

RARE DISEASES COMMUNICATION DOCUMENT



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Rare Diseases: a socially important rarity

A rare disease is a disease with an incidence of fewer than **5 cases per 10,000** persons.

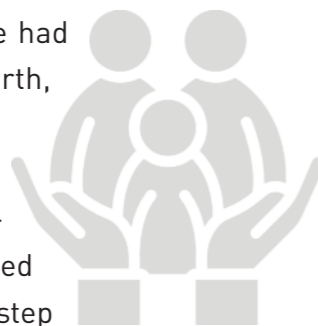
Rare diseases number **between 5,000 and 8,000** worldwide and mainly affect **children** who account **for between 50% and 75%** of all patients. Moreover, the rarity of such diseases is often a complicating factor in the research for pharmaceutical treatments.

Therefore, news of a **medicine** that can **finally treat a rare disease** always elicits an **unforgettable emotion** among rare-

disease patients who have had to coexist, often since birth, with the pathology.

Such treatment, by removing the suffering and limits frequently determined by rare diseases, is a first step toward a better quality of life.

And sometimes **"this" new treatment** can offer a **realistic hope** of recovery.



Think of a better future. Now and after Covid-19

Covid-19 struck the international community and Italy with the force of a **tsunami** and its ongoing effects are subjecting the National Health System to intense pressure.

According to the Centre for Research in Healthcare Economics and Management (CREMS) of the Carlo Cattaneo LIUC University, during lockdown **20.4 million blood analysis** and **more than a million hospital admissions** were **forgone** and **12.5 million diagnostic exams and 13.9 million specialist visits** had to be **reprogrammed or cancelled**. And further consequences include **ever longer waiting lists** for TACS, magnetic resonances and simple ultrasound scans.

We have witnessed the partial **"emptying" of general practitioners' surgeries** but also and to a greater extent specialists' surgeries.

This overview was also confirmed by Cittadinanzattiva's latest national report on chronicity policies, from which it emerged that more than two patients out of five had their examinations or surgery cancelled.

This situation penalizes everybody, but it is espe-

cially punishing for persons afflicted **with rare diseases**: fragile patients who in the first phase of the pandemic were often not able to have their healthcare needs satisfied.



This finding has been confirmed by an investigation conducted by the Superior Health Institute's National Rare Diseases Centre and UNIAMO: **52%** of patients **renounced examinations and routine visits or rehabilitative therapies** for fear of contagion; over **46%** experienced practical **difficulties**: in healthcare and welfare assistance (31%); in accessing medicines as a result of transport and healthcare malfunctions (16%).

For these reasons it is necessary **to make plans for a better future**. Not only now when the pandemic is in full swing, but also when it is finally defeated thanks to the vaccines and pharmacological therapies under development.

Rare Diseases, neglected diseases

All patients are fragile by definition since they must deal with the difficulties and limits determined by their disease. However, as concerns **rare-disease patients** their **fragility** is even **greater** because, in most cases, the patients are children

and their numbers, in terms of each rare disease, are extremely limited.

For this reason, rare-disease patients' **immediate access to rare-disease therapies**, and which are often the only treatment available, constitutes a **non-deferrable right**. And obstacles placed in the way of this right adversely affect both patients and their families.

Unfortunately, **access to pharmacological therapy**, authorised at national level by the Italian Medicines Agency, **is not equally or promptly available** throughout the country as a whole.

Furthermore, nor can the national protection network for persons affected by a rare disease guarantee uniform coverage.

Far too many differences exist not only in terms of links between healthcare centres and territorial structures but also as regards deficiencies in many areas of the national territory that prevent rare-disease patients from enjoying equal access to available medicines.

The pandemic has **lowered the already limited attention** reserved to **rare disease patients even further**: no further information, for example, has been forthcoming both on **the National Rare Diseases Plan**, which has remained inactive since 2016 like its funding, and the regular revi-

sion of the neonatal screening panel. However, Health Vice Minister Pierpaolo **Sileri's** announcement in September **that the ministry has undertaken to produce a new plan as soon as possible** is a highly **positive signal**.



Similarly, the recent decree setting up a **ministerial table** to address the **revision** of the **list** of diseases subject to **neonatal screening** is also encouraging. This is a first step towards accelerating work on improving arrangements for early diagnosis.

The commitment of pharmaceutical companies

Whenever possible and wherever applicable, **pharmaceutical companies** have for years been **implementing patient support programmes and home therapy**. These instruments offer **numerous advantages** for rare-disease patients and their families. They facilitate therapeutic compliance and access to medicines, improve the clinical monitoring of the pathology, minimise risks for fragile patients (for example, by obviating the need for frequent visits to a hospital or a hospital pharmacy) and simplify caregivers' everyday activities by providing them with practical support.

Moreover, the pharmaceutical companies have introduced many other **initiatives designed to reconcile the work and living activities** of their employees, especially those having to cope with fragile and complex situations on a daily basis such as families affected by a rare disease.

During the Covid-19 health emergency, pharmaceutical companies have redoubled their efforts in this field specifically in order to manage the special fragility of persons suffering from rare diseases.

Thinking about patients' future

The first Covid-19 wave has taught us to go beyond accepted practices if we are **to deal with future contingencies better**. Therefore, we must improve access to information, diagnosis, screening, home and hospital care and therapies. However, much important work has already been done and the competent public health authorities should be accorded the merit due for the steps taken.



For example, **the Italian Medicines Agency** has **extended the validity of its therapeutic plans** for medicinal products subject to monitoring,

where the clinical conditions of the patient/pathology make this possible, in order to reduce the risk of infection from Sars-CoV-2 in elderly patients and/or with chronic diseases, and therefore also rare-disease patients, and in so doing has reduced the number of visits to specialised surgeries. **A novelty that should be consolidated and expanded**.

In full Covid-19 emergency, **some regions** implemented measures to facilitate access to therapy by **shifting the distribution of A-PHT medicines from direct distribution to pharmacy-facilitated distribution** for the purpose of reducing the risk of infection in hospital structures for most fragile patients. Another change that could be confirmed refers to the redistribution of healthcare assistance to territorial outlets, especially as regards widely used medicinal products, leaving the public structures with the sole task of distributing medicines for which special conditions apply.

In these times of health emergency, the **Italian Medicines Agency** in view of the fragility of rare-disease patients has deemed it advisable to **provide greater access to home therapy**. This measure has not only **minimized potential risks deriving from discontinuities in treat-**

ment occasioned by fear of contagion in hospital environments, but also bestows **many benefits** upon patients: the guarantee of therapeutic compliance and receiving correct treatment, an improvement in patients' mental health, which, in turn, helps them to cope with their disease better, and the administration of therapy in a family environment.

Home therapy is a perfect example of an **improvement** that has undoubtedly benefited rare disease patients and goes in the direction of **telemedicine** and **proximity medicine**, a matter often addressed by Minister Speranza. This important measure merits greater application especially as it is a means for **strengthening a partnership between the public and private sectors**.



The pandemic has engendered profound changes in all our lives. **Consolidating our best achievements** and learning from past experiences is the right course if we are to continue to work for improved healthcare and **build a better future**: a future that embraces us all, including the most fragile patients.

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