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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 LITHUANIA

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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 Lithunia. 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 LITHUANIA

Definition of a rare disease

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

National plan/strategy for rare diseases and related actions

A working group was established by Order No. V-564 of 1 Jun 2011 of the Ministry of Health of the Republic of Lithuania to frame the National Plan on activities related to rare diseases which will be finalised for the second half of 2012. Representatives from the universities, university hospitals, non-governmental organisations of patients and medical professionals as well as state institutions (Ministry of Health, the National Health Insurance Fund, the State Medicines Control Agency) are involved in the working group.

Expenses for health care services and drugs are reimbursed from the Compulsory Health Insurance Fund budget as for other groups of patients. Additionally, expenses for the treatment of rare cases abroad are reimbursed from Compulsory Health Insurance Fund budget; compensation for orphan medicinal products and medicinal devices for rare diseases and conditions are paid from a selected part of the budget of the Compulsory Health Insurance Fund (Order No. 151 of 20 March 1998 of the Ministry of Health).

Centres of expertise

There are no official centres of expertise in Lithuania, but two centres (Centre for Medical Genetics in Vilnius University Hospital Santariškių Clinics and the Hospital of Kaunas University of Medicine) provide genetics services and diagnostic services for rare diseases to the Lithuanian population. A specialist group has been created, at the Centre for Medical Genetics at Vilnius University Hospital Santariškių Klinikos, for the creation of rare diseases management plans and a rare diseases registry. An outpatient clinic for cystic fibrosis patients was established in 2011.

Pilot European Reference Networks

Lithuanian teams participate, or have participated, in the following European Reference Networks for rare diseases: ECORN CF and PAAIR.

Registries

Lithuania contributes to the EUROCARE CF registry. A specialist group has been created, at the Centre for Medical Genetics at Vilnius University Hospital Santariškių Klinikos, for the creation of a rare diseases registry.

Neonatal screening policy

Newborn screening programmes are in place for phenylketonuria and hypothyroidism (Order No. V-865 of the Minister of Health of Republic of Lithuania of 6 December 2004 "Regarding the Approval of Universal Screening of Newborns for Inborn Metabolism Disorders Procedures").

The basic prices paid from Compulsory Health Insurance Fund budget for the newborn screening programmes for phenylketonuria and hypothyroidism were re-counted and approved by the Order No. V-962 of the Minister of Health of Republic of Lithuania in 10 November 2011.

Genetic testing

Genetic testing is provided for patients of high risk group according to Ministry of Health Decree Nr.V-522 of 23 June 2005; the expenses related to this testing are reimbursed from Compulsory Health Insurance Fund budget.

Diagnostic tests are registered as available in Lithuania for 4 genes and an estimated 3 diseases in the Orphanet database[2]. In December 2011 Lithuania delivered an extended list of genes and diseases (available for diagnostics) and now is waiting for approval.

National alliances of patient organisations and patient representation

Although there is no alliance of rare disease patient organisations In Lithuania, a Council of Representatives of Lithuanian Patient Organisations is in place which brings together about 20 different non-governmental patient

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

organisations (including rare disease) and seeks to achieve common goals and rights. There are several separate patient organisations for patients with rare diseases, including phenylketonuria, rare oncological diseases, Alpha-1-antitrypsin insufficiency disease, haemophilia, patients with impaired hearing, cystic fibrosis.

Patient organisations receive funding mainly from private sponsorship, donations and income tax donations. Patient organisations are represented at the Compulsory Health Insurance Council, and at the Council of Representatives of Patient Organisations under the Ministry of Healthcare. Members of patient organisations are involved into working groups organised by orders of the Health Ministry, Parliament, and the representatives of patient organisations also participate in the Patient's Rights and Damage for Health Compensation Commission at the Health Ministry. Members of patient organisations will be involved in elaborating the national plan for rare diseases as well.

Sources of information on rare diseases and national help lines

Orphanet activities in Lithuania

Since 2004 there is a dedicated Orphanet team in Lithuania, currently hosted by the Department of Human and Medical Genetics at the University of Vilnius. The Ministry of Health designated this team as the official Orphanet team for Lithuania 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. The team also manages the Orphanet Lithuania[3] national website in Lithuanian.

Official information centre on rare diseases

The only official common information source on rare diseases in Lithuania - is Orphanet.

Helpline

There is currently no help line dedicated to providing information on rare diseases, but other general help lines (e.g. providing psychological support) exist.

Other sources of information on rare diseases

Lithuania is a partner in the ECORN-CF project which maintains a website and forum for patients with cystic fibrosis, their relatives and any other interested parties where they can ask questions and obtain answers from experts. Although the EC-funding of this project has ended, the service continues to be sustained through other sources of financing.

