State of the Art for Rare Diseases – Activities in EU Member States and Other European Countries

Finland Report

Definition of a Rare Disease

Finland has adopted the European Commission definition of a rare disease (i.e. a prevalence of no more than 5 per 10,000 people, as per *Regulation (*EC) No 141/*2000)* and the National Plan espouses this definition.

Status Quo of any National Plan or Strategy for Rare Diseases

Finland adopted a National Plan for Rare Diseases. The plan covers the years 2014-2017. The Ministry of Social Affairs and Health had appointed a multidisciplinary Steering Group to prepare a proposal for a National Plan. Once the proposal was completed it was assessed and approved by the Ministry. Unfortunately, no laws or decrees relating to the National Plan were approved and a specific budget was not assigned.

The Ministry, however, did provide the minimal funding required to form and run an Expert Group responsible for the realisation of the National Plan. The Ministry also provided a doctor who, on a part time basis, was tasked with working towards the goals of the National Plan.
All five of the university hospitals in Finland realised that something needed to be done to streamline the diagnostics, treatment and research relating to rare diseases: this required funds and the university hospitals dedicated a budget to perusing this goal. Helsinki University Hospital, the biggest in Finland, dedicated budget to pay for the salary of a specialist MD, one nurse and a part time senior consultant and other personnel.

The national plan addresses the issue of rare disease coding, stating that Finland will wait for the implementation of ICD11. (http://www.who.int/classifications/icd/revision/icd11faq/en/). At present Finland has, by use of ICD10, more RD codes than most other countries. However, some of the university hospitals are in the process of implementing OrphaCodes to their current coding systems.

In Finland, a dedicated advisory body existed to oversee the drafting (and to some extent the implementation) of the National Plan – this body is multistakeholder (including patients) and meets regularly. Its main purpose, post adoption of the Plan, is to promote collaboration between the rare disease units within the University hospitals and to organise the National rare disease forum. It has become the role of the University hospitals to start working towards the implementation of the National Plan rather than the advisory body.

A major success of the National Plan is that every university hospital now has a rare disease unit. Much time and energy has been spent in trying to identify all rare disease experts within each hospital so that patients may find these individuals more easily. It is hoped that this will enable hospitals to facilitate care pathways for suspected rare disease patients and enable them to find experts more quickly. There is also a plan and funding in place to create shared websites, for each university hospital. Collaboration with patient organisations has also been prominent but now it is considered an integral part of the work of the rare disease units. Another notable success has been the Academy of Finland's membership of the IRDiRC.

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Organisation of Rare Disease Health and Social Care (Centres of Expertise; ERNs; Integrated Care and Social Support

Centres of Expertise

Finland has an official policy for the designation of Centres of Expertise for rare disease and has adopted both a national and regional designation process. The formal policy is fully implemented: the healthcare provider (University hospital) has to assess and elect a Centre of Expertise and examine its work plan, quality etc. The Ministry of Social Affairs and Health makes the final designation check by ensuring the centre and the process of designation have conformed to Finnish legislation.

The national criteria used to designate a Centre of Expertise for rare disease is in accordance with the EUCERD criteria. One element of the criteria is that Centres ensure a holistic approach to care and liaise with social services.

- The number of Centres of Expertise complying with the National Policy: 16
- The number of Centres of Expertise within Finland divided by million inhabitants: 3
- The number of Centres of Expertise within Finland which fulfil the EUCERD criteria: 16

European Reference Networks (ERNs)

Finland has a formal process in place for the endorsement of Health care providers to participate as members or a coordinator of an ERN. Currently there are 4 HCPs participating as full members of 11 ERNs.

