NORDIC RARE DISEASE SUMMIT 2023

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SWEDEN ANNOUNCES

First National Strategy for Rare Diseases at Nordic Rare Disease Summit 2023

At the second Nordic Rare Disease Summit taking place on the 17th of April 2023, the main theme was "Rare diseases – setting the scene for change". A total of 663 registered participants from 33 countries gathered to discuss three key focus areas: Early Diagnosis, Patient Empowerment and Access to Innovation. The two main insights from the summit were the announcement that Sweden now will commence work with a first national strategy for rare diseases and that there is a strong commitment across the Nordics to collaborate to find solutions to the three key focus areas.

Participating stakeholders from across Europe gathered at Posthuset in the heart of Stockholm, Sweden as well as online. Attendees included patient representatives, health care professionals, academic researchers and scientists, representatives from the pharmaceutical sector as well as healthcare leaders, policy makers and government officials.

> **The work with early diagnosis and care, support, and treatment for people with a rare disease is a priority for the Swedish government. And therefore, we will start working on an initiative this autumn to develop a national strategy for rare diseases.**
>
> Acko Ankarberg Johansson
>
> SWEDISH MINISTER FOR HEALTH CARE

Nordic Rare Disease Summit 2023 addressed key action areas identified at the first Nordic Rare Disease Summit in 2021 to discuss how to put them into action:

1 Early Diagnosis

On average it takes six to eight years before a person with a rare disease receives the correct diagnosis. Approximately 40% of all patients with a rare disease are initially misdiagnosed. A correct diagnosis empowers patients, enables access to innovative care and improves quality of life for patients living with rare disease.

2 Patient Empowerment

A challenge in rare disease is to fully empower patients in their own health journey. Patients with rare diseases many times have more expertise about their diagnosis than health care representatives they interact with. Hence, systematic involvement of patients and caregivers, acknowledging their unique source of knowledge in their own situation, is essential.

3 Access to Innovation

As part of addressing the unmet medical needs of people living with rare diseases, the access deadlock needs to be broken 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.

National strategies setting ambitions and uniting stakeholders

With 360 million people worldwide and 1 million people estimated to live with rare diseases in the Nordic countries, rare diseases are anything but rare[1]. A national strategy in place could help setting ambitions and unite stakeholders. Hence, the announcement that Sweden will commence work with its first national strategy for rare diseases was much welcomed by all attendees.

Experiences from European countries that already have a national strategy in place provided insights on how to successfully develop and implement this tool. Insights that were highlighted was the importance of placing patients' needs at the center and encourage cross-stakeholder collaborations, from the get-go. Financials are needed to safeguard the long-term implementation of a national strategy for rare diseases and ongoing evaluation essential to help prioritize efficient initiatives.

Satu Wedenoja, Chief Physician at the Finnish Institute for Health and Welfare (THL), provided a real-world perspective on the capability of a national strategy. Today in Finland, the Orpha Code System is used in the electronic patient reporting systems – an initiative developed in Finland's first national strategy for rare diseases. As of June 2023, these codes will transfer directly into the National Registry and enable data collection of all rare diseases in Finland. This is a concrete result of the national strategy and could be replicated to other Nordic Countries.

Birthe Byskow Holm, Chair, Rare Diseases in Denmark quoting the first Danish strategy for rare diseases: "All the new knowledge about rare diseases will not lead to change by itself, you need to do something very special, not only in the health care system but also, in the social system. The person with a rare disease is often a child, and it affects the whole family. It also has an impact on school, education, housing, work, being able to live independently, all aspects of life."

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> **Birthe Byskow Holm**
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> **RARE DISEASES IN DENMARK QUOTING THE FIRST DANISH STRATEGY FOR RARE DISEASES**

Several speakers emphasized a holistic approach on a national strategy and to remember to include extremely important areas such as family support, rehabilitation, physiotherapy.

[1]*Center for Rare Diagnoses (CSD), Karolinska University Hospital*

Europe is buzzing with activities around rare diseases

The Nordic Rare Disease Summit 2023 was arranged under the Swedish EU presidency. During the summit a number of speakers representing the EU-perspective as well as several delegates outside the Nordic countries confirmed the interest in shaping policy in the rare disease area, across Europe.

The European pharmaceutical legislation is undergoing a revision where several parts have a direct impact on the innovation climate for Orphan Medical Products. This provides an unprecedented opportunity to secure ongoing incentives in development of new treatments.

