EURORDIS
RARE DISEASES EUROPE

OUTLINE POSITION PAPER ON MENTAL HEALTH & WELLBEING

A Position Paper by EURORDIS and its Members on behalf of rare disease community in Europe

JUNE 2024

Table of contents

About Rare Diseases and EURORDIS-Rare Diseases Europe 2

Introduction 3

Structure of the Outline Position Paper on Mental Health 4

Policy 5

Recommendations – European Level 7

Recommendations – National Level 10

Annex I: Blueprint of Psychosocial Care 13

1. Holistic Care 15

2. Prevention 16

3. Family orientation 17

4. Person-centred 18

5. Resource-orientation 19

6. Supportive therapy 21

7. Orientation toward the rare disease journey 22

8. Interdisciplinary Cooperation 25

About this position paper 26

Annex II: References 27

Funded by the Euroean Union/ Views ans opinions expressed are however those of the autor(s) only and do not necessarily reflect those of the European or HaDEA. Neither the European Union not the granting authority can be held responisble for the.

Co-funded by the European Union

About Rare Diseases and EURORDIS-Rare Diseases Europe

Rare conditions affect at least 3.5% to 5.9% of the worldwide population (Nguengang et al., 2020), with 30 million people living with a rare disease (PLWRD) in Europe. The European Union (EU) defines a condition as rare when it affects less than 1 in 2000 citizens. Over 6,000 rare conditions have been identified to date, among which less than 6% currently have a treatment (Monaco et al., 2022).

Rare conditions are frequently chronic and complex. People with a rare condition live with disabilities which may be visible, invisible, degenerative, or vary from one day to the next and throughout the course of the disease. Evidence demonstrates that PLWRD and their families face serious challenges to health, economic situation, social inclusion and day-to-day lives, all of which can have a severe impact on their mental health and wellbeing.

EURORDIS-Rare Diseases Europe is a non-profit alliance of over 1000 rare condition patient organisations from 74 countries, including all EU countries, which work together to improve the lives of over 300 million people globally living with a rare condition.

The vision of EURORDIS is for a world in which each person living with a rare condition can have a longer and better life, achieving their full potential in a society that values their well-being and leaves no one behind.

Introduction

Rare conditions are characterised by both their rarity and their complexity, which together accumulate and increase the impacts on both the affected individual and the whole family. The scarcity of knowledge and effective treatments for most rare conditions results in a lack of support and understanding, which increases social isolation, stigma and discrimination.

The complexity of clinical presentation due to the multiple co-morbidities and intersectional needs associated with rare conditions has been documented, as has the complexity of the rare disease pathway, from establishing a diagnosis to determining and sourcing care, treatment and long-term follow-up. The majority of rare conditions affect children and young people.

Furthermore, a large number of people living with a rare disease (PLWRD) live with a disability and face barriers to social participation. Many are part of socioeconomic groups with low levels of income, education and employment. Consequently, PLWRD often have multiple intersectional needs, which increase their vulnerability to poor mental health and wellbeing. **Due to the complexities and multiple intersectional unmet needs of rare conditions, PLWRD are disproportionately impacted by the accumulated risk factors that populations facing vulnerable situations experience.**

PLWRD and their families live with severe, progressive and chronic conditions, which may have visible or invisible disabilities and multiple co-morbidities. Some rare conditions have associated mental health challenges as a primary characteristic or a co-morbidity. Although rare conditions are medically heterogeneous, producing a wide spectrum of needs, PLWRD and their families do live with common challenges that have a detrimental impact on their mental health and wellbeing.

> *For 85% of people living with a rare condition, the condition impacts upon several aspects of their health and everyday life. (Courbier et al., 2017)*

The common daily challenges experienced by PLWRD and their families include lengthy diagnostic journeys with a history of misdiagnosis and unnecessary treatments, and limited access and availability of treatment options and uncoordinated care pathways. The strain of juggling hospital appointments and treatment with everyday life significantly impacts PLWRD and their families.

This can result in reduced/stopped employment for long periods of time, limited access to education, and reduced employment opportunities. The combination of these common challenges, including uncertainties around prognosis and outcome, medical trauma, pain, economic hardship, discrimination, and isolation and stigma, increase the vulnerabilities and psychosocial risks of PLWRD and their families.

The complexities and challenges of rare conditions have been shown to increase the risk to mental health compared with the general population and underscore the particular vulnerability of PLWRD and their families to experiencing mental health issues (Spencer-Tansley et al., 2022).

> *90% of PLWRD surveyed felt worried, anxious, stressed and/or depressed and 19% had suicidal thoughts. (Spencer-Tansley et al., 2022)*

People living with a rare and/or undiagnosed condition live with an accumulative impact on their mental wellbeing. At an individual level, PLWRD can have an associated mental health co-morbidity, whereas at population level, the community lives with the increased psychological impact associated with the rare disease journey across all stages of life. Furthermore, PLWRD and their families have increased exposure to social inequalities and discrimination, which are determinants for poor mental wellbeing.

Despite the high level of needs faced by PLWRD and their families, the mental health of the rare disease community remains hidden and neglected, due to stigma, discrimination or from simply being overshadowed by the other medical complexities associated with rare conditions. The lack of recognition for the unmet mental health needs of the rare disease community in health, social, educational and employment policies further marginalise and excludes this group, putting them at risk for further vulnerability due to a lack of policy actions and available support to address their needs.

The rare disease community has identified as an absolute priority the need to look beyond the physiological aspects of rare conditions and to take concrete action to address the psychological impacts associated with these complex conditions (Rare 2023).

Without addressing the specific psychosocial, economic and environmental determinants that increase the risk factors associated with poor mental health, rare conditions will continue to have a detrimental impact on the mental wellbeing of individuals and families, hindering opportunities to access education, employment, independent living and to participate actively in society. To fully address these unmet needs, mental health and wellbeing must be addressed beyond healthcare sy stems, becoming incorporated in **all policies** and **at all levels of society.**

Structure of the Outline Position Paper on Mental Health

The Outline Position Paper on Mental Health is structured to set out the policy context detailing policy recommendations both at European and at National levels.

The primary aim of the Outline Position Paper is to support the implementation of the United Nations General Assembly Resolution (A/RES/76/132) call to develop effective programmes to promote mental health and psychosocial support for people living with a rare condition. The paper also sets out a proposal for how psychosocial care should be structured in Annex I.

Policy

Mental health has become defined as a global public health priority, with policy action initiated in all institutions from the United Nations, through the World Health Organization and the European Commission. This global call to action has mobilised all stakeholders, from Civil Society Organisations to academic, research and health and social care sectors, cutting across all policy areas including the health, social, education, climate, digital and economic sectors.

1. UNITED NATIONS

PLWRD are vulnerable to discrimination and stigma, experiencing unequal treatment in areas ranging from access to education, employment, leisure and other essential support services, which can be aggravated when the illness and disabilities are invisible or vary over time, which is the case for many rare conditions (Courbier et al., 2017). The United Nations has recognised the increased inequalities, discrimination and stigma faced by PLWRD in a new UN Resolution on addressing the challenges of PLWRD and their families (A/RES/76/132, 2021).

The UN specifically underscores the high level of unmet needs and increased vulnerability of people living with a rare disease, urging Member States "to implement effective programmes to promote mental health and psychosocial support for persons living with a rare condition, and to promote policies and programmes that enhance the well-being of their families and caregivers" (A/RES/76/132, 2021).

