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THE FINNISH NATIONAL PROGRAMME FOR RARE DISEASES 2014–2017

Report of the Steering Group

Helsinki 2014

FACT SHEET

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Authors The Rare Diseases Steering Group Chairperson: Liisa-Maria Voipio-Pulkki Vice-chairperson: Jaakko Yrjö-Koskinen	**Commissioned by** Ministry of Social Affairs and Health, Finland HARE project number and date of appointment of the working body MSH122: 2011
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ABSTRACT

The Rare Diseases Steering Group presents its proposals for the Rare Diseases National Programme for the period 2014-2017. The programme has been drawn up with the involvement of the various stakeholders. The aim of the programme is that individuals, regardless of the rare disease they may have, should be able to live a full life based on their own choices, while receiving the appropriate treatment, rehabilitation and necessary psychosocial assistance.

Finland's social and healthcare services do not currently provide satisfactory responses to the needs of people suffering from rare diseases. Given the low frequency and insufficiency of expertise concerning these diseases, special measures must be applied within the health and social affairs service systems so that the patients concerned can receive the high quality treatment and rehabilitation they are entitled to, based on principles of equality.

The National Programme for Rare Diseases includes action proposals for the areas of research, treatment, rehabilitation and social support. The programme identifies the following measures as priorities: inclusion of special measures in health and social services legislation and preambles to legislation; clarification of the integrated care pathways for rare diseases; creation of rare disease units in university hospitals; increase in availability of medication for rare diseases, with reimbursement of the costs; establishment of a national coordination centre; finally, development of social support and rehabilitation.

An essential feature of the programme is that rare diseases patients, their families, and patient organisations will become more involved in the decision-making and planning of services related to rare diseases. Patient organisations should be offered more opportunities to influence the service system at all levels.

Keywords:

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

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The Ministry of Social Affairs and Health appointed the Rare Diseases Steering Group for the period 01/06/2012 to 31/12/2013.

The goals of the Steering Group were identified as:
1. To promote the use of the existing knowledge and experience within the Finnish healthcare system for the benefit of patients with rare diseases;
2. To promote quality of research and treatment, with equal access for patients with rare diseases;
3. To direct Finland's involvement in EU cooperation projects on rare diseases and address issues concerning the implementation of the EU Patients' Rights Directive.

The Steering Group's tasks were defined as:
1. To plan and direct Finland's implementation of EU recommendations concerning rare diseases;
2. To propose objectives and plans concerning the monitoring of measures for rare diseases, as well as for other potential measures.

Chairperson of the Steering Group: Liisa-Maria Voipio-Pulkki, Director, Ministry of Social Affairs and Health

Vice-chairperson: Päivi Topo, Secretary General, National Advisory Board on Social Welfare and Healthcare Ethics (until 02/04/2013); replaced by Jaakko Yrjö-Koskinen, Counsellor, Ministry of Social Affairs and Health

Members of the Steering Group (alternate delegates in brackets):

Päivi Topo, Secretary General, National Advisory Board on Social Welfare and Healthcare Ethics (until 31/05/2013); replaced by Leila Jylhänkangas, National Advisory Board on Social Welfare and Healthcare Ethics

Pälvi Kaukonen, Counsellor, Ministry of Social Affairs and Health (until 06/11/2012); replaced by Jaakko Yrjö-Koskinen, Counsellor, Ministry of Social Affairs and Health.

Annakaisa Iivari, Director, Ministry of Social Affairs and Health (until 20/03/2013); replaced by Teppo Heikkilä, Senior Physician, Ministry of Social Affairs and Health

Jaana Huhta, Advising Official, Ministry of Social Affairs and Health (Anne-Mari Raassina, Special Advisor)

Helena Kääriäinen, Research Professor, Department of Health and Welfare (Jukka Kärkkäinen, Head of Unit)

Päivi Koivuranta-Vaara, Senior Administrative Physician, Finnish Local and Regional Authorities (Heikki Punnonen, Development Manager)

Pirkko Karttunen-Lewandowski, Senior Physician in Paediatric Neurology, Etelä-Savo Hospital District Federation of Municipalities (Matti Suistomaa, Chief Senior Physician)

Leila Saulamaa, Senior Physician, Pirkanmaa Hospital District (Päivi Keskinen, Paediatric Specialist)

Markku Savolainen, Senior Physician, Pohjois-Pohjanmaa Hospital District (Johanna Uusimaa, Paediatric Neurology Specialist)

Harri Niinikoski, Assistant Professor, Lounais-Suomi Healthcare District

Jarmo Jääskeläinen, Professor of Paediatrics, Pohjois-Savo Hospital District (Reetta Kälviäinen, Clinical Professor in Epileptology)

Riitta Pirilä, Neurology Specialist, Lappi Hospital District (Laura Simontaival, Specialist in Paediatric Neurology)

Leena Vekara, Director of Healthcare, City of Tampere (Anja Jalonen-Männikkö, Director of Nursing)

Emmi Hanhikoski, Senior Social Worker, Town of Jyväskylä

Jorma Rautio, Plastic Surgery Specialist, Lip and Palatoschisis Centre, HUS (Ilkka Kantola, Specialist in Internal Diseases)

Sirkku Peltonen, Assistant Professor, Turku University Hospital (Anna-Elina Lehesjoki, Professor)

Risto Lapatto, Assistant Professor, Unit for Gynaecology and Paediatrics, Helsinki University Central Hospital

Niilo Keränen, Senior Physician, Member of the Steering Network for National Public Health Work, Kuusamo Health Centre

Riitta Salonen-Kajander, Senior Physician, Rinnekoti Foundation (Christina Avela, Genetics Specialist, and from 20/03/2013, Leena Toivanen, Specialist, both of the Rinnekoti Foundation)

Katri Karlsson, Member of the Board, HARSO Association (Päivi Reinikka, Member of the Board)

Eila Niemi, Special Project Manager, Finnish MS Society/Network for Rare Diseases (Jaana Hirvonen, Development Manager, Finnish Rheumatism Association/Network for Rare Diseases)

Jukka Sariola, Chairman, Muscular Diseases Federation/Network for Rare Diseases (Jussi Lindevall, Finnish EB Association/Network for Rare Diseases)

Additional members of the Steering Group, appointed 18/10/2012:

Ulla Närhi, Special Advisor, Ministry of Social Affairs and Health,

Ilona Autti-Rämö, Research Professor, Research Department, Social Insurance Institution of Finland.

The Steering Group met nine times. The Steering Group appointed two working parties, whose mission was to plan the implementation of the National Programme and to draw up proposals for the development of Centres of Expertise. The working groups were to hold seminars focusing on planning the programme.

From 18-19 March 2013, the Steering Group held a seminar at the Hanasaari Cultural Centre, aimed at involving the stakeholder groups in the implementation of the National Programme. The seminar was attended by 78 representatives of stakeholder groups, 29 of them representing patient organisations. The members representing the Steering Group were Jaana Lähdetie, Assistant Professor, Turku University Central Hospital, and Jarmo Wahlfors of the Finnish Academy.

The Ministry of Social Affairs and Health entered into service provision agreements with Katja Aktan-Collan (16/04/2012 to 31/12/2012) and Elina Rantanen (03/04/2013 to 30/12/2013), for assistance in implementation of the National Programme under the direction of the Steering Group.

In October 2013 the Steering Group invited the organisations represented by its members to provide feedback for inclusion in the draft of the National Programme. The feedback received was taken into account when finalising the National Programme.

The Steering Group submits its proposals to the Ministry of Social Affairs and Health and recommends that the Ministry launch the necessary measures for the implementation of the National Programme for Rare Diseases.

Helsinki, 13 March 2014

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

1 INTRODUCTION

The European Union identifies a rare disease as one that affects not more than 5 out of 10,000 individuals. It is estimated that more than 300,000 Finns suffer from a rare disease, injury, syndrome or deformity. These individuals represent about six per cent of the national population, and compose a large share of the heavy users of national health services. The significance size of this population is one of the reasons that the Rare Diseases Steering Group should be recognised in the Act for Provision of Social and Healthcare Services, and in the development of service structures. The rarity of disease and disabling conditions is accompanied by challenges in their diagnosis and treatment, in rehabilitation of the patient, in health and social services, and in everyday life. The rare disease or injury, the more challenging it can be to find expertise and assistance. Large amounts of resources are typically spent in seeking diagnosis of rare diseases, and treatment is often inappropriately directed and longer than necessary. The available resources should instead be better used in more rapid diagnosis and effective treatment of the patients.

The National Programme adopts defined concepts of *rare diseases* and *those afflicted by rare diseases* in order to better focus on the common problems faced by these persons and the measures taken to resolve them.

There are thousands of rare diseases. At least 5,000 have been discovered and new ones are continuously described in the literature. Persons afflicted by rare diseases share similar problems, such as finding the correct diagnosis, receiving correct information and the necessary expert services. Particular attention must be paid to issues related to the quality of the treatment, access to medical and social support, and the functioning of pathways between primary healthcare and specialised healthcare. Vocational and social integration, independent living, normal use of services, and coping with everyday life are key factors in the lives of rare disease patients and their families. Compared to other patient groups, those afflicted by rare diseases encounter greater difficulties in getting peer support and are more susceptible to psychological, social, and economic problems.

In 2009, the Council of the Europe Union issued a recommendation on measures to be applied within the area of rare diseases (Annex 1). The recommendation states that access to high-quality treatment and the recognition of equality and joint responsibility are values confirmed under the Council of the European Union's conclusions on common values and principles for health systems, and are extremely important for rare disease patients. The Member States are encouraged to ratify plans and strategies concerning rare diseases, and each nation is to draw up and adopt a National Programme for rare diseases by the end of 2013. One of the goals of the EU is to establish international centres of expertise for rare diseases and create associated networks for the dissemination of expertise. The Finnish Ministry of Social Affairs and Health appointed the Rare Diseases Steering Group for the period 01/06/2012 to 31/12/2013, for the purpose of implementing a National Programme for Rare Diseases.

The goals of the Steering Group were identified as:

1. To promote the use of the existing knowledge and experience within the Finnish healthcare system to benefit patients with rare diseases
2. To promote quality of research and treatment, with equal access for patients with rare diseases;
3. To direct Finland's involvement in EU cooperation projects on rare diseases and address issues concerning the implementation of the EU Patients' Rights Directive.

The Steering Group's tasks were defined as:

1. To plan and direct Finland's implementation of EU recommendations concerning rare diseases;
2. To propose objectives and plans concerning the monitoring of measures for rare diseases, as well as for other potential measures.

The Steering Group appointed two working parties, whose mission was to plan the implementation of the National Programme and to consider the establishment of centres of expertise. A seminar of the National Programme for Rare Diseases was held between the 18th and 19th of March 2013 in order to make preparations for the National Programme.

Those attending the seminar worked in small groups focusing on identifying the measures necessary for specific sections of the National Programme: research related to rare diseases; disease classification systems and registries; national definitions of rare diseases; diagnosis of the diseases; development of treatment and improving the availability of medication; national centres of expertise; development of social support; empowerment of rare disease patients; implementation of coordination, monitoring, evaluation and funding for the National Programme; international networking. The seminar was attended by 78 representatives from the various hospital districts, the Finnish Network for Rare Diseases (Harvinaiset) and the Rare Diseases Alliance of Finland (HARSO), the Finnish Social Insurance Institution, the municipalities, the Slot Machine Association (a national agency for funding of social and health organisations) and the Ministry of Social Affairs and Health. In September 2013, with the support of the European Project for Rare Diseases National Plans Development (EUROPLAN), HARSO organised a conference to obtain input from patient organisation representatives on the draft programme for rare diseases. The conference was attended by 60 representatives of various patient organisations.

