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

Slovenia Report

Definition of a Rare Disease

Slovenia has adopted the European Commission definition of a rare disease (i.e. no more than 5 per 10,000 people, as per Regulation (EC) No 141/2000).

Status Quo of any National Plan or Strategy for Rare Diseases

Slovenia has adopted a National Plan for rare diseases which covers the years 2012-2020. The Ministry of Health has established a formal group of medical professionals, patients' societies, representatives of medical insurance companies and the Ministry of Health, which prepared the plan. The plan was confirmed and accepted by the highest medical consulting body, the Committee of Medical Healthcare by Ministry of Health.
There is no associated funding for the National Plan; however the Ministry of Health has launched 2 related projects with dedicated funding: the National Contact Point for Rare Diseases; and the Platform for the National Registry for Rare Diseases. The aforementioned formal group established to prepare the National Plan is tasked with the follow-up and oversight of the activities highlighted within the plan. Unfortunately the group only meets once per year and does not currently hold any powers to influence the actions within the plan.

The National Plan has had several notable successes since its creation:

- Increased awareness of rare diseases in Slovenia.
- The arrangement of annual, national conferences for the Rare Disease Day in February.
- The website for rare diseases, www.redkebolezni.si, was launched in August 2016.

The National Plan does not address the coding of rare diseases in health information systems.

Organisation of Rare Disease Health and Social Care (Centres of Expertise; ERNs; Integrated Care and Social Support)

Centres of Expertise

Currently there is no official policy in Slovenia for the designation of Centres of Expertise for rare disease. At present, University Medical Centres are recognised as Centres of Expertise but without a formal designation. There are no plans to adopt a more formal National Policy for establishing Centres of Expertise for rare disease in the future.

European Reference Networks (ERNs)

Slovenia has a formal process in place for the endorsement of Health Care Providers (HCPs) to participate as either coordinators or members of a European Reference Network. **Currently 3 HCPs are members of 9 different ERNs (**https://ec.europa.eu/health/ern_en**):**

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Institute of Oncology, Ljubljana	ERN EURACAN ERN GENTURIS
University Medical Centre, Ljubljana	ERN EURO-NMD MetabERN ERN RITA ERN ReCONNET ERN-RND
Univerzitetni klinični center Ljubljana	ERN PaedCan

However, the procedure for confirmation of the members is still ongoing.

Integration of Rare Diseases into Social Policies and Services

Specific programmes and facilities exist to support people with rare diseases in Slovenia. Activities including training, assistance for patients, and integration to employment and education, are part of the public health system for all patients who require the support of social services, and this includes rare disease patients.
Individual care plans include access to social and support services. There are specific measures in place to support the integration of rare disease specificities into the national system responsible for assessing a person's level of functioning: there is a list of disabilities and rare diseases which require specialist care and attention.

Rare Disease Registration

In Slovenia, regional-level registries are in place for rare diseases. These registries are governed by dedicated legislation, namely the '*Zakon o zbirkah podatkov s področja zdravstvenega varstva (Uradni list RS, št. 65/00 in 47/15*' or 'Law on Healthcare Databases Act' (http://www.pisrs.si/Pis.web/pregledPredpisa?id=ZAKO1419). Slovenian registries of relevance to rare diseases do not capture the OrphaCode.
The oldest relevant registry is the *Cancer Registry of Slovenia* (http://www.onko-i.si/eng/crs/), which is very well-organised: it has been operational for 50 years and has enrolled 90,000 patients (enrolment is mandatory for all cases). Also notable are the *National registry for Occupational Diseases and Infectious Diseases*, and the *Registry of Haemophilia*. All existing registries are active and well defined. Most registries do not receive funding from the state (with the exception of the Cancer registry and the *National registry for Occupational Diseases and Infectious Diseases*).

Genetic Testing and Neonatal Screening

Slovenia has ratified the Oviedo Convention and its Additional Protocol concerning **Genetic Testing** for Health Purposes. National legislation concerning genetic testing in Slovenia is currently being developed. All national laboratories are part of Orphanet database.
Genetic services are a part of the healthcare system and are funded the National health insurance agency. The Specialization of Clinical Genetics for medical doctors has been fully implemented, along with the Specialization in Laboratory medical genetics for others involved in genetic testing.

Genetic testing is only performed after Informed consent has been obtained, evidenced by the signature of the patient or the child's parent. All pre-symptomatic, predictive or prenatal genetic testing is performed with pre-

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and post-test genetic counselling. There are no specific procedures at present to facilitate cross-border genetic testing for rare diseases.

Neonatal screening is mandatory (as a public preventative healthcare measure) for two diseases at present: Phenylketonuria and Congenital Hypothyroidism.

