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Finland's National Plan for

RARE DISEASES 2014–2017

Steering group report

MINISTRY OF SOCIAL AFFAIRS AND HEALTH
Helsinki 2015

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National Plan for Rare Diseases 2014–2017 - Steering group report

Abstract

■ The steering group for rare diseases has prepared a proposal for a National Plan for Rare Diseases 2014–2017. Stakeholders had an opportunity to participate in the preparation of the plan. A key objective of the plan is that people with a rare disease can live a full life of their own choosing regardless of their disease and receive not only appropriate care and habilitation, but also psychosocial support indicated by their needs.

Finnish social and health care services do not currently provide a satisfactory response to the needs of patients with a rare disease. Due to the low prevalence of rare diseases and scarcity of relevant expertise, dedicated measures are needed in the service system to ensure equity of access to quality care and habilitation for people with a rare disease.

The National Plan contains proposals for actions aiming to develop research, care, habilitation and social support. As priority actions, the plan proposes acknowledging in statutes or in their rationale the need for dedicated measures targeted at rare diseases, clarifying the care pathway for people with a rare disease, establishing units for rare diseases in university hospitals, promoting the availability and reimbursement of orphan drugs, setting up a national coordinating centre for rare diseases, and developing social support and habilitation.

The plan's goals include empowering people with a rare disease, their families and patient organisations to participate in decision-making in the field of rare diseases and the planning of services. Patient organisations' opportunities for exerting influence must be improved at all levels of the service system.

Key words

rare diseases, health services, social services, patient organisations, empowerment

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Harvinaisten sairauksien kansallinen ohjelma 2014–2017 - Ohjausryhmän raportti

Tiivistelmä

■ Harvinaiset sairaudet -ohjausryhmä esittää ehdotukset harvinaisten sairauksien kansalliseksi ohjelmaksi vuosille 2014–2017. Ohjelma on laadittu sidosryhmiä osallistaen. Ohjelman keskeisenä tavoitteena on, että henkilö voi harvinaissairaudesta huolimatta elää täysipainoista ja omiin valintoihinsa perustuvaa elämää ja saada siihen asianmukaisen hoidon ja kuntoutuksen lisäksi tarpeen mukaisen psykososiaalisen tuen.

Sosiaali- ja terveydenhuoltopalvelut eivät nykyisellään vastaa tyydyttävästi harvinaissairaiden tarpeisiin. Pienen esiintyvyytensä sekä asiantuntemuksen harvinaisuuden vuoksi harvinaiset sairaudet edellyttävät erityistoimia palvelujärjestelmässä, jotta niitä sairastavien oikeus laadukkaaseen hoitoon ja kuntoutukseen toteutuisi yhdenvertaisesti.

Kansallinen ohjelma sisältää toimenpide-ehdotukset tutkimuksen, hoidon, kuntoutuksen ja sosiaalisen tuen kehittämiseksi. Ohjelmassa esitetään ensisijaisiksi toimenpiteiksi harvinaissairauksien vaatimien erityistoimenpiteiden tunnustamista lainsäädännössä tai niiden perusteluissa, harvinaissairaiden hoitopolun selkeyttämistä, harvinaissairauksien yksiköiden perustamista yliopistosairaaloihin, harvinaislääkkeiden saatavuuden ja korvattavuuden edistämistä, kansallisen koordinoivan keskuksen perustamista sekä sosiaalisen tuen ja kuntoutuksen kehittämistä.

Ohjelman yhtenä tavoitteena on, että harvinaissairaat, heidän läheisensä ja potilasjärjestöt osallistuvat paremmin harvinaissairauksia koskevaan päätöksentekoon ja palveluiden suunnitteluun. Potilasjärjestöjen vaikutusmahdollisuuksia tulee kasvattaa palvelujärjestelmän kaikilla tasoilla.

Asiasanat

harvinaiset taudit, terveyspalvelut, sosiaalipalvelut, potilasjärjestöt, voimaantuminen

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SOSIAALI- JA TERVEYSMINISTERIÖ

PRESENTATIONSBLAD

Utgivare	
Social- och hälsovårdsministeriet	13/03/2014
Författare	Uppdragsgivare
Styrgruppen för sällsynta sjukdomar Ordförande: Liisa-Maria Voipio-Pulkki, SHM Vice ordförande: Jaakko Yrjö-Koskinen, SHM	Social- och hälsovårdsministeriet
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Rapportens titel

Nationella programmet för sällsynta sjukdomar 2014–2017 – Styrgruppens rapport

Referat

■ Styrgruppen för sällsynta sjukdomar framför sina förslag till ett nationellt program för sällsynta sjukdomar för åren 2014–2017. Programmet har utarbetats genom att engagera intressentgrupperna. Det centrala målet för programmet är att en person trots sin sällsynta sjukdom ska kunna leva ett fullödigt liv genom att göra sina egna val och få förutom ändamålsenlig vård och rehabilitering också behövligt psykosocialt stöd.

Social- och hälsovårdstjänsterna kan i detta nu inte tillräckligt väl tillgodose behoven hos personer som lider av sällsynta sjukdomar. Eftersom dessa sjukdomar förekommer i liten omfattning och expertis i dem är sällsynt krävs speciella åtgärder av servicesystemet för att de sjukas rätt till kvalitativ vård och rehabilitering ska kunna tillgodoses på ett likvärdigt sätt.

Det nationella programmet inkluderar förslag till åtgärder för att utveckla forskningen, vården, rehabiliteringen och det sociala stödet. Som åtgärder i första hand föreslås att man ska erkänna inom lagstiftningen eller motiveringen till lagstiftning att sällsynta sjukdomar kräver speciella åtgärder och att man ska förtydliga vårdstigen för sällsynta sjukdomar, inrätta enheter för sällsynta sjukdomar vid universitetssjukhus, främja tillgången till och ersättningen för läkemedel som behövs vid sällsynta sjukdomar, inrätta ett nationellt centrum för koordinering samt utveckla det sociala stödet och rehabiliteringen.

Ett mål för programmet är att personer med sällsynta sjukdomar, deras närstående och patientorganisationer i högre grad ska delta i beslutsfattandet och planeringen av tjänster med anknytning till sällsynta sjukdomar. Patientorganisationernas möjligheter att påverka bör ökas på servicesystemets samtliga nivåer.

Nyckelord

sällsynta sjukdomar, hälsovårdstjänster, socialtjänst, patientorganisationer, brukarmedverkan

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SOCIAL- OCH HÄLSOVÅRDSMINISTERIET

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To the Ministry of Social Affairs and Health

A steering group for rare diseases was appointed by the Ministry of Social Affairs and Health for the period 1 June 2012–31 December 2013.

The objectives of the steering group were the following:

1. Promoting the utilisation of the Finnish health care system's knowledge and experience for improving the lives of people with a rare disease
2. Promoting research and equity in access to quality care
3. Steering Finland's activities in the European Union Committee of Experts on Rare Diseases (EUCERD) Joint Action and issues related to the implementation of the EU Directive on patients' rights cross-border healthcare.

The steering group's tasks were to:

1. Plan and direct the implementation of the Council Recommendation on action in the field of rare diseases in Finland
2. Submit proposals concerning the monitoring of implemented actions and other measures that may be necessary.

Steering group chairperson was Director Liisa-Maria Voipio-Pulkki from the Ministry of Social Affairs and Health. **Vice chairperson** of the steering group was General Secretary Päivi Topo from the National Advisory Board on Social Welfare and Health Care Ethics until 4 February 2013, after which date this role was performed by Ministerial Counsellor Jaakko Yrjö-Koskinen from the Ministry of Social Affairs and Health.

The appointed **steering group members** and their personal deputies (in brackets) were the following:

- General Secretary Leila Jylhänkangas, National Advisory Board on Social Welfare and Health Care Ethics, took over from General Secretary Päivi Topo as from 31 May 2013
- Ministerial Adviser Pälvi Kaukonen, Member until 6 November 2012, was substituted by Ministerial Counsellor Jaakko Yrjö-Koskinen, both from the Ministry of Social Affairs and Health
- Director Annakaisa Iivari, Member until 20 March 2013, was substituted by Senior Medical Officer Teppo Heikkilä, both from the Ministry of Social Affairs and Health
- Ministerial Adviser Jaana Huhta, Ministry of Social Affairs and Health (Ministerial Adviser Anne-Mari Raassina)
- Research Professor Helena Kääriäinen, National Institute for Health and Welfare (Head of Unit Jukka Kärkkäinen)
- Chief Administrative Physician Päivi Koivuranta-Vaara, Association of Finnish Local and Regional Authorities (Development Manager Heikki Punnonen)

- Chief Physician, Paediatric Neurology Pirkko Karttunen-Lewandowski, Etelä-Savo Hospital District Joint Municipal Authority (Medical Director Matti Suistomaa)
- Chief Physician Leila Saulamaa, Pirkanmaa Hospital District (Paediatrician Päivi Keskinen)
- Chief Physician Markku Savolainen, Northern Ostrobothnia Hospital District (Paediatric Neurologist Johanna Uusimaa)
- Docent Harri Niinikoski, Southwest Finland Hospital District
- Professor of Paediatrics Jarmo Jääskeläinen, Pohjois-Savo Hospital District (Professor of Clinical Epileptology Reetta Kälviäinen)
- Neurologist Riitta Pirilä, Lapland Hospital District (Paediatric Neurologist Laura Simontaival)
- Head of Nursing Leena Vekara, City of Tampere (Head Nurse Anja Jalonen-Männikkö)
- Senior Social Worker Emmi Hanhikoski, City of Jyväskylä
- Specialist, Plastic Surgery Jorma Rautio, Hospital District of Helsinki and Uusimaa, the Cleft Palate and Craniofacial Centre (Specialist, Internal Medicine Ilkka Kantola)
- Docent Sirkku Peltonen, Turku University Central Hospital (Professor Anna-Elina Lehesjoki)
- Docent Risto Lapatto, Helsinki University Central Hospital, Gynaecological and Paediatric profit centre
- Chief Physician Niilo Keränen, Kuusamo Health Centre, member of the National Network of Public Health Care Managers
- Chief Physician Riitta Salonen-Kajander, Rinnekoti Foundation (Specialist in Genetics Kristiina Avela, and as from 20 March 2013, Informatician Leena Toivanen, both from Rinnekoti Foundation)
- Board Member Katri Karlsson, Harso Association (Board Member Päivi Reinikka, Harso Association)
- Senior Planning Officer Eila Niemi, the Finnish MS Society / the Finnish Network for Rare Diseases (Development Manager Jaana Hirvonen, the Finnish Rheumatism Association / the Finnish Network for Rare Diseases)
- Chairperson Jukka Sariola, the Finnish Neuromuscular Disorders Association / the Finnish Network for Rare Diseases (Jussi Lindevall, DebRA Finland / the Finnish Network of Rare Diseases)

On 18 October 2012, Ministerial Counsellor Ulla Närhi from the Ministry of Social Affairs and Health and Research Professor Ilona Autti-Rämö from Social Insurance Institution's research division were appointed to the steering group as additional members.

The steering group met nine times. Two working groups were set up to prepare the National Plan and a seminar focusing on it and to draw up proposals for the centre of expertise selection process. On 18–19 March 2013, the steering group organised a seminar in Hanasaari Cultural Centre to engage stakeholders in the preparation of the National Plan. A total of 78 stakeholder representatives, of whom 29 were from patient organisations, took part in the seminar. The steering group consulted Docent Jaana Lähdetie from Turku University Central Hospital and Director Jarmo Wahlfors from the Academy of Finland.

The Ministry of Social Affairs and Health concluded service contracts with Katja Aktan-Collan (16 April–31 December 2012) and Elina Rantanen (3 April–30 December 2013) on providing assistance in the preparation of the National Plan under the direction of the steering group.

In October 2013, the steering group requested feedback on the Draft National Plan from its members' background organisations. The feedback received was taken into consideration in the final version of the National Plan.

The steering group submits its proposal to the Ministry of Social Affairs and Health and urges the Ministry to initiate the requisite further measures to implement the National Plan for Rare Diseases.

