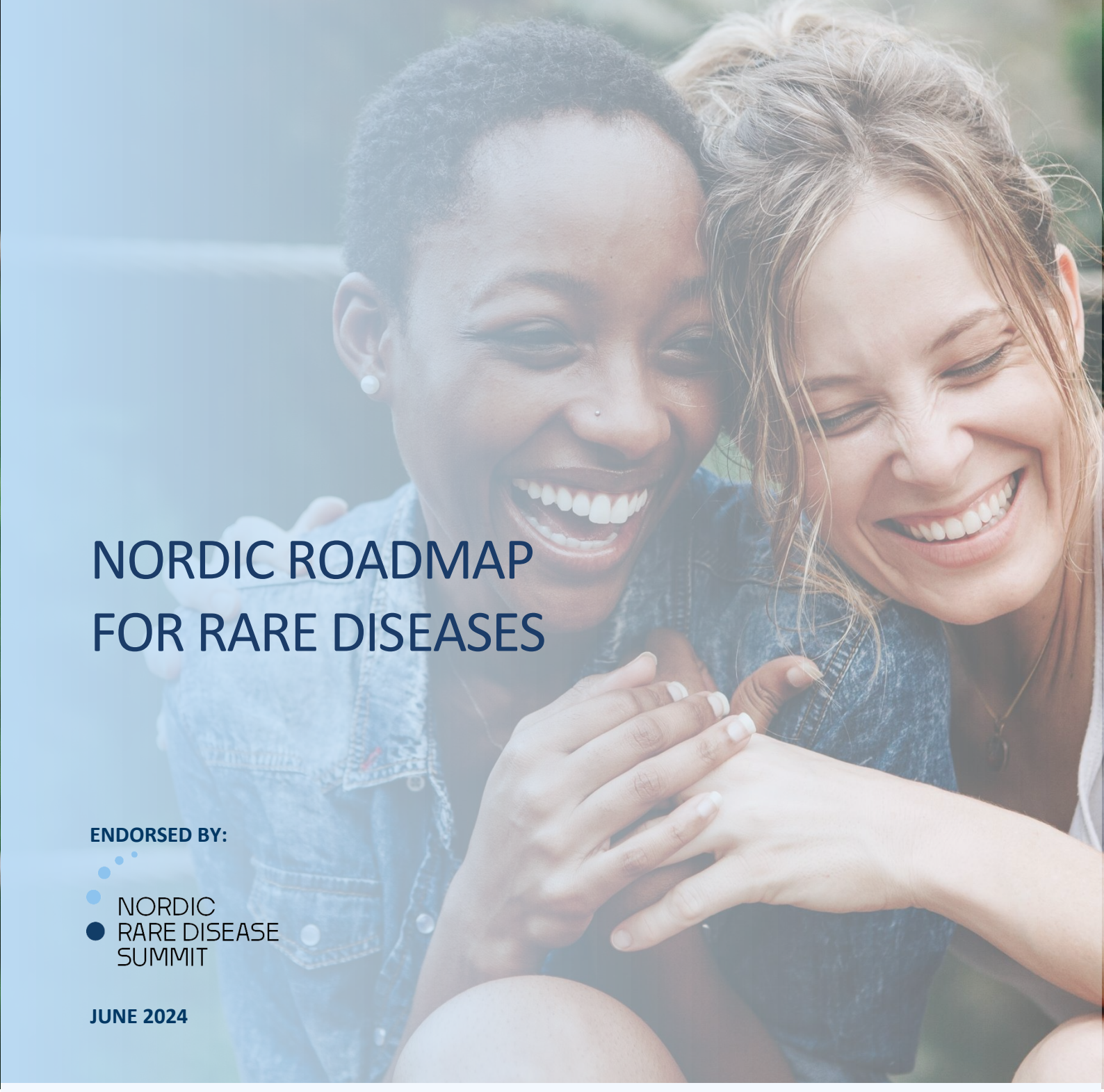


NORDIC ROADMAP FOR RARE DISEASES

ENDORSED BY:

NORDIC RARE DISEASE SUMMIT

JUNE 2024

THE NORDIC RARE DISEASE SUMMIT 2023

was organised in Stockholm on 17 April 2023 by the pharmaceutical company Takeda Pharma with contributions from Alexion AstraZeneca Rare Disease, Novo Nordisk, and Sobi. Additional partners were Rare Diseases Sweden, Lif - the research-based pharmaceutical industry in Sweden, EURORDIS - Rare Diseases Europe, SBONN – a network of six umbrella patient organisations for rare diseases in five Nordic countries, EUCOPE - The European Confederation of Pharmaceutical Entrepreneurs, and EFPIA - The European Federation of Pharmaceutical Industries and Associations.

