

**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
THE SLOVAK REPUBLIC**

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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 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 the Slovak Republic. 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 report¹.

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

RARE DISEASE ACTIVITIES IN THE SLOVAK REPUBLIC

Definition of a rare disease

Stakeholders in Slovak Republic 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

No national plan or strategy for rare diseases currently exists in Slovak Republic and there is no specific budget currently dedicated to rare diseases. However, a working group was established in January 2011 at the Ministry of Health and work has begun on a "Strategy for improving health care for patients with rare diseases". This document is being reviewed at the Ministry of Health level. A workshop was also held in December 2011 concerning rare diseases at the Ministry of Health.

Centres of expertise

There is currently no official policy concerning centres of expertise for rare diseases and no official centres of expertise for rare diseases in Slovak Republic. Health care for several rare diseases is centralised mainly at the Departments of Clinical Genetics (12 in the country), the Centre for metabolic diseases (1 in the country), and in several metabolism or specialised outpatient clinics, as well as in cooperation with some research laboratories of Slovak Academy of Science. A small workshop was held in December 2011 concerning rare diseases at the Ministry of Health and this group is now working on the criteria for centres of expertise in line with those issued by the EUCERD. Several specialised and centralised departments would be appointed as centres of expertise in the near future (e.g. oncogenetics, hereditary metabolic diseases).

Pilot European Reference Networks

Teams from the Slovak Republic participate, or have participated, in the following European Reference Networks for rare diseases: Dyscerne.

Registries

At present, there is no national committee dedicated to dealing with registries and no national rare disease registry. National health registries are financed by Ministry of Health of the Slovak Republic. The National Health Information Center (NCZI) is the operator of most national health registries. Of these the following registries are for rare diseases are: the National registry for congenital disorders (established in 1976), the National registry for congenital heart defects (established in 1992), the National cancer and rare cancers registry (established in 1976), the National child diabetes mellitus and neonatal diabetes registry (established in 1986).

The NCZI provides data from the new registry focused at the monogenic forms of diabetes. The registry has been launched in 2008 based upon the data produced by the DIABGENE Laboratory at the Institute of Experimental Endocrinology, Slovak Academy of Sciences. In 2011 the National child hypertension registry was established as sub register of cardiovascular registry.

There are also several disease-specific registries managed outside of the NCZI (i.e. national registry of haemophilia).

The Slovak Republic contributes to the European Cystic Fibrosis Society Patient Registry,

Neonatal screening policy

Neonatal screening (NBS) policy has been officially established by the Ministry of Health in the Slovak Republic. Screening is in place since 1985 for congenital hypothyreosis (CH), phenylketonuria, congenital adrenal hyperplasia, and cystic fibrosis. Screening is provided in one central National Newborn Screening Centre, in coordination with three regional Recall Centres providing definitive diagnostic procedures and continuous management of confirmed cases. MS/MS technology has been introduced into selective screening. In addition to the screened diseases every newborn/infant is screened for hearing disorder, hip dislocation and the majority of newborns (more than 90%) are screened immediately after birth by means of USG for somatic malformations (CNS, cardiol, obstructive uropathy, etc.) although this is not an official governmental policy. The National Newborn Screening Centre is a member of EUNENBS (European Union Network of Experts on Newborn Screening).

Genetic testing

As a small country, the Slovak Republic does not have a large number of laboratories for genetic testing. Genetic testing is organised by the Departments of Clinical Genetics (12 in the country), specialised genetic outpatient clinics or specialised Departments of Clinical Oncogenetics (2 in the country). There are 3-5 bigger and several smaller DNA laboratories which perform molecular diagnostics for around 350 monogenic mendelian disorders. There are currently no reference laboratories in Slovak Republic. National guidelines for genetic testing have not yet been approved, but are being developed at national level. Slovak Republic participated in elaboration of several international "Best Practice Guidelines", e.g. "Molecular Genetic Diagnosis of Maturity – onset Diabetes of the Young". Specific provisions for the reimbursement of tests are not yet in place and genetic testing for non-medical reasons is paid for by the person requesting the test. Genetic testing also takes place abroad, mainly in the Czech Republic.

National alliances of patient organisations and patient representation

The Slovak Rare Diseases Alliance was established at their first constitutive meeting held in Bratislava in Slovakia on 12 December 2011 and is composed of 12 patient organisations out of the 17 related to rare diseases in the country.

