

Handout: Tailoring Rehabilitation to also fit Rare Disease Populations: Challenges and Solutions

Authors from The Norwegian Centre for Rare Diseases, Sunnaas Rehabilitation Hospital:

Ariane Kwiet, dr.med., Specialist in Physical Medicine and Rehabilitation

Anne-Mette Bredahl, PhD, Clinical Psychologist

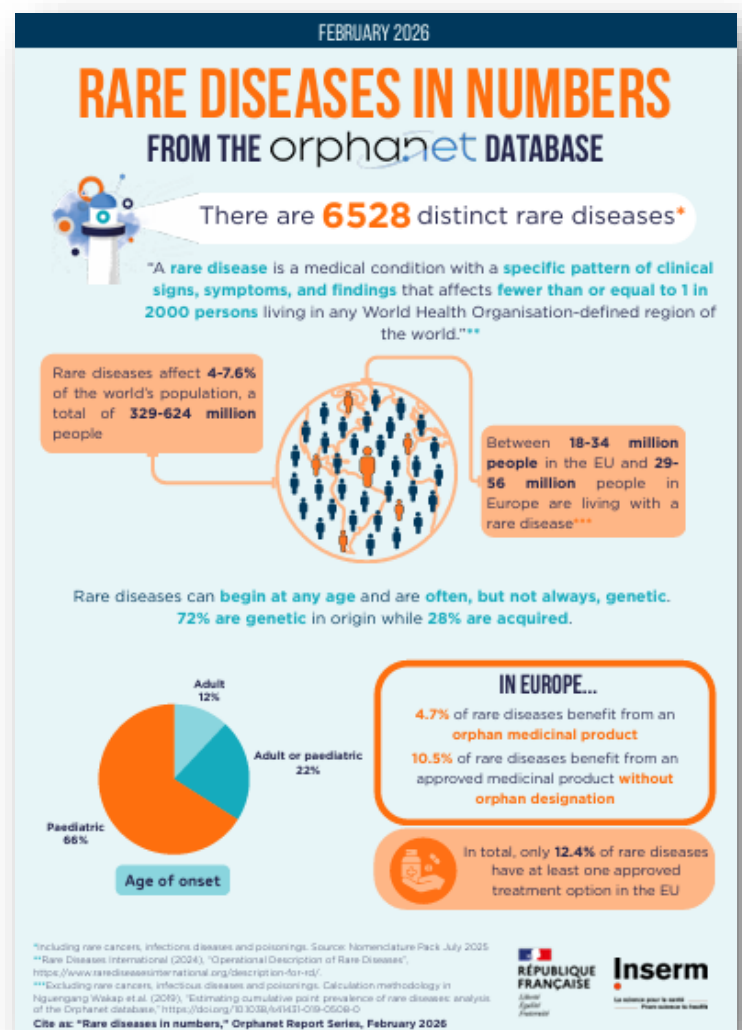
Trine Bathen, MSc, Occupational Therapy Specialist

What are rare diseases?

- Different definitions are used:
 - US: The disease must impact fewer than 200.000 individuals
 - Europe: Life-threatening or chronically debilitating disease with such a low prevalence that special combined efforts are needed to address it (fewer than 1 in 2.000 people)
- Between 6.000 – 8.000 known rare diseases worldwide.
- Rare Diseases affect 4% of the world's population, a total of between 3 to 6,5 million people
- High burden of symptoms with high variability – often chronic, progressive and life-threatening in some cases
 - Often affects multiple organ systems
 - Acute and chronic pain and fatigue often present
 - High psychosocial burden
 - Delayed diagnosis creates stress
 - Reduced work ability
 - Social isolation and stigma
 - Reduced quality of life

References

- [RD in numbers 2026 factsheet from the Orphanet database](#)
- [What is a rare disease? - EURORDIS-Rare Diseases Europe](#)





Barriers in health care systems

- Lack of diagnosis-related knowledge in health care workers
- Limited targeted therapies (and often expensive)
- Treatment development challenges due to small populations
- Limited multidisciplinary care
- Limited possibility to meet others with similar diagnoses



Rare disease knowledge resources

- [Orphanet](#), an international rare disease consortium
- [Genetic and Rare Diseases Information Centre GARD](#). U.S. Dep. Health & Human Services
- [European Reference Networks - Public Health - European Commission](#)



Rare diseases and rehabilitation

- Many patients with rare diseases have unmet rehabilitation needs
- Main challenges are:
 - Lack of information and coordination
 - Lack of knowledge among professionals
 - Little opportunity to meet others with similar diagnoses



Essential partners in building effective rare disease rehabilitation models

- It is essential to include all relevant stakeholders when developing new rehabilitation models for patients with rare diseases
 - Municipal rehabilitation services
 - Specialized rehabilitation services
 - Rare Disease Specialists
 - Patient Associations
 - Rehabilitation Coordinators
 - General Practitioners
 - Health System Planners



Possible solutions

- Multidisciplinary care team with integrated psychosocial support and access to expertise
- The use of e-health can be a solution to have access to expertise
- Coordinated care
- Integrating groups of patients with a rare disease into existing rehabilitation services with slightly adapting the program can be a feasible way to use existing structures
- Group rehabilitation to enhance the possibility of peer effect



Contact information

Ariane Kwiet, arikwi@sunnaas.no

Trine Bathen Trine.Bathen@sunnaas.no

Anne-Mette Bredahl chrann@sunnaas.no

Webpage:

The Norwegian Centre for Rare Diseases, unit Sunnaas: [The Norwegian Centre for Rare Diseases - Sunnaas Rehabilitation Hospital](#)

Ongoing projects on rehabilitation in patients with rare bone diseases

- Project page: [A systematic review about the current evidence about effective rehabilitation for children with rare bone diseases](#)
- Project page: [Pain rehabilitation of patients with a skeletal dysplasia diagnosis – how to take care of their special needs and use existing programs at the same time](#)



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