Helsinki, 13 March 2014

Liisa-Maria Voipio-Pulkki Jaakko Yrjö-Koskinen

Leila Jylhänkangas	Teppo Heikkilä	Jaana Huhta
Ulla Närhi	Helena Kääriäinen	Ilona Autti-Rämö
Päivi Koivuranta-Vaara	Pirkko Karttunen-Lewandowski	
Leila Saulamaa	Markku Savolainen	Harri Niinikoski
Jarmo Jääskeläinen	Riitta Pirilä	Leena Vekara
Emmi Hanhikoski	Jorma Rautio	Sirkku Peltonen
Risto Lapatto	Niilo Keränen	Riitta Salonen-Kajander
Katri Karlsson	Eila Niemi	Jukka Sariola

1 INTRODUCTION

The European Union's definition of a rare disease is a disease affecting no more than 5 per 10 000 persons. It is estimated that over 300 000 Finnish people are affected by a rare disease, disability, syndrome or malformation. They account for some six per cent of the population and a considerably larger share of major users of healthcare services. It is thus justified to take people with a rare disease into consideration as a specific group in the act on the arrangement of social welfare and health care services and in the development of the service structure. Living with a rare disease is associated with challenges encountered in the identification and care of diseases and disabilities, habilitation, services and everyday life. The rarer the disease or disability, the more challenging it may be to find information and assistance. Plenty of resources are misspent when finding a correct diagnosis and suitable care takes an excessively long period of time. With the same expenditure of resources, the disease could be diagnosed and care could be provided much better and faster.

In the National Plan, the concepts *rare disease* and *people living with a rare disease* are used to describe the similar problems encountered by people with a rare disease and actions taken to solve these problems. There are thousands of different rare diseases. At least 5 000 have been discovered, and new ones are continuously being described in the literature. People living with a rare disease often share very similar problems, for example in receiving a correct diagnosis or finding appropriate information and expert specialist services. Particular attention also needs to be paid to issues related to quality of care, access to medical and social support, and the effectiveness of care pathways between primary health care and specialised medical care. Aspects related to professional and social integration, independent coping, using ordinary services and managing everyday routines are also highlighted in the lives of people with a rare disease and their families. Peer support is less readily available for them than for other patient groups, and they are more exposed to psychological, social and financial problems.

The Council of the European Union issued a recommendation on an action in the field of rare diseases in 2009 (Appendix 1). The recommendation notes that the principles and overarching values of universality, access to good quality care, equity and solidarity, as endorsed in the Council conclusions on common values and principles in EU health systems of 2 June 2006, are of paramount importance for patients with rare diseases. The Member States are urged to elaborate their plans and strategies in the field of rare diseases. A national plan in the field of rare diseases should be elaborated and adopted by the end of 2013 at the latest. The objective of the EU is to establish centres of expertise for rare diseases and to create networks for sharing expertise.

A steering group on rare diseases was appointed by the Ministry of Social Affairs and Health for the period 1 June 2012─31 December 2013 and tasked to draw up Finland's National Plan for Rare Diseases. The steering group had the following objectives:

1. Promote the use of knowledge and experience in the Finnish healthcare system for the benefit of patients with rare diseases.
2. Promote research and equity in access to quality care
3. Steer Finland’s activities in the European Union Committee of Experts on Rare Diseases (EUCERD) Joint Action and issues related to the implementation of the EU Directive on patients' rights in cross-border healthcare.

The steering group was tasked with the following:

1. Plan and direct the implementation of the Council Recommendation on action in the field of rare diseases in Finland.
2. Submit proposals concerning the monitoring of implemented actions and other measures that may be necessary.

The steering group appointed two working groups among its members to plan the drafting of the National Plan and to consider the establishment of centres of expertise. A seminar on the National Plan for Rare Diseases was organised on 18–19 March 2013 to support plan preparation. At this seminar, the participants were divided into groups whose task was to identify requisite actions in the following areas of the National Plan: research in rare diseases, disease classification systems and registries of rare diseases, national definition and diagnosis of rare diseases, improvement of care and access to drugs, national centres of expertise, developing social support and the empowerment of people living with rare disease, as well as implementation, coordination, monitoring, evaluation, financing and international networking related to the national plan. A total of 78 representatives from various hospital districts, the Finnish Network for Rare Diseases and the HARSO network, the Social Insurance Institution, municipalities, the Finnish Slot Machine Association RAY and the Ministry of Social Affairs and Health attended the seminar.

With financial support from EUROPLAN (European Project for Rare Diseases National Plans Development), the Finnish umbrella organisation for patients with rare diseases HARSO organised a conference in September 2013 that offered patient organisation representatives an opportunity to comment on the draft National Plan for Rare Diseases. This meeting was attended by 60 NGO representatives.

The outcome of the above mentioned seminar provided the basis for drafting the National Plan for Rare Diseases. Feedback received from the participants to the conference hosted by HARSO and from the organizations that were represented in the steering group was taken into account in the finalisation of the plan. The plan aims to improve the quality of life for people living with a rare disease through their empowerment and by increasing the responsiveness of the Finnish health and social care system to their needs. People with a rare disease are entitled to the same quality of care and habilitation as other patients. A correct diagnosis, habilitation, medication and coping with everyday routines are essential elements of quality of life. Their value cannot be measured in monetary terms. Timely diagnosis of a rare disease and appropriate treatment and care also bring savings in health care expenditure. Expertise in diagnostics and care in the field of rare diseases is available in Finland, but it has not been put to effective use. The objective is to achieve better health outcomes by using available resources more efficiently. The plan aims to promote actions that are of high importance and have a real impact on the quality of life and well-being of people living with a rare disease. In this plan, the concept of rare diseases refers to rare diseases, disabilities, syndromes and malformations.

2 FINLAND'S NATIONAL PLAN FOR RARE DISEASES

2.1 BACKGROUND ON THE FINNISH SITUATION

While no National Plan for Rare Diseases has previously been prepared in Finland, rare diseases have been the object of increasing attention in the 1990s and 2000s. In 1991, the Ministry of Social Affairs and Health designated the Organisation of Respiratory Health in Finland, the Finnish Central Organisation for Skin Patients, the Finnish Association of People with Physical Disabilities, Inclusion Finland KVTL, the Federation of Hard of Hearing, the Finnish Federation of the Visually Impaired, the Rehabilitation Home for Children, the Finnish MS Society and the Finnish Rheumatism Association as resource centres for rare diseases. In 1995, the Finnish Network for Rare Diseases was set up, and its affiliated members included not only the organisations listed above but also other organisations in the field of social welfare and health working with patients with a rare disease.

In spring 2012, HARSO Organisation for Rare Diseases and Disabilities in Finland was established as an umbrella organisation for people with a rare disease. Its members comprise 22 national patient associations. HARSO strives to promote equal participation in society of people living with a rare disease and their families and better social and health care services for people with a rare disease. It also offers peer support for those whose disease is so rare that they have no dedicated patient organisation.

National events on rare diseases are organised in Finland every year. The Orphanet team[1] collects and distributes information about expertise in the field of rare diseases. Health care system actors can access Orphanet information through Terveysportti health portal. A Nordic cooperation project was implemented in 2009−2010 with the purpose of sharing information and organising joint training events. While no centres of expertise for rare diseases have been designated in Finland so far, the university hospitals usually serve in this role, and some of them have expertise in certain diseases. The small population of the country and the Finnish diseases heritage set their own requirements to the National Plan for Rare Diseases and international cooperation.

Finland's National Plan for Rare Diseases is influenced by many on-going national legislative reforms, including preparing the ratification of the UN Convention on the Rights of People with Disabilities and the setting up of a national Comprehensive Cancer Center. The framework for building up and sharing of expertise in the field of rare diseases is provided by the Health Care Act, the objective of which is to ensure equal access to quality health services and patient safety (Government decree 2011). The Health Care Act requires hospital districts within a catchment area for specialised medical care to agree upon the division of responsibilities and coordination (specialised medical care agreement). Some procedures and services that are deemed to constitute highly specialised medical care may be centralised.

The increased freedom of choice affects the organisation of diagnostics and care in the field of rare diseases both on the national and the EU level. Under the Health Care Act the patient may, on certain conditions, choose a treatment facility anywhere in Finland as from 2014. The European context related to care in the field of rare diseases has been transformed by the Directive on the application of patients' rights in cross-border healthcare (2011) implemented in Finland from the beginning of 2014. Centres of expertise for rare diseases are needed in

[1] Orphanet is an open European online service for rare diseases and orphan drugs. Its mission is to improve the diagnostics and care of rare diseases.

Finland to ensure optimal utilisation of national expertise; if patients seek care in other countries, this may prove significantly more expensive for them.

The on-going reform of the social and health care service structure strives to contribute to guaranteeing equity in access to these services. The policies that set the course for this reform assign responsibility for organising social and health services to social welfare and health care regions. The tasks of the catchments areas for highly specialised medical care would include providing social and health care services specified as specialised level tasks, with the aim of eliminating overlaps in the service network and coordinating the activities regionally. An ownership structure that guarantees them adequate resources and an administrative solution that does not result in fragmentation of the functional whole has been proposed for university hospitals. Research, development and teaching are expected to have close links with service provision. The Government Bill on the arrangement of social welfare and health care services is to be submitted to the Parliament in spring 2014.

The extent of and gaps in Finnish expertise in the field of rare diseases were surveyed by means of a questionnaire directed at chief physicians in specialised fields in university and central hospitals and the member organisations of the Finnish Network for Rare Diseases (Ministry of Social Affairs and Health 2011). The responses indicated a need for more cooperation in diagnostics and care in the field of rare diseases. In particular, the respondents called for more cooperation across administrative boundaries between hospital districts that would allow the sharing of responsibility for diagnostics, care and habilitation in the field of rare diseases between university hospitals. Various consultation networks were suggested as ways of addressing the existing shortcomings. While consultations are common today, the respondents would prefer a more clear-cut process based on agreements. The respondents also expressed a need for joint training and meetings where rare diseases could be discussed between different fields on the one hand, and between representatives of university hospitals and central hospitals on the other. They advocated centralisation, both within the special catchment areas and nationally. In addition to cooperation, there were shortcomings in knowledge and resources. The respondents called for training and clear instructions to eliminate gaps in knowledge. Lack of resources was seen to be particularly relevant to the numbers of physicians and their job descriptions: physicians cannot devote any more time to acquiring competence in rare diseases than in common diseases, even if more time is needed to acquire such competence in the field of rare diseases.

A country-.specific study (2009) conducted by EURORDIS, an umbrella patient organisation for people with a rare disease, noted that people living with a rare disease in Finland had encountered shortcomings in access to information and care. One out of three Finnish respondents had been given a diagnosis without being appropriately informed of what the diagnosis means. 70% of the respondents were left without psychological support, even if 91% felt that such support was important. 37% felt that accessing essential health services was difficult or impossible because these services did not exist, they were too expensive, the patient had not been referred to these services, the waiting times were excessive, or the services were too far away and difficult to reach.

2.2 NATIONAL PLAN OBJECTIVES

The steering group proposes the following goals for Finland's National Plan for Rare Diseases:

- People with a rare disease will have access to services of the same quality as other patients
- Preventive, diagnostic, care and habilitation services as well as social services required by people with a rare disease will be delivered to a high standard and equitably across the country
- Units and centres of expertise for rare diseases will be set up

- Rare diseases will be diagnosed more promptly, and fewer patients will be left without a definitive diagnosis
- Premature morbidity and mortality will be reduced
- Multidisciplinary care will be better coordinated
- Communication between professionals involved in the care of a patient will be improved, including during transitions from one care facility to another, or from paediatric to adult services
- Healthcare resources in the field of rare diseases will be utilised more effectively
- The quality of life and socio-economic situation of people living with a rare disease will improve
- The availability of orphan drugs will be improved, and their costs to patients will be reimbursed more fully
- Knowledge of rare diseases will be built up through research
- Social welfare and health care professionals will have more extensive knowledge of rare diseases
- Participation in international co-operation in the field of rare diseases will increase
- Clearer cooperation channels will be created between experts, both nationally and internationally
- People living with a rare disease and their families will be empowered to participate in decision-making in the field of rare diseases
- The National Plan for Rare Diseases will be sustainably integrated into the Finnish social welfare and healthcare system

The National Plan addresses the challenges posed by rare diseases in five areas. To respond to these challenges, the steering group proposes thirteen concrete actions that will promote the achievement of the aforementioned general objectives of the National Plan. In connection with each proposed action, indicators for monitoring progress, the responsible actor, a timeline and the prerequisites for successfully achieving the objective are set out. At the end of the plan document, the methodology for coordinating, monitoring and evaluating the implementation of the plan is described, and a summary of the actions is given in which priority areas are highlighted.

