2014 REPORT ON THE STATE OF THE ART OF RARE DISEASE ACTIVITIES IN EUROPE

STATE OF THE ART OF RARE DISEASE ACTIVITIES IN THE SLOVAK REPUBLIC

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The European Union Committee of Experts on Rare Diseases (EUCERD) was established in 2009 and its mandate ended in 2013. It is replaced from 2014 by the Commission Expert Group on Rare Diseases. The EUCERD Joint Action continues to support the activities of the new Expert Group until 2015.

More information on the activities of the former European Union Committee of Experts on Rare Diseases and the EUCERD Joint Action 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 European Union Committee of Experts on Rare Diseases (EUCERD) was established in 2009 and its mandate ended in 2013. It is replaced from 2014 by the Commission Expert Group on Rare Diseases. The EUCERD Joint Action continues to support the activities of the new Expert Group until 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 2013. 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 Commission Expert Group 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 2013
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

Parts I – V also include a description of the methodology, sources and validation process of the entire report, and 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 report.

Each year, there are around 15 000 downloads of the different sections of the report combined.
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
A working group was established in January 2011 at the Ministry of Health to work on a strategy for improving health care for patients with rare diseases. This strategy includes the basic concepts for the plan and was adopted by the Government of the Slovak Republic on 24 October 2012. However, no funding has been allocated for the Plan as of yet.

Centres of expertise
There is currently no official policy concerning centres of expertise for rare diseases: no official centres of expertise for rare diseases in Slovak Republic exist. Health care for several rare diseases is centralised mainly at the University hospitals (as for example Children’s University Hospitals in Bratislava or Banská Bystrica) or specialised hospitals (as for example National Institute for Cardiovascular diseases, National Institute for rheumatic diseases, National Cancer Institute), Departments of Clinical Genetics (12 in the country), the Centre for metabolic diseases (1 in the country), and in several specialised outpatient clinics for metabolism or few types of rare diseases, as well as in cooperation with some research laboratories of Slovak Academy of Science.

Registries
At present, there is no national committee dedicated to deal with registries and no national rare disease registry; however the future National Plan for Rare Diseases will look at ways of patient data collecting. 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: the National registry for congenital disorders (established in 2011), the National registry for congenital heart defects (established in 1992), the National cancer registry (established in 1976 including rare cancers), the National child diabetes mellitus and neonatal diabetes registry (established in 1986).

The NCZI provides data for 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 juvenile hypertension registry was established as sub register of cardiovascular registry.

During 2013 the Slovak Society of Medical Genetics together with the NCZI worked on reporting congenital anomalies (including ORPHA, OMIM, codes), so as to use this for future information about rare disease patients.

There are also several disease-specific registries managed outside of the NCZI (i.e. clinical registry of haemophilia). Slovak patients are registered also in international registries as REaDy – REgister of muscular dystrophies (http://ready.registry.cz/). Currently, the database registers a total of 51 patients from Slovakia.

Up November 2013 Slovakia is involved in the European Cystic Fibrosis Society Patients Registry.

The use of Orphacodes to code rare diseases is being considered.

The Slovak Republic contributes to the EUROCARE CF and RARECARE 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 hypothyroidism, 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 disorders, hip dislocation and the majority of newborns (more than 90%) are screened immediately after birth by means of USG for somatic malformations (CNS, cardiology, obstructive uropathy, etc.) although this is not an official governmental policy.

In 2012 a document on newborn screening was adopted, expanding the panel of screened diseases from 4 to 13, to include hyperphenylalaninemia (HPA), leucinosis (MSUD), Medium Chain Acyl Co A Dehydrogenase
Deficiency (MCAD), Long Chain Acyl Co A Dehydrogenase Deficiency (LCHAD), VLCAD, Carnitine Palmitoyl Transferase I. Deficiency (CPT I), Carnitine Palmitoyl Transferase II. Deficiency (CPT II), Carnitine Acylcarnitine Translocase Deficiency (CACT), glutaric aciduria type I (GAI), and isovaleric aciduria (IVA). In the year 2013 the expanded newborn screening was launched 4.

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 5 bigger and several smaller DNA laboratories which perform or provide molecular diagnostics for around 350 monogenic mendelian disorders. There are currently no reference laboratories in Slovak Republic.

The Slovak Republic has also 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. Slovak Society of Medical Genetics and health insurance companies are now developing new system of reimbursement of genetic testing. Genetic testing also takes place abroad, mainly in the Czech Republic.

Diagnostic tests are registered in the Orphanet database for 49 genes and an estimated 103 diseases 2.

National alliances of patient organisations and patient representation
The Slovak Rare Disease Alliance 1 (Slovak RD Alliance) was established at their first constitutive meeting held in Bratislava in Slovakia on 12 December 2011 and was composed of 12 patient organisations out of the 17 related to rare diseases in the country at that time. In 2013, 16 rare disease organisations plus one voluntary group of patients form the Slovak RD Alliance.

