EURORDIS open letter to policy makers: Recommendations to protect people living with a rare disease during the COVID-19 pandemic

EURORDIS- Rare Diseases Europe praises the actions, efforts and dedication of all health and social care professionals, as well policy makers at EU, national and regional level, who are facing difficult decisions and exhaustion on the frontline of this COVID-19 pandemic.

We know that no policy maker or public health authority wishes to leave anyone behind during this pandemic. However, the 30 million people in Europe and 300 million worldwide living with a rare disease are among the most vulnerable populations during this crisis.

We would therefore like to bring to their urgent attention the concerns and needs of people living with a rare disease, their families and carers during the current crisis.

Through this open letter, we implore policy makers and authorities in Europe and around the world to take action to protect people living with a rare disease from becoming even more vulnerable during this crisis in line with the recommendations set out further below.

Rare diseases, often chronic, highly complex, progressive and severely disabling, generate specific care needs. The effect of COVID-19 on healthcare and social systems is immense, and directly impacts the care that people living with a rare disease receive during this stressful time, as well as their socio-economic status, education and employment.

Below we set out how COVID-19 impacts the lives of people living with a rare disease in **seven areas listed below** and issue **recommendations** in each area on what policy makers can do to support the rare disease community in the weeks and months to come:

1. **Access to health care**
2. **Screening and diagnosis of rare diseases**
3. **Holistic care, including social services and social support**
4. **Daily life and work conditions**
5. **Safe use of medicines**
6. **Development and Access to medicines**
7. **Research on rare diseases**

This is a time for upholding the guiding principles of solidarity, equity and cooperation. It is a time to see through the promise to 'leave no one behind'[1] and achieve universal health coverage for all[2].

The crisis proves the importance of strong, well-funded, public health and welfare systems that address in particular the needs of the most vulnerable[3] and also demonstrates the added value of coordination in public health policy and collaboration across borders within the EU and beyond.

[1] A/RES/70/1 Transforming our world: the 2030 Agenda for Sustainable Development
[2] Political Declaration on Universal Health Coverage
[3] A/RES/72/139 Addressing the health of the most vulnerable for an inclusive society

Linked to this, EURORDIS, as a member of both organisations, supports the communications on COVID-19 released by the European Patients Forum (EPF) and the European Disability Forum (EDF), and **aims with this open letter to highlight the specific challenges and needs of the particularly vulnerable community of people living with a rare disease.**

1. Access to health care

The surge of patients seeking medical attention due to COVID-19 is stretching the capacity of healthcare systems in terms of human resources, medical equipment (personal protective equipment and devices such as respiratory aid equipment) and hospital beds. **This is causing a number of issues for people living with a rare disease:**

The challenges people living with a rare disease are facing in this area:

- People living with a rare disease are seeing their **regular access to medical advice restricted and their continuity of care disrupted.** This includes routine treatment administration that occurs in the hospital/clinic setting, (e.g.blood transfusions).
- **Certain medical and surgical interventions labelled as 'elective' or 'non-essential' are being cancelled or postponed, and in some instances, other units, such as transplant ones, are having to shut down to leave room for patients infected by COVID-19.** As rare diseases can be highly debilitating and life-threatening, waiting for medical intervention can result in a severe deterioration of symptoms in addition to adding future strains on healthcare systems due to possible complications.
- **People living with a rare disease are facing issues in exercising their right to cross-border health care provided under relevant EU legislation** due to travel restrictions, or because hospitals abroad are cancelling non-COVID-19-related interventions. Accessing specialised health care or treatment in another EU country is especially important to people living with a rare disease due to the scattered expertise on rare diseases and due to certain treatments being only available in some hospitals across the EU.
- **EURORDIS is extremely concerned by reports stating that, in some countries, medical guidelines issued for the COVID-19 pandemic are discriminating against people with disabilities and/or living with a rare disease on the basis of concepts such as 'value of the person to society'.** EURORDIS praises the dedication of all healthcare professionals currently fighting COVID-19, having to take decisions based on their experience and moral compass. Yet, we look with concern at the potential negative impact that following discriminatory guidelines could have on the rare disease and other vulnerable populations.
- **When ill with COVID-19 themselves, people living with a rare disease may face barriers in receiving health care in the hospital setting as there are no protocols set in place for their care.** This is due, in part, to the limited knowledge and scattered expertise on rare diseases.

