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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 ESTONIA

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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

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 Estonia. 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 ESTONIA

Definition of a rare disease
Stakeholders in Estonia accept the definition of the European Regulation on Orphan Medicinal Products of a prevalence of no more than 5 in 10'000 individuals.

National plan/strategy for rare diseases and related actions
There is currently no plan for rare diseases in Estonia. In 2008, Estonian Government adopted Eesti Rahvastiku Arengukava 2009-2020 (*Estonian National Health Plan 2009-2020,* hereafter referred to its Estonian acronym, ERTA). ERTA 2009-2020 provides recommendations and indicates the directions to be taken to improve healthcare and brings together the tasks necessary to achieve this. The plan also assembles a large number of strategic documents which have already been implemented or which are soon to be implemented in other fields that have a role to play in achieving ERTA's objective. In 2012 a working group will be set up to discuss the activities on the field of rare diseases which will be added to the implementation plan of ERTA.

Centres of expertise
Up to Summer 2011 there were two clinical genetics centres specialising in the diagnosis and treatment of rare diseases in Estonia, Tartu University Hospital and Tallinn Children's Hospital. In summer 2011 Tallinn Children's Hospital genetics services joined with Tartu University Hospital, the United Laboratory and the Department of Genetics and thus one common Department of Genetics was formally established at Tartu University Hospital. Tallinn Children's Hospital genetics service became the branch office of Department of Genetics, Tartu University Hospital. All of the genetic specialty services are represented with the diagnosis and treatment of rare diseases provided all over Estonia.

Pilot European Reference Networks
Estonian teams participated, in the following European Reference Network for rare diseases: PAAIR.

Registries
Estonia does not have national registers for groups of diagnoses separately. In Estonia all health-related information is collected to the Tervise Infosüsteem (Health Information System). In case of need there is possibility to extract the necessary information. Estonia participates in the EUROCARE CF and EURO-WABB European registries.

Neonatal screening policy
For early detection of all developmental disorders, including rare disorders, there is a consensus agreement that all infants and children with any developmental disorders should be referred to one of tertiary children's hospitals in Estonia: Children's Clinic of Tartu University Hospital or Tallinn Children's Hospital. Both hospitals have quick access to medical genetics services for early detection and prevention[2]. Early detection or treatment of rare diseases is provided using metabolic testing, chromosomal analyses, DNA diagnostics and neonatal screening programmes are in place for phenylketonuria and congenital hypothyroidism since 1993.

Genetic testing
Genetic testing for 46 different diseases is currently available at the Department of Genetics at Tartu University Hospital[3]. Diagnostic tests are registered as available in Estonia for 160 genes and an estimated 78 diseases in the Orphanet database[4].

National alliances of patient organisations and patient representation
There is currently no national alliance for rare disease patient organisations in Estonia. There are only a few non-profit patients associations in the field of rare diseases (Estonian Spinal Hernia and Hydrocephalus Association, Estonian Cystic Fibrosis Society, Estonian Phenylketonuria Society, Estonian Haemophilia Society, Estonian Prader-Willi syndrome Society), who are also members of *Eesti Puuetega Inimeste Koda* (The Estonian

[2] http://www.kliinikum.ee/geneetikakeskus/
[3] See the regularly updated list in Estonian for further information: http://www.kliinikum.ee/geneetikakeskus/
[4] Information extracted from the Orphanet database (September 2011).

Chamber of Disabled People). Support for patient organisations is provided by The Estonian Chamber of Disabled People and *Eesti Patsientide Esindusühing* (The Estonian Patient Advocacy Association (EPAA)). EPAA is a non-profit NGO established in 1994, with the primary aim of advocating for human and civil rights of health and social care service users. The activity of EPAA is financed as purpose-oriented grants from a state budget. In addition there are possibilities to use funds from the gambling tax for project-based financing for patients' organisations.

Patient organisations are represented on the council of the Estonian Health Insurance Fund and grants are available for patient organisations to attend these meetings.

Sources of information on rare diseases and national help lines

Orphanet activities in Estonia

Since 2004, here is a dedicated Orphanet team in Estonia, currently hosted by the Estonian Biocentre. The team was designated at the Orphanet team for Estonia by the Ministry of Social Affairs 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. In 2011 the Orphanet Estonia national website[5], in Estonian, was launched by the Orphanet Estonia team.

