EURORDIS
Rare Diseases Europe

Inspiration from national conferences: an overview of successes in national rare disease strategies – all topics of good practice

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National Alliances still need

"strong and common advocacy tools"

to promote the good measures to be adopted and implemented in their country in each of the 6 themes addressed in the EUROPLAN conference

How EURORDIS and EUROPLAN Advisors can help?

→ Proposal to identify **1) good practices** & **2) inspiring proposals** on governance, research, access to care (CEs & ERNs), orphan drugs and social care

from Europlan Conference Reports + current RD National Plans + State of the Art on RDs, others…

→ **Ongoing work**: to be discussed at this Membership Meeting

→ **Collective work**: Good Practices & Inspiring proposals reviewed by National Alliances / members

Will inform the implementation phase of RD Plans

Good Practices (and proposals) from :

National Plans/National Strategies
State of the Art Report 2014
EUROPLAN National Conferences for:

Centres of Expertise
Social Care
Research
Orphan Medicinal Products

Best practice and proposals on CoE

- Designation and evaluation of Centres of Expertise
- Scope and functioning of Centres of Expertise
- Multidisciplinary, healthcare pathways and continuity of care
- Research in Centres of Expertise
- Diagnostic and genetic testing
- European and international collaboration
- Sustainability

The Spanish model - based on three pillars

- The model is based on three pillars: 1. **diagnostic systems and early diagnosis, 2. comprehensive care plan and 3. support and assistance for patients and their families.**
- *The Autonomous Communities are responsible for the provision of health care. NHS has established the **procedures for the designation of Centres of Reference** (*Royal Decree 1302/2006 and 1207/2006 for accreditation and management of the Cohesion Fund *). The Reference Centre would act as the leading centre for the confirmation of diagnosis, definition of therapeutic strategies and continued treatment, as well as a reference point for those health care centres which provide the day-to-day care. The involvement of patient associations depends largely on the professionals in the Reference Centre and their interest in encouraging that participation.*
- **CoE in Spain must have a nationwide scope** and access to those centres must be guaranteed on equal terms to all patients;
- Role of the Patient Case Manager at a Centre of Reference

France model - based on 3 level accredited model

1. **National – National federation of the networks on rare diseases** (Centres of Expertise, Network of Reference Laboratories)
2. **Regional – Competence centres** (referral, research, data collection, reference laboratories and research centres)
3. **Local – Primary care services and secondary care hospitals (**National federation of the networks on RD: reference centres, competence centres and future platform of reference laboratories)

The centers are accredited and funded for five years

Evaluation: First round (2009-2012)

Self-assessment after 3 years (assessment manual produced by the French National Authority for Health –HAS)

External assessment (organised by HAS) after 5 years (only the coordinating sites): final report by HAS

Decision by the Ministry of Health: renewal of authorisation granted

Care is managed across this tiered model through inter-hospital agreements

CoE for 1 RD & CoE for group of RDs: Patient care pathways and Case Managers, Registries & Research

The German model - 3 types

Care, centres and networks for systemic complex diseases needing a multidisciplinary approach that are financially sustainable using orphan and off-label drugs. Centres of three types:

Type C, a **centre for a specific disease or for a group of diseases encompassing hospital and non hospital practices and delivering concrete care to patients;**

Type B, a **centre of expertise**, **for a specific disease or group of diseases in an hospital with in-patient care;**

Type A, a **reference centre**, composed of **more than two type B centres** for disease specific and for non disease specific activities such as dealing with patients with an unclear diagnosis, training, research.

