



EURORDIS

European Organisation for Rare Diseases

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“Rare Diseases: understanding this Public Health Priority”



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What is a Rare Disease?

1 The concept of disease

The general definition of a disease is as follows: a disease is an impairment of health or a condition of abnormal functioning¹. It is a pathological condition of a part, organ, or system of an organism resulting from various causes, such as infection, genetic defect, or environmental stress, and is characterised by an identifiable group of signs or symptoms².

A patient will define himself or herself as affected by either a disease or a disorder. In this document the word “disease” is used.

2 The concept of rarity

2.1. Figures of rarity

“A rare disease is a disease that occurs infrequently or rarely in the general population”. In order to be considered as rare, each specific disease cannot affect more than a limited number of people out of the whole population, defined in Europe as **less than 1 in 2,000 citizens** (EC Regulation on Orphan Medicinal Products). This figure can also be expressed as 500 rare disease patients out of 1 million citizens. While 1 out of 2,000 seems very few, in a total population of 459 million citizens this could mean as many as 230,000 individuals for each rare disease. It is important to underline that the number of rare disease patients varies considerably from disease to disease, and that most people represented by the statistics in this field suffer from even rarer diseases, affecting only one in 100,000 people or less. Most rare diseases do only affect some thousands, hundreds or even a couple of dozens patients. These “very rare diseases” make patients and their families particularly isolated and vulnerable. It is worth noting that most cancers, including all cancers affecting children, are rare diseases.

Despite the rarity of each rare disease, it is always surprising for the public to discover that according to a well-accepted estimation, “about **30 million people have a rare disease** in the 25 EU countries”³, which means that 6% to 8% of the total EU population are rare disease patients. This figure is equivalent to the combined populations of the Netherlands, Belgium and Luxembourg.

Quoting from the Background Paper on Orphan Diseases for the “WHO Report on Priority Medicines for Europe and the World” – 7 October 2004: “Unfortunately, the epidemiological data that are available are inadequate for most rare diseases to give firm details on the number of patients with a specific rare disease. In general people with a rare disease are not registered in databases. Many rare diseases are summed up as “other endocrine and metabolic disorders” and as a consequence, with few exceptions, it is difficult to register people with a rare disease on a national or international basis, and in a reliable, harmonised way”. In the case of rare cancers, many registries do not publish sufficient data that break down figures of rare tumours

¹ WordNet

² Answers.com

³ Background Paper on Orphan Diseases for the “WHO Report on Priority Medicines for Europe and the World” – 7 October 2004

by type, even though this information might be available from pathological examination of tissue removed during surgery.

It is worth noting that each and every one of us is, statistically speaking, a carrier of 6 to 8 genetic abnormalities, which are, usually but not always, recessive ones in their transmission. These abnormalities generally have no consequences, but if two individuals carrying the same genetic abnormality have children, these may be affected.

2.2. Paradox of rarity

The above-mentioned figures mean that even though the “diseases are rare, rare diseases patients are many”. It is therefore **“not unusual to have a rare disease”**.

It is also not unusual to “be affected by” a rare disease, as **the whole family of a patient is indeed affected** in one way or another: in this sense it is “rare” to find a family where nobody is - or no ancestor has been - affected by a rare (or “unknown”, “unexplained”, “strange”) disease.

A mother tells:

“At the age of 6, Samuel was diagnosed with a rare metabolic disease. Almost three years after Samuel’s death, we are still a family with a rare disease: I have discovered that I have symptoms linked to the fact that I am a carrier, my marriage broke down due to the stress of losing a child and my daughter was unable to sit her A level exams due to the grief of losing her little brother and her father leaving”.

3 Diversity and heterogeneity of rare diseases

From the medical perspective, rare diseases are characterised by the large number and **broad diversity** of disorders and symptoms that vary not only from disease to disease, but also within the same disease. The same condition can have very different clinical manifestations from one person affected to the other. For many disorders, there is a broad diversity of subtypes of the same disease. It is estimated that **between 5.000 and 7.000 distinct rare diseases exist today**, affecting patients in their physical capabilities, their mental abilities, in their behaviour and sensorial capacities. Many disabilities can coexist for a given person, and this is defined as a polyhandicap.

