



Rare Diseases Day Policy Event at the United Nations *Resource Centres for Rare Diseases – NoRo Center Romania*

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EURORDIS.ORG

There is always a personal story



1985 2019
2014

De ce noi?



Google Photos



It's all the
Little moments
that make
life a ...
BIG adventure!



Our daughter's care pathway



1. Pre-Diagnosis: Poor muscle tone; Distinct facial features
Poor sucking reflex and difficult feeding; Almost no cry.

1st Symptom: Hypotonic and difficulties in feeding;
Poor responsiveness; Sitting up and walking — later Hypotonic and difficulties in feeding;
unusually tired, respond poorly to stimulation, have a hard time waking up or have a weak cry.
Sitting up and walking later.

Ideally:
Improvement of muscle tone
Special technics for feeding and stimulating exercises
Special technics for feeding

Medical treatment and early rehabilitation intervention

3. Diagnosis
Unusual food-seeking behaviors, hypogonadism
Mild intellectual disability;
Delayed motor development
Speech problems
Small hands and feet
Thick saliva and dental problems associated
Overweight

Note: Medication & rehabilitation
Support for education & school integration & inclusion into community

Ideally: GH Treatment
Sex hormone treatment
Permanent rehabilitation program; Education and work
Independent life skills

4. First Treatment
Delayed in motor development
Rehabilitation program, 3 weeks every 3 months

5. Surgery (No)
Scoliosis (gymnastic)
Physiotherapy

Ideally:
Improvement in general health and reduced weight

Not always easy!!!

6. Follow-up Care...
Delayed in motor and intellectual development, behavior problems,
Speech problems
Small hands and feet,
Overweight and sleep apnea;
Medication & rehabilitation
Support for education & school integration & inclusion into community; a proper job and qualification

Ideally: Maintaining the weight, even reducing it
High school, passing exams and training courses to find a proper job;
Integration into a Center for occupational therapy and than a supported job;
Friends, community, understanding and care at NoRo Center;

Complex & often invisible

Missunderstanding &
Discrimination

“The difficulty lies in the **impossibility of carrying a routine** (...). The problem arises when **one day you appear completely healthy, the next day you are sick**, and two days later you appear completely normal again. Many people find it difficult to understand the disease and the process, and the absenteeism that entails” A.H. Romania

”



Time Burden!!!

42% of patients & carers spend **more than 2h/day** on **illness-related tasks**

- ☐ Symptoms vary & can be **invisible** in many ways
- ☐ >90% of RDs have NO TREATMENT
- ☐ Support in coordination of care is a need!
- ☐ Heavy time burden for patients & carers