AUTHORS OF THIS ROADMAP:

Authorship of the Nordic Roadmap lies with multiple stakeholders. The partners behind the Nordic Rare Disease Summits in 2021 and 2023 developed the foundation of this Nordic Roadmap, and contributions to the content derive from presentations and statements from speakers, panelists, and participants, as well as round tables before and thematic working group meetings after the latest summit in 2023. Before finalisation, the content of the roadmap was discussed and validated through conversations with experts from across the Nordics. As a result of the collaborative effort, no organisation or individual should be made independently responsible for, or be cited to support, all recommendations and measures of the Nordic Roadmap.

Introduction

Despite the label of 'rare', around one million people in the Nordics are living with a rare disease.[1] Nevertheless, many people affected by rare diseases, as well as their families and caregivers, face significant challenges in receiving a timely and accurate diagnosis, and securing access to innovative medicines and appropriate medical and social care, often sharing a common experience of feeling marginalised.

The Nordics, consisting of Denmark, Finland, Iceland, Norway and Sweden, is a region with a favorable ecosystem to become a frontrunner in rare disease care. There are good public registries, biobanks and genome centers, which enable individuals to manage their own health data, while also promoting the use of data for enhanced healthcare delivery, research, innovation and policy making. There is further a thriving, innovative life science sector with a dedicated patient community, world-leading experts and highly skilled health care professionals who have a long tradition of working in partnerships across disciplines and functional areas. The Nordics have the added benefit of comparable health care systems, facilitating cross-border collaboration for those seeking care and the right way to address opportunities and challenges of rare diseases.

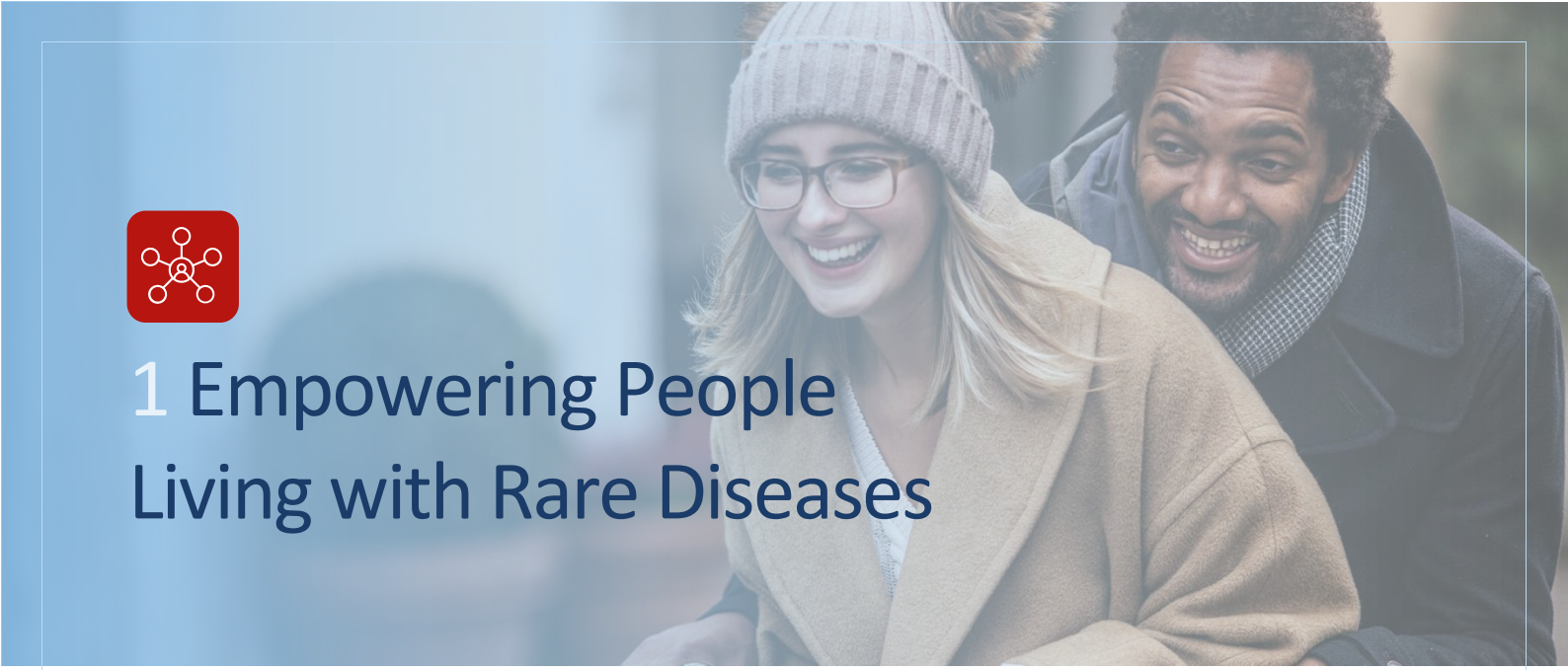
In line with the spirit of this cross-border collaboration was the Nordic Rare Disease Summit which took place in Stockholm in early 2023. This Summit, as well as its predecessor in 2021, calls for a Nordic Rare Disease Roadmap, incorporating vital insights and experiences of people living with rare diseases. This roadmap aims to cement and drive the Summit's long-term vision and commitment to improved quality of life, care, treatment and patient outcomes for individuals affected by rare diseases as well as their caregivers.