Rare diseases also differ widely in terms of severity, but in average the life expectancy of rare disease patients is significantly reduced. The impact on life expectancy varies greatly from one disease to the other; some cause death at birth, many are degenerative or life threatening, whilst others are compatible with a normal life if diagnosed in time and properly managed and/or treated.

80% of rare diseases have identified genetic origins, involving one or several genes or chromosomal abnormalities. They can be inherited or derived from *de novo* gene mutation or from a chromosomal abnormality. They concern **between 3% and 4% of births**. Other rare diseases are caused by infections (bacterial or viral), or allergies, or are due to degenerative, proliferative or teratogenic (chemicals, radiations, etc) causes. Some rare diseases are also caused by a combination of genetic and environmental factors. But for most rare diseases the etiological

mechanisms are still unknown due to lack of research to find the physiopathology of the disease.

There is also great **diversity in the age at which the first symptoms occur**. Symptoms of many rare diseases appear at birth or in childhood, including Infantile Spinal Muscular Atrophy, Neurofibromatosis, Osteogenesis Imperfecta, Rett syndrome and most metabolic diseases, such as Hurler, Hunter, Sanfilippo, Mucopolysaccharidosis Type II, Krabbe diseases, Chondrodysplasia. In some cases, the first symptoms of the disease, such as Neurofibromatosis, may occur in childhood, but this does not prevent much heavier symptoms to occur at a later stage of life. Other rare diseases, such as Huntington disease, Spinocerebellar Ataxias, Charcot-Marie-Tooth disease, Amyotrophic Lateral Sclerosis, Kaposi's Sarcoma and thyroid cancer, are specific to adulthood. Whilst many diseases cause symptoms in childhood, these symptoms may not translate into a specific rare diagnosis for years.

It is also to be underlined that **relatively common conditions can hide underlying rare diseases**, e.g. autism (in Rett syndrome, Usher syndrome type II, Sotos Cerebral Gigantism, Fragile X, Angelman, Adult Phenylketonuria, Sanfilippo,...) or Epilepsy (Shokeir syndrome, Feigenbaum Bergeron Richardson syndrome, Kohlschutter Tonz syndrome, Dravet syndrome...). For many conditions described in the past as clinical ones such as mental deficiency, cerebral palsy, autism or psychosis, a genetic origin is now suspected or has already been described. In fact, a rare disease can be masked by a host of other conditions, which may lead to misdiagnosis.

4 Common characteristics of rare diseases

Despite this great diversity, rare diseases have some major common traits. The main characteristics are as follows:

- Rare diseases are severe to very severe, chronic, often degenerative and life-threatening;
- The onset of the disease occurs in childhood for 50% of rare diseases;
- Disabling: the quality of life of rare diseases patients is often compromised by the lack or loss of autonomy;
- Highly painful in terms of psychosocial burden: the suffering of rare disease patients and their families is aggravated by psychological despair, the lack of therapeutic hope, and the absence of practical support for everyday life;
- Incurable diseases, mostly without effective treatment. In some cases, symptoms can be treated to improve quality of life and life expectancy;
- Rare diseases are very difficult to manage: families encounter enormous difficulties in finding adequate treatment.

5 Clarification of some related concepts: rare diseases, neglected diseases, orphan diseases, orphan drugs

It is not unusual to read documents and publications in which the concepts of rare diseases, neglected diseases, orphan drugs and orphan diseases are not clearly

defined and used as interchangeable concepts. This situation has led to misperception and confusion as to precisely what each of these concepts refers to and/or as to what reality each of them covers.

5.1. Rare diseases

Firstly, rare diseases are characterised by their low prevalence (less than 1/2,000) and their heterogeneity. They affect both children and adults anywhere in the world. Because rare disease patients are a minority, there is a lack of public awareness; these diseases do not represent a public health priority, and little research is performed. The market is so narrow for each disease that the pharmaceutical industry is reticent to invest in research and to develop treatments for rare diseases. There is therefore a need for economic regulation, such as national incentives, as provided for in the EC Orphan Drug Regulation.