3 AREAS OF THE NATIONAL PLAN

3.1 AREA 1: DEFINITION AND REGISTRATION OF RARE DISEASES

The first step towards improving the care and quality of life of people with a rare disease is acknowledging their situation. Compared to the remainder of the population, people with a rare disease have poorer access to expert services. To guarantee people with a rare disease equality with those affected by more common diseases, dedicated measures are required in the service system. Even if each rare disease only affects a handful of people, the total number of patients is large: approximately six per cent of the adult population will be affected by a rare disease during their lives. In specialised medical care, the share of patients with a rare disease is significantly greater than this.

A precondition for acknowledging the situation of people with a rare disease is adopting a uniform definition of rare diseases that enables to identify persons who are within the scope of dedicated measures required by rare diseases. As a result of inconsistencies in this definition at the EU level, problems arise in joint research projects and the sharing of expertise.

Research and the dissemination of information are also hampered by a lack of databases on rare diseases. Since no data has been compiled in Finland on clinics treating rare diseases, on treatment practices, nor on research, the collection of information and exploitation of expertise are random and based on personal contacts. This prevents patients from accessing the best available expertise in diagnostics, care and habilitation.

3.1.1 Action 1: Adopting a uniform definition of rare diseases and acknowledging the need for dedicated measures

A definition of rare diseases will be adopted that is consistent with the EU definition. The need for dedicated measures targeted at rare diseases will be acknowledged in the legislation.

The European Union recommendation proposes the implementation of a common identification of rare diseases by all the Member States, which would facilitate cooperation at Community level in the field of rare diseases. This National Plan uses the EU definition of rare diseases. It defines as rare a disease affecting no more than 5 per 10 000 persons. In Finland, the EU definition means no more than 2 800 persons per each disease. In this plan document, rare diseases are understood in a broad sense to also comprise disabilities, malformations and syndromes. The plan seeks to respond to the challenge of rare diseases that are life-threatening or chronically debilitating.

The majority of rare diseases have an extremely low prevalence. As the greatest challenges related to diagnostics and care are associated with these, the rarest of all diseases, they should be addressed separately in the National Plan implementation. It may be necessary to resort to international cooperation networks to seek expertise in these diseases.

People living with a rare disease have the right to care of a similar quality that is available to other patients, and to social services that meet their specific needs. Because of their low prevalence and the scarcity of relevant expertise, rare diseases call for dedicated measures to ensure equal access to quality services. This has been the justification for European Union action in the field of rare diseases. This principle should be incorporated either in the new act on the arrangement of social and health care services or in its explanatory memorandum.

Objectives:

- Defining rare diseases following the EU definition.
- Acknowledging in the act on the arrangement of social welfare and health care services or its rationale that rare diseases necessitate dedicated measures in the service system.

Indicator: Dedicated measures necessitated by rare diseases are referred to in the act or its rationale.

Responsible actor: Ministry of Social Affairs and Health

Timeline: The definition is introduced with immediate effect; the need for dedicated measures is acknowledged in the act on the arrangement of social welfare and health care services which is to enter into force in 2015, or in its rationale.

Prerequisites: Political support

3.1.2 Action 2: Registry of rare diseases

A registry of rare diseases will be created as part of the current electronic Care Register for Health Care (Hilmo). For this purpose, a disease classification system that covers rare diseases more extensively than the current ICD-10 system must be introduced.

Having a national registry of rare diseases in Finland is a precondition for developing both research and care. While systematic, data collection should be as automatic as possible; completing separate registration forms is labour-intensive and thus does not promote the collection of a comprehensive database. The compilation of registers should not entail additional work; a key feature of the registry is that it makes rare diagnoses more readily identifiable in the health care system.

A separate national registry of rare diseases is not to be established in Finland. The registration of rare diseases will take place using the Care Register for Health Care (Hilmo) maintained by the National Institute for Health and Welfare. Finding rare diseases in the Hilmo register is the most crucial question in terms of following care pathways and disseminating epidemiological information. In the registry of rare diseases, an international diseases classification system will be used. This could be the ICD-11 system which, as far as is currently known, will include codes for a much wider spectrum of rare diseases than the current version. It is expected that ICD-11 will be in use by 2017. The actors responsible for maintaining health care registers in Finland should also take rare diseases into consideration when monitoring the development of codification systems and before making a decision on whether to wait for the completion of ICD-11 or whether some other disease classification system should be introduced in Finland before this date.

In addition to an automated ICD registry, centres of expertise may compile their own registries of individual diseases. Patient consent must be obtained before registering patient data in them.

Objectives:

- People with a rare disease can be more readily identified.

- The possibility of identifying patient groups will facilitate the launching of research projects and improve opportunities for cooperation.

Indicators: Codes for a much wider spectrum of rare diseases included in Hilmo

Responsible actor: National Institute for Health and Welfare

Timeline: Registry compilation will start at the latest when the ICD-11 classification system is introduced

Prerequisites: Implementation of ICD-11 or introduction of some other disease codification system

3.2 AREA 2: RESEARCH IN THE FIELD OF RARE DISEASES

In the allocation of funds for research, research in the field of rare diseases may be overlooked in favour of more common diseases. It is difficult to collect sufficiently large patient data sets for clinical and epidemiological research because patient groups are small and due to the lack of registries and inadequate diagnostics. For the same reasons, research on the quality of life of people with a rare disease and on how they cope with it in their everyday lives is unable to compete for research funding. Development of treatments is driven by the pharmaceutical industry which is not interested in developing drugs for small patient groups as financial gains from this would be uncertain. Information on research projects has not been systematically complied. Researchers are thus missing out on the benefits of cooperation. However, since the development of social and health care services must be based on information, there is a need for research focussing on the services for people with a rare disease and on the impact of the services on their health, well-being and coping with everyday life.

3.2.1 Action 3: Research funding and a research programme focusing on rare diseases

A share of national research funding will be earmarked for research in rare diseases and the Finnish disease heritage. An initiative will be prepared and submitted to the Academy of Finland concerning the development of a multidisciplinary research programme for rare diseases.

Research in the field of rare diseases is needed to improve the quality of patients' lives. The results may also be exploited in the care of patients with less rare diseases. A stronger national foundation for research in rare diseases is needed to enable Finland to take part in European research projects on rare diseases. Adequate funding must also be reserved for research in conditions that are part of the Finnish disease heritage and the related care, as little research in this field is conducted anywhere else.

University level health research is carried out in health care units with national research funding. The priorities and objectives of this research are defined by the Ministry of Social Affairs and Health for a four-year period at a time. The Ministry grants research funding to the

scientific committees of the special catchment areas, which make decisions on allocating funding to research projects in their areas on the basis of applications. Research in rare diseases and the Finnish disease heritage implemented in cooperation between the catchment areas is proposed as one priority area of research.

Through its research programmes, the Academy of Finland channels funding to areas of research that are vital for science and society. The research programmes aim to develop selected fields, improve the scientific standard in these fields and create expertise of a new type. The programmes stress a multidisciplinary and interdisciplinary approach and international cooperation. An initiative will be submitted to the Academy of Finland to propose the establishment of a research programme for rare diseases. This research programme should include research in disease mechanisms, diagnostics, care and social support as well as in the coping and quality of life of people with a rare disease. Information about on-going research projects in the field of rare diseases will be collected in a single database.

Objectives:

- Increasing funding allocated to research in rare diseases.
- Developing a cooperation network on rare diseases and a database of on-going research.
- Promotion of research related to people with a rare disease as users of social and health care services and research on the effectiveness of the service system, which will be implemented in broad-based cooperation with experts in the sector, including patient and client organisations.

Indicators: Share of national research funding earmarked for rare diseases and the Finnish disease heritage; Initiative concerning a research programme for rare diseases submitted to and a decision to prepare this programme made in the Academy of Finland; Data gathered on on-going research.

Responsible actor: Ministry of Social Affairs and Health: Rare diseases as a priority in national research funding; University hospitals: Coordination of the research programme initiative; National coordinating centre for rare diseases: Compilation of a research database

Timeline: Drafting of national research funding in 2014−2015, implementation at the beginning of the forthcoming four-year period in 2016−2019; Initiative concerning the research programme submitted to the Academy of Finland in 2014; Research database in 2015

Prerequisites: Confirming the actors responsible for submitting the initiatives

3.2.2 Action 4: Building up international research cooperation

Possibilities for international research cooperation will be improved by applying for membership in the E-RARE project.

As the numbers of patients affected by each individual rare disease are not adequate in Finland for clinical studies needed to develop treatments, international networking in the collection of information and samples is required. An administrative recommendation on collecting registries of this type is needed in Finland to speed up the permit process and enable cooperation.

Boosting national research funding dedicated to rare diseases will enable Finnish scientists to compete for European Union funding sources. Finnish scientists should tap ERC funding (European Research Council) and the Horizon 2020 funding programme as sources of funding for research projects on rare diseases.

In 2011, the European Commission and US National Institutes for Health Research launched cooperation under the auspices of IRDiRC (International Rare Diseases Research Consortium) to promote international cooperation in research in the field of rare diseases. The objective of this consortium is to develop 200 new treatments for rare diseases and to find a diagnosis for nearly all people with a rare disease by 2020. Each country or organisation participating in the IRDiRC is obliged to fund research in the field of rare diseases by USD 10 million over a five-year period. This funding will be spent on research projects carried out in the member country or organisation. The Academy of Finland joined the IRDiRC in June 2013.

The European Union's E-RARE-1 project succeeded in uniting European research in rare diseases. The E-RARE-2 project that continues until 2014 represents a step further in this cooperation. The steering group proposes that the Academy of Finland take part in the next phase of E-RARE.

Objectives:

- Engaging in international cooperation in order to obtain sufficiently large patient data sets and to improve the lives of people with a rare disease.
- Allocating adequate national funding to research in rare diseases that would allow participation in the international cooperation project through E-RARE membership.

Indicators: Adequate national funding enabling Finland to join E-RARE

Responsible actor: Ministry of Social Affairs and Health, Ministry of Education and Culture, Academy of Finland

Timeline: Application for E-RARE membership at the launch of the next phase in 2014

Prerequisites: Adequate national funding

3.3 AREA 3: BETTER AND MORE EFFECTIVE HEALTH CARE FOR PEOPLE LIVING WITH A RARE DISEASE

People living with a rare disease and their families encounter many types of problems in the health care system. As the disease is rare, finding a correct diagnosis is often delayed. Consequently, a significant share of the patients are assessed by several physicians before a correct diagnosis can be pinned down. Some patients are initially given one or more incorrect diagnoses, as physicians are not always alert to the possibility of the symptoms being caused by a rare disease. In particular, physicians and care personnel working in primary health care do not have enough knowledge about rare diseases. In most cases, a precondition for a timely diagnosis of a rare disease is simultaneous consultation with several specialist fields, which our current health care system does not facilitate. While all tasks requiring special expertise require more time than routine work, this time has not been set aside. Delays in finding a diagnosis often result in significant harm to people with a rare disease and their families. A

more prompt diagnosis would also save society's resources by eliminating the need for being assessed by several experts and allowing the timely initiation of care and habilitation.

Once people with a rare disease have received a correct diagnosis, they meet new challenges. While new symptoms and diagnostic changes may occur after the disease has been diagnosed, they are not always examined. Many rare diseases require a high level of specialist competence and multidisciplinary care. When the care and habilitation of a person with a rare disease is being planned, a single actor to assume overall responsibility may be lacking. While each field of specialisation concentrates on treating certain symptoms, responsibility for the care of the patient as a whole is not necessarily assigned to anyone. As a paediatric patient grows up and the responsibility for his or her care shifts to specialised medical care for adults, the continuity of care is not always guaranteed. There also are regional differences in access to care, and a person with a rare disease is not always referred to a facility with adequate competence.

All health care professionals, for example those in emergency care, do not have enough knowledge about rare diseases and the care procedures indicated by them. Specialised medical care fails to prepare sufficiently detailed instructions for primary health care. In acute situations, patients need rapid access to a unit with skilled staff and adequate experience.