Therefore the 2024 Nordic Rare Disease Roadmap is a shared call to action, exploring four critical aspects: (i) empowering people living with rare diseases, (ii) early diagnosis, (iii) patients' access to innovative medicines, and (iv) the development of impactful Nordic rare disease strategies. We, the partners of the Nordic Rare Disease Summit, have contributed to a set of recommendations with measurable outcomes aiming to enhance accountability, enable monitoring, and track the progress of these aspects.

We acknowledge that important steps have already been taken by several Nordic countries through the development of national rare disease strategies, and by the Nordic Council of Ministers through the establishment of the Nordic Network for Rare Diseases. We now call on the Nordics to build on this momentum and further strengthen cross-country collaboration.

All stakeholders who have jointly contributed to this roadmap share the aspiration to improve the quality of life for people living with rare diseases in the Nordics. We share a vision of making the Nordics a global frontrunner in rare disease care and to make this a reality will require a strong commitment from all key partners of the Rare Disease Summit including patient organisations, industry, policymakers, academia, and healthcare professionals.

1 Centrum för sällsynta diagnoser (CSD), Karolinska Universitetslaboratoriet



1 Empowering People Living with Rare Diseases

CONTEXT

Empowerment has been described by the World Health Organization (WHO) as a 'prerequisite for health' and an enabler of 'improved health outcomes' and quality of life. According to the WHO, empowerment refers to the process by which people gain control over the factors and decisions that shape their lives. It is the process by which they increase their assets and attributes and build capacities to gain access, partners, networks and a voice, in order to gain control[2].

Due to the rarity, high complexity, lack of research and understanding, and the often poor quality of life of people affected by rare diseases, the process of empowerment is particularly critical. People living with rare diseases as well as their close ones are often experts in their field, holding unique knowledge of the rare disease that is not available in the health care system. Therefore, society needs structures to incorporate knowledge from people living with rare diseases and their organisations, in partnership with health care and other relevant professionals.

As suggested by EURORDIS, a non-profit alliance of over 1000 rare disease patient organisations, the expertise of people living with a rare disease should be recognised and promoted as a resource on different levels of healthcare systems: At the micro level, empowerment of the individual can serve as a resource for people living with a rare disease and their close ones, in order to gain greater control over decisions and actions concerning their health and wellbeing. At the community level, empowerment means that people living with rare diseases and their close ones get involved in shaping structures providing care and social services to improve their quality of life. Finally, at the policy level, empowerment can contribute to the development of sustainable and resilient healthcare systems, improving the use of health services, and leading to overall productivity gains.

Living with a rare disease affects all aspects of life. The approach to empowerment should be holistic. So, when planning and carrying out social support, independent living, education and more, inclusion of the experiences and expertise of people living with rare diseases, their close ones, and their organisations, must be taken seriously.

CURRENT CHALLENGES

Today, we are still far from empowering people living with rare diseases in decision and policy shaping on all three levels. Instead, we continue to face a range of barriers, from inconsistent definitions of patient involvement and empowerment to a lack of inclusion, education, training, structures, procedures, and funding. More can be done to empower people living with a rare disease and their organisations.

RECOMMENDATIONS

1. At the individual level

It is essential to implement a personalised and person-centric approach in our healthcare systems, which is tailored to the individual's specific needs and preferences, in order to promote communication between the person living with a rare disease and the health care provider. Supporting health literacy, education, and skill-building can allow for better self-management and reduce health inequalities and

[2] https://www.who.int/teams/health-promotion/enhanced-wellbeing/seventh-global-conference/community-empowerment

treatment errors. In addition, it is essential to enable shared decision-making to further integrate patients' views on managing their disease or diseases. Due to the inequalities present in health care provided to people living with rare diseases today, it is essential to equip them with accessible and simple support systems and cross-border tools and frameworks according to their legal rights.