Good practice guidelines

A "National agreement for cystic fibrosis diagnostic and treatment: evidence based methodical recommendations"[4] was published in the journal *Paediatric pulmonology and allergology* (Vol. XIII, Nr. 2): this agreement was reached in October 2010 between paediatricians and pulmonologists and concerns best practice for cystic fibrosis treatment.

A national agreement for cystic fibrosis diagnostic, treatment and management for adults was reached and published in journal *Pulmonology, immunology and allergology (1(8), 2011*)

Training and education initiatives

A training program for the improvement in rare diseases diagnostics for doctors has been initiated and a training cycle called "Rare diseases" has been introduced for medical students.

National rare disease events in 2011

Various rare disease patient organisations invited the public to participate in various events, and also organised online publications and articles in the newspapers to mark Rare Disease Day.

The Centre for Medical Genetics in Vilnius University Hospital Santariškių clinics issued an online invitation via their internet portal to commemorate the Rare Disease Day 2011. The Orphanet-Lithuania coordinator attended the Morning News talk show on national TV, where he spoke about the problems that patients and their families face when dealing with rare diseases in Lithuania in particular.

The Lithuanian Association for the Genetic Neuromuscular Disorders "Sraunija" produced a play entitled "Little Prince" where the main character was a boy with a rare disease. An interview of the Lithuanian Cystic Fibrosis Association chairman and a pediatric neurologist was released in the press on that day. A

[3] http://www.orpha.net/national/LT-LT/index/prad%C5%BEia/

[4] http://www.pediatrija.org

representative of the Rare Disease Initiative spoke on national radio to raise awareness of the lack of information about rare diseases and the problems of families affected by these disorders. A clinical geneticist was also interviewed on national radio about rare diseases in Lithuania and other countries, and the diagnostic and treatment possibilities.

A section on rare diseases took place in the 2011 Paediatric Congress in Vilnius.

Hosted rare disease events in 2011

No specific activity reported.

Research activities and E-Rare partnership

National research activities

In the recent years funding is available for fundamental research and research concerning medicinal products: this second area of research is in particular targeted by the European Union Structural Assistance Operational Programme 2007-2017 for Economical Growth and research projects for rare diseases may receive financial support by taking part in tendering processes. Additionally, in 2007 the Government of the Republic of Lithuania adopted the Lithuanian Research and Development Priorities for 2007-2010 (Governmental Decree No. 166, 7 February 2007) which also includes as a priority the development of medicinal products, including those targeting rare diseases.

An academic research project in Lithuania entitled "National hereditary childhood cancer research platform" which focuses on six genetic diseases (von Hippel-Lindau syndrome, Li-Fraumeni syndrome, Multiple endocrine neoplasia syndromes - MEN1 and MEN2, Familial adenomatous polyposis and Type 2 Neurofibromatosis), molecular epidemiology and establishing of molecular diagnostic facility as well as information dissemination concerning rare diseases is on-going.

Participation in European research projects

Lithuanian teams participate, or have participated, in the EUROPEAN LEUKEMIA NET research project.

E-Rare

Lithuania is not currently part of the E-Rare consortium.

IRDiRC

Lithuanian funding agencies are not currently committed members of the IRDiRC.

Orphan medicinal products

Orphan medicinal product committee

A committee, set up by the Health Ministry's State Patient Fund, responsible for taking decisions on medicines and medical treatment for very rare diseases and conditions. Lithuania as an EU MS has a national representative at the COMP.

Orphan medicinal product incentives

No specific activity reported.

Orphan medicinal product market availability situation

Orphan medicinal products are available in the same way as the medicines authorised in all EU states. The website of the Lithuanian State Medicines Control Agency[5] provides information including the list of authorised medicines but does not give any other information concerning orphan medicinal products apart from that provided by the EMA concerning orphan medicinal products with EU market authorisations.

29 centrally authorised products were marketed in Lithuania in 2011. These include Arzerra (ofatumumab), Atriance (nelarabine), Busilvex (busulfan), Evoltra (clofirabine), Fabrazyme (agalsidase beta), Gliolan (5-aminolevulinic acid hydrochloride), Glivec (imatinib), Increlex (mecasermin), Inovelon (rufinamide), Litak (cladribine), Lydodren (mitotane), Mozobil (plerixafor), Nexavar (sorafenib), Orfadin (nitisinone), Pedea (ibuprofen), Revatio (sildenafil), Rvlimid (lenalidomide), Revolade (eltrombopag), Sprecel (dasatinib), Tasigna (nilotinib), Tepadina (thiotepa), Thalidomide Celgene (thalidomide), Torisel (temsirolimus), Tracleer (bosentan monohydrate), Trisenox (arsenic trioxide), Ventavis (iloprost), Volibris (ambrisentan), Wilzin (zinc), Yondelis (trabectedin). The majority of these medicines were marketed, other were available on patient basis. In

[5] http://www.vvkt.lt/

addition, nationally authorised anagrelide (Thromboreductin) and inhaled tobramycin (Tobramycin Norameda) were available in 2011.

