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European Union Committee of Experts on Rare Diseases

# 2012 REPORT ON THE STATE OF THE ART OF RARE DISEASE ACTIVITIES IN EUROPE OF THE EUROPEAN UNION COMMITTEE OF EXPERTS ON RARE DISEASES

# STATE OF THE ART OF RARE DISEASE ACTIVITIES IN FINLAND

**This work was financed by the EUCERD Joint Action: Working for Rare Diseases N° 2011 22 01**

EUCERD European Union Committee of Experts on Rare Diseases
2011-22-01
EUCERD Joint Action

This document has been produced by the Scientific Secretariat of the European Union Committee of Experts on Rare Diseases (EUCERD, formerly the European Commission's Rare Diseases Task Force) through the EUCERD Joint Action: Working for Rare Diseases (N° 2011 22 01, Coordinator: Kate Bushby, University of Newcastle, United Kingdom), within the European Union's Second Programme of Community Action in the Field of Health.

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

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**To quote this document:**
Aymé S., Rodwell C., eds., *"2012 Report on the State of the Art of Rare Disease Activities in Europe of the European Union Committee of Experts on Rare Diseases"*, July 2012.

ISBN : 978-92-79-25355-3
DOI : 10.2772/50554

http://www.eucerd.eu/wp-content/uploads/2012/09/2012ReportStateofArtRDActivitiesFI.pdf

# ACRONYMS

**General**

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 (Iceland, Switzerland, Norway)
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

**Pilot European Reference Networks**

Dyscerne - European network of centres of expertise for dysmorphology
ECORN-CF - European centres of reference network for cystic fibrosis
Paediatric Hodgkin Lymphoma Network - Europe-wide organisation of quality controlled treatment
NEUROPED - European network of reference for rare paediatric neurological diseases
EUROHISTIONET - A reference network for Langerhans cell histiocytosis and associated syndrome in EU)
TAG - Together Against Genodermatoses – improving healthcare and social support for patients and families affected by severe genodermatoses
PAAIR - Patients' Association and Alpha-1 International Registry Network
EPNET - European Porphyria Network - providing better healthcare for patients and their families
EN-RBD -European Network of Rare Bleeding Disorders
CARE-NMD -Dissemination and Implementation of the Standards of Care for Duchenne Muscular Dystrophy in Europe project
ENERCA - European network for rare and congenital anaemia – Stage 3

# GENERAL INTRODUCTION TO THE REPORT ON THE STATE OF THE ART OF RARE DISEASE ACTIVITIES IN EUROPE OF THE EUROPEAN UNION COMMITTEE OF EXPERTS ON RARE DISEASES

The 2012 Report on the State of the Art of Rare Disease Activities in Europe 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 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 2011. 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 either the European Commission, its Agencies or national health authorities.

The report is split into five parts:

Part I: Overview of rare disease activities in Europe
Part II: Key developments in the field of rare diseases in 2011
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

Each part contains a 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[1].

[1] http://www.eucerd.eu/upload/file/Reports/2012ReportStateofArtRDActivities.pdf

# 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.

**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). 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, including hospital districts, governmental institutes like the National Institute for Health and Welfare, the Orphanet National Advisory Board, the umbrella organisation for rare diseases HARSO 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).

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 wassupported 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[2] that co-operation 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.

**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.

**Pilot European Reference Networks**

Finnish teams participate, or have participated, in the following European Reference Networks for rare diseases: Dyscerne, EPNET and EPI.

**Registries**

In general, all the main health care registries are under the National Institute for Health and Welfare.

There are two legally specified registries concerning rare diseases: the Finnish register of congenital anomalies and the Finnish register of visual impairment.

The Finnish Heamatology 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

[2] http://www.nordicwelfare.org/filearchive/1/119089/Rapport%20med%20bilagor%20final.pdf

primary and specialised health care[3]) 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. 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 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. 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, a private laboratory Medix Laboratories Ltd offers a selection of genetic tests.

There are no national guidelines for performing genetic testing. 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.

Genetic tests are, as a rule, performed in the framework of public health care. The municipalities then are responsible for paying for the tests. 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 tests are often purchased from abroad. 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 158 genes and an estimated 195 diseases in the Orphanet database[4]. Other tests are available abroad.

**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 planned for 21 January 2012.

The role of the new alliance is yet to be seen. 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, currently hosted by the Medical Genetics Clinic of Vaestoliitto, the Family Federation of Finland. 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[5] in Finnish.

[3] www.thl.fi/tietokantaraportit

[4] Information extracted from the Orphanet database (September 2011).

[5] http://www.orpha.net/national/FI-FI/index/kotisivu/

In 2010 Orphanet and Terveysportti[6] established a collaboration. 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. During 2010 links from Terveysportti's Finnish texts to the Orphanet disease 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.

***Official information centre for rare diseases***

There is no official information centre for rare diseases in Finland other than the services provided by Orphanet.

***Help line***

There is no official help line for rare diseases. The Medical Genetics Clinic of Vaestoliitto, the Family Federation of Finland has a nationwide phone and e-mail service for matters concerning rare diseases, which operates on work-days.