Official information centre for rare diseases

There is no official information centre for rare diseases in Estonia other than Orphanet.

Help line

There is currently no help line for rare diseases.

Other sources of information

No specific information reported.

Good practice guidelines

No specific information reported.

Training and education initiatives

There are special advanced courses for physicians (2-3 courses per year) on rare disorders, aimed at improving the early detection and diagnosis of certain rare diseases (Prader-Willi syndrome, Angelmann syndrome, SMA, Dravet Syndrome, etc). In 2009 -2011 the number of number of advanced courses on rare disorders organised by the Department of Continuing Education at the Tartu Medical University increased, due to rising interest in the subject. This activity is also planned for the future (i.e. a new course is planned for 2012).

National rare disease events in 2011

Due to Estonia's small size, there are no special annual rare disease events, nonetheless rare diseases are given a spotlight during the annual meetings of the Estonian Society of Human Genetics and Estonian Society of Laboratory Medicine.

Hosted rare disease events in 2011

The 11th annual International GeneForum[6] was held on 10-11 June 2011, in Tartu, Estonia. This conference on genetics and genomics brought together experts from Europe and the United States of America for discussion on the progress in the field of genetics and in particular, on interdisciplinary areas within human genetics, epigenetics, modern population genetics, biomedical informatics and personal medicine. The conference also incorporates an exhibition aimed at biotechnology-related companies.

The second OpenGENE Young Investigator Workshop in the Baltic Region was held in Tartu, Estonia, on 23-26 August 2011 and focused on re-sequencing, genotyping and statistical analysis of common complex diseases.

[5] http://www.orpha.net/national/EE-ET/index/avaleht/
[6] http://www.geneforum.ee/GF2011/index.php

Research activities and E-Rare partnership

National research activities

According to the *Inventory of Community and Member States' incentive measures to aid the research, marketing, development and availability of orphan medicinal products*, *Eesti Teadusfond* (Estonian Science Foundation) supports research on rare diseases at national level on the basis of appropriate applications, but there is no distinction from other projects not related to rare diseases (approximately 40,000-65,000 EUR available over four years)[7]. Some projects that involve research on rare diseases are financed by Targeted Financing from the Estonian Government (dysmorphic syndromes, methylation defects such as Prader-Willi, Silver-Russell and Beckwith-Wiedemann syndrome, metabolic diseases such as phenylketonuria, classical galactosemia, mucopolysaccharidoses, fatty acid oxidation defects and mitochondrial diseases, and congenital adrenal hyperplasia).

Participation in European research projects

Estonian teams participate, or participated, in European rare disease research projects, including: AAVEYE, EURAPS, MOLDIAG-PACA and RD PLATFORM. Estonia contributes to the EURO-WABB registry project.

E-Rare

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

IRDiRC

Estonian funding agencies have not yet committed national funding to the IRDiRC.

Orphan medicinal products

Orphan medicinal product committee

There is currently no orphan medicinal product committee in Estonia. On the sphere of drugs the main adviser is the Drug Committee. The main objective of the Drug Committee is to advise the Ministry of Social Affairs on the positive list of reimbursement medicines and to debate about the use of pharmaceuticals and about the need to update the reimbursement rules for some product or group of products. Orphan medicinal products are subject to Drug Committee on the same basis as other medicines.

Orphan medicinal product incentives

There are no specific incentives for orphan medicinal products in Estonia.

Orphan medicinal product market availability situation

In theory, all orphan medicinal products with EU market authorisation can be bought in Estonia. All information concerning drugs, including orphan medicinal products is available in Ravimiamet[8] (*Stage Agency of Medicine of Estonia*).

Orphan medicinal product pricing policy

There is no specific pricing policy for orphan medicinal products in Estonia.