The current proposal for the National Programme for Rare Diseases is the outcome of the March 2013 seminar, the September 2013 conference, and the work of the Steering Group appointed by the Ministry of Social Affairs and Health. The Steering Group has taken into account the feedback received from the patient organisations in preparing the National Programme, which aims to promote the role of rare disease patients in the Finnish healthcare system and in Finnish society as a whole. Rare disease patients need equality of treatment with other disease groups. Correct diagnosis, proper rehabilitation, medication, and normal everyday life are essential, and their value cannot be measured in monetary terms. However, the early diagnosis and treatment of cases of rare disease will in fact permit savings in healthcare expenses. Finland has expertise in rare disease diagnosis and treatment but this expertise has not been efficiently used, and thus is effectively wasted. The National Programme aims to make better use of the existing resources and thereby improve diagnostic and therapeutic outcomes. The essential strategy of the National Programme is to develop measures of significant human value and influence on the well-being of rare disease patients. The National Programme applies the term *rare disease* to all rare diseases, injuries, syndromes and deformations.

2 THE NATIONAL PROGRAMME FOR RARE DISEASES

2.1 BACKGROUND TO THE SITUATION

Until now, Finland has not implemented a National Programme for rare disease, however national attention to rare diseases has increased since the 1990s. In 1991, the Ministry of Social Affairs and Health designated the following associations as rare diseases resource centres: Hengitysliitto ry (Organisation for Respiratory Health in Finland), Iholiitto ry (Central Organisation for Skin Patients), Invalidiliitto ry (Association of People with Physical Disabilities), Kehitys-vammaisten Tukiliitto ry (Inclusion Finland KVTL), Kuuloliitto ry (Finnish Federation of the Hearing Impaired), Näkövammaisten keskusliitto ry (Finnish Federation of the Visually Impaired), Lasten kuntoutuskoti (Norio Centre), Suomen MS-liitto ry (Finnish Neuro Society), and Suomen Reumaliitto ry (Finnish Rheumatism Association). The Finnish Network for Rare Diseases was founded in 1995, thus joining the designated social and healthcare organisations involved in work among people with rare diseases. HARSO, the Rare Diseases Alliance of Finland was founded in the spring of 2012. HARSO's membership currently totals 22 national rare disease patient organisations. The aims of HARSO are to ensure that rare disease patients and their loved ones have equal opportunities in matters such as social participation, and to promote the provision of appropriate social and healthcare services. HARSO also offers a support network for those rare disease patients who do not have their own specific organisation, due to the rarity of their diseases. National rare disease events are organised annually in Finland. The Finnish Orphanet team collects and disseminates knowledge and information on rare diseases, and disseminates information via a Web portal.[1] In 2009-2010, a pan-Nordic project was implemented for exchange of information and organisation of joint training events. No centres of expertise have yet been officially designated, but the various university hospitals generally operate as national centres for rare diseases, with some of them specialising in specific diseases. In view of Finland's small population and its unique legacy of diseases, the National Programme for Rare Diseases must be conceived to meet specific national requirements and to address international cooperation. Finland's National Programme for Rare Diseases is impacted by numerous on-going legislative reforms and national and international initiatives, ranging from preparations for a national cancer centre to the ratification of the UN Convention on the Rights of Persons with Disabilities.

The framework for expanding and disseminating expertise on rare diseases is set by Finland's Healthcare Act. The Act states the goal of "equality in availability and quality of healthcare" (Government Decree 2011), and requires that the nation's specialised university hospital districts agree on the coordination and division of labour for different tasks (*Erikoissairaanhoidon järjestämissopimus*, or "Specialised Healthcare Organisation Agreement"). Section 45 of the Act provides for certain specialised healthcare measures, specifying that in some case treatment can be provided in a centralised manner. However the organisation of diagnosis and treatment of rare diseases is also impacted by a trend towards increasing freedom of choice, both at the national and at the EU level. Since 2014, in accordance with the Healthcare Act and subject to certain preconditions, patients may choose that their place of treatment be anywhere in Finland. The treatment of rare diseases has also changed in the broader European context, under the EU Directive on patients' rights in cross-border healthcare (2011), which has been implemented in Finland as of 2014. Finland needs centres of expertise for rare diseases, so that national expertise can be put to best use, and to avoid the significantly higher costs that result when patients seek treatment abroad.

Finland's on-going reform of social and healthcare service structures is aimed at ensuring universal equality of access to social and healthcare services. The fundamentals of the policy are that the state social and healthcare areas are responsible for the organisation of social and healthcare services, and shall include specialised areas with specific service responsibilities. These areas are used to manage situations of overlap

[1] Orphanet (Portal for Rare Diseases and Orphan Drugs) is an open-access reference portal for information on rare diseases and rare disease drugs, led by a consortium of around 40 European countries and coordinated by the French INSERM team.

in the service network, and to provide regional guidance. University hospitals must have a sufficiently endowed ownership basis and resources, and apply administrative solutions that avoid fragmentation of the functional whole. The research, development and training functions are required to maintain close contact with service providers. The government's proposals for the Social and Healthcare Organising Act are to be submitted to Parliament in the spring of 2014.

Finland's strengths and weaknesses in rare disease expertise at university and central hospitals have been mapped by means of questionnaire submitted to senior specialised physicians and the Network for Rare Diseases member organisations (STM 2011). The participants in the survey called for more cooperation in the diagnosing and treatment of rare diseases. The responses stressed the need for increased cooperation and sharing between the hospital districts to better address the responsibilities for diagnosis, treatment and rehabilitation involving rare diseases. In response to questions concerning how to address shortcoming, the respondents recommended the formation of various consultancy networks. The respondents indicated that consultation is already quite common, but that clearer and more appropriate forms of consultation were needed. In addition, the responses recommended the organisation of more joint training sessions and meetings, during which rare diseases could be discussed among the representatives of the different sectors and university and central hospitals. Central coordination was felt to be necessary within the specific responsibility areas, as well as in the overall national administration of rare disease services. In addition to cooperation being inadequate, it was felt that there was lack of knowledge and resources. To remedy the lack of knowledge, better training and clearer instructions were called for. In terms of the availability of resources, the respondents indicated that one of the key problems is the insufficient numbers of physicians and the inadequacy in their job descriptions. In this regard, respondents noted that physicians are not allocated added time for dealing with individual cases of rare diseases, although rare diseases clearly require more sustained efforts and resources than common diseases.

EURORDIS, the European Organisation for Rare Diseases, conducted country-specific surveys in 2009. The survey for Finland showed that rare disease patients had experienced shortcomings in access to information and treatment. A third of national respondents had been diagnosed without having received appropriate information about the significance of the diagnosis. Some 70% of the respondents stated that they had not received proper psychological support, although 91% of the individuals felt this was an important need. Also, 37% of the respondents stated that access to essential healthcare services was difficult or even impossible. The motivations for the perceived lack of access varied: the services simply did not exist or were too expensive, the individuals had not been referred to receive such services, the waiting periods were too long, or the services were too far away and difficult to reach.

2.2 GOALS FOR THE NATIONAL PROGRAMME

The Steering Group proposes that the Finnish National Programme for Rare Diseases should have the following general goals:

- That rare disease patients receive the same quality of services as other patients
- That prevention, diagnosis, treatment, rehabilitation and the necessary social services for rare disease patients be of high quality and equally accessible throughout Finland
- Establishment of rare diseases units and centres of expertise
- Speedier diagnosis of rare diseases, and constant reduction of cases without diagnosis
- Reduction of premature morbidity and mortality
- Multidisciplinary coordination of care

- Improved flow of information, including potential for transfer of patients between healthcare centres and from paediatrics to adult wards
- Increased efficiency in the use of healthcare resources for cases of rare diseases
- Improved quality of life and socio-economic status for rare disease patients
- Promotion of availability and reimbursement of rare disease medical products
- Ever-increasing knowledge related to rare diseases, through research
- Increased knowledge and ability concerning rare diseases among social and healthcare professionals
- Greater international cooperation on issues concerning rare diseases
- Creation of clearer cooperation channels between experts, both nationally and internationally
- Closer involvement of rare disease patients in decisions concerning rare diseases
- Permanent integration of the National Programme for Rare Diseases within the Finnish social and healthcare systems.

The challenges associated with rare diseases are specified in detail in the National Programme's five sections. To resolve the challenges, the programme presents the above general goals, and proposes 13 concrete measures for their implementation. The programme identifies specific indicators to track the implementation of the 13 proposed measures, naming the parties responsible, time schedules and preconditions for their realisation. Specific coordination, evaluation, and monitoring methods are set out. The report concludes with a summary of the National Programme priorities and the recommended measures for their implementation, as well as an Appendix showing how the programme responds to EU recommendations on action in the field of rare diseases.

3 SECTIONS OF THE NATIONAL PROGRAMME

The proposed National Programme is divided in five sections, numbered “3.1” to “3.5” for the purposes of the current document. Each section includes the proposed measures for implementation, the indicators, parties responsible, scheduling, and necessary preconditions.

3.1 DEFINING AND REGISTERING RARE DISEASES

The first step towards improving the care and quality of life for rare disease patients is the recognition of their status. Cases of rare disease receive relatively less access to specialised services when compared to the rest of cases. In order to achieve equal footing in the service system, rare disease patients require special attention.

Although rare disease patients are not numerous when counted per disease, their total number is large: about six per cent of the Finnish population suffer from a rare disease during their lifetime. When looking at special medical care, the proportion of rare disease patients is still greater.

The status of rare disease patients must be recognised by a uniform definition, enabling the identification of all those who require the types of measures applied to rare diseases. The differences in the definition of rare diseases and rare disease patients at the EU level cause problems in terms of joint research projects and dissemination of expertise. Research and dissemination of knowledge have also been hindered by the lack of relevant databases. Finland is one of the nations that lacks data on its rare disease clinics, research work, and forms of treatment applied. Instead, the approaches to cases and application of expertise are based on personal experience, relationships and to some extent on chance. This means that patients may not receive the best possible expertise during diagnosis, treatment and rehabilitation.

3.1.1 Proposed measure 1: A uniform definition of rare diseases and legislative recognition of the need for special measures

Rare diseases should be defined in accordance with the EU definition and associated special measures should be recognised in the legislation.

The European Union recommends that all Member States adopt a uniform definition of rare diseases, to facilitate cooperation on the relevant issues and practices at the Community level.

The Finnish National Programme applies the EU definition of rare diseases, meaning that rare diseases are those that do not affect more than five out of 10,000 individuals. In Finland, this definition implies a maximum of 2,800 persons per rare disease. This National Programme also considers rare diseases in the broad sense, as covering injuries, deformations and syndromes. The National Programme aims to respond to the challenges posed by rare diseases, including those of disability and daily function in life.

Most rare diseases are extremely rare. The rarest ones are also those that present the greatest diagnostic and therapeutic challenges, and which require special consideration in planning and implementing the National Programme. It will be necessary to seek expertise related to these diseases via international cooperation networks.

Rare disease patients are entitled to treatment and social services that met their needs, of the same quality as those for other patients and social welfare clients. To make this principle a reality, rare diseases require special measures within the service system, particularly given their low rates of occurrence and the scarcities of developed expertise.

The above statements are in line with European Community's policies and measures in the field of rare diseases. The principles reflected should be included in the articles or preamble (statement of principles) of the new Social and Healthcare Organisation Act.

Goals:

- To define rare diseases to comply with the EU's definition.

