FINAL REPORT OF THE EMRADI PROJECT

EMRaDi
EUREGIO MEUSE-RHINE
— RARE DISEASES

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Maastricht UMC+

CHU de Liège

Maastricht University

TABLE OF CONTENT

SCOPE OF THE FINAL REPORT

This Final Report gathers the results, advice and recommendations from the EMRaDi project, with specific information dedicated to the EMRaDi target groups:

- Patients, relatives and patient associations
- Health care providers (primary, secondary and tertiary care)
- Health insurance funds
- Decision makers

In addition to this report, the tools and detailed reports of the activities of the project can be found by visiting the EMRaDi website and then clicking on "Project" -> "Activities and results" (http://www.emradi.eu/en/project-activities). The EMRaDi partners can be contacted for more information (see "Contact details and sources of information").

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DISCLAIMER

The content of this report is based on the project's activities and reports. It has been validated by the members of the EMRaDi Steering Committee composed of project managers, project officers and doctors working for the different EMRaDi partners. The facts and views expressed in this publication are their sole responsibility and do not necessarily reflect the position of the EMRaDi partner organisations.

All the personal data that were gathered throughout the EMRaDi project were processed in accordance with the European Regulation 2016/679 (EU) on the Protection of Personal Data (GDPR). Particularly the data of the patients that were interviewed were dealt with following stricter rules described in a protocol, approved by the Ethical Committees of the 3 University Hospitals participating to the project.

CO-FINANCING

The EMRaDi project was carried out under the Interreg V-A Euregio Meuse-Rhine programme from October 2016 to March 2020 and was supported by the European Union and the European Regional Development Fund for an amount of EUR 1,687,675. The project was also co-funded by regional authorities in the three countries of the Euregio Meuse-Rhine: the Walloon region, the Dutch province of Limburg and the Land North Rhine-Westphalia. All the EMRaDi partners also contributed with their own fund.

The Interreg V-A Euregio Meuse-Rhine (EMR) programme invested almost EUR 100 million in developing the Interreg-region until 2020. This area stretches out from Leuven in the west to the borders of Cologne in the east, and runs from Eindhoven in the north all the way down to the border of Luxemburg. Over 5.5 million people live in this cross-border region, where the best of three countries merges into a truly European culture.

With the investment of EU funds in Interreg projects, the European Union directly invests in the economic development, innovation, territorial development and social inclusion and education of this region.

With the support of

INTRODUCTION

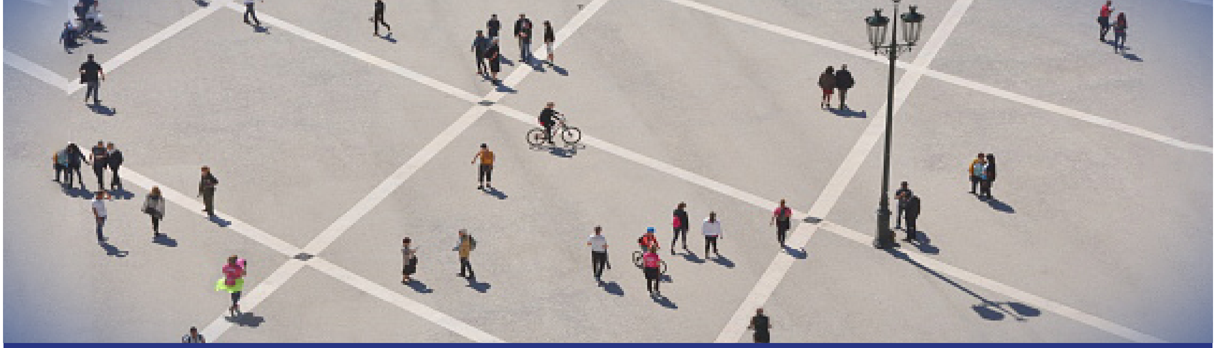
The first part of this report presents the EMRaDi (Euregio Meuse-Rhine Rare Diseases) in short – the context of this cross-border cooperation on rare diseases in the Euregio Meuse Rhine, the main objectives and the activities of the project from October 2016 to March 2020.

