July 2022

Ukraine Emergency Response

Rare Diseases
HUB Ukraine

Hub’s origine

The European Reference Networks for rare and complex diseases respond to the Ukrainian crisis

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The abrupt and ferocious Russian military assault on Ukraine at the end of February 2022 coincided with the Rare Diseases Day and the meeting organized by the French presidency of the European Union Council to specifically address the global public health priority of 6% of the worldwide population affected by nearly 7000 rare and complex diseases.[1] Paediatric onset is observed in 70% of rare diseases which are highly heterogeneous covering almost all fields of medicine with often severe outcomes as acute failures or long-life chronic diseases requiring highly specialized expert multidisciplinary teams and complex therapies. Launched in 2017, the 24 European Reference Networks (ERNs)[2] develop strong links between medical teams from very different disciplines across the EU, facilitating concrete actions for patients suffering from rare and complex diseases. Spontaneously, the coordinators of the 24 ERNs decided to initiate a concerted action to support patients from Ukraine.

In Ukraine, the population of patients with rare diseases is estimated to be about 2 million people. The present humanitarian crisis emphasises the distress of highly vulnerable patients with very immediate or delayed hidden threat when medical care is postponed or stopped due to destroyed healthcare facilities, shortage of therapies or lack of expert medical teams. With more than 6 million Ukrainian refugees and displaced people, this major humanitarian crisis has erupted into unprecedented broad health challenges with a need of urgent responses especially for women and children (but also men) as refugees, and those remaining in Ukraine.[3,4]

In March 2022, ERNs deployed an almost instant response[5] to help Ukrainian patients providing direct contact to the 24 coordinating ERN teams connected to more than 1900 European clinical centers based in more than 500 expert health providers. In addition to remote support during the first 3 months, almost 300 cases have been referred to ERN Healthcare Providers, the majority being patients with rare paediatric cancers, blood or developmental diseases.

Specific medical needs were identified:

- Support for medical diagnosis and advice (destroyed or lost patient files, needs to translate medical reports, difficulties of patient families to contact treating physicians, colleagues in Ukraine in need of support; . . .)
- Support and advice on treatment issues: access to special diets or drug shortages; switch to equivalent alternative medications; access to specialized surgeries or medical devices
- Support to healthcare professionals in Ukraine and patient associations
- Support in triage and referral of refugees to ad hoc EU medical teams
- Support connections between patients and associations, such as EURORDIS

After the first 4 months, we are now also entering a phase during which patients with chronic rare disorders also risk aggravation, and therefore ERNs are partners of centralized HUB.[5]

As a public health concern, rare diseases must always be systematically considered in major health crisis organisation and settings. Patients need to be recognised as affected by a rare disease, to benefit from ad hoc triage towards specialised centres and/or receive ad hoc remote support via on site medical teams. The ERNs fully support the Ukrainian health professionals involved in rare diseases care and plea for a status to integrate the rebuilding/expanding Ukrainian teams as official affiliated partners to the ERNs with the concept of seamless care on the European territories.

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HD, AA, MS wrote & edited the manuscript.

Declaration of interests
N.A.

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Conclusion: There is a need to ensure reliability and acceleration of the response for patients with RD.

Organize RD patients' needs

Hub

Reliably connect the needs with the solution providers

Optimize available patients' solutions

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The Hub:

The Rare Diseases Hub is an innovative project promoted by the European Commission, ERNs, ECHO and EURORDIS.

It is based at the Sant Joan de Déu Children's Hospital (Barcelona).

European Reference Networks

EURORDIS
RARE DISEASES EUROPE

ECHO
European Children's Hospitals Organisation
www.echohospitals.org

SJD
Sant Joan de Déu
Barcelona · Hospital

Generalitat de Catalunya
Departament de Salut

Objective:

To have a dedicated focal point for any Ukrainian affected by the war who needs specialized medical support for their rare and/or complex[1] condition and to help this person and their family to better navigate the support systems available to them.