Clinical Practice Guidelines and Training Activities

Clinical Practice Guidelines (CPGs)

In Slovenia, CPGs have been developed at the national level for Fabry Disease, Haemophilia and Cystic Fibrosis. Slovenia has in place a national policy for the *development* of CPGs but not for the adoption or implementation of CPGs.

Training and Education

The following resources and initiatives are in place for rare disease-related training and education in Slovenia:

- Basic information can be found at wwww.redkebolezni.si
- A professional symposium is held annually to coincide with Rare Disease Day
- Training and education related to rare diseases is incorporated to the national annual education for paediatricians.

Information Resources for Rare Diseases

Orphanet Activities

Slovenia has a national Orphanet team, currently hosted by the University Medical Centre Ljubljana (Clinical Institute of Medical Genetics). The institution has been a partner in RD-ACTION and the team produces information in the Slovenian national language. There is no dedicated funding in place to support the activities of the Orphanet team; however, the Slovenian team has been able to participate in the organisation of rare disease day activities and provides updated information regarding national activities in the field or rare diseases and genetics to the main Orphanet database.

National Helplines

In Slovenia there is a helpline in place which is dedicated to rare diseases: it is publicly funded and is available to both patients and professionals (i.e. it is open to anyone). The helpline is available through the website www.redkebolezni.si and users can seek rare disease-related assistance and information by email or by telephone (the telephone option is available for three hours per day, Monday to Friday).

Official Information Centres

Slovenia has an information centre for rare diseases (www.redkebolezni.si) which forms part of the project for rare diseases initiated by the Ministry of Health; however, the financial support for this centre is only available until the end of 2016 (negotiations for the prolongation of the project are underway).

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Rare Disease Research Activities

Slovenia has a specific research programme for rare diseases - *V3-1505 (C) – Analysis and development in the field of Rare Diseases in Slovenia*. At present Slovenia participates in neither E-RARE nor IRDiRC.

National Alliance of Patient Organisations and Patient Representation

Slovenia has a national alliance of rare disease patient organisations, established in May 2015 (Združenja za redke bolezni Slovenije). It has played a very active role in the organisation of activities for Rare Disease Day in Slovenia.

The medical care of rare disease patients is centralised at UMC Ljubljana. Patient groups are participating at the level of UMC and the Ministry of Health.

Orphan Medicinal Products (OMPs)

As of October 2016, 39 OMPs are available in Slovenia (out of 89 currently approved in the EU); this figure represents distinct product lines, not specific presentations (packs). Broad measures are in place **to facilitate access to Orphan Medicinal Products for patients**, including the following:

- specific pricing instruments;
- regulated list prices on two levels: ceiling price and optional premium exceptional higher ceiling price;
- possibility of negotiated lower pricing;
- possibility of reduced or waiver fees for regulatory procedures, including pricing procedures;
- a reimbursement committee;
- restricted prescribing measures;
- specific hospital medicines reimbursement list with additional monitoring
- specific evaluations by consultative bodies, reporting to the minister
- a health council and a strategic council on medicines

There are **incentives to support research into, and the development of (designated or potential) OMPs**:
these include pricing procedures and regulatory fee reduction or waiver depending on the sales value.
Tax incentives include a flat tax of 9% VAT placed upon all medicinal products, with no exceptions. Specific research programmes receive incentives and, in addition, the 'special hospital medicines reimbursement list' is in place to ensure the monitoring and transparency of the usage of OMPs, and of other high-cost hospital medicines. Scientific advice procedures are in place for all regulatory procedures. 'Out of specification' solutions are possible for foreign pack usage.

Slovenia reported several ***planned*** measures, for future development.

- To support research in future, a Health Technology Assessment process (HTA) is being formed through participation in the third Joint Action for HTA).
- To support the development of OMPs, Slovenia is planning enhanced monitoring of OMP 'penetration and consumption' by gathering on-line data from the health system.

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- To support the availability of OMPs to patients, regulatory solutions are necessary: obligations need to be extended to marketing authorization holders, more specifically than at present (i.e. under Dir 2001/83 Article 81). Slovenia is also putting in place a system for coordinated joint procurement of medicines for all hospitals in the country, which will have relevance for OMPs

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Rare Disease Day

Rare Disease Day events were organised in February 2015 and 2016. Further information can be found on the following website: http://www.drustvo-bkb.si/strokovna-srecanja/dnevi-redkih-bolezni/

Main achievements since 2014:

Slovenia reports several key rare disease achievements in recent years, including:

- The project for the establishment of a National Contact Point for Rare Diseases (available at www.redkebolezni.si)
- The project for the establishment of a National Registry of Rare Diseases is ongoing.
- The active participation of Slovenia in nine different ERNs.

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