



The Norwegian National Strategy for Rare Diagnoses



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Background

In 2009, the EU Commission recommended (1) that member states should establish plans and strategies for the assessment and treatment of rare diseases to ensure that patients with these conditions would have access to high-quality healthcare services. As an alternative to its own strategy, member states were encouraged to develop measures for rare diseases as a part of other healthcare strategies and healthcare legislation.

Norway has thus far not had a its own strategy for patients with rare diseases. However, it has implemented various measures to ensure that all patients receive satisfactory services from both specialist health services and municipal health and care services. Based on both medical advancements and patient needs, specialist health services have established new clinics, departments and units to provide specialist assessments, diagnostics and treatment. Both primary and specialist health services offer habilitation and rehabilitation and have developed various programs to ensure that patients with rare diseases and conditions are able to obtain the help they need. In several fields, individual services have been developed in cooperation with agencies such as the Norwegian Labour and Welfare Administration (NAV) and the Statped state agency for special education services.

Despite this, a number of patients and service users, family members, user organisations and health professionals believe that many of the established services and programmes do not function well enough, or do not adequately meet the needs of people with rare diseases and conditions. This strategy will contribute towards targeted efforts to ensure that such services are improved.

The aim of the National Strategy for Rare Diagnoses

The National strategy for rare diagnoses is the Ministry of Health and Care Services' strategy on how to equip specialist health services and municipal health and care services to address current and future challenges for people with rare diagnoses, diseases and conditions. This strategy primarily encompasses services that fall under the Ministry of Health and Care Services' areas of responsibility.

The main objective of the strategy is to ensure that all people who are born with or later develop a rare diagnosis or disorder have equal access to a high-quality assessment, diagnostics, treatment and follow-up. For many conditions, this would require more knowledge and research, as well as a better organisation, coordination, and customisation of measures based on patient needs and a clarification of responsibilities.

This strategy highlights several issues related to assessment, treatment and follow-up, and it identifies objectives that can help health services provide care that is better adapted to user needs.

The strategy is based on what is already in place and what can be improved. The goal is to create patient-centred health and care services with equal access to high-quality services. Based on feedback from service users and health professionals, this strategy focuses on different objectives that must be achieved to improve services for the patient group.

1 Patient-Centred Health Services

Equal access to assessment, diagnostics and treatment

The Norwegian Government's goal is to create patient-centred healthcare services. This refers to healthcare services that secure help, information and care for patients when they need it, and where they have the opportunity to influence, impact and have control over their own decisions. Patient-centred healthcare services involve equal access to high-quality health and care services. The primary goal of the strategy is to ensure that all people with rare diagnoses or health conditions have equal access to high-quality assessments, diagnostics, treatment and follow-up.

Principles and criteria for the prioritisation of methods and initiatives aimed at patients and patient groups with both common and rare diagnoses are stated in the Report to the Parliament (White Paper) on Prioritisation (9). As stated in (10) Sections 2 and 2a of the Prioritisation Regulations, the prioritisation of special health services shall be based on criteria for severity, benefit and resources. Rarity is not a specific priority criterion, even if diagnostics present special challenges.

Specific priority guidelines have been developed for different medical areas. These guidelines will include conditions covered by the definition of rare diseases and conditions, and that will result in recommendations for granting the right to care with deadlines for the most commonly referred conditions in each medical area. Certain diagnoses and conditions are not mentioned in the priority guidelines. Referrals, however, must still be evaluated on the basis of priority criteria. The use of priority criteria and priority guidelines will contribute to a more equal access to assessments and treatment.

User involvement and integrated services

Service users and patients receiving specialist health services and municipal health and care services have a statutory right to participation and involvement. This applies to both the design of their own services and to the choice of available treatment methods through shared decision making.

In patient-centred health services, the aim is for patients, family members and medical personnel to experience health and care services such that patients:

- are actively involved in their healthcare and are both seen and heard
- experience integrated services across hospitals and municipalities
- find that services are provided by teams created for them

This strategy has five overarching objectives:

1. Equal and faster access to high-quality assessment and diagnostics

Medical advancements and increased expertise make it possible for more people with rare diagnoses, diseases and conditions to obtain better and more precise diagnostics more quickly. Good patient trajectories with equal and faster access to high-quality assessment and diagnostics will help ensure that people with rare diagnoses are given the correct diagnosis as early as possible.

2. Equal access to high-quality treatment and follow-up

Medical advancements and technology help ensure that an increasing number of patients can be offered treatment. An objective is for patients with rare diagnoses and conditions to receive equal access to treatment, including new and experimental treatment. However, most patients with rare diagnoses will have complex needs for which there are no cause-oriented therapies. For all of these patients, multidisciplinary symptomatic treatment, habilitation, rehabilitation and services from other sectors will be essential to ensure a good life. Individual needs should be assessed from a life course perspective. Patients and professional communities will have access to new expertise and assistance for assessments, diagnostics and treatment of rare diagnoses and conditions through participation in the European Reference Networks (ERNs) that facilitate cooperation on rare diseases and highly specialised treatment.

3. Good patient pathways, better cooperation and coordination

People with rare diagnoses and complex needs for health care often experience challenges due to inadequate coordination and cooperation between various services.

An objective is to establish good patient pathways with a clear division of responsibilities and a structure and organisation that contribute to more comprehensive care for these patients. User involvement and participation at the service and system level must be facilitated.