To better support the patient with a rare disease and to improve their pathway, different target groups were addressed during the EMRaDi project:

1. Patients, relatives and patient associations
2. Health care providers (primary, secondary and tertiary care)
3. Health insurance funds
4. Decision makers

In connection to those target groups and based on the project results, the next parts of the report present the observations, challenges, main achievements and the recommendations and pilot projects proposed to sustainably improve the quality of life of people living with a rare disease.

The EMRaDi project has also developed specific tools and information factsheets, for patients and relatives on the one hand, and for primary care practitioners (for general practitioners and paediatricians) on the other hand, which are mentioned in the achievements and available in the appendices of this Report. They aim to share practical information, advice and recommendations for these specific target groups.



1/ THE EMRADI PROJECT IN SHORT

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**

1.1/ What is a rare disease?

"Rare diseases are rare, but rare disease patients are numerous."[1]

In the European Union, any disease affecting fewer than **5** people in **10,000** is considered rare.
Approximately **5,000** to **8,000** distinct rare diseases (RDs) affect between **6%** and **8%** of the EU population, which represents between **27** and **36** million people.[2]
80% of RDs are of genetic origin.[3]
Approximately **50%** of the people affected by RD are children.

RDs are generally complex, chronic, disabling, and degenerative, often threatening life expectancy. Most of them cannot be cured and there are very few accessible treatments. Experts are few and they are geographically dispersed. Access to expertise, knowledge and information is also difficult, despite the technological advances.
Due to the specificity of symptoms and difficulty of diagnosis associated with RDs, most patients undergo an odyssey before they even know that they have a RD.

“Today, the 30 million Europeans living with a RD and their family members (often the main carers) remain a marginalised and largely invisible population, with little information about their diseases and their rights, few treatments, and a high level of psychological, social and economic vulnerability.”[4]

National-level only approaches are inefficient and ineffective; EU regional- and European-level coordination has a clear added value.
European cooperation can help ensure that scarce knowledge can be shared, and that resources can be combined as efficiently as possible to tackle RDs effectively across the EU as a whole.

1.2/ Why a project on rare disease in the Euregio Meuse-Rhine?

In the Euregio Meuse-Rhine[5] (EMR), up to 300,000 people - often children - have RDs, not including the relatives, friends and others impacted by the presence of the disease. Not only patients, but also health organizations, knowledge institutions and health insurance funds often experience difficulties in giving or getting the correct diagnosis, treatment and care, as well as administrative and financial support. RD patients normally desire medical treatment and support as near as possible to their home. In the Euregio Meuse-Rhine (EMR), RD patients might be forced to seek care across borders, within "foreign" healthcare systems. Clarification of available resources (especially appropriate treatment and financial reimbursement) is essential. Joining forces in the EMR is therefore important, despite the challenges of different healthcare systems and languages.

1.3/ What is the EMRaDi project?

"EMRaDi" stands for **E**uregio **M**euse-**R**hine **R**are **Di**seases.
The project started on 1st October 2016 and ended on 31st March 2020 with a total budget of EUR 3,372,669.

This was the first INTERREG cross-border project on Rare Diseases in the Euregio Meuse-Rhine.
This project involved a cross-border cooperation between health insurance funds, university hospitals, patient associations and a university in the Euregio Meuse-Rhine, with the following partners:

Mutualité chrétienne de Verviers-Eupen (lead partner)

Socialistische Mutualiteiten
Solidaris

Alliance nationale des Mutualités chrétiennes

VSOP

UNIKLINIK RWTH AACHEN
Zentrum für Seltene Erkrankungen Aachen (ZSEA)

