



# Regional conference on Rare Diseases

Organized by CHAFEA in collaboration with the Czech Ministry of Health and the active contribution by the Czech National Coordination Centre for Rare Diseases – Department of Biology and Medical Genetics

02-03 December 2014 Prague, Czech Republic



MINISTRY OF HEALTH  
OF THE CZECH REPUBLIC



## Background

The Consumers Health and Food Executive Agency – Chafea (formerly the Executive Agency for Health and Consumers – EAHC) – was created on 1 January 2005. The Chafea mandate includes the implementation of the third EU Health Programme and the dissemination of the results from the second Health Programme 2008-2013.

In accordance with the Annual Work Plan 2013 priority 4.3.2 Communication, promotion and dissemination of information on EU health policies and the results of the Health programmes, Chafea plans a series of activities to widely disseminate results of the second EU Health Programme at EU, national, regional as well as local level.

As part of the dissemination strategy, Chafea collaborates with the National Focal Points (NFPs) to organize events at national level. On the 10 April, the prioritization of the dissemination activities was done during the NFP meeting. This resulted in the planning of the dissemination activities for 2014-2015, including the organization of five regional workshops/conferences, the production of 12 information sheets and the dissemination of the 2<sup>nd</sup> Health programme results in several Public Health events.

In the last NFP meeting, the Central European countries (Austria, the Czech Republic, Hungary, Poland and the Slovak Republic) representatives have selected a workshop in the area of rare diseases (RD) as the topic with higher public health relevance. The Czech NFP representing the Ministry of Health of the Czech Republic proposed to host the event in the autumn 2014, in collaboration with Chafea.

# Objectives

The main objective of the **Regional conference on rare diseases** is to present the results from several actions co-funded under the 2<sup>nd</sup> Health Programme from the field of rare diseases and foster the exchange of knowledge and best practices between European experts and countries. The conference should not only have an informative purpose but also provide a platform for discussion aimed to identify real solutions.

The target audience are experts on rare diseases, health professionals, policy makers and health managers from the EU MS, mainly the Czech Republic, the Slovak Republic, Austria, Hungary and Poland. At least 100 participants are expected.

Five actions coordinators and rare diseases specialists from 10 countries are invited to present their main results and share experiences. They were grouped in 2 plenary sessions and 5 workshop covering the areas of the European reference networks, the role of the patients organizations, the RD diagnosis, treatment and management, RD coding.

Another important aspect to be discussed is the sustainability of achievements beyond the EC funding, and how the actions' knowledge, deliverables (database, information tools, training programmes,) can become permanent, available to support the MS policies and improve their capacities on rare diseases management.

## Day 1 – December 2, 2014

**08:30 – 09:00** Registration

**09:00 – 10:00** Welcome and key note speeches (Chair: **Czech MoH**)

- Deputy Minister of Health of the Czech Republic (TBA)
- **Jaroslav Waligora** – DG SANCO, Health and Consumers, European Commission (EU policy on rare diseases)
- **Hristina Mileva** – Consumers, Health and Food Executive Agency, European Commission (Overview of the rare diseases actions funded under the Second EU Health Programme)
- **Jan Bodnár** – Department of EU funds, Ministry of Health of the Czech Republic

**10:00 – 10:30** Coffee break

**10:30 – 12:00** Special guest presentation (1) (Chair: **Milan Macek**)

- European Reference Networks: relevance for rare diseases – **Till Voigtlaender** (Vienna)
- EUCERD Joint Action – **Stephen Lynn** (Newcastle)
- Orphanet Joint Action – **Ana Rath** (Paris)

