



REPORT ON RARE DISEASES: A CLINICAL PERSPECTIVE

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Almost a year after the Russian invasion of Ukraine, issues faced by the population in Ukrainian cities - affected by the hostilities and their humanitarian consequence - continue to multiply.

EURORDIS has previously published a series of reports highlighting the problems of Ukrainians living with a rare disease (ULWRD), in Ukraine and abroad, from the patient's perspective. This time, we have asked **Ukrainian doctors** to take a closer look at the clinical side of the story and highlight the fantastic work that continues even in times of war.

As this report was compiled in November-December 2022, an important part of the context that should be taken into account when analysing the needs of ULWRD is Russia's attacks on critical infrastructure, which has left most of the country affected by planned and emergency power, heating and water outages. Such outages, depending on the location, can last continuously from several hours to several days. The situation is also much more complicated on the occupied territories and combat areas, where all of these may be absent permanently. This significantly affects Ukrainian doctors and patients, which will be discussed more in detail below.



Patient organisations, EURORDIS, doctors and geneticists in the new premises of the newborn screening laboratory of the National Children's Hospital "Okhmatdyt" (Kyiv, September 2022)



While the difficult winter season is forcing some families to leave their homes in search of safer living conditions and access to healthcare services abroad, the majority of families who chose to stay in Ukraine remain in the country.

As a result, Ukrainian doctors are under heavy pressure. At the same time, the healthcare system, overall, suffers from constant attacks on healthcare facilities, staff outflow and the need to train new specialists, power outages, as well as healthcare budget reduction, among other problems.

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10 months into the invasion, coping mechanisms have been put in place to help overcome some of these challenges. However, many problems require additional resources and crisis response mechanisms.

The information in this report is derived from a series of meetings with Ukrainian clinicians, the European clinicians supporting them, patient organisations, and the Ministry of Health of Ukraine.

It is important to recognise that this is a rapidly-changing situation. This report was compiled between November and December 2022, and what is true today may be different in a few weeks' time. EURORDIS will endeavour to review the situation and provide updates as needed.

DIAGNOSIS OF RARE DISEASES

A diagnosis can serve as the key to unlocking access to appropriate medical and social care. Getting an accurate diagnosis, even when there is no treatment, provides the opportunity for people living with a rare disease and their families to plan their care and future.

Even under normal circumstances, diagnosing a rare disease is a complex process and can take years of testing and doctors' visits. This is further complicated in times of war due to the destruction, absence or insufficiency of laboratories that perform certain tests; the disruption of logistics and supply chains; as well as the inability of patients to pay for expensive tests.

Individual initiatives by the patient community, Ukrainian doctors and their foreign counterparts as well as through corporate donations have enabled sporadic opportunities for diagnostics in case of need. However, it **is important to attract additional resources to improve the diagnosis of rare diseases in a more systematic way.**

At the same time, EURORDIS recognises the Ministry of Health of Ukraine for introducing an expanded newborn screening programme - for 21 diseases - which is an important step for the early detection of rare diseases.

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Genetic tests are rather expensive and not so affordable for our patients. The primary patients who have no initial diagnosis are often incapable to get this diagnosis due to its expensiveness. They may decide that it is better to spend this money on just living costs, which is very sad.

Ukrainian geneticist, the city of Rivne



Patient organisations and EURORDIS at the meeting with the management and doctors of the National Children's Hospital "Okhmatdyt" (Kyiv, September 2022)

AVAILABILITY OF NECESSARY MEDICINES

In 2023, the public healthcare budget in Ukraine is expected to sink by 10-15%, which will also affect rare diseases. Due to currency fluctuations, it is expected that the most affected areas by such deficits will be medicines procurement, which includes the import of medicines and, accordingly, payments in foreign currencies.

Another factor affecting a potential shortage of medicines is the increased mobility of the population, both within Ukraine and abroad - in search of safety and access to necessary services. The return of those who had left earlier, as well as admission to new hospitals in case of internal displacement complicates the procurement and allocation of medicines, even if such procurement is foreseen.



One of the biggest problems we are facing is that we still have a huge number of patients with rare diseases, like Spinal Muscular Atrophy (SMA), that were not given any kind of treatment in Ukraine, and, unfortunately, considering the lack of funding, we understand that those patients are left without any treatment accordingly. It is not just one or two patients all over Ukraine. We are in communication with each and every company about humanitarian aid, but we often hit a dead end.

Ukrainian doctor, Kyiv

The situation, in general, varies greatly from one disease to another, due to disproportional international or industry assistance, as well as different levels of coverage of procurement at the expense of state or local budgets.

In this light, **EURORDIS emphasises the need for more active involvement in the process of supplying orphan medicines, both by manufacturers of such products and through humanitarian organisations working in the field of health care.**



Everybody is getting tired of the war [...] So we are losing the volume of donations. Initially, it was easier to ask for humanitarian aid and some grants but it's becoming more and more difficult now [...] We still have problems, and they are accumulating.