2. WORLD HEALTH ORGANIZATION

The World Health Organization (WHO) has recognised mental health as a leading public health priority that impacts global development, affecting an estimated 1 billion people worldwide, with depression being a leading cause of disability. The global mental health crisis has been the legacy of the recent global COVID-19 pandemic, with a reported 25% of the population affected by anxiety and depression in the first year following the pandemic outbreak.

However, the impact of the pandemic and social distancing measures disproportionately affected people in certain communities, with the WHO recognising "people with an existing health condition" as one of the main groups in vulnerable situations more likely to develop symptoms of mental disorders following the pandemic, along with young people and women (WHO, 2022).

Furthermore, among the risk factors that negatively impact mental health there is a strong association with social inequalities. The WHO recognises that the greater the level of inequity, the higher the risk to mental health and well-being (Social Determinants of Mental Health, WHO, 2014). This is particularly pertinent to rare conditions.

In the wake of the UN Resolution, a WHO Resolution on Rare Diseases is under preparation, and it must address the mental health needs of PLWRD, amongst others. **People with rare conditions have been shown to live with increased exposure to social inequalities and discrimination, which, coupled with the medical complexity associated with rare conditions, increases the emotional impact of living with a rare condition and increases the vulnerabilities and risks to their mental health and wellbeing.**

3. EUROPEAN UNION

Mental health has now been recognised as a public health priority in the European Union. Notably, during the 14 September 2022 State of the Union speech at the European Parliament, European Commission President Ursula von der Leyen committed to adopting a new European Union "Action Plan on Mental Health".

The "Action Plan on Mental Health" was published as a new *Commission Communication – Comprehensive Approach to Mental Health* in all policy areas on 07 June 2023. The European Parliament published a report on Mental Health in 2023 highlighting the need to take a psychosocial approach and focusing on addressing the needs of groups in vulnerable situations. These two initiatives are an important milestone in renewing the political commitment to address the mental health needs of all Europeans.

In November 2023, the European Parliament adopted an Own-Initiative Report on Mental Health which underlined **the heightened vulnerability to mental health problems in specific groups, including recognising individuals with chronic and rare conditions** and called on the European Commission to convert the flagship initiatives, introduced in the comprehensive approach, into concrete actions with adequate financial support for vulnerable groups, including people living with chronic and rare conditions and disabilities.

The Commission Communication will be supported in the coming years with targeted community actions, initiatives and grants to support the priority areas, including for 'those most in need'. It is imperative to continue to advocate for the formal recognition of the unmet needs and accumulated impact on mental health and wellbeing of PLWRD and their families, and to take concrete action across all policy areas to address these unmet needs.

4. TIME TO ACT

The political momentum for addressing mental health as a public health priority both at an international and European level provides a timely opportunity to ensure the visibility of the impact of rare conditions on mental health and wellbeing. This momentum provides a window of opportunity for taking decisive actions to address the unmet intersectional needs of PLWRD at National, European and international level.

EURORDIS wishes to leverage the opportunity of the new *Commission Communication on a Comprehensive Approach to Mental Health* to make visible the social determinants of mental health and the unmet mental health needs of PLWRD and their families, and to take affirmative action to address these needs.

The following section will detail the policy recommendations needed at both European level and National level to address the unmet mental health needs of people living with a rare condition.

Recommendations – European Level

VISIBILITY OF RARE CONDITIONS IN MENTAL HEALTH WITHIN THE EU AGENDA

1. CALL FOR THE EUROPEAN COMMISSION TO ADOPT A MORE INCLUSIVE APPROACH FOR ALL POPULATIONS IN VULNERABLE SITUATIONS, AS PEOPLE WITH AN EXISTING HEALTH CONDITION (CHRONIC AND RARE CONDITIONS) ARE NOT CURRENTLY RECOGNISED IN THE NEW COMMUNICATION

> *People living with a rare condition face a risk more than three times higher than that of the general population for experiencing unhappiness and depression. (Courbier et al., 2017)*

The new Communication identifies populations in vulnerable situations and the need for EU-funded actions to target support, resources and tailored policies to meet the specific mental health needs of these high-risk groups. The high-risk groups recognised in the new Communication include children and young people, older people, women, victims of gender-based violence, victims of trafficking, victims of crime, cancer patients and people with disabilities, refugees and displaced people, people experiencing homelessness and people living in rural or remote areas.

However, **the new comprehensive approach has fallen short of being inclusive of all groups in vulnerable situations, by not explicitly recognising Europeans living with existing physical health conditions, including chronic and rare conditions.** The lack of inclusion and visibility of PLWRD as highly vulnerable members of society in key EU policies will increase the risk of deepening existing inequalities and further marginalise these populations.

RECOMMENDATION: 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.

2. TACKLING STIGMA AND DISCRIMINATION

PLWRD and their families frequently find themselves at the intersection of numerous diverse vulnerable situations, including psychosocial risks at work or school, medical uncertainties, and economic hardship. The focus on tackling stigma and discrimination in the new Communication is welcome as a critical cornerstone to all the objectives and EU-funded actions identified in the Communication. However, not all populations in vulnerable situations are currently recognised, and PLWRD must be included in this initiative to implement a truly comprehensive approach.

RECOMMENDATION: Call on the Commission to be inclusive of all populations in vulnerable situations (including those living with rare conditions) in the actions to implement the Comprehensive Approach to Mental Health.

3. WHILST THE COMPREHENSIVE APPROACH RECOGNISES THE NEEDS OF CERTAIN POPULATIONS IN VULNERABLE SITUATIONS, THE COMMUNICATION ONLY HAS TWO FLAGSHIP INITIATIVES TO ADDRESS THESE NEEDS

Whilst the Comprehensive Approach recognises the needs of (some) populations in vulnerable situations, the Communication only has two flagship initiatives to specifically address these needs: *Flagship initiative 11 for victims of crime and Flagship initiative 12 for young cancer survivors*. Additional new actions that specifically target the other high-risk groups should be identified in the European Commission's follow-up actions to implement the Communication, including new flagship initiatives of EU funding programmes.

RECOMMENDATION: Call on the Commission to 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 to improve access to psychological support.

4. MENTAL HEALTH & WELLBEING TOOLKIT FOR POPULATIONS IN VULNERABLE SITUATIONS

Addressing the needs of vulnerable populations presents an area of added value for the European Union, whereby EU community actions help to strengthen national measures. It is reported that each of the populations in vulnerable situations comprises a significant sized community – young people (67 million), older people (94 million), chronic conditions (50 million) and rare conditions (30 million).

There is a wide discrepancy in available health and social services across EU countries to support these vulnerable populations.

The development of a *Mental Health & Wellbeing Toolkit for Populations in Vulnerable Situations* would help address these gaps by strengthening the capacity of patient organisations and Civil Society Organisations (CSO) through EU shared best practices and tools to support their respective communities, thereby reducing avoidable downstream health, social and economic costs.

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

Strengthening the resilience and wellbeing of populations in vulnerable situations would have a direct benefit on the economies of EU Member States by reducing downstream costs and increasing productivity, as people and their family members would be better able to work and have greater independence.

A joint initiative could bring together experts and leaders from vulnerable populations to share best practices and evidence-based approaches, test their applicability and scalability and co-create a common *Mental Health & Wellbeing Toolkit for Populations in Vulnerable Situations*.