***Other sources of information***

Established in 1993, the Harvinaiset Network is a network of 17 non-governmental, non-profit organisations funded by RAY, Finland's Slot Machine Association. 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 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.rarelink.fi.

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**

Information on 35 monogenic diseases belonging to the Finnish Disease Heritage[7] 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**

No specific information reported.

**National rare disease events in 2011**

Rare Disease Day 2011 was coordinated by the Finnish Network for Rare Diseases, Harvinaiset. Awareness raising campaigns were launched on the website harvinaiset.fi and in social media between 25 and 28 February 2011 in cooperation with local associations and groups. Stories, photos, videos and messages around the theme "rare but equal" were shared. The members of the Finnish Network for Rare Diseases visited schools, institutions and other focus groups to inform about the RDD and issues concerning rare diseases. Members of the Network also launched their own press releases concerning the day.

On 21 October 2011, a rare disease event was organised in Helsinki by medical specialist societies and Swedish Orphan Biovitrum. 150 health professionals, researchers and policy makers attended. The main theme

[6] http://www.terveysportti.fi/

[7] http://www.findis.org/main.php?action=disease

of the meeting was rare disease research and treatment, the current situation and future possibilities. Special national funding into research was requested because funding possibilities have diminished in recent years. Much of the general discussion focused on the assessment of marketed drugs for reimbursement but also on the need for centres of expertise, and involvement and empowerment of patients and their families to the RD field. A 700 name petition was handed over to the minister of Social Affairs and Health. The petition drew attention to the unequal situation of rare disease patients compared to common disease patients.

On 20 October 2011 the Orphanet Finland team organised a one-day meeting for the Orphanet National Advisory Board members which focused on the national plan for rare diseases and allowed members to express their expectations for the plan. Discussion also focused on how to structure centres of expertise in Finland, including their criteria, funding, role of the university hospitals and importance of the research in the context of centres etc. Importance of networks and cooperation between specialists in various fields were highlighted in many speeches.

**Hosted rare disease events in 2011**
No specific information reported.

**Research activities and E-Rare partnership**
***National research activities***
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 (HNPCC), 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, EUGINDAT, EUMITOCOMBAT, EURAPS, EUREGENE, EUROBONET, EUROGENTEST, EUROPEAN LEUKEMIA NET, GEN2PHEN, LYMPHANGIOGENOMICS, NEUROPRION, PEROXISOMES, PROTHETS, 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[8]***
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.

[8] 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)

***Orphan medicinal product market availability situation***

Of the orphan medicinal products with EU market authorisation, 48 are available on the market currently in Finland in at least one form, if not in all forms. The Fimea[9] lists the following orphan medicinal products as available on the market in Finland: Aldurazyme, Arzerra, Atriance, Busilvex, Cystadane, Diacomit, Elaprase, Evoltra, Exjade, Fabrazyme, Firazyr, Firdapse, Gliolan, Glivec, Increlex, Inovelon, Litak, Lysodren, Mepact, Mozobil, Myozyme, Naglazyme, Nexavar, Nplate, Orfadin, Pedea, Prialt, Replagal, Revatio, Revlimid, Revolade, Savene, Soliris, Somavert, Sprycel, Tasigna, Tepadina, Thalidomide Celgene, Tobi Podhaler, Torisel, Tracleer, Ventavis, Vidaza, Volibris, 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*[10] 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 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***

According to the 2005 *Inventory of Community and Member States' incentive measures to aid the research, marketing, development and availability of orphan medicinal products*[11] in the section concerning Finland, *"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 42% of the purchasing price. In certain diseases or conditions, lower (72%) or higher (100%) special reimbursement is available.*

In October 2010 Harvinaiset, the Finnish Network for Rare Diseases, sent a letter[12] 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.

**Orphan devices**

No specific information reported.

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[9] http://www.fimea.fi/medicines/fimeaweb

[10] *Inventory of Community and Member States' incentive measures to aid the research, marketing, development and availability of orphan medicinal products. Revision 2005* (p12)

[11] *Inventory of Community and Member States' incentive measures to aid the research, marketing, development and availability of orphan medicinal products. Revision 2005* (p12)

[12] http://harvinaiset.fi/ajankohtaista/stm-n-vastaus-kannanottoon-laeaekehoidon-korvauksista&rurl=translate.google.fr&twu=1&usg=ALkJrhh-90epJLiXoahxcmk5V47DHOlr_Av

**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.

# DEVELOPMENT OF RARE DISEASE ACTIVITIES IN 2011 IN FINLAND

**National plan/strategy for rare diseases and related actions**

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). 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, including hospital districts, governmental institutes like the National Institute for Health and Welfare, the Orphanet National Advisory Board, the umbrella organisation for rare diseases HARSO 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).

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[13] http://www.findis.org/main.php?action=disease

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**Research activities and E-Rare partnership**

***IRDiRC***

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

# LIST OF CONTRIBUTIONS[14]

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  http://www.orpha.net/national/FI-FI/index/kotisivu/

[14] 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.
[15] All websites and documents were last accessed in May 2012. A more detailed list of sources is available in the full report: http://www.eucerd.eu/upload/file/Reports/2012ReportStateofArtRDActivities.pdf