It **guides patients throughout the system**, provides standard approaches to diagnosis and remote diagnostic procedures, provides resources such as registries, biobanks, innovation, continuing medical education, ERNs, guidelines, support to types B and C and multidisciplinary research. **There is a national network of type A centres.**

The UK Strategy for Rare Diseases

- ***A UK-wide vision:*** equity of access to defined care pathways, patient centered coordinated and integrated care, evidence-based diagnosis and treatment, support, translational research, collaboration, education and training
- **CoE structured around medical specialities, not diseases.**
- The UK has highly specialist clinical centres that are focused solely on ultra-rare diseases.
- The UK Strategy for Rare Diseases identifies the need for specialist clinical centres to have a minimum set of standards that reflect the EUCERD recommendations.
- ***Coordination of care:*** integrating GPs; extensive care plans with defined responsibilities and agreed information exchanges; integrating levels of care, social and voluntary care; timely referrals to specialist clinical centres; seamless transitions; measures of quality and outcome, sufficient case load; involvement of patients

Best practice on Social Care for RD

FOCUS

- The patient in the centre of care: care pathways & individual care plans; case management
- Coordination and networking
- Supporting patients' day-to-day challenges & integrating RDs into specialised social services
- Social research, information/data collection & sharing
- Training professionals and patients/families
- The role of Centres of Expertise in Social Care
- Assessment of (Dis)abilities & (in)capacities
- Quality and good practices

Spain :

- **Complex care protocols** and models that specify the care and referral processes, as well as the health services and social services
- Bodies to coordinate health and social care services (**case manager**)
- State Reference Centre for Rare Diseases Patients and their Families – Burgos
- **Psychological care**, support and guidance throughout the process of the disease as well as at the time of diagnosis and during genetic counselling
- **Help the people close to them** (home assistance services, hospital at home services, day centres or short-stay centres, etc.)
- Promote **access to hospital classrooms** for schoolchildren with RD
- **Official master degrees in Rare Diseases** (Murcia), knowledge of Rare Diseases (Seville), Integral Psychological Intervention in Rare Diseases (Seville)
- **Educational guides** for teaching staff
- **Common platform** is needed for sharing information between health, social services education and employment professionals, enabling access to the necessary data of each person required for different benefits. Plan to have a national law approved on this matter in the next 4 years

Romania

- NoRo – a Resource Centre for RD (RPWA &RONARD)
- Initiation of a National Plan/ strategy to adapt the existing services to patients' needs through training of the staff;
- **Therapeutic Recreation Camps** for rare cancers, haemophilia and other RD
- Accredited trainings for different professionals & online platform: www.edubolirare.ro
- Developing a **standard cost for specialised social services for RD**
- Reassessment and adaptation of criteria for defining the **degree of disability** **Palliative care** for patients with severe disabilities produced by RD
- Establishment of **networks of health care and social assistance clustered around centres of expertise to ensure the continuity of care**
- Cooperation with CoE through **videoconference system and Helpline**
- A study performed for **evaluation of the quality of life** for patients with RD (>600 patients enrolled)

France

- **Linking teams and platforms** connected to the national plan for rare disabilities (CNSA)
- **Case managers** connected to CoE
- Develop **Disability factsheets**
- **Health Networks of Rare Diseases** organised to adapt to specificities of the disease in which they operate will further support social services
- **Better coordinate centres of expertise with the department services for people with disabilities**
- **National Plan** by the Ministry of Economy, Finances and Industry, Ministry of High Education and Research, Ministry of Solidarity and Social Cohesion, Ministry of Work, Employment and Health
- **Develop organisation methods** to respond to the need of respite for people with a rare disease or for their carers by for example reserving places in temporary stays in nursing homes or specialised centres across regions

Scandinavian countries

- Individual care plan & care programme & personalised service plan &patient pathway made for patients and families in need of multi professional services
- One stop shop services (Frambu & Agrenska): organise a central office which would provide information and training on all procedures, rights, means, and support available (medical, social, professional)
- Rare Disorders in Norway: How users experience the health services' (report produced by SINTEF December 2008) – Summary in English (NO);
- Creation of a central information portal for RDs (DE)
- Studies on the psychosocial problems faced by patients with rare diseases as well as on possible new approaches are of particular importance (DE);
- Knowledge of professionals (for example physiotherapists and social workers) concerning rare diseases has to be improved (FI EUROPLAN Conf.);
- There should also be a grant for patient representatives' participation in studies/investigations relevant to rare diagnosis groups (SE EUROPLAN Conf.);

Best practice on Research

FOCUS

- RD funding programs
- Research gouvernance
- Patients involvement in research for RD
- Research infrastructure
- Research area

1. RD funding programmes / calls

GOOD PRACTICES: Funding programmes and/or calls are specifically dedicated to RD research:

FRANCE – Yearly calls of the Research Programme for Hospital Clinical Research (PHRC): thematic priorities include RDs

AUSTRIA - In 2013, the Austrian Research Promotion Agency (FFG) published a specific programme for rare diseases for SMEs, amounting to €5 million in total.

