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

Czech Republic Report

Definition of a Rare disease

The Czech Republic has adopted the European Commission definition of a rare disease, the National Plan espouses this definition.

National Plan/Strategy

Adoption

The Czech Republic adopted a National Plan for Rare Diseases, the plan has evolved over the last few years and is split into different forms; the National Strategy for Rare Diseases for 2010 – 2020, the National Action Plan for Rare Diseases 2012 -2014 to the National Action Plan for Rare Diseases 2015-2017. Currently, the third National Action plan is under preparation.

Content/Funding of the National Plan

There is no associated ("earmarked") funding to support the actions of the National plan, but it is possible to apply for a targeted appropriation / grant within the annual grant programme of the Czech Ministry of Health that supports projects that enable implementation of various national plans.

The National Plan addresses the topic of the coding of rare disease. The Institute of Health Information and Statistics within the Czech Ministry of Health is in the process of incorporating the RD coding into Czech health information systems and finished Czech translation of all Orpha.net terms. This will enable the use of Orphacodes within the Czech health care system monitoring of rare diseases.

Implementation and Evaluation of the National Plan

There is no dedicated advisory body to oversee the drafting or implementation of the National Plan or evaluate its impact. After the approval of the National Strategy for Rare Disease in 2010 an Interdepartmental and interdisciplinary working group (hence Rare Disease Taskforce) was established at the Ministry of Health. This group has been biannually convening since 2012 and monitors implementation of the National plan, prepares centralisation of care and serves as an advisory body for the Minister of Health. The working group was made up of representatives from all expert fields and the Ministry of Labour and Social Affairs. The patient representatives were part of this group from its inception. Once the umbrella patient organisation, the Czech Association for Rare Diseases (CAVO) was founded in 2012, it also became a member of the working group, and later became a member of Eurordis. Rare disease activities have also been fostered by the activities of the National Coordination Centre for Rare diseases that was established in 2012 at Motol University Hospital in Prague.

Major Successes

The National Plan has 11 main activities, these are:

- **Increasing awareness** – within the last 3 years this has been primarily implemented by the Czech Association for Rare Diseases (CAVO), the Ministry of Health has tried to help this task through the provision of a grant.
- **Education** – primarily implemented by CAVO and its expert advisors, now involving several medical professional associations, medical faculties and general paediatricians association.
- **Prevention and prenatal screening** – there has been an improvement of screening and diagnostics. 5 new metabolic diseases have been added to the list of screened diseases (2016) in the neonatal screening programme bringing the total amount of diseases to 18 rare disorders.
- **Improvement of access and quality of care** - a pilot project was launched in 2012 in which Centres of Excellence were established for cystic fibrosis, metabolic diseases and epidermolysis bullosa. These centres expired in 2015 and since then no further centres have been officially established. However

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many other expert representatives have reported to the Ministerial Working Group that they are ready to create centres of expertise for rare diseases according to Art. 112 of Act 372/2011 Coll. by fulfilling EUCERD criteria.

- **Improvements of quality of life and social inclusion** – no improvements to report.
- **Support of elementary and applied research** – There has been a unification and development of data collection and biological samples. UZIS, the Institute of Health Information and Statistics, is in the process of adopting Orphacodes into Czech Republic health care system. In addition, Agency for Medical research is implementing specific rare disease-related research calls. One of the major successes of 2017 is official enlistment of the Czech Republic within E-rare joint calls.
- **Support and strengthening of patient organisations** – the patient representatives form part of the Working Group board and there are opportunities for the representatives to receive financial support in the form of grants from the Ministry of Health.
- **Interdepartmental and interdisciplinary cooperation** – no significant achievements, there is a strong need for the Ministry of Health to collaborate with the Ministry of Labour and Social Affairs mainly in the area of data collection.
- **International cooperation** – the rare disease experts are cooperating on an international level, recently the Czech Republic began the process of becoming part of the European Reference Networks (ERNs). There are 27 health care providers taking part in 17 out of 24 ERNs, which is the best outcome within EU New Member States (EU13) group.

Organisation of RD Health and Social Care

European Reference Networks (ERNs)

There is a formal process in place within the Czech Republic for the endorsement of Health Care Providers (CEs) to participate as members or coordinators of an ERN in collaboration of the Ministry of Health with the National Coordination Centre for Rare Diseases.

At present a total of 8 HCPs participate as full members of 17 ERNs.

Rare Disease Registration

Although there are no official registries for rare disease in the Czech Republic, various research groups (mostly involved in various ERNs) have their own internal research databases. These, however, are not interlinked.

Genetic Testing and Neonatal Screening

Neonatal Screening

Newborn screening is performed in compliance with the valid legislation in the Czech Republic: Act No. 372/2011 Coll. And No. 373/2011 Coll. as amended.

