State of the Art of Rare Disease – Activities in EU Member States and Other European Countries

Slovak Republic Report

Definition of a Rare Disease

The Slovak Republic has adopted the European Commission definition of a rare diseases. However for the purposes of reimbursement the frequency mentioned is less than 1 in 10,000, however rare diseases are not mentioned.

National Plans and Strategies for Rare Diseases

A National Plan for rare disease patients in the Slovak Republic 2016 - 2020 was created by the multi-stakeholder advisory group, including patients, for rare diseases at the Ministry of Health of the Slovak Republic. It is based on the National Strategy for the Development of Health Care for Patients with Rare Diseases for the Years 2012-2013 Act number 578/2012 of the Slovak Government. The EUCERD criteria form the basis of both these papers.

A decision has been made to ensure long-term sustainability of the measures detailed within the national plan. The budget for 2016-2017 is €240,000 however the budget for the activities detailed within the national plan are incorporated into the general budget. An action plan for the NP for RD in the Slovak Republic 2016 - 2020 had defined 9 priorities, based upon the EUCERD criteria. Every year the fulfilment of the priorities will be evaluated and the need for further sources will be updated.

For now ICD10 is obligatory for health care professionals and providers. However for the purpose of registering of rare diseases Orphacodes were introduced.

The multi stakeholder group, the Commission at the Ministry of Health of the Slovak Republic for rare diseases has 15 members. It is composed of representatives of the Ministry of Health, a member of CEGRD, a representative of the new-born screening centre, representatives of health insurance companies, specialists in the RD, specifically from neurology and oncology, a representative of Pharmacists, National Health Information Centre, deputy of ORPHANET and a patient representative from the Slovak Alliance for Rare Diseases.

In line with the National Plan in 2014 the innovative concept of medical genetics in health care was approved. The Ministry of Health decided to modify the guidance documents regarding neonatal screening. In the Slovak Republic, by January 2013, the new-born screening was extended to 13 diseases. Currently the new-born screening programme tests for 23 diseases. These diseases were selected in accordance with the results of the pilot study conducted by the National Screening Centre. There is potential to merge the results from the study with the register of congenital anomalies. This might paint a more precise picture regarding prevalence and incidence of rare diseases in the Slovak Republic.

Organisation of Rare Disease Health and Social Care

Centres of Expertise

There is no policy in place for the designation of Centres of Expertise for rare disease within the Slovak Republic. The organisation of the health care in the Slovak Republic is stepwise, ending with university or faculty hospitals or specialised institutes, for example the Institute for Cardiovascular Diseases, Oncologic or Rheumatologic Diseases or other system specific institutions, the Endocrinology Institute

Co-funded by
the Health Programme
of the European Union

for instance. Patients with rare diseases or undiagnosed diseases are advised to visit these hospitals or institutes for help due to their advanced expertise and multidisciplinary care.

There are plans to adopt a formal national policy for establishing Centres of Expertise for rare disease, the process is currently ongoing. The criteria were approved this year and centres can apply for official designation. In 2017 it is hoped that the first national Centres of Expertise will be declared. The criteria were set according the EUCERD recommendation but they are not as strict. The process should be progressive, motivating Centres of Expertise to strive for excellence and fulfil as much of the criteria as possible.

European Reference Networks (ERNs)

There is currently no formal process in place for the endorsement of Health Care Providers to participate as members or coordinators of an ERN. Since CEs do not exist in the Slovak Republic, no centre has applied to be part of an ERN application. However there is high quality international cooperation in several rare disease areas, for example Alkaptonuria and Pulmonary Hypertension.

Rare Disease Registration

Neither national nor regional rare disease registries exist, but disease specific registries are available in the Slovak Republic.

National laws are currently in place, pertaining to health care professionals, neonatologists and genetics, which stipulate that tests which show inborn errors, genetic diseases and rare diseases need to be reported to the National Centre of Health Statistics and Informatics.

The disease specific registries available in the Slovak Republic are:

- Cystic fibrosis clinician led, sponsored by pts, part of the international one,
- Neuromuscular disease - clinicians led,
- Haemophilia - clinician led,
- Primary immunodeficiency's - clinician led,
- Pulmonary hypertension clinicians led,
- Hereditary metabolic diseases clinicians led,
- Epidermolysis bullosa patients led,
- Duchenne muscular dystrophy - clinicians led, part of international one,
- Pilot register of rare genetic diseases from 2014 is coordinated by the National Centre for health statistics and informatics.

