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EmpoweRARE

Empowerment of People with Rare Diseases

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

PROJECT BULLETIN

SPECIAL EDITION

May 7, 2020

SPECIAL EDITION

of the EmpowerRARE project newsletter

Based on a recent study by

EURORDIS-Rare Diseases Europe

Before the explosion of the COVID-19 pandemic

Rare Disease Day in the European Parliament

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

The COVID-19 pandemic makes it difficult to access care for patients with rare diseases

EURORDIS - The voice of patients with rare diseases in Europe

<https://www.eurordis.org/>

Presentation of EURORDIS-Rare Diseases Europe

EURORDIS-Rare Diseases Europe is a non-profit organization made up of over 900 organizations from and for patients with rare diseases from 72 countries, working together to improve the lives of the 30 million people living with rare diseases in Europe. By connecting patients, families and patient groups, as well as bringing together all stakeholders and mobilizing the rare disease community, EURORDIS strengthens the patient's voice and shapes research, policies and services for patients.

Presentation of the Rare Barometer program

The Rare Barometer program is a EURORDIS research initiative that brings together more than 11,000 rare disease patients, family members and carers who share their experiences and opinions on issues relevant to the rare disease community. The Rare Barometer survey software allows high-quality, secure data collection and analysis. The program is designed to systematically gather patients'

opinions on various cross-cutting issues and incorporate them into policies and decision-making processes, turning patients' and families' opinions and experiences into figures and facts that can be shared with the general public. and politicians.

Once again for rare diseases

The European Union considers the disease rare when it affects less than 1 in 2,000 citizens. To date, more than 6,000 different rare diseases have been identified, affecting about 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 each disease, medical experience in their treatment is scarce, knowledge is scarce, care is insufficient and research is limited. Despite their large total number, patients with rare diseases are "orphans" of health systems, often denied diagnosis, treatment and the benefits of research.

9 out of 10 people living with a rare disease have experienced discontinuation of care due to COVID-19

The study shows a detrimental effect on the community of people with rare diseases due to the coronavirus pandemic

May 4, 2020, Paris

EURORDIS-Rare Diseases Europe has announced the preliminary global results of the first multilateral study on how COVID-19 affects people living with rare diseases, finding that the pandemic is making it very difficult for them to access the care they need.

The COVID-19 pandemic has exacerbated the many challenges that people living with the rare disease are already facing and created additional risks in their daily lives with side effects.

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

- 6 out of 10 of those who announced an interruption of the care they needed in connection with the COVID-19 pandemic

said that it was detrimental to their health or the health of the person they care for.

- 3 out of 10 consider that these care interruptions can definitely (1 in 10) or probably (2 in 10) be life-threatening.
- more than half of those in need of surgery or transplantation to survive have had their procedures canceled or postponed.
- 8 out of 10 have seen their prescribed rehabilitation therapies, such as speech and physical therapies (sometimes the only therapies available when treatment is not available), being postponed or canceled.

- Patients who normally receive care in hospitals have experienced specific difficulties, with almost 3 in 10 reporting that the hospital or unit that normally provides care for their rare disease has been closed.
- 1 in 2 have participated 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 counseling are satisfied with the experience and report that it has been very or relatively useful.

5 000 + patients with rare diseases and their families from all EU countries and beyond, representing 993 diseases responded to the survey, conducted by the program Rare Barometer.

The reported results are based on the responses of the study presented to researchers between 18 and 28 April 2020. **These are preliminary data and the study continues throughout the crisis.** Rare diseases are often chronic and life-threatening and research is needed to address identified deficits in the measures taken.

Sandra Courbier, Director of Social Research at EURORDIS, commented:

"It is clear that the COVID-19 pandemic has a concomitant impact on the health and quality of life of the 30 million people living with the rare disease in Europe and the 300 million worldwide.

For years, EURORDIS has been collecting data on the experiences of people living with a rare disease, demonstrating the enormous difficulties they have 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 and when possible in the post-closure period; to take into account the specific needs and to protect the lives of people living with a rare disease”.

Additional key findings of the study:

Patients with rare diseases have experienced interruption in the care they receive for their rare disease

Since the beginning of the COVID-19 pandemic and for those who need the following aspects of the care provided by healthcare professionals, the data so far show that:

- Almost **6 out of 10** no longer have access to medical therapies at home or in the hospital, such as infusions, chemotherapy and hormone therapy.
- More than half of those in need of surgery or transplantation have survived their interventions to be canceled or postponed.
- More than **6 out of 10** no longer have access to diagnostic tests such as blood or heart tests and medical imaging of their condition, which are often a crucial part of their daily care.
- Nearly **7 out of 10** are those who have had canceled appointments with their GPs or specialists who provide them with care for their rare disease
- Almost **6 out of 10** have experienced interruption of their psychiatric follow-up.