SPAIN – 3 main strands: 1) The Health Research Fund (FIS) in Spain - RD as an area for promotion. Between 2008 and 2013: 260 specific RD projects financed = almost 31,8 MEUR; 2) The State Plan for Scientific Research 2013-2016 - RD as one of priority areas for research and finances research projects in this area 3) CIBERER finances intramural projects. The 2012 and 2014 calls totalled 1,1 MEUR.

→ CIBERER awards 10 scholarships a year with a duration of 1 year for young researchers who want to start their career in CIBERER affiliated groups.

NB – In EUROPLAN National Conferences, most of countries where this does not exist called for a specific RD programmes

2. RD research governance

GOOD PRACTICES: a structure/network exists for fostering and coordinating RD research across the country:

FRANCE – The **"*Fondation Maladies Rares*"** (Rare Disease Foundation)

SPAIN - The Centre for Biomedical Network Research on RD (**CIBERER**)

UK – The Rare Diseases Translational Research Collaboration (**TRC**)

DE - German NP, action 14: "Implementation of a **cooperative platform to broker the engagement between academia and industry**. It should include patient patient organisations as well as SMES as part of a multistakeholder process."

LUX – The **Personalised Medicine Consortium of Luxembourg** (**PMC**) groups entities associated with European organisations and participate in joint projects, coordinates research projects with important multidisciplinary teams.

2. RD research governance

PROPOSAL S

→ Closer relations with **Centres of Expertise** should be established and research in Centres should be fostered

HU, HR, DE, IT

PROPOSAL

→ Areas for which research is lacking and **priorities** for future calls should be identified. The integration of RD research within relevant government research policy and legislation should be supported.

→ A comprehensive **strategy** should be devised.

IT, IE, DK

2.1.Patient involvement in RD research

GOOD PRACTICES: Patients are involved in research programmes, as inputs from patients and their families can improve quality and effectiveness of research:

UK - NIHR , England: patient and public involvement is included within the assessment and awarding of all research across all its funding programmes

IT - "Open days" with research labs, specific meetings with patients and training initiatives to enhance patient participation in research

NB – In many countries, active engagement of PLWRD in research is an essential component at different stages of research; their participation in research decision-making bodies must be institutionalised

3. Infrastructures for RD research governance

GOOD PRACTICE: Interoperable databases and registries are established to pool together scattered data, also across countries:

DK, SW, FI – RAREDIS, the Nordic database for rare diseases, developed in Denmark, is supported by the Nordic Council of Ministers. Centres of rare diseases in Nordic countries use their local version for collecting clinical data on patients, by collecting information that can be pooled and used for research projects and benchmarking on a Nordic level.

GOOD PRACTICE: The national biobanking plan provides coordination and quality standards:

IRELAND - The forthcoming national biobanking plan provides national coordination and quality standards for biobanking and embraces all opportunities for rare disease research.

4. Research areas

GOOD PRACTICE: Investments are made to make whole genome sequencing available to priority groups of patients, including people living with RD:

GB –£100 million pledged to make whole genome sequencing available to NHS patients / 3 to 5 years. Expected to generate 100,000 whole genome sequences. RDs one of the initial priority disease groups.

NB – In GB and other countries it was pointed out that ethical discussion is needed to accompany these measures. Pros and cons should be discussed, as well as right to know of the individual/family, consequences for patient.

GOOD PRACTICE: Research in social and economic sciences is supported with specific calls and interdisciplinary teams, including patients. :

FR – The RD Foundation launched two calls for projects in 2012 and 2013 to increase knowledge on individual /social consequences of living with a RD esp. in terms of limitation of activities and integration in society: 17 projects funded, 76 teams of clinicians & researchers, 20 patient associations.

