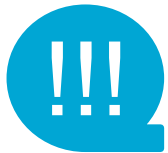


# New Advocate Topic: Rare Diseases & Mental Health



# New EC Health Priority Areas



- **Mental health is now being 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, it is imperative that rare diseases are seen as a vulnerable population requiring specific attention in the new EU Action Plan on Mental Health to be presented in June 2023.



- **Rare diseases are complex multi-system conditions requiring a holistic approach to meet their health and psycho-social related needs.** For many rare diseases, mental health, learning disability and behavioural challenges are common associated co-morbidities.



- The rare disease patient community have highlighted the increased psychological impact and the associated mental, learning and behavioural co-morbidities as being a significant and debilitating unmet need affected not only the PLWRD but also the family and caregivers.

# Rare Diseases & Mental Health

## Psychological Impact along the Patient Journey:

- Undiagnosed disease and impact of the diagnostic odyssey
- At diagnosis
- Living with a Rare Disease (groupings of common characteristics):
  - Functional Impairment (BBS, Rare Epilepsies, RETT)
  - Progressive diseases (MND, SMA)
  - Visible disabilities (NF1, Craniofacial Anomalies)
  - Surveillance (Genetic Tumours Risk Syndromes; Rare Cancers)



MH affecting the person living with a rare disease & the wider family – partner, parents, siblings and carers

## Risk Factors to Mental Wellbeing through increase complexity, rarity, progression, prognosis, visible and invisible disabilities:

- Isolation
- Digitalization
- Age and gender inequalities
- Impacting on education work and independent living
- Reduced economic capacities

# Objectives



**Short-term objectives:** For people living with a rare disease, and their family and carer givers, to be:

- Recognized as a vulnerable group with a higher risk of develop symptoms of mental disorders and associated increased psychological impact to be included in education, work and society.
- Included in the new EU Action Plan on Mental Health to address unmet needs of people living with a rare disease and their families/relatives/carers, across a wide range of rare diseases and across countries.



**Long-term objectives:**

- the psychological health of people affected by rare diseases are considered at each stage of their life journey
- the simple effective social and psycho-social interventions are implemented to prevent the costly down stream health system impacts of unmanaged psychological burdens.

# Gathering Evidence

Spencer-Tansley et al. BMC Health Services Research 2022, 22:449  
https://doi.org/10.1186/s12913-022-08600-9

BMC Health Services Research

RESEARCH Open Access

## Mental health care for rare disease in the UK – recommendations from a quantitative survey and multi-stakeholder workshop

Rosa Spencer-Tansley, Nick Meade, Farhana Ali, Amy Simpson and Amy Hunter\*

**Abstract**  
**Background:** Rare disease patients and carers report significant impacts on mental health. In the UK, 100k have focused on relatively few, specific conditions. Care burden, with an estimated 3.5 million affected individuals in the UK.  
**Method:** We explored the impact on mental health of living with a rare condition through multiple choice questionnaires. The survey assessed the impact on those affected by a rare condition through multiple choice questionnaires. Through a multi-stakeholder workshop that involved lay clinicians and a government advisor, we developed recommendations and an integrated approach.  
**Results:** Eligible responses came from 1231 patients and 564 respondents (50% had felt worried/anxious, stressed, and 10% of carers had had suicidal thoughts). Challenges that are particular to rare conditions and which need the attention of healthcare professionals (88%). Only 23% of respondents felt healthcare professionals consider mental health. Almost half reported never having been asked about mental health access to, and appropriateness of, professional psychological support with rare disease services.  
**Conclusion:** Living with a rare disease substantially impacts mental health. Our recommendations are for healthcare professionals to address patients' and carers' mental health needs, and for psychological support with rare disease services.  
**Keywords:** Rare disease, Undiagnosed, Mental health, Health

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**Rare Disease and Orphan Drugs Journal** Open Access

## The importance of psychological support for parents and caregivers of children with a rare disease at diagnosis

Thomas Kenney<sup>1</sup>, Kathleen Bogart<sup>2</sup>, Albert Freedman<sup>3</sup>, Claire Garthwaite<sup>4</sup>, Susie M. D. Henley<sup>5</sup>, Matt Boto-Johnson<sup>6</sup>, Shamsi Mohammed<sup>7</sup>, Jill Walker<sup>8</sup>, Ryan Wintler<sup>9</sup>, Deborah Woodman<sup>10</sup>

**Abstract**  
Rare diseases are complex and difficult to diagnose, with parents and caregivers often reporting significant delays in receiving a definitive diagnosis. Following diagnosis, parents and caregivers often feel overwhelmed with emotions, including relief, guilt, and shock. The culmination of this emotional burden may lead to a deterioration of psychological health, ultimately reaching a stage where the parents struggle to cope. A systematic literature review was conducted of the articles on this topic by searching the electronic databases, PubMed. Further studies were retrieved from a reference listing of relevant articles and consultation with experts in the field. The review was based on the guidance in the Preferred Reporting Items for Systematic Reviews and Meta-Analyses protocol. The initial search identified 1216 articles, of which 37 met the inclusion criteria and were included in this review. The literature revealed key factors that appeared to contribute to psychological stress, including prolonged diagnostic odyssey, poor diagnostic delivery, lack of information and specialist knowledge, and convoluted healthcare