- People living with a rare disease may **be in fear of visiting hospitals because of the risk of catching COVID-19 o**r because they may not receive the appropriate level of care due to their underlying condition, and consequently may **put themselves at higher risk by staying at home.**

- **People living with a rare disease are not sufficiently considered among the target vulnerable populations for diagnostic testing for COVID-19 (PCR),** in the same way that health professionals, the elderly, people with diabetes and people with cancer are.

- Similarly, testing practices that leave out children as they are considered low-risk or asymptomatic can also be problematic. **70% of genetic rare diseases start in childhood, meaning that there is a vast number of children living with a rare disease who may constitute a high-risk population in the face of COVID-19.**

What policy makers can do:

- **Safeguard the continuity of care and access to medical consultation** for people living with a rare disease by directing funds and efforts towards healthcare systems to reinforce medical workforce and equipment. This will also avoid the need to resort to triage.

- In particular, **in the case of Substances of Human Origin (SoHO, such as organs for transplantation/ blood for transfusions), ensure their free circulation as essential goods/services** and implement measures to mitigate the risks posed by COVID-19 to an adequate and sustained supply of SoHO in accordance with ECDC guidelines.

- **Encourage and facilitate practices like virtual consultations (telemedicine) or the administration of certain therapies in the home setting** to ensure the continuity of care for patients and avoid going to the hospital. These measures must be coupled with the appropriate supply of personal protective equipment to medical practitioners, so that the safety of the patients can be guaranteed during the visit.

- **Revise any guidelines for healthcare professionals that may put into question the rights of people living with a rare disease.** In producing or endorsing medical guidelines, authorities must take into account their commitment to the UN Convention on the Rights of Persons with Disabilities, especially articles 11 on situations of risk and humanitarian emergency and 25 on health.

- **Ensure the adoption of concrete measures/ protocols warranted by the complex needs of rare disease patients in the provision of emergency healthcare during the COVID-19 crisis.** Existing Orphanet resources should be supported and made available to healthcare professionals treating these patients.

    - The on-duty ICU doctor should contact the patient's permanent medical practitioner and/or expert centre to understand the specific disease history and treatment plan of the patient, and make use European Reference Networks' resources (e.g. ERN-BOND COVID-19 helpline).
    - Recognise the role of carers as experts on the disease and the specific needs of the individual patient and allow them as much as possible to attend the ICU.
    - Consider the creation of temporary special hospital wards dedicated to particularly vulnerable rare disease patients affected by COVID-19 whenever possible.

- Where separate hospital wards exist between patients affected by COVID-19 and others, ensure there is adequate communication on this so that patients do not unnecessarily avoid accessing care.
- Encourage and facilitate the use of the Clinical Patient Management System (CPMS) to expedite exchanges between experts in rare disease European Reference Networks for those patients affected by COVID19.

- **Include people living with a rare disease, adults and children, as a priority population in preventative measures (testing, access to PPE, etc.) to stop the spread of COVID-19,** both during the lockdown as well as when these measures are lifted. During the lockdown period especially, carers of rare disease patients most vulnerable to COVID-19 should be systematically tested (or tested on request) with molecular and serological tests. They should receive masks/ personal protective equipment as a priority when quantities allow for this beyond priority health/ social care professionals.

2. Screening and diagnosis of rare diseases

Screening techniques such as medical imaging and laboratory tests can lead to the diagnosis of a rare disease. Not performing them in a timely fashion can be detrimental to the health of those who are yet undiagnosed or those who are affected by a degenerative disease as they are put at risk of severe progression of their disease if it is overseen.

The challenges people living with a rare disease are facing in this area:

- **Appointments for medical imaging and for other screening tests are being postponed or cancelled** due to the lack of medical personnel or the need to use the laboratory facilities for the testing of COVID-19 samples.

