European Reference Network
for rare or low prevalence complex diseases

# The perspective of the Member State on the value of the CPG and knowledge generated by the ERNs, how should be organized/addressed at national level

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# Conflict of Interest

*No relevant conflict of interest in relation to this presentation*

# Clinical Practices Guideline (CPG) and Clinical Decision Support Tool (CDST) – *special concerns for rare diseases (RD) I*

- ***Classification of rare diseases*** – beyond ICD10 ⟹ ICD11 (**orpha codes – essential for reliable registries**)

- The **widest approach** must be used to a disease/syndrome
  - ICD10 codes, MIM codes, genes, age of onset, mode of inheritance, class of prevalence etc

- „***Traditional" subdisciplines/societies*** – e.g. endocrinology, metabolism, immunology, gastroenterology etc– not always, or ***exclusively*** fitting

- The **burden of informations** (e.g. ECFS revised, 2018 guideline: 25 pages, SMA: 65 pages) ... since patients usually have more than one ***exclusive disorder*** - ***if guidelines were strictly followed for each***

***≳1000 pages/patient...***

# CPG, CDST – *special concerns for RD II*

- **”Classical” evidence based** approach not always available
- Not to forget, however: guidelines ≠ fixed protocols, i.e. not to follow but to **consider** *(the physicians’ personal knowledge, decision)*!
  - One might even choose another solution but MUST
    - a) be familiar with the guideline
    - b) written explanation why not (exactly) followed the guideline

- The 8 Standards *(www.nationalacademies.org)* set up for guidelines cannot be completely fullfilled, e.g.
  - **S1** ”only specialists with no COI” **– i*mpossible in the field of RD,***
  - **S2** ”authorship ef external reviews should be kept confidential” - ***highly improbable***

Rare Best Practices

International Course

**Health care guidelines on rare diseases: Quality assessment**

February 23-24, 2015

Rare Best Practices

International Course

**Course for health care guidelines developers on treatments of rare diseases**

February 10-12, 2016

Istituto Superiore di Sanità

Rare Best Practices

2nd International Course

**Health care guidelines on rare diseases: Quality assessment**

December 3-4, 2015

Organized by the
National Centre for Rare Diseases
Istituto Superiore di Sanità
Rome (Italy)

CNMR

*This RARE-Bestpractices course has received funding from the European Union's Seventh Framework Programme (FP7/2007-2013) under grant agreement n° 305690. Sole responsibility lies with the authors and the European Commission is not responsible for any use that may be made of the information contained therein.*

Rare Best Practices

International Course

**Course for health care guidelines developers on diagnosis of rare diseases**

June 6-7, 2016

Organized by
Associazione per la ricerca sull'efficacia dell'assistenza sanitaria – Centro Cochrane Italiano (AREAS - CCI)

Cochrane Italy

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# *Important, recent, developing initiatives*

February 06, 2017

## RARE-Bestpractices and Orphanet collaboration

**Statement of collaboration signed**

RARE-Bestpractices and Orphanet are to collaborate towards the shared goal of promoting and advancing knowledge on rare diseases.

Orphanet is a database offering a comprehensive and freely accessible repertory of information on rare diseases and orphan drugs. Orphanet is led by a consortium of around 40 countries, coordinated by the Institut national de la santé et de la recherche médicale (France).

A statement of collaboration was signed by Dr. Domenica Taruscio, RARE-Bestpractices leader and Dr. Ana Rath, Orphanet director on December 16, 2016.

The collaboration between RARE-Bestpractices and Orphanet involves the creation of links between Orphanet portal and RARE-Bestpractices web site and databases (RAREGUIDELINE and RAREGAP), the dissemination of news of each other's events and activities, the contribution to workshops, meetings, symposia of each other when appropriate and the identification of further areas of interaction

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An International Journal of Public Health
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Istituto Superiore di Sanità

Rare Best Practices

**Clinical Practice Guidelines for Rare Diseases: development and quality assessment**

October 9-10, 2018

Organised by
National Centre for Rare Diseases

Istituto Superiore di Sanità
Rome (Italy)