Hygiene



Administration of treatments



Helping with house
chores



Helping patients to
move



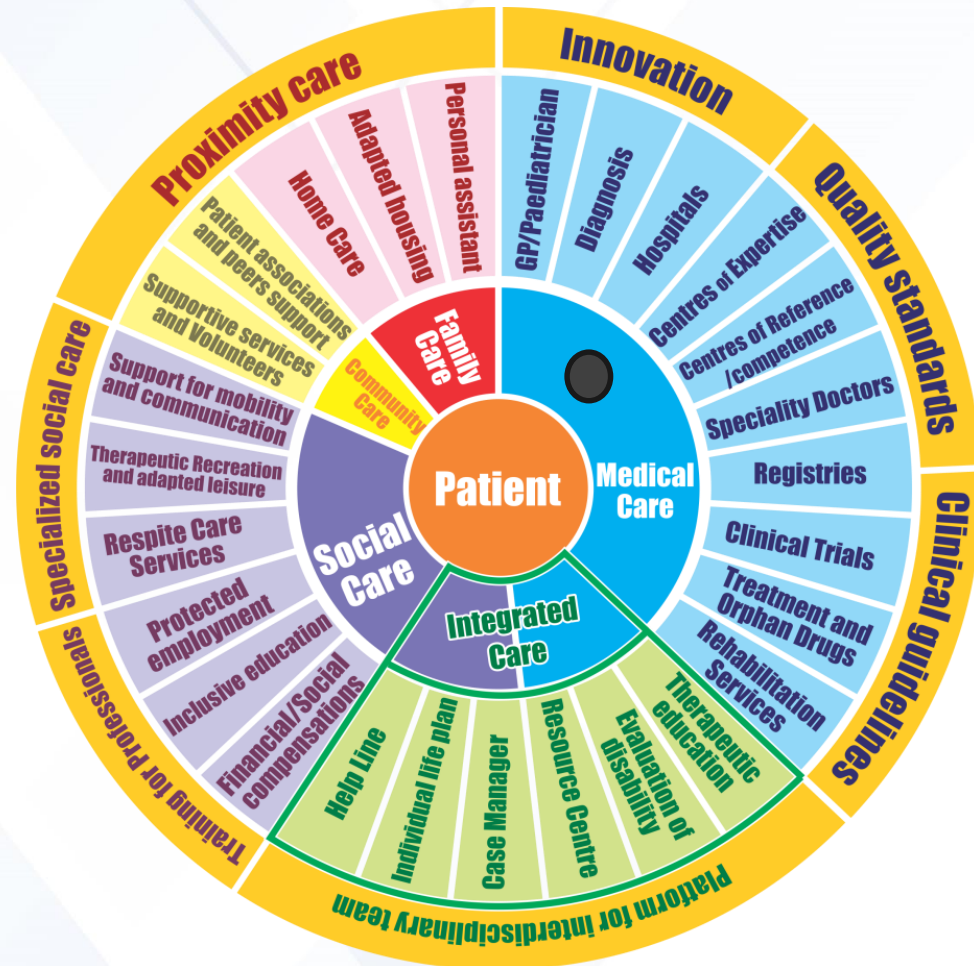
Why do we need integrated care?



Health



Social



Specialised Social Services

Specialised Social Services for RD in EU mapped in 2014



94 Services

22 Countries

Information on activities, target population, contacts, etc.

-  Therapeutic Recreation Services
-  Respite Care Services
-  Adapted Housing
-  Resource Centres

Poster award at ECRD 2014 Berlin:
"Integration of Rare Diseases into Social Services"

The map is available at www.eucerd.eu and www.eurordis.org

Romanian actions in the field of rare diseases

"RD are recognized as a priority for the health care system"



**Alianța Națională
pentru Boli Rare
România**



Patients and professionals as equal partners

NCRD since 2013

2016
MoH Order for
CoE

2014
NPRDs included in
NH Strategy

2008–2012
NPRDs

2008
Partnership
MoH – RONARD

RD School for journalists
2012 - 2018



Definition of resource centers

☐ Resource Centre services include information and guidance services, training courses, provision of information concerning social benefits, and documentation and research services. They can also include daily support therapies, medical/psychological consultations and therapeutic recreation activities.

☐ *in partnership with Centres of Expertise*

☐ *part of a Centre of Expertise themselves*

☐ Resource Centres could be defined as a ‘one-stop shop style’ service for rare disorders.

RPWA brief history:

- ✓ RPWA was established in 2003
- ✓ First international meeting in 2004 in Zalau
- ✓ Attending the 5th International PWS Conference in Christchurch, New Zealand
- ✓ Opened the Center for Information about Rare Genetic Diseases in 2005
- ✓ 1 Session about PWS at the Balkan Congress for Endocrinology Bucharest
- ✓ The 6th International PWS Conference in Cluj- Napoca, June 2007
- ✓ 1st National Conference about Rare Diseases, 1-3 November 2007
- ✓ 1st European RDD – 29 February 2008 –NPRD partnership with the MoH;
- ✓ Renewed partnership 2013; 2014 – National Strategy for Health;
- ✓ 1st National Program for Rare Diseases - 2008;
- ✓ May 2009 – 1st EE Conference on PWS, Timisoara;2010 Zalau;
- ✓ Balkan Congress for RD, Cluj – June 2009
- ✓ Implementation of the NoRo Project: 2009 -2011
- ✓ 2010 – EUROPLAN1 and annual conferences or workshops;
- ✓ 2010 – The 2nd EE Conference on PWS
- ✓ 2011 – June 28, Official opening of the Center
- ✓ **ExpertRARE** Project – to organize a center of expertise
- ✓ **INNOVCare** (Oct. 2015- Sept 2018)