Increasing the capacities among both social care systems and Civil Society Organisations, including organisations representing people living with rare conditions, would support and empower them to better alleviate the risk factors and drivers of poor mental health, prevent mental health comorbidities and address the high levels of unmet needs of these populations in vulnerable situations.

SPECIFICALLY

- Reduce the impact on the lives of individuals and their family members by increasing social inclusion and supporting access to education, employment and independent living.
- Reduce and prevent costs in the health, social and educational systems and employment sectors stemming from unmanaged psychological impact.
- Enable individuals and families to be active in society, continuing to work, contribute taxes and live independently.

RECOMMENDATION: Call on the Commission to allocate funding for a *Mental Health & Wellbeing Toolkit for Populations in Vulnerable Situations* in order to build the capacities and empowerment of these groups to become mentally healthy communities.

This toolkit could be generic in nature, identifying the best and most promising practices and evidence-based tools that could be tailored by each population in vulnerable situations to address their specific needs.

Recommendations – National Level

VISIBILITY OF MENTAL HEALTH IN RARE DISEASE NATIONAL PLANS & STRATEGIES

The impact of rare conditions on mental health and wellbeing should be addressed on two levels:

- **Individual level:** rare condition(s) can present mental health challenges as a primary characteristic or associated co-morbidity.
- **Population level:** Everyone affected by a rare condition including the whole family is affected by living with the increased psychological impact associated with the rare disease journey across all stages of life.

Actions to address these unmet needs therefore need to be organised to complement each other by reducing the co-morbidities affecting the individual as well as the psychological impact on the family as a whole.

RECOMMENDATION: Increase and include the visibility of mental health in the revision of Rare Disease National Plans & Strategies and commit to actions to address the unmet mental health needs of PLWRD and their families.

1. ADDRESSING INDIVIDUAL CO-MORBIDITIES ASSOCIATED WITH A RARE CONDITION

The mental health co-morbidities associated with rare conditions are a source of significant psychological impact and debilitating unmet need. For PLWRD, mental health co-morbidities stemming from their condition present a significant psychological impact, creating a debilitating unmet need, which affects not only the affected individual, but the whole family.

Rare conditions with mental health as a primary characteristic or associated mental health co-morbidity include Rett Syndrome, Prader Willi Syndrome, Pitt Hopkins Syndrome, Sanfilippo Syndrome, Glut1 Deficiency Syndrome, Hypothalamic Hamartoma, Lennox Gastaut Syndrome, Alternating Hemiplegia, Ring 20 Chromosome, KCNT1 Deficit Syndrome, Dravet Syndrome, Neurofibromatosis Type 1, Fragile X Syndrome, Williams Syndrome , Tuberous Sclerosis, among others.

Upskilling the medical and mental health workforce to increase the understanding of the impact of rare conditions with mental health as a primary characteristic or associated mental health co-morbidity is essential in order to tailor effective treatment programmes for PLWRD affected by these conditions. In addition, there is a need to develop new specialists in congenital neurodevelopment (including motor development, mental development and education). This specialty aims to understand the neurodevelopmental needs of the child from birth or onset of symptoms and follow the child to adulthood.

Training in the clinical decision support tools is needed to reinforce early detection of mental health difficulties in children and young people with a learning disability and/or communication differences. There is currently a lack of assessments (Flynn et al., 2017) and interventions (Vereenooghe et al., 2018) available to aid early detection of an underlining mental health issue.

It is critical to target and increase training of mental health professionals and to strengthen the training for healthcare and other professionals, as significant gaps exist in mental health workforces and healthcare professional skills and training in many EU Member States.

EURORDIS proposes measures to better identify and address the training needs of health professionals:

- Specific training in rare conditions for psychologists, psychiatrists and associated mental healthcare professionals to enable them to recognise and understand the organic cognitive problems present in rare conditions, and the way in which they evolve over time.

- Upskill medical healthcare professionals with knowledge and understanding of rare genetic and non-genetic conditions and the associated impact on mental health in this population.

RECOMMENDATION: Support new and targeted medical training to strengthen cross-specialty training for all professionals to better understand the relationship between physical and mental health, specifically to allow mental health practitioners to become more 'rare aware' and medical and nursing care practitioners to become more 'mental health' aware.

2. ADDRESSING THE PSYCHOLOGICAL IMPACT ON THE FAMILY UNIT AS A WHOLE

The main drivers of poor mental health in PLWRD and their families are directly linked to issues in managing the rare conditions from the onset of symptoms, from searching for the right expert and securing a diagnosis through seeking effective treatments and pushing for research to find a cure. PLWRD and their families face daily challenges in managing the condition and living with uncertainty around the prognosis and future progression of the disease symptoms.

The UN General Assembly has recognised the high level of unmet mental health needs and increased vulnerability of people living with a rare condition. **The United Nations General Assembly Resolution A/RES/76/132 (2021) "urges Member States to implement effective programmes to promote mental health and psychosocial support for persons living with a rare condition, and to promote policies and programmes that enhance the well-being of their families and caregivers".**

The majority of EU Member States have adopted or plan to adopt National Rare Disease Plans and Strategies, and it is critical that mental health and wellbeing for PLWRD and their families be firmly recognised in these plans and strategies, specifically including the ways in which national initiatives and actions will fulfil the UN General Assembly commitment to develop psychosocial programmes for rare conditions.

RECOMMENDATION: Call on EU Member States to honour the United Nations General Assembly Resolution (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.

PSYCHOSOCIAL CARE

The United Nations General Assembly call for the development of psychosocial programmes for rare conditions. This does not necessarily equate only to integrating psychosocial personnel as core members of the medical team, but also through enhancing medical care to be psychologically informed.

> *Nearly 50% of people from the rare disease community were not offered psychological support (in specialist services or expert centres). (Courbier et al., 2017)*

The EURORDIS Rare Barometer Voices survey highlights the gap in psychological support in medical care and expert centres. Other published literature sources report that only one in seven rare disease patients state that they receive sufficient psychological support (Nunn et al., 2017).

Member States can include psychosocial programmes as part of their national rare disease plans by enhancing the existing medical care provided in expert centres to become 'psychologically informed medical care'.

For example, in Luxembourg the national authority has commissioned psychosocial care for people living with a rare disease through the national rare disease alliance – ALAN Maladies Rares Luxembourg.

The service was launched under the national rare disease plan in 2009 and is delivered by a psychosocial care team of psychologists and social workers. This service has been recognised by the European Commission under the Best Practice Portal.

RECOMMENDATION

- **Integrate psychosocial support would become an integral standard of medical care for people living with a rare disease, by integrating 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.**

In the following Annex I a proposal is set out for how psychosocial care for people living with a rare disease should be structured. This proposal can be used by Member States to guide the development of psychosocial care, delivering on the commitment set out in the UN resolution.

Annex I: Blueprint of Psychosocial Care

People living with a rare condition live at an intersection of multiple complex needs that cut across all aspects of life, from physical health, mental health, social integration and independent living, making it impossible to separate out any one of the needs.

This outline position paper focuses on the mental health and wellbeing needs of PLWRD and specifically elaborates on the UNGA Resolution, setting out how a psychosocial programme could be structured. This paper has the value to bring together concrete recommendations and actions that can be used by both the rare disease community and Member States to support the development of psychosocial programmes for PLWRD, by enhancing existing medical care to be psychologically informed.

Psychosocial support has been evidenced to improve the mental health and wellbeing of patients and their families, improving their ability to adapt to the course of the disease, increasing adherence to medical care and interventions while reducing related stress factors (Askins & Moore, 2008; Kazak, 2005).