- The social and healthcare organisation act, or its statement of principles, recognises that rare diseases require special measures to be applied within the service system.

Indicator: The requirement of special measures for rare diseases is stated in the legislation or its preamble.

Responsibility: Ministry of Social Affairs and Health

Schedule: Immediate internal identification of the definition; Recognition of the need for special measures is included in the Social and Healthcare Organisation Act or its preamble, due to come into effect in 2015

Preconditions: Political support

3.1.2 Proposed measure 2: Registration of rare diseases

A register of rare diseases should be established and included within the computerised system of Care Registers for Social Welfare and Healthcare (HILMO). The aim is that rare diseases will be more easily found and identified based on the register, rather than relying solely on the International Classification of Diseases (ICD-10) system.

Enactment of a national register of rare diseases is a precondition for the development of both research and care. Data collection must be systematic and as far as possible should be automated. The use of separate registration forms and systems should be avoided, as these are time consuming and fail to develop comprehensive data resources. Collecting data for the register should not mean extra work. The fundamental aim of data collection is to make rare disease diagnoses identifiable within the healthcare system.

Finland will not establish a separate national register of rare diseases. Registration of rare diseases will be carried out by the National Institute for Health and Welfare (THL), using the existing HILMO system. Identifying rare diseases within the HILMO system is a key factor in obtaining epidemiological knowledge and monitoring the care pathways.

An international disease classification system, probably the ICD-11 international system (release date 2018), will be applied when registering rare diseases. The HILMO system will document diagnosed numbers of rare diseases in a far more comprehensive manner than what is accomplished by current data collection. This aspect of the HILMA system is expected to become operational by 2017. In Finland, healthcare register administrators should take rare diseases into account when monitoring the development of codes, in order to resolve the issue of whether to waited for the ICD-11 system to become available or to start using a different disease classification system that is already in use.

In addition to the automated ICD register, centres of expertise may also compile their own disease-specific registers, which will require the advance consent of their patients.

Goals:

- The identification of rare diseases is facilitated.
- Launching of research projects is facilitated and possibilities for cooperation are improved, once patient groups have been identified.

Indicators: HILMO codes for rare diseases are observed as far more comprehensive

Responsibility: THL

Schedule: Collecting of data for the registry to begin no later than when the ICD-11 system is operational

Conditions: Realisation of the ICD-11 system or some other disease classification system

3.2 RESEARCH ON RARE DISEASES

Due to their rarity, research on rare diseases may find itself in a situation of competitive disadvantage when compared to research on common diseases in terms of decision-making on research funding. The collecting of sufficient patient data for clinical and epidemiological research is difficult because the patient groups are small and scattered due to the absence of registers and due to inadequate diagnostics. For the same reasons, research focusing on the everyday life and quality of life of rare disease patients is not able to compete for research funding.

Treatments are being developed under the leadership of the pharmaceutical industry, and the pharmaceutical industry is not interested in the development of the treatment needed by small patient groups because of the uncertainty of the economic yields. Information on research projects has not been compiled systematically anywhere and thus the benefits of cooperation are lost. The development of social and healthcare services should be based on knowledge, which is why research focusing on rare diseases is needed on services and their impacts on people's health, well-being, and coping with everyday life.

3.2.1 Proposed measure 3: Targeted research funding and research programmes on rare diseases

Government funding should be made available to applicants seeking partial funding for research on rare diseases and the Finnish disease legacy. An initiative will be prepared and submitted to the Academy of Finland, proposing the establishment of a multidisciplinary rare diseases research programme.

Research on rare diseases is necessary to serve in the treatment of the diseases and ease the life of the people afflicted. A more solidly organised and funded national research basis is necessary in order to participate in European research programs for rare diseases. Finland must also ensure sufficient funding for research delving into the Finnish disease legacy and the treatment of the diseases specific to this nation, which are not studied elsewhere.

University-level healthcare research of all kinds is generally carried out with the support of government research funding. The National Institute for Health and Welfare (THL) defines research priorities and goals every four years, and grants research funding to committees with specific areas of responsibility. These committees then decide on the use of the research funding for projects under their special areas, based on applications. The National Programme proposes that the THL indicate research into rare diseases and Finland's disease heritage be indicated in the research priorities and goals, and that such research be executed in cooperation among the special responsibility areas.

The programmes of the Academy of Finland direct funding to key fields of research serving science and society. The Academy's programmes are intended to develop selected research areas, raise scientific levels in the fields, and create new skills. The programmes emphasise multidisciplinary and interdisciplinary research and international cooperation. The National Programme for Rare Diseases Steering Group proposes to submit an initiative to the Academy of Finland for the creation of a multidisciplinary research programme focusing on rare diseases. The proposed research programme should deal with disease mechanisms, diagnostics, treatment, and social support, as well as the quality of life of rare disease patients and their means of coping with everyday life.

As part of these initiatives, it is necessary that data from on-going research projects on rare diseases be compiled in a single database.

Goals:

- To strengthen research funding focusing on rare diseases.
- To create a rare disease research cooperation network and a database of on-going research.
- To promote research concerning rare disease patients as social and healthcare service users and examining the effectiveness of service systems, through wide-ranging cooperation with experts from various sectors, including patient and client organisations.

Indicators: State research funding dedicated to research into rare diseases and the Finnish disease legacy; Submission of a rare disease research initiative to the Academy of Finland, with subsequent decision to carry out the research programme; Data compiled from on-going studies

Responsibility: THL, for state funding of research into rare disease priorities; University hospitals, for coordination of the research programme initiatives; Centre for coordinating rare disease research, for compilation of the research database

Schedule: Preparation of government research funding programme in 2014-2015, implementation at the beginning of the new four-year period, 2016-2019; submission of the research programme Initiative to the Academy of Finland in 2014; Research Database in 2015

Preconditions: Ensuring the implementation of the initiatives by the responsible parties

3.2.2 Proposed measure 4: Strengthening international research

To increase the opportunities for international research collaboration by applying for membership in the E-RARE Project.

Finland does not have sufficient numbers of individual patients afflicted by rare diseases to enable the development of treatments, and the country must depend in part on the data, case knowledge and services offered by cooperation in international networks. To participate in such international networks, Finland requires simplified administrative procedures for permits and cooperation.

Strengthening of national rare diseases research funding and structures, as recommended in Proposed measure 3, will facilitate competition for European Union funding in this area. Finland should make use of European Research Council funding, as well as of the Horizon 2020 funding programme for research projects focusing on rare diseases.

In 2012, the European Commission and the US National Institutes for Health Research launched the International Rare Diseases Research Consortium (IRDiRC), to promote international cooperation in research on rare diseases. The goals of IRDiRC are to develop 200 new rare disease treatments and have almost all rare disease patients diagnosed by the year 2020. Each country or organisation participating in IRDiRC is required to fund research on rare diseases in the amount of a specified commitment over a five-year period. This sum is to be used for each member country's or organisation's own research projects, and is not transferred elsewhere. The Academy of Finland joined IRDiRC in June 2013.

The European Union E-RARE-1 project stimulated successful international cooperation on rare disease research within Europe. This cooperation will be further developed in September 2014, under the E-RARE-2 Project. The Academy of Finland should be a participant in the coming stages of the E-RARE-2 Project.

Goals:

- To cooperate in international research in a manner that obtains sufficient patient data, of service to rare disease patients.
- To achieve national rare diseases research funding at sufficient levels for participation in international cooperation projects, such as via E-RARE membership.

Indicators: National funding to enable joining E-RARE

Responsibility: THL, the Ministry of Education and Culture, the Academy of Finland

Schedule: Application for E-RARE membership when the second stage begins in 2014

Preconditions: Sufficiency of national funding

3.3 BETTER AND MORE EFFICIENT HEALTHCARE FOR RARE DISEASE PATIENTS

Rare disease patients and their families face a wide range of problems.

When dealing with a rare disease, achieving a correct diagnosis is often difficult. This leads to situations of delay, where a significant share of patients visit many different physicians before receiving the correct diagnosis. Some patients receive one or more incorrect initial diagnoses, as the physicians are not always in a position to suspect that a rare disease is the cause of the symptoms. Primary healthcare physicians and care staff do not have sufficient knowledge concerning rare diseases. In most cases, the timely diagnosis of a rare disease requires the simultaneous presence of several specialised consultants, which the current healthcare system cannot easily provide. The exercise of highly specialised expertise almost always requires increased time compared to physicians' more routine work, but there is typically little extra time allocated for rare disease tasks. Delays in diagnosis often have significant negative consequences for rare disease patients and their families. Speedier diagnosis would also save public resources, as repeated assessments conducted by going to several different experts would be unnecessary, and treatment and rehabilitation could begin much earlier.

When a rare disease patient has been correctly diagnosed, he or she faces new challenges. New symptoms and diagnostic changes can appear after the initial diagnosis, however these are not always observed or examined. Many rare diseases require high levels of specialist expertise, and patient care must almost always be multidisciplinary. When treatment and rehabilitation of a rare disease patient is planned or undertaken, there is often the lack of a single party to assume overall responsibility.

Each field of specialisation focuses on the treatment of certain symptoms, but wholeness is not necessarily anyone's responsibility. As a child patient becomes an adult and the responsibility for her care is transferred to an adult specialist, the continuity of care is again not guaranteed. There are also regional differences in access to care, and a rare disease patient is not always referred to a Centre of Expertise.

Not all healthcare professionals have sufficient knowledge of rare diseases or the required measures. This can in particular occur in emergency departments. The structures and systems for special healthcare do not provide sufficiently detailed care instructions to basic healthcare personnel. In an acute situation, the patient should be quickly sent to an expert unit with sufficient experience to provide specialised treatment. The majority of rare diseases are such that there currently no assured medical treatments for the patients. Many rare diseases have in recent years been provided with disease-specific drug treatments, but their effectiveness has often remained uncertain because of the small total numbers of patients and their dispersion among many countries. This hampers the reliable detection and documentation of treatment outcomes. In addition, many of the drugs for treating patients afflicted by rare diseases are expensive, in part because the rare disease drug status granted by the European Commission guarantees any new drug a reserved status for 10 years. Pharmaceutical companies have little interest in bringing generic preparations to the market because of the weak demand once the period of exclusive sales for the rare disease drug has ended. However, the pharmaceutical companies' interest in developing rare disease drugs and transforming "common-disease" drugs for use in rare disease illnesses has increased. Surgical treatment of rare diseases requires special technical know-how. Such know-how can only be developed through repetition over time, therefore surgeons and departments should be supported in focusing on procedures for rare diseases.

Some rare diseases can be diagnosed in infants, before visible symptoms have developed. The initiation of treatment at this stage would ensure far superior treatment outcomes, and secondly would also be less expensive. This is why there are now a variety of rare disease screening tests for infants, however in Finland the only such screening is for detection of congenital hypothyroidism. The neonatal screening programme should be expanded to achieve the levels seen in many other European countries. This requires proper resourcing. The Screening Working party appointed by THL has been considering the necessity of establishing a national screening centre.

3.3.1 Proposed measure 5: Clarifying rare disease patient-care pathways

The diagnostic procedures for rare disease cases should be streamlined, along with the associated care pathways, and these pathways should be known and used by all levels of the healthcare system. "Treatment passports" should be issued to the rare disease patients who require them.

To find better solutions to the problems faced by rare disease patients, we must first create clearer care pathways, known to all levels of healthcare services, the supporting organisations, social services, the Finnish Social Insurance Institution, and among rare disease patients and their families. Figure 1 shows the treatment pathway of a rare disease patient, including both the existing healthcare actors as well as new centres of expertise and rare disease units that are still to be founded, as described in Proposed measures 6 and 7.

