President of Eurordis Terkel Andersen presented the necessity of patient empowerment and recommendations on specific mechanisms that can empower people with rare diseases.

Launch of the Nordic Roadmap

On the 13th of April 2021 the Nordic Roadmap for Rare Diseases was launched to ensure a lasting legacy of the Nordic Rare Disease Summit held on the 12-13th of April 2021 with a common aspiration to elevate rare diseases as a national health priority in the Nordic countries.

The Roadmap is the culmination of 2 days of knowledge sharing and debating between rare disease experts, decision- and policymakers as well as representatives from NGO's, patient organizations, academia, and industry from across the Nordic countries. The sessions were followed virtually by more than 480 people across the Nordics and beyond in an engaging manner with inputs and live chat between people connected across borders.

The Summit revolved around 3 pre-identified themes: Diagnostic Delay, Patient Empowerment and Patients Access to Innovative Treatment.

Each theme included presentations that highlighted challenges, solutions and recommendations for the future which experts on the topic discussed thoroughly.

Early diagnosis is key

Among experts it became clear that no matter which disease you may have, a diagnosis is crucial for the patient and the possibility to offer treatment. *"Without the correct diagnosis, the health and well-being of persons living with a rare disease and their relatives will be severely impacted,"* underlined Maria Montefusco, Chair, Rare Diseases Sweden.

US Chief Medical Officer at Microsoft, Clifford Goldsmith, presented the different initiatives launched by the Global Commission. New ways of diagnosing rare diseasesare evolving. The Global Commission is a collaboration between Eurordis - Rare Diseases Europe, Microsoft and Takeda with the purpose of shortening the diagnostic journey for children with rare diseases. To accomplish this, the Global Commission has launched initiatives that combine recent breakthrough diagnostics with state-of-the-art AI capability. *"The Global Commission is committed to harnessing the power of technology and empowering families to accelerate the time to diagnose a child with a rare disease,"* stated Clifford Goldsmith.

Major steps forward in shortening the diagnostic journey of people with rare diseases have been taken in some countries. For instance, Iceland has recently taken a major stride towards using whole genome sequencing to diagnose more people with rare diseases. MD PhD, Associate professor Hans Tómas Björnsson elaborated: *"We have made major strides by collaborating with Decode Genetics who have been doing pro bono whole genome sequencing. This has enabled us to diagnose many more people and it also increased the access to genetic services for people across Iceland".*

In Denmark, considerable advances has also been made, especially a promising project from Copenhagen University Hospital is worth mentioning. At the hospital 320 acutely ill babies were screened for more than 2000 genes using whole genome sequencing. With a diagnostic rate of about 30% and many of the diagnoses being treatable, the project was a major success. Projects like this could hopefully also pave the way for a more systematic use of whole genome screening in the national neonatal screening program. *"We have several efforts that goes in the same direction. An example could be our screening service for acutely ill infants, where we use whole genome sequencing to screen for 2000 genes. We are uncovering diagnoses we have never seen before,"* explained Chief Physician Allan Meldgaard Lund.

The summit is officially endorsed by EFPIA – The European Federation of Pharmaceutical Industries and Associations, EUCOPE – the European Confederation of Pharmaceutical Entrepreneurs, The Danish Haemophilia Society (Bløderforeningen) and IML – Innovative Smaller Life science companies.

The summit was organized by Takeda and hosted in collaboration with Rare Diseases Denmark (Sjældne Diagnoser).

Eurordis, SBONN and Lif joined as partners

Allan Meldgaard Lund especially highlighted the National Genome Center that is a part of the personalized medicine strategy being implemented in Denmark. The center is part of the Ministry of Health, responsible for developing and leading the Danish national infrastructure for personalized medicine. The main infrastructure consists of a national whole genome sequencing center and a national high performance computing center.

These are just a couple of the highly promising projects that are being carried out in some of the Nordic countries. However, there is still much work to be done. Whole genome sequencing is at the cutting edge of diagnostics, and it will take time to disseminate knowledge about new capabilities throughout the healthcare system and the public. *"Education is key, both for the general public and for healthcare providers so they are aware of the capabilities of modern diagnostics,"* stated Prof. (em.) Arvid Heiberg.

Empowerment is key for the wellbeing of the rare patient

All participants agreed that diagnostics are of high importance for the rare disease patient. However, improved diagnostics cannot stand alone since this is only the beginning of the patient journey. Patients are the true experts on living with their disease, and empowerment is of outmost importance on many levels. President of Eurordis Terkel Andersen presented the necessity of patient empowerment and Eurordis recommendations on specific mechanisms that can empower people with rare diseases. *"The World Bank has defined empowerment as the process of increasing the capacity of individuals or groups to make choices and to transform those choices into desired actions and outcomes,"* said Terkel Andersen.

