Information factsheet on rare diseases

EMRaDi
EUREGIO MEUSE-RHINE — RARE DISEASES

TO RAISE THE AWARENESS OF PRIMARY CARE PRACTITIONERS TO BETTER DETECT POTENTIAL RARE DISEASES

among their patients and to help position themselves and support their patients and relatives in their pathway

RARE DISEASES are often chronic, highly complex, progressive and severely disabling, frequently affecting life expectancy and generating specific care needs.

Due to their low prevalence, little is known about most rare diseases. As a result, they are difficult to diagnose and their symptoms under-recognized in healthcare and social systems.

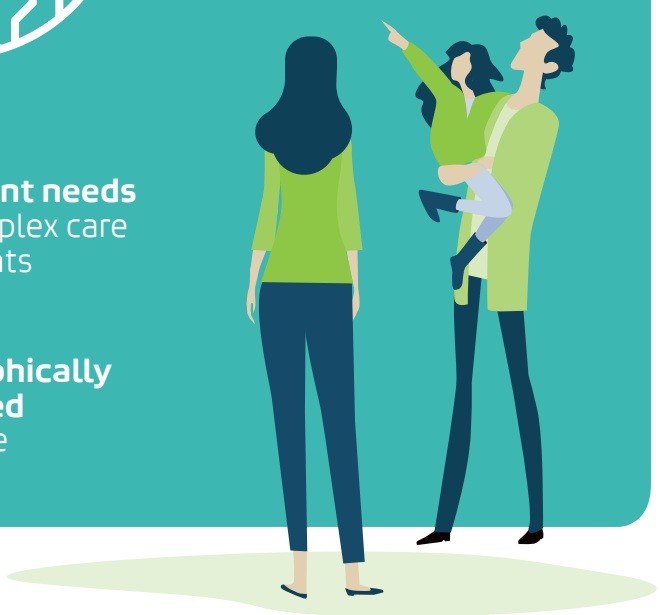
30 MILLION CITIZENS AFFECTED IN THE EU

6000 - 8000 different rare diseases

For each RD, **fewer than 1 out of 2000** people affected

Important needs and complex care of patients

Geographically scattered expertise



PRIMARY CARE PRACTITIONERS - GENERAL PRACTITIONERS (GPS) AND PAEDIATRICIANS

THEY ARE VERY IMPORTANT FOR PATIENTS WITH A RARE DISEASE AT VARIOUS STAGES THROUGHOUT THE PATIENT PATHWAY.

They usually are the first medical contact for patients suffering from health complaints without a diagnosis. They have a very good overview of the overall personal and family situation and are seen at **the start of the diagnostic process and referral**.

After the diagnosis, they accompany the patients and their relatives in the rare disease **follow-up** and in their more common day-to-day medical and social care.

They have a central role as they converse with patients and their relatives and liaise with all the professionals and counterparts involved throughout the patient pathway – medical specialists, other healthcare providers and health professionals, health insurance funds, patient associations, etc.

Only a few RDs can be diagnosed through clear symptoms, most of them leading to very unspecific and generalised health complaints like fatigue, systemic pain and/or dizziness.

DETECTION OF A RARE DISEASE AND REFERRAL TO A CENTRE FOR RARE DISEASES

As early detection of rare diseases and referral to Centres for Rare Diseases is crucial for the patients, these eight questions were created[1] in order to provide you with the necessary awareness to consider the possibility of a potential rare disease:

1. Does the patient show acute or chronic unexplainable, incoherent or unspecific **symptoms** (especially at a younger age)?
2. Does a **family history** exist?
3. Have there been a **number of sick periods** because of differing or same symptoms?
4. Is there a history of senior consultations of **different medical specializations** without a satisfactory result?
5. Are there **pathological or borderline results** that are non-conclusive on first impression?
6. Has there ever been **suspicion of psychosomatic aetiology**?
7. Have there been **periods of illness** in previous years?
8. Are any known **specific exposure scenarios** (concerning nutrition, hobbies, living situation, animals, journeys or relating to work)?

If you can answer most of the questions with "yes" and if your intuition confirms it, it might be advisable to consider the possibility of a **Rare Disease** and to refer your patient to a health specialist or geneticist, preferably connected to a designated Centre of Expertise for Rare Diseases who is better placed to come to a diagnosis.

[1] The 8 questions have been written by RD experts who have participated to the EMRaDi project*.

SUPPORT OF PATIENTS LIVING WITH A RARE DISEASE AND THEIR RELATIVES

When you accompany a patient with a rare disease and their relatives, you are advised to pay particular attention to the following aspects:

- **Your central position in the medical aspects of the pathway:** Your network with local professionals and specialists in Centres of Expertise facilitates access to care, including day-to-day treatment (access to speech therapists and physiotherapists, for example). Accompanying patients with their common (medical) complaints requires collaboration and communication between all professionals involved in the pathway – including a potential liaison nurse/care coordinator.
- **Holistic view:** Having a more holistic approach to cover the 360° spectrum of the medical, practical, psychological and social needs of your patients and their relatives is very important.
- **Arranging a psychological follow-up for the patients and their relatives/informal caregivers** is of high importance not only before, during and after the diagnosis phase, but also during the transition through life stages. Due to the extensive needs of the patients and their relatives, you could offer systematically.
- **Information for your patients:** Talking with your patients is crucial to learn how and at what pace your RD patients prefer to be informed and supported as every patient has different needs and expectations. Support is available, such as the EMRaDi Project* Factsheet for Patients and Relatives, with practical advice and interesting sources of information, not to mention key stakeholders to contact, including patient associations.

