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RÉPUBLIQUE FRANÇAISE

Ministère de la santé
et de la protection sociale

Secrétariat d'Etat
aux personnes handicapées



French National Plan for Rare Diseases 2005 – 2008

**“Ensuring equity in the access to diagnosis, treatment and
provision of care”**

20 November 2004

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Foreword

The definition of the rarity of a disease – less than one person in 2000 affected according to the threshold retained in Europe – should not mask the importance of the challenge that rare diseases represent for public health. Each of these disorders affects less than 30,000 patients in France. Certain diseases that are rare in metropolitan France, like sickle cell anaemia, are quite frequent in French overseas territories and departments. A large number of these pathologies are also called “orphan diseases” because the affected populations do not have access to any therapeutic treatment.

Health professionals have insufficient knowledge of the majority of rare diseases. This lack of knowledge underlies diagnostic error - a great source of suffering for patients and their families - and delayed care provision, which can sometimes be prejudicial.

Early diagnosis and follow-up require multidisciplinary teams, associating both scientific expertise and medical competence, permitting patients to benefit rapidly from advances in research. These specialised teams, in a limited number, should be able to be recognised and clearly identified by patients and health professionals.

Rare diseases present a new medical problem: the need to learn to discover and recognise the exception, to progress in the understanding of the disease, to share information and experience and to organise networks supported by centres of reference.

Faced with 7000 known rare diseases, it is easy to feel invaded by a sense of powerlessness. In terms of cost and efficiency, the efforts made by medical research to treat and overcome these diseases can appear to have little value: for some, spending so much time and energy on a disease encountered once or twice in five years makes little sense.

Rare diseases present a political problem, in the noblest sense: that of taking into account the needs of the weakest and the fewest in number. If we want to guarantee equal access to treatment, if we are looking for the best quality of care and support, then it is obvious that the problem concerns the entire health and socio-medical system.

Rare diseases are, in most cases, serious, chronic and debilitating, often requiring long and heavy specialised treatments. In addition, they often result in some form of handicap, sometimes extremely severe. The impact on families is often major and it is the mobilisation of these families that has helped gain recognition of the importance of these long-ignored diseases. The misreading of various aspects of the disease by social and socio-medical personnel can provoke delays in the provision of appropriate help. For that reason the measures outlined in the *Loi sur l'égalité des droits et des chances, la participation et la citoyenneté des personnes handicapées*¹ integrate the specificity of rare diseases.

Even though knowledge of the origin of these diseases has significantly progressed, notably through the identification of genes that already allow the formal identification of 800 diseases, there is still progress to be made in research.

The pharmaceutical industry also constitutes one of the major sources of innovation, with new drugs entering the market that have benefited from the incitements resulting from the December 1999 European legislation on orphan drugs.

¹ Law on the equality of rights and opportunities, the participation and the citizenship of the handicapped, adopted on 3 February 2005

A determined policy in this domain implicating all partners, notably industry and the voluntary sector, should allow the efforts already underway to continue, thanks to a reinforcement of the means dedicated to research on rare diseases, both in diagnosis and in treatment.

Finally, the associations of people affected by rare diseases, and in particular the *Association française contre les myopathies* (AFM)², play an important role in this domain and must be supported. The AFM was the motor behind a real collective awareness raising; it has become a key player and important partner of health professionals, researchers and public authorities.

Considering the stakes at play, rare diseases have been included as one of the five major priorities of the 9 August 2004 *Loi relative à la politique de santé publique*³. Translating this strong political engagement, the National Plan for Rare Diseases proposes a series of concrete measures that will be coherent and structuring for the organisation of our system of care provision. They should provide a response to the expectations of patients and their families.

This plan is the result of a wide consultation process, associating experts in rare diseases, clinicians and researchers, representatives from patient groups, from the national health insurance⁴, and from the ministries of health and of research.

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François d'AUBERT
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² AFM - French Muscular Dystrophy Association

³ Law relating to public health policy

⁴ *Assurance Maladie* : the system of mandatory national health insurance, part of the French social security system dealing with sickness benefits

Rare diseases – the figures

A disease is said to be rare if **less than one person in 2,000 is affected**. In France, this means less than 30,000 people affected for a given disease.

Nearly 7,000 rare diseases, more or less debilitating, have been identified. Five new pathologies are described each week in the world, 80% of which have a genetic origin.

6-8% of the world's population are likely to be concerned, at some level, by these diseases: more than **3 million people in France**, 27 million in Europe and 27 million in North America.

Around 50 rare diseases each affect a few thousand people in France, whilst around 500 diseases affect only hundreds of people each. There are thousands of other rare diseases that only affect a few dozen people each.

As an example, in France there are:

- 15,000 people suffering from **sickle cell anaemia**
- 8,000 people suffering from **amyotrophic lateral sclerosis**
- 5,000 - 6,000 people suffering from **cystic fibrosis**
- 5,000 people suffering from **Duchenne muscular dystrophy**
- 400 - 500 people suffering from **leukodystrophy**
- a few cases of **progeria** or premature ageing (less than 100 cases in the world)

65% of rare diseases are serious and debilitating. They are characterised by:

- **appearing early in life**, two out of three times before the age of 2 years old
- **chronic pain** in one patient out of five
- a **motor, sensory or intellectual deficiency** in half of all cases, leading to an incapacity which reduces autonomy in one case out of three
- an impairment of **vital prognosis** in half of all cases, with rare diseases responsible for 35% of deaths before the age of 1 year old, 10% between the ages of 1 and 5 years old and 12% between 5 and 15 years old.

Rare diseases : a national plan structured around ten strategic priorities

The improvement in healthcare provision for rare diseases constitutes a major challenge for public health owing to the epidemiological data, the consequences of these pathologies for the quality of life of the patients and their families and the challenges for research in the domains of diagnosis and treatment.

France has taken many initiatives in this area over the last few years in collaboration with patients' associations, thanks in particular to the *Association Française contre les Myopathies* (AFM): creation of an orphan drugs mission (1995); financing of a telephone help line service for patients (1995) and of an online information server, Orphanet (1997); financing of clinical research on rare diseases in the programme of clinical research in hospitals (since 2001); providing access to the list of clinical trials by the *Agence française de sécurité sanitaire des produits de santé* (AFSSAPS⁵, 2002); creation of a *Groupement d'Intérêt Scientifique - Institut des Maladies Rares*⁶ (2002).

The determined policy of France in the domain of rare diseases enabled the adoption, in January 2000, of European legislation on orphan drugs.

The National Rare Diseases Plan 2004 – 2008, part of the 9 August 2004 law relating to public health policy, will develop, reinforce and bring coherence to these different initiatives.