Empowerment can be increased at several levels. At the Summit, empowerment at patient, societal and policy level was explored.

At the patient level, Research Director at The Finnish Association of People with Physical Disabilities, Sinikka Hiekkala, presented research about how web-based adaptation training had significantly impacted the lives of the participants with rare diseases: *"Patients with rare diseases had good experiences with the course "Rare among"*. It helped all participants achieve their goals of improved physical and mental health and self-esteem," said Sinikka Hiekkala.

At the societal level, President of Rare Diseases Denmark, Birthe Byskov Holm, presented Rare Diseases Denmark's Helpline to empower people living with a rare disease and their relatives. The helpline also contributes to capacity building, creating and sharing knowledge: *"Knowledge of rare diseases and how to live with them are very scarce. Rare people are important stakeholders in the rare knowledge landscape,"* said Birthe Byskov Holm.

At the policy level, Founder and President of the Wilhelm Foundation, Helene Cederroth, painted a picture of the absolute necessity of national strategies to help people with undiagnosed diseases receive equal opportunities: *"Don't forget the undiagnosed patients, they need all of us, we all have to collaborate to help them,"* Helene Cederroth stated.

Diagnostics and empowerment can only take us some of the way

Currently, many people with rare diseases face prolonged access to treatment – if any treatment exists at all. This is caused by both the sheer number of rare diseases, but also because of inadequacies in the current fragmented evaluation process and nonaligned common methodologies. *"We are at the stone age regarding patient representation in HTA scientific dialogue and assessments in Europe, it's very diverse and there are agencies where it simply doesn't exist,"* said Yann Le Cam, Chief Executive Officer, Eurordis.

The problems with access to treatment are systematic and is often the result of distrust between payers and industry. The systems need to change if we want to improve access to treatment for people with rare diseases. MP Lina Nordquist elaborated: *"No matter how hard we try, there will always be patients that have difficulties in getting their voice heard. I think we need to secure the system and for instance in quality registries we need to make sure to ask what is important to every single person included".*

Pasi Nevalainen, Head of Rare Diseases Unit, Tampere University Hospital, Nicklas Sandström, Moderate Party, Prof. (em) Arvid Heiberg, Department of Medical Genetics, Oslo University Hospital, and CEO Lene Jensen, Rare Diseases Denmark, reflected on the Nordic Roadmap for Rare Diseases and discussed priorities for the future in one of the many virtual debates at the summit.

Based on nine principles for value assessment and funding processes in rare diseases developed by the European Working Group ORPH-VAL, the UK consultancy DOLON performed an in-depth review in 2018 and updates in 2020 and 2021. This analysis identified three common areas for potential improvement.

1. Value assessment processes should consider all Rare Disease Therapies (RDT) specificities in a consisten way.
2. More consistent disease-specific expertise should be incorporated in current processes.
3. RDT assessment processes should be adaptive and subject to the need and availability of information over time.

A particular hindrance to the access to treatment for patients with rare diseases is the affordability issues of orphan drugs. Part of the issue stems from subnational financing, especially financing at the hospital level, which can be a significant obstacle. Financing of orphan diseases at national level could help improve affordability issues. *"Subnational decision making has fostered inequality in access to treatment in some countries, so we definitely support national processes,"* said Giles Platford, President Europe and Canada Business Unit, Takeda.

The Nordic Roadmap will pave the way forward

The Summit highlighted that we face the same challenges across the Nordics. The strong tradition for working together is a great strength and will be a key factor in improving the lives of people living with rare diseases. With the Nordic Roadmap as a framework, we can build new ambitious national and supranational strategies. *"When it comes to rare diseases, Nordic cooperation is so obvious and it is hard to understand why we do not cooperate even more. When the disease is rare the number of patients, scientists, doctors, nurses and professionals with the newest knowledge are few. This calls for specialization and more collaboration than we have ever had before."* said Bertel Haarder, President of The Nordic Council, (MP) and former Danish Minister of Health.

Only with national strategies and cross-country cooperation – supported by the necessary political and economic ambition - can we make lasting progress within rare diseases: *"All the themes go together; with better empowerment, we get better diagnostics and access,"* concluded CEO Lene Jensen, Rare Diseases Denmark, and encouraged the participants to elevate the rare disease agenda whenever possible.

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