If your patients and their relatives need:

- **Information about the disease itself**: Orphanet is a European database and a unique resource which gathers knowledge on rare diseases and their diagnosis, care and treatment. It provides you with more information on medical descriptions of rare diseases and directories towards professionals and institutions, medical laboratories, expert centres and patient associations: www.orpha.net.
- **Information on reimbursement and the management of their day-to-day life**: Health insurance funds, RD health experts and social services are a good source of information and support to help your patients with their access to care and reimbursement, and with the management of their day-to-day life. In Belgium, health insurance funds and their social services can also often coordinate or cooperate with homecare services and other social and healthcare services.
- **Information on cross-border healthcare (CBHC):** Due to the specificities of rare diseases, it might be beneficial for patients to seek specific expertise abroad. Before going abroad to receive healthcare (CBHC), consulting their respective health insurance funds first is highly recommended as they can provide you and your patients assistance to check the different possibilities, required authorisation, and conditions for reimbursement. Further information for treatment/care and reimbursement can be requested at the National Contact Points for CBHC. Each of the three Centres for Rare Diseases in the Euregio Meuse-Rhine has its own special expertise as shown in Table 1.

More information is included in the EMRaDi Factsheet for Patients and Relatives: https://www.emradi.eu/en/patient-information

CONTACT DETAILS AND EXPERTISE OF THE EMRADI CENTRES FOR RARE DISEASES

FOR MORE INFORMATION on the Centres for Rare Diseases, for accessing specific RD training sessions or for referring your patients, the 3 University hospitals that have participated to the EMRaDi project* are mentioned here. Other experts in your region can be found via:

- the European database **www.orpha.net**
- and through the health specialists and patient associations
  - in Germany, also via **www.se-atlas.de**,
  - in the Netherlands, an overview of all accredited Centres of Expertise, can be found at **www.nfu.nl** (click on "Patiëntenzorg" and then on "350" in the section "Erkende expertisecentra").

UNIKLINIK RWTH AACHEN (UKA), ZENTRUM FÜR SELTENE ERKRANKUNGEN AACHEN (ZSEA):

0049 (0)241 80-85859
zsea@ukaachen.de
▶ www.ZSEA.ukaachen.de

MAASTRICHT UMC+ (MUMC+), POLIKLINIEK KLINISCHE GENETICA:

0031 (0)43 3875855
polikliniek.klinischegenetica@mumc.nl
▶ www.klinischegenetica.mumc.nl

CHU DE LIÈGE, CENTRE DES MALADIES RARES

0032 (0)4 284 36 40
maladierare@chuliege.be
▶ www.chuliege.be

* This factsheet is part of the EMRaDi Final Report and is based on its global results. EMRaDi (acronym of "Euregio Meuse-Rhine Rare Diseases") is an INTERREG-funded cross-border project on rare diseases, the initial phase of which took place between October 2016 and March 2020 in the Euregio Meuse-Rhine. For more information, you can access the complete report at **www.emradi.eu**.

Table 1: Overview of Rare Disease Groups with special expertise at the three UH (overlap at all UHs in dark green and at two UHs in light green, as of March 2020)

UKA (Aachen)	MUMC+ (Maastricht)	CHU (Liège)
Neurology [1]	Neurology	Neurology
Haematology	Haematology	Haematology
Liver and gastrointestinal diseases		
Skeleton	Skeleton	Skeleton
Nephrology		Nephrology
Inflammatory diseases (children)	Inflammatory diseases (adults)	Inflammatory diseases (children and adults)
Retinopathies		Rare eye diseases
Syndromal diseases and infantile breathing regulatory diseases	Syndromal diseases	Syndromal diseases
Rare allergies and skin diseases		
Cardiomyopathies and keratopathies (research only)	Cardiomyopathies and keratopathies	
Cancer	Cancer	Cancer
	Genodermatoses	Genodermatoses
Metabolic disease is partly covered in the other topics	Metabolic diseases, especially galactosaemia	Metabolic diseases
		Endocrine diseases (adults and children) [2]

[1] As 8,000 rare diseases exist, the listed groups indicate focal fields. This does not mean that, for example, all rare neurological diseases are treated at the respective hospital.

[2] This field is partly covered through the other University hospitals too.

Dutch information on various rare diseases for GPs located in the Netherlands: www.zichtopzeldzaam.nl (click on "docs" then on "filtreren" and select "huisartsenbrochures")

E-learning modules: www.huisartsengenetica.nl (click on "Achtergrondinformatie / Scholing" and then on "Nascholing Erfocentrum").

Publisher: Mutualité chrétienne Verviers-Eupen (MCVE) – Michel Halin – Rue Lucien Defays 77 – 4800 Verviers – Belgium – March 2020 edition

With the support of Interreg Euregio Meuse-Rhine European Regional Development Fund, European Union, provincie limburg, Wallonie, Ministerium für Wirtschaft, Innovation, Digitalisierung und Energie des Landes Nordrhein-Westfalen

Mutualité Chrétienne, Solidaris, Socialistische Mutualiteiten, VSOP, CHU de Liège, Uniklinik RWTH Aachen, Maastricht UMC+, Maastricht University