Five new metabolic disorders were added to the screening panel in June 2016 bringing the total number of diseases screened to 18. The diseases screened are as follows:

- Endocrine Disorders
- Congenital hypothyroidism
- Congenital adrenal hyperplasia
- Disturbances of amino acid metabolism
- Argininemia
- Citrullinemia
- Phenylketonuria
- Glutaric aciduria, Type I (glutaryl-CoA dehydrogenase deficiency)
- Homocystinuria from CBS deficiency, pyridoxine non-responsive
- Homocystinuria from MTHFR deficiency
- Isovaleryl-CoA dehydrogenase deficiency (Isovaleric acidemia)

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- Maple syrup urine disease
- Disorders of fatty acid oxidation
- Carnitine uptake/transporter defects
- Carnitine-acylcarnitine translocase deficiency
- Carnitine palmitoyl transferase I deficiency (CPT I)
- Carnitine palmitoyl transferase II deficiency (CPT II)
- Very long chain acyl-CoA dehydrogenase deficiency (VLCADD)
- Long chain L-3 hydroxyacyl-CoA dehydrogenase deficiency (LCHADD)
- Medium chain acyl-CoA dehydrogenase deficiency (MCADD)
- Biotinidase deficiency
- Cystic fibrosis

Guidelines and training activities

Training and Education

Training on rare diseases is provided by the National Coordinating Centre for Rare Diseases in cooperation with CAVO (Czech Association for Rare Diseases). Together they:

- Provide third year medical students with information about rare diseases with participation of patient representatives who are invited to tell their stories.
- Provide postgraduate training to general practitioners on rare disease topics
- Coordinate a project which seeks to discuss early diagnostics for the general public and medical experts. This is provided with cooperation from the Expert Society of General Practitioners for Children and Adolescents.
- Provide a new project which is aimed at providing information on rare diseases to secondary medical schools using Whiteboard Illustration.

Information Resources for RDs

Orphanet Activities

The Czech Republic does have an operational Orphanet team who produces information in the national language since 2006.

National Helplines

There is a helpline in the Czech Republic dedicated to rare diseases which is privately funded and only available to professionals within the healthcare field. CAVO, together with its expert advisors from the National Coordination Centre, have been operating the helpline since 2013 (help@vzacna-onemocneni.cz). This is a voluntary activity, which the Ministry of Health does not financially support.

Official Information Centres for Rare Disease

There are both official and unofficial information centres for rare disease within the Czech Republic.

Rare Disease Research Activities

Participation in E-Rare and International Research Initiatives

The Czech Republic participates within IRDiRC, Professor Milan Maceck is a member of the DSC of IRDiRC.

National Alliance of Patient Organisations and Patient Representation

The Czech Republic has a national alliance of rare disease patient's organisations. CAVO, the Czech Association for Rare Diseases was established in 2012 and currently has 29 members-patient organisations, there are also 50 individual members representing various ultra-diseases. CAVO became a member of the ministerial working

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group and has successfully been implementing the National Plan for Rare Diseases with the help of its expert advisors.

The rare disease patient advocacy has recently started to take a very progressive path and is improving annually. Patient representatives are experienced and well educated in their field.

Integration of Rare Diseases to social policies and services

Specific programmes exist within the Czech Republic to support those with rare disease, these programmes are primarily offered by patient organisations. However there are no specific programmes for people with rare disease, they may apply for financial support and help from social services. Unfortunately however it is not uncommon, due to the rarity of their diagnosis, for people with rare disease to not receive the support and services they deserve.

It is believed that the Czech Ministry of Labour and Social Affairs will resume participation in the InnovCare project which could bring better social care to rare disease patients.

Rare Disease Day

Since 2014 CAVO, together with rare disease experts from the National Coordination Centre for Rare Diseases, has organised a seminar in the Czech Parliament on Rare Disease Day to highlight the importance of the implementation of the National Plan for Rare Diseases. These seminars are fully supported by several parliamentary deputies.

In 2015 the country celebrated 150 years of genetics in Brno, the birthplace of Gregor Mendel, the founder of genetics.

Other

The Czech Republic has had many rare disease related achievements since 2014:

- Approval of the second version of the National Action Plan for Rare Diseases (2015-2017)
- The terms “rare disease” and “orphan drug” have been integrated into the Czech legislation system.
- There has been an increase in awareness of rare diseases within the Czech Republic among the general public and healthcare experts.
- There has been a recognition of a strong rare disease patient platform.
- One of the major successes was introduction of the term “rare diseases and orphan medicinal product” into amended Act 48/1997 on public health insurance in 2015. Art. 11 point 1 f) of this law stipulates (in unofficial translation) “that health care reimbursement according to the provisions of this law also applies to diseases with very low prevalence in the general population (henceforward termed “rare diseases”) in accordance with the directly applicable Art. 3 of Directive 141/2000 on orphan medicinal products and to the related utilisation of orphan medicinal products in line with general provisions of this law”. This law established the “right” for reimbursement of care in rare diseases in the country.

Information on Centres of Expertise, Genetic Testing, Clinical Practice Guidelines (CPGs), and the existence of RD research programmes/projects and Orphan Medicinal Products (OMPs) will be included within the next update.

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