No pharmacotherapy is currently available for the majority of rare diseases. Specific drugs have been developed to treat many rare diseases in recent years, but their effectiveness has often not been established with certainty, as small numbers of patients dispersed in different countries are not sufficient to reliably confirm the results. Many orphan drugs are also expensive, as an orphan drug status granted by the European Commission guarantees drug market exclusivity for 10 years, and due to the low demand, pharmaceutical companies have no interest in bringing generic products to the market after the period of exclusivity has ended. However, pharmaceutical companies are becoming more interested in developing orphan drugs and converting drugs intended for the treatment of ordinary illnesses into orphan drugs. For optimal outcomes, surgical treatment of rare diseases requires technical skills which develop with sufficient repetition of the procedure. Centralisation of rare surgical procedures is thus justified.

Some rare diseases can be diagnosed at birth before the onset of observable symptoms. Starting treatment before the onset of symptoms is less expensive and, most importantly, the outcome is much better. These diseases can be detected by screening tests carried out on newborns, of which only the test for congenital thyroid insufficiency is used in Finland. The newborn screening programme should be expanded, following the example of many other European countries. A precondition for expanding the screening programme is appropriate resource allocation. A working group on screening appointed by the Ministry of Social Affairs and Health has considered the necessity of establishing a national screening centre.

3.3.1 Action 5: Clarification of the care pathway for people with a rare disease

The diagnosis and care pathway for people with a rare disease will be streamlined. Awareness of the pathways will be spread at various levels of the health care system. A care passport should be provided for all people with a rare disease who need one.

In order to find better solutions to problems encountered by people with a rare disease, clear care pathways should be created for them, and all levels of health care, NGOs, social services, the Social Insurance Institution, people living with a rare disease and their families should be informed of them. Figure 1 depicts the care pathway of a person with a rare disease. The

pathway includes both the current health care actors and the new centres of expertise and units for rare diseases to be set up. For a more detailed description of these, see Actions 6 and 7.

The point of departure is a patient with symptoms whose disease is so rare that a diagnosis cannot be found immediately. His or her care pathway will typically start with primary health care or specialised medical care at the primary care level. From this point on, the diagnostics, care and habilitation may take any course on the care pathway that optimally responds to the patient's situation. The purpose of defining the care pathway is clarifying and accelerating diagnostics and care delivery to people with a rare disease, not making the patients' life more difficult by centralising care to a small number of units. Each person with a rare disease must be able to proceed on their personal care pathway without delay from their initial situation to any level required to find a diagnosis and deliver care. The patient must be promptly referred further on the care pathway if a correct diagnosis or a suitable form of care cannot be found in its initial phases. The main principle is, however, striving to provide care and monitoring close to where the patient lives.

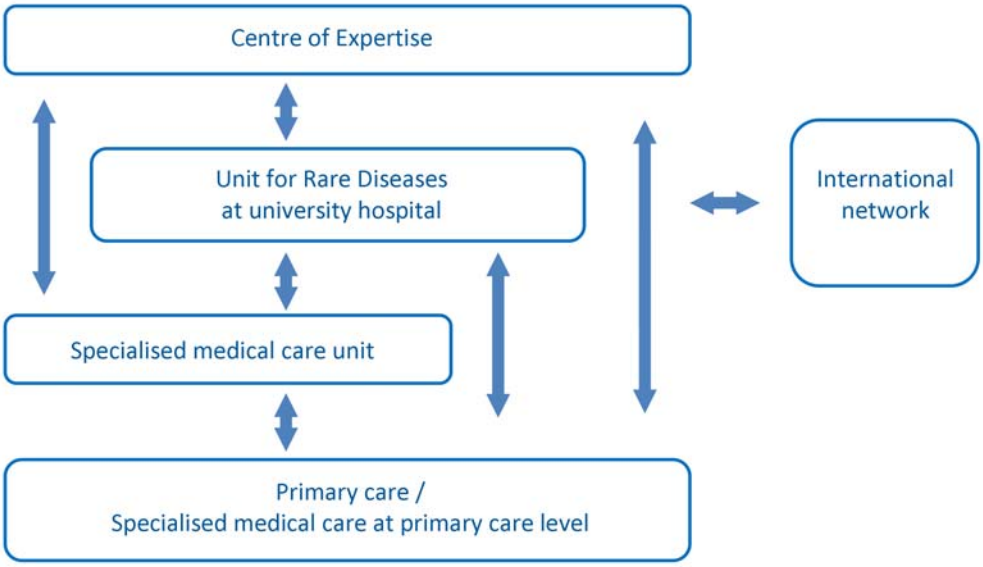

Figure 1. The care pathway of a person with a rare disease from the search for a diagnosis to care delivery and follow-up. In order to find a diagnosis, the pathway must lead to higher levels as soon as possible, and in care delivery it must promptly lead back down and close to the patient's place of residence.

Some diseases can be diagnosed in a certain unit of specialised medical care. Where the disease is so rare that its diagnostics or care are fraught with uncertainties or require multidisciplinary competence, the patient must have access to a unit for rare diseases in the university hospital of his or her catchment area and a diagnostic examination by a multidisciplinary team. This unit will also assume overall responsibility for planning the patient's care and the continuity of care when a patient transitions from paediatric to adult services, if he or she is not referred to a centre of expertise. Centres of expertise are national clusters with expertise in certain groups of diseases. A patient must have access to them if necessary. The centre of expertise may direct a patient's care and monitoring that is delivered by a unit for rare diseases, another clinic, primary health care or the third sector.

All levels of health care will engage in close cooperation with social and habilitation services to holistically address the situation of people living with a rare disease. An assessment of the patient's needs for social services, habilitation and psychosocial support should be initiated as soon as possible when the diagnosis has been found.

The centres of expertise will be part of international networks, and they will be able to draw on the expertise and resources of those networks in the diagnostics and care of a patient. Other health care units may also collaborate with international experts. In diagnostics and care delivery, the primary form of cooperation with international networks of centres of expertise is consultation but, if necessary, patients may also be referred to a European centre of expertise for assessment.

The so-called Cross-border Healthcare Directive applies to patients' rights in cross-border healthcare (2011/24/EU). The provisions of this Directive have been applied in Finland since the beginning of 2014. Under this Directive, a patient is entitled to reimbursement for the costs of care received in another member state on the same grounds as for similar care in the patient's home country. However, the patient will only receive reimbursement for care that would be reimbursed in the patient's home country. In addition, a National Contact Point for cross-border healthcare, which provides information on seeking care in other countries by Finnish residents and by foreign residents in Finland, operates in connection with the Social Insurance Institute. An effort should be made to gradually accumulate expertise in rare diseases at the National Contact Point. The Cross-border Healthcare Directive requires Finland to specify the selection of services offered by the Finnish health care system to establish which costs of care incurred abroad by a patient will be reimbursed in Finland. A new government body will be established under the auspices of the Ministry of Social Affairs and Health in spring 2014 that will be responsible for defining and continuously updating the service selection of the Finnish health care system. While this entity could issue general guidance for applying the service selection, it would not comment on decisions concerning the care of an individual patient. From the perspective of rare diseases, it should be ensured that the service selection includes new treatments of proven effectiveness which are not available in Finland but which are offered in another EU Member State. The government body to be set up to specify the service selection will consist of a council, a permanent secretariat and a network of experts. The participation of centres of expertise for rare diseases or representatives of centre of expertise networks must be ensured so that the views of the centres of expertise, for example on the development of treatments, would be taken into consideration.

An effort will be made to ensure that all actors are clear about the care pathway for people with a rare disease. The parties that are best placed to diagnose a patient's disease, plan and provide care and habilitation, and offer other support will work together. Cooperation may take the form of consultations or referrals. The patients or their data may be sent to any unit for care delivery or consultation, striving to deliver care and carry out monitoring as close to the patient's living area as possible.

The party responsible for the patient's care will make sure that a care passport in Finnish or Swedish is issued to a person with a rare disease if necessary. This passport will facilitate transfers from one care facility to another and be helpful in acute situations. The care passport will be developed as part of the existing electronic information system, or one to be developed. If necessary, the electronic care passport may be printed out and carried by the patient.

Objectives:

- The care pathway of people with a rare disease will be clarified.
- Each level of the health care system will be familiar with the organisation of diagnostics and care delivery for people with a rare disease.
- If necessary, people with a rare disease will have care passports in Finnish or Swedish.

Indicators: Health care professionals have been informed of the care pathway for people with a rare disease; a care passport in the electronic system

Responsible actor: Catchment areas: agreements on care pathways for people with a rare disease; National Institute for Health and Welfare: Care passport

Timeline: During the plan period

Prerequisites: Establishment of the units and centres of expertise for rare diseases, identification of a suitable electronic system for care passports

3.3.2 Action 6: Units for rare diseases in university hospitals

A unit for rare diseases will be set up in each university hospital. Each unit will have a coordinator and a multidisciplinary team.

People with a rare disease need an actor who will assume overall responsibility for their care. This means that patients will receive better care faster, and resources will also be saved. While plenty of resources are currently spent on the diagnostics and care of people with a rare disease, a diagnosis and an optimal care pathway are not necessarily found. A correct care pathway would save resources and speed up the initiation of appropriate care. Adults with a rare disease in particular often lack a party responsible for their care. Specialists in different fields rarely have an opportunity to get together and consider the diagnosis of patients requiring multidisciplinary competence and their care as a whole.

Units for rare diseases need to be established in university hospitals where some of the existing functions will be reorganised to achieve better outcomes. Six per cent of the population will be affected by a rare disease during their lifetime. The share of people with a rare disease is significantly higher in specialised medical care, and they account for a large group among the patients of university hospitals. The unit for rare diseases will facilitate multidisciplinary meetings to assess patients whose diagnosis is difficult to pin down and to coordinate their care. The units will assume responsibility for those paediatric and adult patients in their catchment areas who display symptoms consistent with a rare disease but for whom no diagnosis has been found in primary health care or in a specialised medical care unit. The units will also be responsible for people diagnosed with a rare disease who require multidisciplinary competence and for whom a dedicated centre of excellence cannot be set up in Finland, or who cannot be cared for elsewhere in specialised medical care units. The activities of the unit for rare diseases should cover all age groups.

Each unit for rare diseases will need a coordinator who is a designated person working in the university hospital. He or she will liaise with primary health care and serve as an information channel between various specialisations and social services. The coordinator will organise the multidisciplinary meetings to discuss rare diseases. A multidisciplinary team will operate in the unit for rare diseases. It will deal with undiagnosed patients whose symptoms are consistent with a rare disease. A selection of specialists will be invited to take part in the team as seen necessary in the case of each patient. The units should have expertise in medical genetics. The unit for rare diseases will develop flexible cooperation between various clinics and coordinate the transition of children to adult services if necessary, and the transition of patients to primary health care and social services.

The unit for rare diseases will work together with other similar units, centres of expertise, international networks and patient organisations. In addition to diagnostics and care, the unit will coordinate the habilitation and support measures related to rare diseases for which a suitable coordinating party cannot be found in the centres of expertise or in any specialist field.

If a rare disease is suspected in a patient, the threshold for consulting a unit for rare diseases or a centre of expertise should be low. Such patients are primarily diagnosed and cared for in primary health care or in a specialised medical care clinic. If necessary, however, patients in need of a diagnosis or appropriate care will be referred to a unit for rare diseases or a national centre of expertise without delay.

The special catchment areas will assess the cooperation between the university hospital, central hospital and primary health care. The unit for rare diseases will mainly be responsible for the coordination of care; patients may receive care in a clinic of a university hospital, in a central hospital or in primary health care services close to their homes, unless specialised medical care is needed. The team on rare diseases will periodically organise meetings to which parties involved in providing care for people with a rare disease will be invited from primary health care, specialised fields or units providing habilitation. The unit will promote the availability of information on rare diseases in the social welfare and health care system. Information will also be available on the website of the unit for rare diseases which will list the contact information of experts.

As far as possible, the unit for rare diseases will be structured as a sustainable part of the university hospital, ensuring that the arrangement works well for the hospital. For example, the unit may operate in connection with a medical genetics clinic, which also plays an important role in the diagnostics of rare diseases in other respects. Additional funding is needed for the unit for rare diseases, allowing the establishment of the posts of a coordinator and a coordinating nurse as well as the organisation of sufficiently frequent multidisciplinary meetings. The hospital management must ensure that the multidisciplinary team members have the possibility of taking part in meetings to discuss rare diseases.

