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Experiences and perspectives of collaborations

Romanian National Alliance for Rare Diseases

It is hard to understand
the complexity of Rare Diseases



EURORDIS Photo Contest, 2014

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Low prevalence

Complex

Debilitating

Chronic

Often degenerative

Varying from patient
to patient

Rare Diseases

- * Expertise and knowledge on rare diseases are scarce
- * Research is fragmented & resources are limited
- * The existing treatments are too expensive
- * People with rare diseases struggle to connect with peers and with experts
- * 4000-5000 rare diseases have no treatment

- * Patients have multiple needs and expertise is insufficient
- * Multidisciplinary approach and gathering efforts at national and international level is a real chance for proper management of RD patients

Present situation:

- * Specialists evaluate the patient from their expertise's focus, not the whole person
- * The patient has to repeat his "story" for every specialist involved
- * They get "lost in the system" sometimes, without a clear pathway
- * **Lack of continuity of care**

*** Cross-border healthcare - A patients' right**

“Patients suffering from rare conditions should be entitled to the **SAME quality of treatment as other patients”**

→ they should not be penalised because of the rarity
→ when the treatment/care/ genetic testing is **not there**, they **go & get it** where it is available.

→ Can they **really** do so?

* Art. 13 - a specific article on RDs

Rare diseases

The Commission shall support Member States in cooperating in the development of diagnosis and treatment capacity in particular by aiming to:

- (a) make health professionals **aware of the tools available** to them at Union level to assist them in the correct diagnosis of rare diseases, in particular the Orphanet database, and the European reference networks;
- (b) make patients, health professionals and those bodies responsible for the funding of healthcare aware of the possibilities offered by Regulation (EC) No 883/2004 for referral of patients with rare diseases to other Member States **even for diagnosis and treatments which are not available in the Member State of affiliation.**

* A Regulation and a Directive

1. Regulation (EC) N° 883/2004 on the coordination of Social Security Systems

- Prior Authorisation (PA) to access care “abroad”: always required - based on S2 form (ex E112)
- Authorisation of S2 / E112 cannot be refused where "undue delay" applies
- When OK: actual costs reimbursed

2. Directive 2011/24/EU on CBHC

- Prior Authorisation is not necessarily needed if treatment type is in the “basket of benefits” (the “list”)
- When on the list, reimbursement on the basis of costs “at home”

DO PATIENTS KNOW THIS? DO THEY UNDERSTAND THE DIFFERENCE? DO THEY NEED TO KNOW?



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** What are the key issues associated with Cross-Border healthcare?*

** Patients affected by a rare disease and his family are very vulnerable*

Getting to the specialist they need, should not be a coincidence



Accreditation of Centers of expertise in MS



European Reference Networks

Opportunities for patients:

- ❖ **Shorter time** to get proper diagnosis
- ❖ Confirming clinical diagnosis
- ❖ **Structured** way of having access to expertise needed in management of the disease
- ❖ **Clear pathways** – **traveling patient** only when wanted or necessarily needed



- **Future therapies**
- **Access to more Research projects & clinical trials**

Objective:

- To facilitate the access to information of patients with rare diseases

It is a resource for

- * **patients** with rare diseases,
- * their **families**,
- * **specialists** involved in the diagnosis and management of these diseases.



Why are people calling?

To get information about:

- their rare disease
- procedures of diagnoses and treatment
- how to get in contact with specialists
- how to get in contact patient organizations
- available social and medical services on local, national or international level
- or just to talk to somebody is willing to listen, who can understand...

* Projects for collaboration

RONARD facilitates collaboration:

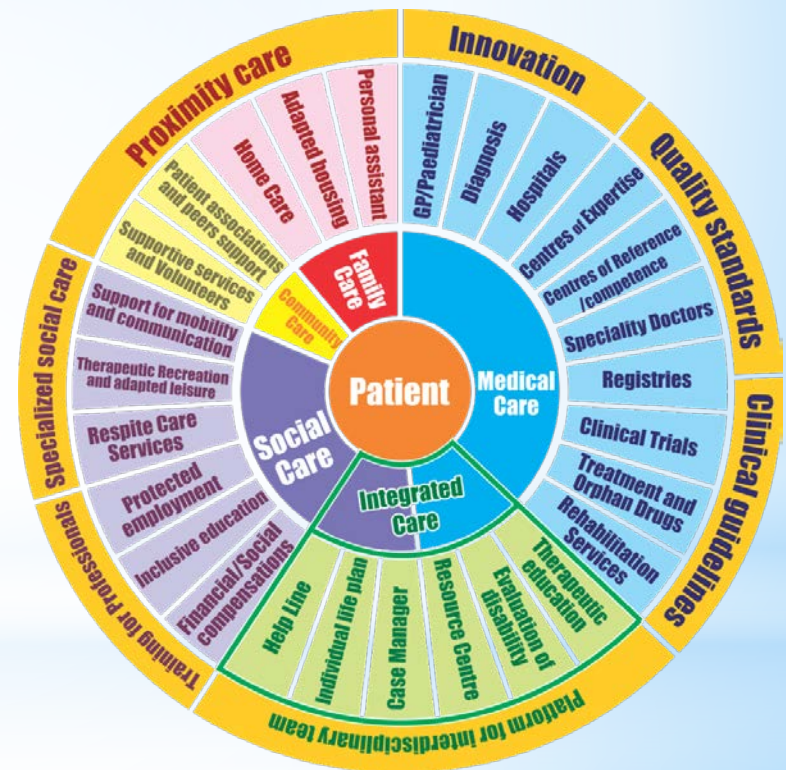
- * Between all stakeholders to have a proper pathway for patients with RD
- * For shared and structured information about available possibilities/ interventions / treatments
- * For shared guidelines for patient management
- * For proper services at local, national and international level



Why?