1. Understanding “complexity” as per ERNs scope

Target

- Population with rare diseases
- Source of contact
- Process of contact

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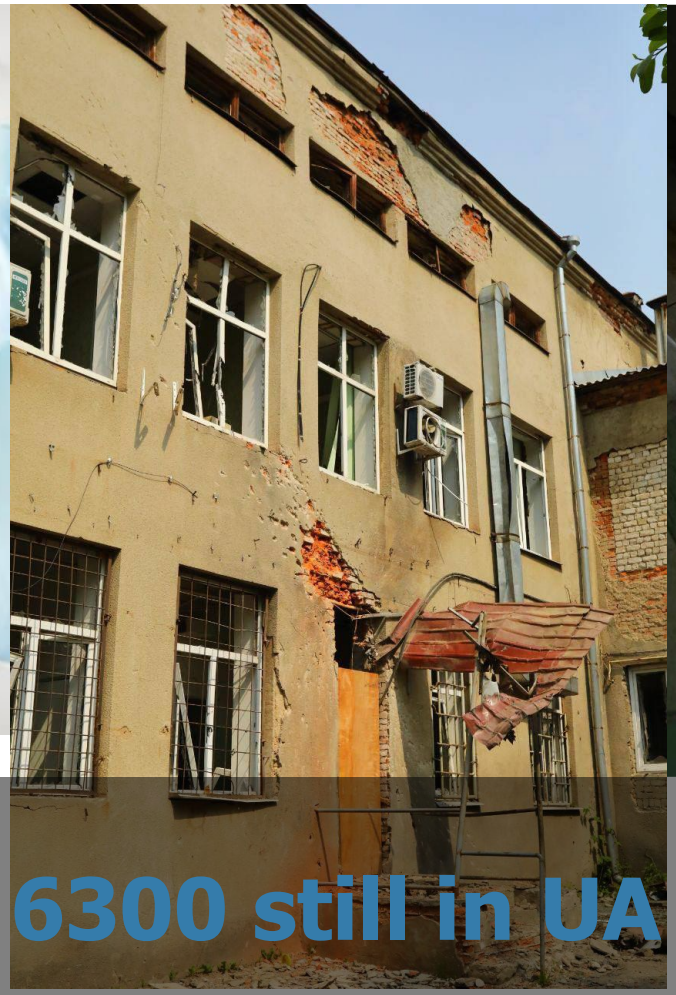
2.000.000 Ukraninian patients with Rare Diseases



6300 still in UA

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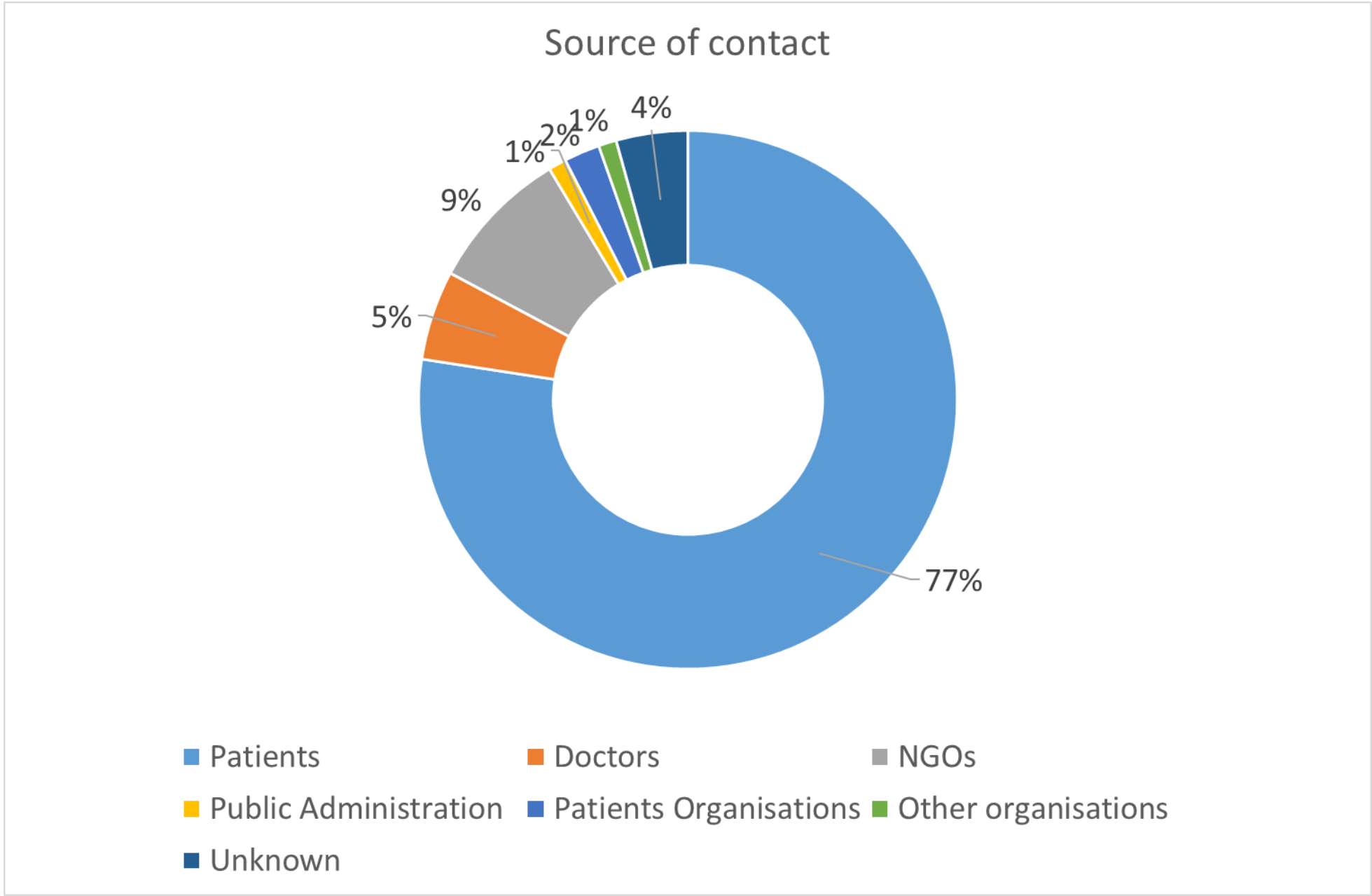

