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European Union Committee of Experts on Rare Diseases

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

STATE OF THE ART OF RARE DISEASE ACTIVITIES IN ICELAND

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EUCERD Joint Action

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.

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

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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.

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http://www.eucerd.eu/upload/file/Reports/2013ReportStateofArtRDActivitiesIS.pdf

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 Iceland. 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.

RARE DISEASE ACTIVITIES IN ICELAND

Definition of a rare disease
In Iceland a condition is defined as rare if it affects 2 or fewer individuals per 10 000.

National plan/strategy for rare diseases and related actions
In Iceland there is not yet a specific plan for rare diseases. In 2012, for the first time, Iceland was able to send a representative to a EUCERD meeting, which already has increased discussion and awareness of rare diseases in Iceland at the level of the Ministry, stakeholders and rehabilitation services. Representatives from these organisations now meet regularly with the purpose to increase awareness of rare diseases within the country. A strategy for disabilities was launched in 2012 and the plan is to revise this document next year and include a chapter on rare diseases within it. Currently, there is no specific funding in place for rare diseases. In 2012 Iceland hosted a Nordic meeting on rare diseases with 120 attendees from all the Nordic countries.

Centres of expertise
Up to now, no centres in Iceland have been designated as national centres of expertise. However, since 1986 one national habilitation centre provides services for children and adolescents with conditions leading to disability, including rare diseases and syndromes. The same accounts for diagnostic and habilitation services for visually impaired and hearing impaired children and adults including those with rare diagnosis. These services are centralized at two national centres. Administrative databases designed to collect and store data on causes of disability (including rare diseases/syndromes) are developed at these three centres and updated regularly. The plan is to further enhance services for individuals with rare diseases at these centres.

Registries
In Iceland there is no formal national committee dedicated to dealing with registries but administrative databases (see above) are organized by directors of the individual national centres. In 2012, a centralised database on causes of deaf blindness (combined hearing and visual impairment) among Icelandic children and adults was established in cooperation of these three national centres servicing people with disabling conditions. These administrative databases are financed by the state.In addition, Icelandic teams contribute to the EUROCARE CF and RARECARE European registries.

Neonatal screening policy
There have not been any developments in the neonatal screening policy in 2012 nor have there been additions to the list of rare diseases tested. Since 1 January 2008 neonates in Iceland have been screened for 42 different rare diseases using tandem mass spectrometry.

Genetic testing
Genetic testing (as well as genetic counseling) in Iceland is centralised at the Department of Genetics and Molecular Biology of the National University Hospital of Iceland. Other than guidelines regarding the newborn screening, Iceland does not have national guidelines regarding genetic testing. Genetic testing abroad is possible as specimens are frequently sent abroad for further testing which is not possible to perform in Iceland. The patients pay only a small proportion of the total cost of genetic testing (universal health care) but there are no specific provisions in place for patients with rare diseases.

National alliances of patient organisations and patient representation
In Iceland there is one patient organisation focusing on children with rare diseases and their families. The organisation is called “Unique children”. In 2012, collaboration was initiated between representatives from the organisation “Unique children”, the Ministry of Welfare and the national habilitation centre with the aim to increase awareness of rare diseases at all service levels in the country.

Sources of information on rare diseases and national help lines
Orphanet activities in Iceland
There is no Orphanet team in Iceland.

Official information centre for rare diseases

In Iceland there is no official information centre for rare diseases. However, the patient organisation, Unique Children, provides information regarding rare diseases and the Nordic cooperation Rarelink (with its Icelandic link, www.rarelink.is) provides information on hundreds of rare diseases (either in the Nordic languages or English). Rarelink also offers networking for people with rare diseases i.e. people can get information on others within the Nordic countries with the same diagnosis. The organisation Unique Children is not funded by the state but by donations and specific fundraising activities.

Help line

There is no rare disease specific helpline in Iceland.

Other sources of information

Iceland has taken part in Nordic activities regarding rare diseases during the past several years. In autumn 2011 the homepage www.rarelink.is was launched and in 2012 information regarding several rare diseases and syndromes was translated into the Icelandic language and published on this website.

Good practice guidelines

Best practice guidelines for rare diseases have not been produced in Iceland.

Training and education initiatives

No rare disease specific or related training/education courses were held in 2012.

National rare disease events in 2012

The patient organisation "Unique children" celebrated Rare Disease Day for the first time in 2012. The main event was the fund raising event "Rare Run" where people ran a certain distance and raised money for the organisation. In the radio there was an hour long program dedicated to children with rare diseases. In the program children were interviewed and their favourite songs were played.

Hosted rare disease events in 2012

On 31 May and 1 June 2012 Iceland hosted a Nordic meeting on rare diseases with 120 attendees from all the Nordic countries[1].

Research activities and E-Rare partnership

Research activities

There are no specific programmes for rare diseases research in Iceland and there are not any fund-raising initiatives for rare diseases research in Iceland.

Participation in European research projects

Icelandic teams participate, or have participated, in European research projects on rare diseases, including: INTERPREGGEN, GEN2PHEN, PSYCHCNVS, EPINOSTICS, EURORETT, EUROSPA, ERCUSYN, and RARECARE.

E-Rare partnership

Iceland is currently not an E-Rare partner and has not yet participated in these calls.

IRDiRC

Icelandic funding agencies are not yet committed members of IRDiRC.

Orphan medicinal products

Orphan medicinal product committee

In Iceland there is no orphan drug committee and there are no official plans to introduce one.

Orphan medicinal product incentives

No specific information reported.

[1] http://www.greining.is/media/nordic-conference 2012/nordicconfernce2012linkar.pdf

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
A new information centre for children with rare disease and their families was opened in autumn 2012. The centre is called Leiðarljós (Guiding Light) and offers parents of children with rare diseases nursing support and detailed information regarding available services.

DEVELOPMENT OF RARE DISEASE ACTIVITIES IN 2012 IN ICELAND

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[2] http://www.greining.is/media/nordic-conference 2012/nordicconfernce2012linkar.pdf

LIST OF CONTRIBUTIONS[3]

Contributions in 2013
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SELECTED BIBLIOGRAPHY AND SOURCES[4]

- Rare link Iceland
  http://rarelink.is
- The State Diagnostic and Counselling Centre
  www.greining.is

[3] 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.
[4] All websites and documents were last accessed in May 2013.