2014 REPORT ON THE STATE OF THE ART OF RARE DISEASE ACTIVITIES IN EUROPE

STATE OF THE ART OF RARE DISEASE ACTIVITIES IN LUXEMBOURG

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

The European Union Committee of Experts on Rare Diseases (EUCERD) was established in 2009 and its mandate ended in 2013. It is replaced from 2014 by the Commission Expert Group on Rare Diseases. The EUCERD Joint Action continues to support the activities of the new Expert Group until 2015.

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

Disclaimer:
The findings and conclusions in this report are those of the contributors and validating authorities, who are responsible for the contents; the findings and conclusions do not necessarily represent the views of the European Commission or national health authorities in Europe. Therefore, no statement in this report should be construed as an official position of the European Commission or a national health authority.

Copyright information:
The “2014 Report on the State of the Art of Rare Disease Activities” is copyrighted by the Scientific Secretariat of the EUCERD Joint Action: Working for Rare Diseases (N° 2011 22 01). This product and its contents may be used and incorporated into other* materials on the condition that the contents are not changed in any way (including covers and front matter) and that no fee is charged by the reproducer of the product or its contents for their use. The product may not be sold for profit or incorporated into any profit-making venture without the expressed written permission of the EUCERD Scientific Secretariat. Specifically:

1) When the document is reprinted, it must be reprinted in its entirety without any changes.
2) When parts of the documents are used or quoted, the following citation should be used.

*Note: The “2014 Report on the State of the Art of Rare Disease Activities in Europe” contains material copyrighted by others. For material noted as copyrighted by others, the user must obtain permission from the copyright holders identified in the document.

To quote this document:


©European Union, 2014
**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 European Union Committee of Experts on Rare Diseases (EUCERD) was established in 2009 and its mandate ended in 2013. It is replaced from 2014 by the Commission Expert Group on Rare Diseases. The EUCERD Joint Action continues to support the activities of the new Expert Group until 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 2013. 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 Commission Expert Group 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 2013
- 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

Parts I – V also include a description of the methodology, sources and validation process of the entire report, and 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 Luxembourg. 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.

Each year, there are around 15,000 downloads of the different sections of the report combined.
RARE DISEASE ACTIVITIES IN LUXEMBOURG

Definition of a rare disease
Stakeholders in Luxembourg 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
The Task Force on Rare Diseases Luxembourg ("Groupe de travail maladies rares") was created in 2005 to analyse the needs of rare disease patients in the country and to develop a national strategy for improvement. This Task Force carried out a survey ("Rare diseases: a national survey on the situation of persons with rare diseases in Luxembourg") carried out between May 2006 and February 2007 aimed at analysing the strengths and weaknesses of the healthcare system and the experiences of rare disease patients. The results of this survey were published on 28 February 2011. The results of the survey show that, as in many other European countries, obtaining a diagnosis is often difficult for rare diseases patients in Luxembourg; that medical and scientific knowledge is often insufficient, as Luxembourg does not have university hospitals or specialised investigation centres; that often there is a lack of information on diseases or specialised treatment centres nearby. Due to the small size of the country, the proximity to a huge number of university hospitals in the neighbouring countries Belgium, Germany and France, and the fact that most medical doctors are trained in these universities, good professional collaboration with specialised centres abroad has been in action for many years. Also the orientation of patients (patients with rare diseases, but also patients needing highly specialised care such as complicated surgical interventions, very specialised diagnostic interventions for more frequent diseases) to a specialist or a specialised centre abroad is a procedure foreseen via the national sickness fund.

According to the results of the survey, 44 % of patients with rare diseases residing in Luxembourg have been oriented by their treating doctor for diagnostic purposes to specialised centres abroad. The medical care of 54 % of the patients is delivered in Luxembourg, 22% are followed for medical purposes in specialised centres in the neighbouring countries (Germany, Belgium and France), and for 24% a shared medical care program has been established between local doctors or hospital departments and abroad specialists. 50% of patients travel 1 to 5 times a year abroad for medical care, knowing however that the distances to these centres in general do not exceed 500km. Patients often lament the lack of coordination between the health professionals; there is a lack of quality care for a large number of rare pathologies; parts of the care and treatment might not be covered by the sickness fund and so there are inequalities in the access to a diagnosis, treatments and care; and rare diseases might have serious social consequences. A list of recommendations have been made on the basis of these results including: the elaboration of a national plan for rare diseases; the improvement of information and awareness of rare diseases; a guarantee of equal access to diagnosis, care and treatment; the provision of specific help services for patients with rare diseases and their families; support of rare disease patient organisations and their involvement in national rare disease actions; intensification of international collaboration; promotion of advanced research; and support for the sustainability of rare disease initiatives at national level.