The strong evidence-base for psychosocial care has resulted in it becoming an integrated component of medical care for cancer. However, PLWRD face significant barriers to accessing psychosocial care, creating increased inequalities which in turn further increase the risk to mental health and wellbeing.

DEFINITION OF PSYCHOSOCIAL CARE

Psychosocial care is concerned with the psychological and emotional wellbeing of the patient and their family/carers, including addressing issues of self-esteem, insights into adaption to the illness and its consequences, communication, social functioning and relationships (Onyeka et al., 2010).

Psychosocial care is conducted in cooperation with the medical treatment team. The emphasis is on supporting the resources of the patient and family throughout the course of the disease, including during therapy and follow-up care. The basis for psychosocial care is a supportive and informative relationship with the affected individual, family and social environment.

Psychosocial care is oriented towards the physical, emotional, social and developmental potential of the affected child, adolescent or adult and their family and social environment, and takes into account individual styles and abilities to cope and adjust (Schröder et al., 2008).

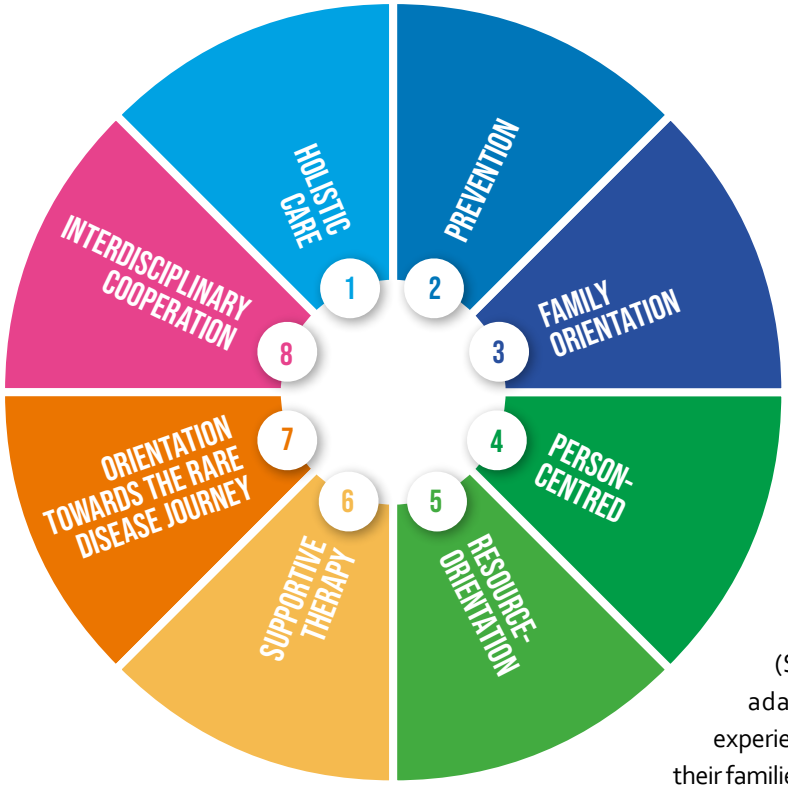
BARRIERS TO EXISTING PSYCHOSOCIAL SUPPORT

Even when psychosocial care is available, there are barriers to accessing this support, including a lack of awareness and information on available psychosocial care resources, such as peer support and/or patient groups (Witt et al., 2023). Other reported barriers include:

- Medical care is not provided in a holistic way and the individual and their family are not routinely asked about the need for psychosocial support.
- Time management constraints due to medical appointments, managing daily life activities and taking care of family needs.
- Difficulties accessing existing therapies due to geographic distances travel, cost implications and/or waiting times.
- Information inaccessible due to linguistic and cultural difficulties.
- Psychosocial support not delivered in an accessible language and format.

Psychosocial care should address these identified barriers around time, distance, language, culture and organisation so that families can benefit from existing care to address and reduce the psychological stress factors associated with rare diseases.

PSYCHOSOCIAL CARE CAN BE STRUCTURED TO ADDRESS THE NEEDS FOR PEOPLE LIVING WITH RARE CONDITIONS UNDER THE FOLLOWING 8 DOMAINS



These 8 domains and supporting standards are based on the PSAPOH Guideline on Psychosocial Care in Paediatric Oncology and Haematology (Schröder et al., 2008) and have been adapted to address the common needs experienced by people living a rare disease and their families.

1. Holistic Care

Rare diseases are complex, multisystemic, progressive conditions that frequently require intensive interventions and treatments. The stresses relating to both the disease and the treatments significantly increase the psychological risk factors and distress of the whole family, requiring a holistic approach to address the complex set of physical, emotional, social and financial needs.

Addressing psychosocial problems is at the centre of psychosocial care and may include helping PLWRD and their families with traumatic life events, social isolation, stigma and discrimination, or grief and loss, all of which have a strong association to anxiety, depression and adjustment.

The EURORDIS Holistic Care position paper (2019) highlighted that PLWRD need follow-up care and support from different health and social care professionals and from a range of services, including rehabilitation, day care, home care, personal assistants, respite services and psychological support (Castro et al., 2019).

Coordinated care across multi-sector services is the cornerstone of holistic care. A lack of coordinated care has been shown to have a psychosocial impact on the patient and carers (Simpson et al., 2021). Existing evidence highlights the importance of flexible, tailored care throughout the rare disease journey.

Key measures identified to reduce the negative psychosocial impact for PLWRD include improvement to the coordination of care by clinician services, the use of technologies, and adopting a multidisciplinary approach (Simpson et al., 2021).

STANDARDS

1. The primary aim of psychosocial care is to identify the psychosocial implications of living with a rare condition, which are frequently complex multisystemic conditions, and the impact of treatment (or lack of access to treatment), which can result in acute or 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.

2. Prevention

Routine and regular mental wellbeing assessments for the early detection and prevention of mental health decline can support the empowerment of individuals and family members with information, knowledge and resources to strengthen coping strategies (McConkie-Rosell & Sullivan, 1999).

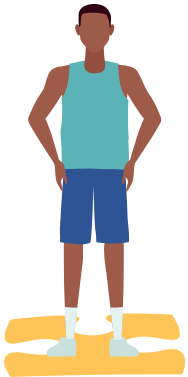
Psychological interventions that are structured around the whole family are needed, such as health promotion and/or prevention of mental health problems, to reduce the emotional strain and potential depression experienced by all family members (Rice et al., 2020; Baumbusch et al., 2019).

STANDARDS:

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.



3. Family orientation

Rare conditions have a significant psychological impact not only for the affected individual, but also for the whole family, throughout all stages of the rare disease journey (Kenny et al., 2022).

Family functioning has been identified as a significant risk factor for the mental health and quality of life of parents (Boettcher et al., 2020). By adopting a family-oriented approach to medical care for PLWRD, the parents, carers and family can be empowered to provide the best support needed for the person affected.

Although research has shown that such empowerment can improve the cost-effective use of health services (Wallerstein et al., 2006), there remains a lack of fundamental support to mitigate the risk factors for poor mental health for affected individuals and their families (Kasparian et al., 2015; Anderson et al., 2013).

STANDARDS

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.

4. Person-centred

Psychosocial care needs to be centred on the individual and their specific circumstances. The heterogeneous nature of rare and undiagnosed diseases and the associated interventions and treatments are critical factors to understand and consider when tailoring psychosocial care to the individual needs and situation of each affected person.

