2013 REPORT ON THE STATE OF THE ART OF RARE DISEASE ACTIVITIES IN EUROPE

STATE OF THE ART OF RARE DISEASE ACTIVITIES IN FINLAND

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More information on the European Union Committee of Experts on Rare Diseases can be found at www.eucerd.eu.

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ACRONYMS

CAT - Committee for Advanced Therapies at EMA
CHMP - Committee for Medicinal Products for Human Use at EMA
COMP - Committee on Orphan Medicinal Products at EMA
DG - Directorate General
DG Enterprise - European Commission Directorate General Enterprise and Industry
DG Research - European Commission Directorate General Research
DG Sanco - European Commission Directorate General Health and Consumers
EC - European Commission
ECRD - European Conference on Rare Diseases
EEA - European Economic Area
EMA - European Medicines Agency
ERN - European reference network
EU - European Union
EUCERD - European Union Committee of Experts on Rare Diseases
EUROCAT - European surveillance of congenital anomalies
EUROPLAN - European Project for Rare Diseases National Plans Development
EURORDIS - European Organisation for Rare Diseases
FDA - US Food and Drug Administration
HLG - High Level Group for Health Services and Medical Care
HTA - Health Technology Assessment
IRDiRC – International Rare Diseases Research Consortium
JA - Joint Action
MA - Market Authorisation
MoH - Ministry of Health
MS - Member State
NBS - New born screening
NCA - National Competent Authorities
NHS - National Health System
PDCO - Paediatric Committee at EMA
RDTF - EC Rare Disease Task Force
WG - Working Group
WHO - World Health Organization
GENERAL INTRODUCTION

This document was produced by the Scientific Secretariat of the European Union Committee of Experts on Rare Diseases (EUCERD), through the EUCERD Joint Action: Working for Rare Diseases (N° 2011 22 01), which covers a three year period (March 2012 – February 2015).

The present report aims to provide an informative and descriptive overview of rare disease activities at European Union (EU) and Member State (MS) level in the field of rare diseases and orphan medicinal products up to the end of 2012. A range of stakeholders in each Member State/country have been consulted during the elaboration of the report, which has been validated as an accurate representation of activities at national level, to the best of their knowledge, by the Member State/country representatives of the European Union Committee of Experts on Rare Diseases. The reader, however, should bear in mind that the information provided is not exhaustive and is not an official position of the European Commission, its Agencies or national health authorities.

The report is split into six parts:

Part I: Overview of rare disease activities in Europe
Part II: Key developments in the field of rare diseases in 2012
Part III: European Commission activities in the field of rare diseases
Part IV: European Medicines Agency activities and other European activities in the field of rare diseases
Part V: Activities in EU Member States and other European countries in the field of rare diseases
Part VI: Activities at National level in each EU Member State and other European countries in the field of rare diseases

Each part contains the following description of the methodology, sources and validation process of the entire report, and concludes with a selected bibliography and list of persons having contributed to the report.

The present document contains the information from Parts II and V of the report concerning Finland. A list of contributors to the report and selected sources are in annex of this document. For more information about the elaboration and validation procedure for the report, please refer to the general introduction of the main report.
RARE DISEASE ACTIVITIES IN FINLAND

Definition of a rare disease
There is no official definition for rare diseases in Finland. At present the parties involved in the field of rare diseases normally use the common EU definition of no more than 5 in 10,000 individuals. In matters concerning orphan medicinal products Finland officially applies the same definition used in European Regulation on Orphan Medicinal Products. Finland has decided (for the draft plan) to use the EU definition of no more than 1 in 2,000 and severe/debilitating diseases.

