National Plans for Rare Diseases: the French experience

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France is the only country in the world to have adopted a national plan with a specific budget to start solving the many challenges raised by the rarity of thousands of rare diseases. Rare diseases (RD) were included as one of the five major priorities in the 9 August 2004 law relating to public health policy. The national plan was elaborated through working groups comprised of health professionals, patient representatives and policy makers. It was published on 20 November 2004 as a four year plan with a budget of 100 millions Euros.
The overall aim was to ensure equity in access to diagnosis, treatment and provision of care for people suffering from a RD through ten strategic objectives:

1- The first objective was to increase the knowledge about RD through an epidemiological surveillance, to allow a better understanding of the natural history of these diseases, an evaluation of the needs, the progress of patients within the health system, and a monitoring of the evolution of indicators relative to the quality of life of patients. To meet this objective a national committee for rare diseases registries was established. It launched recently a call for proposals to identify, certify and support registries in the field of RD.
2- The second objective was to recognise the specificity of RD within the framework of the procedure for long-term disorders. The specific objectives were to simplify the procedure for reimbursement of patients; to broaden the reimbursement of certain prescribed items used to treat RD; to reduce the procedures for the reimbursement of travel cost to attend expert clinics; to improve the knowledge of professionals working in the national health care insurance system. These goals have been partially reached and the result is already assessed as satisfactory, although progress remains to be made in the future.
3- The third objective was to provide accurate information for patients and health professionals through the development of services at Orphanet, through the certification of help lines, and through the dissemination of the information on this national plan. As a result of the national plan's mandate, Orphanet was able to develop review articles in French for the lay public on over 100 RD, create emergency guidelines, and publish emergency cards developed by the Ministry of Health online. A new version of the website has been developed with many additional services. Help lines have also attained professional standards.
4- The fourth objective was to improve the general training of health care professionals and to recognise new professions which could help improve care of patients and their families. Two hours of training on RD have been added to the curriculum of medical studies. The profession of genetic counsellor has been formally established and the corresponding training has been implemented. The position of Coordinator of Centres of Reference has also been formally recognised.
5- The fifth objective was to put in place a coherent policy for the screening of RD. This policy was founded on clearly defined priorities based on a rigorous evaluation of the envisaged screening and on a rationalisation of the decisions taking into account society's opinion. This is the only objective for which no concrete measures have been taken so far.
6- The sixth objective was to improve access to treatment and the quality of patient care through the implementation and funding of centres of reference, and to ensure the availability and reimbursement of orphan drugs and prevent the delay in

commercialisation of certain health products. A national consultative committee to certify centres of reference was established and four annual calls for proposals led to the certification of 132 centres of reference covering all fields of medicine. The task of these centres is to develop clinical guidelines for the diagnosis and the treatment of the RD they cover. They are required to collect epidemiological data on the patients attending their centre and in charge of deciding about the first prescription of any costly treatment for RD. As these centres have a national scope, they are requested to organise their own network of regional centres to avoid travel for patients. This aspect of the national plan has already been evaluated as having introduced a real difference in the quality of care. Centres of reference are certified for five years. An internal evaluation is scheduled after three years and an external evaluation after five years.

7- The seventh objective was to pursue the dynamic development of new orphan drugs through appropriate measures. A funding process has been put into place to support academic clinical research. The French system of ATU (temporary authorisation for use) has been continued to allow early access to orphan drugs in development, and to reimburse off label use of drugs proved to be efficient for certain RD. This system has been assessed as very satisfactory.

8- The eighth objective was to provide an appropriate response to the specific accompanying needs of patients with RD by structuring links between the health care institutions in charge of disability and the centres of reference for RD, and by supporting patient organisations. Thus far, this objective has only been partially accomplished as the reorganisation of the pathways for handicapped people has just recently gone underway. The new structures have been recently implemented and the link to centres of reference is in the process of being established. Patient umbrella organisations are also supported.

9- The ninth objective was to improve research on RD by putting in place a voluntary research policy. An annual call for proposals was launched, entirely dedicated to RD, and funded by all existing research funding agencies (5 million Euros per year). This action is considered as one of the most effective of the national plan.

10- The tenth objective was to further the national coordination of all partners, notably patients associations, and to contribute to a European coordination for RD. At the national level, a Platform for RD was established in Paris in which two patient umbrella organisations (Alliance Maladies Rares and Eurordis) Orphanet, a RD help line and the research funding agency for RD all work in the same building. At the European level, France contributed actively to the dissemination of the concept of centres of reference for RD, to the establishment of a coordinated funding scheme for research, to the development of coding and classification of RD, and to the promotion of the adoption of other national plans.

In conclusion, the national plan is already considered as a major step forward which could serve as an example for other countries, although each country has its specificities.