These goals can be achieved through specific information, tools, guidelines, trainings, and campaigns aimed at healthcare professionals and patients coupled with sufficient funding and resources. In addition, data-driven healthcare can further empower people living with a rare disease by providing them access to their health data, ensuring increased ownership and enabling tracking progress of their conditions.

To unify these efforts across the region, setting Nordic guidelines with a common definition or standard for patient empowerment could prove beneficial in enhancing the quality of person centered care and ensuring consistent support regardless of the country of residence. Finally, further integrating education on rare diseases – particularly on diagnosis and patient care as part of the healthcare education curriculum - can equip the next generation of healthcare professionals with the knowledge, skills, and empathy needed to care for all patients effectively, including those living with rare diseases.

2. At the community level

It is important to enable people living with rare diseases and their organisations to speak with a strong common voice. To this end, the formation of mutual support groups, information-sharing networks, and coalitions is essential. Patient organisations should take the role of educating people living with a rare disease and healthcare professionals to better understand the scientific, medical and policy landscape, and act as a single voice to pool patients' views and facilitate the exchange of best practices. With this in mind, patient organisations should be systematically included in defined healthcare-related policy and decision-making. In addition, these organisations should work closely together with universities, researchers, and other key stakeholders for the co-creation of clinical studies and trials. An example to be followed is the consistent participation of European Patient Advocacy Groups (ePAGs) in the European Reference Networks (ERNs).

These goals can be achieved through sustainable financial support to relevant initiatives and patient advocacy groups to help develop capacity-building programs including helplines, websites and ad hoc trainings. An example is the European Patient's Academy on Therapeutic Innovation (EUPATI) which provides patients with the knowledge and skills to get involved in medical research and development, and secures education of researchers on how to engage with patients.[3] In addition, patient organisations should be invited to participate in the development of innovative healthcare solutions. This would include assistance by governments and international organisations with trainings on how to make use of funds and other tools. Finally, a key recommendation is to improve cross-border and Nordic collaboration between governments, associations and individuals through data and knowledge sharing.

3. At policy level

It is crucial that rare disease patient organisations engage with decision-makers in early dialogue to exchange best practices and coordinate policy solutions.

These goals can be achieved through early and structured dialogues, consultations, interviews and focus groups, as well as a system of appraisals and patient evaluation and feedback. For example, patient representatives should be systematically and more thoroughly involved in all processes in the development, monitoring and revision of health policies and national strategies. This could include becoming partners in drafting and implementing rare diseases strategies and other relevant health policies at local, national or European level. Patient representatives should also receive adequate training, guidance, support, and financial resources to contribute to the legislative process. To enable this, the EU4Health programme should entail a specific budget for patient involvement in regulatory processes. People living with a rare disease should also be represented in steering committees and advisory boards of relevant organisations and multi-stakeholder groups. Patient representatives can also be invited to medical and policy conferences as well as participating in media opportunities to further spread awareness. Finally, there should be clear and accessible ways for patients' experiences to be considered during the national and EU health technology assessment (HTA) processes.

3 European Patients' Academy on Therapeutic Innovation (EUPATI). (n.d.). About us. Retrieved from https://eupati.eu/about-us/

2 Early Diagnosis

CONTEXT

People living with a rare disease often face challenges in obtaining a diagnosis. On average it takes six to eight years before a correct diagnosis is made [4] and more than 40% of patients are misdiagnosed at initial presentation. [5] In addition, 70% wait more than one year for a confirmed diagnosis after seeking medical attention.[6]

A delay in diagnosis means that patients receive inadequate treatment, medical care and social support, potentially causing a deterioration of their condition and decreasing their overall quality of life. In the case of hereditary diseases, a late diagnosis can lead to delayed diagnosis also in younger siblings or family members. Importantly, a late diagnosis often entails psychological strain and suffering for the person living with rare diseases and their loved ones as well as productivity loss and a financial burden for the individual and the healthcare system as a whole.[5]

CURRENT CHALLENGES

A key obstacle for early diagnosis is the small population of individuals living with a specific disease in terms of presentation and cause. This challenge is combined with the limited awareness and scientific knowledge amongst healthcare professionals of each of the thousands of rare diseases, [7] further compounded by the limited availability and sometimes high cost of diagnostic tests. Fundamentally, most physicians will only see a small number of people with a specific rare disease which makes it difficult to build up the necessary clinical knowledge and experience to recognise the symptoms. There is a need to improve cooperation and pooling of knowledge across expertise areas and country borders. In addition, pathways for admittance of patients with a suspected rare disease from primary to specialised care is often unsystematic, risking to further prolong the journey towards diagnosis. Consequently, patients may spend years moving from physician to physician, undergoing a large number of diagnostic tests, and they are often faced with multiple misdiagnoses before eventually obtaining a correct one.

