



1st French National Plan for Rare Diseases 2005-2008 Evaluation after 4 years

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Main objective of the National Plan in France:

- **« ensuring equity in the access to diagnosis, treatments and provision of care »**
- **After 4 years, an evaluation was conducted by the « Haut Comité de santé Publique » : all main stakeholders agreed that the situation has improved**
 - **Rare diseases are a model for the involvement of patient associations, for pluridisciplinary care.**
- **However, need to better link specialised care to proximity care, medical to social care, basic research to clinics, to improve collection of data, develop European partnerships, and ensure sustainability.**

The National Plan in France : Strengths (1)

- **Methodology** : Participation of all stakeholders: patient representatives, health care professionals, researchers, health authorities, pharmaceutical industry.
- Independent evaluation by the « Haut Comité de Santé Publique »
- **Information** : Development of Orphanet database, of a classification and coding for rare diseases with EC and WHO
- Care and information personal cards
- Dissemination of information leaflets, websites by associations and centres of reference
- Support for an information helpline (Maladies Rares Info Service)
- **Promotion of research**: higher budget for longer projects

Strengths (2)

- **Better access to care and orphan drugs:** Designation and funding by the Ministry of Health of 132 « centres de référence » (specialised centres at National level) and 500 « centres de compétence » (provision of care at regional level): pluridisciplinary approach, production of information, clinical research, clinical trials...
- 80 new counsellors in genetics,
- Production of good practice guidelines for 17 diseases, 24 in preparation : slow process, updating necessary, European cooperation needed.
- Availability and reimbursement of orphan drugs by Social Security, compassionate use system

The National Plan in France: Weaknesses (1)

- **Methodology** : poor monitoring during the Plan, lack of indicators
- **Epidemiology**: poor monitoring of activity of « centres de référence », clinical data scattered. Lack of funding for registries. Specialised centres need technical and scientific support and human resources.
- **Access to diagnosis** still difficult, particularly for diseases with intellectual impairment
- **Lack of training of professionals**: GPs, public and private insurance medical experts, school medical professionals, paramedics, social workers, need for continuous medical training...
- **Information**: long term consolidation of Orphanet

Weaknesses (2)

- **Screening**: lack of consensus on technologies, costs, innovations, lack of quality control of gene testing, lack of equity in individual screening.
- **Access to social care**: currently no articulation with medical care provided by centres of reference, lack of information and training of social workers, recurrent problems of reimbursement of transportation, sometimes of consultations in specialised centres
- **Access to drugs prescribed off-label**
- **Lack of support for patient representatives, little support to patient group activities** (capacity building, training...)
- **Research**: need for more research partnerships at National and European level
- **SUSTAINABILITY**: need for a second Plan acknowledged

Main focus of the second Plan

position of the Plateforme Maladies Rares:

Alliance Maladies Rares, MRIS, Orphanet, Eurordis, Institut des Maladies Rares

- **We are asking for**
- **The establishment of an interministerial steering committee for the next plan**
- **The following preparatory working groups:**
 - How to speed up basic and pre-clinical research?
 - Marketing authorisation conditions and access to treatments
 - Development of therapies
 - Diagnosis tests, screening: organisation, development and funding
 - Information systems: databases, cohorts and registries
 - Consolidation and development of reference and competence centers, link with proximity care.
 - Financial and social compensation of disabilities linked to rare diseases