Supporting patients with rare diseases in the Czech Republic

The Czech Republic’s National Strategy for Rare Diseases 2010-2020 is complemented by EU-wide efforts to improve diagnosis and care for patients throughout Europe.

A new dawn

In 2010, the Czech Republic published its first 10-year strategy for rare diseases, prioritising diagnosis, treatment and access to high-quality care. These goals are practically elaborated upon in the first three-year National Action Plan 2012-2014 devised by a dedicated inter-ministerial and interdisciplinary taskforce which meets regularly. It has established a pilot group of Centres of Expertise, for example for cystic fibrosis, epidermolysis bullosa and metabolic disorders, which will raise standards across the country.

Working with Europe

The Czech Republic has actively engaged with rare diseases projects funded under the second EU health programme 2008-2013. Czech experts contribute to European registries, such as EUROCAT for rare birth defects, ECFS.eu for cystic fibrosis, TREAT-NMD for muscular dystrophies, E-HOD for homocystinuria, E-IMD for intoxication types of inborn errors of metabolism, INPDR for Niemann Pick disease and the European Porphyria Network.

Genetic tests are registered for 223 genes and 285 of the diseases listed in the Orphanet database — an EU project in which the Czech Republic is a partner.

Care can be provided, where appropriate, for Czech patients with very rare diseases in European Reference Networks, in line with the EU Directive on cross-border healthcare which was transposed into Czech national legislation in spring 2014.
Pooling knowledge, harmonising standards

**Project name:** European network and registry for homocystinurias and methylation defects (E-HOD)

**Number of partners:** 13 from 11 countries: CZ, DE, IE, ES, FR, IT, LV, HU, NL, SI, UK. **EU funding:** €690 793. **Duration:** 2012-2014.

The goal of the project is to improve access to rapid diagnosis and care for patients with homocystinurias (HCU) and methylation defects (MD) through the compilation of clinical data into a registry. The group, of which Charles University Prague, 1st Faculty of Medicine, is a member, is seeking to develop diagnosis and clinical care recommendations and evaluate new-born screening programmes in seven countries, including the Czech Republic. It will use the data from the registry to develop agreed protocols for HCU and MD. A public website will be produced to share information with communities of health professionals, researchers and patients.

Visit: www.e-hod.org

Enhancing access to specialist care

**Project name:** Dissemination and Implementation of the Standards of Care for Duchenne Muscular Dystrophy in Europe (including Eastern Countries) (CARE-NMD)

**Number of partners:** 7 from 7 countries: BG, CZ, DK, DE, HU, PL, UK. **EU funding:** €905 080. **Duration:** 2010-2013.

By examining how Duchenne muscular dystrophy (DMD) is managed in seven countries, including the Czech Republic, CARE-NMD aimed to improve standards of care. By increasing the number of reference centres for DMD, the project has improved access to specialist treatment, thus reducing inequalities and improving quality of life and life expectancy for patients. Fakultní nemocnice from Brno was a member of the consortium.

Visit: en.care-nmd.eu

Sharing data, caring for children

**Project name:** Single Hub and Access point for paediatric Rheumatology in Europe (SHARE)

**Number of partners:** 11 from 11 countries: BE, CZ, DE, ES, FR, IT, LV, HU, NL, SI, UK. **EU funding:** €860 244. **Duration:** 2013-2016.

The project aims to improve the quality of care for children suffering from rare rheumatology diseases. It is: strengthening networks of research teams and others involved in the development of technologies, medicines and patient care; improving patient access to relevant data about their disease; facilitating the exchange of ideas, information and best practices; and applying the results to the training of health professionals in the Czech Republic.

The project builds on the work of existing networks, national registers and European projects, and will support informed regulatory and policy decisions in this field. Charles University Prague, 1st Faculty of Medicine, participates in this project.

Visit: www.ped-rheum.com/content/11/1/5

Find out more

- Ministry of Health
  - [www.mzcr.cz](http://www.mzcr.cz)
- Czech National Strategy for Rare Diseases
  - [www.vzacenemoci.cz](http://www.vzacenemoci.cz)
- Czech National Alliance for Rare Diseases
  - [www.vzacna-onemocneni.cz](http://www.vzacna-onemocneni.cz)
- EUCERD website for Czech Republic
  - [www.eucerd.eu/?page_id=530](http://www.eucerd.eu/?page_id=530)

Directorate-General for Health and Consumers of the European Commission
- Consumers, Health and Food Executive Agency (Chafea)

Database of actions co-funded under the EU health programmes