4. Mastery of everyday life with a rare disease or condition

Many people with rare diagnoses will have health issues and the need for long-term services throughout much of their lives. An objective is for everyone to receive services that provide support and that promote coping skills, activity and participation, making it easier to live with a rare disease or condition. Health and care services must be made better aware of how various types of disabilities, diseases and conditions can affect a person's need for assistance.

5. Knowledge and expertise –cooperation, research and registries

Assessment, treatment and research all rely on good patient registries and international cooperation. Good patient registries require a common system for coding patients with rare diseases, as well as good national coverage. It is essential for Norwegian professional communities to actively participate in international research, as the patient base for rare diagnoses is very small. It is also important to strengthen efforts to develop and disseminate expertise in each professional area throughout the country.

2 Strategy Follow-Up

The Norwegian Government's objectives:

- that user involvement is facilitated at the individual, service and system levels
- that people with rare diagnoses and conditions will receive equal and fast access to high-quality, multidisciplinary assessments, diagnostics and patient care
- that people with rare diagnoses and conditions receive good, comprehensive patient pathways and coordinated services with a life course approach
- that patients are given equal access to a multidisciplinary evaluation and assessment of cognitive, physical and psychosocial functions with a focus on mastery, activity and participation
- that research and knowledge advancements in the area of rare disorders and diagnoses are strengthened through national and international research and network cooperation
- that knowledge of assessments, diagnostics, treatment and follow-up of rare diseases and conditions are strengthened in healthcare services

The Norwegian Government will implement the following initiatives to achieve the above-mentioned objectives:

- 1. Regional health authorities shall ensure that the infrastructure for genetic testing includes rare diagnoses.***

This initiative will contribute towards better diagnostics and treatment, as well as increased participation in clinical studies.

- 2. The Norwegian Directorate of Health shall, in cooperation with regional health authorities, ensure that we have a dynamic system for including new diseases in neonatal screening.***

This initiative will contribute towards better diagnostics, assessments and treatment, as well as research.

- 3. Regional health authorities are commissioned, in cooperation with user organisations and other relevant actors, to evaluate how equal and fast access to highly specialised assessments, diagnostics and treatment can best be ensured by specialist health services through national and possibly regional services.***

This initiative will contribute towards better and faster diagnostics, treatment and follow-up of patients with rare diagnoses and disorders, and to the dissemination of knowledge and expertise among healthcare personnel and services. It is assumed that the national competence services for rare diagnoses will be discontinued as national competence centres, but that the activities and professional expertise will be maintained and developed. The evaluation must determine the opportunities for better diagnostics, treatment and follow-up from a life course perspective, as well as the development of competence through restructuring and mandates. Highly specialised assessments and competence development should be viewed in the context of Norwegian participation in the ERN, and the establishment of national professional networks associated with this.

- 4. Regional health authorities will facilitate increased Norwegian participation in the European Reference Networks (ERN) and establish formalised Norwegian professional networks in areas where Norway is represented.***

This initiative will contribute towards faster and more accurate diagnostics, as well as increased cooperation on research and dissemination of knowledge.

- 5. *The Norwegian Directorate of Health is commissioned to establish a national forum for participants within and around the work of the European Reference Network, including the participation of user organisations.***

This initiative entails the establishment of a forum as an arena for sharing information about experiences and the status of the efforts to establish and operate the various networks and associated quality registries. This initiative will contribute towards increased coordination between the various networks, and between the networks and the regional health authorities, the Norwegian Directorate of Health, and the Ministry of Health and Care Services in their work with the ERN.

- 6. *The Norwegian Directorate of eHealth is commissioned, in cooperation with the Norwegian Directorate of Health and the regional health authorities, to evaluate whether the current coding system and ongoing initiatives meet the needs of rare diagnoses, including ICD-11 and ORPHA codes.***

This initiative will contribute towards improving and strengthening diagnostics, research and knowledge development.

- 7. *Regional health authorities are commissioned to evaluate and, where appropriate, establish a national registry for rare disorders based on the National Advisory Unit on Rare Disorders established at Oslo University Hospital Trust.***

This initiative will contribute towards improving and strengthening diagnostics, research and knowledge development.

- 8. *The Norwegian Directorate of Health is commissioned, in cooperation with user organisations, the Norwegian Association of Local and Regional Authorities (KS) and regional health authorities, to evaluate how service user needs for coping and mastery from a life course perspective can be better addressed for patients with rare diagnoses and disorders.***

This work must be viewed in the context of ongoing assessments of learning and mastery services for patients. This initiative will contribute towards more comprehensive assessments, evaluations and follow-up of patients with rare diagnoses and disorders from a life course perspective.

- 9. The Norwegian Directorate of Health is commissioned, in cooperation with relevant actors and professional communities, to assess and prioritise Norway's participation in various international forums, and to propose appropriate support from and participation of users, professional communities and health authorities.***

This initiative will contribute towards increased research, knowledge and competence development, as well as better diagnostics and treatment.

- 10. The health sector, education sector, specialist health services and Statped are commissioned with evaluating how cooperation can be improved with the aim of achieving a more coordinated range of services.***

This initiative is a follow-up of the Report to the Parliament (White Paper) 6 Early intervention and inclusive education in kindergartens, schools and out-of-school-hours care.

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