

**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  
SLOVENIA**

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More information on the European Union Committee of Experts on Rare Diseases can be found at [www.eucerd.eu](http://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 Porphyrin 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 Slovenia. 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<sup>1</sup>.

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<sup>1</sup> <http://www.eucerd.eu/upload/file/Reports/2012ReportStateofArRDActivities.pdf>

# RARE DISEASE ACTIVITIES IN SLOVENIA

## Definition of a rare disease

Stakeholders in Slovenia 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

There is currently no national plan for rare diseases in Slovenia and there is no dedicated budget for rare diseases and costs related to rare diseases are covered by the national health care insurance scheme. A working group was created at the Ministry of Health in 2010 which prepared a draft national plan for rare diseases in 2011 which has now passed a professional and lay public consultation phase. The plan<sup>2</sup> was accepted by the Health Council in February 2012 and the next steps will be to elaborate an action plan and its implementation.

## Centres of expertise

There are no official centres of expertise in Slovenia, but the majority of patients with rare diseases in Slovenia are evaluated centrally at the University Medical Centre Ljubljana where there is an efficient system for the referral of genetic, endocrine, metabolic, and neurodegenerative disorders, amongst others. In addition to this, there is a Centre for Fabry disease in Slovenj Gradec. The establishment of centres of expertise is foreseen in the national plan for rare diseases.

## Pilot European Reference Networks

Slovenian teams participate, or have participated, in the following European Reference Networks for rare diseases: Dyscerne, NEUROPED, TAG, Care-NMD and EN-RBD.

## Registries

There is currently no national registry for rare diseases in Slovenia. A new Healthcare Databases Act, which sanctions the establishment of national registries, is under preparation, and the inclusion of registries in the area of rare diseases is expected. Slovenia contributes to the EURO CARE CF European registry.

## Neonatal screening policy

Neonatal screening is available for phenylketonuria and congenital hypothyroidism. A screening policy is also in place for hearing impairments and developmental dislocation of the hip.

## Genetic testing

Genetic testing is offered to patients when there is an indication to perform such tests recognised by a medical specialist. While there are no formally established reference centres in Slovenia, the Institute of Medical Genetics at the University Medical Centre in Ljubljana is the tertiary institution in this area. There are no specific national guidelines regulating genetic testing, those that are deemed necessary are financed by the Health Insurance Institute of Slovenia. In case a specific test not being available in Slovenia, there is a procedure in place, through which patients can obtain approval for reimbursement of genetic testing performed abroad.

Diagnostic tests are registered as available in Slovenia for 47 genes and an estimated 50 diseases in the Orphanet database<sup>3</sup>.

## National alliances of patient organisations and patient representation

There is currently no national alliance of rare disease patient organisations in Slovenia. Patient organisations are financed through different sources: this may include funding from the government/public sector and the private sector (private sponsorships and donations). The Ministry of Health financially supports some programmes within patient organisations through calls for project proposals: the aims of these calls vary.

The role of patient organisations is recognised in national plan. Patient organisation representatives are usually consulted concerning legislative proposals and in some cases are included in the process of drafting

<sup>2</sup> [http://www.mz.gov.si/fileadmin/mz.gov.si/pageuploads/redke\\_bolezni\\_2012\\_-\\_nacrtna\\_dela/Nacrtna\\_dela\\_na\\_podrocju\\_redkih\\_bolezni.pdf](http://www.mz.gov.si/fileadmin/mz.gov.si/pageuploads/redke_bolezni_2012_-_nacrtna_dela/Nacrtna_dela_na_podrocju_redkih_bolezni.pdf)

<sup>3</sup> Information extracted from the Orphanet database (September 2011).

legislation. Patient organisation representatives do not usually receive financial support in order to attend these meetings.

### **Sources of information on rare diseases and national help lines**

#### ***Orphanet activities in Slovenia***

Since 2006 there is a dedicated Orphanet team in Slovenia, currently hosted by the Institute of Medical Genetics at the University Medical Centre Ljubljana. 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. This team was designated by the Ministry of Health in 2010 as the official Orphanet team in Slovenia. The team launched in 2011 the Orphanet Slovenia national website<sup>4</sup>.

#### ***Official information centre for rare diseases***

There is no official information centre for rare diseases in Slovenia other than Orphanet.

#### ***Help line***

There is currently no information help line for rare diseases in Slovenia.

#### ***Other sources of information on rare diseases***

Information on rare diseases is available on some institutions' web sites, and web sites run by patient organisations.

#### **Best practice clinical guidelines**

National clinical guidelines are not available.

#### **Training and education initiatives**

No specific activity reported.

#### **National rare disease events in 2011**

To mark Rare Disease Day, a press conference was organised on 28 February 2011 to raise society's awareness of rare diseases, to inform the general public about rare diseases and to improve cooperation between patients and medical profession for better quality of life of patients. To raise awareness about the rare diseases in Slovenia an article has been written for Wikipedia in the Slovenian language.

#### **Hosted rare disease events in 2011**

No specific information reported.

#### **Research activities and E-Rare partnership**

##### ***National research activities***

The Slovenian Research Agency is a government body which awards grants for research. Although not specifically aimed at rare diseases, in the past rare disease topics have been given research grants.

##### ***Participation in European projects***

Slovenian teams participate, or have participated, in European rare disease research projects including: CONTICANET, EMSA-SG, MYELINET, PNSEURONET and SARS/FLU VACCINE.