Empathic communication and awareness for the specific needs and circumstances of each individual is the foundation of psychosocial care. For example, the manner in which a diagnosis is disclosed can have a long-lasting impact and colour the relationship PLWRD and their families have with medical services going forward. When sensitively handled, PLWRD and their families can be empowered to cope with the demands placed upon them, come to terms with the diagnosis and strengthen their resilience in managing the multiple uncertainties associated with rare conditions.

Conversely, poor communication of a diagnosis has the potential to create long lasting damage to relationships in affected families, or to erode trust between individuals and families and healthcare professionals with potentially devasting implications for both affected individuals and extended family members.

Communication of a rare condition diagnosis must therefore be disclosed in an empathic, psychologically supportive manner, with the presence of a friend or family member in attendance to support the affected individual. Inclusive communication that involves the individual in their own care decisions can enhance the sense of agency and dignity for PLWRD.

The process for disclosing diagnostic results should not be rushed and should include reassurance for parents or caregivers that they will receive information, support and advice to care for the affected person going forward.

Some PLWRD and their families are bombarded with information about the rare condition, both from clinical services as well as from internet sources. Others may be provided with very little information or be unsure where to seek out reliable information and advice. Trusted information and signposting are needed to help individuals and families acquire a deeper understanding and knowledge about a condition, at the point at which they are ready to receive it..

STANDARDS

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.

5. Resource-orientation

Psychosocial care takes a resource-oriented approach, with the aim of promoting the resources of the affected individual and their family throughout the course of the disease and across the healthcare journey, from the initial diagnostic process through care, treatment and follow-up. The basis of psychosocial care is a supportive and informative relationship with the patients, families and the social environment (Schröder et al., 2008).

Uncertainty has been directly linked to anxiety, with greater amounts of uncertainty correlated to a higher level of anxiety. The unpredictable nature and scarcity of knowledge for many rare conditions render daily life uncertain for many affected individuals, making it difficult to plan for the future.

Living with unpredictable events and an uncertain future is a stressful and anxiety-provoking experience for everyone affected by a rare condition. The corrosive emotional impact of living over time with the uncertainty characteristic of rare conditions can be challenging to manage. Coping strategies and support systems are needed to navigate the emotional impact of living with ongoing uncertainty.

One of the best ways to reduce the impact of living with uncertainty and better support PLWRD and their families is to address these issues within mainstream medical services by mitigating uncertainty from the health and social care system. Health and social care professionals can play a large role in alleviating many uncertainties by using appropriate language and systems.

Rare conditions expose PLWRD and their families to both macro and micro uncertainties. The macro uncertainties associated with rare conditions are unavoidable, whereas the micro uncertainties that are often associated with being on the rare disease pathway can be mitigated and/or are preventable.

MACRO UNCERTAINTIES

- living with the onset of symptoms of an undiagnosed condition and the search for a diagnosis.
- condition progression over time and unclear prognosis.
- responsiveness to treatment and symptom control.

MICRO UNCERTAINTIES

- poor coordination between care services and rollercoaster of tests and observations.
- delays in referrals and communication around waiting times for appointments and results.
- capacity of others to understand a rare condition and its impact across statutory services, workplace, and personal relationships.
- money, accommodation and employment.

One of the best ways to reduce the impact of living with uncertainty and better support PLWRD and their families is to mitigate these micro uncertainties from the health and social care system. Health and social care professionals can play a large role in alleviating many of these uncertainties by using appropriate language and systems.

Isolation can be exacerbated due to living with uncertainty and anxiety as well as by the significant time needed to manage care and treatment at home or during medical appointments. The situation can be further compounded by a lack of understanding within both social and wider family networks.

> *The most important problem faced by families is isolation from friends and family, with 54% of people declaring that their isolation was caused or amplified by the rare condition. (EURORDIS Juggling care and life survey, 2017)*

Support from friends and the extended family is critical throughout the rare disease journey. However, lack of understanding or familiarity with rare conditions, compared with more common conditions such as cancer, can result in individuals affected by a rare condition feeling misunderstood, thereby increasing their sense of isolation, and reducing the possibility of support at the time when it is most needed.

For PLWRD and their families, finding a balance between maintaining hope and optimism and bearing disappointment can be challenging. Managing expectations, planning social events with consideration for individual limitations, managing uncertainty and seeking support from others can all help navigate this difficult balancing act.

Patient groups are frequently key providers of psychosocial support to affected families and individuals. Being connected to a network of people with shared lived experience can be an invaluable support for mental wellbeing by mitigating isolation, providing practical resources and information, and perhaps most significantly, by offering hope.

Patient organisations play an important role in mitigating the uncertainties associated with specific rare and undiagnosed conditions by providing a sense of belonging through the provision of community and peer support. As the foundation of psychosocial care, patient organisations also provide access to trusted information, facilitating earlier detection and preventative support.

Peer support has been determined to be the only significant predictor of mental health in mothers of children with a rare condition (Boettcher et al., 2020). Evidence based around peer support from social networks is more effective when it is actively supported by the clinical services treating the person living with a rare condition.

STANDARDS

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.

6. Supportive therapy

PLWRD and their carers have reported that healthcare teams do not routinely support them beyond the clinical aspects of their treatment (EURORDIS, 2021). Although it is evident that rare conditions severely impact the parents, siblings and carers of PLWRD, access to psychosocial support is seldom requested nor made available. In addition, parents and carers have expressed that they find it hard to ask for psychosocial support for themselves as the needs of the child or person living with a rare condition are the priority (Smits et al., 2022).

> *Over half (54%) of survey respondents had not accessed any professional psychological support. (Genetic Alliance UK, 2022)*

The understanding of the psychosocial needs and treatments for anxiety, depression and stress is well documented for existing health conditions such as cancer. This psychosocial support should be an integrated part of the care and management for both affected individuals and their family members / carers (Institute of Medicine, 2008). However, for rare diseases this is not a standard part of care and treatment.

People living with a rare disease and their families should have access to psychosocial care when it is needed (EURORDIS Achieving Holistic Person-Centred Care Paper, 2019). PLWRD and their families should have routine access to the simple, effective psychosocial interventions they need (e.g., peer support, curated information, access to experts, and time and care).

The patient community preference that psychosocial assessment and access to support be fully integrated as part of the coordination of rare condition services and made routinely available following diagnosis is supported by published evidence (Anderson et al., 2013; Zurynski et al., 2017; Spencer-Tansley et al., 2022).

Routine psychosocial assessment would not only detect poor mental health but also be able to identify ways to increase mental health protection, reduce risk factors, promote good mental health, and prevent the precipitation of decline. This would strengthen the capacity of PLWRD and their families to cope, improve quality of life and prevent the costly downstream health system impacts of unmanaged psychological needs.

STANDARDS

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.

7. Orientation toward the rare disease journey

People with rare diseases live with the accumulative effects of the psychological impact throughout the rare disease journey across all stages of life. The common characteristics of the rare disease journey is a long diagnostic odyssey, limited availability or lack of access to treatment, uncoordinated care, living with disabilities, uncertainty around prognosis and future outcomes and facing social stigma and discrimination.

These additional stressors have been shown to increase the risk to mental health compared with the general population and underline the fact that PLWRD are particularly vulnerable to experiencing mental health issues (Spencer-Tansley et al., 2022).

