Working group on rare diseases

National programme for rare diseases 2019–2023

REPORTS AND MEMORANDUMS OF THE MINISTRY OF SOCIAL AFFAIRS AND HEALTH 2020:26

Reports and Memorandums of the Ministry of Social Affairs and Health
2020:26

Report of the Working Group on Rare diseases

National Programme for Rare Diseases 2019–2023

Ministry of Social Affairs and Health, Helsinki 2020

Ministry of Social Affairs and Health
ISBN PDF: 978-952-00-5670-4
Layout:: Government Administration Department, Publications
Helsinki 2020

Published by	Ministry of Social Affairs and Health		2020
Authors	Working group on rare diseases		
Title of publication	National programme for rare diseases 2019–2023		
Series and publication number	Reports and Memorandums of the Ministry of Social Affairs and Health 2020:26		
Register number	STM025:00/2016	**Subject**	
ISBN PDF	978-952-00-5670-4	**ISSN (PDF)**	2242-0037
Website address (URN)	http://urn.fi/URN:ISBN:978-952-00-5670-4		
Pages	46	**Language**	English
Keywords	rare diseases; care chains, rehabilitation, social welfare, health care, equality rare diseases; care chains, rehabilitation, social welfare, health care, equality		

Abstract

It is estimated that six out of one hundred Finns have a rare disease, injury, syndrome or malformation. It can sometimes be challenging and time-consuming to identify them. It may be difficult to provide care, rehabilitation and everyday support for people with rare diseases, and there may be large regional variation in services. Therefore, the regional coordination for the prevention, diagnostics, care and rehabilitation of rare diseases has been centralised to five university hospitals in Finland.

Measures descibed in this programme are still required to improve the inclusion and everyday coping of people with rare diseases, as well as their equity in access to services. The working group also proposes strengthening the national coordination and means for increasing competence and information. Participation of Finnish university hospitals in European reference networks makes expertise available to ever more people with rare diseases. Funding is needed for research on rare diseases. Limited resources for producing and sharing information can be used more effectively by cooperation, in order to provide information about rare diseases in easily understandable form in the native languages of Finland.

Publisher	Ministry of Social Affairs and Health
Distributed by/ publication sales	Online version: julkaisut.valtioneuvosto.fi Publication sales: vnjulkaisumyynti.fi

Julkaisija	Sosiaali- ja terveysministeriö		2020
Tekijät	Harvinaiset sairaudet -työryhmä		
Julkaisun nimi	Harvinaisten sairauksien kansallinen ohjelma 2019–2023		
Julkaisusarjan nimi ja numero	STM raportteja ja muistioita 2020:26		
Diaari/hankenumero	STM025:00/2016	**Teema**	
ISBN PDF	978-952-00-5670-4	**ISSN PDF**	2242-0037
URN-osoite	http://urn.fi/URN:ISBN: 978-952-00-5670-4		
Sivumäärä	46	**Kieli**	englanti
Asiasanat	harvinaiset taudit, hoitoketjut, kuntoutus, sosiaalihuolto, terveydenhuolto, yhdenvertaisuus		

Tiivistelmä

Arviolta kuudella sadasta suomalaisesta on harvinainen sairaus, vamma, oireyhtymä tai epämuodostuma. Niiden tunnistaminen voi olla haastavaa ja aikaa vievää. Harvinaissairaan hoito, kuntoutus ja arjen tuki ovat joskus vaikeita toteuttaa, ja palvelujen alueellinen vaihtelu saattaa olla suurta. Sen vuoksi Suomessa harvinaisten sairauksien ehkäisyn, diagnostiikan, hoidon ja kuntoutuksen alueellinen koordinaatio on keskitetty viiteen yliopistosairaalaan.

Harvinaissairaiden osallisuus, arjessa selviytyminen ja yhdenvertaisuus palvelujen saamisessa vaativat edelleen toimenpiteitä, joita on kuvattu ohjelmassa. Työryhmä ehdottaa myös kansallisen koordinaation vahvistamista sekä keinoja osaamisen ja tiedon lisäämiseksi. Suomalaisten yliopistosairaaloiden osallistuminen eurooppalaisiin osaamisverkostoihin tuo erityisosaamisen yhä useamman harvinaissairaan saataville. Harvinaissairauksien tutkimukseen tarvitaan rahoitusta. Yhteistyö tiedon tuottamisessa ja jakamisessa auttaa kohdentamaan rajalliset voimavarat tehokkaasti, jotta tarjolla olisi mahdollisimman monille helposti ymmärrettävää tietoa harvinaissairauksista heidän äidinkielellään.

Kustantaja	Sosiaali- ja terveysministeriö
Julkaisun jakaja/ myynti	Sähköinen versio: julkaisut.valtioneuvosto.fi Julkaisumyynti: vnjulkaisumyynti.fi

Utgivare	Social- och hälsovårdsministeriet		2020
Författare	Arbetsgrupp för sällsynta sjukdoma		
Publikationens titel	Publikation (även den finska titeln) Nationella programmet för sällsynta sjukdomar 2019–2023		
Publikationsseriens namn och nummer	Social- och hälsovårdsministeriets Rapporter och promemorior 2020:26		
Diarie-/ projektnummer	STM025:00/2016	**Tema**	
ISBN PDF	978-952-00-5670-4	**ISSN PDF**	2242-0037
URN-adress	http://urn.fi/URN:ISBN:978-952-00-5670-4		
Sidantal	46	**Språk**	engelska
Nyckelord	sällsynta sjukdomar, vårdkedjor, rehabilitering, socialvård, hälso- och sjukvård, jämlikhet		

Referat

Uppskattningsvis sex av hundra finländare lider av någon sällsynt sjukdom, skada eller missbildning eller något sällsynt syndrom. Att identifiera dessa kan vara utmanande och tidskrävande. Det kan ibland vara svårt att genomföra vården, rehabiliteringen och stödet i vardagen för personer med sällsynta sjukdomar, och den regionala variationen i servicen kan vara stor. I Finland har den regionala samordningen av förebyggande, diagnostisering och behandling av sällsynta sjukdomar samt rehabilitering för personer som lider av sådana därför koncentrerats till fem universitetssjukhus.

Sådana åtgärder som beskrivs i programmet krävs fortfarande för att de som lider av sällsynta sjukdomar ska vara delaktiga, klara vardagen och ha jämlik tillgång till tjänster. Arbetsgruppen föreslår också starkare nationell samordning och metoder för att öka kompetensen och kunskaperna. Att finländska universitetssjukhus deltar i europeiska referensnätverk gör att allt fler personer som lider av sällsynta sjukdomar kan komma i kontakt med experter med specialkunskaper. Det behövs finansiering för forskning om sällsynta sjukdomar. Samarbete vid produktion och spridning av kunskap och information gör det lättare att använda de knappa resurserna effektivt, så att det finns information om sällsynta sjukdomar i lättbegriplig form och på människors eget modersmål.

Förläggare	Social- och hälsovårdsministeriet
Distribution/ beställningar	Sähköinen versio: julkaisut.valtioneuvosto.fi Julkaisumyynti: vnjulkaisumyynti.fi

Sisältö

TO THE READER 8

1 Introduction 10

2 Implementation of programme objectives in 2014-2017 and changes in the operational environment 13

2.1 Adopting the definition of rare diseases and acknowledging the need for dedicated measures 15

2.2 Units and centres of expertise for rare diseases 15

2.3 Research funding and research cooperation 16

2.4 Disseminating information and strengthening knowledge 17

2.5 National registry for rare diseases and streamlining of care pathway 18

2.6 Social support, rehabilitation and augmentation of involvement 18

2.7 National coordinating centre for rare diseases 19

3 Pivotal objectives of the National Programme 2019–2023 20

3.1 Increasing knowledge on rare diseases and strengthening expertise 20

3.1.1 Increasing knowledge and awareness 20

3.1.2 Strengthening the status of Rare Diseases Units and allocation of resources to them 24

3.1.3 European Reference Networks (ERN) 26

3.1.4 Promoting research 29

30

3.2 Strengthening of patient involvement in rare diseases 30

3.2.1 Empowerment and involvement of people with rare diseases in society and their communities 31

3.2.2 Involvement of people with rare diseases in developing healthcare and social welfare services in their own areas 33

3.2.3 Safeguarding the involvement of people with rare diseases in their care and services 35

3.3.1 National coordination 37

3.3.2 Regional coordination 40

3.3.3 Coordination of care and services of a person with rare diseases 41

3.4 Estimated cost effects of the National Programme 42

3.5 Practical implementation of the National Programme and monitoring it 43

Sources 45

TO THE READER

In Finland, all citizens are entitled to equal access to healthcare and social welfare services. However, equity in health care services is not always easy to achieve for people with rare diseases. Their diseases may be difficult to identify, specialists may be few, there are no effective treatment methods, or the available level of evidence for efficacy may be low.

The first Finnish National Programme for Rare Diseases 2014-2017 followed the recommendations of the Council of the European Union. The programme detailed a plan for various necessary actions, but not all of its objectives were accomplished. Over the past few years, the operational environment in which rare diseases are treated has changed considerably for a variety of reasons, such as the establishment of European Reference Networks. Therefore, it was time to revise the programme.