❑ Projects implemented in partnership with Frambu:

- **2011 – Opening of NoRo Center** (*Norwegian-Romanian, partnership for future in rare diseases*)
- AD AUGUSTA PER ANGUSTA
- NoRo- Frambu, partnership for the future
- TEAM – Together, everybody achieve more
- DocumentRARE – **2017**

- **RareResourceNet – to be continue!!!**



NoRo – a Resource Centre for RDs

Since 2011- addressing health, psychological, social, educational, and daily life needs, abilities and wishes of people with a RD and their carers + case management in INNOVCare project;

Opened thanks to a EEA & Norway grant
and our partnership
with FRAMBU !



NoRo Interdisciplinary team: 24
Medical department – 2 doctors, 2 nurses;
Administrative staff: 4
Therapists - 16

Day care center: 60 beneficiaries

- Children with rare diseases and with autistic spectrum disorders**
- From Zalau and county of Salaj**
- Individual and group therapy**
- Summer camps and therapeutic weekends for families;**

“Trial for flight” Group for independent life skills
12-14 young people with Intellectual disabilities and RD

- From city and county**
- Individual therapies, social, medical and psychological support, occupational therapy and training for life skills, summer camps and therapeutic weekends**

RD Groups / 5 days/ month

- 12-14 Patients with the same RD**
- From all over the country**
- therapies, social, medical and psychological support, support groups, registry and videoconferences;**

Therapies provided by NoRo
 Ergo therapy
 Massage
 Hydrotherapy
 Kinetotherapy (MIRA)
 Speech therapy
 Behavior therapy
 Deep oscillation
 Electric Therapy
 Psychological counseling
 Genetic counseling

Community support network
2 weeks/ year for home visits
Information campaigns



Nutriția în bolile rare

Învățați principalele categorii de alimente necesare pentru o dietă sănătoasă. Învățați cum să evaluați starea de nutriție a copilului, care sunt principalele boli genetice de metabolism, care sunt cauzele și principalele modalități de prevenție și tratament al malnutriției și al obezității.

Tip curs: OnLine Durata: 18 ore



Managementul în boli rare

Învățați cum să depistați și să urmăriți un pacient cu o boală rară, cum să efectuați consultul genetic predictiv și de specialitate, care sunt modele de transmitere a eredității normale și patologice în generații, cum să interpretați unele fenotipuri în relație cu suspiciunea de boala genetică, care sunt posibilități terapeutice și profilactice în bolile genetice, care sunt principalele aspecte de bioetică medicală în bolile genetice rare, în lumina legislației europene.

Tip curs: OnLine Durata: 18 ore



Asistent personal al persoanei cu handicap grav (cod COR 513304)

Învățați cum să asigurați îngrijirea și protecția adultului cu handicap grav sau accentuat la domiciliul acestuia, precum și cum să asigurați condiții pentru implicarea persoanei cu handicap în activitate, în situații de viață reale, pentru participarea la viața comunității și exercitarea tuturor drepturilor cetățenești.

Tip curs: Bleded Durata: 90 ore



Animator socio-educativ (Cod COR: 511307)

Învățați cum să aplicați diverse practici și tehnici de animație în grup, în cadrul unor proiecte generale de animație socială, sportivă, educativă destinate unui public specific: copii și tineri instituționalizați, copii și tineri din cadrul centrelor de urgență, copii ai străzii, copii și tineri cu dizabilități, copii și tineri aflați în dificultate, etc.

Tip curs: Bleded Durata: 54 ore



Pedagog de recuperare (Cod COR: 333003)

Învățați metodele și tehnicile de îngrijire și ocrotire a persoanele cu cerințe speciale, prin proiectarea, organizarea, desfășurarea, monitorizarea și evaluarea activităților de stimulare și recuperare specifice. Învățați cum să stimulați și să formați deprinderi de autonomie personală și socială la persoanele cu cerințe speciale, cum să promovați și să respectați drepturile și interesele beneficiarului, cum să acționați pentru incluziunea acestuia în comunitate.