There are no public funding schemes for patient organisations in Slovak Republic. Some patient organisations are members of the NR OZP SR (National Disability Council in Slovak Republic). A patient representative is present in the rare disease strategy working group.

Sources of information on rare diseases and national help lines

Orphanet activities in the Slovak Republic

In 2010, in the context of the Joint Action Orphanet Europe, the Ministry of Health designated the 2nd Department of Paediatrics of the University Children's Hospital Bratislava as the official Orphanet team for Slovak Republic. 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. The team launched in 2011 the Orphanet Slovakia national website².

Official information centre for rare diseases

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

Help line

There is currently no dedicated help line for rare diseases at the moment.

Other sources of information on rare diseases

Information sources on rare diseases are mostly run by non-governmental organisations with a few projects supported by the state and its municipalities.

Good practice guidelines

Best practice guidelines have been developed for cystic fibrosis, maturity-onset diabetes of the young, Wilson disease and haemophilia. Some molecular genetics laboratories in Slovakia have been participating in the EMQN programme and EQA KRAS programme. The Slovak Republic participated in elaboration of the international Best Practice Guidelines for Molecular Genetic Diagnosis of Maturity-onset Diabetes of the Young.

Training and education initiatives

Currently, there are no training or education initiatives organised systematically in the field of rare diseases.

National rare disease events 2011

To mark Rare Disease Day 2011 several events was organised, mainly through mass media presentations. On the website of Slovak Ministry of Health there was a short advertisement about Rare Disease Day. Short films were broadcasted on TV and wider discussions took place on the radio. Several rare diseases related presentations were organised in the context of local medical conferences and seminars.

Izakovic's Memorial is an annual conference organised in Slovak Republic by the Society of Medical Genetics, related to genetic and rare diseases. In 2011 the conference was specially focused on problem of rare diseases.

² <http://www.orpha.net/national/SK-SK/index/%C3%BAvod/>

Hosted rare disease events 2011

The DIABGENE Laboratory from Slovak Academy of Sciences organised from 30 September to 3 October 2011 the meeting "The Genetic of Diabetes in Post-Genome Wide Association Era" devoted to monogenic forms of diabetes and/or hereditary hyperinsulinism.

Research activities and E-Rare partnership

National research activities

Currently there are no specific programmes for rare disease research in Slovak Republic.

Participation in European research projects

Teams from the Slovak Republic participate, or have participated, in European rare disease research projects including: ANTEPRION and NM4TB.

E-Rare

Slovak Republic is not currently a partner of the E-Rare Project.

IRDiRC

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

Orphan medicinal products

Orphan medicinal product committee

The Slovak Republic does not have a national orphan medicinal product committee, and currently does not have a representative at the COMP. The Slovak Ministry of Health is responsible for pricing and reimbursement of all drugs as well as orphan medicinal products.

Orphan medicinal product incentives

To attract orphan medicinal products manufactures to launch the Slovak market better information about the patients with rare diseases is needed to be able to estimate the real need in this relatively small country. Formation of a special register for patients with rare diseases could solve this problem.

Orphan medicinal product market availability situation

SUKL, the State Institute for Drug Control, is the regulatory body in the Slovak Republic responsible for the regulation and surveillance of human medicinal products and medical devices, including orphan medicinal products. All orphan medicinal products registered at EU level are registered in the Slovak Republic and at the end of 2011 26 orphan medicinal products were on the Slovak market: Myozyme, Fabrazyme, Aldurazyme, Naglazyme, Zavesca, Kuvan, Ventavis, Revolade, Nplate, Firazyf, Volibris, Tracleer, Revatio, Somavert, Increlex, Litak, Glivec, Nexavar, Sprycel, Tyverb, Tasigna, Torisel, Lysodren, Vidaza, Inovelon, Exjade.

The reimbursement level is set in a national process named "categorisation".

Orphan medicinal product pricing policy

No specific information reported.

Orphan medicinal product reimbursement policy

At the end of the year 2011 Slovak Ministry of Health introduced a monthly update of "categorisation list", thus increasing the possibilities for all pharmaceutical companies as well as orphan medicinal product pharma companies to launch their products. The "categorisation list" of all reimbursed drugs is published every quarter.