Three main themes have been highlighted in the revised National Programme for Rare Diseases: the involvement of people with rare diseases, augmenting expertise and strengthening coordination. These themes are examined at different levels of the Finnish service system. This Programme is not a detailed action plan, but it rather outlines the general direction for decision-makers and people working with rare diseases, who on their behalf are responsible for the measures and monitoring of this programme during its implementation.

The programme aims at enhancing the involvement of people with rare diseases in decisions concerning themselves, and at intensifying collaboration of rare disease patient organisations in improving healthcare and social welfare. Other key objectives include providing up-to-date and correct information in a manner that is easy to understand, and reinforcing competence in healthcare and social welfare. The expertise provided by the European Reference Networks will likely improve the availability and quality of diagnostics and care. Promoting cooperation between different parties will improve regional and national coordination. To successfully implement coordination, programme calls for nationally agreed division of tasks.

The National Programme was drafted in the Sub-Committee of the Working Group on Rare Diseases, established by the Ministry of Social Affairs and Health. We would like to thank the Sub-Committee for its invaluable work. The Programme was edited by a representative of the Finnish Network for Rare Diseases, Risto Heikkinen from the Finnish Federation for Allergy, Skin and Asthma. All key stakeholders have commented on it before publication.

It is of paramount importance that all parties working with rare diseases commit themselves to the implementation of this Finnish National Programme for Rare Diseases 2019-2023.

July 2019

Working Group on Rare Diseases

1 Introduction

Approximately more than 300,000 Finns have a rare disease, disability, syndrome or malformation. There are thousands of rare diseases and they are highly variable. Some present in childhood, others only at the adulthood. A rare disease may be barely noticeable or cause severe impairment.

There are between 6,000 and 8,000 known rare diseases with new described constantly in medical literature. In Europe, the definition of rare diseases covers diseases that affect no more than 5 in 10,000 people. In Finland, this implies 2,800 individuals per disease at most. Approximately, over 300,000 Finns or about six per cent of the population have some kind of rare disease, disability, syndrome or malformation. Rare diseases are a very heterogeneous group of illnesses. Some rare diseases are diagnosed soon after birth, others not until adulthood. Some of these progress in severity, while others do not. Sometimes a sub-form of a common disease may be rare. Therefore, one should consider the person's own evaluation of his or her status and outcome and the challenges related to it.

There are strong justifications for updating the National Plan for Rare Diseases and setting new targets and action proposals. In an extensive European survey (Eurordis 2017), more than 50% of people with rare diseases and their families reported that the disease had serious or very serious impacts on their everyday life, such as on their ability to cope with everyday routines or personal care. The UN Convention on the Rights of Persons with Disabilities from 2006 was ratified in Finland in 2016 and has influenced our legislation, the patient's and the customer's status, and the healthcare and social welfare practices in Finland. The Convention emphasises the importance of strengthening the involvement and right to self-determination of people with disabilities and chronic diseases. This also improves the status of people with rare diseases as users of the service system.

The costs incurred by rare diseases in specialised medical care highlight the need to update the National Programme for Rare Diseases and set new objectives.

According to a survey concerning the period 2010-2014, individuals with rare diseases accounted for between 6% and 8% of patients receiving specialised medical care in the Hospital District of Helsinki and Uusimaa and the Northern Ostrobothnia Hospital District. However, the costs related to rare diseases in specialised medical care in these two hospital districts were slightly under 20% of the overall costs of specialised medical care. This estimate did not include the costs of rehabilitation, social services or outpatient healthcare. For example, costs result from delays and failures of diagnostics or appropriate treatment. The methods used in diagnostics and treatment of rare diseases may also be expensive.

In 2014, the Working Group on Rare Diseases established by the Ministry of Social Affairs and Health compiled a proposal for the Finnish National Programme for Rare Diseases. The programme was based on the recommendation issued by the Council of the European Union in 2009, which obligated the Member States to strengthen their plans and strategies for rare diseases. Finland's National Programme described the plight of individuals living with rare diseases and the challenges they faced. It presented 16 general objectives and 13 concrete proposals for the development of research, care, rehabilitation and social support for people with rare diseases. The programme period ended at the end of 2017. The term of the Working Group on Rare Diseases promoting the implementation of the National Programme ended at the same time. Thereafter, the working group was granted an extension from January 1st 2018 to December 31st 2019 with the following tasks to:

1. Define and draw up a proposal for strengthening national coordination in the field of rare diseases.

2. Assess the need to update the National Programme for Rare Diseases and plan the update.

3. Monitor the operation of European Reference Networks in Finland.

4. Monitor and promote the cooperation between the Rare Diseases Units in university hospitals and the implementation of tasks laid down in the Government Decree on the Division of Work in Specialised Medical Care and Centralisation of Certain Tasks (582/2017).

5. Support the operation of the Nordic Network on Rare Diseases.

The preparation and implementation of the tasks were given to the Sub-committee set up by the Working Group. In spring 2018, the Working Group decided to update the National Plan for a four-year period, as proposed by the Sub-Committee. Risto Heikkinen, a member of the working group, was chosen for the task of coordinating the update and writing the revised programme. Individuals with rare diseases, their families and key professionals from the field of rare diseases were involved in the update. A survey to organisations and associations representing rare diseases was conducted during autumn 2018. A patient advocacy group (Harkko) and other member communities of the Finnish Network for Rare Diseases (www.harvinaiset.fi), consisting of 21 social and health organisations, were in various ways involved in evaluating the implementation of the first National Programme and in updating it. In addition, a survey to the five Rare Disease Units in university hospitals and to the 14 Finnish healthcare provider members of ERN networks was conducted. The discussions and results from the national conference on rare diseases

organised in October 2018 further complemented the programme. Before its finalisation, the updated draft programme was sent to the stakeholders for comments.

The revised plan provides a general overview of the current situation, evaluates the progress made in the implementation of the actions proposed in the old programme and, based on this, proposes new actions. The purpose of the programme is to develop the social and healthcare service system to better meet the needs of people with rare diseases. In addition, the intention is to promote the daily coping abilities of people with long-term illnesses and disabilities, who during their lives face similar challenges to people with rare diseases.

The revised programme emphasizes the importance of expertise, involvement and coordination. It aims at improving the position of people with rare diseases, and the ability of their families, friends and themselves to cope in their daily lives by speeding up the diagnosis and promoting their access to high-quality care and rehabilitation. The objectives for the upcoming programme period are to increase the amount of up-to-date and correct information, to reinforce expertise and to promote cooperation between different parties by strengthening national coordination. The specialist expertise and the tools provided by the European Reference Networks are a new way of improving the availability and quality of diagnostics and care. Integration of the Reference Networks into the Finnish service system requires cooperation and descriptions of care pathways. To implement the coordinating tasks in practice, division of work also needs to be agreed at the national level.

2 Implementation of programme objectives in 2014-2017 and changes in the operational environment

General awareness of rare diseases increased during the first programme period. The Units for Rare Diseases and regional coordination were launched, with new centres of expertise at university hospitals. Cooperation at the Nordic and European levels increased. Nevertheless, many objectives were only partially implemented while changes in the operational environment lead to identification of previously unrecognized demands.

Thirteen actions were proposed for the programme period 2014-2017 to promote the definition and registration of rare diseases, research, improved and more efficient healthcare, coordination of expertise, knowledge sharing, provision of holistic support for individuals with rare diseases, and strengthening their involvement.

PROPOSED ACTIONS 2014–2017

- Adopting a uniform definition for rare diseases and acknowledging the need fordedicated measures
- Establishing a national registry for rare diseases
- Focused research funding and research programme for rare diseases
- Strengthening of international research collaboration
- Streamlining the care pathway for an individual with rare disease
- Establishing Units for Rare Diseases in all university hospitals
- Establishing specialized Centres of Expertise for Rare Diseases
- Providing more education and training
- Promoting the availability of orphan drugs
- Establishing a national coordinating centre for rare diseases
- Systematic collection and sharing of information
- Developing social support and rehabilitation
- Augmenting rare disease patient involvement

Implementation of the proposed actions has not comprehensively been evaluated. Docent Ilona Autti-Rämö, a member of the Working Group on Rare Diseases, presented observations on the first National Programme and its implementation at the National Conference on Rare Diseases held in spring 2017. According to her estimate, the only fully implemented actions were the adoption of a uniform definition for rare diseases, and the establishment of Rare Diseases Units and Centres of Expertise for rare diseases. The other proposed actions had progressed variably.

OBSERVATIONS ON THE IMPLEMENTATION OF THE FIRST PROGRAMME OBJECTIVES:

- on a general level, the programme strived towards the identification of development areas and recording the challenges affecting the life of individuals suffering from rare diseases
- the implemented actions were those that were achievable by developing or changing existing operations
- the measures requiring national agreement have been implemented only partially
- as of now, the needs of individuals with rare diseases have been insufficiently acknowledged in legislation to for example enable equitable care pathways
- actions requiring new resources, or redivision of resources or funding, were largely not implemented

EU Member States share the need to develop the diagnostics, care and expertise related to rare diseases, as no single country will be able to improve the situation of people with rare diseases on its own. International and Nordic Cooperation has indeed strengthened considerably between authorities, social welfare and healthcare professionals, and patient organisations, although the measures proposed in the first programme only emphasised the importance of internationalisation in the research community.

Our impression is that for the last few years the concept of rare diseases has become better understood, probably at least partly due to the first National Programme. In the following section, the implementation of the proposed actions in each area of the programme will be assessed in detail.