RECOMMENDATIONS

Receiving a timely and accurate diagnosis is of vital importance to people living with a rare disease. It facilitates access to correct treatment, provides a possible prognosis, and enables a connection to a support community, patient organisation or other network. An accurate diagnosis at an early stage can also help to identify other family members who may be at risk of developing the same disease.

To address this goal of early diagnosis, several recommendations can be implemented including raising awareness of rare diseases, improving education of health care professionals, strengthening the organisation of

4 Martínez-deMiguel, C., Segura-Bedmar, I., Chacón-Solano, E., & Guerrero-Aspizua, S. (2022). The RareDis corpus: A corpus annotated with rare diseases, their signs and symptoms. Journal of biomedical informatics, 125, 103961. https://doi.org/10.1016/j.jbi.2021.103961

5 Vandeborne, L., van Overbeeke, E., Dooms, M., De Beleyr, B., & Huys, I. (2019). Information needs of physicians regarding the diagnosis of rare diseases: a questionnaire-based study in Belgium. Orphanet journal of rare diseases, 14(1), 99. https://doi.org/10.1186/s13023-019-1075-8

6 EURORDIS - Rare Diseases Europe. (n.d.). Earlier, faster and more accurate diagnosis. EURORDIS. Retrieved from https://www.eurordis.org/our-priorities/diagnosis/

7 Benito-Lozano, J., López-Villalba, B., Arias-Merino, G., Posada De la Paz, M., & Alonso-Ferreira, V. (2022). Diagnostic delay in rare diseases: data from the Spanish rare diseases patient registry. Orphanet Journal of Rare Diseases, 17(1), 418.

standardised care pathways, and increasing access to innovative diagnostics as well as genetic counseling.

First, awareness and knowledge of rare diseases should be raised amongst patients, all primary care and social care professionals and specialists, and the public to avoid delayed diagnosis and improper treatment. One approach is through adopting Nordic medical guidelines and best practice recommendations on diagnosis and screening. Other initiatives to improve awareness include the celebration of Rare Diseases Day (28/29 February) as well as the creation of a Nordic Rare Platform that allow people living with rare diseases, professionals and advocates to connect and share information, resources, and best practices. A notable example is the National Advisory Unit on Rare Disorders (NKSD) in Norway.[8] Nordic countries should allocate financial resources towards technical expertise and infrastructure enabling accessible information hotlines for healthcare professionals and patients.

Second, strengthening the development of standardised care pathways can clarify admittance routes between primary care and specialist care, to ensure knowledge transfer between healthcare providers and a patient journey with clear medical ownership. Furthermore, standardised care pathways can guide appropriate scaling up of diagnostic tools, such as whole-genome sequencing and genomic guidance.

Overall, appropriate funding should be dedicated at Nordic level to improve national surveillance mechanisms and registries to gather sufficient information to support research. In addition, internationally harmonised coding system for rare diseases such as OrphaCodes should be implemented – allowing pooling of data through harmonised data entry (preferably across national borders). This data would be supported through establishing quality registries on a national, regional, and international level that can deal with the heterogeneity observed across rare diseases.

Beyond this, access and use of advanced diagnostics in hospitals should be expanded and sufficiently funded to increase the capacity across the Nordics. To shorten the journey towards accurate and timely diagnosis, it is necessary to invest in pioneering diagnostic platforms and exploit digital innovations across all Nordic countries. Methods used within precision medicine are particularly suitable for diagnosing and treating rare diseases and can lead to overall better outcomes by giving the right treatment to the right person at the right time. More specifically, utilising i.e. genomic data can lead to a quicker diagnosis, tailored treatments, reduced expenses, more effective clinical trials, and assist in the development of novel and personalised medicines.

Nevertheless, timely diagnosis also requires timely action, and neo-natal screening programs are effective tools to this end, allowing the identification of diseases as early as possible. These programmes should be expanded, standardised, and offered systematically. An example to be followed is the State Serum Institute (SSI) in Denmark which has recently announced that newborns will be screened for an additional 6 rare diseases, bringing the total to 25 rare diseases now included in the program.[9] Finally, another key example is Screen4Care which uses the power of digital solutions combined with newborn screening programmes to improve diagnosis.[10]

EU LEVEL RECOMMENDATIONS

Finally, advancing diagnosis also relies on innovative approaches, where technology innovators, patient advocates, healthcare providers, and researchers beyond the Nordics cooperate to tackle the challenge of rare disease diagnostics. This cooperation can be supported by European Reference Networks (ERNs) – which connects doctors, researchers and patient advocates virtually across the European Union so that expertise travels rather than the patient. These networks can serve as knowledge hubs where healthcare professionals can find the right information at the right time and can provide doctors with a structure for efficiently sharing experiences and knowledge.