Its priority is “to ensure equity in the access to diagnosis, to treatment and to provision of care” for people suffering from a rare disease through ten strategic priorities:

- ❑ Increase knowledge of the epidemiology of rare diseases
- ❑ Recognise the specificity of rare diseases
- ❑ Develop information for patients, health professionals and the general public concerning rare diseases
- ❑ Train professionals to better identify them
- ❑ Organise screening and access to diagnostic tests
- ❑ Improve access to treatment and the quality of healthcare provision for patients
- ❑ Continue efforts in favour of orphan drugs
- ❑ Respond to the specific needs of accompaniment of people suffering from rare diseases and develop support for patients' associations
- ❑ Promote research and innovation on rare diseases, notably for treatments
- ❑ Develop national and European partnerships in the domain of rare diseases

⁵ AFSSAPS : French Health Products Safety Agency

⁶ Scientific interest group for rare diseases

1: Increase knowledge of the epidemiology of rare diseases

Context

There is currently no epidemiological surveillance organised for rare diseases in France. Putting in place such surveillance requires that their specificities be taken into account. Their diversity prohibits an exhaustive surveillance of all the pathologies concerned.

The nomenclature is poorly adapted to an epidemiological surveillance of these diseases. The large number of different places and types of care provision for the rare disease (specialised centres, but also non-specialised services, specialist practitioners...) makes it difficult to identify and locate these diseases.

The requirements in terms of epidemiological surveillance vary according to the pathologies, their prognosis, the nature of patient care, the emergence of innovative therapeutics, and the cost, whether economic or social.

Certain data sources on rare diseases already exist but they have not been used for epidemiological purposes. If they are to become operational, there needs to be a reflection on the possible ways of using them and feasibility studies.

The objective is to put in place, over five years, a coherent policy of broad epidemiological surveillance and a more specific surveillance for certain rare diseases, in terms of incidence, prevalence, repercussions on mortality and morbidity, quality of life and the circuits of patient care.

Objectives

- To increase knowledge of rare diseases by an epidemiological surveillance, which will allow a better understanding of the natural history of these diseases, an evaluation of the needs and the progression of patients within the health system, and a monitoring of the evolution of indicators relative to the quality of life of patients.

Measures

- **The *Institut de Veille Sanitaire (InVS)*⁷ is responsible for putting in place and coordinating the epidemiological surveillance of rare diseases**, in liaison with all parties concerned. This mission will be included in the work programme of the InVS from 2005, through the intermediary of the contract of objectives and means (COM)⁸. Its missions in this domain are:
 - to develop a nomenclature and a classification adapted to rare diseases, in collaboration with the international bodies: the World Health Organisation and the European bodies, notably the Rare Disease Task Force which brings together European experts on rare diseases;
 - to ensure a coordination between different partners (international bodies, health professionals, notably the centres of reference on rare diseases and the patients associations) in order to:

⁷ InVS : Institute of Health Surveillance

⁸ COM : *Contrat d'objectifs et de moyens*

- establish an order of priority of diseases to monitor in terms of incidence, of prevalence, of severity, of morbidity, of handicap and of mortality;
- define the epidemiological surveillance tools adapted to the task;
- to elaborate, with the professionals concerned and the patients associations, the epidemiological specifications for the centres of reference;
- to contribute methodological expertise to the centres of references, allowing them to fulfil their mission of epidemiological surveillance through:
 - support for the setting up of a collection of consistent and appropriate data, databases, and appropriate statistical methods, with the collaboration of the hospital-based public health services;
 - an organisation adapted to moving information upwards;
 - a logistical assistance for the analysis and synthesis of the collected data;
- to mobilise all existing databases:
 - the recording of death certificates, managed by the *Institut national de la santé et de la recherche médicale* (INSERM)⁹: the “cépiDc¹⁰”;
 - the declarations of *Affections de Longue Durée* (ALD¹¹) that allow certain diseases or groups of rare diseases to be clearly identified;
 - the *Programme Medicalisé des Systèmes d’Information* (PMSI¹²), the files of the *Commissions Départementales d’Education Speciale* (CDES¹³), the *Commissions Techniques d’Orientation et de Reclassement Professionnel* (COTOREP¹⁴) and the databases of laboratories (follow-up of orphan drugs or tracers, data from diagnostic examinations in genetic laboratories...);
- to continue the development of registers of rare diseases by:
 - maintaining the policy of inviting bids for the setting up of rare disease registers, initiated by the *Comité Nationale des Registres* (CNR¹⁵) in May 2004;
 - the creation of a sub-committee of the CNR, which would allow rare disease registers to be designated according to an adapted procedure (modification of the decree defining the missions of the CNR and its internal rules);
- to set up an epidemiological study on mortality due to rare diseases using death certificates;
- to collect data on the level of handicap and the social, educational and professional insertion of patients;

⁹ INSERM : French medical research agency

¹⁰ cépiDC: Epidemiology Centre for Causes of Death. A service which manages the medical information included on death certificates

¹¹ ALD : chronic long-term disorders

¹² PMSI : programme of medical information systems

¹³ CDES: departmental commissions for special needs education

¹⁴ COTOREP: technical commissions for professional orientation and reclassification

¹⁵ CNR: National Committee of Registers

- to produce a synthesis of the epidemiology of rare diseases and to ensure that this is distributed widely, notably to all institutional partners, professionals, associations and the general public.
- **The coordinating structure for research on rare diseases**, born out of the *Groupement d'Intérêt Scientifique* on rare diseases, will promote research projects to develop:
 - generic tools for epidemiology (e.g. database managers);
 - clinical research projects, including therapeutic trials from centres of reference or from patient registers.

Cost

Financial and human means will be specifically allocated to the InVS for this mission: 500,000 euros per year (a total of 2 million euros over the duration of the Plan)

Calendar

New InVS missions: from January 2005

Research programme on rare diseases: 1st trimester 2005

Supervising Body

Direction Générale de la Santé (DGS)¹⁶ in collaboration with the InVS

¹⁶ DGS : Directorate of Health within the Ministry of Health and Social Protection

2 : Recognise the specificity of rare diseases

The recognition of the specificity of rare diseases is a necessary prerequisite to facilitate early diagnosis of the disorder and improvement in the care of patients and their families, factors that can contribute to improved prognosis and quality of life.

Context

Certain patients affected by rare diseases meet with difficulties in getting healthcare reimbursed by the national health insurance. These difficulties arise in four areas:

Differences in procedures for reimbursement in the case of long-term disorders (ALD)

There are differences in procedures for reimbursement because of the absence of a unique “rare diseases” category within the list of long-term disorders that leads to a lack of coherence between the different procedures. Either:

- the pathology or one of its symptoms appears nominally on the list of 30 recognised long-term disorders – the ALD 30 (e.g. hereditary metabolic diseases, cystic fibrosis, amyotrophic lateral sclerosis, neuromuscular disease...);

or

- the pathology is not on this list and the patient is registered as suffering from a “non-listed” disorder (31st ALD) and is reimbursed at 100%. The pathology must be serious, evolving or debilitating and require continuous care for a period greater than 6 months.