At the end of 2011 26 orphan medicinal products (OMP) were on the Slovak market. Out of the 26 OMP, 7 OMP need patients' participation at their costs (Myozyme, Revolade, Firazyf, Volibris, Somavert, Litak, Inovelon). However some orphan medicinal products are at the market in different packages (example Myozyme 1x50mg, 10x50mg, 25x50mg), and one out of them is fully reimbursed. Indeed Myozyme, Somavert and Litak have a fully reimbursed alternative. The highest copayment was for Myozyme plc ifo 25x 50mg (€287.61). However if we express the copayment as percentage of the total price, the highest copayment was in Somavert plv iol 1x20mg - 36.78%. Somavert has other three alternatives (30x20mg, 30x10mg, 30x15mg) on the Slovak market, which are fully reimbursed. The orphan medicinal products are distributed mainly through pharmacies as well as on a centre basis, depending on the reimbursement category which is also set in the "categorisation list".

Other initiatives to improve access to orphan medicinal products

Only categorised drugs are directly available on the Slovak market. In the case of the drug not being 'categorised' the drug can be delivered on named-patient basis. Responsibility for approving delivery on a named-patient basis rests with the Ministry of Health according to Act 140/1998.

Several activities in 2011 stressed the importance of orphan medicinal product availability and to inform care givers, insurers as well as other health care professionals about situation in the Slovak Republic including a seminar in cooperation with State Institute for Drug Control and the Slovak Society for Pharmacoeconomics³ and a publication written in English on the topic⁴. Results of the use of OMP were presented also at 40th Symposium of European Society for Clinical Pharmacy in Dublin (18- 21 October 2011).

Orphan devices

No specific information reported.

Specialised social services

Care services, both government-run and private, are available in Slovak Republic and partial or full reimbursement is available (depending on certain criteria). Therapeutic programmes such as spa stays are available and paid mainly through private health insurance.

DEVELOPMENT OF RARE DISEASE ACTIVITIES IN 2011 IN THE SLOVAK REPUBLIC

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³ <http://www.sukl.sk/sk/o-nas/vzdelavacie-akcie-a-prezentacie/prezentacie-2010/dostupnost-orphan-liekov-v-sr>

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Sources of information on rare diseases and national help lines

Orphanet activities in the Slovak Republic

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⁵ <http://www.orpha.net/national/SK-SK/index/%C3%BAvod/>

LIST OF CONTRIBUTIONS⁶

Contributions in 2010

László Kovács and Ludovít Kádasi (*Orphanet Slovakia, Institute of Molecular Physiology and Genetics, Bratislava*)
Jana Behunova (*University Childrens' Hospital, Kosice*)

Contributions in 2011

Táňa Foltánová (*Department of Pharmacology and Toxicology, Comenius University, Bratislava*)
Katarína Štěpánková (*Slovak Marfan Association*)
František Cisarík (*EUCERD Representative Slovakia, Expert of the Ministry of Health for Medical Genetics*)

Contributions in 2012

František Cisarík (*EUCERD Representative Slovakia, Expert of the Ministry of Health for Medical Genetics*)
László Kovács and Anna Hlavatá (*Orphanet Slovakia*)
Lúdevít Kádaši (*EUCERD Alternat, President of the Slovak Society for Medical Genetics*)
Beáta Ramljakova (*Slovak Rare Disease Patient Alliance*)
Táňa Foltánová (*Department of Pharmacology and Toxicology, Comenius University, Bratislava, COMP Representative*)
Katarína Štěpánková (*Slovak Marfan Association*)
Michal Konečný (*OUSA*)
Iwar Klimeš (*Institute of Experimental Endocrinology, Slovak Academy of Sciences*)
Jana Behúňová (*University Childrens' Hospital, Košice*)
Svetozár Dluholucký (*Newborn Screening Centre*)
Anna Baráková (*National Health Information Centre*)

Validated by: František Cisarík (*EUCERD Representative Slovakia, Expert of the Ministry of Health for Medical Genetics*)

SELECTED BIBLIOGRAPHY AND SOURCES⁷

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<http://www.orpha.net/national/SK-SK/index/%C3%BAvod/>
- SUKL - State Institute for Drug Control
<http://www.sukl.sk/>

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>