5.2. Neglected diseases

Neglected diseases are common, communicable diseases that mainly affect patients living in developing countries. Because they are not a public health priority in the industrialised countries, little research and drug development is performed for these diseases. They are “neglected” by the pharmaceutical industry because the market is usually seen as unprofitable. There is a need for economic regulation and alternative approaches in this field in order to create incentives aimed at stimulating research and developing treatments to fight neglected diseases, which are prevalent in developing countries. Neglected diseases are therefore not rare diseases.

5.3. Orphan diseases

Orphan diseases comprise both rare diseases and neglected diseases. They are “orphan” of research focus and market interest, as well as of public health policies.

5.4. Orphan drugs

Orphan drugs are medicinal products intended for the diagnosis, prevention or treatment of rare diseases. These drugs are called "orphan" because, under normal market conditions, it is not cost-effective for the pharmaceutical industry to develop and market products intended for only a small number of patients suffering from rare conditions. The drugs developed for this unprofitable market would not be financially viable for the patent-holding manufacturer. For drug companies, the cost of bringing an orphan medicinal product to the market would not be recovered by the expected sales of the product. For this reason, governments and rare disease patient organisations have emphasised the need for economic incentives to encourage drug companies to develop and market medicines intended for the "orphaned" rare disease patients.

Living with a rare disease: same difficulties - different specificities

Beyond the diversity of the diseases, rare disease patients and their families are confronted with the same wide range of difficulties arising directly from the rarity of these pathologies:

- **Lack of access to correct diagnosis:** the period between the emergence of the first symptoms and the appropriate diagnosis involves unacceptable and highly risky delays, as well as wrong diagnosis leading to inaccurate treatments: the pre-diagnosis maze;
- **Lack of information:** about both the disease itself and about where to obtain help, including lack of referral to qualified professionals;
- **Lack of scientific knowledge:** this results in difficulties in developing therapeutic tools, in defining the therapeutic strategy and in shortage of therapeutic products, both medicinal products and appropriate medical devices;
- **Social consequences:** living with a rare disease has implications in all areas of life, whether school, choice of future work, leisure time with friends, or affective life. It may lead to stigmatisation, isolation, exclusion from social community, discrimination for insurance subscription (health insurance, travel insurance, mortgage), and often reduced professional opportunities (when at all relevant);
- **Lack of appropriate quality healthcare:** combining the different spheres of expertise needed for rare disease patients, such as physiotherapist, nutritionist, psychologist, etc... Patients can live for several years in precarious situations without competent medical attention, including rehabilitation interventions; they remain excluded from the health care system, even after the diagnosis is made;
- **High cost of the few existing drugs and care:** the additional expense of coping with the disease, in terms of both human and technical aids, combined with the lack of social benefits and reimbursement, cause an overall pauperisation of the family, and dramatically increases the inequity of access to care for rare disease patients.
- **Inequities in availability of treatment and care:** innovative treatments are often unevenly available in the EU because of delays in price determination and/or reimbursement decision, lack of experience of the treating physicians (not enough physicians involved in rare diseases clinical trials), and the absence of treatment consensus recommendations.

The first struggle facing patients and families is to obtain a diagnosis: it is often the most despairing fight. This struggle is repeated at every new stage of an evolving or degenerative rare disease. The lack of knowledge of their rare pathology often puts the life of patients at risk and results in enormous wastage: pointless delays, multiple medical consultations and prescription of drugs and treatments that are inappropriate or even harmful. Because so little is known about most rare diseases, accurate

diagnosis is regularly made late, when the patient has already been treated - during many months or even years - for another more common disease. Often, only some of the symptoms are recognised and treated.

A survey by Eurordis (EurordisCare2)⁴ focusing on diagnostic delays for rare diseases, has revealed that, for Ehlers Danlos syndrome, 1 out of 4 patients waited for more than thirty years before being given the right diagnosis.

