The Second French Plan for Rare Diseases 2011-2014

Major Developments

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General Secretary of the National Plan for Rare Diseases 2011-2014
First French National Plan 2005-2008

Foundations

- Recognize the specificity of rare diseases and develop information on rare diseases for patients, health professionals and the general public (Orphanet database)
- Improve access to health care and quality of care thanks to a network of 631 centres of expertise: 131 at national level called “centres of reference”, and 500 at regional level called “centres of competence”
- Continue efforts in favour of access to orphan drugs and specific measures (compassionate use) for delivery of medicines
- Give specific funding for research on rare diseases and creation of Rare Disease Institute-GIS for coordinating research

2009 assessment: 200 M € spent, 100 M€ for permanent support to centres of reference, 40 millions for research!
Second French National Plan (NPRD2) 2011-2014

After several years of progress, time for consolidation

Consolidation of previous achievements:
- New procedure and assessment criteria for centres of reference and development of coordination between these centres
- Increasing the role of Orphanet
- Development and coordination of research

Coverage for each patient and any rare disease will be guaranteed:
- Improving the quality of patient care
- Better visibility and better access to diagnosis, treatment and social services

Amplifying European and international cooperation
5 working groups created (experts, administration, health agencies, centres of reference, patients organisations, academic societies)

1) New criteria and simpler evaluation for (re)designations of Centres of Reference (CoR)

- Annual activity report of CoR online
- External evaluation of CoR every 5 years by the French National Authority for Health (HAS), using a new set of criteria
- Recommendations to the Minister of Health, made by a permanent working group
- (Re)Designation of CoR by the Minister of Health
2) Creation of 20 to 30 Healthcare pathways, vertical networks called « filières », gathering several CoR, to ensure the coordination and visibility of all CoR in the same domain of diseases, the coverage of all rare diseases, and the equal quality for patients care from diagnosis to provision of healthcare and social services.

- Definition of the main goals of this grouping
- Survey of existing thematic networks
- Facilitating and supporting the network creation, taking into account the existing classification of CoR in 18 groups of rares diseases
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Healthcare pathways : main goals

- Share resources, expertise, information for patients to improve patients care and research
- Collect clinical data for epidemiological and therapeutic research
- Organize patients healthcare pathways in connection with local or regional platforms for information and coordination of healthcare and social services (i.e. complex case management)
- Produce, share and use good practice guidelines (“National Diagnosis and Treatment Protocols”, PNDS)
- Implement or improve training on rare diseases for health professionals, and therapeutic education for patients and family carers
- Improve visibility of CoR networks at the national level, and in the perspective of European Reference Networks
Interconnexion of networks

National network « Filières »
Local / regional platform
Coordination of expertise
Coordination of health and social actors
European Reference Networks
3) Creation of the National Data Bank for Rare Diseases aimed at coordinating and developing the collection of epidemiological data

- Survey of data currently collected in all 131 RC
- Definition of a minimal common data set to collect for each patient in all centres of expertise (CoR and centres of competence), registries and cohorts (RADICO project), using orphacodes
- Taking into account the progress of Epirare project
- In connexion with the new « Fondation Maladies Rares » created on 29 February 2012 to impulse and advise rare diseases research, and collect private funding (executive director Pr Nicolas Levy)
4) Dotation for Next Generation Sequencing (NGS) Technology for rare diseases diagnosis to 25 university hospital laboratories of genetics

- Survey of sequencing technology needs in all university hospital laboratories of genetics
- Recommendations for dotation made by a working group
- Direct dotation of medium high frequency equipment (to analyze 15 to 20 gene for diagnosis) by Ministry of Health (9,6 M€ over 2011 and 2012)
- Discussion on very high NGS needs for care (to analyze exome or total genome)
Second French National Plan Implementation

5) Simplification of development and increase of number of « National Diagnosis and Treatment Protocols » (PNDS), produced by the CoR with a methodological support of HAS

In France, PNDS:

- are good practice guidelines for centres of expertise and also ambulatory care (recommendations for GP)
- are used to define the list of healthcare products and services recognized as necessary for patients, but not systematically fully refunded to patients by the National Health Insurance (NHI)
- may be used to allow derogative coverage of off-label prescription of drugs or non-covered medical devices by NHI (cf. new French law reinforcing the monitoring of safety of drugs and other healthcare products)

Objective: from 40 PNDS produced during the first Plan to 200 PNDS at the end of the second Plan
The second French National Plan 2011-2014

In a nutshell:

3 axes, 15 measures, 47 actions, 4 focus areas, more than 200 M€

French most important assets for progress:

- 25 years success of AFM/TELETHON
- National active role of « Alliance Maladies Rares » (202 patients associations) supported by Eurordis
- ORPHANET: a powerful tool
- Widespread efforts of patient organisations and health professionals to promote rare diseases information, research, quality of care
- Close and exemplary collaboration between patient organisations, health professionals, scientific researchers and public authorities
Rare diseases:

- A model for structuring health care provision and research

- A model for a tolerant society and an open democracy

Rare but exemplary