Psychosocial care should be oriented towards both the course of the disease and treatment as well as the impact the rare disease journey has on the individual and the family as a whole. Tailored psychological support should address the impact of living with a rare disease and identify coping strategies to deal with the uncertainty of the rare disease journey.

THROUGHOUT THE DIAGNOSTIC ODYSSEY

For many PLWRD, a diagnosis is only obtained after a long and arduous process, with an average of 5 years (Genetic Alliance UK, 2020; EURORDIS, 2009) and following visits to more than 6 specialists (Zurynski et al., 2017).

People with an undiagnosed rare disease and their families live with the impact from the diagnostic odyssey, which causes emotional stress, economic loss and significant delay in accessing appropriate treatment (Wu et al., 2020; Zurynski et al., 2017). For some, this odyssey is ongoing, and a diagnosis is never obtained, such as those living with Syndromes Without A Name (SWAN).

> *Living with uncertainty through the diagnostic odyssey results in living with anxiety, fear, stress, anger and frustration, as well as increased social isolation. (EURORDIS, 2009)*

A 2022 EURORDIS survey on the diagnostic journey of over 10,500 PLWRD (under publication) reveals that PLWRD whose needs for psychological support were met accessed a confirmed diagnosis faster. The average time from symptom onset to a confirmed diagnosis was 3.8 years when psychological support needs were met versus 5.1 years for those whose needs were not met.

These findings suggest that accessing psychological support helps PLWRD to better navigate the health system and the diagnostic odyssey (Faye, 2024).

The often-lengthy period of uncertainty during the journey to obtain a diagnosis can be emotionally challenging as individuals grapple with questions about their future and the management of the condition. For families and individuals who never receive a firm diagnosis, the process can be devastating.

> *>1/3 of parents of a child with an undiagnosed condition meet clinical criteria for mild to moderate depression or anxiety. (McConkie-Rosell, 2018)*

The obligation to repeatedly describe symptoms to different health professionals, sometimes facing disbelief or questioning of the validity of a diagnosis are among the traumatic experiences facing individuals and families who travel the rare disease journey.

PLWRD and their families may experience medical trauma, which can lead to post-traumatic stress disorder if left untreated. Addressing and overcoming medical trauma is important and may involve seeking support, establishing patient organisations, and advocating for improved healthcare experiences.

It is commonly reported that one reason for the long diagnostic delay for PLWRD is that many rare diseases have no effective treatment. Although many conditions do not have a treatment, they are actionable. Securing a diagnosis reduces worry and uncertainty and empowers affected individuals/parents/caregivers to connect with a patient community, thereby reducing social isolation and accessing peer and psychosocial support.

AT DIAGNOSIS

The moment of diagnosis is incredibly important for PLWRD and their family and can be one of the most challenging and pivotal periods in the rare disease journey. Many people find this period highly emotional. Upon receiving a definitive diagnosis, family members and caregivers often initially feel relief, which is soon replaced by significant feelings of fear, stress and guilt. Stress can manifest through a range of psychological symptoms that may require health professional intervention (Kenny et al., 2022).

Understanding and absorbing the diagnosis and the related psychological, emotional and practical implications takes time. It is important for health professionals to provide the opportunity for a follow-up appointment for additional information, to answer questions that have arisen in-between appointments and discuss the diagnosis further.

It can take several months for PLWRD and their families to process the news of a diagnosis. Coming to terms with the various implications of the diagnosis and developing the capacity to formulate questions to obtain the understanding and support they may need will vary widely across families, and family members. Inaccurate, out-of-date or worst-case scenario information can heighten fears, worries and anxieties for PLWRD and their families, adversely impacting mental health and wellbeing.

All people diagnosed with a rare condition should routinely have a regular follow-up appointment scheduled that allows PLWRD and their family the time needed to emotionally process the diagnostic results.

A diagnosis often triggers a process of grief and mourning for the 'imagined future' that previously existed. It can also trigger feelings of guilt (particularly when issues of genetic inheritance are involved) and have a range of consequences on the relationships between parents, children and siblings. It can also impact the relationship between the couple, particularly with respect to future life choices and family planning.

DURING CARE AND TREATMENT

Rare conditions typically require individuals and their families to manage living with various symptoms, tests, treatments and numerous medical appointments, often at diverse geographical locations. Disease and symptom management is often logistically burdensome, which can itself take a toll on emotional wellbeing. Repeated tests and unpleasant or intrusive medical interventions over time can lead to trauma in both adults and children. Fear of doctors or hospitals may result in 'hospital avoidance' or 'non-adherence' to treatment or monitoring regimes. Juggling medications, adhering to treatment schedules, and the consequent impact on work life and personal relationships can be overwhelming.

Awaiting test results, the fear of bad news, and living in uncertainty about the future is emotionally and psychologically challenging for PLWRD and their families. It can be disruptive to family life and relationships. Furthermore, the treatment decision processes may not adequately take into account the impact on daily life, potential side effects, or the impact on the overall quality of life of the patient and family.

STANDARDS

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.

8. Interdisciplinary Cooperation

Interdisciplinary cooperation is a vital component of psychosocial care. This can be achieved by integrating psychosocial personnel as core members of the medical team, and through enhancing medical care to be psychologically informed. This would enable the medical team members to provide information and advice in a sensitive manner that is tailored to the individual needs, disease stage and treatment, as well as early detection and access to preventative support when needed. This could reduce the impact of both the disease and the treatment on the individual, optimise their resources and safeguard their emotional wellbeing.

With over 70% of rare conditions having a genetic origin, genetic counsellors play a critical role in providing medical care, psychosocial support and case management (National Society of Genetic Counselors). Genetic counsellors should work closely with medical teams to support the family as a whole unit due to the hereditary nature of rare conditions.

For many PLWRD, issues around mental health are a primary characteristic or co-morbidity of their condition, and can have a significant psychological impact, presenting debilitating unmet needs, including neurodevelopment problems affecting intellectual abilities. Mild to severe psychiatric symptoms may not occur in early childhood or early on in the disease trajectory but appear later. It is critical to support and treat the whole person and not treat physical health and mental health separately. Access to psychiatric and psychological support is vital for many syndromes and it is critical that mental health practitioners have training to become 'rare aware' to understand the impact of rare conditions on mental health.

People living with an undiagnosed or rare condition may experience being dismissed by clinician services and specialists who mistake the rare condition for a psychosomatic disorder, leaving patients with a distrust of psychological support (Boettcher et al., 2020). Integration of psychological support as part of routine medical care for PLWRD would enable psychosocial support to be offered in a more acceptable and de-stigmatising way (Boettcher et al., 2020).

STANDARDS

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.

About this position paper

The co-creation process to develop this position paper started at EURORDIS Membership Meeting 2023 and continued through the engagement of EURORDIS Board of Directors, Council of National Alliances and EURORDIS Mental Health & Wellbeing Partnership Network via a series of online workshops. The position paper was developed from an evidence review of the published literature and by harnessing existing EURORDIS initiatives and various Rare Barometer studies, specifically:

- The material and evidence EURORDIS gathered as part of our advocacy in the development of the Commission's new Communication on a Comprehensive Approach to Mental Health has been used as a basis for this outline position paper.

- Consensus building exercises with EURORDIS Mental Health & Wellbeing Partnership Network to identify unmet needs and evidence and to formulate the recommendations.

- The paper is aligned with the holistic lens of PLWRD and the evidence and recommendations outlined in the EURORDIS position papers on Achieving Holistic Person-Centred Care to Leave No One Behind, with a focus on the psychosocial lens to specifically address the unmet mental health needs of PLWRD.