Members	European Reference Networks (ERNs)
Helsinki University Hospital	ERKNet ERN CRANIO ERN GUARD-HEART ERN ITHACA ERN Skin ERNICA VASCREN
Kuopio University Hospital	ERN EpiCARE ERN PaedCan
Tampere University Hospital	ERN EURO-NMD ERN PaedCan
Turku University Hospital	ERN EURACAN ERN GENTURIS ERN PaedCan

See further https://ec.europa.eu/health/ern_en

Integration of Rare Diseases into Social Policies and Services

In Finland, specific programmes and facilities exist to support patients with rare diseases. Finland has a tradition of organising education courses for patients and families with rare or chronic diseases. These courses are free of charge and often last between several days and a week. Otherwise, those who have a rare disease are directed to support through normal social care. Individual care plans are provided which include access to social and support services however they also exist for patients with common diseases, if required, meaning the services provided for both common and rare disease patients are similar.

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There are no specific measures in place to support the integration of rare diseases' specificities into the national system responsible for assessing a person's level of functioning: the rarity of the condition is not a criterion but rather the scope of the symptoms, degree of disability etc.

Future plans to support social integration are outlined within the Finnish national plan, thus possible improvements are being discussed.

Rare Disease Registration

There is no national registry explicitly for all rare diseases in Finland; however, national healthcare registries do exist, which cover certain rare diseases. These include registries for Cancer, Malformations, and Hospital Discharge. These registries only include rare disease patients if the diagnosis appears in ICD10.

There are also certain disease-specific registries, funded through government institutes, but these exist purely for research purposes and they tend to become obsolete once the research project is complete. Once University hospitals begin to use the OrphaCode, rare disease will automatically become part of the Hospital Discharge registry.

Registries use the ICD10 coding system which allows healthcare professionals to locate certain rare disease patients within registries. The legalisation relating to Healthcare Registries is currently in the process of being amended.

Genetic Testing and Neonatal Screening

Nearly 100% of **genetic testing** is performed within public healthcare services in Finland. Testing is performed within Finnish laboratories or abroad, depending on where the test is available, which incurs no cost to the patient. Very few genetic tests are performed within private clinics and paid for by the patient. Genetic testing is mentioned within legislation which prohibits its use for employment purposes, i.e. to ensure no candidate is excluded. There is no list of reference laboratories, but many of the national laboratories provide details of the services they offer, via the Orphanet database.

Cross border genetic testing is used widely in both directions. There are no restrictions and clinicians may purchase tests from anywhere if they are required. Initially the hospital pays for the testing; however, in the end, it is the community which pays the bill.

The **Neonatal screening** programme in Finland initially screened only for hypothyroidism; however, in recent years the programme has been extended to include a wider set of diseases based on recommendations from the Ministry of Social Affairs and Health 2014. Helsinki University Hospital now offers screening for 20 rare diseases.

- Phenylketonuria (PKU)
- Tyrosinemia type 1
- Maple syrup urine disease (MSUD)
- Congenital adrenal hyperplasia (CAH)
- Congenital B12-vitamine deficiency
- Glutaric acid uria type I (GA I)
- Isovalericaciduria
- Metylmalonicaciduria
- Propionicaciduria

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- CACT (carnitine-asylcarnitinetranslocase deficiency)
- CPT (Carnitinepalmitystranpherase deficiency) type I
- CPT type II CUD (carnitine uptake defect)
- Glutaricaciduria type II (GA II)
- MCAD
- LCHAD
- TFP (Trifunctional Protein Deficiency)
- VLCAD
- Citrullinemia
- ASA-uria
- Argininemia

Clinical Practice Guidelines and Training Activities

Clinical Practice Guidelines (CPGs)

In Finland, no CPGs for rare diseases have been produced at the national level. Ther is no policy for developing or adopting CPGs. There have been unofficial discussions and different centres tend to have the same types of treatment and follow-up with patients but there are no official guidelines. There *is* a policy for implementing CPGs: there is a rich collection of official guidelines for common diseases, but this collection, produced by Duodecim, is based on Cochrane and similar large studies which cannot be applied to rare diseases.