Orphan medicinal product reimbursement policy

There is no concrete list of orphan medicines for reimbursement and no specific programmes to facilitate the provision of medicines to rare disease patients. Reimbursement of the cost of medicines to patients comes from joint medical-insurance funds on the basis of *Eesti Haigekassa's* (Estonian Health Insurance Fund) medicine reimbursement budget in accordance with the diagnosis, where the criterion for establishing the selection of corresponding diagnoses is not so much the incidence of the disease as its seriousness and mortality, the possibility of an epidemic, the need for alleviating the associated pain or other humane considerations, the chronic nature of the disease together with the impairment caused to the quality of life, and the match with the financial possibilities of the medical insurance scheme. Children under the age of 4 are entitled to 100% drug reimbursement. Rare diseases are also included in the catalogue of described diagnoses for reimbursement. Currently[9] *Haigekassa* reimburses patients 100% of the costs of 21 orphan medicinal products.

[7] This section is 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* (p 11)

[8] http://www.sam.ee/

[9] Information accurate in December 2011.

Due to the high cost of these orphan medicinal products, only those which are reimbursed by *Eesti Haigekassa* (Estonian Health Insurance Fund) are easily accessible. Patients can access all other orphan medicinal products if they are willing to pay the cost of the drug.

Other initiatives to improve access to orphan medicinal products
There are no specific programmes to facilitate the provision of medicines to rare disease patients.

Orphan devices
No specific information reported.

Specialised social services
The Estonian Agrenska Foundation, founded by several sources including Agrenska Sweden, the University of Tartu, the Estonian Board of Disabled People, the Tartu University Hospital Foundation, and *Stenstroms Skjortfabrik Eesti* provides counselling and care for families with children with rare disorders. Like its Swedish counterpart, the Estonian Agrenska Foundation[10] targets the family, offering a family-centred counselling system that should be able to cover all of Estonia in the coming future. The service focuses on families of children with disabilities, offering psychosocial, educational and medical information and support. In 2011, several respite camps were organized by the Estonian Agrenska Foundation. The reimbursement of these services varies from full reimbursement to partial payment by patients. Every family with a disabled child is entitled to a fixed sum per year from the government for respite care services. The Maarja Village Foundation (founded by the Tartu Toome Rotary Club) runs a residential centre which accommodates up to 33 young people with mental disabilities. Therapeutic recreational programmes exist for certain rare diseases (Prader Willi for example) and are provided by patient organisations and are partially reimbursed. Services exist to promote the integration of patients with disabilities in schools and in the work place and are financed by the government.

DEVELOPMENT OF RARE DISEASE ACTIVITIES IN 2011 IN ESTONIA

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Sources of information on rare diseases and national help lines
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In 2011 the Orphanet Estonia national website[11], in Estonian, was launched by the Orphanet Estonia team.

[10] http://www.agrenska.ee/
[11] http://www.orpha.net/national/EE-ET/index/avaleht/

Training and education initiatives

In 2009-2011 the number of number of advanced courses on rare disorders organised by the Department of Continuing Education at the Tartu Medical University increased, due to rising interest in the subject. This activity is also planned for the future (i.e. a new course is planned for 2012).

Research activities and E-Rare partnership

IRDiRC

Estonian funding agencies have not yet committed national funding to the IRDiRC.

Specialised social services

In 2011, several respite camps were organised by the Estonian Agrenska Foundation[12].

[12] http://www.agrenska.ee/

LIST OF CONTRIBUTIONS[13]

Contributions in 2010
Andres Metspalu and Merike Leego *(Orphanet Estonia,Tartu University)*
Vallo Tillman *(COMP Representative, University of Tartu)*

Contributions in 2011
Vallo Tillman *(Orphanet Estonia, COMP Representative, University of Tartu)*
Inna Vabamäe *(Ministry of Social Affairs)*

Contributions in 2012
Vallo Tillman *(Orphanet Estonia, COMP Representative, University of Tartu)*
Inna Vabamäe *(Ministry of Social Affairs)*
Riina Žordania and Katrin Õunap *(Department of Genetics under Tartu University Hospital)*
Validated by: Inna Vabamäe *(EUCERD Representative Estonia, Ministry of Social Affairs)*

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  http://www.agrenska.ee/?setlang=15
- Ravimiamet – State Agency for Medicines
  http://www.sam.ee/
- Orphanet Estonia national website
  http://www.orpha.net/national/EE-ET/index/avaleht/

[13] 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.
[14] 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