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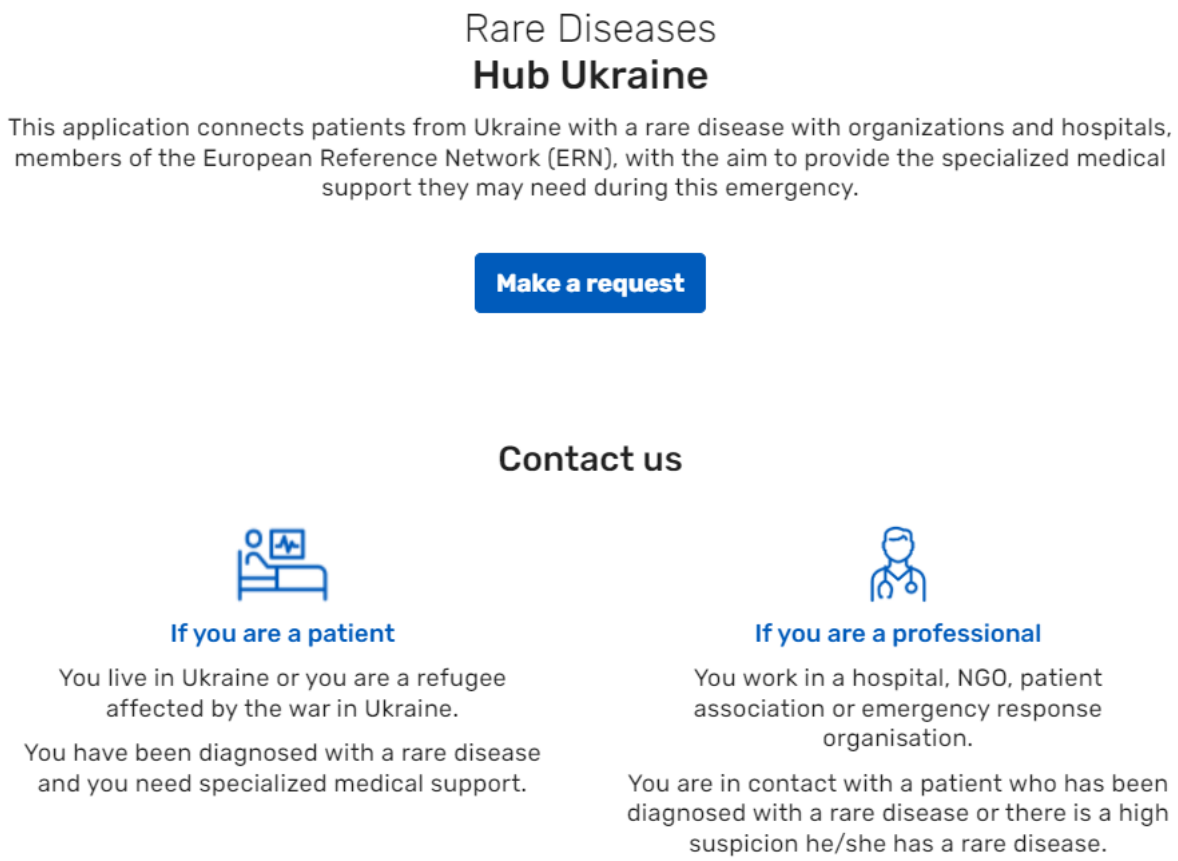
Our web page:



https://www.rarediseaseshub4ua.org/

Available in English, Ukrainian and Spanish

Rare Diseases
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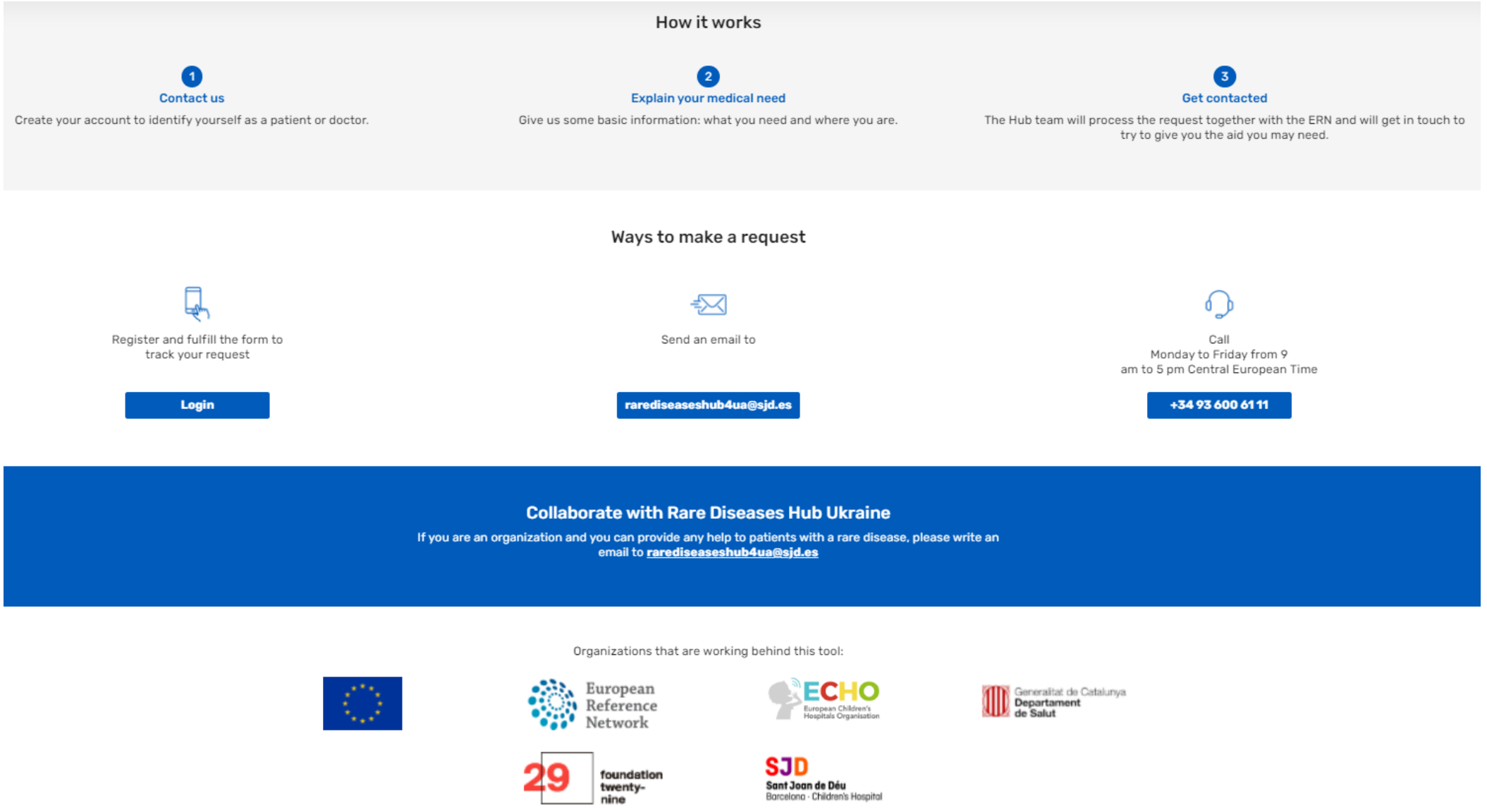
Our web page:



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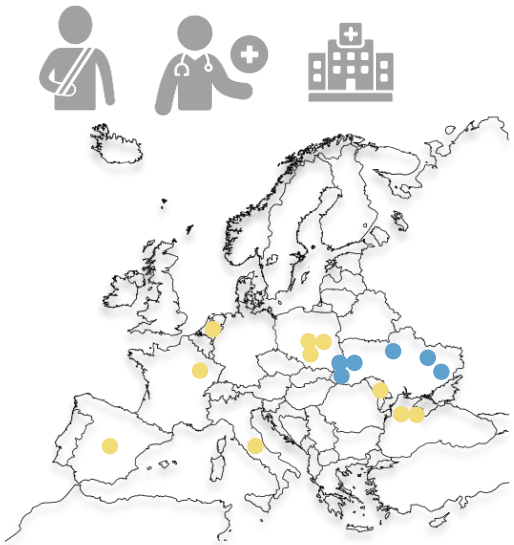
Contact process & operations

Entry channels



Needs could be identified by any channel in any location

APP

API

Web form

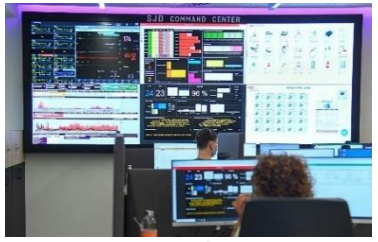
Telephone

Mail

Contact Center

Will be in charge of performing a **triage** over the requests received and **define the answer strategy**

CRM Salesforce



Command Center

Resources

Clinical needs

Providing advice to solve clinical situations will be the **core activity** of the Hub

Logistical/ other needs

Suport in logistical or other needs will only be provided to ensure the clinical response

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Patients journey

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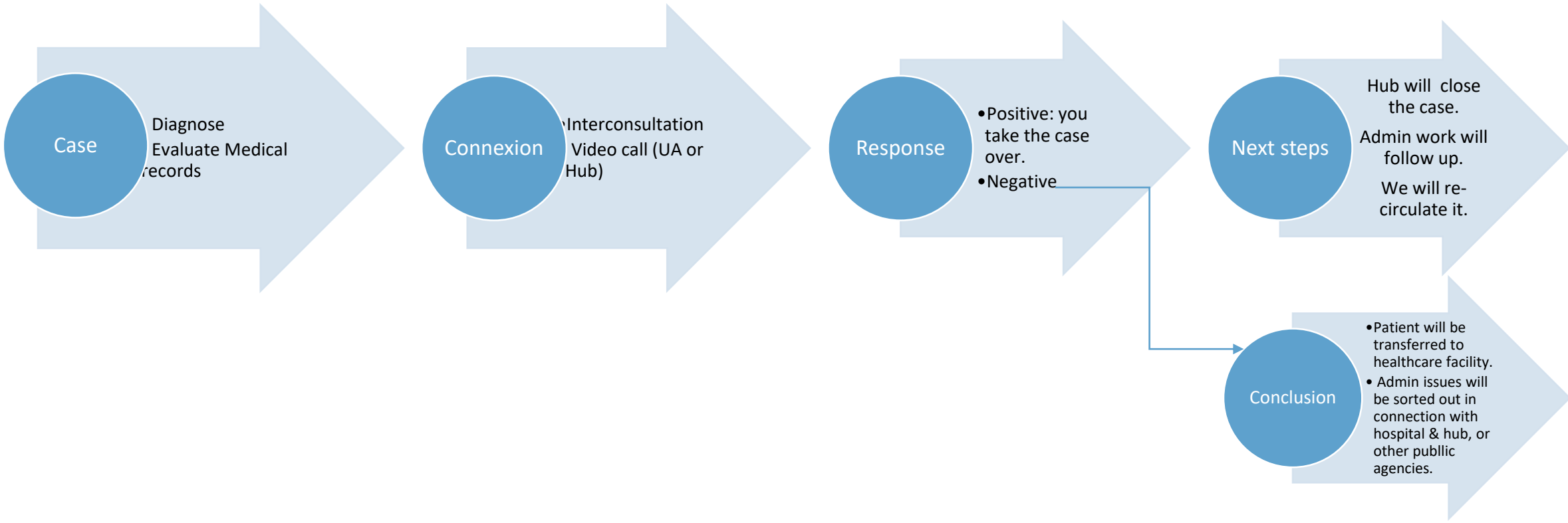
What is it that we need from ERNs?



