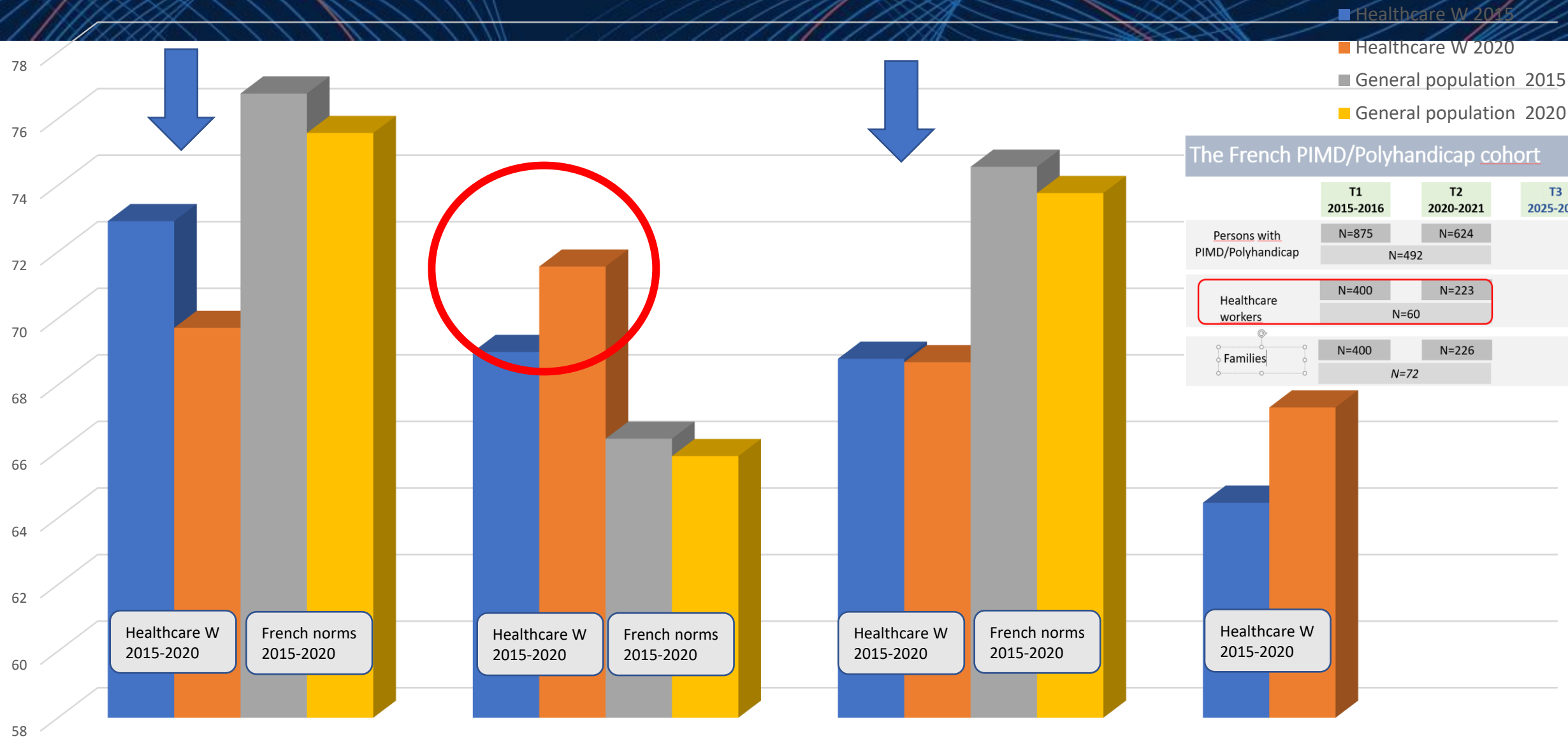
Healthcare workers face specific working conditions

- Decisions on behalf of person with a rare disease/disability



- Challenging personal histories of these persons and their families
- Restricted feeback and recognition of the care provided due to limitation of communication with these persons
- Frequent physical tasks due to complete physical dependence of the disabled persons with a rare disease/disability

Healthcare workers' QOL of persons with PIMD/Polyhandicap : longitudinal assessment