What policy makers can do:

- **Ensure sufficient funds and workforce are allocated to hospitals, so that a minimum screening service is guaranteed during the time of the crisis** to test for conditions that may greatly affect the life expectancy of the patient if left untreated, such as cancers, immunodeficiencies and degenerative diseases. Funds from the EU Coronavirus Response Investment Initiative Plus (CRII+) could be allocated to this.

3. Holistic care, including social services and social support

Holistic care covers the 360° spectrum of the health, social and everyday needs of people living with a rare disease and their families. **The implementation of confinement measures and quarantines to avoid the spread of COVID-19 can have a serious impact in the provision of holistic care for people living with a rare disease:**

The challenges people living with a rare disease are facing in this area:

- **The delivery of essential care services for people living with a rare disease such as at-home support or personal assistance is being disrupted,** due to lack of available personnel and personal protective equipment (PPE) for both service providers and the patient. As a consequence people with rare disease and disability, who need intensive support to live independently, are facing long waiting hours for their basic and personal hygiene needs to be attended to.

- A number of **resource centres that offer rehabilitation therapy, physiotherapy, respite care and day care are having to close completely or reduce their offer of services**. They do not have the necessary PPE or sufficient staff to provide them in the home setting. The **sustainability of these essential resource centres is being impacted** by the measures imposed due to COVID-19.

- **Confinement measures can have a severe psychological impact on people living with a rare disease due to the isolation.** Under normal circumstances, being affected by a rare disease impacts mental health (in a 2017 survey[4] 37% of the respondents declared that they often felt unhappy and depressed, compared to 11% of the general population), and the crisis can exacerbate this impact.

- In addition, confinement is very problematic in the case of **diseases and disabilities for which outdoor activity is part of the therapeutic routine, particularly in the case of intellectual disability.** This can cause high distress for the person affected by the disease and for their carers.

What policy makers can do:

- **Guarantee a minimum support service and personal assistance service for vulnerable populations,** including people living with a rare disease, in the same way that minimum childcare services are being maintained in a number of Member States.

- **Take measures to promote the prioritisation of PPE for professionals working in social services.**

- **Develop clear guidelines for social care providers to use during the crisis.**

- **Ensure that the funding of social services across Europe, including independent living services and resource centres for rare diseases, is guaranteed as a matter of urgency.** These services will remain essential when the crisis is addressed and their sustainability must be ensured.

- Ensure that national and regional authorities tap at the **resources made available by the European Coronavirus Response Investment Initiative Plus (CRII+) to fund social care and support providers for persons with disabilities** to ensure continuity of care and support in this time of crisis.

[4] "Juggling care and daily life: The balancing act of the rare disease community", Rare Barometer survey 2017

- **Support existing rare disease national helplines both financially and with guidelines/most accurate up-to-date information**, so they can guide and inform people living with a rare disease during the crisis.

- **Engage with and support patient organisations** which are connected to their grass-roots communities and can back the healthcare services in ensuring information flows and good communication, peer support, and developing creative solutions.

- **Accommodate for people living with conditions affecting behavioral functions which are aggravated by confinement measures**, by for example giving special dispensation allowing them to go outdoors with a carer during lockdown periods, inasmuch as hygienic and security measures are followed.

- **Ensure that people living with a rare disease are not confined to residential institutions and psychiatric units and institutions, due to the lack of independent living services**. If they must be for a limited period of time, ensure that these institutions follow safety guidelines and are well equipped with PPE in the same way as it is being done for institutions for the elderly, while facilitating contact with family members when visits need to be discontinued for safety reasons.

4. Everyday life and work conditions:

People living with a rare disease often face difficulties balancing work life and organisation of care. In addition, they often experience lack of understanding from employers or school, which leads to a lack of adaptation of these settings[5]. These issues are multiplied during challenging times such as the COVID-19 crisis.

The challenges people living with a rare disease are facing in this area:

- **The closure of specialised child care facilities and schools means that families of people living with a rare disease are unable to work and obliged to take full time responsibilities of care for their children or adult relatives.**

- The fact that people living with a rare disease are not systematically reported under the 'at risk population' to COVID-19, means that **employers do not necessarily offer special accommodations for them, such as flexible working arrangements or working remotely.** This is also true for carers who, as a consequence, put the people affected by the rare disease at risk or are obliged to take time off work.