Tip curs: Bleded Durata: 54 ore



Lucrător prin arte combinate (cod COR 513905)

Învățați cum să oferiți servicii de stimulare/dezvoltare prin mișcare, artă, muzică, joc, teatru pentru persoane cu cerințe speciale, promovând și respectând drepturile beneficiarilor de servicii sociale dându-le valoare ca indivizi și ajutându-i să capete încredere în sine.

Tip curs: Bleded Durata: 54 ore



Evaluator de competențe profesionale (cod COR 241219)

Învățați cum să planificați și să organizați activități de evaluare de competențe, să înregistrați și să raportați rezultatele evaluării, să efectuați evaluări, să analizați informațiile obținute în timpul evaluărilor și să luați decizii privind performanța profesională a celui evaluat.

Tip curs: Bleded Durata: 54 ore

CURS GRATUIT: Metode și tehnici de integrare a persoanelor cu dizabilități



Învățați cum să planificați, organizați și să desfășurați activități de socializare cu scopul integrării în comunitate a persoanelor cu dizabilități, indiferent dacă acestea sunt generate de o boală rară sau nu.

Tip curs: OnLine Durata: 2 ore

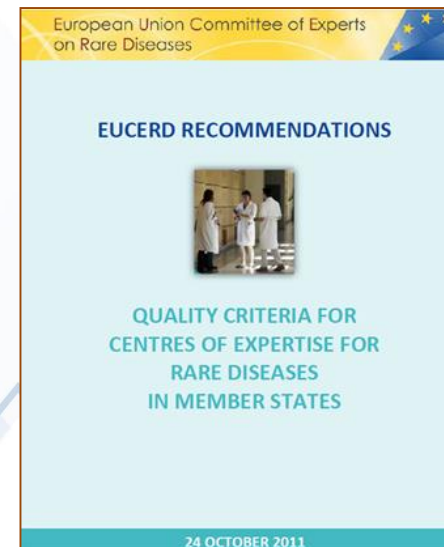
Integrated care provision (NoRo)

Medical services in contract with NHI – CoE Accreditation since 2017 Part of RO-NMCA ID & ERN ITHACA	I N T E R D I S C I P L I N A R Y	Different specialties, 24 permanent staff: therapists, administrative staff, medical doctors: pediatric psychiatry and geneticist;
Individual therapies:		Psychological intervention, hydrotherapy, medical gymnastic (MIRA), speech therapy, behavior, sensorial, electric therapy and social counseling;
Group therapy and interventions: Case management!!!		Psychological counseling, occupational therapy, ergo therapy; therapeutic weekends, summer camps, etc.
Help Line		Part of the Eurordis network of Help Lines
Trainings and online platform: www.edubolirare.ro & 2 magazines (www.rjrd.ro)		Diseases management, “Trial for flight”- life skills, trainings for professionals, including RD School for journalists;
Registry		NoRo electronic Patient registry
Information		Website, Facebook, blog, magazines, Radio NoRo – the voice of patients;

Quality in our services

QUALITY CRITERIA

- ✓ Demonstration of a multi-disciplinary approach
- ✓ Capacity to participate in data collection – patient registry
- ✓ Organisation of collaborations to assure the continuity of care between childhood, adolescence and adulthood – Trial for flight
- ✓ Continuity of care between all stages of the disease: diagnostic, treatment, rehabilitations, HCP, social services, education and social economy
- ✓ Links and partnerships with other CoE at national, patient organisations, Resource centres for rare diseases at EU level
- ✓ Consideration of e-Health solutions: training platform, videoconferences, mapping services, elaboration of guidelines, factsheets and documents
- ✓ Sharing resources and best practice with other HCPs
- ✓ Creating networks at national level & defining patient pathways



[www.participare.ro](#)

Cautare în site

EN | RO

ACASĂ • DESPRE NOI • BIBLIOTECĂ • REȚEAUA DE RESURSE PENTRU BOLII RARE • EVENIMENTE • CONTACT

Asculta Radio NoRo - Vocea Pacientilor

ParticipRARE este un website al Alianței Naționale pentru Boli Rare România (website-ul alianței este www.boliareromania.ro)