National plan/strategy for rare diseases and related actions
There is currently no national plan/strategy for rare diseases, though the first steps have been taken in the process. Funding specifically focused on national plan related activities was applied for in late 2011 from the Ministry of Social Affairs and Health (this was accepted in 2012). A national advisory committee and a pilot have been appointed for the elaboration of the plan. During 2011 a nationwide survey was performed to identify centres who consider themselves as experts related to a rare disease or disease group. The Ministry of Social Affairs and Health decided to invite stakeholders in the field to become members of the steering committee, including hospital districts, governmental institutes like the National Institute for Health and Welfare, Väestöliitto, the Orphanet National Advisory Board, the umbrella organisation for rare diseases HARSO, Helsinki University Hospital, and the Harvinaiset Network for Rare Diseases to name representatives for a steering committee for elaborating the national plan. There will be a focus on establishing centres of expertise, with a step which will include patients before they reach the centres (i.e. early health care pathways to diagnostic processes). However, a healthcare reform is underway which may slow down the work on the plan. A meeting is planned in March 2013 to advance with the elaboration of the plan. The aim is to have the plan ready during 2013, the designation of expert centres will be a much longer process to follow.

Current expenditures for rare diseases fall within the general health system budget with additional ad hoc funding on the basis of rare disease projects.

Finland participated in a project (which ran from 2009 to 2010) focusing on cooperation possibilities between Nordic countries in the field of rare diseases. The project was supported by the Nordic Council of Ministers, and was entitled “Kartläggning av möjliga nordiska samarbetsområden anknutna till små och sällsynta diagnosgrupper” ("Report on possibilities for co-operation between the rare disease groups in Nordic Countries"). The goal of the project is to create recommendations for Nordic cooperation in all fields: medical, social, psychological and pedagogical. The project came to the conclusion that cooperation with the Nordic countries should involve continuous exchange of experiences and knowledge of rare diseases through regular conferences and seminars, increasing co-operation with small separate projects in the field of rare diseases, and joint Nordic training in the field.

Planning is also underway for a national plan for cancer treatment and research with the hope that the process for the rare disease and cancer plans to feed into one another.

Centres of expertise
There are currently no official centres of expertise for rare diseases in Finland. However, the departments for different medical specialities in university hospitals act as reference centres for rare diseases, and certain university hospitals specialise in specific rare operations related to rare diseases, such as congenital heart defects, cleft lip or palate, craniofacial malformations, glaucoma, retinoblastoma and biliary atresia according to the decree of the Ministry of Social Affairs and Health (767/2006) based on a law for specialised medical treatment (1062/1989).

The establishment of centres of expertise and healthcare pathways will be one of the first topics to be dealt with in the elaboration of a national plan for rare diseases, with hope for the first official centres of expertise by 2013. There are already officially designated expert centres, though not for specific diagnoses but for specific treatments (craniofacial surgery, childhood rheumatoid arthritis, hematologic malignancies in children).

The Ministry of Social Affairs and Health carried out a nationwide survey in 2011 to identify unofficial centres of expertise which fueled discussions on the subject at the 2012 meeting of the Orphanet Scientific Advisory Board.

Registries
In general, all the main health care registries are under the National Institute for Health and Welfare, for example Hospital Discharge Registry, Cancer Registry, Malformation Registry and Birth Registry. There is a need for new legislation related to registries; this is in the pipeline in the Ministry of Social Affairs and Health. Finland has not decided how to approach the question of rare disease registries, this will be part of the national plan. The other registries are financed by the government.

There are two legally specified registries concerning rare diseases: the Finnish register of congenital anomalies and the Finnish register of visual impairment. However, there is no designation process for rare disease registries.

The Finnish Haematology Registry and Clinical Biobank was established in 2010 by the Finnish Association of Haematology (FAH). In addition, rare inherited cancers are included in the Cancer Register and rare kidney diseases are included in the Finnish registry for Kidney Diseases. There is a national registry for primary and specialised health care but in this registry rare diseases are difficult to trace due to the problems of ICD10. Finland contributes to European registries including TREAT-NMD and EUROCAT.

Neonatal screening policy
All newborns are screened for hypothyroidism but not for phenyketonuria as it is practically absent in the Finnish population. A pilot scheme for screening additional metabolic diseases including congenital adrenal hyperplasia (CAH), MCAD deficiency, LCHAD deficiency, Glutaricaciduria type 1 (GA1), and phenylketonuria was started in 2007 in the Turku area, concerning around 3000 newborns per year. No decision has at present been made concerning the continuation of the pilot beyond the year 2012 or widening of the pilot to other areas in Finland, though experts representing university hospitals now suggest that screening for newborn congenital metabolic diseases should be widened in 2015, so that the screening practice in Finland would be similar to other western countries. A screening recommendation was handed over to the Ministry of Social Affairs and Health in 2012. In addition to this, hospitals organise screening for phenylketonuria in newborns of non-Finnish origin.