The Slovak RD Alliance leads the activities in the field of rare diseases in Slovakia and thus was the main organiser of the EUROPLAN National Conference on the Rare diseases day in February 2013. The fundamental challenge for the Slovak RD Alliance is to raise public awareness about rare diseases. The representatives of the Slovak RD Alliance are actively involved in the Working group for rare diseases at the Ministry of Health and in 2013 participated in the formation of National Plan for Rare Diseases. The RD Alliance also publishes a newsletter MINORIT (quarterly).

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).

Sources of information on rare diseases and national help lines
Orphanet activities in the Slovak Republic
Since 2006 there is a dedicated Orphanet team in the Slovak Republic, hosted before 2010 by the Institute of Molecular Physiology and Genetics in Bratislava. 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 engaged in 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 website 5. Part of the Orphanet Encyclopedia has also been translated to Slovak language and this initial set rare disease descriptions is now available on the Orphanet Slovakia website. In the year 2013 the Slovak Orphanet team was partner of the EUROPLAN National Conference held on the Rare Disease Day (2nd Slovak Rare Disease Conference 5). In April 2014 the Slovak Orphan team organised the 2nd Slovak Rare Disease Conference.

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

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1 Dluholucký S./Knapková M. Newborn Screening in Slovakia – From 1985 Till Today
2 Data extracted from the Orphanet database in January 2014.
4 http://www.sazch.sk/ www.zriedkave-choroby.sk
5 http://www.orpha.net/national/SK-SK/index%C3%A0Vod/ www.zriedkave-choroby.sk
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 patient’s organisations with a few projects supported by the state and municipality. One well developed information source is internet page of Slovak RD Alliance. More used sources of information for professionals in clinical genetics are the websites OMIM (Online Mendelian Inheritance in Man), GeneTests, and many other web sites for another medical specialities.

Guidelines
Good practice guidelines have been developed for cystic fibrosis, maturity-onset diabetes of the young, Wilson disease and haemophilia, as well as for new born screening. 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. National guidelines for genetic testing were developed and adopted by the Slovak Society of Medical Genetics (SSLG) in June 2012. SSLG, oncogenetics laboratories and Association of Health Insurance Companies developed guideline for diagnostics and clinical management of HBOC.

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

National rare disease events 2013
The main activity of the National Rare Disease Day 2013 in Slovakia was the EUROPLAN National Conference (27-28 February 2013)\(^6\). This second edition was organised by the Slovak RD Alliance in cooperation with the EUCERD representative and under auspices of the Chair of Health Care Committee National Council of the Slovak Republic and member of Parliament of the Slovak Republic - Richard Raší, MD., PhD, MPH., Ministry of Health of the Slovak Republic - Dr. Zuzana Zvolenská and EURORDIS.

The special website dedicated to Rare Disease Day was launched by Slovak RD Alliance (http://www.zriedkave-choroby.sk) which is regularly updated mainly with information and news regarding activities run by Slovak RD Alliance on Rare Disease Day.

On 27 of February there was a press conference, where the representative of Health Care Committee National Council of the Slovak Republic: Richard Raší MD, the representative of EURORDIS Dorica Dan, the representative of professionals Anna Hlavatá MD from University Children’s Hospital, the representative of EUCERD Slovakia Frantisek Cisarik MD and the press conference host from Slovak RD Alliance and DebrA SR Beata Ramjaková took part. After the press conference the speakers moved to the Ministry of Health, where together with others involved, the first meeting of the Working Group for Rare Diseases was held. The EUCERD representative from the Czech Republic Milan Macek gave a talk about the experience with the health care for rare disease patients in the Czech Republic. The representatives of the Slovak RD Alliance gave the official translation\(^7\) of the National strategy into English to the representatives of the Ministry of Health. On the next day the Forum of Experts with 164 guests and 16 talks was held. The Conference was divided into three main parts, according the guideline for EUROPLAN conferences. In the first one, the starting points for the creation and formation of the National plan were explained and an update on the legislative status given. Then an update on Orphanet activities was given and professionals were encouraged to register their services. The availability of orphan medicinal products from the regulatory point of view as well as from the point of view of the health insurance company was discussed. In the next part experience with the newborn screening policy was presented. The issue of registries for rare diseases was also tackled. Finally the Slovak RD Alliance presented its activities and importance of active participation in the creation of the National Plan. Representatives of the patient organisations presented their experience with the specialised social services provided. The national scientific journal Acta Facultatis Pharmaceuticae Universitatis Comenianae published the proceedings\(^8\). The conference report is available online\(^9\).

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\(^{1}\) http://www.eurordis.org/sites/default/files/flags/finalreport-slovakia.pdf

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On 24 April 2013, the 2nd Slovak Rare Disease Conference was held in Bratislava. One of the aims was to reflect on the creation of a national network of expert centres for rare diseases. A number of scientific lectures with emphasis on therapeutic approaches were also given.

Izakovic’s Memorial is an annual conference organised in Slovak Republic by the Slovak Society of Medical Genetics, related to genetic and rare diseases.

Hosted rare disease events 2013
No reported events.

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
There are currently no teams from Slovakia participating in FP7 rare disease related projects.