CNMR

# Member States *and* ERN CPGs I

- ***Beside ERN Guidelines do we need National Guideline versions as well?***
- The answer is a clear yes, because of
  - Local circumstances, care might be different
  - The prevalence of the disease might not be the same
  - English may not be perfectly understood by local doctors, HCPs and patients...
  - etc

# Member States and ERN CPGs II

- **The way to adopt international (ERN) guidelines**
  - ***Participation*** in the writing, developing committees
  - Once accepted translate into the language of the country meanwhile adopting it to ***fit national expectations/facilities***
  - Taking care, however that ***essential parts*** remain intact

- **Implementation**
  - ***Distributing and discussing Guidelines on several occasions*** („roadshows", position papers, conferences etc.) It is very important that *all participants;* specialists and patients' organizations as well as health authorities, stake-holders should be addressed.
  - To distribute Guidelines ***both*** **electronically and in printed versions**

# RD – Guidelines -*legislation developments* in Hungary

- 141/2000. (VIII. 9.) Gov. Ord. **Financial support** for severe disabled due to **chromosome aberrations/mutations**.
  https://net.jogtar.hu/jr/gen/hjegy_doc.cgi?docid=a0000141.kor

- **Lifelong data collection**, preparation for the introduction of **Orpha codes**;
  https://net.jogtar.hu/jr/gen/hjegy_doc.cgi?docid=a1400021.emm

- **Electronic database** on rare and complex diseases
  https://www.antsz.hu/felso_menu/temaink/vrony_rbk/evrony_bemutatasa.html

- **National Contact Point** (http://www.eubetegjog.hu), (eubetegjog = ***patient's rights***) according to EP and EC decision (2011/24/ EU) on transborder health care

- **Rare Disease Expert Centres** 4 Medical Schools + Nat.Oncol.Inst., 2015
- Hungarian HCPs participating in **9 ERNs** (Bond, Vascern, Metabern, Skin etc)

2017 National Plan Report

# RD – Guidelines: *further developments* in Hungary I

- ***Patient-helpline*** („life-belt”) e. http://mentoov.rirosz.hu

- RD as ***self standing entities*** in the regulations of the Ministry of Human Capacities with allocated human resources http://www.kormany.hu/download/b/7e/21000/SZMSZ%20egys%C3%A9ges%20szerkezetben.pdf

- EFOP-1.9.5-VEKOP-16-2016-00001 program for ***interdisciplinar promotion of early childhood intervention medicine*** https://www.onyf.hu/hu/hivatal/projektek/2233- efop-1-9-5-vekop-16-2016-00001.html

- ***GINOP-***2.3.2-15-2016-00039 program (1.2 Md Ft) launched in 2017 for ***new diagnostic and therapeutic tools*** in the field of RD (Szeged, Pécs, Debrecen) https://www.u-szeged.hu/fejlesztesiprojektek/ginop-2-3-2-15-2016-170718- 2/ginop-2-3-2-15-2016,http://pii.pte.hu/content/ginop-232-15-2016- 00039,https://kancellaria.palyazatok.unideb.hu/hu/node/108

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# RD – Guidelines: ***further developments*** in Hungary II

- The system of **Electronic Health Records (EHRs)** was launched, Orpha codes are part of the e-profile (EESZT, https://e- egeszsegugy.gov.hu/eeszt)

- „**Off-label**” orphan drug availability
  https://www.ogyei.gov.hu/engedelyezes_elotti_gyogyszeralkalmazas

- Beside **Debrecen Inpatient Ward (2001)**, a second one was opened at **Semmelweis University (2017)** http://semmelweis.hu/genomikai-medicina/fekvobeteg-osztaly

- Hungarian contribution to the foundation of ***UNO Rare Diseases Civil Organizing Committee***, action plan (Agenda 2030) 17 goals for sustainable development out of 8 are relevant for RD