- Because we want to avoid accessing services by luck
- To integrate rare diseases patients in existing social, medical and educational services
- **Equal access to services!**

* This is why we need networks of services!





Sabina G.

- * "My diagnose is : **generalized dystonia**
- * When my first symptoms appeared, most doctors said **it is only my impression** that my head won't stay still. A patient with dystonia has better and worse days, this hardens the evaluation and the integration process.
- * The signs appeared early (fainting, tremor of the hand at writing, emotional changes), at the age of 16.
- * First I was diagnosed with epilepsy, and after eight years of treatment, only in 2004, at age 24, I was told that **I never had this disease**. I suffered **in fact** from **dystonia**.
- * 2004 - 2007 were hard years of suffering, I ended up in a situation that I couldn't do anything alone. I couldn't even walk because of the involuntary movements of the body.
- * As my condition was very severe, I had a neurosurgery procedure in 2006, without success. I learned only later on, that my chance of getting better was the **deep brain stimulation (DBS)**, procedure **not available at that time in the country**.



- * I wrote many letters to many clinics abroad. I got a positive answer from a clinic in Hannover, Germany and in December 2006 I went for my first consultation there. After a detailed evaluation, I was proposed for DBS. The intervention implied high costs, and I could not afford it. But it seemed to be my only chance.
- * Thanks to many kind people - youth, artists, colleagues, friends and well known people of my city - I could have the intervention in February 2007.
- * After I had DBS surgery in Hannover, Germany, my quality of life was much improved, but it is essential to have proper rehabilitation services, for which we do not have specialists and which cost a lot.
- * After the first intervention, I needed to go to Germany for reevaluation every 3 months.
- * Having DBS and strengthening the muscles - body building, aerobics - helped me because in 2009 my posture was O.K. I recovered 90%. I could control the posture of my body and the pain was less.

- * The first battery lasted barely a year and a half, so I had to replace it. For this I got **E112 form**. The second battery lasted a year and eight months and the third two and a half years.
- * **I had great results**. I could control my posture. I had no pain anymore. I noticed an almost complete recovery of the trunk and neck posture until the February of 2011 doing these therapies together and having the device. I had seven good years, but unfortunately the disease advanced.
- * In November 2012 I had a replacement of both IPGs with one Rechargeable ACTIVA RC and my condition remained stable until SEPTEMBER 2014 when I had a high fever and a terrible coughing and started having pneumonia very often my **dystonia symptoms were worsening**.
- * I was sent in 2015 for further evaluation at a specialized clinic in Milan then again in Germany at Hanover. I was finally proposed for a **new implant**. It was performed in January 2017. The intervention was more complicated.
- * I am still in recovery, not well yet, but I have to be patient and after three months I have to go to a reassessment.





Since I started to have symptoms in 2004, I had to face many difficulties:

- * - getting a diagnosis was difficult because symptoms were interpreted as psychiatric problems even though the problems are neurological
- * - the personal feeling of embarrassment of not being able to control the movements of the body can lead to isolation and/or depression
- * - I need a multidisciplinary teams to follow my health, as I met abroad (in Hannover), where I got my DSB surgery
- * - I need permanent rehabilitation services but for the moment what I can find is without medical supervision or specific knowledge about the disease
- * What helps in increasing quality of life is a series of therapies, ex. physical therapy, swimming in warm water, massage, reflexology, horse therapy, psychotherapy, mental relaxation therapies, ergo therapy. We also need a nutritionist, to avoid foods that increase spasticity."



Dóra A. - past

- * I was 14, it was 2000, and my arm started to hurt. In Romania **the diagnosis wasn't set up correctly**, wasn't set up at all. Thanks to the fact that in my parents sent my x-ray to Hungary for my relative who was a nurse in an oncologic institute, asked me to go as fast as I can, because the prior diagnose was bone cancer. While doing my diagnosis in Budapest, my parents started to search for funding my treatment. At that time The Helping Right Foundation (Segito Jobb Alapítvány) had a programme for **funding treatments unavailable** in Romania, so we reached them.
- * I needed an operation to remove the tumor, which couldn't been done in Budapest only with amputation, so I needed to go more west. My oncologist from Budapest arranged my operation in Vienna, in the AKH (Allgemeines Krankenhaus), where I get my humerus prosthesis also my tumor was removed totally. **The funding for the operation though was a problem of my family**. With the help of a lady (acquaintance of my oncologist) the city of Vienna overtook the whole cost.
- * The **costs of the controls of the prosthesis** also were a problem of my family. In that matter communication with the Ministry of Health of Romania started, but very **few costs were overtaken** by them, and the communication could be described as very slow and very bureaucratic.



Dóra A. - now

- * **After 16 years** some parts of my prosthesis have to be replaced. I went for control directly back to the AKH, they are the competent of managing this operation. One single document was required from me, so that my whole costs of operation to be covered by the Romanian health insurance. This is the **E112 form**, which allows treatments within the EU what cannot be performed in Romania due to the lack of logistics, material or even medical proficiency (though this is not really acknowledged).
- * So I had to prove than my operation cannot be done in Romania, so the Health Insurance House could release the E112 form for me. **I, personally think the method is transparent and not bureaucratic. My operation will be on 2nd of March, 2017, so fingers crossed!**

*Did the rare disease patient quality of life improve in Romania?

Yes, thanks to

- *Improved service provision*
- *More diagnostic procedures treatments and therapies available in National programs / other programs (NPRD, Regional Genetic Centers)*
- *Cross-border healthcare*
- *Centers of expertise / European Reference Networks*

....but there is still a long way to go...

- *To fulfill all the needs rare disease patients have*
- *To raise their quality of life*
- *To have **continuous multidisciplinary care available** for everybody*



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Thank you!