ROADMAP FOR RARE DISEASES

NORDIC RARE DISEASE SUMMIT 2021

The Nordic Rare Disease Summit, organized as a virtual meeting on 12th and 13th of April 2021, gathered a wide range of rare disease experts, decision- and policymakers as well as representatives from NGO's, patient organizations, academia and industry from across the Nordic countries. All coming together to debate how to best overcome the special challenges characterizing the rare disease area.

This Nordic Roadmap for Rare Diseases sums up key messages, discussions and new knowledge from the summit – with an aspiration to provide policy guidance relevant for the Nordic countries, recommendations for future co-operation and a shared call for action.

WHAT WE ARE FACING: The special challenges of rare diseases are…

The summit discussions revolved around three overarching themes "Diagnostic Delay", "Patient Empowerment" and "Patients' Access to Innovation". Despite progress in these areas, a stronger focus and an increased collaboration amongst all stakeholders is needed to enable better lives for people living with a rare disease. This need is reinforced by the COVID-19 pandemic which has negatively impacted quality of life and challenged access to necessary care.

Diagnostic Delay

The journey to diagnosis for people with rare diseases can often be long and uncertain. On average it takes six to eight years before a person with a rare disease receives the correct diagnosis and more than 40% of rare disease patients are misdiagnosed at initial presentation.[1,2,3]

Due to the rarity of the diseases there is limited awareness and knowledge of the signs and the symptoms of rare diseases. Most physicians will only see a small number of people with a rare disease, which makes it difficult to build up the necessary clinical knowledge and experience to recognize the symptoms.

In addition, the cooperation and pooling of knowledge across expertise areas and across countries is unsystematic and therefore the potential for knowledge exchange between healthcare specialists remains untapped.

The issue of diagnostic delay and the need for better tools, knowledge and cooperation to secure earlier diagnosis of people with rare diseases was elaborated throughout the summit.

Need for patient empowerment

Due to low prevalence, lack of expertise and poor quality of life amongst many people living with a rare disease, empowerment plays a crucial role.

Patients and their relatives hold unique knowledge of living with a rare disease and should be involved and consulted on all levels.[4]. There is a need for empowerment on individual patient level in the form of personal skills and knowledge, on community level in the form of mutual support groups, information sharing and coalition building, and on policy level to improve the shaping, prioritization and implementation of policies targeted people with rare diseases.

These needs are far from met today. People living with rare diseases are facing a lack of involvement and empowerment, and more can be done to develop disease specific information, guidelines and tools to strengthen patient engagement.

Uneven access to innovative and adequate treatment Rare diseases typically have few – or no – treatment options, and too often people living with a rare disease have delayed or no access to the treatment they need.

In the Nordic region, the assessment systems are typically not designed for rare diseases, and they often take a narrow approach to assessing the value of new treatments[5]. The current approach lacks a holistic view, that in a consistent way includes patient experiences, preferences, needs and life quality when assessing the value and price. The populations are small, and rare disease trials often lack the large scale datasets, which are requested by the authorities to demonstrate treatment value. Consequently, some innovative treatments for rare diseases never make it to the patients, who need them most. At the summit, it was recognized that delays and failure to gain access to innovative treatments may root in insufficient assessment systems.

1 Ronicke S, et al. Orphanet J Rare Dis 2019;14(1):69.
2 Vandeborne L, et al. Orphanet J Rare Dis 2019;14(1):99.
3 The Global Commission to End the Diagnostic Odyssey for Children with a Rare Disease
4 Marta De Santis, Clara Hervas, Ariane Weinman, Valentina Bottarelli, National Centre for Rare Diseases, Istituto Superiore di Sanità, Rome, Italy, EURORDIS – Rare Diseases Europe
5 Nordic Rare Disease Summit – An Assessment of Alignment of P&R Systems with the ORPH-VAL Principles, DOLON March 2021

WHAT WE SHOULD STRIVE TO DO:

We can overcome the special challenges of rare diseases by…

At the summit leading experts shared best practices and reflected on potential solutions to the challenges of rare diseases.