Genetic testing
Genetic testing on the national level is not centrally organised but has developed partly based on needs for certain tests but partly due to local desire to have a molecular laboratory also for training/educational purposes. There are laboratories offering selections of tests (especially the founder mutations of the so called Finnish Disease Heritage) in University Hospitals, the largest in Helsinki. Genetic tests are performed in all five University towns either in the University (Turku) or in the University Hospital or a linked state-owned laboratory company (Helsinki, Kuopio, Oulu, and Tampere).

In addition, there is one major private laboratory offering testing, also to public health care.

There are no national guidelines, but most of predictive testing (including familial cancer) happens in genetic clinics. According to the law on the patient’s status and rights (1992/785) informed consent is always sought for medical tests but it does not have to be written. Clinical geneticists have agreed among themselves that tests for adult-onset diseases or carriership are not performed in minors. Most physicians representing other specialties agree to this principle. Some of the laboratories are accredited, some are still in the process of being accredited but they all belong to larger laboratory units which are, at least partly, accredited.

Tests are part of the hospital fee of which the patients pay a nominal sum. The municipalities then are responsible for paying for the tests: the rest is paid by the municipalities. It is rather rare that the payment would create a problem: usually if the physician in charge of diagnosis/treatment of a patient suggests genetic test(s), they are always paid without any discussion.

Genetic testing abroad creates usually no problems, many even quite common diseases like NF1, Marfan and related disorders etc are regularly bought elsewhere, from Europe or the USA. Then, usually, a laboratory that performs the required test is sought for from Orphanet. Also Finnish laboratories carry out genetic tests for foreign customers, especially in case of the diseases of the so called Finnish Diseases Heritage.

Diagnostic tests are registered as available in Finland for 182 genes and an estimated 230 diseases in the Orphanet database. Other tests are available abroad.

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2 www.thl.fi/tietokantaraportit
3 Information extracted from the Orphanet database (December 2012).
National alliances of patient organisations and patient representation
Representatives of patient associations decided to set up a national alliance at their meeting at the Family Federation Finland, in Helsinki on 6 June 2011. During this meeting it was decided to set up a work group, led by Elina Nykyri, head of the Finnish Turner Association, to prepare a constitutive meeting held on 8 October 2011. A first statutory meeting was held on 21 January 2012. The new alliance, named HARSO, HArvinainen (rare) Sairauksien (diseases) Organisaatio (organisation) welcomes all Finnish patient organisations that represent one or more rare diseases or disabilities. Harso is run by patients themselves. There were 29 organisations out of a total of 51 in Finland present at the launch of the association. The new umbrella group will advocate for the rare disease patients, their families and their organisations in Finland, aiming for the best possible health and social care for the entire rare disease community. One of the main objectives will be to raise awareness of rare diseases and disabilities in order to facilitate diagnosis. Rare diseases and/or disabilities affect the daily lives of approximately 250 000 people in Finland. Harso will provide the rare disease community with strength in numbers for the first time. The organisation unites the rare disease community, creating a common voice and more visibility. So far, some rare disease organisations have chosen to stay outside Harso.

At present, individual patient organisations may be consulted on their opinion about forthcoming legislation, but the bodies to be consulted on such matters have not been defined. The Ministry of Social Affairs and Health has a council of the disabled which represents all groups of the disabled including rare disease patients.

Sources of information on rare diseases and national help lines

**Orphanet activities in Finland**
Since 2004 there is a dedicated Orphanet team in Finland, previously hosted by the Medical Genetics Clinic of Vaestolitto, the Family Federation of Finland, and now hosted by the Norio-centre The team was designated as the Finnish national Orphanet team by the Ministry of Social Affairs and Health in 2010. This team is in charge of collecting data on rare disease related services (specialised clinics, medical laboratories, ongoing research, registries, clinical trials and patient organisations) in their country for entry into the Orphanet database, as well as maintaining the Orphanet Finland national website in Finnish.