##### ***E-Rare***

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

##### ***IRDIRC***

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

#### **Orphan medicinal products**

##### ***Orphan medicinal product committee***

In Slovenia, orphan medicinal products are included in public funding in the same manner as any other drug. A decision on their financing from public funds is adopted by a commission of experts in the field of medicine and

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<sup>4</sup> <http://www.orpha.net/national/SI-SL/index/domov/>

pharmacy within the Health Insurance Institute of Slovenia. Additionally a Strategic Council for Drugs operates within the Ministry of Health. It is responsible for policy and funding availability of medicinal products - particularly expensive drugs, including orphan medicinal products. The Strategic Council for Drugs in 2010 provided additional government budget funds of €1'000'000 to finance two orphan medicinal products for the treatment of two patients with a rare haemolytic condition and to treat one patient with Hunter syndrome. For the year 2011 additional funding was provided for two orphan medicinal products for the treatment of children suffering from acute lymphoblastic anemia (Evoltra) clofarabine and in preparation for bone marrow transplantation (Busilvex) busulfan. Both drugs are already available and in use within the public healthcare system.

#### **Orphan medicinal product incentives**

In Slovenia, there are several measures concerning national incentives for orphan medicinal products according to the *Inventory of Community and Member States' incentive measures to aid the research, marketing, development and availability of orphan medicinal products*, including "reduced fees for marketing authorisation procedure (if the centralised procedure was not followed)<sup>5</sup>."

#### **Orphan medicinal product market availability situation**

The orphan medicinal products launched on the market up to the end of 2011 were: Afintor, Busilvex, Cystadane, Diacomit, Elaprased, Evoltra, Exjade, Fabrazyme, Glivec, Kuvan, Litak, Lysodren, Mozobil, Myozyme, Naglazyme, Nexavar, Nplate, Replagal, Revatio, Revlimid, Revolade, Savene, Soliris, Somavert, Sprycel, Sutent, Tassigna, Tepadina, Thalidomide, Torisel, Tracleer, Ventavis, Vidaza, Volibris, Wilzin, Xyrem, Zavesca.

#### **Orphan medicinal product pricing policy**

Pricing of orphan medicinal products is subject to the same procedure as other medicinal products, which are financed from public funds. Determination of maximum prices is the responsibility of the Agency for Medicinal Products and Medical Devices of the Republic of Slovenia (JAZMP), whereas the Health Insurance Institute of Slovenia negotiates prices that are lower than those set by JAZMP. The latter sets the maximum prices taking into account those set in a selection of EU countries (Germany, France and Austria).

#### **Orphan medicinal product reimbursement policy**

In Slovenia, one of the criteria for including a drug among those covered by health insurance is an "ethical criteria" which applies in particular to severe and rare diseases: this has a positive influence on the accessibility of drugs for rare diseases patients.

The following orphan medicinal products were covered (without the need for any co-payment by the patient) by the Health Insurance Institute of Slovenia in 2010: Afintor, Busilvex, Cystadane, Diacomit, Elaprased, Evoltra, Exjade, Fabrazyme, Glivec, Kuvan, Litak, Lysodren, Mozobil, Myozyme, Naglazyme, Nexavar, Nplate, Replagal, Revatio, Revlimid, Revolade, Savene, Somavert, Sprycel, Sutent, Tassigna, Tepadina, Thalidomide, Torisel, Tracleer, Ventavis, Vidaza, Volibris, Wilzin, Xyrem, Zavesca.

In 2010, the public expenditure for orphan medicinal products increased by 26,31%, which is considerably more than the average increase in expenditure for other drugs. In 2010 (the latest data available) 900 patients were receiving orphan medicinal products in Slovenia, however, the number of patients in Slovenia with rare diseases is not known at this stage.

#### **Other initiatives to improve access to orphan medicinal products**

In Slovenia, there are several measures concerning national incentives for orphan medicinal products according to the *Inventory of Community and Member States' incentive measures to aid the research, marketing, development and availability of orphan medicinal products*, including "permission to use medicines labelled in any EU language with stickers in Slovenian language; [and] negotiation on drug prices."<sup>6</sup>

#### **Orphan devices**

No specific information reported.

#### **Specialised social services**

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<sup>5</sup> *Inventory of Community and Member States' incentive measures to aid the research, marketing, development and availability of orphan medicinal products* (2005 revision) (p19)

<sup>6</sup> *Inventory of Community and Member States' incentive measures to aid the research, marketing, development and availability of orphan medicinal products* (2005 revision) (p19)

Some respite care services are available in Slovenia for patients with disabilities, and are provided by governmental and non-governmental organisations with either government or private financing. Some services are available in Slovenia for patients with disabilities. Therapeutic recreational programmes are available for patients with disabilities in Slovenia, and are provided by governmental and non-governmental organisations with government and private financing. Services are in place promoting the social integration of patients with disabilities in the workplace: most activities are provided through government institutions.

## DEVELOPMENT OF RARE DISEASE ACTIVITIES IN 2011 IN SLOVENIA

### **National plan/strategy for rare diseases and related actions**

A working group was created at the Ministry of Health in 2010 which prepared a draft national plan for rare diseases in 2011 which has now passed a professional and lay public consultation phase. The plan was accepted by the Health Council in February 2012 and the next steps will be to elaborate an action plan and its implementation.

### **Sources of information on rare diseases and national help lines**

#### ***Orphanet activities in Slovenia***

The team launched in 2011 the Orphanet Slovenia national website<sup>7</sup>.

### **National rare disease events in 2011**

To mark Rare Disease Day, a press conference was organised on 28 February 2011 to raise society's awareness of rare diseases, to inform the general public about rare diseases and to improve cooperation between patients and medical profession for better quality of life of patients.

### **Research activities and E-Rare partnership**

#### ***IRDiRC***

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

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<sup>7</sup> <http://www.orpha.net/national/SI-SL/index/domov/>

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- Orphanet Slovenia national website  
<http://www.orpha.net/national/SI-SL/index/domov/>

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<sup>8</sup> 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.

<sup>9</sup> 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>