2.1 Adopting the definition of rare diseases and acknowledging the need for dedicated measures

Finland has now put to use the European definition for rare diseases, according to which a disease is rare if it affects no more than 5 in 10,000 people. Like the first National Programme for Rare Diseases, this programme uses the concept of rare diseases to also refer to rare disabilities, malformations and symptoms.

The legal status of people with rare diseases has been addressed in legislative amendments, government bills concerning the health and social services reform and

in the background material for the reform. An Act amending the Health Care Act (1516/2016), which entered into force in 2017, made section 45 of the Health Care Act (1326/2010) more specific, with an obligation to centralise certain medical tests, procedures and care requiring highly specialised expertise to fewer than five university hospitals. The Government Decree on the Division of Work in Specialised Medical Care and Centralisation of Certain Tasks (582/2017) obligates the five hospital districts maintaining a university hospital to perform, plan and coordinate certain tasks in specialised medical care regionally. The prevention, care, diagnostics and rehabilitation of rare diseases are part of this obligation.

The ratification of the UN Convention on the Rights of Persons with Disabilities in 2016 improves the status of people with disabilities by prohibiting all kinds of discrimination against them. The Convention emphasises the human rights and equity of people with disabilities and requires that people with disabilities and the organisations representing them to be included in the planning and decision-making concerning them. The principles described in the Convention also touch on the position of individuals with rare diseases, The Convention emphasises the importance of the accessibility of the physical, social and mental environment. For example, ensuring equal access to information and interaction are an important part of accessibility.

2.2 Units and centres of expertise for rare diseases

The establishment of Rare Diseases Units in all five university hospitals and the membership of a total of 14 centres of expertise from four university hospitals as healthcare provider representatives in the European Reference Networks for Rare and Low Prevalence Complex Diseases (European Reference Networks, ERN) have been important reforms in promoting the diagnostics and treatment of individuals with rare

diseases. The status of Rare Diseases Units (and/or Centres) has been strengthened with the Governmental Decree on Centralization of Specialist Health Care and Some Other Actions (582/2017). For their part, these measures have improved the status and involvement of individuals living with rare diseases. The importance of the status and tasks of these units and centres is emphasised in the objectives and proposed actions of this National Programme.

To implement the health sector growth strategy, four National Centres of Excellence that have been highlighted as key strategic focus areas are in the process of being established in Finland. The goal of a project entity aimed at better utilisation of genomic data is to establish a national Genome Centre. Legislation in the use of

genomic data and legislative clarification of the status of biobanks is currently in preparation. Among other things, biobanks collect and store biological samples for rare disease research. In addition to five regional cancer centres, a national cancer centre

(Comprehensive Cancer Centre Finland, FICAN) has started in 2019 while the establishment of a Neurocentre Finland is in preparation. These projects will improve the operating conditions of personalised medicine in healthcare. Personalised medicine plays an important role also in the treatment of rare diseases.

2.3 Research funding and research cooperation

Finland has conducted no comprehensive study of research focusing on rare diseases. According to the estimate by the Academy of Finland in 2017, the Academy's Research Council for Biosciences, Health and the Environment had granted just over EUR 20 million to 57 research projects focusing or touching upon rare diseases between 2011 and 2017. Excluding research in rare cancers and infectious diseases, the overall funding for research in rare diseases was EUR 19 million. This is less than 10% of funding from the Research Council. Of the 24 rare disease groups included in the ERNs, 10 research fields remained completely unsupported during the time of the estimate, despite internationally effective research in many of them. These disease groups include also diseases enriched in Finland.

There are other funding sources for research conducted on rare diseases in ERN centres and universities. In the total funding, the importance of short-term funding from research foundations has increased while – due to cuts in government research funding – the number of research personnel has reduced and fewer research equipment updated. Research focusing on the genetics and epigenetics of rare diseases requires that genomic variation be reliably and precisely determined, especially in populations with a narrow genetic base.

A significant part of scientific research in rare diseases also focuses on patients identified during clinical practice in university hospitals and on the ensuing translational studies collaboratively carried out by the clinician and basic researchers. In such studies, continuous governmental research funding granted to hospital districts plays a vital role.

In 2013, the Academy of Finland joined the International Rare Diseases Research Consortium IRDiRC (www.irdirc.org). The consortium brings together actors whose work involves rare diseases research. Furthermore, the Academy has participated in the European E-RARE-3 research funding cooperation between 2017 and 2018. In 2019, a five-year funding project by the European Commission, the European Joint Programme on Rare Diseases (EJP RD, www.ejprarediseases.org) will be launched. The objective of this project is to improve the quality of life for individuals with rare diseases by increasing knowledge about the diagnostics, treatment and care of rare diseases. The project brings together research sponsors, research institutes, universities, university hospitals, ERN Centres of Expertise and patient organisations. The Academy of Finland has participated in this from its beginning. Rare diseases have been the object of international clinical pharmaceutical research projects, while Finland's participation in them, and in pharmaceutical research in general has declined.

2.4 Disseminating information and strengthening knowledge

The main yield of the joint Virtual Hospital 2.0 project (2016-2018) of the five university hospitals, funded by the Ministry or Social Affairs and Health, is Health Village, a web service of special health care (www.terveyskyla.fi). It provides information and support for different diseases, symptoms and life situations in Finnish and Swedish (hälsobyn.fi). Health Village contains a hub providing information on rare diseases (harvinaissairaudet.fi, sällsyntasjukdomar.fi), with sections open to all and exclusively to professionals. On its open web pages, information on rare diseases in 10 different disease groups has thus far been made available. Also, information on health and social services has been compiled on the website. Further, information on six rare disease groups can be found in the other hubs of Health Village. Additional information in Finnish related to rare diseases is available on the Finnish websites of Orphanet, in Terveysportti and Terveyskirjasto maintained by Duodecim, and on the homepages of the Finnish Network for Rare Diseases, containing information produced by patient organisations and associations.

The Rare Diseases Units have organised training for healthcare professionals in their own specific catchment areas. Rare diseases have been introduced in medical journals, at physician's seminars in different regions and at conferences and events organised by different stakeholders. The Nordic Conference on Rare Diseases was held in Finland 2014.

Finnish actors from the field of rare diseases have participated in the Nordic Conference on Rare Diseases in Copenhagen in 2016 and in the

conferences of Eurordis (www.eurordis.org), a European umbrella organisation of rare disease organisations. Eurordis conferences are held every two years; the most recent one was held in May 2018 in Vienna. Finland also participates in the Nordic Network on Rare Diseases aimed at promoting Nordic cooperation in the field of rare diseases. The network is funded by the Nordic Council of Ministers. The network of Nordic patient organisations (Sällsynta Brukerorganisationers Nordiska Nätverk, SBONN) promotes cooperation between groups representing people with rare diseases.

2.5 National registry for rare diseases and streamlining of care pathway

Establishing a National Rare Diseases Registry has been promoted through cooperation between the National Institute for Health and Welfare (THL) and the Rare Diseases Units in university hospitals. The Orpha nomenclature, based on the diagnosis database of the European Orphanet website (www.orpha.net) collecting data on rare diseases will be integrated with the Finnish code server parallel to the ICD-10 disease classification. The codes will make the identification of rare diseases easier. There are also plans to make Orpha codes a part of the Care Register for Health Care (Hilmo). A project to design alerts for those rare diseases in which special vigilance or expertise is required from healthcare professionals is ongoing; such alerts should be implemented into all electronic patient record systems in use.

To enhance the diagnostics, care and rehabilitation of rare diseases and to promote service provision, information about these has been made available in the professional section of the Health Village rare diseases hub. However, adequate information on available rare disease rehabilitation and social welfare services covering all levels of the service system is still lacking. The ERNs have started to produce information and guidance on the treatment of rare diseases. These can also be used nationally.

2.6 Social support, rehabilitation and augmentation of involvement

After the publication of the first National Programme, special health care has seen progress that significantly has enhanced the care of rare diseases. New practices and

operating procedures have been created to facilitate and promote the diagnostics and adequate treatment of rare diseases. Still, many individuals with rare diseases remain without appropriate needs assessment in which for example the functional challenges in their daily life and the factors affecting their income would be taken into account. Not everyone with rare diseases may enjoy the benefits of multiprofessional work. Although representatives with rare diseases have been chosen to steering groups of Rare Diseases Units, first programme's proposals regarding strong involvement of patient groups in the planning and evaluation of centres of expertise and in the establishment of customer panels have not been implemented sufficiently. Increasing and more systematic efforts are required to involve representation of people with rare diseases, their families and patient organisations in the planning and evaluation of care and services.

2.7 National coordinating centre for rare diseases

The national coordinating centre proposed in the first National Programme was not implemented. In the course of the programme period, the importance of coordination in promoting the objectives of the National Programme and of implementing equitable treatment for people with rare diseases has only become more prominent. The responsibilities of regional coordination of rare disease activities belongs to university hospitals, but the need for national coordination extends widely over various stakeholders. Until now and partly randomly, coordination has relied on the Ministry of Social Affairs and Health, on unofficial cooperation between Rare Diseases Units, and

active operators in the third sector. In coming years, there is a need for clarification of the roles and coordination tasks of different authorities. The present Programme introduces the active operators participating in such coordination, the coordination tasks, and includes proposals for further development of coordination. The Ministry of Social Affairs and Health and the Working Group on Rare Diseases will continue the work to define necessary coordination structures during 2019.