Reimbursement of certain specific health products

Certain health products (medicines, medical equipment) prescribed for rare diseases are not reimbursed. They include, notably:

- medicines with a marketing authorisation which are reimbursed under certain conditions but which have been prescribed to treat a non-reimbursable condition as part of the treatment for certain rare diseases (e.g. vitamins for cystic fibrosis);
- medicines which have a marketing authorisation, but which are prescribed to treat a rare disease for a condition that does not appear on the marketing authorisation.

There is a special reimbursement procedure for hereditary metabolic diseases that enables, with expert advice, the establishment of the list of medicines prescribed for conditions not covered within the marketing authorisation that can be reimbursed. However, this procedure is not applicable to other categories of rare diseases.

Reimbursement of transport costs

The national health insurance’s rule specifying that only the cost of transport to the nearest treatment centre will be reimbursed presents an obstacle for the reimbursement of transport costs for certain patients.

The specificity of the patient care required for rare diseases that necessitate visits to centres of reference or of expertise, very often far from the patient’s home, mean that the current procedure for reimbursement of travel costs needs to be adapted.

Differing practices within the medical services of the national health insurance

The national health insurance services' unfamiliarity with rare diseases explains, for the most part, why certain patients have their requests for reimbursement refused. A system of national expertise for hereditary metabolic diseases has been set up by the national health insurance, but this procedure does not meet the needs of other categories of rare diseases.

Objectives

An improvement in the recognition of rare diseases, within the framework of the procedure for long-term disorders, will require the following measures:

- simplifying the procedure for reimbursement of patients recognised as suffering from long-term disorders;
- broadening the reimbursement of certain prescribed items used to treat rare diseases;
- lightening the procedures for the reimbursement of travel costs;
- improving knowledge of rare diseases, and their specificities, within the medical services of the national health insurance, so as to limit as far as possible the unjustified refusal of requests for reimbursement.

Measures

- **The *Haute Autorité de Santé*¹⁷ will be asked to:**
 - deliberate with the aim of ensuring that rare diseases are reimbursed within the framework of the procedure for long-term disorders, when these diseases are serious, debilitating and costly. This deliberation could focus notably on the criteria for inclusion of rare diseases in the long-term disorders procedure, as well on the recognition of the expertise of the centres of reference for rare diseases, so that they can be considered to be the preferential interlocutor for the medical consultants;
 - give an opinion on the pertinence of the reimbursement by the national health insurance of medicines prescribed for conditions not covered within the marketing authorisation or non-reimbursable for treatment of patients with rare diseases. In the case of a positive opinion, the ministerial services will put in place a procedure for the reimbursement of these medicines by the national health insurance when they are prescribed outside the current legal framework;

Over and above this measure, the centres of reference should, in collaboration with the *Haute Autorité de Santé*, progressively put in place national protocols for the diagnosis and treatment of rare diseases, so that medicines and medical products which are essential for rare diseases, including those prescribed outside the conditions covered by the marketing authorisation, can be reimbursed by the national health insurance.

- **Improvement in the reimbursement of transport costs by the national health insurance:**

¹⁷ Higher Health Authority - an independent public authority in France which oversees the scientific evaluation of medical practice and diagnostic and therapeutic procedures and which promotes best practice in the health system

- identification of the centre of reference or of expertise as the nearest treatment centre;
 - a decree being prepared in the *Conseil d'Etat*¹⁸ will abolish the rule concerning the nearest treatment centre and allow reimbursement of travel costs to the centres of reference.
- **Creation of a structure dedicated to rare diseases** within the national framework of medical services in the different divisions of the national health insurance to ensure these services are made aware of the specificity of the reimbursement needs of patients suffering from rare diseases.

Cost

No additional costs identified

Calendar

2005

Supervising Body

*Direction de la Sécurité Sociale (DSS)*¹⁹

¹⁸ *Conseil d'Etat* : the Supreme Court for administrative justice in France

¹⁹ DSS : Social security directorate within the Ministry of Health and Social Protection

3: Develop information for patients, health professionals and the general public concerning rare diseases

Context

For a number of years, media campaigns developed by associations, and in particular, *Téléthon*²⁰, and the *Opération nez rouge*²¹, have increased the French public's awareness of rare and orphan diseases.

However, patients and their families meet with major difficulties in getting information and finding their way within the healthcare system. This leads to diagnostic errors, which alter the quality of their care, and increases their feelings of isolation.

In addition, the information aimed at professionals is dispersed and difficult to access in a context where knowledge is evolving rapidly. They need to have real-time access to validated and updated recommendations for clinical practice, and should be able to identify the specialised services to which they can send patients in their care when necessary.

The availability of validated information, which is pertinent and easy to access, is therefore essential to improve patient care.

Sources of information on rare diseases do exist but they need support for their development:

- Orphanet is a multilingual Internet-based information server, created in 1997, and financed by the *Direction Générale de la Santé*, the *Caisse Nationale d'Assurance Maladie des Travailleurs Salariés (CNAMTS)*²², INSERM, the European Commission, *Les Entreprises du Médicament (LEEM)*²³, the *Fondation Groupama pour la santé*²⁴ and the associations, notably the *Association Française contre les Myopathies*. This server provides families and professionals with information on rare diseases and on the related services available.
- Two specific telephone information services, *Maladies Rares Info Service* and the *Fédération des Maladies Orphelines (FMO)*²⁵, provide information to patients and health professionals on a daily basis but are not sufficiently well known.
- Educational documents and educational therapeutic tools are being developed by specialists and patients associations.
- Socio-cultural information relays which inform patients who have linguistic or social difficulties have been put in place, though these initiatives are not sufficiently widespread (e.g. the women-run "information relays" in hospitals which inform families about sickle cell anaemia).

²⁰ Téléthon : an annual fundraising event organised by the AFM which runs over 30 hours and is covered by the French television channel, France 2.

²¹ Opération nez rouge : a fund-raising and awareness-raising campaign organised by the *Fédération des Maladies Orphelines (FMO)*

²² CNAMTS: national health insurance fund for salaried employees

²³ LEEM : French pharmaceutical industry association

²⁴ Fondation Groupama: a benevolent foundation set up by Groupama insurance company

²⁵ FMO: Federation of Orphan Diseases

Objectives

- To develop information aimed at all publics concerned
- To provide certification for the telephone information services
- To develop therapeutic education
- To make the Plan for Rare Diseases known to professionals and the general public

