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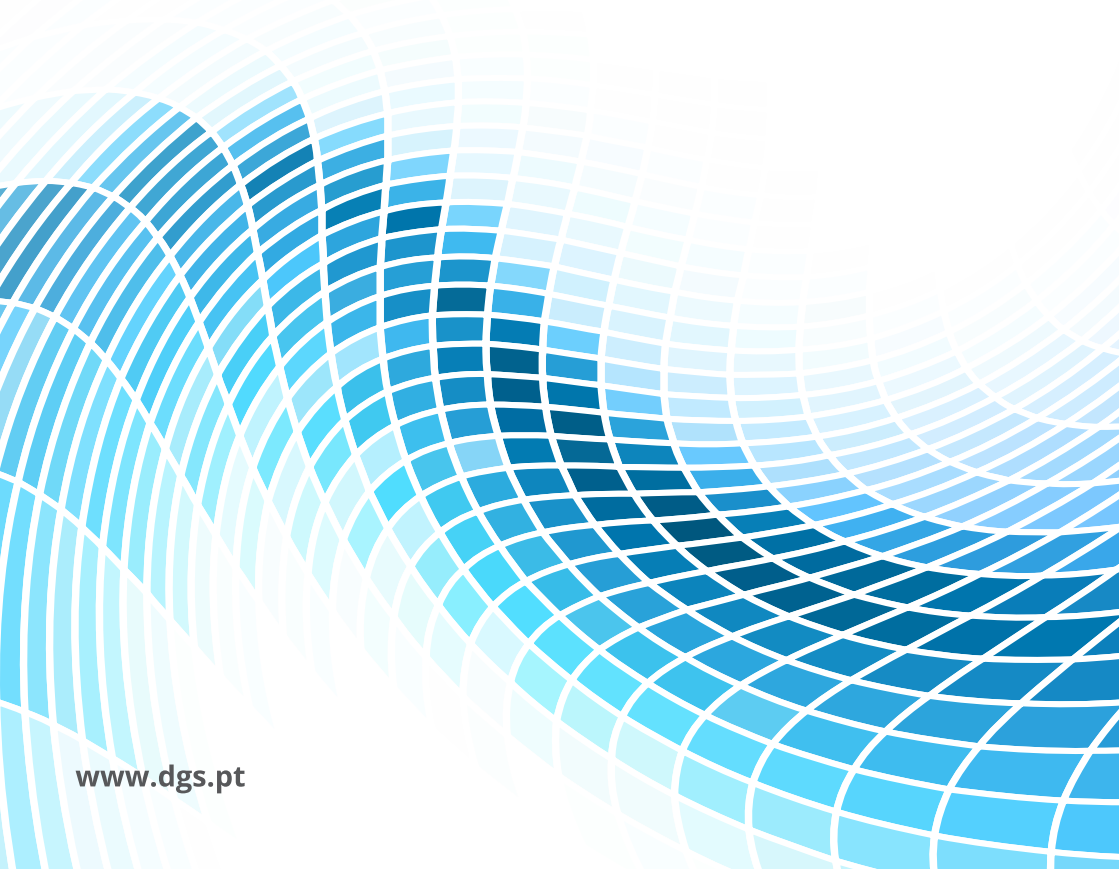
INTEGRATED STRATEGY FOR RARE DISEASES

2015>2020



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I - Framework

In the European Union, diseases having a prevalence inferior to 5 cases for every ten thousand persons, are considered rare diseases, and occasionally also referred to as orphan diseases.

There are, approximately, five to eight thousand rare diseases. Each of these diseases reaches less than 0,1% of the population. Most of them are serious, and occasionally also highly incapacitating, while others, do not hinder the regular intellectual development and have a benign and even functional evolution, when diagnosed and treated at an early stage.

Overall, rare diseases affect around 6% to 8% of the population, and in Portugal, the estimate is of around six hundred to eight hundred thousand persons carrying these diseases. Around 80% of rare diseases have a genetically identified origin, and 50% of new cases are diagnosed in children.

The total number of rare diseases is increasingly bigger. The real dimension of this problem is yet unknown due to inexistence of an adequate codification of these diseases, as well as due to the reduced number and size of the epidemiological studies performed until now.

The estimate is that 5 new diseases are described every week on a worldwide level. Most of them have a serious and incapacitating clinical expression, appearing at an early age before the age of 2 years, associated to multiple disabilities (physical, sensorial or intellectual disability). The prognosis is generally unfavourable, being these diseases responsible for 35% of child mortality in children with less than one year of age.