40% of patients participating in the survey received a wrong diagnosis before being given the right one. Among them:

- **1 out of 6 underwent surgical treatment based on this wrong diagnosis;**
- **1 out of 10 underwent psychological treatment based on this wrong diagnosis.**

The consequences of diagnosis delay are tragic:

- Other children born with the same disease;
- Inappropriate behaviour and inadequate support from family members;
- Clinical worsening of the patient's health in terms of intellectual, psychological and physical condition, even leading to the death of the patient;
- Loss of confidence in the healthcare system.

In the absence of correct diagnosis, emergency units are not in a position to treat the patient appropriately, e.g. headache treated as migraine in a neurological emergency unit, whereas a brain tumour is the underlying cause of the pain. Without a diagnosis, when the patient is a child, the family feels particularly guilty because the child is "acting weird" and is not performing normally in terms of mental and psychomotor development. Any abnormal eating behaviour, which accompanies many rare diseases, is frequently blamed on the mother, causing guilt and insecurity. Incomprehension, depression, isolation and anxiety are an intrinsic part of the everyday life of most parents of a child affected by a rare disease, especially in the pre-diagnosis phase.

The whole family of a rare disease patient, whether children or adults, is affected by the disease of the loved one and becomes marginalized: psychologically, socially, culturally and economically vulnerable. In many cases, the birth of a child with a rare disease is a cause for parental splitting.

Another crucial moment for rare disease patients is the disclosure of diagnosis: despite the progress made over the last ten years, the diagnosis of a rare disease is all too often poorly communicated. Many patients and their families describe the insensitive and uninformative manner in which diagnosis is given. This problem is common among health care practitioners, who are too often neither organised nor trained in good practice for communicating diagnosis.

Up to 50% of patients have suffered from poor or **unacceptable conditions of disclosure**. In order to avoid face-to-face disclosure, doctors often give the terrible diagnosis by phone, in writing - with or even without explanation – or standing in the corridor of a hospital. Training professionals on appropriate ways of disclosure would avoid this additional and unnecessary pain to already anguished patients and

⁴ More information about the EurordisCare 2 survey can be found on the following websites: <http://www.eurordis.org> and <http://www.rare-luxembourg2005.org/>

families. Further schooling in “breaking in bad news” to patients constitutes an important aspect of medical training.

A father tells:

“When I went to pick up my one year-old daughter in the hospital after I had to leave her for many hours of examinations and testing, I anxiously asked the paediatrician what my baby was suffering from. The doctor hardly looked at me and, rushing down the corridor, shouted: “This baby, you better throw her away, and get another child”.

Whatever the conditions of disclosure are, the diagnosis of a rare disease means that **life is toppling**. In order to help rare disease patients and their families face the future and avoid their world collapsing, **psychological support** is greatly needed. Every mother and father knows how many worries and hopes are involved in having a child. But what it means to be diagnosed - or having a child diagnosed - with a rare disease cannot be explained.

Parents’ words:

“All parents worry about their child’s future. When you have a child who is profoundly and multiply disabled, these worries are very different and assume enormous proportions. The future is so daunting that often families do choose to live one day at the time. To think about the future is too painful”.

A testimony:

When Jake was diagnosed with this serious life-threatening metabolic liver disease our hopes and dreams for his future were shattered. When your first son is born you have dreams about him becoming a professional football player or maybe a doctor. These dreams were replaced by new dreams such as hoping we will be able to take him home from hospital and hoping that he will live long enough to hear our child saying “Mummy” and “Daddy”.

Of course, the level of existing knowledge varies greatly between “rare” and “very rare” diseases. How well a rare disease is known determines both how rapidly it is diagnosed and the quality of medical and social coverage. Patients’ perception of the quality of their lives is linked more to the quality of care provided, than to the gravity of the illness, or the degree of the associated disabilities. Because of the limited level of knowledge of the medical community, the coverage provided by the public health care system is generally totally inadequate. Lack of effective treatment is due both to a scarcity of research and to the fact that developing drugs for a small population is not commercially viable without economic incentives. But it has to be underlined that many rare diseases are transmitted along different generations, and therefore investing in the fight against rare diseases today may be a very profitable investment.