To reduce the diagnostic 'odyssey' and fully benefit from the breadth of knowledge provided by these networks, it is critical to ensure that Nordic university hospitals and research centres contribute to existing ERNs, and that the ERN's resources, working methods, and information network become an incorporated part of the Nordics' healthcare systems. Individuals must be referred to the most relevant national specialist centres where ERNs could help facilitate a diagnosis.[11] A flagship action is the European Commission's Joint Action on the integration of ERNs into national healthcare systems (JARDIN).[12]

8 Oslo University Hospital. (2023). National Advisory Unit on Rare Disorders. Retrieved from https://www.oslo-universitetssykehus.no/fag-og-forskning/nasjonale-og-regionale-tjenester/nasjonal-kompetansetjeneste-for-sjeldne-diagnoser/national-advisory-unit-on-rare-disorders

9 Statens Serum Institut. (2024). Nyfødte screenes nu for 25 alvorlige sygdomme. SSI. https://www.ssi.dk/aktuelt/nyheder/2024/nyfoedte-screenes-nu-for-flere-alvorlige-sygdomme

10 Screen4Care. (n.d.). What is Screen4Care? Retrieved from https://screen4care.eu/the-project/screen4care

11 EURORDIS - Rare Diseases Europe. (2020). Rare2030 Recommendations. EURORDIS. https://download2.eurordis.org/rare2030/Rare2030_recommendations.pdf

12 European Commission. (2023). European Reference Networks: a flagship EU action for patients with rare diseases. Health and Food Safety. https://ec.europa.eu/newsroom/sante/items/805612/en

3 Patients' Access to Innovation

CONTEXT

Rare diseases typically have few or no treatment options and people living with a rare disease tend to be underserved both scientifically and clinically. The latter often results in a delay in treatment, should one exist. Additionally, people with rare diseases face an impaired access to the innovative medicines that do exist as only 39% of orphan medicinal products are commercially available across Europe, with rates in the Nordics varying between 69% (Denmark), 41% (Sweden), 34% (Norway and Finland) and 20% (Iceland).[13]

CURRENT CHALLENGES

The root causes of lack of treatment, access inequality and delay are often multifactorial, ranging from low levels of knowledge and resources, and slow regulatory processes to late initiation of health technology assessments (HTA), duplicative evidence requirements, delays in pricing and reimbursement procedures, as well as local formulary decisions. In the Nordic region, the national health technology assessments are not always adapted to accommodate the specific challenges associated with treatments for rare disease.[14] These challenges include small populations, a lack of validated outcome parameters, large heterogeneity in patient populations, limited knowledge of the diseases' natural course as well as short follow-up periods in clinical studies. Consequently, there is often a limited amount of clinical evidence. Moreover, the rarity of some diseases could also be the reason for a potential lack of attention and investment, resulting in a limited understanding of potential underlying mechanisms. Taken together, the uncertainties surrounding the benefits of a certain treatment for rare diseases mean that cost-effectiveness analyses are often associated with a higher degree of uncertainty compared to other, more common, treatments. To conclude, the current methods of evaluating cost effectiveness are not fully suitable for assessing treatments for rare diseases. It is important to note that availability of treatments for rare diseases can also be constrained by pharmaceutical budgets, even if a treatment would be considered as cost-effective.

Additionally, the current approach to assessing cost-effectiveness often lacks a holistic view that in a consistent way incorporates suitable parameters that can estimate the experiences, preferences, needs and quality of life of people living with rare diseases, their family and caregivers, alongside the healthcare systems requirements.

Unfortunately, these challenges relating to rare diseases are often overlooked in national and regional reimbursement evaluations leading to innovative treatments for rare diseases being inaccessible to those who need them the most.

RECOMMENDATIONS

As part of addressing the unmet needs of people living with rare diseases, the value of treatment must be recognised in a more holistic way in order to pay for innovation that adds important value for patients and society, while securing financially sustainable healthcare systems.

