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EmpowerRARE

Empowerment of People with Rare Diseases

The disease may be rare, but care shouldn't be

Newsletter

Extra Issue

Special edition on the challenges of COVID-19

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THE SITUATION CONCERNING RARE DISEASES AND THE CHALLENGES PRESENTED BY COVID-19

Who are we - people so with rare diseases?

We are people affected by rare diseases - those that affect a small percentage of the population, but because of the significant number of different rare diseases they affect a large total population. According to the American organization Global Genes, there are about 300 million people in the world living with a rare disease. Most of them are genetic and are thus present throughout life. Many of the rare diseases appear early in life.

The European Commission in Public Health defines rare diseases as *"life-threatening or chronically debilitating diseases that are so prevalent that special combined efforts are needed to tackle them"*. Later, the term "low prevalence" was defined to mean less than 1 in 2,000 people. Diseases that are statistically rare but not life-threatening, chronically disabling or inadequately treated are excluded from this definition.

The European Union defines a disease as rare when it affects less than 1 in 2,000 people. To date, **more than 6,000 different rare diseases** have been identified, **affecting around 30 million people in Europe and 300 million worldwide. 72% of rare diseases are genetic**, while others are the result of infections (bacterial or viral), allergies and environmental causes, or are degenerative and proliferative. **70% of these genetic rare diseases begin in childhood.**

Due to the low prevalence of any disease, medical expertise is rare, knowledge of them is scarce, care is insufficient and research is limited. Despite their large total number, patients with rare diseases are orphans of health systems, who are often denied diagnosis, treatment and the benefits of research.

The **European Joint Program on Rare Diseases (EJP RD)** brings together 130 institutions in the 27 EU Member States, as well as Canada, Armenia, Georgia, Israel, Norway, Serbia, Switzerland and Turkey, to achieve their two main goals:

1. **To improve the** integration, efficiency, effectiveness and social impact of research on rare diseases by developing, demonstrating and promoting through exchange in Europe and worldwide research and clinical data, materials, processes, knowledge and know-how.

2. **Implementation and further development** of an effective model of financial support for all types of research on rare diseases

(fundamental, clinical, epidemiological, social, economic, health services), combined with accelerated exploitation of research results for the benefit of patients.

<https://www.intechopen.com/books/rare-diseases/international-data-sharing-and-rare-disease-the-importance-of-ethics-and-patient-involvement>

The expected high overall number of patients with rare diseases, orphans in health systems who are often denied diagnosis, treatment and research benefits is the reason for the **US National Organization for Rare Disorders (NORD)**, an organization for the Protection of Patients, dedicated to persons with rare diseases and the organizations that serve them, with more than 300 member organizations, choose the motto

Alone we are rare. Together we are strong.

We ourselves are rare (a little common). We are strong together.



NORD[®]
National Organization
for Rare Disorders

30 million in Europe ...

300 million in the world ...

The construction of awareness about rare diseases is so important, because **one of every 20 people will live with a rare disease in a moment of his life.** Despite this, for most rare diseases there is still no treatment and many of them remain undiagnosed.

Although we are still not treated enough, we are recognized as a challenge

Rare Disease Day is celebrated **on the last day of February** each year.

Rare Disease Day was celebrated for the first time by EURORDIS and its Council of National Alliances in **2008** and later was recognized at a global scale.

The main purpose of Rare Disease Day is to raise awareness among the general public and people, taking decisions, for rare diseases and their impact on the lives of patients.

The campaign is directed primarily to the general public and also so is striving to raise awareness among politicians, public authorities, representatives of industry, researchers, health professionals and anyone, who has a genuine interest to rare diseases.

Special attention is paid to **the participation of the patient** (hereinafter also "engaging a patient") **in the management** of scientific research, the challenges for effective and responsible involvement of patients and how patients with rare diseases cannot be included specifically in the management of biobanks and sharing of data.

Respect for communities is an important ethical principle in health research. According to Charles Weijer et al. researchers have "the obligation to respect the values and interests of the community in scientific research and, when it is possible, to protect the community from harm". This principle is evident in international guidelines for ethical research in the field of health at the Council for International Organizations of Medical Sciences and the World Health Organization: *"The researchers, sponsors, health authorities and relevant institutions should engage with potential participants and communities in meaningful participatory process which involves them in an early and sustainable way in the design, development, implementation, design of the informed consent process and monitoring of research, and in the dissemination of its results"* (Guideline 7).