A woman with a rare disease in Belgium comments:

"So far, we no longer have follow-up by nephrologists on transplantation. One appointment was canceled and the last blood test was in January, although it should be done every 2 months. Psychological and psychiatric follow-up for ADHD and anxiety was canceled because there were no more consultations at the hospital. And private care is too expensive for me. I live with a hyperactive son in my house for 24 hours a day. "

Patients with rare diseases treated in hospitals experience specific difficulties

For patients who need to receive follow-up care in hospitals, gaining access to the care they normally receive in the COVID-19 situation is difficult, as it is clear that hospitals are not in a position to provide this necessary care:

- Almost 3 out of 10 report that the hospital or unit that provides care for their rare disease is closed.
- More than one declares that the materials needed to care for rare diseases are missing, as they are now used for patients affected by COVID-19.
- Finally, more than 3 in 10 are even explicitly told not to go to hospital if they or the person they care for become ill for reasons other than being 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 receiving follow-up care in hospitals did not go to the hospital because they feared that they or the person being cared for care, can become infected with COVID-19.

These interruptions in care have a threatening effect on the health of people with rare diseases and 6 out of 10 declare that the interruptions in care related to the COVID-19 pandemic they have experienced have been a threat to their health or the health of the person being cared for and 7 out of 10 state that this has affected their condition. The interruption of care mentioned above, in particular of urgent surgery or transplantation, the cancellation of medical therapies or diagnostic tests, is perceived as life-threatening for a significant proportion of patients. 3 out of 10 of the respondents declare that such interruptions related to the COVID-19 pandemic are perceived definitely (10%) or probably (22%) as having a life-threatening impact on them or on the person they care for. Respondents appreciate the extra effort made by their medical professionals to care for their rare disease, despite the obstacles generated by the COVID-19 pandemic.

A patient from the United Kingdom said:

" I had to go to the hospital to go to clinics and get rituximab. The staff took all possible measures to protect me. I know the staff and they made every effort to help me. "

An international study of the impact of the COVID-19 pandemic on the care and quality of life of people with rare diseases continues.

We will keep you informed!

Follow our periodic project newsletters on the EmpowerARE project website

<https://www.EmpowerARE.eu>

Among people with rare diseases, there are those who are further marginalized due to discriminatory factors such as race and minority ethnicity.

Ethnic minorities at higher risk of COVID-19 infection says new study. In the largest study to date on risk factors associated with COVID-19, researchers found that people of Asian ethnicity and black ethnicity were exposed to a higher risk of death from the virus, but have not yet been able to fully explain why. In the largest study of risk factors associated with COVID-19 to date, researchers found that people of Asian and Afro-ethnic descent were at higher risk of dying from the virus, but have not yet been able to fully explain why. The study conducted by researchers at Oxford University and the London School of Hygiene and Tropical Medicine (LSHTM), analyzed data from the National Health Service of 17.4 million adults in the UK between early February and late April. This makes it the largest COVID-19 study conducted by any country to date and offers the strongest evidence currently available for COVID-19 risk factors. Remarkable findings from the study include the fact that people of Asian and black ethnic origin were at higher risk of death than whites. Commentators and researchers have previously speculated that this may be due to a greater prevalence of medical problems such as cardiovascular disease or diabetes, or socioeconomic factors such as the high rate of deprivation among black and ethnic minority (BAME) communities. However, the study concludes that this higher risk is only partly due to existing clinical risk factors or deprivations. Researchers are therefore calling for further work to be done to fully understand why BAME people are at such an increased risk of death.

COVID-19 SPECIFIC ADVICE FOR PEOPLE WITH DISEASE Huntington AND THEIR FAMILIES

The information provided below is adapted from the **HDA** recommendations available at <https://www.hda.org.uk/getting-help/covid-19-information-advice> to the measures in Bulgaria.

COVID-19 can also be spread when people are infected but do not yet have symptoms of the disease (cough, fever, shortness of breath).

Experts believe that people are most contagious when they have the most symptoms (when they are most ill).

How long should you be self-isolated if you have symptoms?

If you have symptoms of **COVID-19**, you should isolate yourself for 14 days.

After 14 days:

- If you do not have a fever, you do not need to continue to isolate yourself
- If you still have a high temperature, continue self-isolating until your temperature returns to normal

There is no need to continue with self-isolation if after 14 days your cough continues. The cough caused by **COVID-19** may persist for several weeks after the infection has resolved.

If you live with someone who has symptoms of COVID-19

If you live with someone who has symptoms of **COVID-19**, you should isolate yourself for 14 days from the day your symptoms first appeared.

For families and children

It is important that you speak openly and honestly in the family about **COVID-19** to provide a space for children and other family members to express their concerns so that you can deal with them.

If you or a loved one suffers from Huntington's disease and needs to interact with medical staff or emergency services, you can explain to them what Huntington's disease is and how **COVID-19** can adversely affect people with this disease.

People with Huntington's disease are considered a vulnerable group as far as **COVID-19** is concerned, because as the disease progresses, they become prone to chest infections that can lead to pneumonia, as well as to the development of heart problems.

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



Keep up to date

To subscribe to the newsletter or to receive more information please visit the project website: www.empowerare.eu

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