> **Over two decades ago, before EUCOPE was founded, there were only approximately eight products in the European market indicated for a rare disease, so not really a success story. Since the Orphan Medicinal Products (OMP) legislation was launched in 2000, up until now, more than 200 new therapies have been authorized for people living with a rare disease. In that regard, we can say that the Orphan Regulation has been successful. Based on this, it is extremely important to be active and participate in the shaping of policies and issues now, to build on these successes and improve the system for the next twenty years.**
>
> Alexander Natz
>
> SECRETARY GENERAL OF EUCOPE

> **The Nordic Rare Disease Summit 2023 in Stockholm is the main event on rare diseases under the Swedish EU Presidency.**
>
> Yann Le Cam
>
> RARE DISEASES EUROPE

As stated by Ricardo Marek, President Europe & Canada at Takeda, the pharmaceutical company that initiated the Nordic Rare Disease Summit, a topic that needs to be addressed across Europe is how to define value from a broader societal perspective than today. Exchanging experiences across countries could provide insights on how to collaborate and shift towards innovative payment models and value-based contracts to secure patients access to treatments.

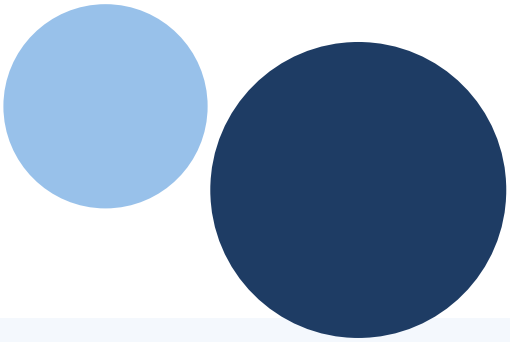
Professor Anna Wedell, medical doctor, and director of Precision Medicine Center at Karolinska University Hospital, provided a further view on how to work on a holistic approach, describing how clinical genomics data is shared all the way to tailored treatment. Diagnostics and treatment are thus more integrated. In Sweden, this way of working does not fit into the existing models as value is generated somewhere else in the health care system. Thus, structural changes, new reimbursement models and a better coordinated healthcare is required.

> **Pharmaceutical innovation in Europe is coming to a crossroad, and we must decide if we're still going to be the first choice of continent for public partnerships, for research and development, for the Pharma Industry and all life science industries.**
>
> Pernille Weiss
>
> MEMBER OF THE EUROPEAN PARLIAMENT REINFORCED THE IMPORTANCE OF CHOOSING A DIRECTION FOR EUROPE

Early diagnosis remains a challenge

As a patient with a rare disease the road towards diagnosis is often long and winding. Delegates confirmed patient surveys have shown that many patients for years, sometimes even decades, are unable to receive accurate help. Delayed diagnosis entail delayed and erroneous treatment or simply, no treatment at all, allowing diseases to progress and possibly cause serious harm. Thus, early diagnosis was chosen as one of the tree focus areas of the summit.

Equal care remains a challenge within many health care systems and with rare diseases, the availability of diagnostics tests varies in Europe. Through coordinating regulations and compensation models across geographical areas the availability of these important diagnostic tools could possibly increase. In addition, payers, decision makers and health care professionals are in need of an increased level of awareness to place rare diseases on the radar.



> "Childhood patients are usually diagnosed within the first three years or so, usually even earlier. For adult patients, on average in Finland, it takes 10 to 18 years to get a disease diagnosis. That's not early, that's a lifetime. For childhood and newborn patients, we need screening a lot earlier. We have those tools in our pack, but we don't utilize them enough."

Katri Asikainen

CEO, HARSO – THE FINNISH ALLIANCE OF RARE DISEASES AND DISABILITIES ORGANIZATIONS

> "The rare disease patients have the biggest challenges because they will always be beyond the most likely case scenario, because it isn't likely, but they still have it."

Stephanie Juran

PROJECT MANAGER AND RESEARCHER, RARE DISEASES SWEDEN

Health data and AI becoming beacons of hope

Nordic Rare Disease Summit 2023 also uncovered opportunities in pooling data in registers, harmonizing diagnose coding and utilizing high-tech solutions.

Common standards and frameworks are crucial in maximizing the potential in pooled data. Hence, the ongoing work of the European Health Data Space is, and will continue to be, of great importance. In turn, the Nordic countries are ready to participate and take action to accumulate relevant data and develop legislations to optimize targeted use of registers. Here, building of databases was identified as one key enabler towards early diagnosis.

> **AI is a little bit of a black box, and as a classically trained scientist you need to be able to demonstrate something, describe how you did things and explain what came out of it. Because that's how you do science.**

Jón Jóhannes Jónsson

MEDICAL DIRECTOR, DEPARTMENT OF GENETICS AND MOLECULAR

> **Adding clinical and real-life data over time can add to our understanding of disease progression, and how different diseases can be associated with similar conditions. And every time we define a new rare disease, we can increase the database which will help to solve diagnostic issues.**

Daniel Scherman

PROFESSOR, DIRECTOR OF THE FOUNDATION FOR RARE DISEASES IN FRANCE

High-tech solutions such as the use of artificial intelligence (AI) becomes additional tools within rare diseases. Innovative digital software can as of today already provide face and language recognition. AI solutions, such as apps analyzing facial traits, can become additional diagnostic tools. A person taking a picture of themselves could send it to a secure big data processing center, where face recognition algorithms may detect possible diagnoses. At this point, these solutions are in an innovative phase outside of clinical practice but could, in a near future, provide further beacons of hope for patients with rare diseases.