Genetic Testing

The Committee of the Slovak Society of Medical Genetics adopted genetic testing principles derived from international recommendations. Information on facilities and laboratories of Medical genetics is on the website of the Slovak Society of Medical Genetics. http://www.sslg.sk/.

Reimbursement for genetic testing is not specifically regulated. If laboratories within the Slovak Republic cannot perform a particular test, testing abroad is available to patients.

There is no special policy in place governing Cross Border Genetic testing, which is provided by Central Laboratories. A request for a special examination or test is assessment on a case by case basis by health insurance companies.

Co-funded by
the Health Programme
of the European Union

Neonatal Screening

The Ministry of Health has decided to modify the guidance documents regarding neonatal screening and in the Slovak Republic, by January 2013, the neonatal screening programmes had been extended to include a total of 13 disease. Currently, the new-born screening programme offers tests for 23 diseases. This is obligatory to all children in the Slovak Republic.

The following diseases are tested for as part of the neonatal screening programme:

- Phenylketonuria,
- Hyperphenylalaninemia,
- leucinosis ,
- Tyrosinemia I., II.
- Hypermethiononemia,
- Citrullinemia,
- Argininemia,
- Isovaleric aciduria,
- Propionic aciduria,
- Methylmalonic aciduria,
- 3- Deficit Methylcrotonyl CoA carboxylase,
- Glutaric aciduria I,
- 3 - hydroxy 3 -
- Methyl glutaric aciduria,
- Dehydrogenase deficiency FA medium chain,
- Dehydrogenase deficiency FA long-chain ,
- Dehydrogenase deficiency FA with very long-chain,
- Dehydrogenase deficiency FA short chain,
- Primary carnitine deficiency.

<u>Guidelines and training activities</u>

Clinical Practice Guidelines (CPGs)

Guidelines have been produced to provide information on the standardisation of genetic diagnostics of hereditary breast and ovarian cancer, Lynch syndrome and KRAS. There are national policies in place for the development of CPGs, adoption of CPGs and the implementation of CPGs.

Training and Education

The Slovak Republic has organised the following activities with the purpose of educating and raising aware of rare diseases.

- Several conferences have been organised.
- Case reports on rare diseases have been written.
- The Annual Genetics conference - Izakovic memorial
- Rare Diseases have been included as part of the agenda of national conferences, for instance the Neurological Disorders conference and the Dietologists and Nutritionists conference.

Co-funded by
the Health Programme
of the European Union

Information Resources for Rare Diseases

Orphanet Activities

There is an operational National Orphanet team within the Slovak Republic which is hosted by the Faculty of Science within Comenius University in Bratislava. The institution is a partner of RD-ACTION and produces information in the national language of the Slovak Republic. The team have written translation abstracts for the Orphanet Encyclopaedia, presented updates on national rare disease conferences and produced articles which have appeared in national journals for professionals.

There is currently no dedicated funding in place to support the Orphanet team however they have been able to complete the following activities since the beginning of 2014:

- Once a year (11/2014, 11/2015) the rare disease conferences were organized. Within the conference the team presented updates on Orphanet were presented.
- The national team have updated the Orphanet database regarding registered professionals, expert centres, diagnostic laboratories, patient organizations, research activities and registries from the national area.
- Abstracts of the encyclopaedia of rare diseases were translated into Slovak.
- Articles about Orphanet and its services have been published in journals for professionals within the field of paediatrics.

National Helplines

There is currently no helpline in place dedicated to rare diseases in the Slovak Republic. Since there is no official helpline for rare disease patients, most of the questions are answered in charge of the patient umbrella organisation. In the future Slovak Alliance for rare diseases, the umbrella organisation might change this.

Official Information Centres

There are no official or unofficial information centres for rare disease in the Slovak Republic.

Rare Disease Research Activities

Existence of Rare Disease Research Programmes/Projects

The Slovak Republic has no programmes or projects which fund or facilitate rare disease research. Nor do they have any ongoing or future plans to conduct research into socio-economic issues relating to rare diseases. Research in the area of rare diseases is minimal, topics in clinical research appear sporadically.

Participation in E-Rare and International Research Initiatives

At present the Slovak Republic does not participate in either E-rare no IRDiRC.

National Alliance of Patient Organisations and Patient Representation

There is a national alliance for rare disease patient organisations in the Slovak Republic. Representatives of the Slovak Alliance for Rare Diseases form part of the Multistakeholder Commission for Rare Diseases at the Ministry of Health. They actively contributed to the final version of the National Plan for Rare Diseases and the criteria for the designation of Centres of Expertise.