Measures

- **To improve information available on Internet in the French language, by:**
 - making Orphanet the reference portal giving access to all information on rare diseases;
 - creating new services for health professionals (diagnostic aids or help with orientation within the health system; guidelines to adopt in emergency situations...) and for social partners (help with administrative processes for educational or professional integration, handicap compensation, technical or financial help...);
 - developing the existing medical information by the extension of Orphanet's medical encyclopaedia aimed at professionals and patients: detailed information will be available for all identified rare diseases;
 - developing new categories of information adapted to the needs of patients and professionals including:
 - the organisation of health systems and of patient care in France and in the rest of the world, particularly in Europe;
 - patient care and access to therapeutics and to state sickness benefits
 - best practice for treatment
 - available educational documentation
 - systems of expert assistance for diagnosis
 - therapeutics and their development
 - making Orphanet better known to health professionals and patients through targeted communication campaigns.
- **To improve the information on rare diseases available by telephone, in collaboration with the *Institut National de Prévention et d'Éducation pour la Santé (INPES²⁶)* and the patients associations by:**
 - certifying the telephone information services;
 - making the telephone services better known through communication campaigns aimed at health professionals, patients and the general public.
- **To develop specific information on rare diseases adapted to the needs of certain publics, particularly teachers, social workers...**
- **To develop therapeutic education by:**

²⁶ INPES: National Institute for Health Education and Prevention

- improving the quality, the quantity, the availability and the accessibility of the educational documents used for therapeutic education in association with the INPES, health professionals and patients associations;
 - bringing together and sharing experiences of health education, under the aegis of the INPES;
 - improving information aimed at different publics with specific difficulties (linguistic or social problems, those in precarious situations) by training health professionals working in the community (doctors and nursery nurses in child healthcare centres, school doctors...);
 - supporting the Education Ministry's online database "Intégrascot", which provides information on rare and chronic diseases to teachers, in association with school doctors;
 - promoting research on health education.
- **Make the Plan for Rare Diseases known to health professionals, patients and the general public by:**
- spreading information on the measures to health professionals, patients, the general public and disability centres;
 - making the Plan known at the international and particularly European level;
 - organising, in collaboration with the European Union and patients associations, an international conference in Luxembourg in 2005;
 - ensuring the follow-up of information concerning the Plan's implementation and the evaluation of its measures by health professionals, patients and associations.

Calendar

Orphanet: 2005 – 2007

Therapeutic education: 2005 – 2007

Telephone information service: 2005

Cost

Information measures: 300,000 euros per year (1.2 million euros over the duration of the Plan);

Telephone information service: 400,000 euros per year (1.6 million euros over the duration of the Plan).

Supervising body

Direction générale de la Santé (DGS), in association with the INPES, INSERM and patients associations.

Examples of new tools to be developed:

- a directory of services that will help guide patients through the healthcare system based on a broad description of their clinical symptoms when no precise diagnosis has been given;
- online up-to-date information on available healthcare (centres of reference, care networks...);
- a bibliographic survey of the organisation of healthcare and patient care for rare diseases across the world;
- online information on best practice developed by healthcare professionals, notably the centres of reference, in association with the *Haute Autorité de Santé* ;
- online tools for diagnostic aid and help with decision-making in emergency situations in association with the professionals concerned, the *Haute Autorité de Santé* and the associations;
- online information on the development and the availability of orphan drugs in association with the AFSSAPS and the LEEM;
- online downloadable educative and informative documents for patients and their families;
- online regularly updated social and administrative information.

4: Train health professionals to better identify rare diseases

Context

Patients frequently complain of diagnostic error, and the problems they and their families encounter in the continuity of care and in day-to-day support.

Patients associations underline the importance of the way information is provided, as much as its content, in helping patients and their families cope with the disease: the announcement of the diagnosis, the reception in emergency situations, the care provided throughout the illness to help ensure an adapted social insertion, periods of disease aggravation or the end of life, all require a quality accompaniment by health professionals.

The programme of study in medical training doesn't mention rare diseases.

The circulation of medical information concerning the patient within the healthcare system is sometimes difficult, notably in emergency situations: faced with insufficient reliable medical information on the diagnosis and care of the disease, a health professional in the emergency services may find it difficult to take the appropriate therapeutic steps (e.g. osteogenesis imperfecta, Marfan syndrome...)

Objectives

- To adapt initial training and continuing education for health and social sector professionals
- To recognise new professions which could help improve the care of patients and their families
- To improve the circulation of medical information

Measures

- **To adapt initial medical training by:**
 - introducing the theme of rare diseases into the national classifying end of study examination programme in 2nd and 3rd cycles of medical studies;
 - introducing the theme of rare diseases into the training programmes of specialists in collaboration with the training establishment of the specialisation concerned;
 - setting up seminars to raise awareness of rare diseases during the 3rd cycle of medical studies;
 - creating a teaching module specifically on rare diseases in the framework of the "virtual medical university";
 - developing specific educational tools and putting them online on Orphanet.
- **To organise continuing medical education, obligatory since the 9 August 2004 public health policy law, in the area of rare diseases, by:**

- developing continuing education modules in association with the *Collège national de la formation médicale continue des médecins libéraux*²⁷, the *Collège des enseignants de médecine générale*²⁸, the *Collège national de la formation médicale continue des médecins salariés*²⁹, the learned societies concerned, and the *Conseils nationaux de la formation continue des médecins hospitaliers*³⁰;
 - putting the training programmes online on Orphanet.
- **To raise awareness of the problems concerning rare diseases amongst other health and social sector professionals by:**
- modifying the programmes of initial training and continuing education of paramedical personnel: nurses, therapists (ergotherapists, speech therapists, psychomotor therapists) social workers, psychologists...
- **To organise a training programme for new professions by:**
- setting up a programme of initial training for genetic counsellors and include issues relevant to rare diseases (announcing the diagnosis, patient care, accompaniment of patients and their families, ethical reflection);
 - setting up a complementary university course (inter-university diploma, masters) for “care coordinators” aimed at health professionals working with patients with rare diseases.
- **To improve circulation of medical information** concerning the patient, whilst respecting the principles of medical and professional secrecy, the *dossier médical personnel* (DMP³¹) is the best adapted means.

Cost

100,000 euros per year (400,000 euros during the duration of the Plan)

Calendar

Development of training: 2005 – 2006

Tools for the circulation of medical information: 2005 – 2006

Supervising body

Direction générale de la Santé (DGS)

²⁷ National college of continuing medical education for doctors

²⁸ College of teachers of general medicine

²⁹ College of continuing medical education for salaried doctors

³⁰ National councils of continuing education for hospital doctors

³¹ DMP: personal medical file

5: Organise screening and access to diagnostic tests

Context

The large number of genetic tests now available, thanks to the rapid growth of molecular biology and the progress made in genetics, makes it possible to diagnose more and more rare diseases.

The availability of these tests also makes it possible to envisage their use for systematic screening of the population.

Organised population screening (prenatal, neonatal or adult screening) or targeted screenings depend upon public health and social choices where many issues are at stake:

- technical: the tests available need to be feasible, accurate, predictive and acceptable to those taking them;
- ethical: the taking of the test and its consequences need to be beneficial for the person being tested and the familial and social implications must be ethically acceptable;
- organisational: people screened within the targeted population must be able to be cared for. The choice of a screening policy poses the problem of the health priorities that society is ready to accept;
- societal: choosing to screen the population is also a societal choice. Society can choose to promote screening, over and above purely public health considerations, as an act of solidarity with a group of patients that it wants to support.