Patient empowerment – all the way

Acknowledge, enable, and increase patient empowerment and keeping patients at the center of both care and policy making. Patient advocacy groups and organizations will need further support and defined collaborative opportunities to continue to be the voice of the patient, as well as an appreciated support for both patients and their families.

> **One of the essential things we can do is to get together, to relate to someone who has a similar disease. Someone who can talk about the challenges they have met, and how they have navigated the health care and social system."**

Terkel Andersen

IMMEDIATE PAST PRESIDENT, EURORDIS

Another important role for patient advocacy groups and organizations is to become part of the decision-making processes, and of national and European initiatives on rare diseases. Additionally, they play an important part in raising awareness and increase knowledge on rare diseases.

As evident in developing the national strategy for rare diseases in Denmark, a key factor mentioned at the summit was the collaboration with the national patient organization. A collaboration resulting in strategic points in the actual national strategy, highlighting both the importance of involving patient representatives in policy making and that patient representatives needs to be heard as new proposals are being developed.

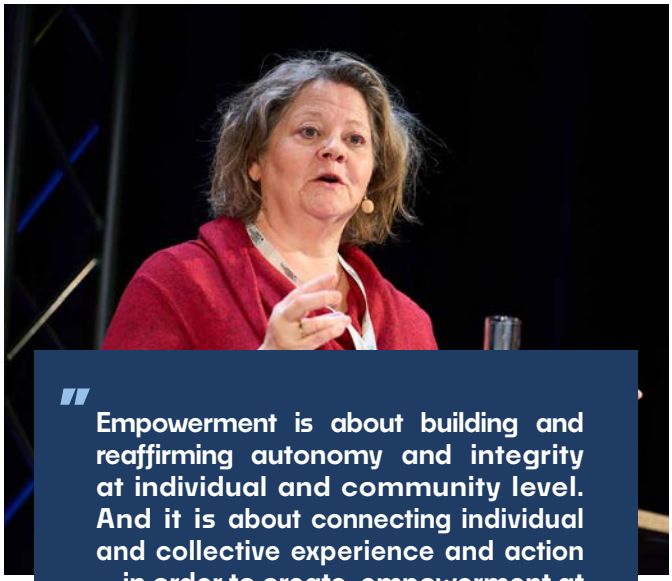
In early 2023, the Danish strategy was evaluated and as stated in the strategy, the patient organization was represented in this working group. That enabled a cross-stakeholder understanding of the strategy's effects and relevance for patients.

> **It is devastating if health care professionals are not given the conditions to listen and collaborate with their patients. Even worse, if you as an individual are not being believed in and thought to be making things up.**

Stephanie Juran

PROJECT MANAGER, RARE DISEASES SWEDEN

The summit also called attention to the difference between patient empowerment on an individual level and patient empowerment on an organizational level. It comes as no surprise that patients, and those around them, many times have a vast knowledge about their disease. Patients' needs must therefore be addressed and respected, and their knowledge utilized and enabled.



> **Empowerment is about building and reaffirming autonomy and integrity at individual and community level. And it is about connecting individual and collective experience and action – in order to create empowerment at all levels.**

Lene Jensen

CEO, RARE DISEASE DENMARK

A consensus during the summit was that this could be achieved by inviting patient representatives to the table, as patients' rights are as fundamental as any rights. To further strengthen patient empowerment, the European Commission is also funding projects to inform, educate and further strengthen patients living with or around rare diseases.

> **It is extremely important to educate society so that patients are able to state what they need and get accurate care.**

Gudrún Helga Hardardóttir

CEO, UNIQUE CHILDREN ISLAND

When patients are caught between local and EU legislations, the enforcement of patient rights becomes a challenge in all Nordic Countries. A challenge that needs to be addressed on governmental and EU level is to provide legislations that support patient rights, rather than prevent. One key to unlocking these legislations is to open a dialogue with patients and those around them – providing insights and knowledge on obstacles as well as opportunities for change.

> **Patients need to be regarded as experts and as such, be respected in different forums**

Geir Lippestad

LAWYER AND RARE DISEASE ADVOCATE

Call for improved access to medicine for people with rare diseases

More innovation and new research resulting in medical breakthroughs calls for a progressive health care system to give access to these innovations. It is one thing to make scientific progress and produce novel therapies that can treat a rare disease and another to have access to novel treatment.