Objectives:

- The unit for rare diseases will be responsible for the diagnostics and coordination of care in case of those patients for whom no clear specialised field exists.
- Cooperation between university hospitals will be facilitated.
- Transitions from paediatric clinics to adult services and primary health care will be smooth.
- The possibility to follow a multidisciplinary approach to diagnosis and care of rare diseases will be secured in the health care services, even if such an approach is often time-consuming.
- The availability of information on rare diseases in the social welfare and health care system will be promoted.

Indicators: Units for rare diseases established (5); Coordinators and coordinating nurses for rare diseases appointed (5 + 5)

Responsible actor: Helsinki University Central Hospital, Turku University Central Hospital, Tampere University Central Hospital, Oulu University Hospital, Kuopio University Hospital

Timeline: Planning of units for rare diseases in 2014, appointment of coordinators and coordinating nurses and unit establishment in 2015, regular operation of teams and online information activities in 2016

Prerequisites: Finding the resources to form units for rare diseases in university hospitals

3.3.3 Action 7: Centres of expertise for rare diseases

Centres of expertise for rare diseases meeting EU criteria and national centres of expertise will be established in Finland.

As the number of people affected by any single rare disease is low, it would be useful to compile information on their management. This would prevent the fragmentation of resources and competence, and each person with a rare disease would have access to the best possible care. The steering group proposes that national centres of expertise focusing on certain groups of rare diseases be established in Finland. The centres of expertise would bring cost savings by reducing unnecessary visits to doctors, periods of in-patient care and costs of malpractice, and by making patient guidance and care more efficient. They can be centres of expertise focusing on particular groups of diseases that meet EU criteria and are part of international cooperation networks, or national centres for diseases or groups of diseases with the primarily task of facilitating the sharing of expertise at the national level rather than meeting EU criteria. These centres of expertise will be designated as EU level or national centres of expertise.

Some centres of expertise already are up and running in Finland: university hospitals have special competence related to certain diseases, which other hospitals draw upon by consulting the specialists. Hover, clear cooperation and the possibility of focusing on a certain disease or group of diseases have so far been lacking. The centres of expertise may be formed in cooperation between various units of university hospitals or third-sector actors. The centres of expertise will be set up sustainably as part of the health care system structures. This way, they will depend on structural arrangements for their continuity rather than on individual specialists.

As the centre of expertise must have a specified, systematic and continuous position in the Finnish health care system, third-sector actors cannot usually form independent centres of expertise; they may, however, form centres of expertise in cooperation with hospitals. One centre of expertise will be set up for each disease or group of diseases. When a rare disease is suspected, the centre of expertise will be contacted either in the diagnostic phase or subsequently. The centre of expertise will direct the diagnostics, care, habilitation and monitoring related to the disease, but once the patient's care has been planned, responsibility for it will revert to a dedicated health care professional in the patient's own hospital district, who will liaise with the centre of expertise. Units for rare diseases in university hospitals will work closely together with the centres of expertise. The centres of expertise may also take their expertise to the patients: the experts may travel to different hospitals, rather than the patients travelling to the centres.

The network consisting of centres of expertise, units for rare diseases, various specialisations and primary health care will work together to ensure that competence is transferred to the party responsible for the patient's care. The centres of expertise will offer training to all professional groups of the health care system. The main principle of delivering care to people with a rare disease should be that patients have access to diagnoses and care regardless of where they live, information is shared effectively, and various hospitals draw on the competence of the centres of expertise. A database of the centres of expertise will be put together where social welfare and health care professionals and people with a rare disease can easily find information about expertise available in Finland in each rare disease.

Cooperation between centres of expertise and patient organisations will be vital, and determined efforts will be made to develop it. The centres of expertise will engage in regular cooperation with patient organisations whose expertise will also be utilised in the planning phase of the centres. The centre of expertise will put together a client panel in cooperation with patient organisations. The panel will comment on the general policies of the centre of

expertise and, if necessary, it may bring up issues on its own initiative. Patient organisations will also take part in periodic evaluations of the centres of expertise, and they will be actively engaged in the development of social support.

The centres of expertise currently have no legal status, and the work must thus start with an assessment of the need to amend legislation. EU level centres of expertise must meet the European Commission's criteria which are to be laid down in spring 2014. These centres of expertise must be part of the European networks of centres of expertise which play a key role in promoting diagnosis and care as expertise and patients move across the borders. The centres of expertise will not only comprise physicians and nurses, but also professionals of other fields, including physiotherapists and social workers. The centres will have competence in diagnostics, care, habilitation and social services.

A formal negotiation mechanism will be put in place between the special catchment areas and the Ministry of Social Affairs and Health to designate centres of expertise. The process of designating both national and EU level centres of expertise will begin with joint negotiations between the special catchment areas. The Ministry of Social Affairs and Health will inform hospital districts and non-governmental organizations that provide health services to people with a rare disease about the launch of the designation process and request proposals for centres of expertise. When a call for applications is launched, no groups of diseases will be named, as in the initial phase the aim will be to form centres around existing expertise. In order to register as applicants, candidates must have engaged in negotiations with fields of specialisation in the special catchment areas, ensuring that the establishment of the centre of expertise has broader support. When applying for the status of a EU level centre of expertise, a unit must also describe how it intends to meet the European Commission's criteria. The establishment of a centre of expertise may be proposed by the management of a university hospital, senior physicians in fields of specialisation, experts in rare diseases or third-sector actors as a result of a negotiation process. Each centre must cover a clear-cut group of rare diseases or, in the case of a national centre, a single rare disease. At a later stage, centres of expertise may also be established by way of invitation: if needs for a centre of expertise for a certain group of diseases emerges, the Ministry of Social Affairs and Health may declare an application round for the centre in question open and launch negotiations with units that could serve in this role.

Centres of expertise will be designated for a term that is long enough to allow for long-term financial planning and enable the centre to develop. This time period will be specified in compliance with the forthcoming EU legal act. When centres of expertise are designated based on application, representatives of specialist fields in all hospital districts and the relevant patient organisation will be consulted. The designation of centres of expertise will be drafted by the national coordinating centre for rare diseases. The university hospitals and patient organisations will be represented in the national centre's board. The centres of expertise will be designated by the Ministry of Social Affairs and Health.

Objectives:

- Examining any legislative amendments required to establish centres of expertise.
- Initiating the establishment of centres of expertise for rare diseases during the plan period in Finland.
- Some of the centres will have strategies for meeting the EU criteria.
- All levels of health care have been informed about the centres of excellence for rare diseases. The centres of expertise are sharing their expertise with all levels of the social welfare and health care system.

Indicators: Centres of expertise for rare diseases have been established; Database on the centres of expertise has been created; Number of referrals to centres of expertise

Responsible actor: Ministry of Social Affairs and Health, hospital districts and university hospitals

Timeline: Examination of the need for legislative amendments in 2014, more detailed planning and drafting of centres of expertise in 2014, round of applications and negotiations in 2015

Prerequisites: Putting the required legislation in place, financial resources for establishing the centres of expertise

3.3.4 Action 8: Providing more education and training

Social welfare and health care professionals will be provided with more education and training in the field of rare diseases.

According to a study (2009) conducted by EURORDIS, the European umbrella organisation for patients with a rare disease, taking the possibility of a rare disease into consideration speeds up the correct diagnosis of a rare disease. A key method for spreading awareness among physicians who diagnose rare diseases and other health care personnel is providing them with more education and training in the field of rare diseases. This education and training should be offered as part of both initial and continuing education.

In basic medical training, content related to rare diseases could be partly integrated in medical genetics studies, and many educational institutions already do so. Due to a lack of personnel resources, a comprehensive course in medical genetics is not included in the syllabi of medical faculties in all universities.

The steering group proposes that a concept focusing on rare diseases be added to the education programme of medical students and students of other social welfare and health care fields. This concept would direct the students in finding out what types of rare diseases there are, how to suspect a rare disease rather than a more common one, how to deal with a rare disease, where the patients can be directed and how significant thinking of the possibility of a rare disease is in terms of a prompt diagnosis. To make sure that professionals can encounter people with a rare disease holistically and as indicated by the patient's situation, teaching about the multi-channel habilitation system will be included in health care and social welfare qualifications in all fields.

Continuing education related to rare diseases should be organised for all fields of social welfare and health care. The units and centres of expertise for rare diseases will provide training for different levels of the health care system and for professionals of habilitation and social services. The Finnish Society of Medical Genetics has already decided to put together a training package intended for professionals working in primary health care and specialised medical care.

The existing service user trainer networks should be exploited more extensively in education. In Eastern Finland, service user training has already been fully integrated in the education, allowing students to benefit from first-hand knowledge about rare diseases as part of their theoretical studies. The service user trainers receive careful coaching, and the only costs incurred by the educational institution are the trainer's travel costs.

Objectives:

- Knowledge of rare diseases among social welfare and health care sector professionals will be built up.

- Health care, social welfare and habilitation professionals who deal with rare diseases will have up-to-date information about rare diseases.

Indicators: Emphasis on rare diseases in the education and training of students in the social welfare and health care sector; Training events related to rare diseases organised for social welfare and health care personnel

Responsible actor: Ministry of Education and Culture, universities and polytechnics: Emphasis on rare diseases in the education and training of students in the social welfare and health care sector; Units and centres of expertise for rare diseases: Training courses intended for social welfare and health care professionals

Timeline: Preparing a study concept focusing on rare diseases to be included in qualifications in the social welfare and health care sector in 2014−2015; annual training events from 2014

Prerequisites: Universities' and polytechnics' approval for modifying education programmes

3.3.5 Action 9: Promoting the availability of orphan drugs

The availability and reimbursement of drugs and medicinal products used to treat rare diseases will be promoted.

As the patient numbers are low and the development of new drugs is expensive, the pharmaceutical industry is unwilling to invest in the research and development of orphan drugs. Dedicated support measures are thus needed. Due to the small markets, pharmacotherapy used to treat rare diseases usually is more expensive than that of more common diseases. People with a rare disease experience long-term and often lifelong needs for medication. As people with a rare disease are entitled to the same quality of care as other patients, the authorities in various countries have developed incentives for the biotechnology and pharmaceutical industry in order to support the research, development and marketing of orphan drugs.

The Regulation on orphan medicinal products adopted in 1999 lays down the EU policy on orphan drugs in the Member States. Under this regulation, a medicinal product shall be designated as an orphan medicinal product if it is intended for a condition affecting not more than five in 10 thousand persons in the Community. The condition must be life-threatening or chronically debilitating. If the condition is less severe than this, a product may be designated as an orphan medicinal product if it is unlikely that the marketing of the medicinal product in the Community would generate sufficient return to justify the necessary investment. Another condition is that there exists no other method of treatment of the condition in question or that the product will be of significant benefit to those affected by that condition.

Finland is committed to the incentive system for developing orphan drugs. The European Commission may grant an exclusive marketing authorisation for an orphan drug for ten years. During this time, generic drugs will not be granted a marketing authorisation unless they are significantly more effective. While the marketing authorisation is granted at the European level, eligibility for reimbursement must be applied for separately in each country. In Finland, a drug must be eligible for basic reimbursement before special reimbursement can be paid. Its therapeutic value, necessity and cost-effectiveness in use must be proven. Eligibility for special reimbursement may be granted for medicinal products that are used to treat severe and

long-term illnesses listed in a Government Decree. Another criterion for eligibility for special reimbursement of a medicinal product is that the wholesale price proposed for it is reasonable. When decisions on eligibility for special reimbursement are being made, the funds available for special reimbursements will be taken into account.

The availability of orphan drugs is reasonably good in Finland, and their continued availability should be safeguarded. The EU Directive on patients' rights in cross-border healthcare includes mutual recognition of prescriptions; in other words, holders of a European prescription issued in Finland can purchase drugs in other European countries. Prescriptions written in another EU Member State will be filled if the drug has a marketing authorisation in the Member State where it is to be purchased.

Almost all orphan drugs available in Finland that are used in outpatient care are eligible for reimbursement. As orphan drugs are expensive, they should also qualify for reimbursement in the future. Due to ambiguities in funding responsibilities, in certain situations an individual patient may end up paying significant amounts for their drugs. According to a working group preparing an overall reform of the drugs reimbursement system, funding responsibilities related to orphan drugs and their reimbursement from the health insurance should be examined in detail (Final report of the working group on developing the drugs reimbursement system, Reports and memorandums of the Ministry of Social Affairs and Health 2012:33).