The starting point is a symptomatic patient, who is not immediately provided with a diagnosis due to the rarity of his disease. His treatment pathway usually begins at the primary healthcare level or the basic special healthcare level. From there, diagnosis, treatment, and rehabilitation should proceed along a pathway suited to the patient's situation. The purpose of determining the care pathway is to clarify and accelerate the diagnosis and treatment of rare disease patients, not to complicate the patients' lives by making treatments available at only selected units. Each rare disease patient needs to be able to initiate the care pathway without delay, from their own unique own starting point, and reach the levels of diagnosis and treatment that answer his rare disease requires.

The patient must be placed on the treatment pathway quickly in the event that the correct diagnosis or the appropriate care cannot be provided within the first few steps along the pathway. However a fundamental principle is that the care and monitoring should be implemented near the patient's place of residence.

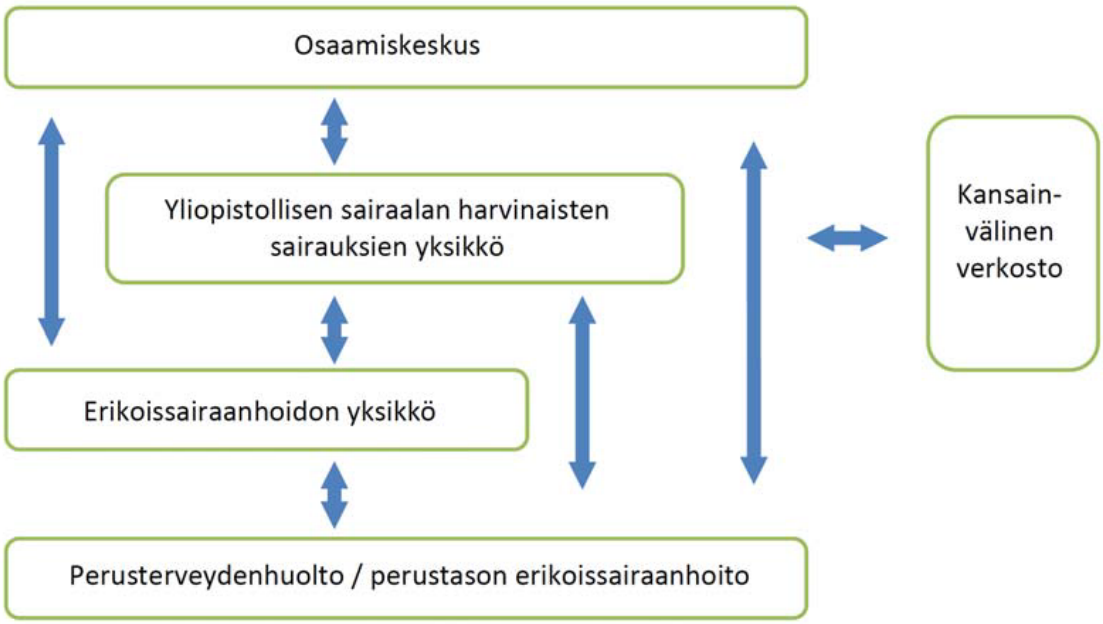

Figure 1: The care pathway for a rare disease patient. from seeking diagnosis to the realisation of care and monitoring. There is an urgent need to proceed upwards from diagnostics, and when administering care to back-track quickly to providers that are near the patient's home.

Some rare diseases can only be diagnosed in a particular special care unit. In cases of diagnosis with persistent uncertainties, or where multidisciplinary expertise clearly required for diagnosis or care, the patient should have access to her university hospital's special responsibility unit for rare diseases, for multidisciplinary examination and receipt of a diagnosis.

This unit also assumes overall responsibility for the planning of the care by the different actors, , unless the patient is sent to a further centre of multidisciplinary expertise. The unit also oversees the continuation of the care when the patient is transferred from the paediatric ward to the adult ward.

The centres of expertise are national centres focus on specific disease groups. Patients are entitled to have access to these when necessary. From the centres of expertise, the patient's care and monitoring may directed to a specific rare disease unit, other clinics, primary healthcare services or the third sector (non-governmental, community organisations and associations).

All levels of healthcare must engage in close cooperation with social and rehabilitation services in order that rare disease patient are considered and provided for in a comprehensive way. Social services, rehabilitation and psychosocial support needs must be assessed as rapidly as possible, in conjunction with the diagnosis.

The centres of expertise participate in international networks, which offer channels of assistance in matters involving the patient's diagnosis or treatment. Other health units may also engage in cooperation with international experts. Consultation in the diagnosis of the patient's illness and on decisions for local care are the primary forms of international cooperation, however where necessary the patient can also be referred to European centres of expertise, for assessment.

The EU Directive on patients' rights in cross-border healthcare (2011) has been applied in Finland since 2014. According to the EU Directive, the patient is entitled to reimbursement for treatment received in another EU Member State under the same terms as for similar treatment received in the patient's home country, for those treatments that are eligible in the home country. The Finnish Social Insurance Institution has established a cross-border healthcare service desk to provide information to persons seeking treatment abroad, and for those from abroad seeking treatment in Finland. Expertise on rare diseases should gradually be collected at this contact point. The EU Patient Directive requires that Finnish healthcare service be defined in a manner that Finnish users can understand which costs incurred abroad will be compensated in Finland. In early 2014, a new body of the Ministry of Social Affairs and Health will be established, with responsibility for defining the range of the Finnish healthcare service and continuous updating of the definitions. This body will be able to provide general guidelines regarding the application of services, but will not issue opinions on individual patient care decisions. From a rare disease patients' point of view, it should be ensured that the definitions of eligible services includes new, scientifically proven therapies, which are not available in Finland but are available in other EU countries. The new body for defining the range of services will be composed of a council, a permanent secretariat, and a network of experts. It must be ensured that the centres of expertise for rare diseases or representatives of the networks of these centres belong to this expert network, so that the assessments of the centres in matters such as the development of therapies are heeded.

The rare disease patients' clinical pathway must be made clear to all participants. The parties that are best suited to carry out patient diagnoses, planning and implementation of treatment and rehabilitation, as well as other support measures, must cooperate with one another. Cooperation must be done by consultation, and may also be by sending the patient onward within the system. The pathway should be tailored so that the patient and her data are sent to the facilities that can provide the necessary treatment and monitoring as close as possible to the patient's home.

The party responsible for care will ensure that the rare disease patient is issued a "care passport" (in Finnish or Swedish language) facilitating his transfer from unit to unit, and to assist in possible acute situations. The care passport will be developed as part of the existing and future computerised information system. Where necessary, a copy of the passport can be printed out to accompany the rare disease patient.

Goals:

- The rare disease patient's care pathway becomes clearer.
- Personnel at each healthcare level know how rare disease diagnostics and treatment are organised.
- Where necessary, the rare disease patient is issued a care passport in Finnish or in Swedish.

Indicators: The rare disease patient's care pathway is known to all healthcare professionals; the care passport is developed and included in national computerised systems

Responsibility: Ervat: Agreement regarding the rare disease patient's clinical pathway; THL: Care passport

Schedule: During the period of effect of the programme

Preconditions: Implementation of the rare disease units and centres of expertise, development of suitable electronic system within which to implement the passport

3.3.2 Proposed measure 6: Creating rare disease units at university hospitals

Every university hospital should have a rare disease unit, employing a rare disease coordinator and a multidisciplinary working group.

Each rare disease patient needs a health services party that takes overall responsibility for their care, so that they receive faster, better treatment and support, at less cost. Currently, the diagnosis and care of rare disease patients consumes substantial amounts of resources, but this is in part because the diagnoses may not be clear and the care pathways are not always optimal. Proper care pathways would save resources and speed the start appropriate therapies. The problem of lack of clear responsibility is especially acute in the case of adult patients. For both children and adults, experts representing different fields rarely have the opportunity to meet and resolve diagnoses requiring multi-profession knowledge, and to consider the care as a whole.

Rare disease units are needed within university hospitals. A rare disease unit is composed of a multi-professional working group, and attends to undiagnosed patients whose symptoms could conform to a rare disease. Such units can achieve better results by reorganising the current functions. Six per cent of Finland's population are afflicted by a rare disease sometime during their lifetime. This means that the proportion of rare disease cases among special care patients is still higher, and that they make up a large group of patients at university hospitals. Having a unit for rare diseases at each university hospital enables the calling together of multi-professional experts to diagnose difficult-to-diagnose patients and coordinate their care.

The units assume responsibility in the university hospital's special areas of responsibility, for both paediatric and adult patients showing symptoms indicating a rare disease, but who have not received a diagnosis in primary healthcare or at a special care unit. The units assume responsibility for diagnosed rare disease patients who are in need of multi-professional expertise and for whom a specialised centre cannot be established in Finland or who cannot be treated in any other specialised care centre. The operation of rare disease units should accommodate all age groups.

Each unit needs a rare disease coordinator. This figure would be someone already employed at the university hospital, who serves as a coordinating link in the direction of the elements of basic healthcare and as an information channel between the different fields of specialisation and social welfare personnel. The coordinator organises multi-professional meetings to address rare disease cases. Within each unit, the coordinator can call together a specific group of specialists compose of the experts deemed necessary for the particular case. The availability of a genetics specialist must be ensured for every unit.

A rare disease unit enables flexible cooperation between different clinics, and where necessary, coordinates the transfers of children to the adult ward or of any patient to primary healthcare and social services. Each rare disease unit cooperates with other rare disease units, centres of expertise, international networks, and patient organisations. In addition to diagnostics and care, the units coordinate the patients' rehabilitation and support measures when another suitable coordinating party cannot be found in the centres of expertise or from a field of specialisation.

If there are suspicions that a patient is afflicted by a rare disease, the case will be readily attended by consulting a rare disease unit or a centre of expertise, so that the patient is diagnosed and treated, as much as possible at the primary healthcare or special-care clinic levels. Patients presenting difficulties in diagnosis or treatment decisions will be sent without delay to a rare disease unit or national centre of expertise.

Cooperation between university hospitals, central hospitals and primary healthcare services will be addressed within the special responsibility areas.

The rare disease unit is the primary coordinator of the rare disease patient’s care. The patient can be treated at a university hospital clinic, central hospital or a primary healthcare facility close to the patient's home, if the care needed does not require special care. The rare disease unit at each university hospital organises rare disease meetings from time to time, and invites representatives of primary care, specialist fields or rehabilitation units to attend these meetings.

Each unit promotes access to information concerning rare diseases for social welfare and healthcare personnel. The rare disease units will maintain websites for transmission of information, also showing the experts' contact details.

A rare disease unit shall be organised at each university hospital in the manner that best ensures its effective operation given the context of that particular hospital. For example the unit can be based in the genetics clinic, which is generally involved in a large share of rare disease diagnostics work.

Each rare disease unit requires funding to establish and maintain the positions of rare disease coordinator and coordinating nurse, as well as convening multi-professional meetings with sufficient frequency. The hospital administration must ensure that the necessary multi-professional team members participate in the meetings concerning rare diseases.

Goals:

- Each rare disease unit is responsible for the diagnosis of and coordination of care for rare disease patients for whom there is no clear field of specialisation
- Cooperation among university hospital is facilitated
- Transfer from paediatric clinics to adult care in primary healthcare proceeds smoothly
- Healthcare professionals are able to devote the necessary time to diagnosing and care of rare diseases
- Circulation of information on rare diseases is promoted within social welfare and healthcare services

Indicators: Five rare disease units are established; Five rare disease coordinators and five coordinating nurses are designated

Responsibility: The five Finnish university hospitals: HYKS, TYKS, TAYS, OYS and KYS

Schedule: Planning for rare disease units in 2014; Appointment of coordinators and coordinating nurses, founding of the units in 2015; Operation of the individual patient working groups and network-based information dissemination in regular operation in 2016

Preconditions: Assembling of resources for a rare disease unit at each university hospital

3.3.3 Proposed measure 7: Centres of Expertise for rare diseases

Centres of expertise for rare disease patient groups should be established, as well as national centres of expertise fulfilling EU criteria.

