# National programme for rare diseases 2024–2028

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Finnish institute for health and welfare

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## Foreword

Rare diseases form a significant group of more than 7,000 different diseases. Whatever the disease, the challenges patients face related to the diagnostics and treatment of their rare disease and coping with their everyday life are often similar.

After two national programmes for rare diseases, many of the objectives set are yet to be achieved. The present third programme has set the objectives that should be achieved in the next five-year period as its strategic priorities. The appendices describe the current state of the structures supporting the treatment and diagnostics of rare diseases in Finland.

In the future, we will be increasingly moving towards the common European strategy for rare diseases. European and international cooperation plays an important role in achieving the objectives described in this programme. It is important that Finland stays on track in the European development, and at the national level, rare diseases must be taken into account as a special group in the planning and implementation of social welfare and health care services.

On behalf of the national expert working group on rare diseases,

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## Abstract

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More than 7,000 different rare diseases have been described. An estimated 6-8% of the population has a rare disease, disability, or syndrome. Together, rare patients form a large group of patients whose diagnosis, treatment and services require special attention. At the EU level, the challenges of rare diseases are being met with the help of virtual knowledge networks for rare diseases (ERN, European Reference Networks for low prevalence and complex diseases) established by the European Commission. In addition, the EU countries have drawn up their national programs to support diagnostics, treatment, implementation of services and patient participation in rare diseases. In Finland, two national programs for rare diseases have been prepared in the past, for the years 2014-2017 and 2019-2023.

The third national program for rare diseases complements previous programs. The program highlights the most important areas for development and strategic goals aimed at strengthening the treatment and participation of people with rare diseases. In the next five-year period, the focus will be on developing registries and the data base, strengthening structures, creating treatment and service paths, ensuring the availability of medicines and treatment, securing research conditions and ensuring patient participation. In addition, one of the goals is to consider patients without a diagnosis and try to meet the challenges of this patient group as well.

**Keywords:** Rare diseases, diagnostics, care, register information, participation

## Contents

# Introduction

A disease is defined as a rare disease when it affects no more than five persons per 10,000 (so-called European definition) (1). The majority of rare diseases are still much rarer than this (2). It is estimated that 6–8% of the population have a rare disease.

More than 7,000 different rare diseases have been described. This group includes various diseases, injuries and syndromes already diagnosed in the neonatal period or in early childhood. However, many adult-onset diseases, such as certain neurodegenerative diseases, metabolic diseases and even cancers, are also rare (3).

At the recommendation of the Council of the European Union, the Member States have drawn up their national programmes for the treatment of rare diseases (4). Finland has drawn up two national programmes, for the periods 2014–2017 and 2019–2023. The key objectives of these programmes include increasing knowledge and competence, improving diagnostics and treatment, supporting patient participation and equality, developing the service system, promoting research and coordinating activities (5, 6).

The newly-found interest in the challenges of rare diseases at the EU level has also led to the establishment of European Reference Networks (ERNs) (7). These virtual reference networks aim to promote the diagnosis and treatment of rare diseases and the collection of data. The members of each of the 24 networks include at least one Finnish centre of excellence operating in a university hospital. At the national level, the units for rare diseases operating in each of the five university hospitals help coordinate the treatment of rare diseases.

After the completion of the two national programmes, there has been progress in many areas in rare diseases. Awareness of diseases has increased and the efficiency of the coordination of treatment and activities has improved. Networking has increased both nationally and through the European Reference Networks. However, there are still challenges and development needs related to patients' everyday coping, inclusion and the implementation of diagnostics and treatment. As a result of the health and social services reform, the links between the new wellbeing services counties and the centres of expertise for rare diseases are only beginning to emerge.

The present third national programme for rare diseases highlights the key points of the previous programmes as the strategic priorities for the next programme period. Instead of drawing up a new extensive programme, the aim is to focus on such tangible objectives that can be pursued with measures that can be taken with the current resources. The aim is also to set strategic objectives in a way that fosters easy monitoring and assessment at the end of the programme period. The results will be described at the end of the programme period in a separate report or as part of the next national programme.

# Strategic objectives

## 1. Building registers and knowledge base and sharing information

The aim is to introduce ORPHAcodes for rare diseases throughout specialised medical care as well as to produce training and materials related to the codes. A further aim is to examine the prerequisites and legislation for national and European quality register activities related to rare diseases and to prepare guidelines. Systematic data registration enables the monitoring of the treatment of rare diseases and patient participation.

