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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 LUXEMBOURG

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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 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[1].

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

# 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 will work out a national plan for rare diseases based on the results of a survey 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[2].

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 abroad. Whereas the orientation of patients to a specialist or a specialised centre abroad (when these are identified) is a procedure foreseen in the national sickness fund, patients regret an lack of coordination between the health professionals; there is a lack of quality care for quite a number of rare pathologies; as parts of the care and treatment might not be covered by the sickness fund there are inequalities in the access to a diagnosis, treatments and care; and that rare diseases 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; the guarantee of equal access to diagnosis, care and treatment; the provision of specific help services for patients with rare diseases and their families; to the support of rare disease patient organisations and their involvement in national rare disease actions; to intensification of international collaboration; the promotion of advanced research; and the support for the sustainability of rare disease initiatives at national level.

According to the results of the survey, 95% of patients with rare diseases residing in Luxembourg have sought or have been oriented by their treating doctor for medical care in neighbouring countries (such as Germany, Belgium and France), with 50% of patients travelling 1 to 5 times a year abroad for medical care.

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

**Pilot European Reference Networks**
Luxembourg does not currently participate, or has not participated, in any European Reference Networks for rare diseases.

**Registries**
Luxembourg contributes to the EUROCARE CF European registry.

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

**Genetic testing**
No specific information reported.

[2] *Les maladies rares: Enquête sur la situation des personnes atteintes de maladies rares au Grand-Duché de Luxembourg* http://www.sante.public.lu/fr/catalogue-publications/maladies-traitements/maladies-rares/index.html

**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 rare diseases: 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.

**Sources of information on rare diseases and national help lines**
***Orphanet activities in Luxembourg***
Since 2006, there is a dedicated Orphanet team in Luxembourg, currently hosted by the Ministry of Health. 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 Luxembourg for entry into the Orphanet database.

***Official information centre for rare diseases***
The Task Force has plans to soon 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***
No specific information reported.

**Good practice guidelines**
No specific information reported.

**Training and education initiatives**
No specific information reported.

**National rare disease events in 2011**
To mark Rare Disease Day 2011, the Ministry of Health of Luxembourg together with the National Interdisciplinary Rare Disease Working Group has organised a press conference on 28 February 2011 to launch the national report : "Rare diseases: a national survey on the situation of persons with rare diseases in Luxembourg.[3]" In 2005, following the European Conference on Rare Diseases held under Luxemburg's Presidency of the EU, an interdisciplinary working group was constituted, gathering medical experts, neurologists, paediatricians, biological specialists, experts in metabolic diseases, the patient's association ALAN and the Ministry of health. The objective was the constitution of a platform for the interdisciplinary exchange, the study of the situation in Luxembourg and the elaboration of initiatives for improvement

The analysis of the situation of rare diseases in Luxembourg was a priority. The objectives of the study were to evaluate the burden of disease of rare diseases in Luxembourg, to analyse the medical and psychosocial care the patients receive, to collect feedback on problems and needs (in the medical, social, education, professional and leisure area), to evaluate the strengths and weaknesses of the health and social security system as concerns rare disease patients. Based on the results of the inquiry recommandations were formulated for the elaboration of a National Plan for Rare Diseases.

**Hosted rare disease events in 2011**
No hosted events were reported in 2011.

**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, including some in Luxembourg.

[3] *Les maladies rares: Enquête sur la situation des personnes atteintes de maladies rares au Grand-Duché de Luxembourg* http://www.sante.public.lu/fr/catalogue-publications/maladies-traitements/maladies-rares/index.html

***Participation in European research projects***
Luxembourg does not currently participate, or has not participated, in any European research projects for rare diseases.

***E-Rare***
Luxembourg is not currently a partner of the E-Rare project.

***IRDiRC***
Funding agencies from Luxembourg are not currently partners of the IRDiRC.

**Orphan medicinal products**
***Orphan medicinal product committee***
The Task Force aims to create a national medical commission to consult on issues regarding access to and reimbursement of orphan medicinal products.

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

***Orphan medicinal product market availability situation***
No specific information reported.

***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.

**Specialised social services**
No specific information reported.

# DEVELOPMENT OF RARE DISEASE ACTIVITIES IN 2011 IN LUXEMBOURG

**National plan/strategy for rare diseases and related actions**
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[4] *Les maladies rares: Enquête sur la situation des personnes atteintes de maladies rares au Grand-Duché de Luxembourg* http://www.sante.public.lu/fr/catalogue-publications/maladies-traitements/maladies-rares/index.html

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**National rare disease events in 2011**

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**Research activities and E-Rare partnership**

***IRDiRC***

Funding agencies in Luxembourg are not currently partners of the IRDiRC.

[5] *Les maladies rares: Enquête sur la situation des personnes atteintes de maladies rares au Grand-Duché de Luxembourg* http://www.sante.public.lu/fr/catalogue-publications/maladies-traitements/maladies-rares/index.html

# LIST OF CONTRIBUTIONS[6]

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Contributions in 2011
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  http://www.sante.public.lu/fr/catalogue-publications/maladies-traitements/maladies-rares/index.html
- Orphanet Luxembourg national website
  http://www.orpha.net/national/LU-LB/index/homepage/
- ALAN
  http://www.alan.lu/


[6] 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.
[7] 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