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Services offered by the Hub

Clinical resources

	Needs	Supplier
	Finding a centre in EU for a RD	ERN members / Other hospitals
	Specialized medical consultation	ERNs
	Assessing medication issues	NGOs, Pharmas, Other suppliers
	Other medical supplies	NGOs, Pharmas, Other suppliers

Major logistic resources

	Needs	Supplier
	Transportation	Transportation companies / agencies, NGOs, etc.
	Accommodation (temporary)	Accommodation companies, NGOs, etc.
	Support with bureaucratic issues	Public Agencies
	Translation services	Translators

We understand as **major logistic needs** anything that is not directly providing clinical support to the patient but it is indeed needed to ensure that the clinical support can be provided. Always subject to the availability of providers in the “response side” ecosystem.

The **function of the Hub** is to **connect with a provider of this solution**, not to provide the solution itself.

Other minor logistic needs such as arranging times to meet patients, providing communication support (e.g., VC), etc. is out of the scope of the Hub and should be managed by the provider of the clinical solution.

Activity (8 weeks)

- Volume of cases
- Type of cases

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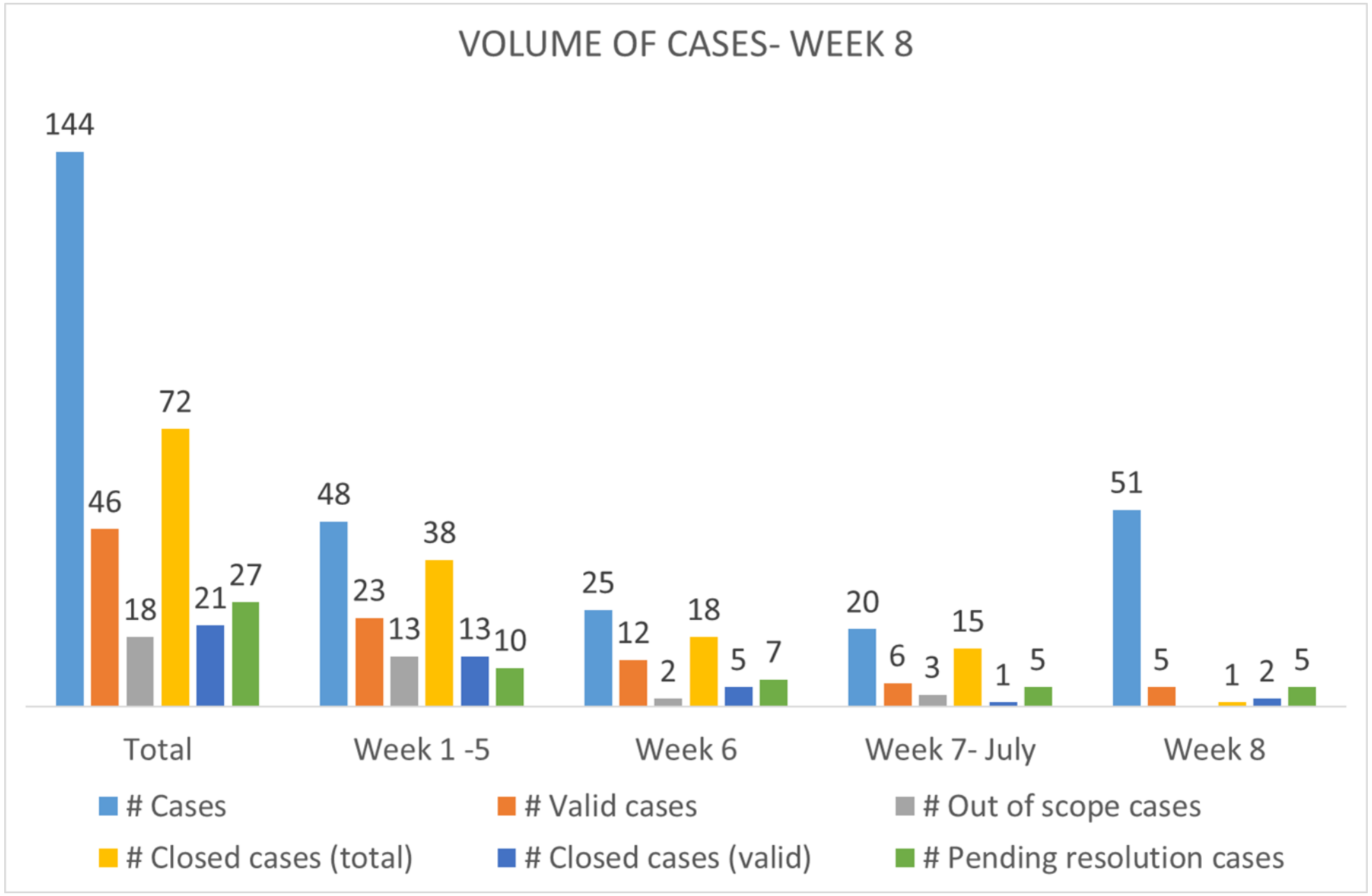

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Qualitative report on main cases. Week 8 (1)

- **Case about Myastenia Gravis:**
  - Meetings held with ERNs coordinators and Ukrainien doctors. Pending response from UA doctors.
  - Medevac mechanism activated for medication supply.
- **Case about Intestin reconstructive surgery need:**
  - Hospital in Mannheim (Germany) is taking the patient over.
  - Admin issues pending to close the case.
- **Case about Lung transplant:**
  - ERNs especialists in Vall Hebrón (Spain) are deciding on the case.
- **Case about Epilepsy:**
  - ERNs coordinators have been contacted.