3 Pivotal objectives of the National Programme 2019–2023

The pivotal objectives of the National Plan 2019-2023 are to increase expertise and its communication, to build up involvement of patients with rare diseases in decision-making concerning them, and to establish national coordination firmly. Resources are needed also for scientific research on rare diseases with special emphasis on the Finnish disease heritage.

3.1 Increasing knowledge on rare diseases and strengthening expertise

Knowledge and expertise in rare diseases is unevenly distributed and occasionally difficult to find. A person with rare disease may know more about her or his disease than a health professional. The importance of experiential knowledge on rare diseases is highlighted by the lack of medical knowledge. Due to lack of knowledge, unusual combinations of symptoms and findings may not be identified as a rare disease. Diagnostic delays may impair the implementation of effective treatments and rehabilitation. Furthermore, insufficient expertise of health professionals to advise and direct a person with rare diseases to employ necessary aids and services may increase the costs of social welfare and healthcare. Since information may be hard to come by, centralisation of available information and expertise would improve accessibility.

Strengthening rare disease expertise and increasing accessibility of information provide ways of improving the diagnostics and care of rare disease patients. As part of cross-border healthcare, increasing the availability of information and expertise in rare diseases has been entrusted to European Reference Networks (ERNs). In addition to the ERN healthcare providers, further units and centres of expertise in rare diseases also operate in hospital districts. To find best available expertise, nationwide cooperation and sharing of information will be pivotal. Critically, scientific research increases expertise and makes the development of new treatments possible.

3.1.1 Increasing knowledge and awareness

Patient organisations and foundations have traditionally produced information targeted at individuals living with rare diseases, and their families. Experiential knowledge is collected

both nationally and internationally. Eurordis maintains the Rare Barometer Voices website (www.eurordis.org/voices), By registering on it, one can respond to surveys on healthcare and social welfare experiences and on everyday challenges met by people with rare diseases. The Finnish Network for Rare Diseases and rare disease patient organisations in various ways collect experiential knowledge gained to help identify problem areas in the service system and to develop solutions.

Table 1. Information on rare diseases on websites (in Finnish and Swedish):

Health Village, rare diseases	www.harvinaissairaudet.fi sällsyntasjukdomar.fi, a national website providing information on rare diseases, targeted at the general public and professionals
Duodecim, Terveysportti and Terveyskirjasto	www.terveysportti.fiwww.terveyskirjasto.fi, the website of the Finnish Medical Society Duodecim, targeted at the general public and professionals
Orphanet	www.orpha.net/national/FI-FI/index/kotisivu/, an open website with comprehensive information on rare diseases and orphan drugs
Finnish Network for Rare Diseases	www.harvinaiset.fi, a website compiling and communicating information on rare diseases by organisations and associations
National Institute for Health and Welfare (THL)	www.thl.fi/fi/web/vammaispalvelujen-kasikirja, Handbook on Disability Services, an open website containing information related to the life of people with disabilities and available services

The rare diseases hub in Health Village provides information to individuals with rare diseases, their families and professionals.

Ultimately, the responsibility for generation of information on rare diseases lies with the public sector. The development of the rare diseases hub in Health Village to meet the needs of individuals with rare diseases, their families and professionals will be vital. During development, other reliable sources of information on rare diseases should be taken into account by linking, whenever possible.

In addition to open data, Health Village has a dedicated section for healthcare and social welfare professionals. This section contains information on disease diagnostics and treatment, and on care and service pathways. To further support consultation and referral, it has a search function to find rare disease experts. The Rare Diseases Units participate in the production of information to it.

It will be possible to develop the rare diseases hub in Health Village into a versatile information portal for rare diseases in cooperation with different operators. This will make it easier to find and take advantage of information in Finnish.

Care instructions and descriptions of service pathways

As there are very few evidence-based clinical practice guidelines for rare diseases, guidelines for diagnostics, care and service pathways should be formed through cooperation and consideration the experience gained by different operators. The European Reference Networks (ERNs) produce information on the diagnostics and treatment of diseases in their own disease groups and promote rare disease-focused scientific research. The Finnish centres of expertise participate in this. Such information should be utilised when creating national practices. Medical information is supplemented with information about social welfare services on disease- or disease group-specific basis. General knowledge about social welfare services does not always suffice when services for an individual with rare disease are designed. The Health Village hubs for rare diseases and rehabilitation (www.kuntoutumistalo.fi) contain information on social welfare services, among other things.

The care and service pathways of people with rare diseases vary according to disease characteristics and the person's life situation. To better enable people with rare diseases to timely receive the needed treatments, individualized care and service pathways should be designed. Such a pathway describes the key procedures, issues and operators for which individuals' needs should be considered and availability ensured while organising their care and services. For the largest and especially challenging groups of rare diseases, on top of general descriptions diagnostic and care instructions which include general quality requirements and list special challenges at different stages of the care and service pathway will be needed. Such pathway descriptions should be delineated in collaboration with patient organisations and networks. The challenges in everyday life vary between different rare diseases. One therefore will need to describe in disease-specific ways how coping with everyday life could be supported best. The nature of social support needed will likely vary in different rare diseases.

The Orphanet website provides information on the most common rare diseases in Finnish

The open access Orphanet website (www.orpha.net) is a joint project of 40 countries. The website is funded by the EU and it is open to everyone. Presently, Orphanet contains information on approximately 6,100 rare diseases, related research projects and services (patient organisations, centres of expertise, laboratories) and orphan drugs. In addition, Orphanet maintains Orpha coding, the most comprehensive classification of rare diseases available. Norio Centre of Rare Diseases is currently responsible for adding Finnish information about rare diseases to Orphanet service.

The introductory pages of Orphanet database and approximately 200 article summaries on the most common rare diseases have been translated into Finnish. Currently available resources are insufficient to translate and maintain the entire database. In future, Orphanet will seek cooperation with ERN networks and other information producers on rare diseases in its production of information. To ensure the continuity of the Orpha nomenclature, EU is looking for ways to commit the Member States to providing stronger support for Orphanet. Orphanet funding needs to be regularised in Finland as well. To avoid overlapping work, it is important to coordinate between the information contained in Orphanet and the contents of Health Village.

OBJECTIVES AND PROPOSED ACTIONS IN RARE DISEASES

- increasing cooperation between website administrators producing information (e.g. Duodecim, Health Village, National Institute for Health and Welfare (THL), Finnish Network for Rare Diseases (FNRD), Orphanet) to improve the availability and coverage of information in Finnish and Swedish, and to avoid overlap
- developing the content of the open hub of rare diseases in Health Village in ooperation with representatives of Rare Diseases Units, stakeholder organisations and associations
- increasing experiential knowledge reported by people with rare diseases (PROM, PROMIS) through cooperation between THL, development personnel of healthcare units and patient organisations.
- by regular surveys conducted by patient organisations assess and monitor the everyday challenges of individuals with rare diseases and flexibility of their care pathways
- improving and expanding related information on available social welfare, aids, services and guidance, and advice about its availability, in collaboration with THL, the Social Insurance Institution of Finland (Kela), Health Village and FNRD
- disseminating information on and increasing awareness of rare diseases, and organising training in cooperation with different operators in the field
- sharing through Health Village the patient information and instructions produced by the European Reference Networks (ERN) in Finnish and Swedish
- ensuring continued Orphanet activities in Finland

The website of the Finnish Network for Rare Diseases contains information produced by patient organisations

People with rare diseases need information in their own language. For several decades, organisations in the field of healthcare and social welfare have produced rare disease-related information. Materials produced by the member communities of Finnish Network for Rare Diseases have been linked to the network's website (www.harvinaiset.fi). Currently, the website carries or links to information in Finnish on about 300 rare diseases, together with further rare disease-related information, This includes contact details of relevant associations or interest groups and advice on available services and peer support. Adding such information systematically to other databases, especially to the rare diseases hub in Health Village, would be recommendable.

3.1.2 Strengthening the status of Rare Diseases Units and allocation of resources to them

In accordance with the Government Decree on the Division of Work and Centralisation of Certain Tasks (582/2017, section 4), rare diseases units were established in each of the five university hospitals, between 2015 and 2017. A physician in charge and a healthcare professional with background in nursing work in each unit.

Table 2. Units for rare diseases and their personnel resources 2019

Unit	Established	Physician in charge	Other personnel
HUS (HAKE)	1 January 2015	physician 100%	nurse 100%
TYKS	1 January 2016	physician 100%	nurse 20%
KYS (HARSY)	1 February 2016	physicians jointly 50%	nurse 100%
OYS (Harvi)	2 June 2016	physician 30%	nurse 100%
TAYS (HarSY)	1 January 2017	physician 60%	nurse 100%

The Rare Diseases Units (Centres) have developed at different paces, according to their resources. To promote equity at the national level, it will be necessary that the basic tasks of the units be determined in a uniform manner, to further delineate the obligations laid down in the Centralisation Decree. However, due to regional and hospital-specific differences, unit activities include different tasks and focus areas.

