Rare disease networks: Lessons learned

Kate Bushby
Newcastle University
EUCERD Joint Action
@bushbykate

This presentation is part of the project / joint action '677024 / RD-ACTION' which has received funding from the European Union's Health Programme (2014-2020).

Rare diseases: common issues

- 30 million Europeans have one of 6-8,000 rare diseases (affecting < 2/1000 people)
- >80% are genetic
- 75% present in childhood
- Scarcity of patients matched by scarcity of experts, specific therapies
- Lack of visibility in healthcare systems
- PH priorities frequently elsewhere

RD ACTION
DATA AND POLICIES FOR RARE DISEASES

EU strategies for holistic approach to RD offers huge opportunities

- Ideals encompassed in Commission Communication and Council Recommendation
- Span the whole pathway from incentives for orphan drug development to delivery of care
- Major investments in RD research via RTD funding
- Additional investment in public health agenda via EAHC

RD ACTION
DATA AND POLICIES FOR RARE DISEASES

Overlap in imperatives for therapy development and care delivery are acute in Rare Diseases

DEVELOPING THERAPIES

- Access to patients
- Access to experts
- Knowledge of natural history

- Best delivered via expert centres, patient registries and **networks**

DELIVERING CARE

- Critical mass of patients
- Multidisciplinary experts
- Ability to monitor care and impact on natural history

- Best delivered via expert centres, patient registries and **networks**

RD ACTION
DATA AND POLICIES FOR RARE DISEASES

Networking is a strength of the RD community

- Multiple grant funded and previously funded international networks
- National networks (different models, including via national plans)
- Many important tools in place (registries, guidelines, telemedicine, educational tools…)
  - Huge diversity
  - Sustainability challenge

RD ACTION
DATA AND POLICIES FOR RARE DISEASES


CURENet
TREAT-NMD
Neuromuscular Network
EN-RBD
EuroCareCF
European Coordination Action for Research in Cystic Fibrosis
eb Clinet
Linking clinical expertise in EB.
ExPO-r-Net
e-pilepsy
EUROGLYCANET
CDG
EuroCleftNet
CARE-NMD
European Skeletal Dysplasia Network
Epnet
EuroCYST
bringing knowledge - improving outcome
ecorn-cf.eu
Expert Advice on Cystic Fibrosis
EUHANET
Improving the care of European citizens with inherited bleeding disorders.
EUROFEVER PROJECT
dyscerne
A network of centres of expertise for dysmorphology
NEUROPED
Euro-Histio-Net
A reference network for Langerhans Cell Histiocytosis and associated syndromes.
EHN
EUROPEAN HUNTINGTON'S DISEASE NETWORK
EUNEFRON
European Network for the Study of Orphan Nephropathies
www.eunefron.org
PRINTO
EIMD
European registry and network for Intoxication type Metabolic Diseases
PEDDCReN
Paediatric European Digestive Diseases Clinical Research Network
Enerca
European Network for Rare and Congenital Anaemias
EuroWilson
EURenOmics
SWEET
PEDIATRIC DIABETES
www.rd-action.eu
RARECARENet
Information Network on Rare Cancers

Work of RDTF, EUCERD and Commission Expert Group on Rare Diseases Networks

- Series of in depth analyses
- Rounds of workshops (member states, experts, networks)
- Interactions with CBHC group
- Publication of Recommendations for RD networks 2013
  - Addendum 2015

Core principles

- RD are a key priority area for ERNs and vice versa
- Multidisciplinarity and patient involvement are critical issues
  - EUCERD centre of expertise recommendations support these principles
- A structured approach to grouped thematic areas for RD ERNs offers
  - The best possibility for all RD patients to have an “ERN home”
  - To ensure best sharing of resources
  - Critical mass in particularly low prevalence disease areas