## 2. Strengthening structures

The aim is to establish and strengthen the activities related to rare diseases at the national and international levels. This particularly applies to units for rare diseases in university hospitals, Finnish ERN centres of excellence, organisations and associations, the coordination of national activities as well as the networking and international cooperation between the operators. To achieve this objective, the integration of ERN centres of excellence into the national service system is essential. Efforts will be made to strengthen competence in rare diseases as a part of further education for social and health care professionals.

## 3. Preparing care and service pathways

The aim is to prepare care and service pathways to guide the diagnosis, treatment, and rehabilitation of rare diseases as well as the access to services, special aids and care supplies. A further aim is to ensure the equal implementation of diagnostics, treatment and accessible services in wellbeing services counties. Efforts will be made to improve the situation of people with a rare disease without a diagnosis by identifying them as a specific group and by responding to their diagnostic challenges.

## 4. Ensuring the availability of medications and care and assessing their benefits individually

The aim is to develop the national monitoring of the impacts and adverse effects of orphan drugs and to ensure the availability of medication provided in outpatient care. A further aim is to promote an individual assessment of the benefits of Orphan drugs and their discretionary reimbursability.

## 5. Safeguarding the conditions for the research of rare diseases

The aim is to collect and publish up-to-date information from key national research projects on rare diseases and international cooperation projects. A further aim is to promote opportunities for allocated research funding and international research cooperation.

## 6. Strengthening the inclusion of patients with rare diseases

The aim is to provide patients and their families as well as professionals with information on rare diseases and relevant associations and organisations as well as to organise training events. A further aim is to examine and strengthen the inclusion of patients with rare diseases in activities promoting a client-driven approach to social welfare and health care, such as client panels and peer support activities.

# Appendices: The current status of the field of rare diseases

## Knowledge and awareness of rare diseases

Discussion on rare diseases has increased, and a growing number of key operators in the health and social services sector have integrated rare diseases more strongly into their planning of activities.

Information on rare diseases in Finnish is most comprehensively available in Health Village (Healthvillage.fi), Duodecim Terveysportti (in Finnish, Terveysportti) and Terveyskirjasto (in Finnish, Terveyskirjasto) and on non-governmental organisations' websites (Norio – Centre of Rare Diseases, The Finnish Network for Rare Diseases and HARSO – The Finnish Alliance of Rare Diseases and Disabilities Organizations). In addition, the European Orphanet database is presented on the Orphanet website available in Finnish (in Finnish, Orphanet).

The national coordination of rare diseases at the Finnish Institute for Health and Welfare (THL) has its own project website (thl.fi/rarediseases), and the THL Disability Services Online Handbook includes a section focusing on rare diseases (in Finnish, Harvinaissairaudet). THL has also published Know & Act cards for decision-makers and social workers in the wellbeing services counties on the special features of rare diseases and issues to be considered in decision-making and the ORPHAcodes (8–10). An online course on rare diseases has been prepared for the Duodecim Oppiportti learning platform. The previous national programmes for rare diseases have compiled information related to rare diseases and the situation in the field of rare diseases from various perspectives (5, 6).

So far, register data related to rare diseases has not been available at the national level. However, the Orpha classification system used in identifying rare diseases has been added to THL's code server in connection with the ICD-10 diagnostic classification used in health care. In addition, the code server has been made to include alarms for any rare diseases whose acute treatment differs from normal. The use of ORPHAcodes in patient information systems was introduced in all university hospitals in 2023. A data field designated to the ORPHAcodes was included in the THL Care Register for Health Care (Hilmo) in June 2023. This makes it possible to integrate the ORPHAcodes recorded in university hospitals into national register data.

## National operators and cooperation networks

Key national operators in the field of rare diseases include social welfare and health care units, units for rare diseases and the ERN centres of expertise in university hospitals. Organisations play an important role in the national field of rare diseases, and they also operate as part of international patient organisation networks. Official tasks related to rare diseases are jointly managed by the Finnish Institute for Health and Welfare and the Ministry of Social Affairs and Health, which are also responsible for Finland's activities in international networks of authorities (Figure 1).