- Lack of adaptation in working practices and lack of Personal Protective Equipment also affect people living with a rare disease **who work in essential services and have to attend the workplace, at risk of infection.**

- **People living with a rare disease are particularly vulnerable to negative economic consequences (lay-offs and reduction in income) ensuing from the crisis' impact on the labour market**. Precarity is already a reality under normal circumstances for many families, with

[5] "Juggling care and daily life: The balancing act of the rare disease community", Rare Barometer survey 2017; pages 18-21

73% of the respondents of a 2017 survey[6] revealing that the cost related to the management of the disease was considered high.

What policy makers can do:

- **Put in place measures for the protection of all workers, with particular priority for those in vulnerable situations such as people living with a rare disease.** The European Pillar of Social Rights should be fully implemented in order to combat poverty and social exclusion.
- **Ensure that people living with a rare disease can work from home, and, if this is not possible due to the nature of the job or any other reason, ensure a special leave that guarantees 100% of the employee's income.**
- **Ensure financial compensation also for families and caregivers who need to take time off work to care for loved ones.**
- **Support schools, other educational facilities and resource centres** that are taking measures to guarantee continued education from home for students living with a rare disease and are providing advice on care to family members, helping them to balance their work and care duties.

5. Safe use of medicines

A number of medicines which have indications for other diseases (such as lupus and rheumatoid arthritis) are currently being tested to verify their efficacy against COVID-19. However, the intake of these medicines in combination with others, or outside of the prescription's indication can put the patient's life at risk.

The challenges people living with a rare disease are facing in this area:

- People living with a rare disease also affected by COVID-19 might be hospitalised in centres that are not their usual centre of expertise and where no link to their permanent medical practitioner is established. In these cases, the administration of medicines to treat COVID-19 (anti-inflammatory, anaesthetics, curare, pain killers etc.) is more challenging than for the general population. There could be **severe drug interactions** between the treatment of people living with a rare disease and the ones provided to treat COVID-19.
- People living with a rare disease are frightened that contracting COVID-19 might worsen the rare disease. They therefore might be tempted to **self-medicate** to avoid the virus, especially if they do not have access to the health care and advice they need. They may also risk using **falsified medicines bought online.**
- Some of the treatments that are essential for the well-being of certain people living with a rare disease (eg. immunosuppressants) may render them more vulnerable to infections. People living in fear of catching COVID-19 may therefore doubt whether to continue their usual treatment for a rare disease without appropriate guidance.

[6] "Juggling care and daily life: The balancing act of the rare disease community", Rare Barometer survey 2017; page 15

What policy makers can do:

- **Make sure that national health authorities are monitoring and putting in place the right measures, guidelines or/and protocols during the COVID-19 pandemic in order to ensure appropriate distribution, delivery and safe use of medicines. For example,** the delivery of medicines that are usually only dispensed in hospitals to local community pharmacies, post offices or shipment to where the patient lives, provided the patient and/or a carer can administer the treatment.
- **Ensure that pharmacovigilance systems are appropriately resourced and that the available information on risks of adverse reactions, drug interactions and appropriate use of existing chronic treatment are communicated in a clear manner to the patients.**

6. Development and access to medicines

The manufacturing of and access to medicines is being impacted during the COVID-19 crisis. On the one hand, there is increased worldwide demand for all medicines used to treat COVID-19 and there are a number of limitations that make meeting this demand difficult. On the other hand, there is also a race towards the development of new or repurposed treatments and a vaccine for COVID-19. All these elements can have an impact on the lives of people living with a rare disease.