Fight for recognition

1 Rare diseases as a reality

It is fundamental to realise that rare diseases can affect any family at any time. It is not just “something terrible that happens to other people”. It is a very cruel reality that can happen to anyone, either when having a child or in the course of one’s own life.

In fact, the terminology “rare diseases” only highlights the characteristic of rarity of the complex and heterogeneous mosaic of an estimated 7,000 life-threatening and heavily debilitating conditions. This terminology, which only underlines rarity, immediately puts a **reassuring distance** between the “poor people to whom something so terrible has happened” and the vast majority of citizens who feel protected by the low prevalence of rare conditions. If these diseases were officially called “terrible diseases that slowly kill your child - or **yourself and you are alone**”, which is more like the truth, the existence of about 30 million people directly affected would strike public opinion more.

Fortunately and mainly thanks to the relentless work of patient and parent organisations, things are slowly changing. Until recently, public health authorities and policy makers have largely ignored rare diseases. Today, and even though the number of specific rare diseases which are known is still very limited, we can witness an **awakening of some parts of public opinion** and, as a consequence, some actions are being taken by public authorities. The rare diseases for which a simple and effective preventive treatment is available are even being screened for, as part of public health policy. But this is not enough, and it is time for public authorities to consider rare diseases as a Public Health priority and take action to concretely support patients and families affected by rare diseases. As we know, most of these diseases involve sensory, motor, mental and physical impairments. These difficulties can effectively be reduced by the implementation of **appropriate public policies**.

As underlined in the Background Paper on Orphan Diseases for the WHO Report on Priority Medicines for Europe and the World, “despite the growing public awareness of rare diseases in the last one or two decades, there are still many gaps in knowledge related to the development of treatment for rare diseases. Policymakers have to realise that rare diseases are a **crucial health issue** for about 30 million people in the EU”.

The **social aspects and implications** of rare diseases also have to be kept in mind: the territorial and financial services to support families and patients have to be organised and developed on a local basis, such as day care services, respite centres, emergency units, socialisation and rehabilitation centres, summer camps, education services and professional training. The problems related to the “after us” - when the life-long carer and/or parent of a rare disease patient disappears - have to be tackled and brought to the attention of national and European decision-makers. The current experiences in this field have to be evaluated and valid organisational and managerial models have to be defined. It has to be underlined that the challenges and problems in relation to the social services do last for the whole life of a rare disease patient and become so important that medical aspects of the disease can be given second line priority.

2 Need for raising public awareness and for appropriate public policy

The reasons why rare diseases as a whole have been ignored for so long are better understood today. Clearly, it is impossible to develop a national public health policy specific to each rare disease. However a global - rather than piecemeal - approach can give rise to suitable solutions. **A global approach to rare diseases enables the individual rare disease patient to escape isolation. Appropriate public health policies** can be developed in the areas of scientific and biomedical research, industry policy, drug research and development, information and training of all involved parties, social care and benefits, hospitalisation and outpatient treatment. In order to foster clinical research, the public funding of rare disease clinical trials should be promoted through national or European measures. Healthcare professionals, public health experts and policy makers cannot apply traditional responses and prioritisation to greater need. This approach is not valid for rare diseases and is not ethically sustainable.

Concerning scientific research, there is a striking need for **increased international cooperation**. The existing research efforts are still scattered and fragmented research is being performed with little coordination between research laboratories. For rare diseases, the resources being very limited and patient populations being very small, the lack of coordination is particularly detrimental to the increase of knowledge on rare diseases. In this particular context, unnecessary duplication of research raises important ethical concerns.

Medical and scientific knowledge about rare diseases is lacking. While the number of scientific publications about rare diseases continues to increase - particularly those identifying new syndromes - less than 1,000 diseases benefit from a minimum of scientific knowledge, and these are essentially the “most frequent” amongst rare diseases. The **acquisition and diffusion of scientific knowledge** is the vital basis for identification of diseases, and most importantly, for research into new diagnostic and therapeutic procedures.