Orphan medicinal product pricing policy
No specific activity reported.

Orphan medicinal product reimbursement policy
According to the *Inventory of Community and Member States' incentive measures to aid the research, marketing, development and availability of orphan medicinal products*[6] concerning Lithuania, *"compensation for orphan medicinal products and medicinal products for rare diseases and conditions is paid for out of the funds earmarked for that purpose in the budget of the compulsory health insurance fund (Ministry of Health Decree No 151 of 20 March 1998; Official Gazette, 1998, No 33-894; 1999, No 7-159). A list of reimbursed medical products is available (the last update is dated 2 February 2009, Ministry of Health Decree No V-52, regarding the amendment of Order No.49 of 28 January 2000 "Regarding the Approval of the List of Reimbursed Medicinal Products"). Individuals are compensated for the purchase of medicinal products for rare diseases and conditions on presentation of specialist doctors' reports, following a decision by the committee, set up by the the National Health Insurance Fund under the Ministry of Health, responsible for taking decisions on medicines and medical treatment for very rare diseases and conditions and on cases for which no provision has been made (Decree of the Director of the National Health Insurance Fund under the Ministry of Health No 1K-149 of 22 November 2005; Official Gazette, 2005, No 139-5037)."*

Other initiatives to improve access to orphan medicinal products
No specific activity reported.

Orphan devices
The Committee at *the National Health Insurance Fund under the Ministry of Health* responsible for taking decisions on medicines and medical treatment for very rare diseases and conditions also makes decisions on compensation of orthopaedic hardware in rare diseases and conditions.

Specialised social services
Respite care services are available and are organised by municipalities and hospital clinics: the Kaunas Children's Development Clinic, the Centre of Children's Development at Vilnius University Children's Hospital and day care centres for mentally disabled patients at municipal level. Therapeutic recreational programmes are available and are provided by local authorities under the administration of municipalities and directed at government level by the Ministry of Social Security and Labour. Some municipalities provide public services but these services are mostly run by private bodies (either companies or patient groups) commissioned by the social authorities. Educational camps are available for children, organised by the Ministry of Education. Rehabilitation issues are regulated by Healthcare Ministerial Order No. V-50 (17 January 2008) "Regarding the Organisation of Medical Rehabilitation and Sanatorium". Governmental services are available to promote social integration including integration in schools and the work place of patients with disabilities: this includes personalised secondary training syllabi and a special integration programme for sick and disabled persons for the labour market.

[6] *Inventory of Community and Member States' incentive measures to aid the research, marketing, development and availability of orphan medicinal products (2005 revision)* (p17)

DEVELOPMENT OF RARE DISEASE ACTIVITIES IN 2011 IN LITHUANIA

National plan/strategy for rare diseases and related actions

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Centres of expertise

An outpatient clinic for cystic fibrosis patients was established in 2011.

Neonatal screening policy

The basic prices paid from Compulsory Health Insurance Fund budget for the newborn screening programmes for phenylketonuria and hypothyroidism were re-counted and approved by the Order No. V-962 of the Minister of Health of Republic of Lithuania in 10 November 2011.

Sources of information on rare diseases and national help lines

Orphanet activities in Lithuania

In 2011, the Orphanet Lithuania national website was launched by the Orphanet Lithuania team, in Lithuanian[7].

Good practice guidelines

A national agreement for cystic fibrosis diagnostic, treatment and management for adults was reached and published in journal *Pulmonology, immunology and allergology (1(8), 2011*)

National rare disease events in 2011

Various rare disease patient organisations invited the public to participate in various events, and also organised online publications and articles in the newspapers to mark Rare Disease Day.

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Research activities and E-Rare partnership

IRDiRC

Lithuanian funding agencies are not currently committed members of the IRDiRC.

[7] http://www.orpha.net/national/LT-LT/index/prad%C5%BEia/

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- Lithuanian State Medication Control Agency
  http://www.vvkt.lt/
- Orphanet Lithuania national website
  http://www.orpha.net/national/LT-LT/index/prad%C5%BEia/

[8] 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.
[9] 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