Orphanet and Terveysportti have established a collaboration to add links from Terveysportti’s Finnish texts to the relevant Orphanet disease page were added: Orphanet is thus included in Terveysportti’s searches for these 300 “most common rare diseases” and will make Orphanet better known amongst Finnish healthcare professionals. Terveysportti is maintained by Duodecim, the Finnish Medical Society, a scientific society adhered to by almost 90% of Finnish doctors and medical students. The Terveysportti portal is for healthcare professionals and is used nationwide in public health care units, hospitals, private practices and pharmacies as well as the universities’ medical faculties. The service consists of more than 35 databases and helps professionals find day-to-day medical information quickly and reliably from one source.

**Official information centre for rare diseases**
There is no official information centre for rare diseases in Finland other than the services provided by Orphanet, however the Norio-centre serves unofficially this function for health care providers, students, teachers, parents etc.

**Help line**
There is no official help line for rare diseases. The Norio-centre has a nationwide phone and e-mail service for matters concerning rare diseases, which operates on work-days: the Norio-centre serves unofficially this function for health care providers, students, teachers, parents etc.

**Other sources of information**
Established in 1993, the Harvinaiset Network is a network of 18 non-governmental, non-profit organisations funded by RAY, Finland’s Slot Machine Association. The members of the network have signed a partnership agreement and are regarded as equal partners in the network.

The Network provides information on rare diseases and services, raises awareness of the needs of people with rare diseases and organises courses for patients and their families. Harvinaiset also maintains an internet portal with information about rare diseases in Finnish. An updated website was launched in 2010 as

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1 [http://www.orpha.net/national/Fi-FI/index/kotisivu/](http://www.orpha.net/national/Fi-FI/index/kotisivu/)
well as a web based service to help individuals without patient organisations for their disease to find other individuals with the same rare disease. The Harvinaiset network also participates in the maintaining of the Nordic website www.rarelifi.

Most providers of services for rare diseases also have web-based information and phone or web answering services: they provide general information about diseases, contacts for treatment, advocacy, rehabilitation, psychological support and support from patient organisations or peer support groups.

**Good practice guidelines**

Finland has a strong tradition of producing best practice guidelines. They, however, are written based on Cochrane reviews and as this is not possible in case of rare diseases, and such guidelines have not been produced. Health care personnel in Finland, especially medical doctors, have no difficulties in accessing and using guidelines written in English.

Information on 35 monogenic diseases belonging to the Finnish Disease Heritage\(^6\) can be found at a database findis.org. For each disease, the prevalence or incidence and a short description of clinical symptoms are provided, as well as genetic locus and a molecular description for identified mutations. As the character and consequences of all known mutations, Finnish and foreign, are described at the DNA and polypeptide level and disease allele frequencies reported for Finnish mutations, the database can be used as a best practice guideline for molecular diagnostics of these diseases. However, this database does not provide guidelines or information related to treatment or follow up of these diseases.

The database follows the Quality Criteria for Health Related Websites recommended by the European Commission: funding for the database has been provided by the Academy of Finland, Centre of Excellence in Disease Genetics.

**Training and education initiatives**

There are regular “dysmorphology afternoons” twice a year, especially planned to support young doctors in training.

**National rare disease events in 2012**

International Rare Disease Day 2012 was coordinated by The Finnish Network for Rare Diseases, Harvinaiset-verkosto. Together with the patient organisations the Network organised a webinar entitled “Rare Disease Day 2012” at the Finnish Parliament Annex, called the Little Parliament on 29 February 2012. Ms. Paula Risikko, The Minister of Social Affairs and Health as a guest speaker. Dr Päivi Kaukonen, Ministerial Adviser from the Ministry of Social Affairs and Health, presented survey which has been carried out among university and central hospital chief physicians and Harvinaiset Network member organisations of diagnostics, care and rehabilitation with rare disease patients in Finland. Dr. Kaukonen announced the establishment for April 2012 of a steering group for the preparation of the national plan by the Ministry. The seminar was also an opportunity to hear the results of the Harvinaiset survey launched in January 2012 on rare diseases which received around 700 responses.