13 7 IQVIA Report. EFPIA Patient WAIT Indicator 2022 survey. April 2023. Available from: https://www.efpia.eu/media/s4qf1eqo/efpia_patient_wait_indicator_final_report.pdf

14 Nordic Rare Disease Summit – An Assessment of Alignment of P&R Systems with the ORPH-VAL Principles, DOLON March 2021

Importantly, access to treatment needs to be supported by providing funding at national level. People living with rare diseases could get immediate access through models that allows for controlled uptake, risk-sharing, and on-going assessment of added value using real-world evidence (RWE). These mechanisms should be designed to maintain sustainable healthcare systems, preventing the costs of rare disease treatments from becoming a disproportional burden on local health authorities. Governments, payers and HTA bodies could engage in early and open dialogue to discuss how to best integrate these innovative payment models within their national pathways.

Moreover, the assessment processes across the Nordic region also need to be more transparent and must take into account the frequently limited evidence of efficacy for treatments for rare diseases. Linked to this, a holistic view of patient value should be implemented which considers alternative elements of products' value, e.g., quality of life, societal preferences, rarity, budget impact, and sustainability of innovation in rare diseases. During development of treatments for rare diseases, these parameters could be included in clinical trials to a greater extent in orderto help quantify the value. The use of RWE in the value assessment process can further address uncertainties and allow adjustments over time.

Linked to the above, the required data structure must be in place and ownership of, and access to, data should be clarified.

Identifying root causes of delays and inequality is a further important step in developing targeted solutions. Proposals such as the industry-led initiative 'European Access Hurdles Portal' aims to increase transparency in this field.[15] These insights may contribute to streamlining this vital process and expediting access to innovative medicines.

Lastly, the expertise of health care professionals and patient groups should be systematically used, and there have been increasing number of calls that all Nordic countries should join Orphanet and OD4RD (Orphanet Data for Rare Diseases) to improve data sharing. Complementary cross-border initiatives such as the Nordic Network for Rare Diseases and Sällsynta Brukarorganisationers Nordiska Nätverk (SBONN) can further foster regional collaboration.

EU LEVEL RECOMMENDATIONS

Beyond this, the Nordic countries should support, improve and facilitate the collaboration between ERNs, industry and other researchers, especially when it comes to the exchange of data and research collaboration. Multi-stakeholder platforms, such as the 'Together for Rare Diseases initiative', are prime examples of how ERNs and industry can also leverage and consolidate their wealth of knowledge, guiding the development and use of innovative medicines where they are most needed. For clinical trials, pooling of resources and patients to develop and test innovative medicines, can ensure that promising treatments are identified and made available more quickly.

15 European Federation of Pharmaceutical Industries and Associations (EFPIA) & Charles River Associates (CRA). (2023). European Access Hurdles Portal: EFPIA-CRA Report. https://www.efpia.eu/media/677291/european-access-hurdles-portal-efpia-cra-report-200423-final.pdf

4 Impactful Nordic Rare Disease Strategies

CONTEXT

A national rare disease strategy is important to raise awareness of rare diseases, ensure better funding for national patient associations and rare disease treatments, as well as secure a higher medical, political, and financial ambition. The framework of national rare disease strategies can also facilitate a continuous dialogue between rare disease stakeholders and drive the efforts to implement recommendations forward.

While an increased acknowledgement of the importance of national strategies for rare diseases has been observed, the implementation, including the financial support and political willingness, still varies within the Nordic countries. Notably, while Sweden and Iceland are currently working on their first national strategy, Finland has launched an updated strategy in January 2024. These strategies provide a potential action plan to prepare for further cross-border alignment and collaboration in the future.

CURRENT CHALLENGES

Despite national budgetary pressures and competing priorities, the Nordics need to commit themselves to setting ambitious goals and monitoring implementation and the effect of existing and future strategies, in order to overcome the current challenges within empowering people living with rare diseases, diagnostic delay and access to innovative treatments.

RECOMMENDATIONS

To ensure truly impactful national strategies, people living with rare diseases should be included in all processes of a strategy's development. It is crucial to harness these individual's expertise and combine this with a multi-stakeholder governance model (including patients and industry) to ensure regular monitoring, evaluation, and revision of strategies as well as identifying good practices to support national implementation.

The establishment of rare disease registries is a further important step towards achieving sufficient data that can improve early diagnosis and care. As it stands currently, the Nordics have a strong ecosystem to take the lead in Europe and show "best practice" on rare diseases. With excellent pre-requisites for pooling and sharing health data, a tradition of quality registries, well-established genomic centers, biobanks, strong patient communities, advanced academic centers as well as an advanced life science industry, the Nordics have a unique opportunity to systematically gather and connect these in rare disease registries on a national, regional, and international level.

There are multiple initiatives across the Nordics aimed at strengthening healthcare structures for patients living with rare diseases. A strategy should highlight the need of standardised care pathways, the establishment of resourceful expert centers for rare diseases, and ensure access and usage of diagnostic methods.