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Case#1: Patient with multiple congenital malformations needing reconstructive surgery

Anamnesis:

- Female patient, 5 years old, term delivery with intrauterine growth retardation syndrome.

- Diagnosed with
  - Multiple congenital malformations: bladder exstrophy, atresia of the anus and rectum, pelvic dystopia of the right kidney, incomplete duplication of the left kidney, agenesis of the large intestine, embryonic hernia.
  - Spina bifida of the sacrum
  - Secondary episodes of malnutrition grade III, deficiency anemia of moderate severity and urinary incontinence
  - Hypoxic-ischemic lesion of the central nervous system
  - Minimal cerebral dysfunction, asthenic-neurotic syndrome.

- Previous surgeries: foramen ovale, intestinal and urinary reconstructive surgery with stoma

- Unknown regular treatment

- Currently in Ukraine, with family support

Recent history:

- Displaced fracture of the humerus, currently undergoing rehabilitation
- Planned reconstructive surgery of internal organs that cannot be performed, no contact with main surgeon

Contact with the hub:

- NGO contacts the Hub in June 20th, seeking for advice to perform the pending surgery

Evolution of the case:

- **ERNs Eurogen and Ernica contacted. We are currently in conversations with Eurogen**

Case ID: 00034716

Case#2: Idiopathic pulmonary fibrosis with indication of lung transplantation

Anamnesis:

- Patient female, 55 years old
- Diagnosed with:
  - Idiopathic pulmonary fibrosis with lymphangioleiomyomatosis, with long term oxygen therapy.
- Unknown surgeries
- Current treatment: Sirolimus (rapamycin)
- Now in Poltava, unknown family support

Recent history:

- The patient is hospitalized in Poltava, reference doctor has indicated lung transplantation in the shortest possible time due to significant worsening of life quality

Contact with the hub:

- The reference doctor contacts the hub in June 24th seeking for advice on how to proceed to list the patient for lung transplantation.