Co-funded by
the Health Programme
of the European Union

Most of their activities is focussed on the Rare Disease Day. The Slovak Alliance for Rare Diseases organises and coordinates all activities on Rare Disease Day in the Slovak Republic. In 2014 the alliance organised a press conference, and a travelling exhibition on the topic of rare diseases, entitled 'Rare Diseases are less rare than you could think'.

In 2015 the Alliance organised a successful beneficial concert for the public, its aim was to increase the awareness of rare diseases amongst the general population. It was tailored to not only adults but also children and families.

In both 2014 and 2015 a national newspaper dedicated a specific section to rare diseases.

For the scientific community, a special edition of the Journal of the Pharmaceutical Faculty from the Comenius University in Bratislava was published. The Slovak Alliance for Rare Diseases is also producing a quarterly newsletter about rare diseases bringing news from the fellow organisations.

Integration of Rare Diseases into Social Policies and Services

Specific actions exist to enable real access for people with rare diseases to general social/disability programmes. Those individuals living with a rare disease are not given a specific category in existing programmes in the Slovak Republic. They are usually incorporated into a bigger group of disabled individuals. Many rare disease patients however find it difficult to obtain disability status due to a lack of understanding and information on rare diseases. Care plans do exist but these plans are not specialised for rare disease patients. There were plans to introduce incentives which would introduce a common view of health and social care of rare disease patients, but until now these plans have been unsuccessful. It is felt that through further cooperation with the Ministry of Labour and Social Affairs this situation will change and rare diseases will feature more heavily on the national agenda.

Orphan Medicinal Products (OMPs)

Only 15 OMPs are directly available with the Slovak Republic. Information regarding these products is publicly available through the Ministry of Health website. It forms part of the list of priced/reimbursed drugs in the Slovak Republic which is updated on a monthly basis. The MAH has to apply to the Ministry of Health to be reimbursed.

There are measures in place to facilitate access to Orphan Medicinal Products. One such measure is a compassionate use programme, which seem to be classes as more formal than practical due to their numerous administrative and legal obstacles.

Generally national drug research is very rare, thus no OMP have special support and incentives to encourage OMP oriented research.

Rare Disease Day

The following events were organised to celebrate Rare Disease day in 2014 and 2015:

In the year 2014 there was a press conference which discussed the exhibition 'Rare diseases - are less rare than you could think.' The press conference and exhibition was organised to highlight the lives of the approximately 300, 000 rare disease patients living in the Slovak Republic. Once the press conference was complete it was moved to the Slovak Parliament and during throughout 2014 it

Co-funded by
the Health Programme
of the European Union

travelled around the country stopping at the Ministry of Health, University hospitals and other public places

In 2015 the Slovak Alliance for Rare Diseases organized a successful beneficial concert for the public, to increase the awareness of rare diseases. The campaign introduced 'Lay rare', an exhibition which presented different public figures laying or laying on strange places. It was intended that the exhibition would raise awareness of rare diseases through the use of laying as a symbol of helplessness. Not only depicting what is felt by the patients but also their families. It is also a symbol of desperateness and desolation of rare diseases patients who cannot do everyday activities so easily. As a sign of solidarity the general public were asked to lay down too. For every minute of laying without any movement they could give one euro on the transparent account of the Slovak Alliance for Rare diseases. The people could also invite or ask their friends to take part in to the campaign. Although it was popular tool in increasing the awareness, the financial objective was not as successful.

The following conferences were organised during 2014 and 2015:

- 06/11/2015 - 4th Slovak Conference on Rare diseases - Case reports
- 07/11/2014 - Rare Disease - Case reports (3rd Conference)
- 01/010-03/10/2014 Izakovic Memorial - Conference of the Slovak Society of Clinical Genetics
- 30/09/-02/10/2015 Izakovic Memorial- Conference of the Slovak Society of Clinical Genetics.

Other

The Slovak Republic has had many rare disease related achievements since the beginning of 2014:

- Gaining the cooperation of the Ministry of Health
- Work on the concept of Centres of Expertise

The most successful achievement was the approval of the National Plan and its first budget for the current year, as well as gaining the active cooperation of the multistakeholder committee at the Ministry of Health, the expansion of the new-born screening, the development of the molecular diagnostics of hereditary diseases, the launch of the national registry of rare diseases, expanding the cooperation with Slovak Alliance for rare diseases and raising awareness about rare diseases within Slovak society as a whole.

Co-funded by
the Health Programme
of the European Union