The French PIMD/Polyhandicap cohort

	T1 2015-2016	T2 2020-2021	T3 2025-2026
Persons with PIMD/Polyhandicap	N=875	N=624	
	N=492		
Healthcare workers	N=400	N=223	
	N=60		
Families	N=400	N=226	
	N=72		

Results and final conclusions

Dorica Dan,
Tanja Zdolšek Draksler

European Commission

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Helping patients with rare or low-prevalence complex diseases

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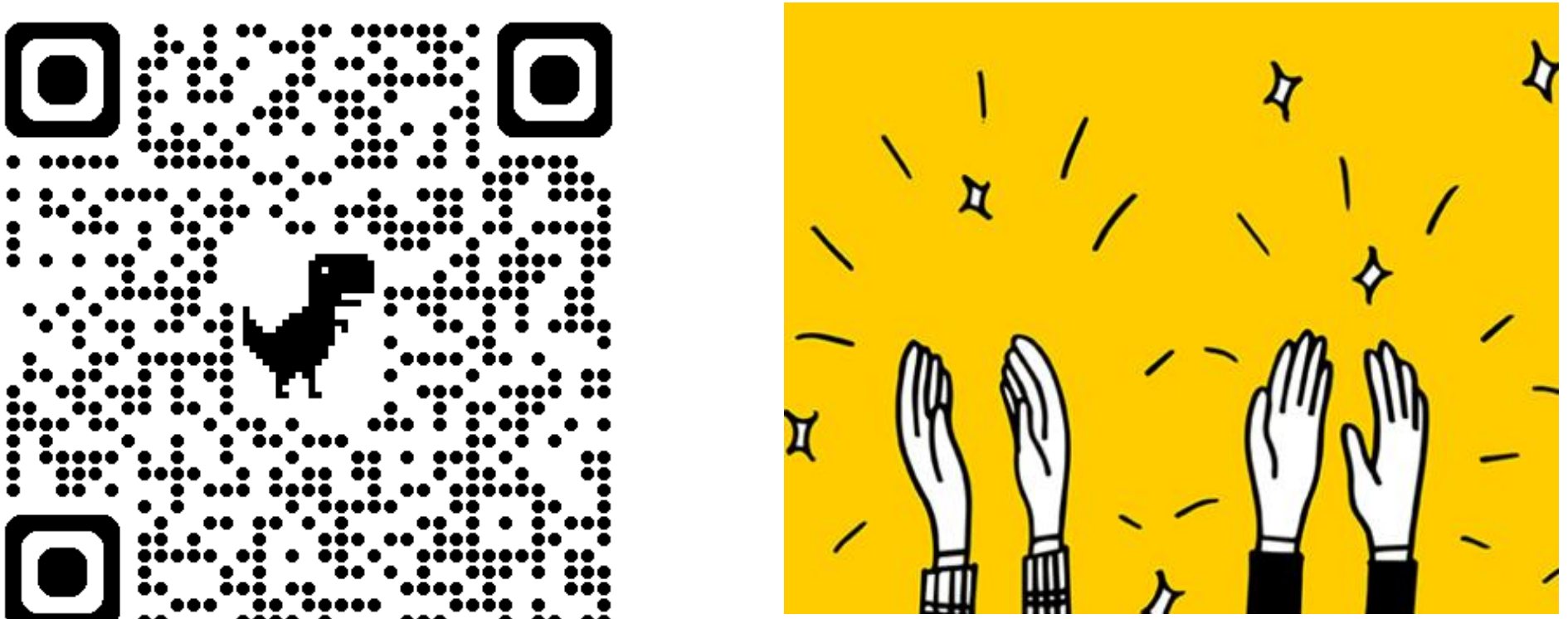
Your feed back



Survey

https://docs.google.com/forms/d/1b74f2q5ELB_ZnQ_DQPPs4gv06qiwGdtGo_5AsbnZh44/

Thank you !

Centrul NoRo - Zalău 2024

https://www.youtube.com/watch?v=ygTZkKtMCe8

- info@ern-ithaca.eu

Cocktail reception

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BOARD MEETING

BUCHAREST, ROMANIA
12-14 DECEMBER, 2024

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