Evolution of the case:

- Hub contacted ERN Lung and is currently in conversations with them. Vall Hebrón Hospital is taking the case over.
- The hub is reaching out to the pharmaceutical industry to find out medication needed.

Case ID: 00034796

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Case #4

ID AND PATIENT DATA: 00035128 (A 12-year-old girl currently in Odesa (Ukraine)

NEEDS: We think the case needs correction of epilepsy therapy in conditions of lack of control over the neurostimulator and difficulties in accessing medication (drugs are expensive).
This corrective therapy necessarily involves visiting the patient in the center, controlling the neurostimulator and facilitating the patient stays at a place where he has access to neuropediatric control and adequate antiepileptic treatment (possibly also physiotherapy treatment) with monitoring and correction of nutrition, etc.

EXTENDED CLINICAL INFORMATION:

Brief anamnesis, diagnostic studies, course of the disease, treatment performed

Diagnosis: Congenital malformation of the brain. Microcephaly with Schizencephaly. Agenesis of the corpus callosum (ACC), aplasia of the parietal lobes, hypoplasia of the frontal lobes and left temporal lobe. Spastic tetraplegia. Pseudobulbar syndrome. Focal symptomatic epilepsy with complex partial secondary-generalized therapeutically resistant seizures. Condition after implantation of a vagus nerve stimulator (2018). General underdevelopment of speech. Bilateral dysplasia of hip and knee joints. Equinus of feet.

Case#3: Patient with Fukuyama myodystrophy needing logistic support

Anamnesis:

- Patient male, 13 years old

- Diagnosed with:
  - Mitochondrial encephalopathy and secondary polyneuropathy due to Fukuyama myodystrophy
  - Secondary respiratory insufficiency, tracheostomy carrier needing ventilator 12h/day what causes many hospitalizations in the ICU due to respiratory decompensation
  - Limited swallowing ability, mainly liquids and some solids
  - Preserved cognitive function with slower than normal but understandable speech

- Unknown surgeries or current treatment

- Now in Odesa, with family support. Supervised by a Neurologist and in Hospital.

Recent history:

- Due to the war situation at hometown, the patient is experiencing a critical stress situation that is affecting the use of the ventilator, causing fear and anxiety

Contact with the hub:

- The patient was referred by NGO. The family is seeking for transportation to a quieter place to decrease the anxiety and allow the patient to breathe normally
- Transportation would require medical support.

Evolution of the case:

- ERNs- RND and ERNICA were contacted

Case ID: 00034722

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Qualitative report on main cases. Week 8 (cont.)

- **Case about Adult Colon Cancer:**
  - Patient is been transfered to an NGO specialised in that type of cases in UA.
- **Case about Cistic Fibrosis**
  - We are working in a multilateral agreement to get medication donations and logistics arranged. This case will be consider a pilot for similar cases.
- **Cases about Duchenes Muscular Dystrophy**
  - We are still at the stage of collecting clinical data
- **Case about Multiple Sclerosis**
  - In conversations with ERNs
- **Case about Femur Tumour**
  - Waiting for a biopsy results
- **Case about Solitary plasmacytoma with Torax damage**
  - Case is being currently treated
- **Case about Fukuyama Myodistrofia:**
  - Prague's Hospital especialist Dr Haberlova considers diagnose is unclear, since there is a lack of genetic testing nor muscles biopsy that would state the case is more than compatible with Fukuyama.
  - Case remains open in search of ERN or ECHO Member that would take it over.

Q&A

- Who’s paying for the treatments?
- What do ERNs have to do?
- Your preferred contact form?
- Can we grant access to UA doctors?
- Can we grant access to the patient’s medical records?
- How are the patients going to get to the UE hospitals?
- Who is taking care of admin issues?
- How is medication going to be supplied to them?
- How is GRPD guaranteed in the process?
- Are you interested in including Hub’s patients in research studies about RD currently going on?
- ….

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Hub's team contact details

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- Response in max 48h work days