National strategies should include a transparent governance model including realistic timelines marked by clear, measurable, and timebound goals secured through adequate funding and resources. Only with national strategies and cross-country cooperation supported by the necessary political and economic ambition can there be lasting progress on rare diseases. This will not only empower people living with rare diseases but empower a much needed systematic shift in how to better support these individuals.

EU LEVEL RECOMMENDATIONS

On a European level, EU, Nordic experts and patient organisations should be connected and exchange knowledge and data across EU Member States. To achieve this strengthened European cooperation and to ensure prioritisation of people living with rare diseases across Europe it is important to create a European Action Plan on Rare diseases, a comprehensive framework of EU actions underpinned by indicators to monitor rare disease diagnostics and treatment success.

To this end, the action plan should also provide a common policy with common goals to develop effective national plans. As Denmark will lead the Presidency of the Council of the EU in 2025, they could promote the importance of an EU strategy, prompting the other Nordic countries to support its inception, and ensure synergies with existing EU efforts such as the ERNs. The European Commission could also play a more significant role in asking questions to EU Member States and tracking progress, to ensure that national rare disease strategies are being implemented and that national budgets are allocated for this.

Conclusion

All stakeholders who have contributed to this roadmap share an aspiration to improve the quality of life for people living with rare diseases in the Nordics. Making this a reality will require a strong commitment from all partners of the Rare Disease Summit including industry, policymakers, patient organisations, and healthcare professionals. We hope this Roadmap can secure the ambition to make the Nordic region the best place in Europe for people with rare diseases to live a fulfilling life, with the best possible support, opportunities, and access to care and treatments.

We promise to continue working together across borders and expert fields to overcome the unique challenges posed by rare diseases. Due to the rarity and high complexity, we need to recognise that rare diseases must be treated differently to secure the same progress as we take for granted for people living with more common diseases. In the Nordic countries, we follow the first steps taken with the previous two rare disease Summits and roadmaps and will strive to pave the way for future international collaboration. Ultimately, we will keep aiming at the long-term vision of enabling better lives and treatment for people living with rare diseases.

Recommendations and Measurable Outcomes

LEGEND

- Patient Empowerment
- Early Diagnosis
- Patients' Access to Innovation
- Impactful Rare Disease Strategies

RECOMMENDATION	MEASURABLE OUTCOME
The expertise of people living with rare diseases should be recognised as a resource on all levels of the Nordic healthcare systems	Before 2030 people living with rare diseases in the Nordics should be allowed access to their own health data to increase ownership and to be able to better track progress
	Joint Nordic guidelines should be developed with a common definition for patient empowerment
	Education on rare diseases should be a standard module at any medical school in the Nordics
	Earmarked national funding should be provided for patient associations representing people with rare diseases in the Nordics
	Patient organisations should be systematically included as partners in defined healthcare-related policy and decision-making following inclusion of ePAGs in ERNs

RECOMMENDATION	MEASURABLE OUTCOME
To significantly reduce the current time to receive a correct diagnosis for people with rare diseases in the Nordics	Joint Nordic medical guidelines, including national surveillance mechanisms, should be developed before 2027 to improve diagnosis and screening across the Nordics
	Before 2027 hospitals in the Nordics should allocate national healthcare budget for increased use of genetic counselling and molecular diagnostics to improve diagnosing of rare diseases
	Neonatal screening programs for newborns should standardise newborn screening across the Nordics and be best in class among European countries by screening for 25 or more rare diseases
	European Reference Networks (ERNs) should be fully incorporated into the Nordic healthcare systems

	Rare Disease registries should be introduced using internationally harmonised coding system for rare diseases such as OrphaCodes	
RECOMMENDATION Nordic countries should improve access to innovative orphan medicinal products, more than 70% of orphan medicinal products approved in EU should be available for people with rare diseases in the Nordics	MEASURABLE OUTCOME The Nordic countries should aim at using more models with risk-sharing or on-going assessments of patient outcomes to allow for faster access to innovative treatments	
	The use of real-world evidence should be considered in the value assessment process for treatments with limited clinical data due to small patient populations	
	Nordic countries should join Orphanet and OD4RD (Orphanet for Rare Diseases)	
RECOMMENDATION National rare disease strategies should be adopted, including clear policy recommendations with earmarked funding, for all Nordic countries by 2027	MEASURABLE OUTCOME People living with rare diseases should be included in all processes and in decision-making bodies related to national rare disease strategies	
	National rare disease strategies should include a transparent governance model and clear accountability, budget and political ownership of each recommendation	
	The European Commission started their mandate in 2024 should include into this new mandate a European Action Plan on Rare Diseases	

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