2013 REPORT ON THE STATE OF THE ART OF RARE DISEASE ACTIVITIES IN EUROPE

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

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 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 2012. 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 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 2012
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

Each part contains the following 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.
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 is working on a national plan for rare diseases based on the results of 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 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 a 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.

Concertation on a plan was reinforced at Ministry level in 2012.

Centres of expertise
There are currently no official centres of expertise for rare diseases in Luxembourg. However a new system for referrals has recently been introduced which should improve access to healthcare.

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 (since 2001) and Medium-Chain Acyl-CoA Deficiency (since 2008).

Genetic testing
No specific information reported.

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

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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 organisations. 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, 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 2012
To mark Rare Disease Day 2012, a week of awareness-raising activities were organised by ALAN absl from 22 to 29 February 2012 at the Belle Etoile shopping centre: this event included a photo exhibition including photos of people with rare diseases posing next to celebrities from Luxembourg. A number of personalities participated, including the Deputy and Bourgmestre of the town of Luxembourg Mr. Bettel.

Hosted rare disease events in 2012
No hosted events were reported in 2012.

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.

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 have not yet committed funding to 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

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.

Other therapies for rare diseases
No specific information reported.

Specialised social services
No specific information reported.

DEVELOPMENT OF RARE DISEASE ACTIVITIES IN 2012 IN LUXEMBOURG

National plan/strategy for rare diseases and related actions
Concertation on a plan was reinforced at Ministry level in 2012.

Centres of expertise
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\(^3\) As of April 2013.
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LIST OF CONTRIBUTIONS

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Contributions in 2011
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- Report Les maladies rares: Enquête sur la situation des personnes atteintes de maladies rares au Grand-Duché de Luxembourg
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  http://www.orpha.net/national/LU-LB/index/homepage/
- ALAN
  http://www.alan.lu/