Given that patients afflicted by an individual rare disease are so few in number, it would be useful to gather the specialised knowledge and expertise associated with their care, so that scarce resources would not be scattered and every patient would receive the best possible care. It is proposed that Finland establish centres of expertise, each focusing on a specific group of rare diseases. The establishment of such centres will result in cost savings, as more efficient patient guidance and treatment are achieved and the numbers of unnecessary doctor visits and hospitalisation cycles are reduced. There will also be reduced costs due to malpractice.

The Centres of Expertise may be such that they fulfil the EU criteria, and will take leadership roles in international cooperation networks and among other disease group centres of expertise. In other cases they may be affiliated with national disease groups or disease-specific centres of expertise whose primary aim is to facilitate the dissemination of expertise at the national level and not to fulfil the EU criteria. The centres of expertise can be designated as EU or national centres of expertise.

Finland already has some centres of expertise in partial operation, particularly in the form of university hospitals with special skills in regards to certain diseases. Such hospitals provide consultants for access to their expertise in other parts of the healthcare and social welfare systems.

However clearly planned cooperation, offering opportunities for the centres to focus on specific diseases or disease groups, has so far been lacking. The proposed Centres of Expertise can be formed in cooperation with the rare disease units of the different university hospitals, or with third-sector parties (e.g. research groups, patient organisations). The centres of expertise will be set up in a sustainable manner within healthcare structures. They will not rely on the ongoing presence a single expert, and will instead be structured for continuity. The centres of expertise will be required to have defined, systematic, continuous roles within the Finnish healthcare system, therefore third parties cannot serve independently as the basis of centres of expertise, but they can form a centre in cooperation with hospitals. One centre of expertise will be formed per disease or disease group.

A Centre of Expertise will be will be contacted when suspicion arises that diagnosis involves a rare disease, or when a rare disease has definitely been diagnosed. The centre of expertise in question then guides the disease diagnosis, patient care, rehabilitation and monitoring, however following the initial planning the care of the patient is returned to her own hospital district's designated public healthcare professionals. The hospital professionals assume responsibility and cooperate with the centre of expertise. The rare disease units of the university hospitals cooperate closely with the centres of expertise. The expertise of the centres can be put into practice by the experts coming to the different hospitals, rather than having patients transferred to the centres.

The national network of centres of expertise, rare disease units, specialised experts and primary healthcare providers ensures that know-how is transmitted to the place where patient care is provided. The centres of expertise will provide training to all healthcare professionals. The fundamental principle in the care of rare disease will be that patients receive diagnosis and care in all parts of Finland, because knowledge is mobile and the expertise of a centre of expertise can be applied in several hospitals.

A database will be formed of the centres of expertise, and this database will be readily accessible by social welfare and healthcare professionals, as well as by rare disease patients, for information on expertise in Finland concerning each rare disease.

Cooperation between the centres of expertise and patient organisations is very important and the aim is to vigorously develop it. The centres of expertise will regularly collaborate with patient organisations, whose advice is already being used at the planning stage of the centres of expertise.

Each centre of expertise will set up a customer panel in cooperation with patient organisations. The panel will raise questions for discussion and provide supportive advice on the centre of expertise's general policies. Patient organisations will also participate in the regular evaluation of the activities of the centres of expertise. The implementation of social support activities also requires cooperation with organisations.

The proposed centres of expertise require a legal basis, and so the development of the necessary Finnish legislation must be addressed first. European-level centres of expertise will have to fulfil the criteria of the European Commission, to be released in early 2014. The Finnish centres must achieve the status of European Network of Centres of Expertise, essential for patient diagnosis and promotion of care as experts and patients move between nations.

Not only are there doctors and nurses at centres of expertise, but also other professionals such as physical therapists and social workers. The centres cover expertise related to diagnostics, medical care, rehabilitation and social services.

A regular consultation mechanism for the designation of the centres of expertise will be created between the special areas of responsibility and the Ministry of Social Affairs and Health. The designation of both national and EU-level centres of expertise will commence in the form of mutual joint negotiation between the special areas of responsibility. The Ministry of Social Affairs and Health shall inform the hospital districts and third-sector parties dealing with rare diseases of the launching of this process of designating the centres of expertise, and invite them to put forward proposals for centres of expertise. The announcement inviting applications will not name specific disease groups; instead, the formation of the centres of expertise will at first capitalise on the existing expertise. To be able to take part in the designation process, the applicant must first engage in negotiations between the fields of specialisation and hospital areas of responsibility in order to ensure broad support for the establishment of the centre of expertise. When applying for the establishment of a centre of expertise at the EU level, the proponent shall describe how it intends to comply with the European Commission's criteria for centres of expertise. The establishment of a centre of expertise may be proposed by the administration of a university hospital management, by specialised senior physicians, by experts in rare diseases or by third-sector parties based on the outcome of the negotiation process. An individual centre is required to cover a distinct group of rare diseases; a national centre will be required to cover one rare disease. At a later stage, centres of expertise may also be established through an invitation process: if a need arises in Finland for a Centre of Expertise for a specific disease group, the Ministry of Social Affairs and Health will launch an application process for such a centre of expertise and commence negotiations with likely units.

The centres of expertise shall established under long-term structural, strategic and financial plans, so that long-term development opportunities are assured. The period of time shall be specified in accordance with EU regulations and recommendations. When designating centres of expertise on the basis of invitation, the representatives of all hospital districts, special fields and relevant patient organisations shall be consulted. The designation of the centres of expertise shall be prepared by the coordinating centre for rare diseases , whose executive board includes representatives of both university hospitals and patient organisations. The centres of expertise shall be appointed by the Ministry of Social Affairs and Health.

Goals:

- To identify the legislation necessary for the establishment of the centres of expertise.
- To begin establishing centres of expertise for rare diseases in Finland during the programme period.
- Some of the centres of expertise will have a strategy in place for fulfilling the European Commission's criteria for centres of expertise.
- All levels of healthcare will be informed of the role and availability of the centres of expertise for rare diseases and the centres will disseminate their expertise to all levels of social welfare and healthcare.

Indicators: Presence of established centres of expertise for rare diseases; Presence of database of centres of expertise; Numbers of referrals to centres of expertise

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

Schedule: Determining of necessary legislation in 2014; more detailed planning and preparation of the centres of expertise in 2014; announcement seeking applications and launching of negotiations in 2015

Preconditions: Implementation of the legislation, financial resources for the establishment of centres of expertise

3.3.4 Proposed measure 8: Increased training

More rare diseases training for social welfare and healthcare professionals.

According to the 2009 survey by EURORDIS, the European rare diseases umbrella organisation, correct diagnosis in cases of rare diseases is achieved faster when the initial diagnosing physicians takes into account the possibility of a rare disease as the cause of the symptoms. One of the essential means to increase awareness of rare diseases among diagnosing physicians and other healthcare personnel is to offer more training on rare diseases. Such training should be provided both during basic education and as continuous upgrading for those already employed in this sector.

Education on rare disease is already integrated to some extent in basic medical education on genetics. However, due to lack of resources, not all the university faculties of medicine include a comprehensive genetics module.

The steering group proposes that rare disease concepts and issues be included in the university curricula for medicine and other healthcare and social welfare professions. The curricula should ensure that the student understands the different kinds of rare diseases, their potential presence within groups commonly diagnosed diseases, the means of dealing with them, where to direct patients, and the importance of the correct diagnosis when the presence of a rare disease is suspected.

To ensure that rare disease patients receive comprehensive provision of treatment and support, training in rehabilitation must be integrated in all channels of the healthcare and social welfare training system. Continuing education on rare diseases must also be provided, throughout the social welfare and healthcare system.

Units and centres of expertise for rare diseases will provide training for healthcare professionals at different levels, as well as for personnel serving in rehabilitation and social services. The Finnish Association of

Genetics Physicians has already decided to compile a training package targeted at primary healthcare and specialised medical professionals.

All training in the rare disease area should make use of the existing experience-based training networks. In Eastern Finland, experience-based training is already fully integrated in the education programme, and students obtain first-hand knowledge of rare diseases alongside their theoretical studies. Experience-based lecturers will be meticulously trained, and the costs that educational institutions incurred in connection with experience-based training shall not include the lecturer's travel costs.

Goals:

- Social and healthcare professionals' knowledge of rare diseases is enhanced.
- Healthcare, social services, and rehabilitation professionals dealing with cases of rare diseases obtain updated knowledge about rare diseases.

Indicators: Emphasis on rare diseases in social and healthcare training; Training associated with rare diseases targeted at social welfare and healthcare personnel.

Responsibility: Ministry of Education and Culture, universities and universities of applied sciences, responsible for emphasising rare diseases in social and healthcare degree programmes; Units for rare diseases and centres of expertise, responsible for courses for social welfare and healthcare professionals

Schedule: Preparation of the concept of rare diseases for inclusion in social welfare and healthcare studies, in 2014- 2015; Annual training events starting in 2014

Preconditions: Approval from universities and applied sciences educational institutions, for modification of their study programmes

3.3.5 Proposed measure 9: Promoting the availability of rare disease drugs

Promotion of the availability of drugs used in the treatment of rare diseases and of equivalent Substitute drugs.

Given the low numbers of patients and high costs of developing new drugs, the pharmaceutical industry expresses little willingness to invest in research and development of drugs for the treatment of rare diseases. Specific support measures are required.

Lacking economies of scale, rare disease drug treatments are usually more expensive than those for more common diseases. Rare disease patients generally need drugs for long periods, often for their entire lives. Since rare disease patients have the right to the same standard of care as other patients, authorities in various countries have developed incentives for biotechnology and pharmaceutical enterprises to participate in the research, development and marketing of rare disease drugs.

In 1999, the European Union adopted a common policy on rare disease drugs. EU regulations determine that a product can be deemed to be a rare disease pharmaceutical if it targets a disease affecting at maximum five persons out of ten thousand. The disease must be life-threatening or chronically life-complicating. If the disease is less severe, it is still possible to have rare disease status recognised for a drug on the grounds that the low consumption is insufficient to compensate its development costs. A further condition of designation is that the product will be either the only drug available for treating patients afflicted by the rare disease, or that it can be expected to yield significant improvement in outcomes when compared to existing therapies.

Finland supports the incentive system for the development of rare disease drugs. The European Commission grants sales permits for rare disease drugs with exclusive rights for 10 years, during which generic drugs will not be awarded permits unless they would result in major improvements in treatment. Sales permits are granted at the European level, but each country determines its own standards of eligibility for patient reimbursement. For any drug to be designated as reimbursable in Finland it must be demonstrated that its use offers therapeutic value, that such treatment is necessary, and that it is economically efficient. Eligibility as "special reimbursability" may be granted to medicinal products for the treatment of persons afflicted by severe and chronic diseases, listed in the relevant decree issued by the Finnish government. To achieve the status of special reimbursability, the proponent must also demonstrate that the product has a reasonable wholesale price. The decision-making process regarding special reimbursability also considers the overall funding available for this purpose.