THE BASIC TASKS OF RARE DISEASES UNITS (CENTRES) IN THEIR SPECIFIC CATCHMENT AREAS:

- facilitating the development of clear care and service pathways
- participation in the coordination of the diagnostics and care of people with rare diseases, and of associated referral practices
- providing a consultation service to healthcare and social welfare professionals
- general advisory services and guidance for people with rare diseases to suitable care or service pathways
- increasing information and awareness of rare diseases
- promoting research in rare diseases
- cooperation with different healthcare units, authorities and organisations

The standardization of basic tasks may require stronger resources and harmonised activities. Description of uniform contact and referral practices will be needed. Information about various tasks of Rare Diseases Units should be made available to individuals with rare diseases, their families and healthcare and social welfare professionals. The units have advocated increased awareness and information about rare diseases in their specific catchment areas by for example organising training to professionals. In their areas, the Rare Diseases Units play important roles in promoting understanding on the need for healthcare and social welfare services by people with rare diseases

OBJECTIVES AND PROPOSED ACTIONS FOR RARE DISEASES UNITS IN THEIR CATCHMENT AREAS

- in all university hospitals, ensuring the allocation of resources to Rare Diseases Units and consolidating their tasks and positions
- building up the knowledge base: a physician in charge and a healthcare professional with background in nursing will be needed and need to be familiar with rare diseases; it should be possible to consult a social worker
- supporting the centres of expertise and units possessing expertise in rare diseases; sharing information on available rare disease specialists
- supporting ERN members, facilitating ERN activities together with strengthening national and international cooperation and exchange of information
- involving the representatives of people with rare diseases in the activities of the unit and the university hospital to promote customer-oriented approaches
- communicating information on rare diseases and on unit's activities in cooperation with patient organisations and networks

Pivotally, the regional roles of Rare Diseases Units should be strengthened according to the Centralisation decree. Mutually cooperatively, national practices such as electronic consultations and descriptions of service pathways should be developed between the units. Rare Diseases Units will disseminate information about the centres of expertise and ERN centres (healthcare providers) in their area and will need to agree on the division of responsibilities and tasks, regionally and nationally.

3.1.3 European Reference Networks (ERN)

In 2014, the European Commission set up criteria according to which a healthcare unit can apply to the European Reference Networks (European Reference Networks for Rare and Low Prevalence Complex Diseases, ERN; https://ec.europa.eu/health/ern_en). The catalyst for establishing ERNs was the EU Directive (2011/24) on the application of patients' rights in

cross-border healthcare. Aims of ERNs were to empower choices to care pathways, organise joint training and consultations, and to enable cross-border discussions on the situation of a person with rare diseases without need to travel to another country,

apart from few exceptional cases. Expertise in each network relates to its specific group of rare diseases. Eurordis, an umbrella organisation of European rare disease patient organisations, participated in establishing criteria for ERN healthcare providers (www.eurordis.org). Strengthening of patient involvement in the operations of ERNs and its healthcare providers was among the objectives.

In 2017, 24 networks started their operations in Europe. More than 900 different healthcare providers from 25 Member States and Norway participate. In 2018, 14 units from four Finnish university hospitals are members of 12 different European Reference Networks (ERN). Active participation in the ERNs by Finnish university hospitals will promote expertise in rare diseases and development of patient care. Aim will be to have at least one university hospital representative in each reference network.

OBJECTIVES AND PROPOSED ACTIONS:

- nationwide, to define the duties and responsibilities of ERN centres, to create cooperation practices between an ERN and other operators in its field of expertise, to integrate ERNs into the national service system
- Finland's participation in all 24 ERNs, either as a healthcare provider or an affiliated partner
- with Rare Disease Units, to communicate up-to-date information about ERN centre operations to other actors in healthcare and social welfare
- in collaboration with Rare Disease Units, to strengthen the role of patient organisations and representatives in the ERN application process and operations
- agreeing on the assessment and monitoring of the national integration and other activities of the ERN centres at the national level
- to develop Nordic cooperation of ERN healthcare providers and to participate in the development of European registries in cooperation with the National Institute for Health and Welfare (THL) and Rare Diseases Units
- to ensure necessary resources and support functions for ERN healthcare providers to participate in ERN activities

Based on the Treaty on the Functioning of the European Union, the content of healthcare services and the reimbursement of costs caused by them are subordinate to decision-making of the Member States. This also applies to medicinal products granted an EU-wide marketing authorisation in a centralised procedure. The Member States are not directly and legally bound by results of the work carried out by the European Reference Networks.

Table 3. ERN member centres in Finland and elsewhere in Europe.
Reference: Kääriäinen Helena, Lääkärilehti 24-31/2018 Vol. 73, p. 1580.

ERN and its subject area	Finnish centre of expertise	Centres /countries **total**
ERN BOND: rare bone disorders	-	39/9
ERN CRANIO: craniofacial anomalies; rare ear, nose and throat disorders	HUS	29/11
Endo-ERN: rare endocrine conditions	-	71/19
ERN EpiCARE: rare epilepsies	KYS	27/13
ERKNet: rare kidney diseases	HUS	37/12
ERN-RND: rare neurological diseases	–	32/13
ERNICA: rare inherited and congenital anomalies	HUS	20/10
ERN LUNG: rare lung diseases	–	60/12
ERN Skin: rare skin diseases	HUS	56/18
ERN EURACAN: adult rare tumour cancers	TYKS	65/15
ERN EuroBloodNet: rare haematological diseases	–	66/15
ERN eUROGEN: rare urogenital diseases	–	29/11
ERN EURO-NMD: rare neuromuscular diseases	TAYS	45/14
ERN EYE: rare eye diseases	–	29/13
ERN GENTURIS: rare genetic tumour risk syndromes	TYKS	23/12
ERN GUARD-HEART: rare diseases of the heart	HUS	24/12
ERN ITHACA: congenital malformations and rare intellectual disability	HUS	36/13
MetabERN: hereditary metabolic disorders	–	68/17
ERN PaedCan: paediatric cancer	TYKS, TAYS, KYS	57/18
ERN RARE-LIVER: rare hepatological diseases	–	28/11
ERN ReCONNET: rare connective tissue and musculoskeletal diseases	–	25/8
ERN RITA: rare immunodeficiency, autoinflammatory and autoimmune diseases	–	24/10
ERN TRANSPLANT-CHILD: transplantation in children	–	17/10
VASCERN: rare vascular diseases	HUS	32/11

3.1.4 Promoting research

Research enables the development of more specific diagnostics and new treatments in rare diseases. Essential knowledge on the prognoses of rare diseases, on their treatment options, associated diseases and effectiveness and necessity of screening for these is lacking. Even less information is available on issues related to rare diseases like the extent of decline in the quality of life, on the efficiency of non-pharmaceutical therapies, on the burden to healthcare and social welfare or the total costs of rare diseases to people suffering from them and to their families. Some of these rare diseases are a part of the Finnish Disease Heritage, while novel and rare inherited diseases, which are less prominently concentrated in our population are being discovered constantly. There is dire need for comprehensive research data on the total burden to society and to individuals caused by these diseases.

While Finnish researchers participate in many international research projects, the greatest challenge to Finnish rare diseases research has become that researchers move abroad towards better funding opportunities, mainly to other Nordic, Central European or Northern American countries. The establishment of the European Reference Networks will likely increase international research cooperation. However, any Europe-wide research funding potentially received will often be divided between multiple centres in different countries. It will be especially challenging to obtain European research funding for diseases specifically enriched in Finns. Funding of rare diseases research should therefore remain a focus area in national funding.

Rare diseases research is important not only to augment knowledge on rare diseases but also to understand the development of common non-communicable diseases. By providing unique information on the normal functioning of the human body at molecular level, rare diseases research enables us to model disturbances in the function of the organs and cells and to pinpoint pathogenetic mechanisms in more common diseases.

OBJECTIVES AND PROPOSED ACTIONS IN RARE DISEASE RESEARCH

- to keep rare diseases research as a focus area in national research funding
- to emphasise the importance of research on diseases concentrated in Finland, on their treatment and total burden to society and an individual
- to keep top-notch research in Finland, increasing governmental research funding for high-quality studies on rare diseases
- targeted funding for the study of research methods used in rare diseases research

3.2 Strengthening of patient involvement in rare diseases

The UN Convention on the Rights of Persons with Disabilities from 2006 obligates Finland to support and promote the involvement of people with disabilities or chronic illnesses in matters concerning them. In addition, the Convention obligates to empower patient organisation involvement at various levels and the involvement of people with disabilities or chronic illnesses in the service system. As recommended by the European Commission (2009), people with rare diseases, their representatives and patient organisations should be consulted and involved in all societal decision-making concerning rare diseases. The Council demands the support of work carried out by patient organisations to improve the status of people with rare diseases. In the selection criteria, ERN healthcare providers have been obligated to cooperate with patient organisations and thus heed the viewpoint of people with rare diseases in the planned activities.

The RD-Action project (2014-2018) related to the EU Health Programme developed a uniform European approach to challenges associated with rare diseases. The Patient Empowerment Report (2018) recommends measures for empowering and engaging people with rare diseases at the national, regional and local levels. In the report, empowerment is a prerequisite to enabling persons with rare diseases to promote matters concerning them. Empowerment is associated with promoting knowledge, education and necessary support. The empowerment and engagement of persons with rare diseases may take place at three different levels:

1. societal decision-making, prioritisation and development in health and social policies
2. planning, implementation, evaluation and development of health and social services
3. personal care and service pathways, increased quality of life and holistic wellbeing of persons with rare diseases

Progress in the involvement of persons with rare diseases should be monitored regularly. For example, a rare diseases barometer could be designed in cooperation with parties working with rare diseases. In such a barometer, especially the objectives set for patient involvement and engagement in this programme could be assessed.