The challenges people living with a rare disease are facing in this area:

- People living with a rare disease may face difficulties in accessing their medicines as a number of countries are facing **supply difficulties**. **Manufacturing** of products may be affected because measures to increase production can take 6 months to one year to produce effect. In addition, the raw materials may be produced in a different country than the one where the manufacturing takes place and current confinement and border control measures make their transport difficult. Last, transport and distribution from the manufacturing site to wholesaler and dispensing pharmacies may also be affected.
- The **repurposing of medicines indicated for other therapeutic areas in the management of COVID-19** is stretching the resources available for their usual dispensation (e.g. products missing in community pharmacies and wholesalers). Some of these products such as Hydroxychloroquine and Azithromycin are widely used in the rare disease community.
- EURORDIS has expressed concern over the **possible abuse of orphan drug regulations for the expeditious development of COVID-19 treatments.** Granting orphan designation to these treatments is detrimental to efforts to bring genuine orphan medicines to market and will have a negative impact on rare disease patients' access to other much-needed medicines.

What policy makers can do:

- **Put in place measures to guarantee the manufacturing and distribution of medicinal products and avoid supply issues.** Authorities should follow the practical guidance issued by the EU Commission on green lanes for the continuous flow of goods across the EU and make use of the tools and measures coordinated by the European Medicines Agency to support availability of medicines used in the COVID-19 pandemic.

- **Ensure the use of channels other than orphan drug regulations to accelerate the availability of treatments for COVID-19, and facilitate global collaboration on research as pooling of knowledge and resources is the best way to expedite the development of these medicines.** Authorities should refer to the recommendations and tools issued by the European Medicines Agency that point in this direction.

- For the management of manufacturing difficulties, **provide support to hospital pharmacists for compounding products that can be prepared in hospital pharmacies under high quality requirements,** depending on the nature of the product. Similarly, military pharmacies could help increase supply.

- **Ensure fair and equitable access to investigational and/or authorised products to treat COVID-19 and ensure fair access to compassionate use programmes.** Measures should include:
    - Making information on clinical trials and on compassionate use programmes public and transparent, including elements like the participating centres, the conditions to access, the number of patients enrolled, and the interim results.
    - Starting specific and relevant research on the benefits/risks of treatments for COVID-19 needed in the context of some rare diseases as early as possible.
    - Establishing a European mechanism to coordinate access and ensure a fair distribution among Member States if supply is limited.
    - Member States should ask for an opinion to the European Medicines Agency so that the criteria and conditions of Compassionate Use programme is common in EU Member States (e.g. see the EMA's recommendations on Compassionate Use for Remdesivir).
    - Supply for compassionate use programmes should be made available at no cost for patients in recognition of the public emergency posed by this pandemic.

- **Encourage developers to share the license of medicines which will demonstrate a positive benefit/risks ratio with Low and Middle Income Countries through the Voluntary Patent Pool Initiative (UNITAID)**, as done for HIV, malaria and tuberculosis

7. Research on rare diseases

Most rare diseases lack effective treatments and there is a great need to continue fostering research and innovation for rare diseases. The COVID-19 pandemic poses a threat to the continuation of vital clinical trials.

The challenges people living with a rare disease are facing in this area:

- Clinical trials are being cancelled on the basis of the safety of patients who are unable or not willing to travel to the clinical sites, or because healthcare professionals are re-assigned to other tasks. **However, for some rare diseases, cancelling a clinical trial is more detrimental than being exposed to the virus in terms of life-expectancy.**

- In the case of clinical trials that were already at an advanced stage, developers may be struggling to put in place **measures to protect the patients that were already enrolled and to preserve as much as possible the information that had been collected so far.**

- **The crisis may require the use of decentralized or hybrid trials using digital tools or apps but this practice is not widely used.** For already planned or ongoing clinical trials, introducing these types of designs requires significant amendments of the trial protocols and a revision of the endpoints to be used to conclude on the trial.

What policy makers can do:

- **Ensure the dissemination and follow-up of guidance issued by the EMA to developers on the management of clinical trials during the pandemic,** as particularly in the field of rare diseases, this is crucial so that the efforts that patients put into clinical research (either as advisors to clinical developers when giving input to the design of a study or as trial participants) are not lost.

- **Encourage developers to seek advice from regulators on when to stop, reschedule and/or adapt their clinical trials designs** (including advice on the feasibility and resources needed to perform clinical trials remotely with the use of innovative/decentralized/digital tools) and **ensure also that regulators are ready to provide this type of advice.**