Ukrainian doctor, Kyiv



New equipment installed in the premises of the "Okhmatdyt" newborn screening laboratory

POWER SUPPLY INTERRUPTIONS

As mentioned in the introduction, since October 2022, Ukraine has been subject to attacks on critical civilian infrastructure. In consequence, all regions regularly experience power outages and other disruptions, including water supply, heating, and even mobile communication and Internet access.

While their impact on the general population is undeniably severe, hospitals and patients using electrical devices at home, for treatment purposes or to maintain body functions, are even more vulnerable.

The territory of the National Children's Hospital "Okhmatdyt"



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In particular, EURORDIS has learned that patients who use inhalers, oxygen concentrators or other mains-powered devices do not have any solution in case of energy blackouts.

In this context, an important aspect of supporting ULWRD is the **procurement and provision of generators and power stations, not only to institutions but also to individual families. We urge humanitarian and other organisations working in Ukraine to prioritise the provision of such devices to ULWRD, who often remain invisible in needs assessments in times of crisis.**

Providing Ukrainian hospitals with generators to maintain their autonomy in case of power outages is equally important.

PSYCHOLOGICAL SUPPORT FOR DOCTORS

As already outlined in [our previous report](#), psychological support is an important element of assistance to ULWRD affected by the war.

Psychological support resources are also needed for caregivers of ULWRD, as well as doctors who work with them.

During wartime, doctors face daunting challenges, including the need to work (sometimes overtime) with complex diseases in appropriate conditions and in the absence of basic guarantees of personal safety. All this requires great adaptability and resilience, both physically and psychologically.

For its part, EURORDIS expresses its readiness to expand its psychological support programme to include both family members of ULWRD and doctors working with them.



One of the buildings of the National Children's Hospital "Okhmatdyt"

BETTER COORDINATION OF MEDICAL CARE IN THE FIELD OF RARE DISEASES

Finding an appropriate specialist, in the case of rare diseases, is a difficult task - due to a lack of research and expertise in diseases that are not widely known.

In the midst of war, when many patients are forced to leave their homes, it is extremely important to be able to find a suitable specialist in the new place of residence or transit.



We are now in a situation when patients are lost, basically. They do not know what to do, or where to go. As are we [doctors], to be honest. We started providing our support with communication. We emailed our colleagues when it all started. Patients called us saying, 'We are coming'. 'Coming where?' And they say, 'We don't know'. So, you call Poltava, or Vinnytsia, or Lviv and you say, 'There are patients on the way to you, maybe you can accept them'. Then, eventually, they are abroad somehow.

Ukrainian doctor, Kyiv

Patient organisations, EURORDIS and doctors during the visit to the Centre for Rare Diseases of the National Children's Hospital "Okhmatdyt" (Kyiv, September 2022)





Patient organisations, EURORDIS, doctors and geneticists in the new premises of the newborn screening laboratory of the National Children's Hospital "Okhmatdyt" (Kyiv, September 2022)

In July 2022, a Virtual Centre (Hub) for Rare Diseases for Ukraine was launched in Barcelona, which dealt with requests of finding a medical institution, treatment or specialist, both abroad and in Ukraine.

In December 2022, a similarly structured **Hub was established in Ukraine, on the basis of the National Children's Specialised Hospital OKHMATDYT**, to provide more coordinated assistance to ULWRD in need. This Hub was built on the initiative of Barcelona's Children's Hospital, EURORDIS, the European Reference Networks (ERN), the European Children's Hospitals Organisation (ECHO), with the approval of the Ministry of Health of Ukraine and the European Commission.



We are about 200 kilometres from the capital [...] where patients would stop in transit, coming from other regions of Ukraine on their way to Western Ukraine or abroad. They did not know where they were going. one day they would be in Poland, the following day they would be in Germany. And initially we were trying to find someone who would help us in this regard.

Ukrainian doctor, Vinnytsia



All that [coordination and communication] was first put in motion based on personal contacts. And then, there was the Hub that took that function and started implementing that. Now, we want to have a virtual Hub here in Ukraine, and we want to engage all the doctors that work with rare diseases.

Ukrainian doctor, Kyiv

EURORDIS WOULD LIKE TO EXPRESS ITS GRATITUDE TO THE MINISTRY OF HEALTH OF UKRAINE, THE EUROPEAN REFERENCE NETWORKS, AS WELL AS UKRAINIAN AND EUROPEAN CLINICIANS FOR PARTICIPATING IN THE NUMEROUS DISCUSSIONS THAT LED TO THE PUBLICATION OF THIS REPORT.



The Ministry of Health of Ukraine, EURORDIS and Ukrainian doctors during a visit to the Lviv Regional Clinical Perinatal Centre conducting newborn screening (Lviv, June 2022)