The delay in diagnosing rare diseases means that timely opportunities for intervention may be wasted. Besides, the relatively common symptoms may hide underlying rare diseases and diagnostic errors may occur.

These diseases have generally a late diagnosis, which is caused by their rarity, to a lack of information from the doctors, inefficiencies in the referral of patients to the most adequate specialized services, and also due to the fact that in a lot of countries Expertise Centres are yet to be defined and recognized. According to the international literature, around a quarter of the patients waits for the definite diagnosis between five and thirty years after the first symptoms. This fact shows the difficulties endured by patients affected with such diseases.

However, the international community recognizes that the patients are not the only ones that need access to high quality information. The mere fact that these diseases are rare means that doctors, nurses, therapists, and pharmacists, as well as social assistants and teachers, often do not have the necessary information that would help them to adequately refer patients and their caregivers, amongst the possible and available responses.

Thus, it is necessary to ensure that doctors are aware of the possibility of verifying a rare disease in a patient, even though they may not have the due competences to perform a specific diagnosis, since that they do not need to have an extensive knowledge about each of these diseases. Furthermore, efforts should be made, to enable all medical and multi-professional specialties the access to a general knowledge about rare diseases, so that they may, whenever necessary, promptly refer their patients to specialized or Expertise Centres.

An early diagnosis in rare diseases and the monitoring of patients, especially in very complex situations, is most efficient when performed in highly specialized centres, which comprehend multidisciplinary teams with advanced scientific and clinical competences, thus allowing patients to promptly benefit from new treatments and knowledge, resulting from research.

A rare disease is a chronic disease, mostly debilitating and often potentially fatal at an early stage, which requires special combined efforts from several intervention areas, where a key role is played by the genetic and pharmacological research, as well as by the support products and the social responses or the way special learning needs are attended, in order to allow patients to be treated, rehabilitated and efficiently integrated in the society.

It should be underlined that medical genetics is inextricably linked to the diagnosis of rare diseases, due to the true outburst of new knowledge and technology, especially in the area of molecular genetics, which is transversal to all medical fields and has opened new opportunity windows to better understand these diseases. Nevertheless, rare diseases may be identified in all medical specialities, whose practitioners are responsible for the diagnosis, monitoring and correct referral of these patients. Many doctors in clinical hospital services have acquired competences in specific diseases or groups of rare diseases, meaning that it is important to develop and assure that they have the necessary resources so that they may be identified as Expertise Centres for rare diseases, and eventually linked to European networks that may be created.

The rarity of such diseases is also the reason for the great difficulties found when carrying out fundamental clinical research, which is necessary in order to develop the adequate drugs for the treatment of these patients.

The involvement of scientific research in this Strategy is thus essential in order to improve the diagnosis, the treatment options and the understanding of these diseases, while the life sciences should help developing new technologies that improve the services and the responses lacking for individuals with rare diseases.

Rare diseases are therefore a particular health problem, and there is a permanent need to research, learn, share knowledge and draft strategies for supporting diagnosis, therapy, rehabilitation, and social inclusion, including - whenever relevant - the access to special education, duly adapted to the exceptional nature of these diseases.

In order for a patient with a rare disease or for its caregiver to make informed choices, he or she must have access to correct and reliable information.

Establishing a good communication between patients, families and professionals is thus essential in order to guarantee that the individual plan of care is consensual, that everyone possesses the correct information and, moreover, that the patient has proper specialized care, what implies a close articulation with the associations representing these patients.

The use of new information technologies, as enablers for the said communication, means - increasingly more - that patients may access remotely to highly differentiated virtual services of specialists who are geographically distant from one another, thus reducing the need for dislocation of patients.

Therefore, the management of rare diseases demands the highest level of partnership between sectors, allowing the possibility of progressively removing unnecessary barriers and offering patients the possibilities that international investigation is providing in terms of effective and sustainable diagnosis, treatments and monitoring.

That is the reason why a joint intersectoral action is mandatory, apart from the provision of health care, towards an agreement on good practices for the efficient use of resources, the sharing of data, the defining of priorities, and finally, the providing of more detailed information to patients and the monitoring of the results of the achieved outcomes.

II - Mission

The Integrated Strategy for Rare Diseases, based on an interministerial, intersectoral and interinstitutional cooperation, which makes a complementary use of medical, social, scientific and technological resources, has as mission the development and improvement of:

1. Coordination of care;
2. Access to early diagnosis;
3. Access to treatment;
4. Clinical and epidemiological information;
5. Research;
6. Social integration and citizenship.

III – Strategic Priorities

Coordination of care

1. Improve interministerial, intersectoral and interinstitutional coordination of care, based on the complex needs of patients and their caregivers, and on the better use of national and regional resources, organizing a coordinated approach of clinical and social services of both general and specialized support, by means of integrated plans of personal care.