Colaborare pentru managementul bolilor rare
ColaboRARE

Proiect finanțat în cadrul Acordului pentru Centri de Referință - Runda 2 în cadrul Instrumentului Financiar al Spitalului Economic București-Fondul ONG-Congresul al DISPOZIȚIEI, CAPACITĂȚI ONG-uri, REȚELE ȘI CALITATE Subcomponenta S.4. Sperăm proiectul nostru să ajute la o mai bună înțelegere și soluționare în problemele dintr-un timp scurt.

IUNIE 2018
EVENIMENTE

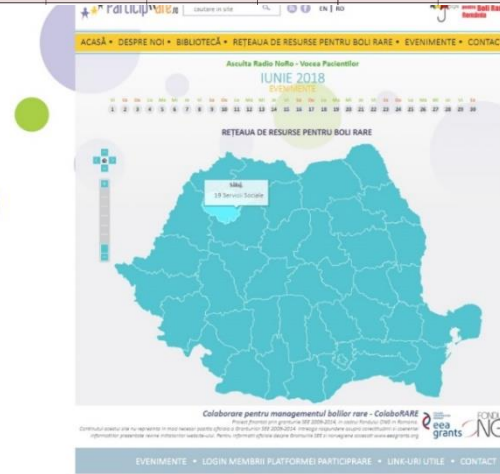
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ParticipRARE is a platform that aims to facilitate the involvement, collaboration and communication among all interested parties in the field of rare diseases.