The recommendations related to the European and national actions and the evidence provided in the blueprint for psychosocial care are based on the existing publications and recommendations EURORDIS submitted to the Commission's call for evidence in Q1 2023.

The 8 domains and supporting standards are based on the PSAPOH Guideline on Psychosocial Care in Paediatric Oncology and Haematology and have been adapted to address the common needs experienced by people living a rare disease and their families.

This outline position paper represents a 'holding' position and will be revised and completed based on the results and population needs identified in EURORDIS Rare Barometer survey on mental health and wellbeing planned from end 2024 to 2025.

Annex II: References

Anderson M, Elliott EJ, Zurynski YA. *Australian families living with rare disease: experiences of diagnosis, health services use and needs for psychosocial support*. Orphanet J Rare Dis 2013;8:22. 10.1186/1750-1172-8-22.

Askins MA, Moore BD 3rd. *Preventing neurocognitive late effects in childhood cancer survivors.* J Child Neurol. 2008 Oct;23(10):1160-71. doi: 10.1177/0883073808321065. PMID: 18952582; PMCID: PMC3674758.

Baumbusch J, Mayer S, Sloan-Yip I. Alone in a Crowd? Parents of Children with Rare Diseases' Experiences of Navigating the Healthcare System. *J Genet Couns* 2019;28:80–90. 10.1007/s10897-018-0294-9.

Boettcher J, Denecke J, Barkmann C, Wiegand-Grefe S. Quality of life and mental health in mothers and fathers caring for children and adolescents with rare diseases requiring long-term mechanical ventilation. Int J Environ Res Public Health. 2020;17:8975. doi: 10.3390/ijerph17238975.

Castro R, Senecat J, De Chalendar M, et al. Client Group Rare Diseases. in: Amelung V.E., Stein V., Goodwin N., et al. eds. 2017. Handbook Integrated Care. Springer, Cham. 413-427. Retrieved January 4, 2019 from https://link.springer.com/chapter/10.1007%2F978-3-319-67144-4_32.

Courbier S, Berjonneau E. *Juggling care and daily life: The balancing act of the rare disease community.* EURORDIS – Rare Diseases Europe. A Rare Barometer survey, 2017.

Dellve L, Samuelsson L, Tallborn A, Fasth A, Hallberg LRM. Stress and well-being among parents of children with rare diseases: A prospective intervention study. J Adv Nurs. 2006;53:392–402. doi: 10.1111/j.1365-2648.2006.03736.x .

Dunkelberg S. A patient's journey: Our special girl. BMJ. 2006;333:430–1. doi: 10.1136/bmj.38937.455949.55.

Elliott EJ, Zurynski YA. Rare diseases are a 'common' problem for clinicians. Aust Fam Physician. 2015;44:630–3.

European Commission's Communication – Comprehensive Approach to Mental Health in All Policy Areas on 06 June 2023.

EURORDIS-Rare Diseases Europe. *Improve our experience of healthcare! Key findings from a survey on patients' and carers' experience of medical care for their rare diseases*. Rare Barometer (2021).

EURORDIS-Rare Diseases Europe. *The Voice of 12,000 Patients: Experiences and Expectations of Rare Disease Patients on Diagnosis and Care in Europe.* 2009.

EURORDIS-NORD-CORD Joint Declaration. *International Joint Recommendations to address the specific needs of Undiagnosed Rare Disease Patients* 2016.

Evans WRH, Tranter J, Rafi I, et al. How genomic information is accessed in clinical practice: an electronic survey of UK general practitioners. J Community Genet 11, 377–386 (2020). https://doi.org/10.1007/s12687-020-00457-5.

Faye F, Crocione C, Anido de Peña R, et al. *Time to diagnosis and determinants of diagnostic delays of people living with a rare disease: results of a Rare Barometer retrospective patient survey.* Eur J Hum Genet (2024). https://doi.org/10.1038/s41431-024-01604-z.

Flynn S, Vereenooghe L, Hastings RP, Adams D, Cooper SA, Gore N, Hatton C, Hood K, Jahoda A, Langdon PE, McNamara R, Oliver C, Roy A, Totsika V, Waite J. *Measurement tools for mental health problems and mental well-being in people with severe or profound intellectual disabilities: A systematic review.* Science Direct (2017) DOI: https://doi.org/10.1016/j.cpr.2017.08.006.

Genetic Alliance UK. Rare Experience 2020. *The lived experiences of people affected by genetic, rare and undiagnosed conditions.* Genetic Alliance UK; 2020.

Institute of Medicine (US) Committee on Psychosocial Services to Cancer Patients/Families in a Community Setting. Adler NE, Page AEK, editors. Cancer Care for the Whole Patient: Meeting Psychosocial Health Needs. Washington (DC): National Academies Press (US); 2008. 1, The Psychosocial Needs of Cancer Patients.

Kenny T, Bogart K, Freedman A, Garthwaite C, Henley S, Bolz-Johnson M, Mohammed S, Walton J, Winter K, Woodman D. *The Importance of Psychological Support for Parents and Caregivers of Children with a Rare Disease at Diagnosis. 2022* Rare Disease and Orphan Drugs Journal.

Kasparian NA, Rutstein A, Sansom-Daly UM, et al. *Through the looking glass: an exploratory study of the lived experiences and unmet needs of families affected by von Hippel-Lindau disease.* Eur J Hum Genet 2015;23:34–40. 10.1038/ejhg.2014.44.

Katon W, Lin EHB, Kroenke K. *The association of depression and anxiety with medical symptom burden in patients with chronic medical illness*. Gen Hosp Psychiatry 2007;29:147–55. 10.1016/j.genhosppsych.2006.11.005.

Kazak AE, Simms S, Alderfer MA, Rourke MT, Crump T, et al. (2005). *Feasibility and preliminary outcomes from a pilot study of a brief psychological intervention for families of children newly diagnosed with cancer.* Journal of Pediatric Psychology 30(8): 644-655.

Kazak AE. (2005). *Evidence-Based Interventions for Survivors of Childhood Cancer and Their Families.* Journal of Pediatric Psychology 30 (1) 29-39.

Knott M, Leonard H, Downs J. *The diagnostic odyssey to Rett syndrome: The experience of an Australian family.* Am J Med Genet. 2012;158A:10–2. doi: 10.1002/ajmg.a.34372.

Kole A, Hedley V. *Recommendations from the Rare 2030 Foresight Study. The future of rare diseases starts today.* EURORDIS – Rare Diseases Europe. 2021.

Lichtman JH, Froelicher ES, Blumenthal JA, Carney RM, Doering LV, Frasure-Smith N, Freedland KE, Jaffe AS, Leifheit-Limson EC, Sheps DS, Vaccarino V, Wulsin L. *Depression as a risk factor for poor prognosis among patients with acute coronary syndrome: systematic review and recommendations: a scientific statement from the American heart association.* Circulation 2014;129:1350–69. 10.1161/CIR.0000000000000019.

McConkie-Rosell A, Sullivan J. *Genetic counseling—stress, coping, and the empowerment perspective.* Journal of Genetic Counseling. 1999; 8:345–358. doi: 10.1023/A:1022919325772.

Mori Y, Downs J, Wong K, Anderson B, Epstein A, Leonard H. *Impacts of caring for a child with the CDKL5 disorder on parental wellbeing and family quality of life.* Orphanet J Rare Dis 2017;12:16. 10.1186/s13023-016-0563-3.