Only an evaluation of all screening, before and after it is carried out, and taking into account the totality of these issues, makes it possible to propose a coherent policy of screening for rare diseases.

Individual screening depends upon a personal decision being taken in the context of a patient's overall healthcare and where the appropriate indications have been validated. Furthermore, the self-tests available through Internet and which don't require any medical prescription do not currently have any scientific validation nor legal framework and pose many ethical problems.

Objectives

- To put in place a coherent policy for the screening for rare diseases, founded on clearly defined priorities based on a rigorous evaluation of the screenings that are envisaged and on a rationalisation of the decisions which takes into account society's choices.
- To improve access to diagnostic tests.
- To propose a concerted policy in Europe on the screening for rare diseases.

Measures

1. *To provide a framework for screening for rare diseases:*

➤ **Improve the organisation of screening programmes in the general population by putting in place:**

- a general regulatory procedure which defines the stages of evaluation for the implementation and follow-up of screening programmes;
- a systematic evaluation before and after screening programmes in association with the bodies concerned (*Ministère de la Santé et de la Protection Sociale, Haute Autorité de Santé, Haut Comité de Santé Publique*³², sanitary agencies...). A guide to the methodology of *a priori* evaluation of a screening programme³³ has recently been developed by the *Agence Nationale d'Accréditation et d'Évaluation en Santé* (ANAES³⁴);
- an independent consultative committee, backed by the *Haute Autorité de Santé* and by the *Haut Conseil de Santé Publique*³⁵, instructed to give an opinion to the Minister for Health and Social Protection on screening policy;
- training and promotion of teams which could contribute their competences to the development of evaluation of programmes via a call for projects;
- a legal and institutional framework for screening programmes and individual screening with:
 - clarification of the legal status of the act of prescribing a screening test;
 - including screening in the nomenclature of medical acts.

➤ **Improve the organisation of individual screening by:**

- validating the indications of screening tests for individual screening aimed at identifying a health risk for a person and their family, in association with the *Agence française de sécurité sanitaire des produits de santé* (AFSSAPS), the *Haute Autorité de Santé*, the *Conseil National Consultatif d'Éthique* (CNCE)³⁶, the *Agence de Biomédecine*³⁷ and the health professionals concerned;
- providing scientific validation for self-tests, in association with the *Haute Autorité de Santé*, the health professionals and the patients associations;
- clarifying the ethical, legal and regulatory status of self-tests for screening.

II. *To improve the access to diagnostic tests and genetic counselling:*

➤ **Improve the availability of diagnostic tests**, by launching an invitation for bids in 2005, aimed at laboratories that undertake complex biological procedures, notably in molecular

³² Higher Committee on Public Health

³³ “*Guide méthodologique: comment évaluer a priori un programme de dépistage, guides pratiques*”

[Hhttp://www.anaes.fr](http://www.anaes.fr)H

³⁴ ANAES: National Agency of Health Accreditation and Evaluation

³⁵ Higher Council for Public Health

³⁶ CNCE: National Consultative Committee on Ethics

³⁷ Biomedicines Agency

biology, in collaboration with the *Comité National de Génétique Clinique*³⁸ and the appropriate learned societies.

A call for projects was undertaken in 2003 to facilitate the development of particularly complex genetic tests for the diagnosis of rare muscular, neurological and neurosensory diseases and mental retardations.

Thirty-four molecular genetics laboratories currently receive an annual financial support of 4.5 million euros.

- **Improve the accessibility to medically assisted reproduction techniques** within the context of a pre-implantatory diagnosis, by making them exempt from the “*ticket modérateur*”³⁹.
- **Raise awareness amongst health professionals and the public about screening tests** and how they are set up (INPES) by:
 - carrying out information and communication campaigns on screening;
 - organising training for health professionals on screening and the ethical and public health problems it raises.

III. To propose a concerted European policy on screening for rare diseases

- Call on the European Commission’s Directorate General on Health (DG SANCO), and notably the “Rare Diseases Task Force”, **to formulate European recommendations.**

Cost

Call for bids for reference laboratories: 5 million euros per year, starting in 2005 (20 million euros over the duration of the Plan).

Calendar

2005 - 2006

Supervising bodies

Supervision of screening: *Direction générale de la Santé* (DGS) in collaboration with the INEPS

Improvement in access to diagnostics: *Direction de l’hospitalisation et de l’organisation des soins* (DHOS⁴⁰) and *Direction de la Sécurité Sociale* (DSS)

³⁸ National Committee for Clinical Genetics

³⁹ *ticket modérateur*: the part of the cost of medical treatment which is not reimbursed by the national health insurance

⁴⁰ DHOS: Directorate of hospitalisation and organisation of treatment – within the Ministry of Health and Social Protection

6: Improve access to treatment and the quality of patient care

Context

The number and the diversity of rare diseases necessitates a specific approach to patient care. Patients and their families meet with difficulties in getting a diagnosis and in getting suitably adapted, coordinated care for their condition.

The access to treatment is often difficult because of:

- patients', and their doctors', unfamiliarity with the pathology and ignorance of the existence of specialised centres for these diseases;
- the multiplicity of actors necessary for the care of the disease;
- the insufficiencies of the community-hospital interface which facilitates global and coordinated patient care by the health and socio-medical networks;
- the absence of clear identification of community-based medical and paramedical centres.

The routing of patients is therefore often the result of individual logic rather than a truly organised care system.

The result is inadequate patient care and diagnostic wavering, particularly painful for families and sometimes the cause of a delay in diagnosis.

Objectives

- To put in place centres of reference for the care of patients with rare diseases.
- To create a structure for the organisation of patient care, starting with these centres, which allows patients to benefit from care that is the best adapted to their needs.
- To ensure coherence between the organisation of patient care with the procedures for reimbursement by the national health insurance and those for the attribution of other types of benefit.
- To ensure the availability and reimbursement of orphan drugs and prevent the halt in commercialisation of certain health products.