The availability of rare disease drugs in Finland is quite good and the same or better level of availability should be continued in the future. The EU's Patient Directive includes the obligation of mutual recognition of prescriptions, meaning that a prescription written in Finland can be used to obtain the prescribed drug elsewhere in Europe, and that prescriptions from other EU countries will be filled in Finland if the sale of the drug in question is permitted here.

Almost all of the rare disease drugs sold in Finland and used in the treatment of outpatients are reimbursable. Since rare disease drugs are expensive, they should continue to be reimbursable in the future. However in some cases, due to lack of clarity in the responsibility for funding, individual patients experience significant costs for medicines. The working group addressing the matter of comprehensive reform of the drug reimbursement system is thus of the opinion that the responsibility for costs and reimbursement of rare disease drugs through health insurance deserves closer examination (source: Final report of the Working party for the Development of the Drug Reimbursement System, Ministry of Social Affairs and Health, Reports and Memoranda, 2012: 33).

One problem is that certain drugs normally intended for the treatment of common diseases have also been demonstrated effective in the treatment of rare diseases, through credited research, yet the drugs have still not been designated reimbursable as rare disease drugs. The root causes and extent of this particular problem should be explored. In particular, the steering group proposes that the Ministry of Social Affairs and Health should assume national and international leadership in promoting extended indications for the use of commercially available drugs in treating rare diseases, where there is sound scientific evidence of the drug's effectiveness.

If a drug does not have a sales permit in Finland, the Finnish Medicines Agency (FIMEA) may grant a special permit for a medicinal product for the treatment of an individual patient or group of patients, for up to one year. Reimbursability for the drugs delivered via such special permits may then be requested by the user himself, by the pharmacy on behalf of the patient, or by the wholesaler or pharmaceutical company. Rare disease patients, families and doctors need more information and advice about the conditions for issue of these special permits, as well as regarding the reimbursement of medications delivered under a special permit.

The availability and reimbursement of non-pharmaceutical nutritive preparations also presents problems. Nutritive preparations and related products are vital for some rare disease patients, but not all of the preparations have been included in the reimbursement system. The identification of eligible severe and chronic diseases is determined by government decree, and the costs arising from the use of clinically-prescribed nutritive preparations in treatment is then reimbursed at the levels of 35% or 65%. As an example of the difficulties, the decree does not cover nutritive preparations used to treat difficult and rare cases of epilepsy. The decree should be amended to incorporate all those difficult diseases whose treatment with nutritive preparations is confirmed by sound research-based evidence. In severe cases of absorption deficiency, only 35% of the costs of nutritive products are reimbursed. The steering group proposes that the nutritive preparations used in such treatments be reimbursed at a higher rate.

Goals:

- To ensure the availability of rare disease drugs.
- To conduct a survey of the drugs used in the treatment of rare diseases.
- To promote extension of the indications for commercially available drugs to permit their use for rare diseases, where the effectiveness of the pharmaceutical treatment has been scientifically demonstrated.
- To extend the government decree on reimbursement of nutritive preparations to include all difficult diseases, where the efficacy of clinical-prescribed treatment using the nutritive preparation is demonstrated by research-based evidence.
- To increase the reimbursement for nutritive preparations for patients afflicted by severe nutrient absorption deficiency to 65% of costs.
- To increase knowledge of the terms and means to apply for reimbursement of costs of a medical product, delivered under special permit.

Indicators: The number of reimbursable rare disease drugs and the portion of these of all rare disease drugs used in outpatient treatment; Knowledge of criteria and procedures for reimbursement for a medical product delivered under special permits (patient survey)

Responsibility: Ministry of Social Affairs and Health, for amendments to the Health Insurance Act, and for providing information on applications for reimbursement for medical products delivered under special permits; Social Insurance Institution of Finland, for the survey of drugs used in the treatment of rare diseases

Schedule: Amendments to legislation in force in 2017

Preconditions: Changes in reimbursement legislation

3.4 KNOWLEDGE SHARING AND COORDINATION OF EXPERTISE

One of the main problems concerning rare diseases is the fragmentation of the associated knowledge and expertise. Healthcare professionals, rare disease patients and social welfare personnel all suffer their own difficulties in finding the existing knowledge and expertise, because the resources have never been compiled. Moreover, knowledge of rare diseases is incomplete in many areas, and this delays diagnosis and access to appropriate care.

Finland lacks a body that would take responsibility for coordinating expertise and knowledge on issues related to rare diseases. In addition to the collecting and disseminating knowledge, such a body would attend to the administration of measures for rare diseases and the monitoring of their implementation, to link rare disease expertise at different levels of the healthcare system as well as to serve as a centre in the debate between the various interest groups and to maintain international cooperation networks.

3.4.1 Proposed measure 10: A national rare disease coordination centre

A national rare disease coordination centre should be established.

A national coordination centre should be established, to serve as the administrative link between primary healthcare, the various specialised sectors, rare disease units, centres of expertise, the third sector, the Finnish Social Insurance Institution, social welfare services, and rare diseases programmes and centres elsewhere in the EU. The coordination centre should be set up within the structure of the National Institute for Health and Welfare (THL). Its executive board should include representatives of the healthcare and social welfare sectors, as well as the centres of expertise, the Social Insurance Institution, the third sector and patient organisations. As necessary, the coordination centre should organise a rare disease national forum, to provide statements of constructive advice and recommendations on current issues relating to rare diseases. Such forums should be held at least once a year. The coordination centre board should operate on the basis of the statements received from the forums. The national forum should include broad representation of all rare disease organisations. The coordination centre will require staffing with a coordinator and expert.

The national coordination centre's main task should be the development of the Rare Diseases National Programme proposals, and development of the service systems in a manner as effective as possible for rare disease patients.

The coordination centre should be responsible for the National Programme's division of responsibilities, its progress, monitoring, assessment, implementation of measures, and setting of new goals. The coordination centre should bear the responsibility for initiating the procedures and strategies for designating of the centres of expertise. In particular, it should coordinate the process of establishing the centres of expertise and propose the designations of centres for final decision by the Ministry of Social Affairs and Health.

The national coordination centre's other tasks would be the development of an independent and wide-ranging system of education and training, the coordination of the preparation of rare disease clinical treatment guidelines, as well as coordination of expertise and knowledge in cooperation with the centres of expertise, rare disease units, and organisations. The coordinating centre would also coordinate cooperation in international research. The national coordinating centre would belong to European bodies for and relay knowledge from international expert forums.

Because of the expectation of significant reductions in resources over the coming years, the establishment of the coordination centre at the THL is not possible at this stage, but should be identified as part of a long-term plan. For the present, the Steering Group recommends the establishment of a free-form smaller group for the implementation of the programme, to meet several times during 2014. The group should include

representatives from the Ministry of Social Affairs and Health, THL, the Social Insurance Institution, all university hospitals, local and regional Authorities, the Network for Rare Diseases, Orphanet, and HARSO. A survey would be conducted at the end of 2014 to see whether the conditions are suitable for the permanent establishment of the national coordination structure. It is further proposed that the university hospitals negotiate among themselves as to which one of them could temporarily take on the responsibility for the coordination of rare disease clinical activity.

Goals:

- To assemble and coordinate Finland's rare disease expertise and resources in a systematic, effective and continual manner.
- To have a centre responsible for national administration related to rare disease programmes and services.
- To receive the advice of the national rare disease forum on a regular basis.

Indicators: The decision to establish a national centre; The hiring of a coordinator and an expert; A database of knowledge relating to rare diseases has been compiled

Responsibility: Ministry of Social Affairs and Health, THL, university hospitals

Schedule: The establishment of the coordination centre and meeting of the national forum, to guide the implementation of the programme as of 2015

Preconditions: Financial resources for the establishment of the centre

3.4.2 Proposed measure 11: Systematic collecting and dissemination of information

A comprehensive rare disease database should be available on the Internet, in both Finnish and Swedish languages. Rare disease patients, their families, and social welfare and healthcare professionals should be able to find online help for their information needs. A rare disease seminar should be held annually in cooperation with patient organisations.

There is already an abundance of information on rare diseases available on the Internet, but the knowledge is fragmented and incomplete. There are also a number of rare diseases for which there is no information in Finnish or Swedish. The proposed national coordination centre (Measure 10) would be responsible for the systematic collection and online publication of data and information on rare diseases. The centre would also collect information on the different rare diseases units, centres of expertise, and on rare disease patients, in cooperation with patient organisations. The centre would also collect information concerning the treatment, rehabilitation and social services for rare disease patients, again in cooperation with patient organisations. Patient organisations are important sources of information for rare disease patients and their families, and this should be taken into account when disseminating the information.

A comprehensive Finnish rare disease Internet-based database can be accomplished by translating the existing rare disease patient and treatment network service, Orphanet, into Finnish and Swedish. Orphanet is a cooperation project involving about 40 countries. Each country’s Orphanet group collects information on specialised clinics, medical laboratories, research projects and patient organisation operating in their own country. Finnish participation in Orphanet is currently managed by the Norio Centre. The information is

available free of charge. The creation of the national database should build on and avoid overlapping with Orphanet's existing work. The creation of a database must be agreed by the coordination centre for rare disease, the Ministry of Social Affairs and Health, and third-sector parties. The database must be updated on a continual basis and in this regard, the roles of all those who involved in work with rare diseases must be communicated.

In addition to the computerised database, there must be communication of broader information on rare diseases and the existing knowledge, both for the general public and for social welfare and healthcare workers. This kind of service is currently provided by third-sector parties, through help-lines and Internet Q&A services. The existing services provide good models. Another possibility is to establish a general level helpline at a rare diseases unit agreed by the special areas of responsibility.

The coordination centre would be responsible for organising the annual rare disease seminar. This seminar is organised as part of the national forum and encourages discussion of current issues in rare diseases. All parties dealing with rare diseases are invited to this seminar.

Goals:

- Information on rare diseases from research, healthcare and social welfare services are systematically collected and made available, including information on the supports for rare disease patients.
- Finland's Orphanet activities are taken into account when creating a rare diseases database in Finnish and Swedish.
- The actors in the field of rare diseases meet annually at the rare disease seminar.

Indicators: A database in Finnish and Swedish exists; Orphanet is continuously updated; organisation of annual rare disease seminars; rare disease patients can find the information they need (patient survey)

Responsibility: The coordination centre for rare diseases

Schedule: Continuous, as of the launching of the programme in 2014

Preconditions: None are perceived

3. HOLISTIC SUPPORT AND INTEGRATION OF RARE DISEASE PATIENTS

Once a rare disease patient has received the correct diagnosis and, where appropriate, even prior to this, the search for appropriate healthcare and social welfare services and rehabilitation is initiated together with the client, and where necessary with his or her family members. A common problem is the lack of an overall grasp of the task of improving a rare disease patient's quality of life. Rare disease patients and their families may also face a variety of challenges as users of ordinary services, e.g. maternity clinics, day-care centres or educational institutions. Instead of a diagnosis-based approach to the assessment of service needs, individual needs should be taken into account. The goal should that regardless of her rare disease, the individual can live a full life based on her own choices, and receive the appropriate care and rehabilitation in addition to any necessary psychosocial support.

Social services personnel do not always respond adequately to the needs of rare disease patients: information regarding the available social services may be inadequate, and the majority of rare disease patients and their families are dissatisfied with the quality of social services. There are also regional differences as regards access to social support.

Information on social security and services for rare disease patients is typically fragmented. The experience of administrative authorities in such diseases may be minimal. There is a lack of comprehensive study of the ability of social services personnel to respond to the needs of rare disease patients. Because of these problems, there is also a need for research and identification of the effectiveness of the current services regarding rare diseases.