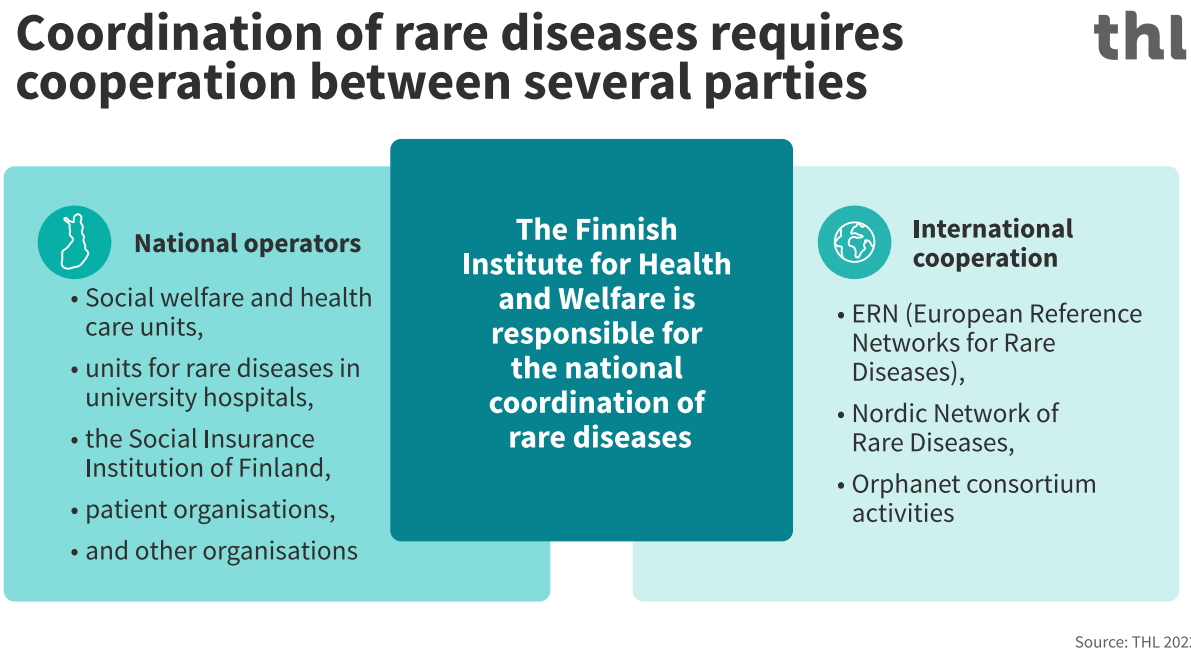


**Figure 1. Structure and operators in the field of rare diseases.**

## Implementation of diagnostics, treatment and services

National and EU legislation has gradually started to identify rare diseases as a separate group. However, in Finland, the implementation and staggering of care are influenced by the Decree on the division of labour in specialised medical care and centralisation of certain tasks (1242/2022). The Decree specifies that the prevention, treatment, diagnostics and rehabilitation of rare diseases are the responsibility of the university hospitals.

Screening for rare diseases is carried out as a part of neonatal screening (11). As many rare diseases are genetic and appear in the neonatal period or in early childhood, child health clinics play a key role in suspected cases of rare diseases and referring patients to further treatment. It appears that we are more prone to suspect rare diseases in children than adults.

While carrier screening for gene defects related to rare diseases is not more widely used in Finland, some private companies offer carrier studies for Finnish disease heritage (12). For other diseases, carrier studies are mainly conducted by the hereditary clinics of university hospitals in families found to have a hereditary disease or susceptibility to cancer. Carrier screening can also be carried out as necessary with a referral from the treating physician to a laboratory offering genetic testing.

Suspicion of diseases and referral of patients to university hospitals play a key role in the diagnosis of rare diseases. If necessary, the units for rare diseases in university hospitals and the Finnish and European ERN centres of excellence provide assistance in the coordination and implementation of treatment (Figure 2). For individual diseases, treatment pathways have been described neither in Finland nor at the European level. Little is known about the situation of rehabilitation, psychosocial support and the need for social services among people with rare diseases post medical diagnostics and the start of treatment.