The participation of patients and patient protection organizations in research, drug development and care is increasingly practiced and is the subject of numerous national and international guidelines for ethical research. Patient involvement is when patients "collaborate meaningfully and actively in the management, prioritization, or design and conduct of research."

Rare Disease Day in the European Parliament

People living with SMA, ALS and other rare diseases were at the center of the event on rare diseases in the European Parliament on 18 February. The event was organized by Biogen in collaboration with EURORDIS, the patient organization for rare diseases.

The challenge of the COVID-19 pandemic for people with rare diseases

Sandra Courbier, Director of Social Research at **EURORDIS**, the voice of people with rare diseases in Europe, (<https://www.eurordis.org/>) commented: "It is clear that the COVID-19 pandemic has a specific impact on the health and quality of life of the 30 million people living with a rare disease in Europe and indeed around the world.

For years, EURORDIS has been collecting data on the experience of people living with a rare disease, which shows the enormous difficulties they experience in accessing care, finding the right specialist and the right therapies.

By creating new barriers, the current pandemic is exacerbating this already difficult situation. We see cases where this causes a strong sense of anxiety among families. We call on politicians and civil servants to remember how vulnerable our community is and to make efforts, where possible, in the post-closure period; to be careful and protect people living with a rare disease".

She adds that this situation is complicated by the fact that **people living with rare diseases** are less well known and therefore "**not sufficiently recognized by the system**", which means that patients are even more vulnerable and less accessible to personalized help. Courier emphasizes that **delays in diagnosis and routine treatment will place an unusual burden on health systems after COVID-19.**

"Diagnosis without COVID-19 is difficult enough, as it can sometimes take up to 15 years to make an accurate diagnosis. This crisis means that cases are not being investigated or tracked properly, which means that there is an accumulation of backlogs. "

"Moreover, **many of these diseases are degenerative. The later patients are diagnosed, the further their condition can worsen,** which means that they need more complex treatment and this will burden the health systems in the future."

She also said that **although patients are affected by this across Europe, some countries are disproportionately affected.**

For example, **eight out of ten operations have been canceled or postponed in Romania, well above the European average.** This is something **that is also observed in other Eastern European countries.**

[The COVID- 19 pandemic makes it difficult to access care for patients with rare diseases](#)

Nine out of ten people living with rare diseases across Europe have been negatively affected by the coronavirus pandemic, including three out of ten saying it has caused them a ' life-threatening ' healthcare disorder, according to a recent EURORDIS study - Rare Diseases Europe.

More than half of those surveyed reported **failing to gain access to vital life-sustaining treatments** such as infusions, chemotherapy and hormone therapy.



Royalty-free [stock photo](#) ID: 287168744 - Stock photo by Sumroeng Chinnapan, Thailand

9 of 10 people, living with a rare disease, have undergone interruption in care because COVID-19

Study shows harmful impact of coronaviruses on the community of people with rare diseases

EURORDIS-Rare Diseases Europe announced on 4 May 2020 Paris preliminary general results of the first study in several countries for this how COVID-19 affects people, living with a rare disease, as established, that pandemic severely hampers access to care.

Over 5000 patients with rare diseases and members of their families from all countries of the EU and outside them, representing 993 diseases, responded to the survey, conducted by the Program **Barometer of rare diseases**. These results are based on responses to surveys, submitted between 18 and 28 April 2020. These are preliminary data and research continues throughout the entire time of crisis. Rare diseases often are chronic and life-threatening.

The results of multilateral study for this how COVID-19 affects people, living with a rare disease, found, that **the pandemic severely hampers access to care** and **exacerbates many already existing challenges**, including access to treatment and drugs.

Concerning the program Barometer of rare diseases

The program **Barometer of rare diseases** is the initiative for the study of EURORDIS, which brings together more than 11,000 patients with rare diseases, members of their families and carers, who share their experience and views on the issues, which are important for the community of rare diseases. The software for study Rare Barometer enables high-quality, safe collection and analysis of data. The program is designed for systematic collection of the views of patients on transversal themes and introducing them into the policy and the process of making a decision, the transformation of the views and experiences of patients and families in figures and facts, which cannot be shared with the wider community and politicians.

The pandemic of COVID-19 exacerbate the numerous challenges with which people, living with a rare disease, already is facing and create additional risks in everyday them with subsequent consequences.