2. Improve the offer of therapeutic, rehabilitation and diagnosis, and social inclusion interventions, which should be fast, effective, equitative and sustainable.

3. Design and implement integrated care, clearly defined, accessible and effective.

4. Define referral criteria for highly specialized centres.

5. Promote ways of joint work between patients and their families, their representing associations and supporting professionals, together with research and industry, in order to obtain better outcomes and benefits for individuals with rare diseases.

Access to early diagnosis

1. Promote access equity to early diagnosis and treatments based on scientific evidence, through specialized clinical centres.

2. Define early diagnosis criteria for rare diseases.

3. Improve the identification of individuals with an increased, individual, familiar or geographic risk, given the probability of being carriers of specific hereditary diseases, namely through the implementation of common acting protocols.

4. Design cost-effective national programme proposals for the screening of rare diseases, based on the scientific evidence, internationally recognized as robust, on the economical evaluation and on the concrete treatment possibilities.

5. Improve the graduate and post-graduate professional training in rare diseases.

6. Implement clinical guidelines on genetic testing prescription, promote the quality of medical genetic laboratories and, furthermore, improve the access to genetic testing for hereditary diseases.

Access to treatment

1. Improve the access to early treatment by means of surgery, drugs or nutrition of serious rare diseases.

2. Implement adequate proceedings, both transparent and robust, for evaluation of cost-benefit innovating therapeutics for rare diseases.

3. Improve the information on new, available therapeutics.

4. Implement the systematic production of individual care plans after the diagnosis, based on evidence describing the expected course of the disease and also establishing the responsibilities of the several institutions and professionals involved in care provision.

5. Promote the use of telemedicine and IT (information technologies) as aid tools to the coordination of care, with the purpose of giving a better and faster access to merged specialized services, regardless of the geographical area where those services are located.

6. Cooperate in the identification and in the proposal for recognition of national Expertise Centres for rare diseases and further promote their integration in European reference networks.

Clinical and epidemiological information

1. Promote the systematic use of the European information system Orphanet, embracing it as the reference portal and credible information source about rare diseases, their characteristics, diagnosis and treatment possibilities.

2. Design national registries of rare diseases, specifically through precise methods and genetic information gathering tools that allow incidence and prevalence calculations, which sustain the national strategic plan and the need to resort to European or international cooperation.

3. Increase the epidemiological, clinical and economic data reliability.

4. Develop at a national, European and international level, the share of information, knowledge, good practices and experience, in terms of diagnosis and treatment of rare diseases.

Research

1. Promote research through collaborative activity amongst health and social services with the scientific and academic community, and with the industry.

2. Promote patients' participation in every stage of the research process, through the awareness and availability of accurate, adequate and helpful information, respecting the law and its dignity,

3. Improve the connection between research and the care provided to patients, thus promoting a culture of innovation.

4. The fastest access to care based on evidence.

5. Create partnerships in the scope of research to identify development opportunities for new drugs, which will help to improve the treatment and the evolution of the most common rare diseases.

6. Promote European and international cooperation in basic and translational research, mainly in the scope of extremely rare diseases.

Social inclusion and citizenship

1. Develop training and education pro-grammes for the health and social sector professionals, which will help their empowerment in the identification, treatment and rehabilitation of rare diseases.

2. Encourage active cooperation of patients' associations of rare diseases in the definition of integrated responses and in their achievement.

3. Empower individuals with rare diseases and their caregivers, through the support from a coordinated and complementary

action of patients' associations and - when relevant – through the satisfaction of special educational needs of those patients.

IV Strategy Coordination

The Strategy is coordinated by an inter-ministerial commission, presided by the Director-General of Health, which sets out its functioning rules, and it is composed by the following members:

a) One representative of the Directorate--General of Health;

b) One representative of the National Institute of Health, Dr. Ricardo Jorge, I. P.;

c) One representative of the Central Administration of the Health System, I.P.;

d) One representative of the National Authority for Medication and Health Products, I.P.;

e) One representative of the Social Security Institute, I.P.;

f) One representative of the National Institute for Rehabilitation, I.P.;

g) One representative of the Foundation for Science and Technology, I.P.;

h) One representative of the Directorate--General of Education, in the area of special educational needs.

Source:
MINISTRIES OF HEALTH, EDUCATION AND SCIENCE, AND OF SOLIDARITY, EMPLOYMENT AND SOCIAL SECURITY: Order no. 2129-B/2015 in Official Journal, 2nd Series – No.41 – 27th of February 2015

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