3.2.1 Empowerment and involvement of people with rare diseases in society and their communities

It is important that as soon as the diagnosis has been made, people with rare diseases receive information about their patient organisation, its services and various forms of support offered. People with rare diseases without an organisation can be advised to contact the Finnish Network for Rare Diseases.

There are dozens of associations and organisation representing people with rare diseases in Finland. The Finnish Network for Rare Diseases, consisting of 20 associations, is a national cooperation network of various health and social welfare organisations in the field of rare diseases. HARSO is an umbrella organisation of associations supporting patients with rare diseases and disabilities. Consortiums of organisations and associations lobby matters concerning people with rare diseases at the national level. Information on organisations, associations and interest groups representing people with rare diseases is available on the Finnish Network for Rare Diseases website (www.harvinaiset.fi). Each of these represents its target group and lobbies issues important to them and to everyone with rare diseases. Several receive support from the Funding Centre for Social Welfare and Health Organisations (STEA). It channels net revenue from the Finnish gaming company Veikkaus. Enabling support to small associations and to rare disease patients without any remains challenging.

Individuals with rare diseases are citizens, residents of a municipality, customers and patients. They and their families need information about their rights, to equitably reach appropriate services and to safeguard their lives. Dissemination of information reduces prejudices and increases understanding about the full diversity of rare diseases. To boost involvement of people with rare diseases, cooperation between Rare Diseases Units, ERN centres, patient organisations and other operators in the healthcare and social welfare is

required. To disseminate and obtain information, participation of social welfare and healthcare professionals in events organised by patient organisations is vital. Reciprocally, people with rare diseases and their families bring their experiential knowledge to dialogue.

Associations and organisations train experts by experience. Their experiential knowledge is used in targeted training of professionals, in planning of social welfare and healthcare services and assessment of activities. Kokemustoiminta Network consisting of 36 organisations (www.kokemuskoulutus.fi), cooperates in the briefing and training of experts by experience and in communicating experiential knowledge, Associations and organisations also train people with rare diseases and their families to be involved, fight for their rights and to speak out in contemporary issues. For example, a panel of experts by experience in rare diseases (Working Group Harkko) and their family members operates in the Finnish Network for Rare Diseases.

WORKING GROUP HARKKO

- each of the 20 member communities of the Finnish Network for Rare Diseases (FNRD) names 1-2 candidates
- meets 2-3 times per year
- the group's proposals factor in in the development of FNRD activities and national lobbying
- the experts by experience participate in Rare Disease Unit's steering groups and customer panels, and in Harkko meetings
- matters discussed in the meetings represent general contemporary issues related to the status of people with rare diseases and the state of their services.

The Finnish Network for Rare Diseases conducts yearly surveys to target groups of its member communities. The network uses the surveys and the results of Harkko's work when issuing opinions and meeting healthcare and social welfare professionals. Surveys and other collection of information should be further strengthened through cooperation between healthcare professionals and patient organisations. To support

lobbying, also HARSO collects information from its member associations. Individual organisations utilize expert panels (or like) that consist of individuals with rare diseases and their family members, enabling discussions on contemporary concerning issues.

OBJECTIVES AND PROPOSED ACTIONS:

- to encourage and direct individuals with rare diseases to contact relevant organisations and to seek peer support and other available services
- to disseminate information on patient associations and organisations to healthcare and social welfare professionals and population in general
- to promote regional and local engagement and involvement of people with rare diseases through the help of patient organisations and regional operators
- to plan and implement annual Rare Diseases Days in specific catchment areas, organised jointly by the Rare Diseases Units and patient organisations
- to share experiential knowledge during national and regional training days and fairs organised for social welfare and healthcare professionals, with the National Institute for Health and Welfare, the Rare Diseases Units and patient organisations
- to highlight the challenges faced by people with rare diseases in Vammaisfoorumi and the Advisory Board for the Rights of Persons with Disabilities (VANE)
- to safeguard the operation of associated patient organisations

3.2.2 Involvement of people with rare diseases in developing healthcare and social welfare services in their own areas

Enabling people with rare diseases to influence the content of healthcare and social welfare services provided by their own hospital or municipality plays a central role in involvement. The care and service pathways and service contents should be created in cooperation with people with rare diseases. People with rare diseases should be able to moot their own needs and experiences into the development and assessment of services routinely. Consequently, our service system should adapt to changing demands according to given feedback and customer satisfaction.

Some Rare Diseases Units have selected representatives of people with rare diseases into their steering groups. Such involvement of people with rare diseases and patient organisations should in all units be harmonised and strengthened. Healthcare professionals are responsible for bringing up issues in which rare disease representatives may utilize their

experiential knowledge to improve activities. To enable a genuine and mutual dialogue between professionals and rare disease representatives, issues discussed must be intelligible.

Activities in Rare Diseases Units utilise the knowledge of experts by experience. In addition, units have organised events and other forms of cooperation with rare disease representatives and organisations. Open access events should be organised regularly in varying locations and planned in cooperation with patient organisations. People with and patient organisations for rare diseases find the activities of Rare Disease Units important; there is need for stronger reciprocal engagement. The voice of rare disease patients should be amplified by customer panels in the units and, for example, by developing feedback systems that assess unit activities. Shared views on which unit activities should be elaborated together with rare disease representatives are needed. Shared matters could include ensuring operational efficiency of care and service paths or issues related to encounters between healthcare or social welfare professionals and people with rare diseases:

RARE DISEASE CUSTOMER PANELS

- to establish customer panels to ERN centres and/or university hospitals
- to invite representatives of patient organisations, experts by experience, their family members and other actors in rare diseases to customer panels
- to strengthen the role of customer panels in communicating needs and wishes of people with rare diseases and in planning and assessment of unit activities
- to utilise in customer panels the experience and expertise of organisations and networks like the Finnish Network for Rare Diseases and HARSO

The roles of people with and patient organisations for rare diseases need to be strengthened in activities of national ERN centres (healthcare providers). In the report on the integration of the European Reference Networks into the national healthcare systems (2018), Eurordis proposed that customer panels consisting of representatives of the patient organisations in the centre's target group would be established in connection with the centres. A national customer panel would correspond to the European Patient Advocacy Groups (ePAG) established in the European Reference Networks, playing important roles in the networks activities. Between national ERN centres, a systematic cooperation model is called for, with strong representation of people with and patient organisations for rare diseases.

OBJECTIVES AND PROPOSED ACTIONS:

- to strengthen the involvement of people with rare diseases by establishing customer panels (CP) in university hospitals. CPs will convene regularly to plan and to assess contemporary issues related to healthcare and social welfare services in rare diseases, together with Rare Diseases Units, centres of expertise and other actorsto reach shared apprehension of matters to be discussed with experts by experience
- to plan and develop the care and service pathways of people with rare diseases jointly with healthcare and social welfare professionals, and patient organisations.
- to boost cooperation and communication by Rare Diseases Units to healthcare, social welfare and other professionals working with rare diseases, and to general public
- to establish regular dialogue by Rare Disease Units and patient organisations to the Social Insurance Institution of Finland (Kela), notably about issues concerning rehabilitation, disability benefits and competitive tendering of services
- to organise in all university hospitals regular annual events directed at patient organisations and people with rare diseases
- to establish and develop patient feedback systems of the Rare Disease Units and ERN centres (healthcare providers): development of electronic feedback system as part of services
- to define the roles of patient organisations and representatives in the operation of the ERN healthcare providers already during the application process

3.2.3 Safeguarding the involvement of people with rare diseases in their care and services

The involvement of people with rare diseases in the planning and implementation of their care and services should be strengthened and their right to self-determination taken into account. Many people with rare diseases feel vulnerable, particularly in situations when the professional taking care of them does not possess sufficient knowledge about the disease or about patients' right to choose their place of care, or if the professional has negative or belittling attitudes towards them or their disease. The prerequisite for stronger involvement of persons with rare diseases in implementation of his or her care and services

is that sufficient information on the health, social and rehabilitation services available. At best, patient involvement will be achieved through an equal dialogue and cooperation between the professionals and the individuals with rare diseases. When supporting daily lives of persons with rare diseases, attention must also be paid to their social milieu including family members, close friends and work communities.

Orderliness of rare disease care and services support individuals' daily life. Under the Social Welfare Act (1301/2014, section 39), a client plan for assessing the need for services must be drawn up for the client. Furthermore, a plan for examinations, care, medical rehabilitation and other corresponding activities is mentioned in the Act on the Status and Rights of Patients (785/1992, section 4a). Such a plan must be drawn up during the implementation of healthcare and medical care, and must indicate the treatment and its implementation schedule. The importance of patient involvement and the right to self-determination are emphasised by the fact that the plan must be designed in mutual understanding with the patient, family member, close friend or legal representative.

OBJECTIVES AND PROPOSED ACTIONS:

- to increase guidance, advice and information about the services available provided by professionals to persons with rare diseases and about the status of patients and clients in healthcare and social welfare
- to develop and strengthen group peer support through cooperation between patient organisations and healthcare
- to increase and disseminate information about advisory and peer support services provided by patient organisations and associations

3.3 Coordination of activities related to rare diseases

Awareness of rare diseases has increased since the publication of the first National Programme. The general public as well as professionals have become increasingly interested in rare diseases and the status of people suffering from these. The public, third and company sectors and the media have all shown interest in people living with rare diseases. However, the multi-layered information, expertise and activities related to rare diseases are challenging to master.