Furthermore, “history shows that a major part of universal medical knowledge we have gained over centuries started with rare disease research. The model of a rare disease has helped to understand more common diseases. Also new techniques are developed using rare diseases for models. For example, research on gene therapy is being done with rare diseases such as X-linked severe combined immunodeficiency, cystic fibrosis, Gaucher disease and haemophilia”⁵.

Easily overlooked by doctors, research scientists and politicians, only the rare diseases that have succeeded in attracting public attention benefit from a public research policy and/or medical coverage. It is mainly patient associations that have raised public awareness. And when this has been possible, progress was made in treating the disease. Patients and families together with health professionals – doctors, scientists and healthcare providers – are co-producing a knowledge base.

From the Agrenska Center in Sweden, we can quote the following reactions from patients and families who have participated in the Family Program:

- **We finally get a true perspective on our children’s disability;**
- **We now feel “normal”;**
- **Exchange of experience is as important as expertise knowledge.**

⁵ Background Paper on Orphan Diseases for the “WHO Report on Priority Medicines for Europe and the World” – 7 October 2004

3 Need for appropriate public health care systems and professional care

After the appearance of the first symptoms, there is an initial battle for diagnosis, which can last for years. After the diagnosis, comes for patients and their families the fight to be heard, informed and directed towards competent medical bodies, where they exist, in order to get the most adequate existing treatment. After the diagnosis, patients and their families are too often faced with **seriously inadequate health and social care**.

For the vast majority of rare diseases, no protocol exists for good clinical practices. Where it does exist, the completeness of dissemination may not be optimal: not all healthcare professionals are always adequately trained, not all EU countries have adopted and shared the protocols. Additionally, the segmentation of medical specialities is a barrier to the comprehensive care of a patient suffering from a rare disease.

Families and health care workers frequently complain about the extreme difficulty in taking the necessary **administrative steps required to receive social benefits**. Major and arbitrary disparities exist between countries - and even between regions within the same country - in the allocation of financial aid, income support and reimbursement of medical costs. Usually in Europe, treatment costs incurred are often higher than they are for other diseases because of the rarity of the disease and the limited number of specialised centres. In most cases, a significant proportion of these expenses is born exclusively by the families, thereby generating an **additional inequality** between rich rare disease patients and poor rare disease patients. Travel costs to specialised centres are high in terms of time off work and financial cost. Furthermore, the anxiety is amplified because usually only one parent can travel whilst the other looks after other children or has to work.

It is also important to underline that, in a family where a child has a rare disease, most often one of the parents – usually the mother – either completely stops or significantly reduces work remunerated outside home. As a consequence, **while expenses increase dramatically, incomes is considerably reduced**. In the case of an adult rare disease patient who is well enough to be able to work, the work hours must be adapted to allow for medical visits and appropriate care. In terms of logistics, much remains to be done to ensure real equality between a disabled and a healthy citizen. It is well accepted that impairment leads to a disability if the environment and regulations do not take into account the special needs of people with impairment to participate in society. The impairment is a part of our being. The **disability comes from outside by disabling factors**.

For some rare diseases, such as familial Mediterranean fever, fragile X syndrome and cystic fibrosis, treatment protocols and defined medical, social and educational programmes exist in certain countries, as well as more or less well-targeted screening programmes.

When antenatal and asymptomatic phase screening methods for rare diseases allow for early and effective medical coverage, they should be implemented because they can significantly improve quality and length of life. Other **screening programmes** should be introduced as part of Public Health policies as soon as simple and reliable tests and effective treatments exist. Qualitative and quantitative progress in prognosis and clinical treatment is raising new public health questions about policies on generalised and targeted screening for some diseases.

From isolation to empowerment of patients

Of course great hopes are raised and profound changes expected from scientific and therapeutic progress. But currently, the existing public research programmes for rare diseases are not sufficient; drug development to treat a small number of patients remains very limited and adequate non-medicinal healthcare is also lacking.