Measures

I. To put in place centres of reference for rare diseases:

- Create a **Comité National Consultatif de Labellisation (CNCL⁴¹)** made up of representatives of health professionals with expertise in rare diseases, institutions and of patients associations. This committee's mission will be to advise the Minister of Health and Social Protection on the certification of the centres of reference;

⁴¹ CNCL: National Consultative Committee for Certification

- Give national certification to around 100 centres of reference for rare diseases. **These centres, made up of multidisciplinary teams, have the following missions:**
 - to facilitate diagnosis and define a strategy of therapeutic and psychological care and of social accompaniment;
 - to define and circulate care protocols, in association with the *Haute Autorité de Santé* and the *Union Nationale des Caisses d'Assurance Maladie* (UNCAM⁴²);
 - to coordinate research and participate in epidemiological surveillance, in association with the *Institut de Veille Sanitaire* (InVS)
 - to participate in training and information initiatives for health professionals, patients and their families, in association with the *Institut National de Prévention et d'Education pour la Santé* (INPES);
 - to manage and coordinate the networks of health and socio-medical care providers
 - to be the main interlocutors for the ministries and patients associations.
- **To ensure wide circulation of the list of centres of reference** to patients and their families in order to facilitate the routing of patients to certified centres. The geographical directory of these centres will be put online on the websites of the Ministry of Health and Social Protection and Orphanet. It will also be publicised to patients associations and health professionals.
-
- **To ensure permanent funding for these centres**, on the one hand through an “activity rate” (T2A⁴³) and on the other hand through a fixed allowance under the “*Missions d'intérêt general et d'aide à la contractualisation*” (MIGAC⁴⁴) for those resources consecrated to the missions of the coordination of care, research and teaching, and of epidemiological surveillance and expertise.

II. *To structure the organisation of care:*

- **To create health and socio-medical networks** starting with the centres of reference by:
 - getting rare diseases recognised by the regional agencies (*Agence Régionale d'Hospitalisation*, ARH⁴⁵ and *Union Régionale des Caisses d'Assurance Maladie*, URCAM⁴⁶) as a priority for funding of care networks emanating from the centres of reference;
 - supporting the development of these inter-regional and national networks through the *Dotation Nationale de Développement des Réseaux* (DNDR⁴⁷) on condition that a partnership with a centre of reference exists.

⁴² UNCAM: National union of national health insurance funds

⁴³ T2A: *tarification à l'activité*

⁴⁴ MIGAC: Missions of general interest and assistance with contractualisation

⁴⁵ ARH: Regional hospital agency

⁴⁶ URCAM: Regional union of health insurance funds

⁴⁷ DNDR: National Allowance for the Development of Networks

- **To develop tools for coordination within the care networks by:**
 - encouraging the institutions which have a centre of reference to set up inter-hospital agreements between the services involved in the care of patients with rare diseases. These agreements should be based on communal medical projects;
 - experimenting with personal medical files (DMP) from 2005 onwards, in one or two centres of reference;
 - paying for the transport costs of hospital practitioners who have to travel to another institution as part of their work as advisors or experts.
- III. *To ensure coherence between the organisation of patient care, the procedures for reimbursement by the national health insurance and socio-medical support.*
- **Identify the centres of reference as sources of expertise** for the medical control services of the national health insurance;
 - **Ensure these centres fulfil an information role** for patients and their families with respect to the different procedures for support and social accompaniment.

Calendar

From now until 2008 more than 100 centres of reference for rare diseases will be certified.

Cost

40 million euros over the duration of the Plan:

10 million euros in 2004
 10 million euros in 2005
 10 million euros in 2006
 10 million euros in 2007

Supervising bodies

Certification of the centres: *Direction de l'hospitalisation et de l'organisation des soins* (DHOS) and *Direction générale de la Santé* (DGS)

For orphan drugs: DHOS, DGS and the *Mission des médicaments orphelins*⁴⁸

Social and socio-medical support: *Direction générale de l'action sociale* (DGAS⁴⁹)

⁴⁸ Orphan drugs mission

⁴⁹ DGAS: Directorate of social action, part of the Ministry of Health and Social Protection

**Call for projects for the certification of centres of reference for rare diseases
(Ministry note DHOS/DGS 27 May 2004)**

As a continuation of work already done on support for cystic fibrosis and amyotrophic lateral sclerosis, a scheme for the certification of centres of reference for the care of patients with rare diseases has been launched. The call for projects provides for the certification of centres of reference by pathology or by group of rare pathologies.

A centre of reference for a rare disease or a group of rare diseases brings together a group of multidisciplinary hospital-based competences, organised around highly specialised medical teams. Its missions are detailed in the call for projects.

A **Comité National Consultatif de Labellisation (CNCL) for the centres of reference**, associating health professionals with expertise in the care of rare diseases, representatives of patients associations and of institutions, nominated by the Minister of Health and Social Protection, will advise the Minister on the certification of the centres. It can be called on to advise on any question relative to the organisation of care with respect to rare diseases.

The certified centres will undergo an evaluation procedure which will be determined in association with the *Agence d'accréditation et d'évaluation en santé* (ANAES).

7: Continue efforts in favour of orphan drugs

Context:

The criteria for the designation of an orphan drug is based on the prevalence of the disease which it is supposed to treat: 5/10,000. It is accorded by the Committee for Orphan Medicinal Products of the European Medicines Agency (EMA).

A ministerial mission for orphan drugs was created in March 1995 with two objectives:

- to promote a European policy in favour of orphan drugs;
- to accompany this project with appropriate national measures.

This mission achieved its main objective because it led to the adoption of the European regulation on orphan drugs on 16 December 1999.

This new regulation, operational within the European Medicines Agency since April 2000 has allowed the designation⁵⁰ of **240 drugs** and the attribution of a European marketing authorisation to **16 health products**, with another 20 or so requests for marketing authorisations currently being examined.

This regulation, and the measures of the ministerial mission, have facilitated the **creation of new pharmaceutical companies** and boosted the existing companies. In France, more than 30 companies are involved in the development of orphan drugs.

In parallel, the *Assistance Publique - Hôpitaux de Paris (AP-HP⁵¹)* has created the *Etablissement Pharmaceutique des Hôpitaux de Paris⁵²* to enter orphan drugs on the market.

Objectives

To pursue the dynamic development of new orphan drugs through appropriate measures.

Measures

- The European Commission will present an assessment of the orphan drugs regulation at the beginning of 2006. This assessment may lead to the re-examination by the European authorities of the measures adopted in 1999.

The French government, deeply attached to the preservation of the existing measures, will follow this assessment procedure with extreme vigilance to avoid any risk of the special regulations in favour of orphan drugs being called into question.

- **The current exemption** for the promoters of orphan drugs, with respect to **taxes and other payments due** by the pharmaceutical industry and destined for the national health insurance and the AFSSAPS, will be continued;

⁵⁰ Designation: procedure by which the Committee for Orphan Medicinal Products accepts that the presented project for drug development is aimed at a rare disease (prevalence inferior to 5 in 10,000) and that this disease is serious and debilitating and that no other treatment is currently available.