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[www.rdojournal.com](http://www.rdojournal.com)

Call to National Alliances, and your Members, for evidence on rare diseases and mental health:

- Please share any publications, surveys, workshop reports
- Send to [matt.johnson@eurordis.org](mailto:matt.johnson@eurordis.org)

Call for leads from National Alliances who have been active in MH

**\*Deadline: end of January 2023 (preferable earlier)**

# New EURORDIS Mental Health Advocates Network

EURORDIS Board supports the **need to implement a specific strategy on mental health and wellbeing for people living with a rare disease, their family and caregivers** to achieve the following objectives:



- Support and reinforce a **united** and **empowered** rare disease community affected by mental health to **come together and be seen, learn and supported each other**;

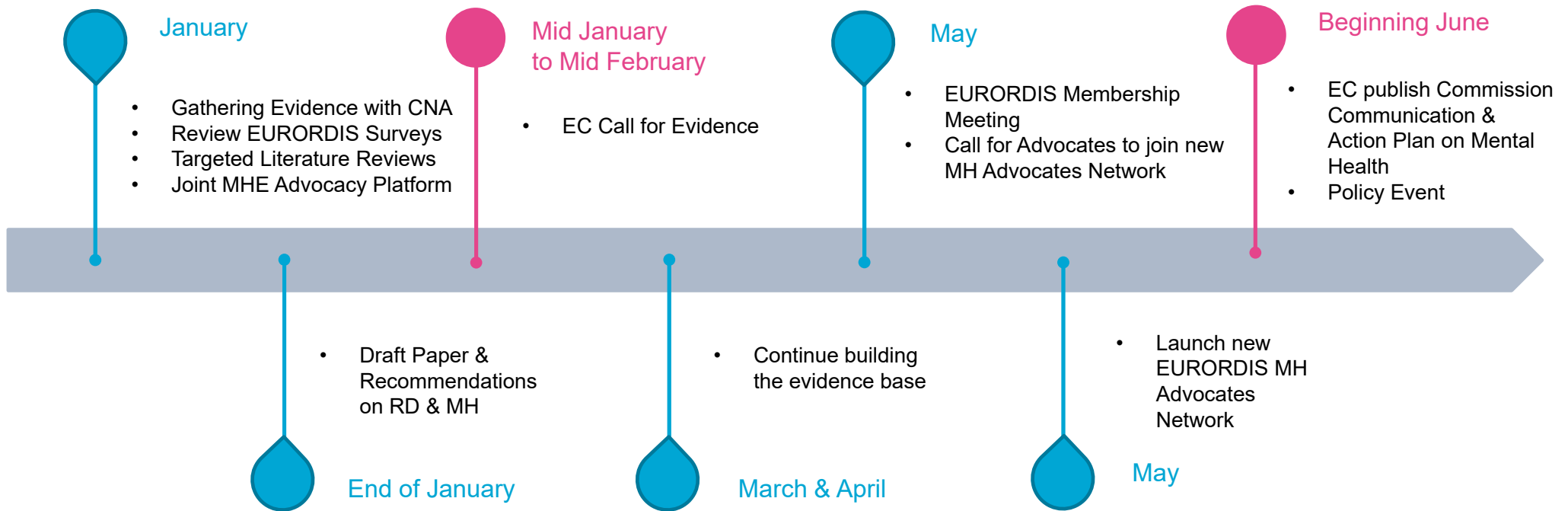


- **Better support and integrate mental health in the all policy areas** and initiatives carried out by EURORDIS for all rare diseases;



- **Help increase visibility of the psychological impact and associated co-morbidities** of mental health, learning difficulties and behavioural disorders in:
  - the world of rare diseases and rare cancers
  - the world of mental health

# Timeline and milestones – Q1-2 2023



# Rare Diseases & Mental Health

PLWRD are exposed to increased inequalities due to the intersections between living with a rare disease, disability and mental health disorder, that impact on all aspects of daily lives and affect the ability to earn a living and live independently and affecting their ability to be active participant in education, work and society.

- Rare disease of often **diagnosed late and after many delays**. Impact of the diagnostic odyssey.



- Receiving a definitive diagnosis, parents often feel **initial relief**, which is soon **replaced by overwhelming feelings of stress and guilt**, that manifests through a range of psychological symptoms.

- **Progression** of disease, living with a **visible disabilities, functional impairment** and poor **prognosis** and the impact due to ongoing **surveillance**.