Since the beginning of the COVID-19 pandemic, 9 out of 10 patients with rare diseases have experienced interruptions in the care they receive for their rare disease:

6 out of 10 of those who declared cessation of care related to the COVID-19 pandemic stated that it was detrimental to their health or the health of the person they cared for, and during COVID-19 they no longer had access to medical therapies. already at home or in the hospital, such as infusions, chemotherapy and hormonal treatment.

3 out of 10 respondents believe that these interruptions of care can definitely (1 out of 10) or are likely (2 out of 10) be life-threatening.

More than half of those who needed surgery or transplantation experienced these interventions being canceled or postponed.

More than 6 in 10 did not have access to diagnostic tests, such as blood or heart tests and medical diagnostics - which was often a key part of their daily care.

Nearly 7 out of 10 have had cancellations of appointments with their GPs or specialists caring for their rare disease.

In 8 out of 10, their appointments for rehabilitation therapies such as speech and physical therapies (sometimes the only therapies available when no treatments were available) were postponed or canceled.

In almost 6 out of 10, the monitoring of their mental health was interrupted.

Patients who normally received care in hospitals experienced specific difficulties, with almost 3 in 10 reporting that the hospital or ward that normally provides care for their rare disease had been closed.

1 in 2 have been involved in online consultations or other forms of telemedicine since the beginning of the pandemic. This is new for 2 out of 10 patients. Almost 9 out of 10 of those who have experienced this type of consultation are satisfied with the experience and find it very or very useful.

Patients treated in hospitals experience specific difficulties

For patients receiving follow-up care in hospitals, access to the care they normally receive is difficult, as hospitals are clearly not in the same position to provide this necessary care:

- Almost 3 out of 10 report that the hospital or ward that provides care for their rare disease is closed.

- More than 1 in 10 state that medicines needed to care for rare diseases were missing, as they are now used for patients affected by COVID-19.

- Finally, more than 3 in 10 have even been explicitly told not to go to hospital if they or the person they care for become ill for other reasons, unless they are affected by COVID-19. Fear of infection with COVID-19 is also a major barrier to receiving the necessary care in hospitals: half of those who receive follow-up care through hospitals did not go to the hospital because they feared that they or the person they cared for, can catch COVID-19.

<https://www.euractiv.com/section/coronavirus/news/covid-19-pandemic-hinders-access-to-care-for-patients-with-rare-diseases/>

One of the biggest fears of people with rare diseases is HOW they will be treated by the health system in case of a COVID-19 pandemic

Norwegian good practice for proper triage care

On May 8, 2020, a new publication was published by the research team of COVID-19 of Public Health Gibraltar - **COVID-19 Triage Systems and Fatality Rates: A Comparative Simulation Study** between Singapore, Japan, Norway, USA and UK, co-authored by F. Mansab, S. Bhatti and Daniel K. Golan (F. Mansab, S. Bhatti and Daniel K. Golan).

As **the mortality rate from COVID-19 cases varies considerably around the world** and there are likely to be a number of factors that may contribute to such a difference, not least the different health care models and approaches, the authors examined and compared the effects of COVID-19 health tips from six different countries, **especially exploring ways to Triage governing is otpatsientite in each country.**

Statistical analysis examines the potential relationship between triage outcomes and case mortality.

Research results

Patient-guided triage systems from Singapore, Japan, and Norway maintain a low threshold for clinical contact counseling for patients with possible COVID-19 (88 to 100% of cases were targeted). Patient-guided triage systems in the United States and the United Kingdom maintain high thresholds for health contact counseling (28 and 33% of cases, respectively). Detected is a strong inverse relationship between the percentage of referral cases, the mortality rate of the nation (Pearson's Correlation=-0,642, p=0,01).

Conclusion

In this simulation study, **the triage algorithms of Singapore, Norway, and Japan successfully identified severe COVID- 19** and analyzed such cases of medical care. The US triage system and the UK triage system performed poorly, failing to identify severe COVID- 19 infection and sepsis. **The effectiveness of national triage systems at COVID-19 correlates with the mortality rate** in this cohort of countries.

Triage of patients during a pandemic is perhaps the most central activity of any non-medical response. Proof material should be infused in him when there, especially when it is an infectious disease that never before met. **Changing the availability of resources will define the boundaries and boundaries of such triage systems.** Public information will focus on the key symptoms that are included in the triage system.

Triage criteria are a critical factor in mortality

The World Health Organization (WHO) has provided guidance for both triage of primary care and triage in low-resource settings. **The WHO advises that a small proportion of potential cases of COVID-19 can be attributed to "staying at home"**. The advice is to examine the majority of patients with potential COVID-19 - an unknown and potentially fatal disease - in a clinic or hospital specializing in this purpose.