⁵¹ AP-HP: Parisian public sector hospitals

⁵² Paris Hospitals' Pharmaceutical Establishment

- The *Comité Economique des Produits de Santé* (CEPS⁵³) has included a section on orphan drugs in its “*accord sectoriel*”⁵⁴ **between the pharmaceutical industry and government**. This initiative will be continued in any future policy contracts between industry and the government;
- The development of orphan drugs is closely associated with research supported by the **hospital programme for clinical research**. In keeping with the measures described under the “Research” section of this Plan, rare diseases will remain a specific thematic priority in this research programme;
- Orphan drugs will be **included on the list of innovative and expensive health products** in the context of the reform of the “activity rate” (measure described in the Plan under access to care). This initiative should allow the distribution of orphan drugs as soon as they have their marketing authorisation. It necessitates an update of the list of products concerned to avoid any delay in the access to innovative therapeutics;
- The *Conseil de l’Hospitalisation*⁵⁵ should advise the Minister in charge of health on the **health products reserved for hospital use without marketing authorisations** which could be financed out of the hospitals budget (ONDAM *hôpital*⁵⁶);
- **The untimely halt to the commercialisation of certain health products prescribed and useful for rare diseases will be prevented**. This measure will be put in place in 2005 by the *Mission des médicaments orphelins* in association with the *Comité Economique des Produits de Santé* (CEPS) and the *Agence Française de Sécurité Sanitaire des Produits de Santé* (AFSSAPS);
- **The “*autorisation temporaire d’utilisation*” (ATU⁵⁷) scheme, particularly advantageous in France, will be continued**. This scheme allows drugs which don’t have a marketing authorisation, and which are presumed to be efficient and of an acceptable level of safety, to be made available to patients with rare diseases.

Cost

No extra costs identified.

Calendar

Early 2006: assessment by the European Commission of the application of the 1999 regulation on orphan drugs;

Early 2005: measures to prevent the untimely halt to the commercialisation of drugs that are useful for rare diseases.

Supervising bodies:

Mission des médicaments orphelins, the *Agence Française de Sécurité Sanitaire des Produits de Santé* (AFSSAPS), the *Direction de l’hospitalisation et de l’organisation des soins* (DHOS) and the *Direction générale de la Santé* (DGS)

⁵³ CEPS: Economic Committee on Health Products

⁵⁴ Industrial agreement

⁵⁵ Council for Hospitalisation

⁵⁶ ONDAM: *Objectif national des dépenses d’assurance maladie* – mandatory national health insurance spending target

⁵⁷ ATU: temporary authorisation for use

8: Respond to the specific accompaniment needs of people suffering from rare diseases

Context

Rare diseases can bring about many types of handicap, some of them very severe. In addition, the rarity of the disease results in difficulties in diagnosis, uncertainties regarding prognosis and ignorance amongst social and socio-medical staff about many aspects of the disease, which can provoke a delay in assistance being sought.

A certain number of difficulties have been identified:

- poor knowledge of rare diseases amongst professionals (doctors, paramedics, social workers...);
- poor knowledge of the means and structures available, notably in the field of disability, by users and professionals;
- absence of a systematic meeting with the patient (or their family, in the case of a child), during the first request made to the departmental evaluation commissions. This meeting is of importance to the families who want to be able to present the particularities associated with the disease. On the other hand, the procedures for renewal are considered too heavy and too frequent;
- poor reactivity of the support services in emergency situations or when a sudden aggravation of the disease occurs;
- inappropriateness of the current responses and compensatory benefits with respect to the human and technical support needs of patients;
- insufficient psychological accompaniment of the patient and their family when the diagnosis is announced and throughout the illness;
- lack of knowledge of the mediation role that the school doctor can play between the family and the school;
- long distances between care centres and place of residence, with accommodation costs charged to the families.

Objectives:

- To provide an appropriate response to the specific accompaniment needs of patients with rare diseases and their families by:
 - improving patients', associations' and professionals' access to information on available support;

- structuring the links between the *maisons départementales des personnes handicapées*⁵⁸ and the care networks for people suffering from rare diseases, in particular around the centres of reference;
- supporting the patients associations in their mission of information provision.

The improvement in the accompaniment and the care of people suffering from those rare diseases leading to invalidity are addressed in the bill on equal rights and chances, participation and citizenship for handicapped people. This bill provides for the creation of *maisons départementales des personnes handicapées*, as well as the development of socio-medical accompaniment for handicapped people, particularly those suffering from rare diseases. The government will be particularly vigilant to ensure that the bill takes into account the specific difficulties of people suffering from rare diseases and with this aim is proposing the measures outlined in the present Plan.

Measures

➤ **To develop information sources for people, users and professionals by:**

- making known to patients, families and associations the different sources of information on support measures: ministerial websites, specialised telephone information services;
- continuing and reinforcing the support for patients associations in their mission of information provision;
- giving the centres of reference a mission to provide information on different support and accompaniment measures for patients and their families;
- training and raising awareness amongst doctors working in *Protection Maternelle and Infantile* (PMI⁵⁹) and *Centres d'Action Medico-Sociale Précoce* (CAMSP⁶⁰), school doctors, and occupational doctors, of the problems associated with rare diseases.

➤ **To structure links between the *maisons départementales des personnes handicapées*, centres of reference and care networks for people suffering from rare diseases by:**

- ensuring a strong relationship between the centres of reference and the *maisons départementales des personnes handicapées* so that the multidisciplinary technical teams based there can have access to the information necessary for the evaluation of patients' needs;
- creating health and socio-medical networks emanating from centres of reference;
- organising a scheme which would allow accommodation and transport costs of patients and their families to be covered for consultations at centres of reference when these are far from the patient's home.

➤ **To integrate the specificities of rare diseases and the personal needs of patients in the elaboration of projects on support and accompaniment by:**

⁵⁸ regional centres for handicapped people

⁵⁹ PMI: Infant and child healthcare centres

⁶⁰ CAMSP: early years socio-medical action centres

- organising psychological care for patients and their families in the hospital structures where diagnosis and the announcement of the diagnosis take place;
- improving information for doctors and other members of the technical teams in the future *maisons départementales des personnes handicapées*, by facilitating their access to information sources (Orphanet) and by including rare diseases in their training, notably in the use of evaluation tools (e.g. using a person with a handicap linked to a rare disease as a case study for training on the “*guide barème*”⁶¹);
- taking the medical assessment of the centre of reference into account in the evaluation of the consequences of a rare disease for the elaboration of personalised compensation plans, as defined in the bill on equal rights and chances, participation and citizenship for handicapped people;
- by identifying professionals with expertise in rare diseases amongst staff in the *maisons départementales des personnes handicapées*.

All of these measures should together help to change patients' experience of social support and accompaniment from an obstacle course into a chosen project for life.

Cost:

No extra costs identified

Calendar:

2005-2007

Supervising body

Direction Générale de l'Action Sociale (DGAS)

⁶¹ a disability measurement scale

9: Promote research on rare diseases

Context

Through a better understanding of the physiopathological mechanisms implicated in rare diseases, an improvement in diagnostic methods leading to earlier disease detection, and the development of more efficient treatments, research generates the hope of delivering improvements in patient care. It is therefore a priority.

However, the large number, low prevalence and heterogeneity of rare diseases makes the development of research difficult, implicating a multidisciplinary approach bringing together teams working in clinical, genetic, physiopathological, therapeutic and social sciences research, as well as the optimal use of existing tools and technological platforms.