Swedish Orphan Biovitrum Finland organised the 4th Harvinaiset Sairaudet-päivä (Rare Disease Day) together with stakeholders in Helsinki on 19 October 2012. This day provided a forum for questions concerning research and management for Finnish decision-makers and specialists. The principal goal of the day was to discuss how Finland would become a model country for rare disease research and management and which actions would ensure that patients suffering from rare conditions would be entitled to the same quality of treatment as other patients.

**Hosted rare disease events in 2012**

No specific information reported.

**Research activities and E-Rare partnership**

**National research activities**

There are no specific programmes for rare disease research and projects compete with other topics in the calls of Finnish Academy and various foundations. Fundraising events do not belong to the research funding traditions in Finland, except for the research related to cancer and paediatric diseases.

Research in the field of rare disease has been focused on diseases of so-called Finnish Disease Heritage; nearly 40 rare inherited diseases are over-represented in Finland in comparison to other populations.

Most of the genes associated with these diseases have been mapped and cloned in Finland during the last 20 years. Also rare forms/founder mutations amongst more common ones, like hereditary nonpolyposis colorectal cancer (HNPPC), hereditary connective tissue diseases, and long QT syndrome, have been studied.

Many different bodies fund medical research programmes in Finland. There are no specific programmes for research of rare diseases, which compete with more common diseases for the funds. Part of this funding for research goes towards research on orphan medicinal products. Five universities with medical faculties have programmes of their own, which are partly funded by a special State contribution (EVO). The Finnish Academy and private foundations finance substantially medical research and some rare disease research programmes amongst others.

**Participation in European research projects**

Finland participates, or has participated, in European rare disease research projects including: BNE, CLINIGENE, DEMCHILD, EUGINDAT, EUMITOCOMBAT, EURAPS, EUREGENE, EUROBONET, EUROGENTEST 1 & 2, EUROPEAN LEUKEMIA NET, GRIP, GEN2PHEN, LYMPHANGIOMATOGENIC, NEUROPRION, PEROXISOMES, PROTHETS, INTERPREGEN, INTREALL, PULMOTENSION, TREAT-NMD and RD PLATFORM.

**E-Rare**

Finland is not currently a partner of the E-Rare consortium.

**IRDiRC**

Finnish funding agencies are not yet committed members of the IRDiRC.

**Orphan medicinal products**

**Orphan medicinal product committee**

No specific information reported.

**Orphan medicinal product incentives**

The Finnish Medicines Agency (Fimea, which before the 1st November 2009 was known as the National Agency for Medicines Lääkelaitos) gives free administrative and scientific advice to bodies developing orphan medicinal products. Furthermore, the special status of orphan medicinal products has been taken into account in inspection and authorisation procedures. Fimea also maintains a registry of clinical trials.

The evaluation criteria are the same for all medicinal products; no exceptions for orphan medicinal products are stated in the Health Insurance Act. However, the health economic evaluation is not always required from the marketing authorisation holder of orphan medicinal product if justified by the applicant.

**Orphan medicinal product market availability situation**

Of the orphan medicinal products with EU market authorisation, 51 are available on the market currently in Finland in at least one form, if not in all forms. The Fimea lists the following orphan medicinal products as available on the market in Finland: Adcetris, Aldurazyme, Arzerra, Atriance, Busilvex, Cystadane, Diacomit, Elaprase, Esbriet, Evoltra, E unpaid, Fabrazyme, Firazyr, Firdapse, Glion, Increlex, Inovelon, Jakavi, Laxite, Lysofren, Mepact, Mozobil, Myozyme, Naglazyme, Nexavar, Nplate, Orfadin, Peeda, Prialt, Revatio, Revlimid, Revolve, Savene, Signon, Soliris, Somaver, Spycel, Tasigna, Tepadin, Thalidomide Celgene, Tobi Podhaler, Torisel, Tracleer, Ventavis, Vidaza, Volibris, Votubia, Vpriv, Xagrid, Yondelis, Zavesca.