3.3.1 National coordination

The establishment of a national coordinating centre was proposed in the first National Programme none has been implemented. Some coordination has been performed by the Working Group on Rare Diseases and its Subcommittee. One of the tasks of the current Working Group is to propose a model for national coordination.

During the update of the National Programme, our understanding of the importance of national coordination in the development of care and comprehensive support of people with rare diseases has been reinforced. In the surveys conducted, rare disease associations, the Rare Diseases Units and ERN centres all supported the establishment of a coordination centre almost unanimously. The importance of national coordination was stressed in many of the speeches held in the National Conference on Rare Diseases in autumn 2018. A variety of tasks have been proposed for the coordinating centre, including gathering information on rare diseases and promoting the networking of operators in the field of rare diseases. Tasks could include systems research related to rare diseases and surveys of needed services, for example by utilising registry data.

Management of the overall status of rare diseases would be improved by a national database tagging rare diseases. Presently, there are occasional registries of individual rare diseases or disease groups. The Rare Diseases Units and the National Institute for Health and Welfare (THL) are jointly developing the possibility to add not only the ICD-10 but also the exact Orpha code to the diagnosis sections of electronic health records. A national rare diseases registry would enable to combine the data to EU and ERN registries. Without international registries, the diagnostics, care and monitoring of ultra-rare diseases cannot be developed adequately. The legislative amendment concerning national THL quality registers would permit the creation of such national registry. Table 4 describes various proposals for rare diseases, inclusive of bolstered coordination tasks of THL.

Table 4. Current or proposed role of different actors in the development and coordination related to rare diseases

Ministry of Social Affairs and Health	steering of legislation, resources and information, general supervision, assessment, EU cooperation, cooperation in Nordic networks
STEA	funding of patient organisation activities in the field of rare diseases, assessment of effectiveness of such measures
Healthcare and social welfare	production of services, improving the availability of expertise, agreeing on service pathways and referral practices
University Hospital Districts	regional cooperation and coordination tasks in accordance with the Centralisation Decree (582/2017), information production to Health Village, international research activities, coordination of centres of expertise and ERN health care providers, care instructions
National Institute for Health and Welfare (THL)	information guidance, maintenance of networking, project steering, comprehensive research, assessment and maintenance of services, functioning measures, Orpha codes, knowledge-bases and registries
COHERE Finland	knowledge steering, assessment of inclusion of services and treatments in rare diseases in the service palette of national healthcare
Social Insurance Institution of Finland	reimbursement of medicines, costs of cross-border care, other benefits and services, rehabilitation
Fimea, Pharmaceuticals Pricing Board (Hila)	marketing authorisation of medicinal products, assessment of benefits to treatment from orphan drugs, conditions for reimbursement of medicines (Hila)
Finnish Network for Rare Diseases	to influence matters promotion of joint lobbying of organisations, communication, compiling and disseminating information
HARSO	promotion of organisations' joint activities to influence matters
Norio Centre of Rare Diseases	maintenance of the Finnish Orphanet website, international cooperation

Furthermore, once general agreements on national rehabilitation policies have been reached, the development of rehabilitation for rare diseases should be implemented through cooperation between university hospitals, Kela and others. Presently, multiple operators participate in the coordination of guidance, supervision, regulation and responsibilities in rare diseases. Nevertheless, coordination of these activities to avoid overlaps is lacking. Neither can operators monitor equitable implementation of medical and social welfare services nationwide, and the interconnectedness of current operators needs improvements. Also, principles guiding the division of labour between the Rare Diseases Units and ERN healthcare providers require national cooperation.

After systematic assessment of tasks of various operators and the potential to develop national coordination in rare diseases, the Working Group proposes that national coordination of rare diseases should be assigned to the National Institute for Health and Welfare (THL). By its assessment THL would be best positioned to coordinate cooperation between experts, service providers, patient representatives and the third sector operating in the field. It would also be able to coordinate the communication of reliable information. THL is also responsible for the maintenance of necessary coding systems and definitions, has the capacity to promote registries, collection and analysis of information, and research and

development at the national and international levels. Besides ensuring the required statutory basis, the Ministry of Social Affairs and Health should continue to bear the main responsibility for international cooperation, especially at the level of EU.

OBJECTIVES AND PROPOSED ACTIONS:

- clarification of national coordination and division of labour between the Ministry of Social Affairs and Health, THL and service providers
- university hospitals will strengthen regional coordination in the field of rare diseases according to the obligations of the Health Care Act and the Centralisation Decree: more detailed description of mutual cooperation and division of labour, strengthening of expertise
- integration of ERNs into our service system, cooperation with other centres of expertise
- in addition to disability services, extending the general coordination tasks of THL to rare diseases
- development of registries and knowledge bases, establishing continuity of Orpha coding and other operations of Orphanet
- to avoid overlap, boost cooperation and networking between the public and third sectors
- boosting cooperation in rehabilitation between the Rare Disease Units and Social Insurance Institution of Finland (Kela) in order to design national plans for various rare disease groups

Diversity and characteristics of rare diseases require special attention with regard to orphan drugs. Special characteristics of pharmacotherapies in rare diseases should be catered to the currently ongoing implementation of rational pharmacotherapy plan and associated legislative amendments (Ruskoaho 2019).

The term of the Working Group on Rare Diseases continues until the end of 2019. The group will monitor and support the implementation of the National Programme. In accordance with its appointed tasks, it will also give a proposal for the implementation of national coordination.

3.3.2 Regional coordination

University hospitals bear the regional responsibility for, plan and coordinate the prevention, care, diagnostics and rehabilitation of rare diseases. The clinical units and centres of expertise responsible for the diagnostics and care provide the services required by people with rare diseases in their field of specialisation. If necessary, the services are provided in

multidisciplinary and multiprofessional cooperation. The tasks of the Rare Diseases Units include participation in the coordination of the diagnostics and the range of services provided to people with rare diseases, advising and referring them to the right experts and to the proper care or service pathway, supporting the clarification of the care and service pathways in their own specific catchment area, and cooperation with different healthcare units, authorities and organisations.

The units' task is to help create smooth consultation and referral opportunities and to participate in the coordination of the care and diagnostics of those patients who have symptoms consistent with rare diseases but for whom no diagnosis has been found in basic or specialised medical care. Consultation models for improving the diagnostics, care and coordination of the care of rare diseases may be developed in the electronic development projects in the field of healthcare and social welfare.

University hospitals can establish centres of expertise for groups of rare diseases or gather diverse professional working groups, for which they find the required expertise and through which they collaborate regionally or nationally with other parties providing care. The tasks of the Rare Diseases Units do not include outpatient clinic activities as the examinations and care choices are the responsibility of various specialist units of the university hospital districts. If necessary, these consult the ERN healthcare providers or refer the patients to them. The care of patients in a stable phase can also be implemented in other healthcare units in the area, as long as sufficient consultation support is available.

OBJECTIVES AND PROPOSED ACTIONS:

- harmonising the regional operation of the units for rare diseases and strengthening the position of the units located in the specific catchment areas

- establishing and strengthening the cooperation of Rare Diseases Units with different healthcare units in their specific catchment areas, both in specialised medical care and basic healthcare, and communicating information about the Rare Diseases Units and centres of expertise to basic healthcare

The Rare Diseases Units can be contacted without a formal referral about questions related to general advice and guidance. The contact details can be found on the websites of the hospitals. Advisory services are provided for example in situations in which an individual with rare diseases has not received the required services or treatment, or has not found information on how to proceed in the personal care and service pathway. Physicians also make referrals to the units.

By clarifying the care and service paths of people with rare diseases, units respond to the need to streamline diagnostic and care pathways, as proposed in the first National Programme. At the same time, they further increase awareness in their own specific catchment areas. Adequate resources are required to strengthen the cooperation of the Rare Diseases Units with basic healthcare, the hospital districts and specific catchment areas, social welfare and organisations,

3.3.3 Coordination of care and services of a person with rare diseases

The importance of the planning of care and services is emphasised in situations when multidisciplinary services are needed but available information is scarce. A comprehensive customer plan makes it possible to provide many people with rare diseases with adequate support for smoothly daily life. The expertise of healthcare social workers and rehabilitation instructors should also be used when drawing up a plan: a comprehensive plan should include both the healthcare and the social welfare services. When drawing up the plan, advantage should be taken of the expertise of the Rare Diseases Units, ERN centres, organisations and associations, as necessary.

According to the Social Welfare Act (1301/2014, section 42), a case manager must be designated to a client if the client's need for services is not short-term or temporary. The objective of the INNOVCare project funded by the EU was to develop structural reforms and comprehensive and customer-centred service pathways improving the social support of people with rare diseases. The project ended in 2018. It emphasised the importance of a case managers supporting people with rare diseases and their families in the coordination of the services and in facing the challenges of everyday life. The case manager helps to find the local experts, works as a contact person for local and national expertise and gives empowering support to persons with rare diseases and the family members. Based on the experience gained in the project, consideration should be given to when it is important to designate a case manager to a person with rare diseases, in which tasks the role of the case manager should be used and how the case manager could best support the person's involvement in his or her care and service pathways.