Best practice on Orphan Medicinal products

FOCUS

- Overview of the best practices identified Access to OMPs
- Incentives to OMP Development
- Pricing / reimbursement
- Exchange on Clinical Added Value
- Innovative approaches to MA
- Access to non-approved OMPs: compassionate use and off-label use

Best practices identified for Access to OMPs

Incentives to OMP Development:

Best Practice 1:

"Fondo AIFA 5%" in *Italy*. AIFA established an innovative funding scheme operational since 2005, by which pharma companies are required to donate 5% of their promotional expenditure to an independent research fund → ½ for the reimbursement of orphan or life saving drugs waiting market entry and ½ support independent research, drug information programme and pharmacovigilance.

Best Practice 2:

In **Belgium,** revenues of OMPs are no longer subject to the so-called "pharmaceutical taxes" on sales of reimbursable drugs.

Best Practice 3:

In **the Netherlands,** Dutch pharmaceutical SMEs are given a subsidy for the costs of submitting a dossier for designation and MA to the EMA.

Pharmacovigilance, including reporting tools on adverse reactions

Best Practice 4:

In **France**, the law provided for the opening of the pharmacovigilance *system* to direct reporting of undesired effects from patients, through an online form.

Best practices identified for Access to OMPs

Pricing / Reimbursement

Best Practice 5

In the Netherlands, when the therapeutic value is not clear, the reimbursement should be conditional for a period of 4 years (this period could/should be shorter)

→ **Conditional MA** for producers, reduces prices for payers and ensures reimbursement / access to patients.

→ During this period, LT data generation post-conditional MA. **Strong link with establishment of registries.**

Best practice 6:

In Italy, according to the "Balduzzi Law" (2012), pharmaceutical companies can apply for P&R to AIFA as soon as CHMP gives a positive opinion

→ Quicker decision-making starting before the EC has granted MA.

Best practice 7:

OMPs have to show therapeutic added-value but there is no obligation for pharmaceutical companies to provide pharmaco-economic data/evaluation for OMPs to address a rare disease with no alternative treatment exit. This is valid in *Belgium, Romania and in the Netherlands*.

Best practices identified for Access to OMPs

Exchange on Clinical Added Value:

Best Practice 8: In **Spain, the Therapeutic Product Reports** (TPRs) provide **single assessment of the added value of medicinal products for the entire territory to secure equal access to medicinal products in all regions of Spain.**

Innovative approaches to MA:

Best Practice 9: In ***Italy***, there is a fast-track mechanism for ODs to enter Pricing / reimbursement negotiation before MA is granted at EMA level.

Access to non-approved OMPs: compassionate use and off-label use

Best practice 10: ***In France*****,** there is a system ATU whereby a MP without MA can be prescribed under exceptional circumstances for severe and life-threatening diseases, either for cohorts (group, phase III) or for individuals (phase II, no therapeutic option, under the responsibility of prescribing doctor).

through the **Special Solidarity Fund (SSF)** (OMP = 35%).

Best practices identified for Access to OMPs

Best practice 11: In Italy, when a MP is not authorised, there are two possibilities:
1. Use of OMP on a national basis (Law 648): **compassionate use** allows for prescription – **paid by the producer** – of drugs not yet authorised (Phase II or III Clinical Trials) but where favourable evaluation expected in terms of efficacy/safety.
2. Off-label: in hospital, by a doctor, personal basis, no other treatment option. Prescription on an individual ad hoc basis for one patient through the **Fondo AIFA 5%, where** ½ of the Fund goes to purchase OMPs and drugs representing a hope for treatment for severe pathologies.

Best practice 12: In Belgium, there is a Compassionate Use programmes or Medical Need Programme (off-label) are provided for in ***Belgium*** since 2006 and the possibility to reimburse OMPs which are not covered by the basket of benefit

Thank you!
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- Questions?
- Advices?
- Suggestions?
- Examples?