**Orphan medicinal product pricing policy**

According to the 2005 Inventory of Community and Member States’ incentive measures to aid the research, marketing, development and availability of orphan medicinal products in the section concerning Finland, “A reasonable wholesale price refers to the maximum price at which the product may be sold to pharmacies and hospitals. The holder of marketing authorisation must be able to justify the reasonableness of the proposed wholesale price for a medicinal product that is to serve as a basis for the reimbursement payments. The application must include a detailed, comprehensive assessment of the cost of the drug therapy and the benefits expected to be gained thereby. Moreover, the application must include an evaluation of the product in relation

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1 This section was written with information from the Inventory of Community and Member States’ incentive measures to aid the research, marketing, development and availability of orphan medicinal products. Revision 2005 (pp11-12)
2 http://www.fimea.fi/medicines/fimeaweb
3 Inventory of Community and Member States’ incentive measures to aid the research, marketing, development and availability of orphan medicinal products. Revision 2005 (p12)
to alternative drug treatments and other therapies. The application must also include the validity period of the pharmaceutical patent or a supplementary protection certificate, an estimate of the sales volume and number of users of the product over the next three years as well as the approved price and ground for reimbursement of the product in other EEA countries.

“Applications concerning medicinal products containing a new active substance must contain a health economic evaluation. When considering the reasonableness of the proposed wholesale price, the Pharmaceuticals Pricing Board takes into account the cost of the drug therapy and the benefits to be gained from its use as regards both the patient and the overall health care and social costs. The Pricing Board will also consider the cost of the treatment alternatives, the prices of comparable medicinal products and the price of the medicine in question in other EEA countries. Manufacturing, research and development costs are also taken into consideration when making a decision on application, if they are considered relevant by the applicant, as are the funds allocated for reimbursement payments.”

**Orphan medicinal product reimbursement policy**

All medicines with a wholesale price approved by the Pharmaceuticals Pricing Board are automatically entitled to reimbursement under the basic refund category. The basic reimbursement is currently 35% of the purchasing price. In certain diseases or conditions, lower (65%) or higher (100%) special reimbursement is available.

In October 2010 Harvinaiset, the Finnish Network for Rare Diseases, sent a letter to the Ministry of Social Affairs and Health concerning the reimbursement of orphan medicinal products in Finland in order to expose the need for an improved approach to the issue especially for Fabry disease, Myasthenia gravis and Long QT syndrome. The Ministry wrote back to assure that the pharmaceutical policy foreseen for 2020 would deal with many of the concerns raised by the network including the development of medications towards more specific treatments (including orphan medicinal products), price regulations, updating the list of diseases for which reimbursement is provided through the Government Regulation in place, uniform payments and payment caps for social and health care.

**Other initiatives to improve access to orphan medicinal products**

No specific information reported.

**Other therapies for rare diseases**

No specific information reported.

**Orphan devices**

No specific information reported.

**Specialised social services**

Respite care services are available and local authorities are responsible for their provision, but some are equally provided by private institutions: patients and families often have to provide co-payment. Therapeutic recreational programmes are available under different forms and patients have to partially pay for these services though some funding can be provided by RAY. Services for transport, modifications for housing arrangements, day-care, interpreter (sign language etc), personal assistants etc are available for those with handicaps by local authorities, provided for by the law 380/1987 in 1987 (updated 1267/2008 and 981/2008).

Patients with a rare disease, as well as all others with a severe disability, were given new possibilities 2009. The legislation for personal assistance was updated as of 1 September 2009. This update follows the principles of Independent Living Movement. Personal assistance for persons with a severe functional disability is free of charge. Besides the support in the daily living, work and education this now also includes assistance with participation in recreational activities, social activities and education. The service is financed by the municipalities.

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10 [http://harvinaiset.fi/ajankohtaista/STM-n-vastaus-kannanottoon-laakkeiden-korvauksista&rurl=translate.google.fr&twu=1&usg=ALkJrhD90epLjIJoahxcmkK5V47DHOfIr_Av](http://harvinaiset.fi/ajankohtaista/STM-n-vastaus-kannanottoon-laakkeiden-korvauksista&rurl=translate.google.fr&twu=1&usg=ALkJrhD90epLjIJoahxcmkK5V47DHOfIr_Av)
DEVELOPMENT OF RARE DISEASE ACTIVITIES IN 2012 IN FINLAND

National plan/strategy for rare diseases and related actions
A meeting is planned in March 2013 to advance with the elaboration of the nationa plan for rare diseases. The aim is to have the plan ready during 2013, the designation of expert centres will be a much longer process to follow.