Muir E. The Rare Reality – an insight into the patient and family experience of rare disease. 2016. http://www.raredisease.org.uk/media/1588/the-rare-realityan-insight-into-the-patient-and-family-experience-of-rare-disease.pdf. Accessed 10 Nov 2016.

Nguengang Wakap S, Lambert DM, Olry A, et al. *Estimating cumulative point prevalence of rare diseases: analysis of the Orphanet database.* Eur J Hum Genet 28, 165–173 (2020). https://doi.org/10.1038/s41431-019-0508-0.

Onyeka TC. *Psychosocial issues in palliative care: a review of five cases.* Indian J Palliat Care. 2010 Sep;16(3):123-8. doi: 10.4103/0973-1075.73642. PMID: 21218001; PMCID: PMC3012234.

Palmer CGS, McConkie-Rosell A, Holm IA , LeBlanc K, Sinsheimer JS, Briere LC, Dorrani N, Herzog MR, Lincoln S, Schoch K, Spillmann RC, Brokamp E; Undiagnosed Diseases Network. *Understanding Adult Participant and Parent Empowerment Prior to Evaluation in the Undiagnosed Diseases Network.* J Genet Couns. 2018 Sep;27(5):1087-1101. doi: 10.1007/s10897-018-0228-6. Epub 2018 Mar 1.

Pelentsov LJ, Fielder AL, Laws TA, Esterman AJ. *The supportive care needs of parents with a child with a rare disease: Results of an online survey.* BMC Fam Pract. 2016;17:88. doi: 10.1186/s12875-016-0488-x.

Rare Minds 2020. *Data Impact Report 2020-21.*

Rice DB, Carboni-Jiménez A, Cañedo-Ayala M, Turner KA, Chiovitti M, Levis AW, Thombs BD. *Perceived benefits and facilitators and barriers to providing psychosocial interventions for informal caregivers of people with rare diseases: a scoping review.* Patient 2020;13:471– 519. 10.1007/s40271-020-00441-8.

Rohani-Montez C, Bomberger J, Zhang C, Cohen J, McKay L, Evans WRH. *Educational needs in diagnosing rare diseases: A multinational, multispecialty clinician survey.* Genetics in Medicine Open, Vo. 1, Issue 1, 2023. https://doi.org/10.1016/j.gimo.2023.100808.

Royal College of Psychiatrists and Centre for Mental Health. *Bridging the gap: the financial case for a reasonable rebalancing of health and care resources.* Centre for Mental Health, 2013.

Schröder HM, Lilienthal S , Schreiber-Gollwitzer BM , Griessmeier B. *Psychosocial Care in Paediatric Oncology and Haematology.* Arbeitsgemeinschaft der Wissenschaftlichen Medizinischen Fachgesellschaften (AWMF). Psychosoziale Arbeitsgemeinschaft in der Pädiatrischen Onkologie und Hämatologie (PSAPOH). 2008.

Simpson A, Bloom L, Fulop N, Hudson E, Leeson-Beevers K, Morris S, et al. *How are patients with rare diseases and their carers in the UK impacted by the way their care is coordinated? An exploratory qualitative interview study.* Orphanet J Rare Dis. 2021;16:76. doi: 10.1186/s13023-020-01664-6.

Smits RM, Vissers E, Te Pas R, Roebbers N, Feitz WFJ, van Rooij IALM, de Blaauw I, Verhaak CM. *Common needs in uncommon conditions: a qualitative study to explore the need for care in pediatric patients with rare diseases.* Orphanet J Rare Dis. 2022 Apr 4;17(1):153. doi: 10.1186/s13023-022-02305-w.

Spencer-Tansley R, Meade N, Ali F, Simpson A, Hunter A. *Mental health care for rare disease in the UK - recommendations from a quantitative survey and multi-stakeholder workshop.* BMC Health Serv Res. 2022 May 14;22(1):648. doi: 10.1186/s12913-022-08060-9.

Thompson AL, Young-Saleme TK. Anticipatory Guidance and Psychoeducation as a Standard of Care in Pediatric Oncology. Pediatr Blood Cancer. 2015 Dec;62 Suppl 5:S684-93. doi: 10.1002/pbc.25721. PMID: 26700925.

The King's Fund. *Long-term conditions and mental health: the cost of co-morbidities.* London: The King's Fund; 2012.

United Nations (UN) *Resolution on Addressing the Challenges of Persons Living with a Rare Disease and their Families.* Adopted by all 193 UN Member States. (RES/76/132, 2021)

Vereenooghe L, Flynn S, Hastings RP, Adams D, Chauhan U, Cooper SA, Gore N, Hatton C, Hood K, Jahoda A, Langdon PE, McNamara R, Oliver C, Roy A, Totsika V, Waite J. *Interventions for mental health problems in children and adults with severe intellectual disabilities: a systematic review.* MBJ Journal 8:6 (2018) DOI: http://orcid.org/0000-0001-9772-1151.

Wallerstein N. *What is the evidence on effectiveness of empowerment to improve health?* 2006. Copenhagen, WHO Regional Office for Europe (Health Evidence Network report).

Witt S, Schuett K, Wiegand-Grefe S, et al. *Living with a rare disease - experiences and needs in pediatric patients and their parents*. Orphanet J Rare Dis 18, 242 (2023). https://doi.org/10.1186/s13023-023-02837-9World Health Organization & Calouste Gulbenkian Foundation. Social Determinants of Mental Health. Geneva, WHO, 2014.

Wu AC, McMahon P, Lu C. Ending the diagnostic odyssey—is whole-genome sequencing the answer? JAMA Pediatr. 2020;174(9):821–822. doi: 10.1001/jamapediatrics.2020.1522.

Yang G, Cintina I, Pariser A, Oehrlein E, Sullivan J, Kennedy A. *The national economic burden of rare disease in the United States in 2019.* Orphanet J Rare Dis. 2022 Apr 12;17(1):163. doi: 10.1186/s13023-022-02299-5.

Zurynski Y, Frith K, Leonard H, Elliott E. *Rare childhood diseases: How should we respond?* Arch Dis Child. 2008; 93:1071–4. doi: 10.1136/adc.2007.134940.

Zurynski Y, Deverell M, Dalkeith T, Johnson S, Christodoulou J, Leonard H, Elliott EJ; APSU Rare Diseases Impacts on Families Study Group. *Australian children living with rare diseases: experiences of diagnosis and perceived consequences of diagnostic delays.* Orphanet J Rare Dis. 2017 Apr 11;12(1):68. doi: 10.1186/s13023-017-0622-4

EURORDIS
RARE DISEASES EUROPE

EDITORIAL COMMITTEE EURORDIS

EURORDIS-RARE DISEASES EUROPE

Plateforme Maladies Rares ◆ 96 rue Didot
75014 Paris ◆ France

EURORDIS BRUSSELS OFFICE

Fondation Universitaire ◆ Rue d'Egmont 11
1000 Brussels ◆ Belgium

EURORDIS BARCELONA OFFICE

Recinte Modernista Sant Pau ◆ Pabellón de Santa Apolonia
Calle Sant Antoni Mª Claret 167 ◆ 08025 Barcelona ◆ Spain

EURORDIS.ORG

This paper was produced thanks to funding received under an operating grant from the European Union's Health Programme (2014-2020).

Co-funded by the Health Programme of the European Union