Securing advanced diagnosing

The improvement of diagnosis relies on innovative approaches, where technology innovators, patient advocates, healthcare providers and researchers cooperate to tackle the challenge of rare disease diagnostics. This cooperation should be supported by European Reference Networks in EU as much as in wider Europe, which can provide doctors with a structure for sharing experiences and knowledge in an efficient way. ERNs can serve as databases where healthcare professionals can find the right information at the right time.[6]

To shorten the journey towards accurate and timely diagnosis it is necessary to invest in pioneering diagnostic platforms and exploit digital innovation to find new solutions. Timely diagnosis requires timely action, and we need to include and offer neo natal screening programs systematically in all Nordic countries. Further, all countries need to secure implementation of innovative technologies and easily accessible information hotlines for HCPs and patients. Finally, we need to implement internationally harmonised coding systems for rare diseases like OrphaCodes and establish registries on a national, regional and international level.

Empowering patients at all levels

Patients' knowledge and expertise should be recognized and promoted as a resource on all levels of the healthcare systems: At micro level, empowerment will serve as a resource for patients and their relatives to gain greater control over decisions and actions concerning their health and wellbeing. At community level, empowerment can improve social services and quality of health. Finally, at policy level, empowerment is important to allow patients to contribute as a resource in development of sustainable healthcare systems.

This requires development of health policies that acknowledges and focuses on patient empowerment, as well as sustainable financial support systems for patient organizations and education, employment and inclusion of patient representatives in decision processes.

We need to recognize chronic patients as experts of their own care by involving them as "co-managers" of their condition in partnership with HCPs. By putting empowerment on the agenda, health policies should guarantee education and training of all stakeholders, patients, HCPs and institutions, and thus contribute to a cultural change towards patient empowerment on all levels.

Securing access to innovative treatment

As part of addressing the unmet medical needs of people living with rare diseases, we need to break the access deadlock by recognizing value of treatment in a holistic way and paying for innovation that adds true value for patients, while securing financially sustainable healthcare systems.

The assessment processes across the Nordic region need to be more transparent and must take the often limited evidence of effect for medicines for rare diseases into account. Linked to this, a holistic view of patient value should be implemented considering all relevant elements of product value, e.g. quality of life, societal preferences, rarity, budget impact and sustainability of innovation in rare diseases. The expertise of health care professionals and patient groups should be systematically integrated.

Access to treatment could also be supported by provision of funding at national level and by combining immediate access for patients with models that allows for controlled uptake, risk-sharing and on-going assessment of added value through use of real world evidence.

National plans and strategies for rare diseases to secure progress and commitment in all areas

Despite an increased acknowledgement of the importance of a national strategy for rare diseases, the implementation, including the financial support and political willingness, still varies within the Nordic countries. To overcome the challenges, all countries need to commit themselves to set ambitious goals and monitor the implementation and effect of the existing and future plans and strategies, including any additional challenges resulting from the COVID-19 pandemic. Further, establishment of rare disease registries is an important step towards achieving sufficient data that can improve diagnostics and care.

In addition to this roadmap, the national plans and strategies can take inspiration from the Rare 2030 recommendations which sets out the need for a new European policy framework for rare diseases to guide the implementation of national plans for rare diseases with the same measurable objectives.[7]

Only with national strategies and cross-country cooperation - supported by the necessary political and economic ambition – can we make lasting progress within rare diseases.

WE PROMISE EACH OTHER….

Today, we promise each other to continue to work together across borders and expert fields to overcome the special challenges of rare diseases. Due to the rarity and high complexity, we need to recognize that rare diseases must be treated different in order to secure the same progress as we take for granted for people living with more common diseases. In the Nordic countries, we have taken the first steps, and will strive to pave the way for future international collaboration to support the long-term vision of enabling better lives for people living with rare disease.

6 The *EU Directive on Patients' Rights in Cross-Border Healthcare* requires the EU Commission to support the development of ERNs throughout Member States.
7 The Rare 2030 "Recommendations: The future of rare diseases starts today, http://www.rare2030.eu/recommendations

Authors and endorsers of this Roadmap: *Authorship of the Nordic Roadmap lies with multiple stakeholders. The hosts and partners of the Nordic Rare Disease Summit 2021 developed the foundation of the Nordic Roadmap, and contributions to the content derive from presentations and statements from speakers, panelsts and participants at the Summit. As a result of the collaborative effort, no organization or individual should be made independently responsible for the Nordic Roadmap. The Nordic Rare Disease Summit was hosted by Takeda in collaboration with Rare Diseases Denmark and the partners of the Summit were EURORDIS, SBONN and LIF.*