Planning is also underway for a national plan for cancer treatment and research with the hope that the process for the rare disease and cancer plans to feed into one another.

Centres of expertise
The establishment of centres of expertise and healthcare pathways will be one of the first topics to be dealt with in the elaboration of a national plan for rare diseases, with hope for the first official centres of expertise by 2013.

The Ministry of Social Affairs and Health carried out a nationwide survey in 2011 to identify unofficial centres of expertise which fueled discussions on the subject at the 2012 meeting of the Orphanet Scientific Advisory Board.

Neonatal screening policy
A pilot scheme for screening additional metabolic diseases including congenital adrenal hyperplasia (CAH), MCAD deficiency, LCHAD deficiency, Glutaric aciduria type 1 (GA1), and phenylketonuria was started in 2007 in the Turku area, concerning around 3000 newborns per year. No decision has at present been made concerning the continuation of the pilot beyond the year 2012 or widening of the pilot to other areas in Finland, though experts representing university hospitals now suggest that screening for newborn congenital metabolic diseases should be widened in 2015, so that the screening practice in Finland would be similar to other western countries. A screening recommendation was handed over to the Ministry of Social Affairs and Health in 2012.

National alliances of patient organisations and patient representation
Representatives of patient associations decided to set up a national alliance at their meeting at the Family Federation Finland, in Helsinki on 6 June 2011. During this meeting it was decided to set up a work group, led by Elina Nykyri, head of the Finnish Turner Association, to prepare a constitutive meeting held on 8 October 2011. A first statutory meeting was held on 21 January 2012. The new alliance, named HARSO, HArvinainen (rare) Sairauksien (diseases) Organisaatio (organisation) welcomes all Finnish patient organisations that represent one or more rare diseases or disabilities. Harso is run by patients themselves. There were 29 organisations out of a total of 51 in Finland present at the launch of the association. The new umbrella group will advocate for the rare disease patients, their families and their organisations in Finland, aiming for the best possible health and social care for the entire rare disease community. One of the main objectives will be to raise awareness of rare diseases and disabilities in order to facilitate diagnosis. Rare diseases and/or disabilities affect the daily lives of approximately 250,000 people in Finland. Harso will provide the rare disease community with strength in numbers for the first time. The organisation unites the rare disease community, creating a common voice and more visibility. So far, some rare disease organisations have chosen to stay outside Harso.

Sources of information on rare diseases and national help lines
Help line
The Norio-centre has a nationwide phone and e-mail service for matters concerning rare diseases, which operates on work-days: the Norio-centre receives part of its funding from Finland’s Slot Machine Association (RAY). The main purpose of RAY is to raise funds through gaming operations to promote Finnish health and welfare.

National rare disease events in 2012
International Rare Disease Day 2012 was coordinated by The Finnish Network for Rare Diseases, Harvinaiset-verkosto. Together with the patient organisations the Network organised a webinar entitled “Rare Disease Day 2012” at the Finnish Parliament Annex, called the Little Parliament on 29 February 2012. Ms. Paula Risikko, The Minister of Social Affairs and Health as a guest speaker. Dr Pälvi Kaukonen, Ministerial Adviser from the...
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Swedish Orphan Biovitrum Finland organised the 4th Harvinaiset Sairaudet–päivä (Rare Disease Day) together with stakeholders in Helsinki on 19 October 2012. This day provided a forum for questions concerning research and management for Finnish decision-makers and specialists. The principal goal of the day was to discuss how Finland would become a model country for rare disease research and management and which actions would ensure that patients suffering from rare conditions would be entitled to the same quality of treatment as other patients.
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- Finnish Disease Database
- Finnish Medicines Agency
  http://www.fimea.fi/medicines/fimeaweb
- Rarelink Finland
  www.rarelink.fi
- Orphanet Finland national website
  http://www.orpha.net/national/FI-FI/index/kotisivu/

The contributors and validators of the report have contributed information which is accurate to the best of their knowledge. However, readers should take note that the contents of this report are illustrative and not exhaustive.

All websites and documents were last accessed in May 2013.