Access pathways to orphan medicinal products challenge a system once built for therapies with large patient populations. The nature of a rare disease is that it affects few people. A small patient population leads to smaller studies showing effect and value of the treatment. Advanced therapies where each medicine is treating a few patients is a driver for price and thereby challenge pharmaceutical budgets. These aspects trigger a discussion on whether it is time to further develop value assessment processes and methods when it comes to rare diseases and how costs should be handled. This was a task that the speakers at Nordic Rare Disease Summit considered suitable for cross-country alignment.

> **"We have seen entirely new paradigms of treatment in the shape of cell and gene therapies. What about policy? It seems to me that the policy has somewhat lagged behind the science."**
>
> **Adam Hutchings**
> **CEO, DOLON**

The value-based-pricing-system in Sweden was taken as an example; There is an ethical platform laying the ground for decisions, taking human value, medical need, and cost-effectiveness into consideration. Evaluation of new medicines targeting rare diseases are however proven harder to evaluate given the lesser quantities of data. There is an increased willingness to pay for "true innovation", but the data quantities available are making it harder to evaluate what therapies actually constitutes such an innovation. Also, the speakers pointed out, the willingness to pay differs from the ability to pay.

> **"We need to talk about priorities and premiere efforts that bring true clinical benefit for patients. Affordability is key from a health care perspective – it is our responsibility towards taxpayers but also fundamental to sustainable health care systems."**
>
> **Elham Pourazaar**
> **HEALTH CARE STRATEGIST, INNOVATION AND PARTNERSHIPS, REGION VÄSTERBOTTEN**

The discussion involved use of complementary data sources in a structured way as well as expert involvement from patient representatives, and health care professionals. Patient reported outcome measures could play an even more important role when possibly integrated into a study design as well as in terms of real-world evidence (RWE). Value based contracts, such as adaptive payment models, was raised as another way to ensure early introduction of new therapies and that society is paying for the true value of new treatments. It was clear that there is broad alignment on that such contracts would be a good way forward to share the financial risk, however alignment on the ´how to get there´ still needs more thinking and collaborations across borders.

Early collaboration between industry, regulators, benefit agencies, payers, healthcare providers and patients could establish how to define and measure the value of a new treatment when data is limited, and then connect it to a payment model that is accepted by all parties involved. There are still challenges with administrative resources and legislations of handling health data, but it is clear that a good intention and ambition is in place across all stakeholders.

The trade associations present at the summit expressed that they were more than open to discussing innovative payment models, however a structured system for how to do this needs to be established.

> **"We all want the same thing, and we are open to find solutions together across borders. And it is important according to LIF to start pressure test together and learn together what is working and what is not."**
>
> **Karolina Antonov**
> **HEAD OF ANALYSIS, LIF SWEDEN**

The power to make a change

Since the 2021 summit, significant improvements have been made to improve the lives of patients with rare diseases; rapid development in treatments, improved patient empowerment and a surge of interest from politicians to improve access to novel treatments.

The main focus areas remain the same, early diagnosis, empowering patients, and access to innovation. Nordic Rare Disease Summit 2023 uncovered ways forward on these areas as well as what part stakeholders could play in making a change for patients living with rare diseases.

Attendees joined the final keynote speakers in appreciating the ongoing work conducted by individuals, organizations, and collaborative entities in the last few years.

Finally, concluding this year's Nordic Rare Disease Summit was Anders Olauson, founder of Ågrenska. In his speech he addressed the importance of working on a larger systematic level while staying focused on the personal level.

> **"It is only by conducting both of them that we can really change the life of patients which ultimately is what matters here."**
>
> **Anders Olauson**
> **FOUNDER, ÅGRENSKA**

Supported by all attendees Anders Olauson encouraged to keep talking and working, and that all of us together has the power to make a change now – and an opportunity and possibility to appreciate and discover what can be accomplished by the next Nordic Rare Disease Summit in 2025.

A notable recommendation from this year's summit was for the development of an updated Nordic Roadmap for Rare Diseases, with measurable recommendations. This will be a forward-looking roadmap, taking into account all dialogues, and possibly agreed upon by the end of 2023 by all ten participating partners of this year's Nordic Rare Disease Summit.

The Nordic Rare Disease Summit 2023

The Nordic Rare Disease Summit 2023 was organized 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 organizations for rare diseases in five Nordic countries, EUCOPE - The European Confederation of Pharmaceutical Entrepreneurs, and EFPIA - The European Federation of Pharmaceutical Industries and Associations.

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Nordic Rare Disease Summit 2023 is organized and financed by Takeda Pharma with contribution from Alexion AstraZeneca Rare Disease, Novo Nordisk and Sobi.