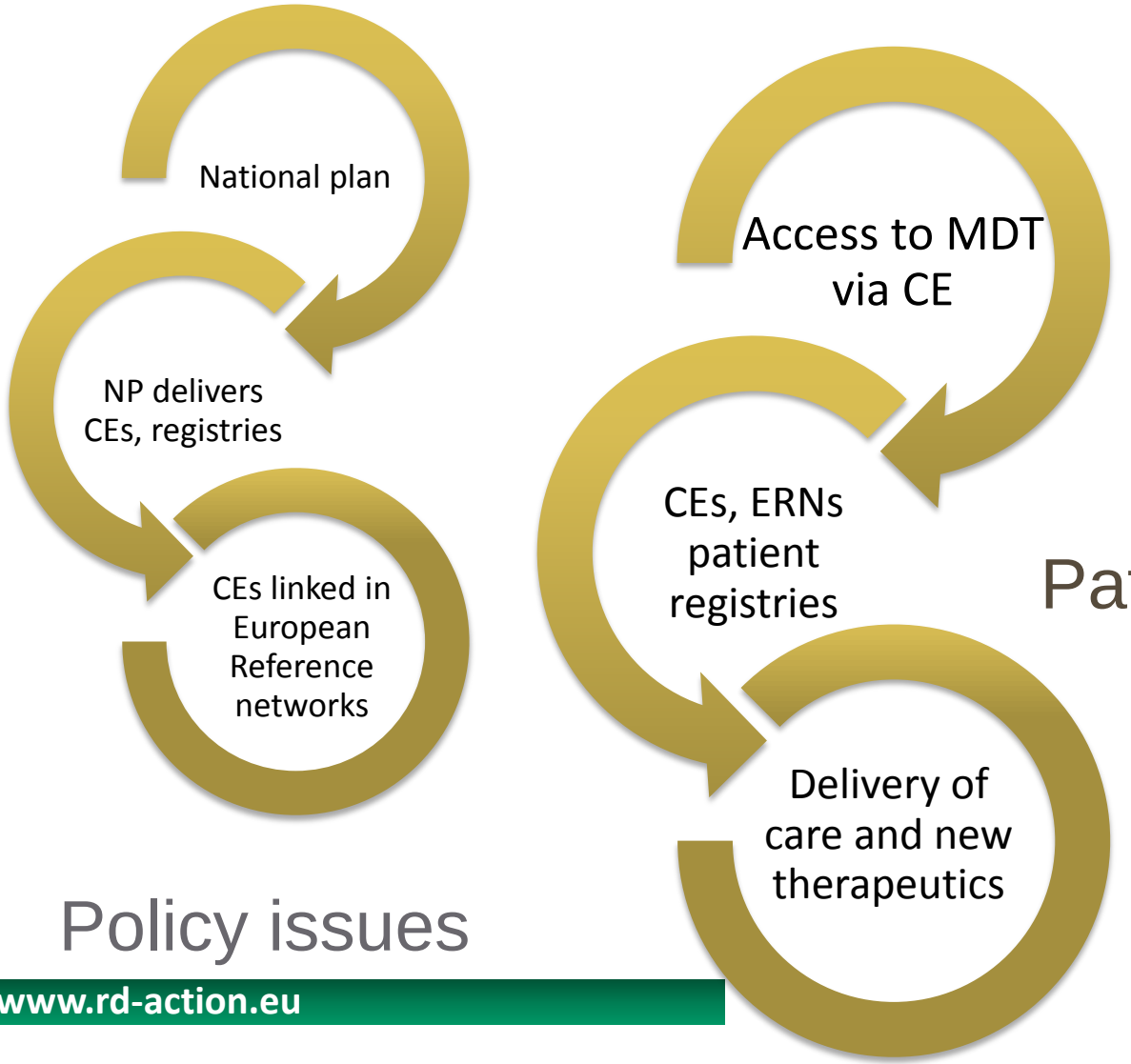
Moving to ERNs: Challenges for already networked fields

- Many current networks are based on research projects: research will be a role of ERNs but not the primary deliverable
- There are many assets in place: how will these play into the new tools and services?
- Healthcare authorities now have a formal role in every case
- Membership has to be flexible and open to any qualifying HCP
- Reorganisation and merging will be needed to “fit” into grouping