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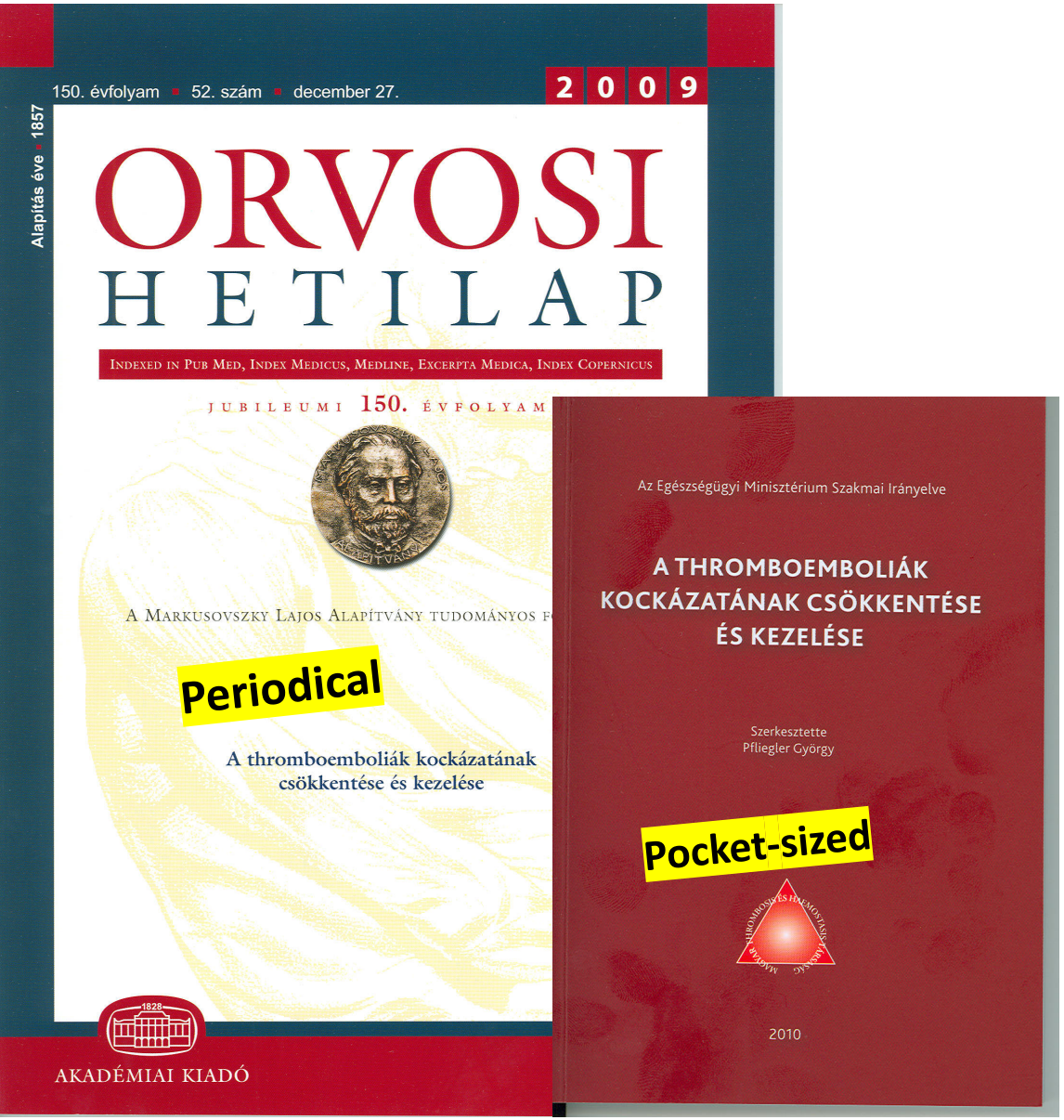
# Hungarian examples for guidelines




**Spinal musculary atrophies (SMA)**

**Chaired: Prof. MJ Molnar**

**11 Medical Colleges/Societies**

**2 Patient Organizations**

**Based on International Consensus Statement adopted to Hungary**

# Expectations and conclusions

- **Unequivocal** patient **pathways** to the centres⟹ ERNs
- **Responsibility, knowledge and competence** (Guidelines)
- All efforts have to be done to **integrate ERN and the National Health Care System**; CPMS

- ERN must be the most important and generally accepted knowledge generating, CPG presenting body in the field of RD
- Broad cooperation with local scientific and clinical societies, earlier guideline makers, patients and HCPs
- Even **Guidelines** must fullfill the criteria of the 4P of modern medicine:

**Predictive, Preventive, Personalized, Participatory...**

**”Guidelines should integrate *fragmented clinical knowledge” (V.Andriukaitis)***