One aspect of this problem is drugs intended for the treatment of common illnesses where research-based evidence of a high quality indicates that the drugs are effective in the treatment of rare diseases but they are not eligible for reimbursement when used to treat a rare disease. The extent of this problem and the factors behind it should be studied. The steering group proposes that if necessary, the Ministry of Social Affairs and Health should act with initiative both in the national and the international context and promote the extension of therapeutic indications of drugs in the market to also include those rare diseases in whose treatment the drug is effective in the light of scientific evidence.

If the drug does not have marketing authorisation in Finland, the Finnish Medicines Agency may on certain conditions grant a special authorisation to a medicinal product for the treatment of an individual patient or a patient group for a maximum period of one year. Eligibility for reimbursement for a medicinal product delivered under a special authorisation may be applied for by an individual user, a pharmacy on behalf of a patient, a wholesaler, or a pharmaceutical company. Physicians, as well as people with a rare disease and their families, need more information and advice about the possibility of applying for a special authorisation and eligibility for reimbursement for a medicinal product delivered under a special authorisation.

The availability and reimbursement of costs of not only medicinal products but also nutritional products are fraught with problems. A Government decree lists the severe diseases where either 35% or 65% of the costs incurred for therapeutically used clinical nutritional products are reimbursed to the patients. For example, this decree does not cover severe rare epilepsies treated with nutritional products. An effort should thus be made to include in the decree all severe illnesses where research-based evidence exists to support treatment with clinical nutritional products. Patients with severe nutrient malabsorption only qualify for the lower rate of reimbursement at 35%. The steering group proposes that nutritional products used to treat these conditions be made eligible for reimbursement based on the higher rate. While nutritional and similar products are a lifeline for some people with a rare disease, not all products are yet covered by the reimbursement scheme.

Objectives:

- Ensuring the availability of orphan drugs.
- Carrying out a survey of drugs used to treat rare diseases.

- If necessary, promoting the extension of therapeutic indications of drugs already on the market to rare diseases for which there is scientific evidence of a drug's therapeutic effectiveness.
- Including all severe illnesses where there is scientific evidence supporting treatment with clinical nutritional products in the decree on the eligibility for reimbursement of nutritional products.
- Reimbursing 65% of the costs incurred for clinical nutritional products used to treat severe malabsorption.
- Spreading awareness of the possibility of applying for eligibility for reimbursement in case of medicinal products supplied under a special authorisation.

Indicators: The number and share of orphan drugs eligible for reimbursement in all orphan drugs used in outpatient care; Awareness of the possibility of applying for eligibility for reimbursement in case of a medicinal product supplied under a special authorisation (patient survey)

Responsible actor: Ministry of Social Affairs and Health: Amendments to the Health Insurance Act, spreading information about the application procedure for eligibility for reimbursement in case of medicinal products supplied under a special authorisation; Social Insurance Institution: A survey of drugs used to treat rare diseases

Timeline: Legislative amendments in force in 2017

Prerequisites: Amendments in legislation on drugs reimbursement

3.4 AREA 4: COORDINATION OF EXPERTISE AND KNOWLEDGE SHARING

One of the key problems associated with rare diseases is the sporadic nature of knowledge and expertise. Health care professionals and people with a rare disease, to say nothing about social welfare actors, cannot access the knowledge and expertise that is available, as it has not been collected in one location. There also are gaps in knowledge about rare diseases in many areas, which delays diagnosis and access to appropriate care.

An actor to assume responsibility for coordinating rare diseases issues is lacking in Finland. In addition to collecting and spreading information, an actor of this nature would be needed to administrate the dedicated measures required by rare diseases and to monitor their implementation, to create links between competence in rare diseases at different levels of the health care system, to serve as a forum for dialogue between various stakeholders and to maintain international cooperation networks.

3.4.1 Action 10: A national coordinating centre for rare diseases

A national coordinating centre for rare diseases will be set up.

A national coordinating centre for rare diseases will be set up in Finland to serve as an administrative link between primary health care, different fields of specialised medical care, units for rare diseases, centres of expertise, third-sector actors, the Social Insurance Institution, social services, and other EU programmes and centres for rare diseases. It would be appropriate to establish this centre under the auspices of the National Institute for Health and Welfare. Its board should include representatives of the health care and social welfare systems, centres of expertise, the Social Insurance Institution, the third sector and patient organisations. The centre will convene a national forum on rare diseases that will meet as necessary but at least once a year and comment on topical issues in the field of rare diseases. The national centre's work would be based on the forum's opinions. The national forum should bring together all stakeholders in the field of rare diseases across a very broad spectrum. The centre will need a coordinator and an expert.

The centre's key task will be developing the service system to meet the needs of people with a rare disease as effectively as possible and as set out in the National Plan for Rare Diseases. The national centre will see to the division of responsibilities for the implementation of National Plan actions and be in charge of the progress, monitoring and evaluation of the actions and the setting of new objectives. The national coordinating centre for rare diseases will be responsible for drafting the designation of centres of expertise: it will coordinate the process aiming to establish centres of expertise and make proposals for the designation of centres to the Ministry of Social Affairs and Health.

Other tasks of the national centre will include developing independent and extensive education and training, coordinating the preparation of clinical care recommendations for rare diseases, and coordinating expertise and knowledge in cooperation with centres of expertise, units for rare diseases and NGOs. It will also coordinate international research cooperation. The national coordinating centre for rare diseases will be a member of European cooperation organs and pass on information about international expert forums.

As the National Institute for Health and Welfare will be faced with significant cuts in its resources, setting up the centre within the Institute is not possible at this stage, and its establishment has to be postponed to a later date. The steering group recommends that a smaller informal group be set up to implement the programme that would meet a few times in 2014. This group should contain representatives from the Ministry of Social Affairs and Health, the National Institute for Health and Welfare, the Social Insurance Institution, all university hospitals, the Association of Finnish Local and Regional Authorities, the Finnish Network for Rare Diseases, Orphanet and the HARSO network. At the end of 2014, the possibilities of establishing a more permanent national coordination structure would be examined. The steering group further proposes negotiations between university hospitals to establish which one of them could assume responsibility for coordinating clinical activities in the field of rare diseases on a temporary basis.

Objectives:

- Information on Finnish expertise in rare diseases will be collected systematically and in a coordinated fashion.
- A national centre will be established that will assume responsibility for good governance and planning in the field of rare diseases.
- A national forum on rare diseases will be set up, and it will meet regularly.

Indicators: Decision to establish a national coordinating centre for rare diseases; Recruitment of a coordinator and a specialist; Compilation of a database containing information relevant to rare diseases

Responsible actor: Ministry of Social Affairs and Health, National Institute for Health and Welfare, university hospitals

Timeline: A centre coordinating rare diseases and a national forum set up to coordinate plan implementation from 2015

Prerequisites: Financial resources for establishing the centre

3.4.2 Action 11: Systematic collection and sharing of information

A comprehensive online database of rare diseases in Finnish and Swedish will be created. Information will be provided to people with a rare disease, their families and social welfare and health care professionals by a dedicated helpline. A yearly seminar on rare diseases will be organised in cooperation with patient organisations.

Plenty of information on rare diseases is already available on the Internet, but this information is to some extent scattered and there are gaps in it. There also is a number of rare diseases on which no information can be found in Finnish or Swedish. The national coordinating centre for rare diseases will be responsible for the systematic collection and publication of information on the Internet. It will organise the collection of information on units and centres of expertise for rare diseases, and from people with a rare disease and patient organisations. The national centre will also collect information about care, habilitation and social services in cooperation with patient organisations. Patient organisations are often important sources of information for people with a rare disease and their families, a fact that should be taken into account in the dissemination of information.

A comprehensive Finnish online database on rare diseases can be created by translating into Finnish and Swedish the contents of Orphanet, the existing online service for rare diseases and orphan drugs, currently maintained by the Norio Centre. Orphanet is a cooperation project in which some forty countries take part. The Orphanet team in each country collects information on specialist clinics, medical laboratories, research projects and patient organisations in their own country. This information is accessible to everyone free of charge. When creating the database, Orphanet should be taken into consideration to avoid overlapping work. The national coordinating centre for rare diseases and the Ministry of Social Affairs and Health will agree upon database implementation and cooperation with the third sector. The database should be regularly updated, and all those working with rare diseases should be informed about its existence.

In addition to an electronic database, general advice on rare diseases and expertise in them available in Finland will be needed for both citizens and social welfare and health care actors. Currently, this type of service is provided by third-sector actors through helplines and electronic query services, which could also be a good operating model in the future. Another possibility is that the special catchment areas agree to set up a general helpline at a certain unit for rare diseases.

The national centre will assume responsibility for organising an annual seminar. This seminar will be organised in connection with the national forum, and it will discuss topical

issues in the field of rare diseases. All parties to whom rare diseases are relevant will be invited to the seminar on rare diseases.

Objectives:

- Information related to rare diseases, including information on research, health care and social services and support needed by people with a rare disease, will be collected and offered systematically.
- The activities of the Finnish Orphanet will be accounted for when creating a Finnish and Swedish database on rare diseases.
- Actors in the field of rare diseases will meet every year at an annual seminar.

Indicators: A database in Finnish and Swedish exist; Orphanet continues to be updated; Annual seminar on rare diseases; People with a rare disease can find the information they need (patient survey)

Responsible actor: National coordinating centre for rare diseases

Timeline: Continuously, starting from the beginning of the plan period in 2014

Prerequisites: No obstacles

3.5 AREA 5: OFFERING HOLISTIC SUPPORT TO AND EMPOWERMENT OF PEOPLE WITH A RARE DISEASE

Once a person with a rare disease has received a correct diagnosis and even earlier if necessary, suitable health and social services will be sought for him or her, and habilitation will be initiated together with the client and, where necessary, his or her family. The problem often lies in the lack of a holistic approach to improving the quality of life of a person with a rare disease. People with a rare disease and their families may also encounter many types of challenges as users of ordinary services, for example at a maternity clinic or in a day-care centre or an educational institution. Instead of an approach based on the diagnosis, the evaluation of service needs should address the special needs of the individual patient. The objective should be that each person can live a full life of his or her own choosing regardless of a rare disease, and receive not only appropriate care and habilitation but also the psychosocial support indicated by his or her needs.

Social services do not always fully meet the needs of people with a rare disease: information provided about the available social services may be inadequate, and a large share of people with a rare disease and their families are dissatisfied with the quality of social services. There also are regional differences in the availability of social support. Information about social security and welfare services is fragmented, the authorities may have little or no experience of rare diseases, and no big picture exists of the social services' capabilities for meeting the needs of people with a rare disease and their families. Because of these problems, research in the effectiveness of services in the field of rare diseases is also needed.

Increasing the clients' inclusion and participation in their own lives and supporting their coping with everyday life are key objectives in the development of social welfare and health care legislation and service system. The provision of social services and support start from the client's needs, regardless of the underlying diagnosis.

The habilitation system is complex and fragmented. Habilitation is organised by the health care system, the social services, the education services, the Social Insurance Institution and the labour administration, and each actor defines their target groups and objectives differently. Rare diseases are often diagnosed already in childhood. During their lives and in various phases of their disease, patients may need habilitation offered by several different actors. Extensive cooperation is needed in the planning of care and habilitation to ensure timely habilitation that would lead into the client's optimal functional capacity and work ability.

Directing people with a rare disease to timely habilitation services indicated by their needs is difficult, and cooperation between the care facility, the Social Insurance Institution and the habilitation service provider is not always adequate. Competences in different fields do not always meet: health care personnel have inadequate knowledge about and competence in habilitation, and information about the Social Insurance Institution's habilitation services is not always successfully communicated to the health care services. On the other hand, the health care system's knowledge about habilitation needs related to rare diseases and new forms of treatment and habilitation do not always reach those in charge of planning the Social Insurance Institution's habilitation services on a sufficiently broad front. The Social Insurance Institution does not have consistent information about the care facilities, and thus correct actors are not always consulted.

People with a rare disease or their families often are themselves highly knowledgeable about their situation, and they know what kind of services the person with a rare disease needs. Many patient organisations also have plenty of knowledge about the experiences and needs of people with a rare disease. However, they are not always adequately taken into consideration in the planning of social and health services. Availability of psychosocial support to people with a rare disease and their families and stronger participation of patients and families in the planning of care and services would improve the holistic implementation of care and support.

The client and patient organisations offering peer support and other psychosocial support receive funding intended for NGOs for the development of these activities from such funding providers as Finland’s Slot Machine Association (RAY).