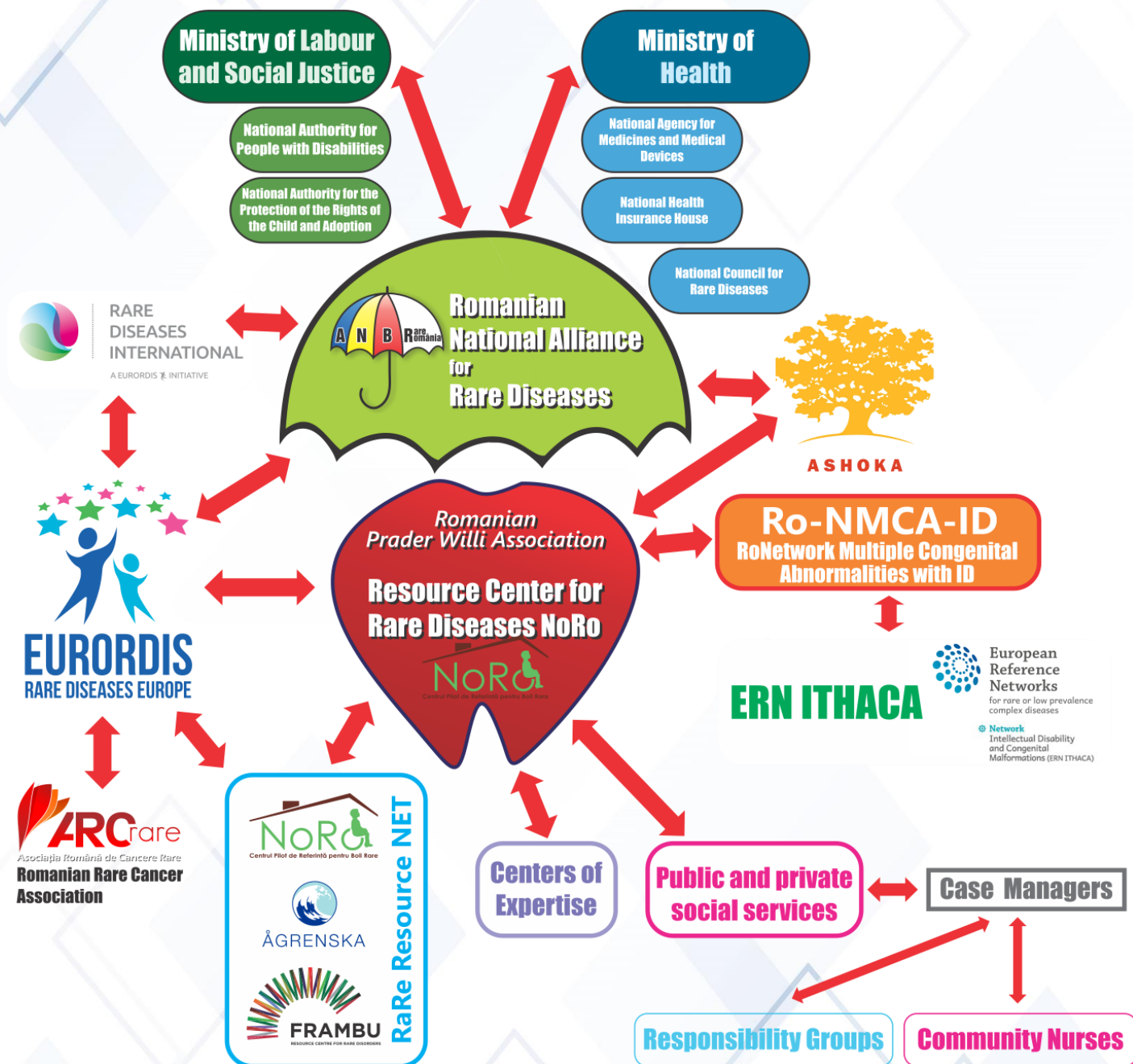


HelpLine NoRo

A	B	C	D	E	F	G	H	I	J	K	L	M	N	O	P	Q	R	S
Columns	Date	Orphanet code 1	Name of disease	Age of Inquirer	Name of Inquirer	Age of patient	Name of patient	Gender of Inquirer	Category of Inquirer	Type of contact	Purpose of contact	Response given	How did inquirer hear about help line	Region/Country	Refuel/citas	Score rating	Time taken	date de contact
NoRo HelpLine Cod:PO-01-AZ19 2018																		
Respondent	Date of call/email received	Orphanet code	Name of disease	Age of inquirer	Name of inquirer	Age of patient	Name of patient	Gender of inquirer	Category of inquirer	Type of contact	Purpose of contact	Response given	How did the inquirer hear about your help line?	Region/Country		Score rating	Time taken to manage query from start to finish	
Please mention an identification for the respondent. This can be a code, not necessarily the name of the person	Format: dd/mm/yyyy		Please refer to www.orpha.net codes of rare diseases	Please enter the age of the inquirer, not the age of the patient.		Please enter the age of the patient, not the age of the inquirer		Please mention the gender of the person who called/sent an email		Please mention if the enquiry was done via one of the following media:	Please mention the reason why the person contacted you (several answers possible)	Several answers possible		Please mention the region of inquiry, not the city of residence. If from another country, please mention country and region		How do you evaluate the quality of the response you gave? 0 -10, 10 being the best score	How long did it take to reply? (in minutes)	
				<2		>2		Male	Patient	Letter	Information on disease	Provide contact with relevant organization	Relative					
				2-4		4-6		Female	Children	e-mail	Specialist/Genip	Provide information on how to create an organ	Professional					
				5-9		5-9		Unknown/other	Relative	Contact with other patient	Provide info on disease and care	Other persons						
				10-14		10-14			Other relative/friend/partner	Telephone	Support	Provide information on scientific literature or studies						
				15-19		15-19			Professional		Legal advice	potent cure						
				20-29		20-29			Student		Orientation to expert	flyers & posters						
				30-39		30-39			Association		Association	Provide information on access to treatment or therapies						
				>40		>40			Unknown		Follow up	Provide information on cross border care						
				Unknown		Unknown			Unknown		Signposting	Psychological support						



Networks & Partnerships:



https://www.youtube.com/watch?v=678_ul0B738&feature=youtu.be

*Rare Disease Day 2019 is an opportunity to be part of a **global call for action** on policy makers, healthcare professionals, and care services to better coordinate all aspects of care for people living with a rare disease!*

Thank You