**12:00 – 13:30** Lunch

**Press event** – a meeting of journalists with key note speakers and project coordinators (12:00 – 12:30)

**13:30 – 15:00** Special guest presentation (2) (Chair: **Jaroslav Waligora**)

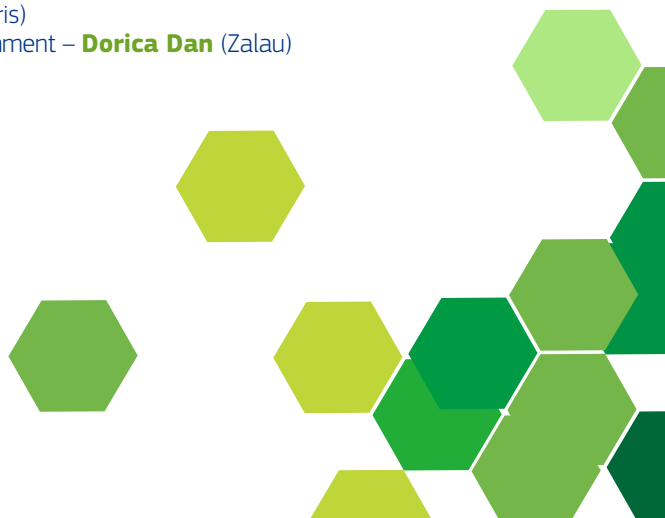
- Care-NMD / Treat-NMD – **Jana Haberlová** (Prague)
- SHARE Joint Action – **Pavla Doležalová** (Prague)
- E-HOD Joint Action – **Viktor Kožich** (Prague)

**15:00 – 15:30** Coffee break

**15:30 – 17:30** Cross-sectional topics (Chair: **Milan Macek**)

- Coding of rare diseases – **Segolene Ayme** (Paris)
- Crossborder genetic testing – **Helena Kääriäinen** (Helsinki)
- EURORDIS: Role of patient organisations – **Matt Johnson** (Paris)
- Development of a rare disease centre in a low resource environment – **Dorica Dan** (Zalau)

**17:30 – 18:00** Day I Conclusions



## Day 2 – December 3, 2014

**08:30 – 10:00** **Workshop session no. 1:** Short overview of Central European rare disease activities: key lessons learned (Chair: **Judit Molnar**)

- Czech Republic – **Milan Macek** (Prague)
- Hungary – **Judit Molnar** (Budapest)
- Poland – **Jolanta Sykut-Cegielska** (Warsaw)
- Slovakia – **František Cisárik** (Žilina)

**10:00 – 10:30** **Coffee break**

**10:30 – 12:00** **Workshop session no. 2:** Role of patient support groups (Chair: **Matt Johnson**)

- Czech Rare Disease Association – **René Břečťan** (Prague)
- HUFERDIS – **Gabor Pogány** (Budapest)
- Slovak Rare Disease Association – **Radoslav Herda** (Bratislava)
- Polish Rare Disease Association – **Mirosław Zielenski** (Warsaw)

**12:00 – 13:00** **Lunch**

**13:00 – 14:30** **Workshop session no. 3:** Patient access to orphan medicinal products (Chair: **Tatiana Foltánová**)

- COMP EMA Activities – **Kateřina Kubáčková** (Praha)
- Health Technology Assessment – right and wrong use of that tool to assess treatment of ultrarare diseases – **Krzysztof Landa** (Warsaw)
- Bulgarian expert centers and reference networks – **Rumen Stefanov** (Plovdiv)
- Orphan medicinal products regulatory aspects – **Tatiana Foltánová** (Bratislava)

**14:30 – 15:00** **Coffee break**

**15:00 – 16:30** **Workshop session no. 4:** Improving health care services – opportunities and challenges (Chair: **Segolene Ayme**)

- Epidermolysis bullosa centre care – **Helmut Hintner** (Salzburg)
- Rare disease teaching and case management: ontological approaches to inborn errors of metabolism – **Johannes Zschocke** (Innsbruck)
- Cystic fibrosis: a Slovak example – **Katarina Štěpánková** (Košice)

**16:30 – 17:00** **Refreshments**

**17:00 – 18:00** **Workshop session no. 5:** Access to diagnostic services (Chair: **Helena Kääriäinen**)

- State of the art of rare diseases activities in Serbia – **Dragica Radojkovic** (Belgrade)
- State of the art of rare diseases in Croatia – **Anja Kladar** (Zagreb)
- Clinical exome sequencing for diagnostics of rare Mendelian disorders: Slovene experience – **Borut Peterlin** (Ljubljana)
- DNA diagnostics: example of Romania – **Cristina Rusu** (Iasi)

**18:00 – 18:30** **Conclusions of Day 2 and Summary of the meeting**