Potential incentives

- Sustainability (5 year cycle depending on evaluation)
  - But funding unclear
- Provision of supporting tools
- Network will be a legal entity
- Links with national healthcare authorities provides a formal mechanism to improve care

Keeping an eye on patient impact



RD ACTION
DATA AND POLICIES FOR RARE DISEASES

Further information and resources

www.eucerd.eu

Kate.bushby@ncl.ac.uk

@bushbykate

@rarediseasesEU


RD ACTION
DATA AND POLICIES
FOR RARE DISEASES

A model for the purposes of grouping RD thematically

Rare immunological and auto-inflammatory diseases
Rare bone diseases
Rare cancers* and tumours
Rare cardiac diseases
Rare connective tissue and musculoskeletal diseases
Rare malformations and developmental anomalies and rare intellectual disabilities
Rare endocrine diseases
Rare eye diseases
Rare gastrointestinal diseases
Rare gynaecological and obstetric diseases
Rare haematological diseases

RD ACTION
DATA AND POLICIES FOR RARE DISEASES

Rare craniofacial anomalies and ENT disorders
Rare hepatic diseases
Rare hereditary metabolic disorders
Rare multi-systemic vascular diseases
Rare neurological diseases
Rare neuromuscular diseases
Rare pulmonary diseases
Rare renal diseases
Rare skin disorders
Rare urogenital diseases

*Note: The networking of rare cancers is currently under discussion in EC Expert Group on Cancer Control.

Relevant Recommendation Text

Recommendation 15: Ahead of the designation process for ERNs, consideration should be given to the possible economies of scale of developing shared platforms across RD ERNs such as core components for registries, QA etc. (as listed in recommendation 4).

Recommendation 16: A clear process for the designation of RD ERNs should be established. Criteria for the evaluation of prospective ERNs should include their inclusiveness and plans for expansion, excellence of the network, leadership qualities of the proposed co-ordinator, and numbers of MS involved, amongst others

Recommendation 17: A step-wise strategy for RD ERN designation should be delineated so that all patients with a rare disease will have access to an appropriate ERN in a defined period of time. This should include access to an ERN for those rare disease patients still seeking a diagnosis.

Recommendation 18: As it will only be possible to establish a limited number of RD ERNs at the beginning of the process, it is recommended to give priority to ERNs which meet the following 3 priority criteria as a robust starting point: 1. Existing formal or informal networks of experts have reached maturity; 2. There are patient registries established and willing to interoperate; and 3. There are existing networks of patient groups. Each thematic RD ERN would still need to expand over the course of its first five years of designation to include other centres, expert groups, patient groups and ultimately diseases.

Recommendation 19: Based around the concept of medical specialties and body systems, diagnostic and therapeutic areas can be identified each covering a wide range of rare diseases. Comparison of the systems in place in MS with well-developed services for rare diseases shows that the number of diagnostic and systemic areas which might cover the majority of diagnoses could be approximately 20-30. By the end of the Health for Growth Programme (in 2020), the 20 to 30 ERNs should be established and covering a wide range of RD. These first established ERNs will be the ones meeting the “priority criteria” as defined above and will then progressively expand in order to cover all RDs by the end of the two next EU Public Health Programmes (by 2025), through integration of appropriate centres and expertise.

Recommendation 8: *Patient organisations should play an integral role, especially in the evaluation of RD ERNs, where patient organisations exist.*

Recommendation 4.v.: Mechanisms for evaluation and clear indicators of performance: *The groups providing evaluation of RD ERNs should be multi-stakeholder and include patient organization.*

*****

The *EUCERD Recommendations on Quality criteria for Centres of Expertise for Rare Diseases in Member States:*

Recommendation 7: *CEs collaborate with patient organisations to bring in the patient perspective*

Recommendation 29: *Link and collaboration with patient organisations where they exist*