The key objective in social welfare and healthcare legislation and in the development of the service systems is to strengthen the client's inclusion and involvement and to provide support in coping with everyday life. The point of departure in the organisation of social services and support is the client's needs, regardless of the underlying diagnosis.

The rehabilitation system is complex and fragmented. The organisers include public healthcare and social welfare institutions, education authorities, the Social Insurance Institution and employment authorities, which all have differently defined target groups and objectives. Rare diseases are often diagnosed in early childhood and patients may need rehabilitation services from a number of different providers over the course of their life and the stages of the disease.

Broad cooperation is required in the planning of care and rehabilitation, to ensure that rehabilitation measures are applied at the right time and that the individual can achieve the best possible functional and working capacities in their personal situation. Timeliness in directing rare disease patients to the benefits of different rehabilitation services is difficult, and cooperation between the Social Insurance Institution and the service providers in the implementation of rehabilitation is not always sufficient. Experts representing different areas do not always meet.

On the one hand, healthcare professionals often lack information and expertise concerning rehabilitation. The dissemination of information from the Social Insurance Institution’s rehabilitation services is not always sufficient. On the other hand, the healthcare professionals’ knowledge on the rehabilitation needs of rare disease patients, including on new forms of treatment, does not reach the Social Insurance Institution's rehabilitation service planners in sufficient depth. The Social Insurance Institution does not have integrated units dealing with relevant information or treatment of rare diseases, and its attempts to consult with the necessary parties have not always met with success.

Rare disease patients and their families are clearly familiar with their own situations and often know what kind of services are necessary. Patient organisations have a great deal of information on rare disease patients’ experiences and needs. However this information is not adequately taken into account in the planning of social welfare and healthcare services. The possibility for rare disease patients and their families to access psychosocial support, along with their clear involvement in the planning of treatment and services,

would help in the implementation of a more comprehensive system of care and support. The role of customer and patient organisations as providers of peer and other psychosocial support is currently being reinforced. In particular, Finland's Slot Machine Association is providing funds for development of this role.

3.5.1 Proposed measure 12: Development of social support and rehabilitation

There should be promotion of awareness of the rare disease patient's needs for rehabilitation, improvement in predicting their needs, in rehabilitative and social services planning, and in comprehensive support for the patient. Such promotion and improvement must involve social welfare and rehabilitation services, supported by closer cooperation with centres of expertise, rare disease units, organisations, the Social Insurance Institution, and municipal social services. Rare disease patients, and when appropriate their families, shall participate in planning of care, rehabilitation, and social welfare services.

The rare disease patient's rehabilitation needs should be taken into account through cooperation with the rare disease unit and the centres of expertise, to ensure that both physical and psychological rehabilitation support the patient's treatment and the overall objectives for the individual, in the best possible way. There should be activation of joint negotiations between the person being rehabilitated, the care unit, the rehabilitation service providers and the social services. The centres of expertise are used to record the contacts with the rehabilitation parties, the Social Insurance Institution, the social services, and organisations, when planning the care, rehabilitation and social support for the patient. The assessment of the opportunities for rehabilitation is now the organisational responsibility of the Social Insurance Institution. This unit or another representative must thus be represented in the organisational development of the rare disease units. Each rare disease patient requiring multi-profession expertise and broad support shall be appointed a person who is responsible for drawing up an appropriate care and rehabilitation plan and for keeping in touch with social welfare services.

Rehabilitation is often demanding and the treatment of diseases is continuously evolving. Without proper exchange of information, patients do not receive the appropriate services and guidance, and rehabilitation is not implemented in a manner that matches their true needs. The centres of expertise and rare disease units shall operate as the coordination units for the planning of care and rehabilitation, together with the Social Insurance Institution's rehabilitation group and the social welfare services. There will be also be a coordinating body for rehabilitation at the primary healthcare level. The party administering the care for the rare disease patient shall inform the municipality and the rare disease patient of the needs to be considered during the rehabilitation process.

Ever closer cooperation between the various parties is important, to ensure there is targeted training for the social welfare and healthcare workers involved and for the persons making decisions in rehabilitation cases, so that the patient can be directed to the appropriate services and assistance. Rare disease patients themselves must be given opportunities to receive information about all stages of their care and service process from all parties, as well as to participate in and influence the process. An information dissemination service needs to be provided concerning the social security and service system, as well as the rehabilitation services. Rare disease units and centres of expertise should have a data bank on rehabilitation and social welfare services, accessible by other healthcare units. Information on the social security system and rehabilitation should be provided by rehabilitation workers at hospitals, by social and municipal welfare workers and services, and particularly by service personnel directly in contact with the disabled. For example, information and support is available for customers and for services personnel working with the disabled, from the electronic manual for services which is part of the THL's Internet-based system.

In 2013 a new section of the manual was launched, dedicated to services for children and young people. Similarly to this, rare diseases should also be taken into account as an independent section. Public health organisations, patient organisations, and disabled persons' organisations have drawn up a joint social

security guide concerning the chronically ill and disabled, which is at the disposal of the organisation's members and for workers who provide advice and guidance. The national rare disease coordination centre should gather the existing information in one place, and disseminate it throughout the social welfare and healthcare services.

When developing cooperation and information dissemination related to rehabilitation and social welfare services, the strategy will be to take advantage of ongoing initiatives under the Ministry of Social Affairs and Health, aimed at reforming the social welfare and healthcare service structures, the Social Welfare Act and legislation on services for the disabled, as well as the development of informal care and multi-profession rehabilitation.

A proposal has been made that the appointment of so-called “personal” social workers for customers requiring more than one social welfare service should be included in the new Social Welfare Act. This operational model would ensure that the individual needs of rare disease patients would be better taken into account, contributing to better planning of service packages. The needs for support to the families of rare disease patients should also be examined.

The peer support provided by patient organisations is very important, however, the Finnish Slot Machine Association (RAY) no longer provides sufficient support to these patient organisations. The RAY grants to patient organisations should increase, ensuring the preconditions for these organisations to continue and increase peer support.

Goals:

- Cooperation between health services, social welfare services, and rehabilitation services becomes closer and more systematic, so that
- the rare disease patient's needs are taken into account in a comprehensive manner.
- Rare disease patients and their families become engaged in the planning of the service packages and get to influence the organisation of services.
- Multi-profession expertise for demanding rare disease patients are provided with service plans.
- Up to date information on rehabilitation and social services is made readily available and this is disseminated to social welfare and healthcare professionals as well as to the rare disease patients.
- New information on rare diseases is disseminated to the Social Insurance Institution, patient organisations, insurance companies, and to both the organizers and providers of social welfare and healthcare services.
- The possibility for patient organisations to implement peer support is reinforced.

Indicators: Link between healthcare, social welfare and rehabilitation services when planning rare disease patients’ treatment and rehabilitation (centres of expertise reports); Completed service plans (Patient survey); Compiled data bank covering social welfare and rehabilitation services; Development of Finnish Slot Machine Association's grants to patient organisations

Responsibility: The national coordination centre, rare diseases units, centres of expertise, SII, municipalities’ social services; THL Disabled persons’ service manual’s rare disease section

Schedule: Following the establishing of the national coordination centre for rare diseases, the rare disease units, and the centres of expertise, starting in 2016

Preconditions: Designation of the coordination body; cooperation to supplement the manual for disabled persons

3.5.2 Proposed measure 13: Empowerment and involvement of rare disease patients

Rare disease patients, their families, and patient organisations to become more involved in the decision-making and planning of services concerning rare diseases.

According to the EU Council's recommendations (2009, see Appendix 1 to this document), patients and patient organisations should be consulted in regards to policies impacting on rare diseases. The EUCERD quality criteria for national centres of expertise identify the same priority (Appendix 2).

The centres of expertise must collaborate with patient organisations in such a manner that the patients' perspectives are taken into account in the measures proposed. Empowering patients and their families and involving them in service planning and increasing of patient organisation influence must be promoted. This is also the starting point when developing social welfare and healthcare legislation and the service system.

Individual rare disease patients and their families must receive adequate information about care, rehabilitation, social welfare and support services. Both the centres of expertise and rare disease units must operate as readily accessible, “low threshold” information centres. There must be comprehensive collection of information in cooperation with patient organisations and the entire healthcare service, social welfare service and rehabilitation service network. The information in question will be available through computerised access to the coordination centre's databank, as well as via help-lines.

Experts from the centres of expertise should visit rare disease peer support groups in person, to give and receive information. Reciprocity is important, so that rare disease patients and their families can contribute effectively to the development of care, rehabilitation, and other services. Rare disease patients and their families are necessary sources of empirical knowledge. The knowledge they contribute will be used in rare disease training modules for healthcare professionals.

The main channels of influence for rare disease patients and their families are the patient organisations. Both the individual organisations and the national umbrella groups of all organisations offer a wide range of services. However their role in the preparation, development and evaluation of services is yet sufficient, because there is a lack of a clear participation model. One of the weaknesses is that there is not enough information on the current function of patient organisation in cooperation with university hospitals, and so a first task is to chart this area.

The participation of patient organisations should be implemented at three different levels.

1) The representatives of the various organisations should participate in the planning process when centres of expertise are being created.

2) Customer panels, including patient organisations, should evaluate operation and make development proposals.

3) The rare disease forum, organised at least annually by the national coordination centre for rare diseases, should involve broad participation by the representatives of patient organisations.

The executive board of the coordination centre must itself include adequate representation from patient organisations, meaning at least two organisations, representing the situations of different diagnosis groups.

In addition to these measures, the Advisory Board of the National Council for Disabled Persons and Rehabilitation Affairs should advise on preventing overlap of efforts and ensuring that all aspects of expertise are involved in planning the service pathways and operation of the centres of expertise.

Goals:

- Rare disease patients and their families as well as the professional staff receive sufficient knowledge regarding social welfare and healthcare services.
- Rare disease patients and their families are increasingly involved in the planning of the service pathways.
- The involvement of patient organisations in influencing the development of service structures increases.
- Patient organisations participate in the planning, assessment and operational development of centres of expertise.
- Patient organisations participate in the evaluation and development of rare disease services.

Indicators: Rare disease patients' and their families' knowledge of existing social welfare and healthcare services (Patient survey); Patient organisations' representatives involved in the planning working party centre of expertise, in the national rare disease forum, as members of the executive board of the national coordination centre, and as members of customer panels of the centres of expertise.

Responsibility: The coordination centre, for coordination of dissemination of information, establishing the executive board, operating the national forum, and for preparation and designation of centres of expertise; Centres of expertise, for customer panels

Schedule: 2015−2017

Preconditions: Obstacle-free; the establishing of expertise of the centres of expertise

4 COORDINATION, MONITORING, AND EVALUATION OF THE NATIONAL RARE DISEASES PROGRAMME

The first measure that must be carried out under Finland's National Programme for Rare Diseases is the creation of a rare disease coordination centre, which underlies all other measures. It is proposed that this be done as soon as the programme period begins. The centre can then proceed to coordinate the implementation of the National Programme's other measures. The establishment of the centre requires steps by the Ministry of Social Affairs and Health, including the empowerment of the centre to coordinate the implementation of the National Programme.

The other priority measures of the National Programme are as follows: legislative recognition of the special measures required for treatment of rare diseases; clarification of the rare disease treatment pathway; establishing of rare disease units at university hospitals; promotion of access to and reimbursement for rare disease drugs; development of social service support and rehabilitation.

The results from the national rare disease forums will influence and guide the national coordination centre's activities and decisions. The forums will include representatives from university hospitals, patient organisations, municipalities, social services and the Social Insurance Institution. The forums provide opportunities for dialogue with the interest groups, and guide and assist in assessing the implementation of the National Programme. The forums are organised by the coordination centre. The forum will meet as necessary but at least once a year there will be a meeting in conjunction with the Rare disease Seminar.

