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

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.

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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 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.
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 not more than 5 in 10,000 individuals. The same definition is also accepted in The Work Plan for Rare Diseases.

National plan/strategy for rare diseases and related actions
In 2012 a national plan for rare diseases was accepted by the Health Council and the next steps will be to elaborate an action plan and its implementation, as well as to identify funding. The Work Plan for Rare Diseases\(^1\) is designed to serve as a roadmap until 2020 and is qualified as “... an opportunity for better coordination of efforts of all partners involved, establishing health care that will be comprehensive, accessible, timely and patient-focused”. The major objectives of the plan centre around the identification and monitoring of rare diseases; improving early diagnosis and access to appropriate medical treatments; mechanisms to improve an integrated approach to rare diseases; and improving access to information for patients, the general public, and professionals. Amongst identified actions include the establishment of a national registry for rare diseases; establishment of national reference centres integrated with international networks; examining cross-border cooperation for genetic testing and other services; introducing a system of evidence-based clinical guidelines; defining orphan drug policy and developing decision guidelines for competent authorities; identifying additional funding sources for orphan drugs; establishing an umbrella organisation of patient groups; and establishing a national centre for rare diseases in the country.

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.

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.

Registries
Slovenia contributes to the EUROCARE CF European registry and RARECARE 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 62 genes and an estimated 73 diseases in the Orphanet database\(^2\).

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\(^2\) Information extracted from the Orphanet database (December 2012).
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 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.

Official information centre for rare diseases

There is no official information centre for rare diseases in Slovenia other than Orphanet at the moment. Establishment of national contact point for patients and professionals to get some extensive information on rare diseases diagnosis and treatment is foreseen. This will be the first action to be implemented in the Work Plan for Rare Diseases. There are on-going negotiations with the stakeholders.

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

The national clinical guidelines are available for the 4 inborn errors of metabolism which are treated with enzyme replacement therapy (Fabry disease, Pompe disease, MPS II and VI).

Training and education initiatives

No specific activity reported.

National rare disease events in 2012

On 29 February 2012 a press conference was organised by Ministry of Health as a way to raise awareness of rare diseases to mark Rare Disease Day 2012. The event was also the opportunity to inform the general public on rare diseases and to improve cooperation between patients and medical professions for a better quality of life. On the same day gathering of different stakeholders on the field of rare diseases (patient organisations, health care providers, medical doctors, ministry representatives, representatives of local community, press) was carried out by one of patient organisations.

Hosted rare disease events in 2012

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.

3 http://www.orpha.net/national/SI-SL/index/domov/
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, PARENT Joint Action, European Fabry Registry Advisory Board.

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

IRDiRC
Slovenian funding agencies have not yet committed funding to 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 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. In 2012 The Health Insurance Institute of Slovenia put on positive hospital list both orphan medicinal products: clofarabine (Evoltra) and busulfan (Busilvex) that were in 2011 covered by additional funding. The Strategic Council for Drugs in 2012 provided additional government budget funds of 794.537,00 € to finance two orphan medicinal products: eculizumab (Soliris) for 2 patients and idrusulfase (Elaprase) for 1 patient.

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)”.

Orphan medicinal product market availability situation
The orphan medicinal products launched on the market up to the end of 2012 were: Adcetris, Afintor, Arzerra, Busilvex, Cystadane, Diacomit, Elaprase, Evoltra, Exjade, Fabrazyme, Firazyr, Glivec, Kuvan, Litak, Lysodren, Mozobil, Myozyme, Naglazyme, Nexavar, Nplate, Pedea, Peyona, Replagal, Revatio, Revlimid, Revolade, Savene, Soliris, Somavert, Sprycel, Sutent, Tasigna, Tepadina, Thalidomide, Tobi Podhaler, Torisel, Tracleer, Ventavis, Vidaza, Volibris, Vpiv, Wilzin, Xyrem, Yondelis, 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.

All orphan medicinal products are covered mainly by compulsory health insurance and some partly by complementary health insurance, without the need for any co-payment by the patient.

The expenditure for orphan medicinal products increased by 44,9 % from 2010 to 2012, whereas total expenditure for other drugs was in 2012 lower in comparison to 2010 due to systematic price regulation.

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1 Inventory of Community and Member States' incentive measures to aid the research, marketing, development and availability of orphan medicinal products (2005 revision) (p19)
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.”

Other therapies for rare diseases
In Slovenia other therapies for rare diseases are also available, for example: implantation of subdermal pump for treatment of epilepsy, surgical corrections of rare inborn heart defects, cranial synostosis, inborn facial defects and transplantation of hematopoietic stem cells.

Orphan devices
No specific information reported.

Specialised social services
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 2012 IN SLOVENIA

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Centres of expertise
The establishment of centres of expertise is foreseen in the national plan for rare diseases.

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.

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5 Inventory of Community and Member States’ incentive measures to aid the research, marketing, development and availability of orphan medicinal products (2005 revision) (p19)
Sources of information on rare diseases and national help lines

**Official information centre for rare diseases**

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**Orphan medicinal products**

**Orphan medicinal product committee**

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**Orphan medicinal product reimbursement policy**

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**Other therapies for rare diseases**

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SELECTED BIBLIOGRAPHY AND SOURCES

- Work Plan for Rare Diseases in Slovenia (September 2011)
- Orphanet Slovenia national website
  http://www.orpha.net/national/Sl-SI/index/domov/

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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 2013.