In addition to the general shortage in terms of treatment and care, the surrounding **psychological desert** is particularly painful for individual patients and parents. Not only are you – or your child or family member – affected by a disease about which hardly anything is known, but also nobody understands what you personally – as patient or parent – endure in your daily life. Just to hear the words “I understand” and being able to share stories and tips about your daily experience can be of tremendous help. Sometimes it is even possible to laugh together about painful realities in order to release the unbearable tension that is part of the everyday life of rare disease patients and parents, but this important “outlet valve” is only possible if you first feel totally understood, in absolute security and in an environment safe from judgement. This is why patients and parents have developed a variety of **“self-help support groups”**. There could be value in patient organisations actively initiating and promoting support groups and electronic email discussion groups which link patients and also families and carers, as a means of overcoming the triple problem of geographical, sensorial isolation and rarity.

In this context of **insufficient scientific knowledge base and human isolation**, rare disease patients and their families are known to be more pro-active than patients suffering from other common illnesses because they have often become as knowledgeable - or even much more - about their affliction as health professionals supposed to alleviate their sufferings.

Rare disease patient and parent organisations have been created as a result of experience gained by patients and their families from being so often excluded from health care systems and thus having to take charge of their own disease themselves. The scientific community's inadequate knowledge of rare diseases and the scarce attention given to them by national competent authorities and the pharmaceutical industry has led to the **creation of associations of patients and parents**. Rare disease patient associations aim at gathering, producing and disseminating the limited existing information on their disease and making patients and parents' voices heard. Their **actions have already ensured progress** in healthcare and social assistance provided to patients and parents, development of treatments, and in raising public awareness of rare diseases. In some cases, few patient organisations have managed to raise funds in order to fund research projects that otherwise would not have been carried out.

Conclusion

- The rare disease patient is the orphan of health systems, often without diagnosis, without treatment, without research: therefore, without reason to hope⁶.
- In this general framework studded with difficulties, it has to be underlined that there is **always something useful that can be done** even with the existing, limited but growing, knowledge and means: with the help of re-education and rehabilitation schemes, the recently designated and authorised Orphan Drugs, the progress made in the fields of physiotherapy, nutrition and dietetics, pain management, psychology, medical devices, advanced therapies, information exchange practices, the national public health systems could achieve much more in order to improve quality of life and life expectancy of rare disease patients.
- At national level, some EU Member States have developed **specific public policies on rare diseases**: these countries are Denmark, France, Italy, Sweden, Spain and the UK.
- At EU level, in addition to the **Regulation on Orphan Medicinal Products**, the current reflection on **Centres/Networks of Reference** to tackle conditions requiring a high concentration of expertise shows a growing awareness by EU decision-makers of the intrinsic European added value of rare diseases, which by their nature require action to be taken at international level. Centres of Reference could be either disease-specific, or specialised by group of rare diseases. Existing and developing networks of health professionals and embryonic co-operation between laboratories demonstrate that some **best practices** do already exist, even though there are limited, and have to be shared and disseminated. A main effort still remains to foster and stimulate research in order to increase the existing knowledge, which is far from being enough to meet the challenges of rare diseases.

About EURORDIS

The European Organisation for Rare Diseases (EURORDIS) represents more than 200 rare disease organisations in 24 different countries, covering more than 1,000 rare diseases. It is therefore the voice of the 30 million patients affected by rare diseases throughout Europe.

EURORDIS is a non-governmental patient-driven alliance of patient organisations and individuals active in the field of rare diseases, dedicated to improving the quality of life of all people living with rare diseases in Europe. It is supported by its members and by the French Muscular Dystrophy Association (AFM), the European Commission, corporate foundations and the health industry. EURORDIS was founded in 1997. Further details concerning EURORDIS and rare diseases are available at: <http://www.eurordis.org>

⁶ "Avis et Rapports du Conseil Economique et Social", edition 2001, Journaux officiels de la République Française.