OBJECTIVES AND PROPOSED ACTIONS:

- planning and piloting a case manager model to ensure seamless care and services for those people with rare diseases requiring large numbers of services
- strengthening the position of healthcare social work in the multidisciplinary cooperation carried out in the care of rare diseases
- piloting the use of personal budgets in those rare diseases that require diverse services

3.4 Estimated cost effects of the National Programme

We propose that the harmonisation of the activities of the Rare Diseases Units be included in the National Programme. Currently, different units have varying personnel resources. Only one of the units has a full-time physician and nurse. If there is a desire to develop the activities, it is important to ensure that the units have adequate resources and expertise. To achieve this, we estimate that approximately 1.6 additional person-years would be needed for physicians and 0.8 person-years for nurses at the national level. The cost effect would be approximately EUR 200,000 per year.

National coordination requires personnel resources in the national coordination unit. According to Subcommittee calculations, at least two person-years are required for the national coordination tasks, including the communication of information, coordination of networks, update and maintenance of the Orpha codes, and Orphanet activities. Resources are also required for development of the database and the maintenance of registry for rare diseases. In total, these amount to EUR 200,000 - 300,000 per year.

The European Commission grants the ERN centres operating in Finland a small amount of financial support for ERN activities. For healthcare providers to fulfil the quality requirements for ERN centres, an additional amount between EUR 20,000- 40,000 per year is needed, for maintenance of required procedures and quality systems. Participation in an ERN uses an input equal to 0.1 person-years for each sub-group in the network. In total, there may be up to ten of them. If centres of expertise joined all the 24 ERNs during the programme period, the number of Finnish centres would total approximately 30. The total amount of work would be between 20 and 25 person-years, divided between several specialists. The imputed costs are about EUR 2 million per year. It has been estimated that an input of less than EUR 1 million is used to maintain the necessary systems in the year a centre is established and about half of that in the following years.

Of note, when estimating the costs of the activities in the field of rare diseases, deficiencies in diagnostics and care may lead to large amounts of unnecessary costs. Also, the costs of an individual treatment may exceed EUR 1 million per year. By investing in expertise, treatment and care decisions based on knowledge gained by studies and flexible services enables allocation of costs in the most appropriate way.

3.5 Practical implementation of the National Programme and monitoring it

The National Plan for Rare Diseases is based on the principle of equitable access to appropriate treatment, care and services by people with rare diseases. The promotion

of involvement improves the status of people with rare diseases and implementation of their rights. The Rare Diseases Units established to university hospitals play a key role in the coordination of the services required by people with rare diseases.

In addition to the promotion of involvement, the National Programme for Rare Diseases 2019-2023 emphasises the importance of expertise and national coordination. The Ministry of Social Affairs and Health and the National Institute for Health and Welfare (THL) will be key actors in national coordination. THL has been developing national coordination of the services for persons with disabilities and the Working Group is of the opinion that many tasks in the national rare disease coordination would be suitable for THL. The role of THL as a national authority maintaining registries responds to the need for coordination in the field of rare diseases. THL has already promoted the introduction of Orpha codes. However, the maintenance of the Orphanet contact point in Finland remains to be solved. For its part, the Ministry of Social Affairs and Health focuses on enabling cross-border cooperation by promoting involvement in ERNs, enhancing the utilisation of their expertise and promoting cooperation between the Nordic countries.

Patient organisations and Rare Diseases Units promote stronger patient involvement. The healthcare and social welfare professionals encountering people with rare diseases are in key positions to transmit information about available opportunities for people with rare diseases to participate in the activities of patient organisations and to access up-to-date and high-quality information. The opportunities to disseminate information have improved in the 2010s. Coordination is needed to increase cooperation and sensible division of labour between various information producers. If overlapping work and production of information could be avoided, the resources could be used efficiently to facilitate the daily life of as many people with rare diseases as possible.

The implementation of the programme requires all parties to commit themselves to achieving the jointly drawn up objectives. The Working Group did not want to make this National Programme into a detailed list of actions. Instead, the group wanted to describe the key principles and objectives guiding decision-making to improve the position of people living with rare diseases. As changes are needed in the healthcare and social welfare services, cooperation is required between regional and national operators to implement the objectives and actions proposed in this programme. In the time when the structure and organisation of healthcare and social welfare services is undergoing a major transformation at the national level, different operators must work together to identify regionally and nationally methods to implement the objectives.

Sources

De Santis, Marta & Hervas, Clara & Weinman, Ariane & Bottarelli, Valentina (2018) Patient Empowerment. Policy Brief. RD-ACTION WP2-TASK 2.5 Output.

European Commission. European Reference Networks (ERN) https://ec.europa.eu/health/ern_en

Council of the European Union (2009) Council Recommendation on an action in the field of rare diseases. 2009/C 151/02

Eurordis (2017) Juggling care and daily life. The balancing act of the rare disease community. A Rare Barometer survey.

Eurordis (2018) Recommendations on the integration of European reference networks (ERNs) into national health systems. November 2018.

National Plan for Rare Diseases 2014 - 2017. Steering group report. Ministry of Social Affairs and Health. Reports and memorandums 2015:43 http://julkaisut.valtioneuvosto.fi/handle/10024/74774

Kääriäinen, Helena (2018) Osaamiskeskusverkot tueksi harvinaissairauksien diagnostiikassa ja hoidossa [Networks of centres of excellence to support diagnostics and treatment of rare diseases]. Lääkärilehti 73, 24–31.

Ruskoaho, Heikki (2018) Selvityshenkilön loppuraportti. Lääkekorvausjärjestelmän kehittäminen [Development of the medicine reimbursement scheme. Examiner's final report]. (Abstract in English). Reports and memorandums of the Ministry of Social Affairs and Health 20/2018.

Saastamoinen, Leena & Saarelainen, Laura & Autti-Rämö, Ilona & Martikainen, Jaana (2015) Lääkkeiden ja ravintovalmisteiden käyttö harvinaisten sairauksien hoidossa. Kysely harvinaissairaita hoitaville lääkäreille [Use of medicines and nutritional supplements in the treatment of rare diseases. Survey of physicians treating people with rare diseases]. Kela Research. Working papers 76/2015.

UN Association of Finland (2015) UN Convention on the Rights of Persons with Disabilities and its Optional Protocol (in Finnish). https://www.ykliitto.fi/sites/ykliitto.fi/files/vammaisten_oikeudet_2016_net.pdf

Government Decree on the Division of Work in Specialised Medical Care and Centralisation of Certain Tasks (582/2017) (in Finnish) https://www.finlex.fi/fi/laki/alkup/2017/20170582

Working Group on Rare Diseases 1 January 2018–31 December 2019

Tuija Ikonen, Ministry of Social Affairs and Health, Chair
Ritva Halila, Ministry of Social Affairs and Health, Deputy chair
Helena Kääriäinen, National Institute for Health and Welfare
deputy member: Jukka Kärkkäinen, National Institute for Health and Welfare
Ilona Autti-Rämö, Social Insurance Institution of Finland, until 30 November 2018
deputy member: Katariina Kallio-Laine, Social Insurance Institution of Finland
Jari Petäjä, Hospital District of Helsinki and Uusimaa
deputy member: Mikko Seppänen, Hospital District of Helsinki and Uusimaa
Pekka Riikonen, Kuopio University Hospital
deputy member: Reetta Kälviäinen, Kuopio University Hospital
Jukka Moilanen, Oulu University Hospital
deputy member: Johanna Uusimaa, Oulu University Hospital
Kari Nieminen, Tampere University Hospital
deputy member: Jaakko Antonen, Tampere University Hospital
Heikki Lukkarinen, Turku University Hospital
deputy member: Samuli Rautava, Turku University Hospital
Elina Rantanen, Norio - Centre of Rare Diseases
deputy member: Leena Toivanen, Rinnekoti Foundation
Katri Asikainen, HARSO
deputy member: Sanna Leppäjoki, HARSO
Risto Heikkinen, Finnish Network for Rare Diseases, Allergia-, iho- ja astmaliitto ry
deputy member: Päivi Vataja, Finnish Network for Rare Diseases, Kuuloliitto
Reima Palonen, Ministry of Social Affairs and Health, Council for Choices in Health Care in Finland

Subcommittee on Rare Diseases 1 January 2018–31 December 2019

Ritva Halila, Ministry of Social Affairs and Health, Chair
Mikko Seppänen, Hospital District of Helsinki and Uusimaa
Jarmo Jääskeläinen, Kuopio University Hospital
Outi Kuismin, Oulu University Hospital
Pasi Nevalainen, Tampere University Hospital
Johanna Lempainen, Turku University Hospital
Helena Kääriäinen, National Institute for Health and Welfare
Risto Heikkinen, Finnish Network for Rare Diseases, Allergia-, iho- ja astmaliitto ry
Marianne Eronen, Social Insurance Institution of Finland
Elina Rantanen, Norio - Centre of Rare Diseases
Päivi Nurmi-Koikkalainen, National Institute for Health and Welfare
Katri Asikainen, HARSO

PUBLICATIONS CAN BE DOWNLOADED AT:
julkaisut.valtioneuvosto.fi

ISSN 2242-0037 (PDF)
ISBN 978-952-00-5670-4 (PDF)