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

IRDiRC
Slovak funding agencies do not currently commit funding to the IRDiRC.

Orphan medicinal products

Orphan medicinal product committee
Also in the year 2013 the Slovak Republic did not have a national orphan medicinal product committee, and a representative at the COMP neither in EUnetHTA. The Slovak Ministry of Health is responsible for system of pricing and reimbursement of all drugs as well as orphan medicinal products.

Orphan medicinal product incentives
To attract orphan medicinal products manufacturers 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 national strategy/plan might help to 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. The data about the adverse events is submitted to the European database of suspected adverse drug reactions.

The reimbursement level is set in a national process named “categorisation”. The “categorisation committee”, established at the Ministry of Health, is responsible for pricing and reimbursement.

All orphan medicinal products registered at EU level are available in the Slovak Republic. However since 2011 changes in the national legislation make it difficult for OMP to be launched on the Slovak market. This is reflected in the fact that since this change only one drug launched the Slovak market (2012 - Tobi Podhaler). Thus OMP are available on individual basis via an administratively and time demanding process. First the physician has to submit a special application on a patient named basis at the Ministry of Health, afterwards individual negotiation between marketing authorisation holder and the Insurance company starts.

Directly available on the Slovak market at the end of 2013 were 22 orphan medicinal products: Myozyme, Aldurazyme (since June 2013 not OMP anymore), Naglazyme, Zavesca, Kuvan, Ventavis, Revolade (not OMP anymore), Nplate, Firazyr, Volibris, Tracleer, Revatio,), Increlex, Litak, Nexavar, Sprycel, Tasigna, Torisel, Revlimid, Lysodren, Vidaza, Inovelon, Exjade, Tobi podhaler. The information about the amount of OMP distributed at patient named-basis is not public (it is the subject of the individual contract between the marketing authorisation holder and the insurance company); data concerning this issue was presented at the EUROPLAN conference in February 2013. A systemic approach is lacking.

**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 to launch their product, however this is not the case of orphan medicinal product pharmaceutical companies, which have problems to meet the criteria for categorisation (Act No 36/2011 in the Collection of Laws of the Slovak Republic). The criteria for categorization are applicable only for high prevalent diseases.

At the end of 2013 out of the 22 orphan medicinal products, 6 orphan medicinal products require patients’ participation towards costs (Myozyme plc ifo 10x50 mg, plc ifo 25x50 mg, Tobi podhaler plv icd 224x28 mg, Firazyr sol inj 1x3ml, Inovelon tbl flm 50x200 mg, tbl flm 50x400 mg). The highest copayment was for Tobi podhaler plv icd (€497, which means 20.61% of the total price). The average copayment is 1.51% of the total price (0.87 – 20.61%). 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**
Thanks to the participation in the REaDy – REgister of muscular dystrophies, 2 patients with Duchenne muscular dystrophy were involved in the clinical trial. Because of unavailability of Centres of expertise, the treatment was given in Czech Republic, in Brno.

**Orphan devices**
No specific information reported.

**Other therapies for rare diseases**
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. However the definition of rare disease patient per se is not included in the indication list for such stays.
DEVELOPMENT OF RARE DISEASE ACTIVITIES IN 2013 IN THE SLOVAK REPUBLIC

Registries
During 2013 the Slovak Society of Medical Genetics together with the NCZI worked on reporting congenital anomalies (including ORPHA, OMIM, codes), so as to use this for future information about rare disease patients.

The use of Orphacodes to code rare diseases is being considered.

National alliances of patient organisations and patient representation
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National rare disease events 2013
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11 http://www.orpha.net/national/SK-SK/index/HC%3BAvod/
and formation of the National plan were explained and an update on the legislative status given. Then an update on Orphanet activities was given and professionals were encouraged to register their services. The availability of orphan medicinal products from the regulatory point of view as well as from the point of view of the health insurance company was discussed. In the next part experience with the newborn screening policy was presented. The issue of registries for rare diseases was also tackled. Finally the Slovak RD Alliance presented its activities and importance of active participation in the creation of the National Plan. Representatives of the patient organisations presented their experience with the specialised social services provided. The national scientific journal Acta Facultatis Pharmaceuticae Universitatis Comenianae published the proceedings\textsuperscript{14}. The conference report is available online\textsuperscript{15}.

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Izakovic’s Memorial is an annual conference organised in Slovak Republic by the Slovak Society of Medical Genetics, related to genetic and rare diseases.

\textsuperscript{14} http://www.degruyter.com/view/j/afpuc.2013.60.issue-supplementum8\textline{}issue-files/afpuc.2013.60.issue-supplementum8.xml
\textsuperscript{15} “\textcolor{blue}{http://www.eurordis.org/sites/default/files/flags/finalreport-slovakia.pdf}”
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16 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.
SELECTED BIBLIOGRAPHY AND SOURCES

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  http://www.sukl.sk/
- Slovak Alliance for Rare Diseases  
- National Strategy for Rare Diseases  
- Final Report of the 2013 Slovakian Europlan National Conference  

17 All websites and documents were last accessed in May 2014.