Although many new initiatives in research on rare diseases have been launched over the past few years (emergence of multidisciplinary research networks, creation of the GIS *Institut des Maladies Rares...*) mainly thanks to the work of patients associations, and notably the *Association Française contre les Myopathies* (AFM), it is still necessary to reinforce these measures.

Objectives

The main objective is to improve research on rare diseases by putting in place a number of measures which aim to:

- promote a voluntarist research policy, particularly in the domain of clinical trials;
- ensure appropriate coordination of researchers' work within a specific research programme, which will be created in accordance with the Ministry of Research;
- allow this programme to develop incentive measures in partnership with institutions, industry and associations.

Measures

- **Reinforce the rare diseases priority in the *Programme hospitalier de recherche clinique* (PHRC⁶²) :**

Every year, the PHRC, managed by the Ministry of Health and Social Protection (DHOS), provides an opportunity for the development of research concerning thematic priorities within health institutions, via a call for projects.

Rare diseases have been included in the themes of the PHRC since 2001. However, up until now only those research projects consisting of clinical trials were considered for funding.

From 2005 onwards, rare diseases will be considered as a specific thematic priority of the PHRC, allowing research projects other than only clinical trials, such as impact studies of diagnostic and therapeutic strategies or of care practices on patients' state of health or quality of life, to be funded. The number of research projects on rare diseases supported by hospital institutions should therefore rise considerably.

⁶² PHRC: hospital programme for clinical research

- **Ensure the coordination of research on rare diseases within the research programme financed by the *Agence Nationale de la Recherche*⁶³ in association with INSERM, and with partners from the institutions, associations and industry.**

This programme, which will replace the *GIS maladies rares*, will give more coherence and a greater visibility to the national research policy on rare diseases, both in France and internationally. The programme will be linked to INSERM and will benefit from flexible management methods. Its organisation will be defined with the different partners within the framework of the *Agence Nationale de la Recherche*'s incentive policy.

This research programme, which will run over a number of years, will launch a call for bids concerning different aspects of research into rare diseases.

- **Develop measures within this programme, over a number of years, via a call for bids in six research domains:**
- *Epidemiology (multidisciplinary networks in association with the centres of reference)*
Promotion of an underdeveloped area of research in descriptive and analytical epidemiology in the domain of disease natural history and clinical nosology.
 - *Genetic and molecular characterisation of rare diseases*
The research effort in this domain, essential for the development of diagnostic tests or as an approach to the study of the physiopathology of rare diseases, will be continued.
 - *Physiopathology*
Research into physiopathology will be reinforced because it is essential to understand the mechanisms implicated in the development of the diseases and the phenotypes observed in patients.
 - *Development of diagnostic tests*
The improvement in diagnostic performance in the area of rare diseases is a major challenge and necessitates the development of new methods.
 - *Therapeutic research*
The discovery of new therapeutics, particularly gene and cell therapies, is the most long-awaited objective of research into rare diseases. The diversity of pathological situations, coupled with the lack of knowledge of the physiopathology of a large number of rare diseases and the relative lack of interest of the pharmaceutical industry, makes evident the complexity of this area of research and the need for a very wide variety of approaches.
 - *Evaluation of the performances of treatment and care systems and the psychosocial consequences of rare diseases*
Teams of researchers in human and social sciences wishing to be involved in this area of research will be supported.

Cost

Ministry of Health and Social Protection

PHRC: 22.5 million euros over the duration of the Plan

⁶³ National Research Agency

Support for the programme of research: 150,000 euros per year (600,000 euros over the duration of the Plan)

Ministry of Research

5 million euros (20 million euros over the duration of the Plan)

Calendar

2004 -2008

Supervising bodies

Direction de l'hospitalisation et de l'organisation des soins (DHOS) and the Direction générale de la Santé (DGS)

Ministry of Research and the *Agence Nationale de la Recherche*, in association with INSERM and the patients associations, particularly the *Association Française contre les Myopathies (AFM)*.

10: Develop national and European partnerships

Context

The implementation of the different measures in the Plan for Rare Diseases calls upon many partners. It cannot happen without integration into the European policy that France largely helped to initiate. The European dimension of the solutions that can be brought to patients is particularly evident in the areas of epidemiology, the development of diagnostic tests, the certification of clinical and biological centres of reference, information and research.

France has taken a number of initiatives at the European level over the last few years. It has a duty to continue to support all European measures which could bring about an improvement in the state of health of people with rare diseases.

Objectives

- To further the national coordination of all partners and notably the patients associations.
- To develop a European coordination for rare diseases.

Measures

To maintain and develop the mission of the *Plate-forme Maladies Rares*⁶⁴, in particular by hosting all partners in a single location. This platform, created in October 2001, at the initiative of the *Association Française contre les Myopathies* (AFM) - its main source of funding - is co-financed by the *Direction générale de la Santé* (DGS), the *Union Nationale des Caisses d'Assurance Maladies* (UNCAM) and INSERM. It brings together different actors:

- the *Alliance Maladies Rares*, a French collective of 135 patients associations;
 - EURORDIS, a European collective of more than 200 rare disease associations, including 10 national alliances, from 16 countries;
 - *Maladies Rares Info Service*, an information, listening and orientation service;
 - Orphanet, an online information server on rare diseases and orphan drugs;
 - the *GIS - Institut des Maladies Rares* which coordinates and promotes research into rare diseases.
- **To contribute to the coordination of the different associations at the national level.**
 - **To develop European-level coordination in the following areas:**
 - availability of biological tests for very rare diseases and the promotion of a quality control for these tests;
 - certification of clinical and biological centres of reference;
 - funding of research into rare diseases;

⁶⁴ Rare Diseases Platform

- development of public health indicators to allow evolutions in the state of health of people suffering from rare diseases to be followed;
- sharing of the experiences of professionals and associations in the care of patients.

Calendar

Plate-forme Maladies Rares: 2005 – 2006

Coordination of European research programmes: 2005 – 2008

Cost

40,000 euros per year (160,000 euros over the duration of the Plan) to support the *Plate-forme Maladies Rares*

Supervising bodies

Direction de l'hospitalisation et de l'organisation des soins (DHOS) and the Direction générale de la Santé (DGS)

The *Association Française contre les Myopathies (AFM)* is currently the main funding source for the *Plate-forme Maladies Rares*. A financial contribution of 40,000 euros per year starting in 2005 is planned.

Monitoring the Plan

A **comité de suivi des maladies rares**⁶⁵ has been created to follow up the implementation of measures outlined in the National Plan, under the auspices of the Ministry of Health and Social Protection and in association with the different partners, notably the patients associations.

An evaluation of the proper distribution of the funds allocated to different establishments in the Plan will be undertaken regularly by the *Direction de l'hospitalisation et de l'organisation des soins* (DHOS) in collaboration with the *Agences Régionales d'Hospitalisation* (ARH).

The committee will also be responsible for evaluating the impact of the different measures and for making propositions.

⁶⁵ monitoring committee for rare diseases