The coordination centre coordinates and monitors the implementation of each proposed measure in the National Programme, in accordance with the time scheduling. If the centre cannot be established at the beginning of the programme period, the Ministry of Social Affairs and Health should appoint a temporary steering group to guide the coordination of the National Programme.

Finland's National Programme for Rare Diseases should proceed as follows during the period 2014-2017:

Year	National Rare Diseases Programme
2014	Planning of the National Programme and ensuring of the preconditions
2015	Implementation of the National Programme and interim assessment
2016	Implementation of the National Programme
2017	Final assessment of the National Programme for the current programme period, planning of the next period

The implementation of the National Programme should be assessed using measure-specific indicators and the key indicators recommended by EUCERD (Appendix 2). In addition to the EUCERD criteria, new national indicators for monitoring the National Programme and the effectiveness of its goals are required, and the development of these indicators is one of the tasks of the coordination centre. Seeing as the current situation is in many respects unknown, the application of indicators will initially be difficult. The coordination centre must therefore conduct a survey of the current situation of rare disease patients, as the first step in monitoring the impact of the National Programme on future developments. Key themes in developing the indicators include the character of feedback from professionals and patient organisations,

objective information on delays in diagnoses, and data on changes in care practices, on the status of marginalised groups, the intactness of care chains, and the presence of continued, coordinated expertise. The satisfaction of rare disease patients and their families should be determined on a regular basis.

The steering group proposes that an interim assessment be performed by the national coordination centre at the end of 2015 using measure-specific indicators and the key indicators as defined by EUCERD. The interim assessment will serve in part to verify the functionality of the set of indicators and further develop them. When the programme period ends in 2017, consideration should be given to obtaining an external international assessment of the National Programme, with the results from this to be taken into account in setting goals for the new programme period.

5 SUMMARY OF MEASURES

	Measure	Key content	Goals	Obstacles /Priorities
1	Uniform definition of rare diseases and legislative recognition of special measures	EU's definition is adopted and special measures are recognised in legislation	Increase in cooperation at EU level; Recognition of special measures in legislation	Political support **Priority**
2	Register of rare diseases	Creation of register of rare diseases in HILMO, applying ICD-11 disease classification	Collecting of research data is facilitated; Cooperation increases both in Finland and internationally	Adoption of ICD-11 disease classification
3	Targeted funding and research on rare diseases	State research funding for research on rare diseases and Finnish disease legacy. Research programme under the Academy of Finland	Strengthening of research funding; Establishing of research cooperation and database; Promotion of research on effectiveness and functionality of service system and services; information obtained is then used in development work	Definition of responsible parties, acceptance of resonsibility
4	Strengthening international research cooperation	Application for E-RARE Project membership	Cooperation in collecting patient data; Sufficient research funding to enable participation in E-RARE Project	Sufficiency of national funding
5	Clarification of rare disease patient-care pathways	Streamlining of rare disease patient care pathways, including diagnosis. Personnel at all levels know the pathways. Issue of "care passports".	Rare disease patients' care pathway becomes clearer; All healthcare levels familiar with how to diagnose and implement appropriate care; Care passport.	Implementation of rare disease units and centres of expertise; Development of suitable computerised systems **Priority**
6	Creation of rare disease units at university hospitals	Each university hospital establishes a rare disease unit, with a rare disease coordinator, coordinating nurse and multi-profession working group	The unit takes responsibility for the patient's diagnoses and care in cases where there is no clear special field; Horizontal cooperation is facilitated; Transfer from paediatric clinic to adult ward is smoothed; Professionals are allocated time to work with rare disease cases, as necessary for diagnosis and care.	Allocation of resources to university hospitals **Priority**
7	Creation of rare disease centres of expertise	Finland establishes EU-compliant rare disease centres of expertise and national disease-specific centres of expertise	This begins with establishing of centres of expertise in Finland; Some of the centres of expertise have strategies fulfilling European Commission criteria; All levels of healthcare and social welfare informed about role of the centres, the centres disseminate their expertise	Financial resources
8	Increased training	More rare disease training for social welfare and healthcare professionals.	Healthcare professionals' knowledge of rare diseases increases; Professionals dealing with rare diseases are up to	Approval of changes to curricula by universities and universities of applied sciences

			date on rare diseases information	
9	Promotion of availability of rare disease drugs	Promoting of availability and reimbursability of drugs for treating rare disease	Ensuring access to drugs; Drugs to be eligible for reimbursability when used to treat rare diseases; Knowledge of reimbursement system increases	Changes in reimbursement legislation **Priority**
10	Establishment of a national coordination centre for rare diseases	Finnish expertise in rare diseases is compiled and coordinated systematically	The centre coordinates the national programme on rare diseases; Rare disease forum meets regularly	Financial resources **Priority**
11	Systematic collecting and sharing of information	Comprehensive rare disease database is created Information is provided via helpline. Rare disease seminar held annually	Rare disease related knowledge and information on research, healthcare and social services, and on support needed by rare disease patients is collected and offered systematically; Orphanet's operation is taken into account; Rare disease field meets annually at rare disease seminar	Obstacle-free
12	Development of social support and integration	Improved information on services and reinforced cooperation between different parties	Cooperation between healthcare, social welfare and rehabilitation services reinforced; Service plans for cases requiring multi-profession skills; Up to date information on rehabilitation and social services	Designation of coordination party **Priority**
13	Empowerment of rare disease patients	Rare disease patients and patient organisations participate in rare disease decision-making and planning of appropriate services	Sufficient information about services; Inclusion of rare disease patients when planning service pathways; Patient organisations have opportunities to influence service structure development; Patient organisations participate in assessment and development of services related to rare diseases	Obstacle-free

LIST OF ABBREVIATIONS USED

ERC	European Research Council
EU	European Union
EUCERD	European Union Committee of Experts on Rare Diseases (replaced by EC Expert Group on Rare Diseases as of 2014)
EUROPLAN	European Project for Rare Diseases National Plans Development
EVO	Special State Subsidy for Research in University Hospitals
HILMO	Care Registers for Social Welfare and Healthcare
HUS	Hospital District of Helsinki and Uusimaa
HYKS	Helsinki University Central Hospital
ICD	International Standard Classification of Diseases and Related Health Problems
IRDiRC	International Rare Diseases Research Consortium
FSII (Kela)	Finnish Social Insurance Institution
KYS	Kuopio University Hospital
MEC	Ministry of Education and Culture
OYS	Oulu University Hospital
RAY	Slot Machine Association
MSH	Ministry of Social Affairs and Health
THL	National Institute for Health and Welfare
TAYS	Tampere University Hospital
HWI	National Institute for Health and Welfare
TYKS	Turku University Hospital

ANNEX 1: The Council of the European Union's recommendation on an action in the field of rare diseases

Note: the comments inserted in italics indicate the sections and measures proposed in the current document, which respond to the recommendations of the Council of the European Union

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 FOR 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:

- ***The entire programme***

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;

- ***The entire programme***

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

- ***Proposed measure 10: The national coordinating centre for Rare diseases***

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

- ***The entire programme; Priority measures summarised in the abstract and summary***

d) take note of the development of guidelines and recommendations for the elaboration 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) (1) selected for funding over the period 2008-2011 in the first programme of Community action in the field of public health.

- ***The period is no longer timely, apart from this the target applies to the whole programme***

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 five per 10,000 persons;

- ***Proposed measure 1: A uniformed definition of rare diseases and the recognition of their required specific measures***

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 national healthcare and reimbursement systems based on the ICD while respecting national procedures;

- ***Proposed measure 2: Register of rare diseases***

4. Contribute actively to the development of the EU's 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;

- ***Proposed measure 11: Systematic collecting 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, registers and databases, whilst being aware of an independent governance;

- ***Proposed measure 2: Register 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;

- ***Proposed measure 3: Research funding and research programmes targeted at rare diseases / Proposed measure 4: Strengthening 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 cooperative approaches to the complementarity addressed through national and Community programmes;

- ***Proposed measure 3: Research funding and research programme targeted at rare diseases***

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

- ***Proposed measure 3: Research funding and research programmes targeted at rare diseases / Proposed measure 4: Strengthening international research cooperation***

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

- ***Proposed measure 3: Research funding and research programmes targeted at rare diseases / Proposed measure 4: Strengthening 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.

- ***Proposed measure 4: Strengthening international research cooperation***

IV

CENTRES OF EXPERTISE FOR RARE DISEASES AND EUROPEAN REFERENCE NETWORKS

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

- ***Timetable later; Proposed measure 6: Unit for rare diseases in university hospitals / Proposed measure 7: Centres of expertise for rare diseases***

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

- ***Proposed measure 7: Centres of expertise for rare diseases***

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

- ***Proposed measure 5: Clarification of the rare disease patients' care pathway / Proposed measure 6: Rare disease unit for university hospitals / Proposed measure 7: Centres of expertise in rare diseases***

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

- ***Proposed measure 5: Clarification of rare disease patients' care pathway / Proposed measure 6: Rare disease unit for university hospitals / Proposed measure 7: Centres of expertise for rare diseases***

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

- ***Proposed measure 5: Clarification of rare disease patients' care pathway / Proposed measure 11: Systematic collecting and sharing of information***

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

- ***Proposed measure 7: Centres of expertise for rare diseases***

V
COLLECTING 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;

- ***Proposed measure 10: National coordination centre for rare disorders / Proposed measure 11: Systematic collecting 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;

- ***Proposed measure 8: Increased training***

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

- ***Proposed measure 8: Increased training***

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

e) the sharing of 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;

- ***Proposed measure 9: Promoting the availability of rare disease drugs,***

VI
EMPOWERMENT OF PATIENT ORGANISATIONS

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

- ***Proposed measure 13: Promotion of involvement of rare disease patients***

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;

- ***Proposed measure 13: Promotion of involvement of rare disease patients***

VII
SUSTAINABILITY

20. Together with the Commission, 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;

- ***The entire programme***

ANNEX 2: EUCERD criteria for national centres of expertise

- Capacity to formulate and adhere to instructions pertaining to good diagnosis and care practices.
- A quality management system ensuring quality care is in place and compliance with national level and European level legislation; participation, where appropriate, in internal and external quality programmes.
- Competence to propose use of quality-of-care indicators for use within the territory and to assess the effectiveness of care and patient satisfaction.
- High level of expertise and experience, evidenced by, for example, the annual number of referrals and consultations, peer-reviewed publications, grants, research posts, and education and training activities.
- Sufficient capacity for managing rare disease patients' care and for offering expert advice.
- Participation in high-level and latest research.
- Capacity to participate in data collection for clinical studies and public healthcare work.
- Capacity to participate, if necessary, in clinical trials.
- Application of multi-profession approach, where necessary, so that medical, healthcare, psychological and social needs are treated as a whole (e.g. committee on rare diseases).
- Organisation of cooperation in order to ensure continuity of care from childhood through teen years and into adulthood, as necessary.
- Organisation of cooperation in order to ensure continuity of care in all stages of the disease.
- Contacts with other centres of expertise and cooperation with these on national, European, and international level.
- Contacts with potential patient organisations and cooperation with them.
- Appropriate arrangements for referring patients to receive care within the Member State or (where appropriate) in another EU country.
- Appropriate arrangements whereby offering of care is improved and especially the time spent on diagnosing is shortened.
- Taking into account of electronic health service applications (e.g. common electrical case management systems, expert systems for tele-expertise service provision and a common case database).