Training and Education

There *are* initiatives within Finland related to rare disease training and education; however these activities, which include training courses or problem case meetings, are not held on a regular basis. The Medical Specialist Societies will usually include 'rare diseases' as a topic within their annual training courses.

Information Resources for Rare Diseases

Orphanet Activities

Finland has an operational national Orphanet team which is housed within the Norio Centre in Helsinki. There is no dedicated funding for this team from within the National Plan. The team is able to produce information in the Finnish national language and is responsible for updating the Orphanet database, creating Finnish summaries of disease information and presenting annual updates at the Finnish Medical Convention on Orphanet's services.

National Helplines

There is a helpline in place in Finland which is publicly funded and available to both professionals and patients. The helpline is part of the Norio-Centre which is funded by Finland's Slot Machine Association; however, the decisions on how to use the funds are (partially) made by the Ministry of Social Affairs and Health, thus the funding is classed as 'public'.

Official Information Centres

There are both official and unofficial information centres for rare diseases within Finland. Helsinki University hospital has an official website for rare diseases which is funded through the hospitals budget.

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The Norio Centre in Helsinki is also considered an information centre for rare diseases. http://www.norio-keskus.fi/fi/briefly-english

Harvinaiset (rare) Network collects information and links can be found on their website. They also collaborate with RareLink, which is a Nordic knowledge centre.

Rare Disease Research Activities

Unfortunately Finland does not have specific programmes or projects to fund or facilitate rare disease research. Traditionally Finland has invested significant effort and funding into the Clinical-Genetic study of so called "Finnish Disease Heritage". This term refers to 35 rare diseases which seem to be more prevalent in Finland than in other countries. There once existed special funding for this area of research, but this is no longer the case. This has meant that rare disease researchers are now competing for the same funding as researchers of more common diseases.

Finland participates in IRDiRC.

National Alliance of Patient Organisations and Patient Representation

There is a national alliance of rare disease patient organisations within Finland. The Umbrella Organisation of rare disease Harso RY (https://www.harso.fi/), also known as the Finnish Rare Diseases and Disabilities Organisation, houses 25 rare disease patient organisations and has a representative in the Rare Disease Expert Group within the Ministry of Social Affairs and Health. It is also an increasingly collaborative organisation, working with University hospitals and their individual rare disease units.

Harso also:

- Collaborates and works with third sector rare disease players, for example clinics, data centres, and other patient organisations.
- Organises seminars and demonstrations on annually on rare disease day

Rare disease patient organisations as well as Harvinaiset (http://www.harvinaiset.fi/english) have collaborated with similar organisations in Nordic countries, including the Nordic Rare Diseases Network SBONN (http://www.rare-diseases.eu/wp-content/uploads/2013/09/181.-SBONN-%E2%80%93-Rare-Diseases-Nordic-Network-of-Patient-Organisations.pdf)

The National alliance as well as other patient organisations and third sector rare disease services are represented in the National Expert Group for rare disease.

Orphan Medicinal Products (OMPs)

In Finland an official website provided by the Finnish Medicines Agency (http://www.fimea.fi/web/en) states that the number of OMPs with European Union marketing authorisation available in Finland is more than 100. Measures such as compassionate use programmes, named patient supply, possibility of accelerated review, etc., are not currently in use.

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Rare Disease Events

Since 2012 there have been array of national and local events taking place to celebrate Rare Disease day in Finland. These include:

- Two National rare disease events, both in January 2014 and 2016, organised with the support of Swedish Orphan Biovitrum (http://www.sobi.com/)
- In September 2014 the 3rd Nordic Conference of Rare Diseases took place in Helsinki.

Main achievements since 2014:

Every University hospital, there are a total of 5 in Finland, has established specific units for coordinating the treatment of rare disease. This is especially important if patients require many specialised medical treatments to tend to their disease. The 5 units collaborate well but so far there has not been any division of tasks/diseases between them.

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