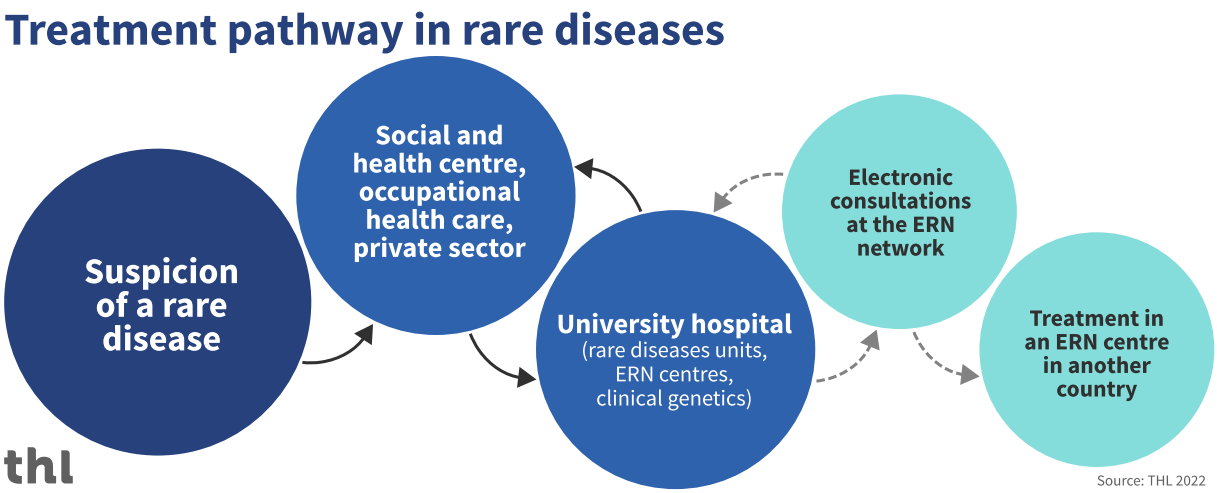


**Figure 2. Treatment pathway in rare diseases.**

A rare disease affects not only the patient's life but also often the lives of people close to them and the whole family. Many patients with rare diseases seek the services offered by organisations to obtain information and support. The organisations have special expertise related to rare diseases and living with them and offer specialised services for people with rare diseases and their families. The organisations also have long-term experiential knowledge of the need for rehabilitation, psychosocial support and social services.

## Orphan drugs

Marketing authorisations for orphan drugs have been granted to over one hundred medications. Orphan drugs are expensive and assessing their cost impacts is challenging due to their small user group and limited research data (13). An orphan drug can also be granted conditional marketing authorisation if its use can be justified by an unmet medical need.

The costs of these medical treatments are covered by the funding of medical treatment of public health care or, in the case of drugs prescribed in outpatient care, by Kela's system of reimbursements for medicine expenses. As with other medications, the Pharmaceuticals Pricing Board (Hila) decides whether orphan drugs are reimbursable by Kela. The reimbursability status requires that the drug has a reasonable wholesale price. Since the beginning of 2018, there has also been an opportunity for the conditional substitution of drugs based on an agreement between the Pharmaceuticals Pricing Board and the pharmaceutical company and which involves agreeing on risk-sharing models in connection with the introduction of the new drugs. Pharmaceutical companies decide on the prices of drugs outside the reimbursement system used in outpatient care.

The costs of orphan drugs are too high for patients to pay. While the pricing and reimbursability process for orphan drugs is the same as for any other medicines in Finland, but in many countries, the introduction of orphan drugs is a faster and smoother process.

## Research on rare diseases

According to an estimate by the Academy of Finland in early 2023, the Academy's Scientific Council for Health granted a total of EUR 38 million (approximately EUR 6.3 million per year) to 117 research projects related to rare diseases between 2017 and 2022. In an earlier report covering the period 2011–2017, the number of research projects was 57 and funding amounted to more than EUR 20 million (approximately EUR 2.9 million per year). The Academy of Finland is no longer involved in the EJP RD (European Joint Programme on Rare Diseases) project focused on rare diseases or in the international IRDiRC (International Rare Diseases Research Consortium) consortium.

Most of the research related to rare diseases is carried out with project or separate funding granted by foundations, universities or hospitals. Information on Finnish research projects on rare diseases has been compiled on the Orphanet website in connection with the description of each disease and, for individual projects, on the websites of hospitals and universities, for instance.

## Participation of persons with rare diseases

Medicine cannot solve all the challenges faced by people with rare diseases. People with rare diseases often experience multiple symptoms, and their treatment requires professionals and specialised employees from various fields. Coping with everyday life requires different social and health care services and social benefits. Support and peer support provided by the third sector also play a major role in the everyday lives of people with rare diseases.

The participation of people with a rare disease in their own care and the planning of services and in research was discussed extensively in the previous national programme. It is important to ensure participation at the levels of society and local communities alike. At the national level, the most important thing is to ensure the participation of persons with rare diseases in the development of social welfare and health care services and the implementation of their own treatment and services.

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