Concertation for the elaboration of a national strategy was reinforced at national level in 2013 based on a broad interdisciplinary Europlan conference. A number of meetings were held before the November 2013 Europlan conference to raise awareness amongst key stakeholders from different domains (political, medical, patient associations, education, social services, reimbursement etc.) regarding specific problems linked to rare diseases. A great majority of the addressed stakeholders participated very actively in the Europlan conference which aimed to contribute to the identification of concrete proposals for the elaboration of a national strategy. This endeavour will be carried on in 2014/2015.

Centres of expertise
There are currently no official centres of expertise for rare diseases in Luxembourg.

---

1 Les maladies rares: Enquête sur la situation des personnes atteintes de maladies rares au Grand-Duché de Luxembourg

2 Les maladies rares: Enquête sur la situation des personnes atteintes de maladies rares au Grand-Duché de Luxembourg

3 http://www.eurordis.org/sites/default/files/flags/finalreport-luxembourg.pdf
Registries
Luxembourg contributes to the EUROCARE CF European registry. A cancer registry is under construction, which will include also the registration of rare cancers. A special effort has been made since 2013 to include Orphacodes when possible to the cause of death registration.

Neonatal screening policy
A national neonatal screening programme is in place for phenylketonuria (since 1968), congenital hyperthyroidism (since 1978), congenital adrenal hyperplasia (since 2001) and Medium-Chain Acyl-CoA Deficiency (since 2008).

Genetic testing
Modifications of the regulations concerning the sickness fund are undertaken to improve the reimbursement of genetic tests.

National alliances of patient organisations and patient representation
The Luxembourg Association for Neuromuscular and Rare Disorders (ALAN absl.) was established in 1998 to represent patients with neuromuscular diseases: since 2006 they have provided support for other rare disease patients for whom there is no other patient organisation. The association organises informative events, counselling, guidance and self-help groups and is involved in the work of the Task Force on Rare Diseases Luxembourg. There are other patient organisations for single or groups of rare diseases. There is the intention to create an alliance of rare disease patient organisations.

Sources of information on rare diseases and national help lines

Orphanet activities in Luxembourg
Since 2006, Luxembourg cooperates with Orphanet, through one contact member situated at the Ministry of Health.

Official information centre for rare diseases
The Task Force has plans to put into place a national rare diseases platform which offers medical and social services, a rare disease hotline, counselling, self-help groups, specialised information on rare diseases and guidelines of best practices. A guide to all medical, paramedical and social services available to rare disease patients and their family should also be made available online.

Help line
A rare disease help line is one of the activities to be hosted by the national rare diseases platform.

Other sources of information on rare diseases
Increasing knowledge and use of the Orphanet database by medical professionals and the general public has been observed.

Good practice guidelines
In general health professionals will adopt good practice guidelines existing abroad, guidelines identified by the Orphanet database or elaborated by recognised centres of expertise abroad.

Training and education initiatives
No specific information reported.

National rare disease events in 2013
During 2013 several awareness-raising activities were organised by ALAN absl partly together with other partners. To mark Rare Disease Day 2013, a day of festivities was organised to raise public awareness and funding. An art exhibition dealing with different aspects of rare diseases ran from December 2013 to February 2014

A major event was the organisation of a Europlan national conference⁴ on 19-20 November 2013 with the participation of members from the European Commission, Eurordis and EUCERD, gathering a huge number of national key stakeholders from the medical, the political and the social security fields. Many patient

⁴ http://www.eurordis.org/sites/default/files/flags/finalreport-luxembourg.pdf
associations participated, as well as research centres and social services. The aim of the conference was to contribute to: increasing the awareness of the specific problems linked to rare diseases, identifying key persons for the contribution in the existing taskforce, identifying the main problems existing in the area of rare diseases, defining priorities for action, and beginning the formulation of the national rare disease strategy.

**Hosted rare disease events in 2013**
The meetings of the EUCERD were hosted by the European Commission in Luxembourg in 2013.

**Research activities and E-Rare partnership**

**National research activities**
An annual rare disease telethon, organised by the Lions Club, raises money and pools this with that of the AFM (*Association française contre les myopathies*) which then redistributes these funds to research projects, a small amount comes back to Luxembourg for work with patients.