Different countries have adopted different approaches to the triage process. **Singapore, Norway and Japan have introduced a triage system in line with WHO recommendations.**

NORWAY

Norway maintains a low CFR 2. Patients with mild illness are advised to isolate themselves and contact their personal physician if symptoms worsen or resolve within 7 days. **All patients with shortness of breath are advised to contact their GP or go to the emergency department.**

Conclusion

Following a review of public health policies and patient-led triage systems, it appears likely that patient-led triage systems will have an impact on case mortality during the COVID-19 pandemic.

The most critical aspect of the social response to COVID-19 is the proper assessment of patients' condition. Few problems are more important during the outbreak of a new pathogen than identifying those who need medical attention. **Most critical triage incidents use the highest level of experience.** This is where the most experienced and perceptive guide people's destinies.

Useful sources:

1. Schuetz P, Hausfater P, Amin D, et al. *Optimizing triage and hospitalization in adult general medical emergency patients: the triage project.* *BMC Emerg Med.* 2013; 13:12. Published 2013 Jul 4. doi: 10.1186 / 1471-227X-13-12 7.
2. **WHO Technical Guidance. Role of primary care in the COVID-19 response**
3. <http://iris.wpro.who.int/handle/10665.1/14510.8>
4. **WHO Technical Guidance. Algorithm for COVID-19 triage and referral : patient triage and referral for resource-limited settings during community transmission.**
5. <http://iris.wpro.who.int/handle/10665.1/14502>

Research and innovation

The University Hospital of Oslo maintains a special program **Tests and projects related to COVID-19 and SARS CoV -2** to contribute to better treatment of patients.

One of the current projects is **“Development of an international questionnaire to assess the symptoms and concerns reported by patients related to COVID- 19.** According to the literature and the press, COVID- 19 disease suggests a complex symptomatic severity in the acute and subacute phase, but probably with long-term side effects. In addition, treatments that have been tested in clinical trials can have serious side effects. There is currently no disease-specific questionnaire available for this group of patients.

A specific questionnaire for COVID- 19, developed in accordance with internationally recognized guidelines, will by definition cover relevant health issues for these patients, be more sensitive to changes in patients' condition over time and provide a better ability to capture of differences between treatment groups. There is therefore an urgent need to develop an appropriate tool.

The aim of the study is to develop and validate an international questionnaire for COVID- 19, which can be used alone or in conjunction with other generic questionnaires for patients with verified COVID- 19 disease, in clinical trials and clinical practice, in diagnosing, during active illness and treatment and in the recovery phase.

People living with rare diseases have high hopes that such a questionnaire will better address their health if they need treatment with COVID- 19.

Another project of the University Hospital in Oslo is the decisions for priority of intensive medicine during the pandemic of COVID- 19 (INTENSIVE TRIAGE) - **Intensive medicine priority decisions during the COVID-19 pandemic (INTENSIVE TRIAGE)**

Decisions about whether patients should be offered intensive care are made daily by the doctors responsible for intensive care. When in doubt, the general strategy is to offer hospitalization in an intensive care unit and to attempt treatment for a limited period. During the stay in the intensive care unit, the benefits of the treatment will be regularly re-evaluated by a multidisciplinary team.

Most deaths in intensive care units worldwide occur after a decision to abstain from treatment.

In a crisis situation, such as a pandemic, changes should be expected in relation to which priority decisions / " **triage to intensive care** " are given importance. More patients are expected to be rejected than during periods of normal activity.

The aim of the study is **to increase knowledge about the ethical considerations of physicians, in particular about the reasons and considerations related to decisions to refrain from intensive care.**

In Norway, methods have been established for systematically prioritizing patients according to how urgently they need care, including triage of acute care requests. Ensuring that the sickest patients are treated at the right time is one of the key functions of ED. **Triage is used to identify these patients.**

Evidence of good results from proper triage care in Norway raises the question of the level of triage in Bulgaria - Triage systems for emergency medical care - **pre-hospital** and **hospital admission.**

**Proper triage can be life-saving,
the wrong one is life-threatening.**

Is the Bulgarian medical staff on the front line sufficiently trained and well instructed?

The positive answer is our chance at life.

Additional information about the project, goals, activities and achievements can be found on the official website of the project: <https://www.empowerare.eu>



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