3.5.1 Action 12: Developing social support and habilitation

More information will be disseminated about social and habilitation services. Centres of expertise, units for rare diseases, NGOs, the Social Insurance Institution and municipal social services will work together in closer cooperation in order to anticipate the habilitation needs of a person with a rare disease, to plan services and to address the need for holistic support of the person with a rare disease. People with a rare disease and, if necessary, their families will take part in the planning of care, habilitation and social services.

Cooperation during the habilitation period of a person with a rare disease will be taken into consideration in the work of units and centres of expertise for rare diseases to ensure that physical and psychological habilitation alike optimally support the care and overall objectives of the person with a rare disease. For example, this will enable network-based negotiations between the rehabilitee, the care facility, the habilitation service provider and social services. Liaising with parties providing habilitation services, the Social Insurance Institution, social services and NGOs will be made part of the operating model of the centres of expertise when planning the care, habilitation and social support of a person with a rare disease. Assessment of the possibilities offered by habilitation that the Social Insurance Institution is responsible for providing will be included in the operating model of units for rare diseases. In case of people with a rare disease requiring multidisciplinary competence and extensive support, the

duty of drawing up a care and habilitation plan as indicated by the client's needs and liaising with social services will be assigned to a dedicated person.

Rehabilitation is often demanding, and treatments are advancing constantly. Without information exchange, habilitation cannot be provided as indicated by the client's needs, and people with a rare disease will not have access to habilitation measures indicated by their needs and guidance that enables habilitation. Centres of expertise and units for rare diseases will serve as coordinating units for the planning of care and habilitation together with health care services, the habilitation team of the Social Insurance Institution and social services. Primary health care will also strive to appoint an actor to coordinate habilitation. The actor delivering care to a person with a rare disease will inform the municipality and the person him/herself about aspects that need to be taken into consideration in the habilitation process. Closer cooperation between various parties is vital, so that education and training can be targeted at those making decisions about habilitation and social welfare and health care personnel working with people affected by a rare disease, and people with a rare disease can be directed to suitable habilitation services and appropriate type of assistance. People with a rare disease should themselves have the possibility of participating in their care and service process and influencing it in all of its phases, as well as being informed about it by the actors involved in the process.

Information about the social security and service system and habilitation should be distributed more effectively. The units and centres of expertise for rare diseases should have data banks on habilitation and social services where other health care units can find information. Information about the social security system and habilitation is provided by habilitation advisers and social workers at hospitals and the municipal social services, especially services for the disabled. Support for client work in the field of services for the disabled and information for clients is available from the electronic Handbook on Disability Services, which is part of the National Institute for Health and Welfare's web service. In 2013, a separate section on services for children and young people was launched. Rare diseases should also be included as a separate section in the handbook. National health and patient organisations and organisations for people with disabilities have joined their forces in preparing a guide to social security for persons with long-term illnesses and disabilities. This guide supports NGO employees who provide advice and guidance to their members. The national coordinating centre for rare diseases will collect existing information in a single location and share it with all levels of the social welfare and health care system.

When developing cooperation and information about habilitation and social services in the field of rare diseases, interfaces with the on-going projects of the Ministry of Social Affairs and Health aiming to reform social and health care service structures, the Social Welfare Act and legislation on services for the disabled and to develop informal care and multidisciplinary habilitation will be taken into consideration. It has been proposed that under the new Social Welfare Act, a dedicated employee could be assigned to those clients who need several social services. This operating model would promote the planning of service packages so that the individual needs of people with a rare disease could be addressed better. The possible need for support of family members assisting a person with a rare illness should also be investigated.

Peer support offered by patient organisations to people with a rare disease is extremely important. However, Finland's Slot Machine Association no longer offers sufficient funding for patient organisations. RAY grants to patient organisations should thus be increased to provide NGOs with better possibilities of offering peer support.

Objectives:

- There will be closer and more systematic cooperation between health, social and habilitation services so that the needs of a person with a rare disease can be addressed holistically.

- People with a rare disease and, if necessary, their families will take part in the planning of service packages, and they will be able to exert influence in the way the services are arranged.
- A service plan will be drawn up for people with a rare disease requiring multidisciplinary competence.
- Information about timely habilitation and social services will be easily available, and it will be distributed to social welfare and health care professionals and people with a rare disease.
- New information about rare diseases will be disseminated to the Social Insurance Institution, patient organisations, insurance companies and social and health service organisers and providers.
- Patient organisations' possibilities of organising peer activities will be supported.

Indicators: Liaising between health, social and habilitation services when planning the care and habilitation of people with a rare disease (reports from centres of expertise); Service plans drawn up (patient survey); Data bank of social and habilitation services compiled; Trends in RAY grants to patient organisations

Responsible actor: The national coordinating centre for rare diseases, units for rare diseases, centres of expertise, the Social Insurance Institution, municipal social services; the National Institute for Health and Welfare: A section on rare diseases in the Handbook on Disability Services

Timeline: After the establishment of the national centre, units for rare diseases and centres of expertise from 2016 on

Prerequisites: Appointment of a coordinating party; cooperation to complement the Handbook on Disability Services

3.5.2 Action 13: Empowerment of people with a rare disease

People with a rare disease, their families and patient organisations will have better opportunities to participate in decision making and in the planning of services in the field of rare diseases.

In the European Commission Recommendation (2009), it is stated that patients and patient organisations should be consulted on policies in the field of rare diseases. According to the quality criteria for centres of expertise formulated by EUCERD (European Union Committee of Experts on Rare Diseases), the centres of expertise collaborate with patient organisations to bring in the patients' perspective (Appendix 2). The empowerment of patients and their families and their participation in planning services should be promoted, and patient organisations' possibilities of exerting influence should be improved at various levels. This should also be the point of departure for developing the social welfare and health care legislation and service system.

Individual persons with a rare disease and their family members should be provided with adequate information about care, habilitation and social and support services. This role of low-threshold information centres will be assumed by the centres of expertise and units for rare diseases. The information will have been collected in cooperation with patient organisations

and the entire health, social and habilitation service network. It will cover all these areas extensively. Information will also be available electronically from a data bank collected by the centre and through helplines. Centre of expertise specialists will visit peer support groups for people with a rare disease to distribute and receive information. A reciprocal attitude is important to ensure that people with a rare disease and their families have sufficient possibilities for participating in developing care, habilitation and services. In other words, people with a rare disease and their families must be involved in producing experiential knowledge. This knowledge will also be drawn upon in teaching modules on rare diseases intended for health care professionals.

Patient organisations are a key channel of influence for people with a rare disease and their families. They are federations that produce many types of services. Their members include associations for rare diseases and other patient organisations. Their role in preparing, developing and evaluating services is not adequate, as there is no clear model for their participation. Currently, adequate information about the effectiveness of cooperation with patient organisations at the level of university hospitals is lacking, and the first step would thus be studying this area.

Patient organisation participation should be realised at three different levels:

1) When centres of expertise are being set up, NGO representatives will be involved in the planning processes.
2) Client panels, in which NGOs are involved, will evaluate the operation of centres of expertise and propose improvements.
3) The forum for rare diseases maintained by the national coordinating centre for rare diseases will have broad-based representation of patient organisations. The board of the national centre will also have adequate numbers of NGO representatives (from several organisations, as the patients' situations vary according to the diagnosis group).

In addition, the roles of the National Council on Disability and the Advisory Board for Rehabilitation in drafting issues related to rare diseases should be clarified to avoid overlapping efforts and to ensure that all possible expertise can be harnessed in the planning of service pathways and centre of expertise operation.

Objectives:

- People with a rare disease, their families and professionals will be provided with adequate information about social and health care services.
- People with a rare disease and their families will have more involvement in the planning of service pathways.
- Patient organisations will have better possibilities of exerting influence on the development of service structures.
- Patient organisations will be involved in launching, evaluating and developing the operation of centres of expertise.
- Patient organisations will participate in evaluating and developing services in the field of rare diseases.

Indicators: Information obtained by people with a rare disease and their families about existing social and health services (patient survey); Participation of patient organisation representatives in the working group for planning the centres of expertise, the national forum for rare diseases, the board of the national coordinating centre for rare diseases and the centre of expertise client panels.

Responsible actors: National coordinating centre for rare diseases: Coordination of information activities, establishment of the national forum and the board of the national centre, preparation and designation of centres of expertise; Centres of expertise: customer panel

Timeline: 2015−2017

Prerequisites: No obstacles; regarding centres of expertise, the establishment of the centres

4 NATIONAL PLAN COORDINATION, MONITORING AND EVALUATION

The first action to be implemented under the Finnish National Plan for Rare Diseases is establishing a national coordinating centre for rare diseases. The steering group proposes that this should go ahead at the very beginning of the plan period, as the centre will coordinate the implementation of other National Plan actions. In order to establish this centre, the Ministry of Social Affairs and Health must take action and authorise the centre to coordinate National Plan implementation. As other priority measures of the programme, the steering group proposes acknowledging the special measures required by rare diseases in legislation, clarifying the care pathway for people with a rare disease, establishing units for rare diseases in university hospitals, promoting the availability and reimbursement of orphan drugs, and developing social support and habilitation.

The operation of the national centre will be influenced by the national forum for rare diseases, in which university hospitals, patient organisations, municipal social services and the Social Insurance Institution will be represented. The forum will facilitate dialogue between stakeholders and direct and evaluate National Plan implementation. The activities of the forum will be organised by the national centre. The forum will meet as necessary, however no less than once a year, in connection with the seminar on rare diseases.

The national centre will coordinate and monitor National Plan implementation, observing the timeline proposed for each action. In the event that it proves impossible to establish the national centre at the beginning of the plan period, the Ministry of Social Affairs and Health should appoint a temporary steering group to manage plan coordination. In 2014−2017, the National Plan for Rare Diseases will progress as follows:

Year	National Plan for Rare diseases
2014	Planning of National Plan implementation and capacity building
2015	Plan implementation and mid-term evaluation
2016	Plan implementation
2017	Final evaluation of the plan and planning of a new period

Indicators for each action and key indicators drawn up by EUCERD will be used to evaluate National Plan implementation. In addition to EUCERD criteria, national indicators will be needed to facilitate monitoring and to gauge the effectiveness of planned actions. Developing these indicators will be one of the national centre's tasks. As there is little information available on the current status in many areas, assessing the impact of actions will be difficult. Consequently, the national centre should use any available means to analyse the current status of rare disease issues. Key themes in developing indicators will be feedback from professionals and patient organisations, delays in finding a diagnosis, changes in care practices, groups that fall through the cracks, integrity of care pathways and ensuring continuity of expertise. The satisfaction levels of people with a rare disease and their families should be surveyed at regular intervals.

The steering group proposes that a mid-term evaluation be carried out by the national coordinating centre for rare diseases in 2015 using EUCERD's key indicators and the indicators specified for each action. In conjunction with the mid-term evaluation, the appropriateness of the selected indicators will be assessed, and the indicators will be developed further. At the end of the plan period in 2017, it would be justified to conduct an external evaluation which will be taken into account when setting the goals for the forthcoming plan period.