**Participation in European research projects**
Teams from Luxembourg participate in 3 FP7 rare disease related projects, involving a number of research institutes such as the Luxembourg Centre for Systems Biomedicine and the Centre for Public Health Research (in particular for cancers and rare cancers).

**E-Rare**
No information

**IRDiRC**
A member from the National Taskforce participates as an observer in IRDiRC. However, there are no funding has been committed by agencies from Luxembourg.

**Orphan medicinal products**

**Orphan medicinal product committee**
The Pharmacy and Medicines division of the Directorate of Health/Ministry of Health is dealing with the accessibility of orphan drugs in Luxembourg. The Task Force aims to create a national medical commission to consult on issues regarding exceptional access to and reimbursement of orphan medicinal products.

**Orphan medicinal product incentives**
No specific information reported.

**Orphan medicinal product market availability situation**

**Orphan medicinal product pricing policy**
No specific information reported.

**Orphan medicinal product reimbursement policy**
No specific information reported.

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

**Orphan devices**
No specific information reported.

---

5 As of April 2013.
Other therapies for rare diseases
No specific information reported.

Specialised social services
No specific information reported.

DEVELOPMENT OF RARE DISEASE ACTIVITIES IN 2013 IN LUXEMBOURG

National plan/strategy for rare diseases and related actions
Concertation for the elaboration of a national strategy was reinforced at national level in 2013 based on a broad interdisciplinary Europlan conference. A number of meetings were held before the November 2013 Europlan conference to raise awareness amongst key stakeholders from different domains (political, medical, patient associations, education, social services, reimbursement etc.) regarding specific problems linked to rare diseases. A great majority of the addressed stakeholders participated very actively in the Europlan conference which aimed to contribute to the identification of concrete proposals for the elaboration of a national strategy. This endeavour will be carried on in 2014/2015.

Registries
A special effort has been made since 2013 to include Orphacodes when possible to the causes of death registration.

National rare disease events in 2013
During 2013 several awareness-raising activities were organised by ALAN absl partly together with other partners. To mark Rare Disease Day 2013, a day of festivities was organised to raise public awareness and funding. An art exhibition dealing with different aspects of rare diseases ran from December 2013 to February 2014.

A major event was the organisation of a Europlan national conference on 19-20 November 2013 with the participation of members from the European Commission, Eurordis and EUCERD, gathering a huge number of national key stakeholders from the medical, the political and the social security fields. Many patient associations participated, as well as research centres and social services. The aim of the conference was to contribute to: increasing the awareness of the specific problems linked to rare diseases, identifying key persons for the contribution in the existing taskforce, identifying the main problems existing in the area of rare diseases, defining priorities for action, and beginning the formulation of the national rare disease strategy.

Hosted rare disease events in 2013
The meetings of the EUCERD were hosted by the European Commission in Luxembourg in 2013.

---

1 http://www.eurordis.org/sites/default/files/flags/finalreport-luxembourg.pdf
2 http://www.eurordis.org/sites/default/files/flags/finalreport-luxembourg.pdf
LIST OF CONTRIBUTIONS

Contributions in 2010
Yolande Wagener (Orphanet Luxembourg, Ministry of Health)

Contributions in 2011
Yolande Wagener (Orphanet Luxembourg, Ministry of Health)

Contributions in 2012
Yolande Wagener (Orphanet Luxembourg, Ministry of Health)
Guy Weber (Ministry of Health)
ALAN

Contributions in 2013
Yolande Wagener (Orphanet Luxembourg ECEGRD, National Taskforce for Rare Diseases, Ministry of Health)
Olivier Lepanto (Division de la Pharmacie et des Médicaments, Ministry of Health)
Shirley Feider & Jos Even (ALAN, National Taskforce for Rare Diseases)

Validated by: Yolande Wagener (ECEGRD Representative Luxembourg, Ministry of Health)

SELECTED BIBLIOGRAPHY AND SOURCES

- Groupe de travail maladies rares
  http://www.maladiesrares.lu/start.html
- Report Les maladies rares: Enquête sur la situation des personnes atteintes de maladies rares au Grand-Duché de Luxembourg
- Orphanet Luxembourg national website
  http://www.orpha.net/national/LU-LB/index/homepage/
- ALAN
  http://www.alan.lu/
- Luxembourg Europlan National Conference Report 2013
  http://www.eurordis.org/sites/default/files/flags/finalreport-luxembourg.pdf

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 2014.