5 SUMMARY OF ACTIONS

	Action	Key content	Objectives	Obstacles / priorities
1	Consistent definition of rare diseases and need for dedicated measures	Adoption of EU definition of rare diseases; Acknowledg-ment of the need for special measures in legislation	Increasing EU level cooperation; Ensuring equal access to quality services	Political support **Priority**
2	Registry of rare diseases	Creating a registry of rare diseases in Care Register for Health Care (Hilmo) based on the ICD-11 classification	Facilitating the collection of research-based data; Increased cooperation both nationally and internationally	Introduction of the ICD-11 classification
3	Research programme for rare diseases	A share of national research funding allocated to research in rare diseases and the Finnish disease heritage; Submission to the Academy of Finland of a proposal for a multidisciplinary research programme on rare diseases	Reinforcing research funding; Creating a network for scientific cooperation and a research database; Promoting research on service quality and on the impact and effectiveness of the service system	Specifying the responsible actor
4	Reinforcing international scientific cooperation	Applying for membership of the E-RARE project	Collection of sufficiently large patient data sets for clinical studies; Adequate research funding that enables participation in E-RARE	Adequate national funding
5	Clarification of the care pathway for people with a rare disease	Streamlining the pathway aiming for the diagnosis and care of people with a rare disease, and dissemination of information on this; Development of a care passport	Accelerate diagnosis and care delivery to people with a rare disease; Health care services at all levels will know how diagnostics and care are implemented; Care passports will facilitate the transfer of patients from one care facility to another and be useful in acute situations	Establishment of units and centres of expertise for rare diseases; An electronic system for care passports **Priority**
6	Setting up of units for rare diseases in university hospitals	A unit for rare diseases will be set up in each university hospital with a rare diseases coordinator, a nurse and a multidisciplinary team	Coordination of care and habilitation of patients requiring multidisciplinary competence; Horizontal cooperation will be facilitated; Smooth transition from paediatric to adult services; Safeguarding the possibility of working to improve diagnostics and care in the field of rare diseases	Resource allocation in university hospitals **Priority**

	Action	Key content	Objectives	Obstacles / priorities
7	Establishing centres of expertise	Centres of expertise for rare diseases meeting EU criteria and national centres of expertise will be established in Finland	Centres of expertise share their expertise with all levels of the social welfare and health care system; Some centres have strategies for meeting the EU criteria	Financial resources
8	Education and training of professionals	Social welfare and health care professionals will be provided with more education and training related to rare diseases	Professionals dealing with rare diseases have up-to-date knowledge about rare diseases	Universities' and polytechnics' approval for modification of education programmes
9	Promoting the availability of orphan drugs	The availability and reimbursement of drugs used to treat rare diseases will be promoted	Availability of drugs will be safeguarded; Drugs eligible for reimbursement when used to treat a rare disease; Increased awareness of the reimbursement system	Legislative amendments concerning reimbursements **Priority**
10	National coordinating centre for rare diseases	A national coordinating centre for rare diseases will be set up	Good governance and planning in the field of rare diseases; Systematic collection of information on expertise in rare diseases in Finland; A forum for rare diseases meets regularly	Financial resources **Priority**
11	Collection and sharing of information	A comprehensive database on rare diseases will be created; Information will be offered through a helpline; A seminar on rare diseases will be organised annually	Information on rare diseases related to research, health care and social services and the support needed by people with a rare disease will be collected and offered systematically; Orphanet activities will be taken into account; Actors in the field of rare diseases will meet annually at a dedicated seminar	No obstacles
12	Development of social support and habilitation	More information will be distributed about the services; Increasing the cooperation between various actors	Closer cooperation between health care, social and habilitation services; Service plans for those requiring multidisciplinary competence; Availability of up-to-date information about habilitation and social services	Appointment of a coordinating body **Priority**

	Action	Key content	Objectives	Obstacles / priorities
13	Empowerment of people with a rare disease	People with a rare disease and patient organisations participate in decision-making in the field of rare diseases and the planning of services	Provision of adequate information about the services; Participation of people with a rare disease in planning service pathways; Patient organisations are able to influence the development of service structures; Patient associations will participate in the evaluation and development of services related to rare diseases	No obstacles

ABBREVIATIONS

ERC	European Research Council
EU	European Union
EUCERD	European Union Committee of Experts on Rare Diseases
EUROPLAN	European Project for Rare Diseases National Plans Development
EURORDIS	European Organisation for Rare Diseases
Evo	Special central government transfer
Hilmo	Care Register for Health Care
HUS	Hospital District of Helsinki and Uusimaa
HUCH	Helsinki University Central Hospital
ICD	World Health Organisation classification system (International Statistical Classification of Diseases)
IRDiRC	International Rare Diseases Research Consortium
Kela	Social Insurance Institution of Finland
KYS	Kuopio University Hospital
OKM	Ministry of Education and Culture
OYS	Oulu University Hospital
RAY	Finland's Slot Machine Association
STM	Ministry of Social Affair and Health
TAYS	Tampere University Hospital
THL	National Institute for Health and Welfare
TYKS	Turku University Central Hospital

APPENDIX 1

COUNCIL OF THE EUROPEAN UNION RECOMMENDATION ON AN ACTION IN THE FIELD OF RARE DISEASES AND HOW IT IS TAKEN INTO ACCOUNT IN THE NATIONAL PLAN FOR RARE DISEASES

COUNCIL RECOMMENDATION of 8 June 2009 on an action in the field of rare diseases (2009/C 151/02)

THE COUNCIL OF THE EUROPEAN UNION RECOMMENDS THAT MEMBER STATES:

I

PLANS AND STRATEGIES IN THE FIELD OF RARE DISEASES

1. Establish and implement plans or strategies for rare diseases at the appropriate level or explore appropriate measures for rare diseases in other public health strategies, in order to aim to ensure that patients with rare diseases have access to high-quality care, including diagnostics, treatments, habilitation for those living with the disease and, if possible, effective orphan drugs, and in particular:

(a) elaborate and adopt a plan or strategy as soon as possible, preferably by the end of 2013 at the latest, aimed at guiding and structuring relevant actions in the field of rare diseases within the framework of their health and social systems;

⇨ Entire plan

(b) take action to integrate current and future initiatives at local, regional and national levels into their plans or strategies for a comprehensive approach;

⇨ Action 10: A national coordinating centre for rare diseases

(c) define a limited number of priority actions within their plans or strategies, with objectives and follow-up mechanisms;

⇨ Entire plan; priority actions are highlighted in the abstract and the Summary of Actions

(d) take note of the development of guidelines and recommendations for the elaboration of national action for rare diseases by relevant authorities at national level in the framework of the ongoing European project for rare diseases national plans development (EUROPLAN) selected for funding over the period 2008-2011 in the first programme of Community action in the field of public health.

⇨ Entire plan

II

ADEQUATE DEFINITION, CODIFICATION AND INVENTORYING OF RARE DISEASES

2. Use for the purposes of Community-level policy work a common definition of rare disease as a disease affecting no more than 5 per 10 000 persons.

⇨ Action 1: Adopting a uniform definition of rare diseases

3. Aim to ensure that rare diseases are adequately coded and traceable in all health information systems, encouraging an adequate recognition of the disease in the national healthcare and reimbursement systems based on the ICD while respecting national procedures.

⇨ Action 2: Registry of rare diseases

4. Contribute actively to the development of the EU easily accessible and dynamic inventory of rare diseases based on the Orphanet network and other existing networks as referred to in the Commission Communication on rare diseases.

⇨ Action 11: Systematic collection and sharing of information

5. Consider supporting at all appropriate levels, including the Community level, on the one hand, specific disease information networks and, on the other hand, for epidemiological purposes, registries and databases, whilst being aware of an independent governance.

⇨ Action 2: Registry of rare diseases

III

RESEARCH ON RARE DISEASES

6. Identify ongoing research and research resources in the national and Community frameworks in order to establish the state of the art, assess the research landscape in the area of rare diseases, and improve the coordination of Community, national and regional programmes for rare diseases research.

⇨ Action 3: Research funding and a research programme focusing on rare diseases
Action 4: Building up international research cooperation

7. Identify needs and priorities for basic, clinical, translational and social research in the field of rare diseases and modes of fostering them, and promote interdisciplinary co-operative approaches to be complementarily addressed through national and Community programmes.

⇨ Action 3: Research funding and a research programme focusing on rare diseases

8. Foster the participation of national researchers in research projects on rare diseases funded at all appropriate levels, including the Community level.

⇨ Action 3: Research funding and a research programme focusing on rare diseases
Action 4: Building up international research cooperation

9. Include in their plans or strategies provisions aimed at fostering research in the field of rare diseases.

⇨ Action 3: Research funding and a research programme focusing on rare diseases

Action 4: Building up international research cooperation

10. Facilitate, together with the Commission, the development of research cooperation with third countries active in research on rare diseases and more generally with regard to the exchange of information and the sharing of expertise.

⇨ Action 4: Building up international research cooperation

IV
CENTRES OF EXPERTISE AND EUROPEAN REFERENCE NETWORKS FOR RARE DISEASES

11. Identify appropriate centres of expertise throughout their national territory by the end of 2013, and consider supporting their creation.

⇨ Action 6: Units for rare diseases in university hospitals
Action 7: Centres of expertise for rare diseases

12. Foster the participation of centres of expertise in European reference networks respecting the national competences and rules with regard to their authorisation or recognition.

⇨ Action 7: Centres of expertise for rare diseases

13. Organise healthcare pathways for patients suffering from rare diseases through the establishment of cooperation with relevant experts and exchange of professionals and expertise within the country or from abroad when necessary.

⇨ Action 5: Clarification of the care pathway for people with a rare disease
Action 6: Units for rare diseases in university hospitals
Action 7: Centres of expertise for rare diseases

14. Support the use of information and communication technologies such as telemedicine where it is necessary to ensure distant access to the specific healthcare needed.

⇨ Action 5: Clarification of the care pathway for people with a rare disease
Action 6: Units for rare diseases in university hospitals
Action 7: Centres of expertise for rare diseases

15. Include, in their plans or strategies, the necessary conditions for the diffusion and mobility of expertise and knowledge in order to facilitate the treatment of patients in their proximity.

⇨ Action 5: Clarification of the care pathway for people with a rare disease
Action 11: Systematic collection and sharing of information

16. Encourage centres of expertise to be based on a multidisciplinary approach to care when addressing rare diseases.

⇨ Action 7: Centres of expertise for rare diseases

V
GATHERING THE EXPERTISE ON RARE DISEASES AT EUROPEAN LEVEL

17. Gather national expertise on rare diseases and support the pooling of that expertise with European counterparts in order to support:

(a) the sharing of best practices on diagnostic tools and medical care as well as education and social care in the field of rare diseases;

⇨ Action 10: A national coordinating centre for rare diseases
Action 11: Systematic collection and sharing of information

(b) adequate education and training for all health professionals to make them aware of the existence of these diseases and of resources available for their care;

⇨ Action 8: Providing more education and training

(c) the development of medical training in fields relevant to the diagnosis and management of rare diseases, such as genetics, immunology, neurology, oncology or paediatrics;

⇨ Action 8: Providing more education and training

(d) the development of European guidelines on diagnostic tests or population screening, while respecting national decisions and competences;

(e) the sharing Member States′ assessment reports on the therapeutic or clinical added value of orphan drugs at Community level where the relevant knowledge and expertise is gathered, in order to minimise delays in access to orphan drugs for rare disease patients.

⇨ Action 9: Promoting the availability of orphan drugs

VI
EMPOWERMENT OF PATIENT ORGANISATIONS

18. Consult patients and patients′ representatives on the policies in the field of rare diseases and facilitate patient access to updated information on rare diseases.

⇨ Action 13: Empowerment of people with a rare disease

19. Promote the activities performed by patient organisations, such as awareness-raising, capacity-building and training, exchange of information and best practices, networking and outreach to very isolated patients.

⇨ Action 13: Empowerment of people with a rare disease

VII
SUSTAINABILITY

20. Together with the Commission, aim to ensure, through appropriate funding and cooperation mechanisms, the long-term sustainability of infrastructures developed in the field of information, research and healthcare for rare diseases.

⇨ Entire plan

APPENDIX 2

EUCERD RECOMMENDATIONS ON QUALITY CRITERIA FOR CENTRES OF EXPERTISE FOR RARE DISEASES IN MEMBER STATES

- Capacity to produce and adhere to good practice guidelines for diagnosis and care.
- Quality management in place to assure quality of care, including National and European legal provisions, and participation in internal and external quality schemes when applicable.
- Capacity to propose quality of care indicators in their area and implement outcome measures including patient satisfaction.
- High level of expertise and experience documented, for instance, by the annual volume of referrals and second opinions, and through peer-reviewed publications, grants, positions, teaching and training activities.
- Appropriate capacity to manage rare disease (RD) patients and provide expert advice.
- Contribution to state-of-the-art research.
- Capacity to participate in data collection for clinical research and public health purposes.
- Capacity to participate in clinical trials, if applicable.
- Demonstration of a multi-disciplinary approach, when appropriate, integrating medical, paramedical, psychological and social needs (e.g. RD board).
- Organisation of collaborations to assure the continuity of care between childhood, adolescence and adulthood, if relevant.
- Organisation of collaborations to assure the continuity of care between all stages of the disease.
- Links and collaboration with other centres of expertise at national, European and international level.
- Links and collaboration with patient organisations where they exist.
- Appropriate arrangements for referrals within individual Member States and from/to other EU countries if applicable.
- Appropriate arrangements to improve the delivery of care and especially to shorten the time taken to reach a diagnosis.
- Consideration of eHealth solutions (e.g. shared case management systems, expert systems for tele-expertise and shared repository of cases).