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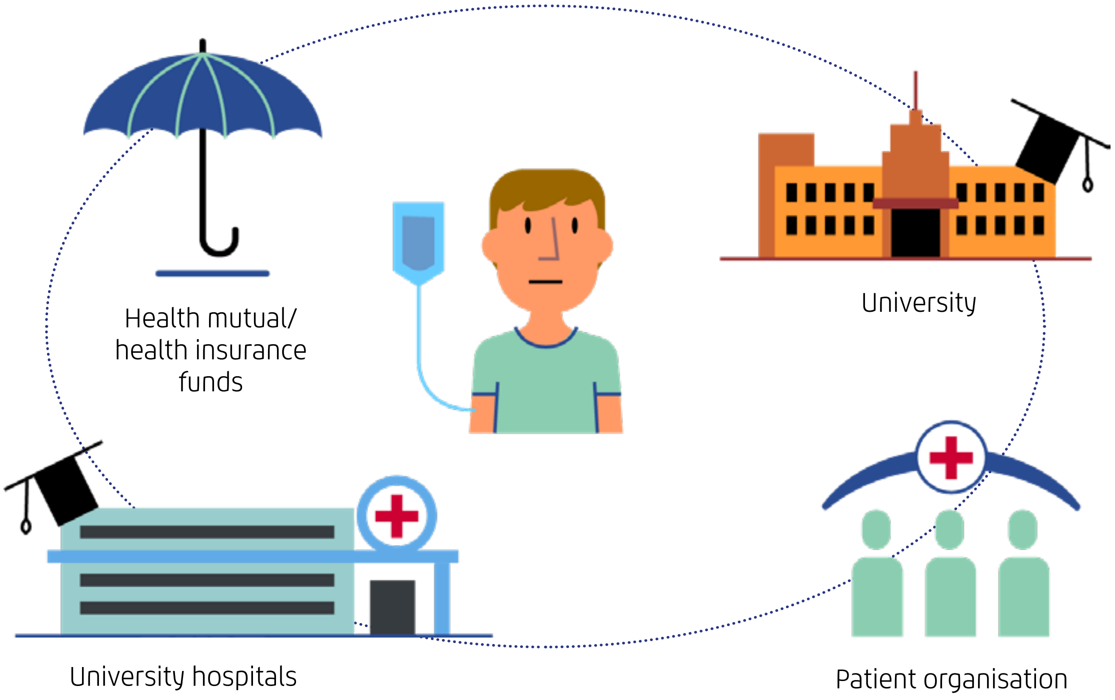
The project was **patient-oriented and cross-sectoral as the consortium of partners** included the major health players who support RD patients and their relatives in their day-to-day RD patient pathway.



Several associated partners also contributed to the development of the project objectives and activities, the umbrella associations for RD patients, RaDiOrg in Belgium and Achse in Germany, as well as other health insurance funds in the EMR, AOK in Germany, CZ in the Netherlands and MLOZ in Belgium.

The EMRaDi project aimed to carry out substantive improvements in integrated care for RD patients through **three objectives:**

1. Increase the transparency of needs and the availability of services in the field of RDs in the Euregio Meuse-Rhine (EMR)
2. Develop EMR models of RD patient pathways in order to draw up patient-oriented recommendations in synergy with national and European developments
3. Improve the network of health care providers, health insurance providers and patient organisations and raise (public) awareness of RDs

The overall long-term goal was **to improve the quality of life of these patients**.

1.4/ Definition of the EMRaDi RD Patient pathway

The EMRaDi project partners have defined the pathway of a person living with an RD as: "A rare disease patient pathway [that] encompasses all the steps from the first symptoms experienced by the patient, along his or her path through the health system towards a (hopefully) fast, successful diagnosis and including every aspect of the care – whether organizational, medical, social, psychological, legal and/or vocational – to finally ensure the best possible quality of life for the patient."

1.5/ Focus on 8 rare diseases

During its implementation, the EMRaDi project focused its attention on eight RDs from the neurological, haematological, syndromic and metabolic groups of RDs.

The objective was to use the results obtained for these RDs to help groups of patients with other RDs, and to make global recommendations and bring about developments for the cross-border healthcare of RD patients in the Euregio Meuse-Rhine (EMR).

The selection of the 8 RDs was a long process based on different criteria, principally on the cooperation possibilities and the expertise of the different project partners. Choosing specific RDs was difficult but necessary as it enabled obtaining as concrete results as possible for these diseases. More information on the selection process and on the 8 RDs can be found in Appendix 5.

The eight RDs that were selected in June 2017 are the following:

RD Group	8 RDs	University hospitals' expertise
Neurological	**1.** Huntington disease	UKA, CHU, MUMC+
	2. Duchenne muscular dystrophy	(UKA), MUMC+
Haematological	**3.** Chronic myeloid leukaemia (CML)	UKA, CHU, MUMC+
	4. Polycythaemia vera (PV)	UKA, CHU, MUMC+
Syndromic	**5.** Silver-Russell syndrome	UKA, CHU
	6. Rett syndrome	MUMC+
Metabolic	**7.** Galactosemia type 1	CHU, MUMC+
	8. Phenylketonuria (PKU)	CHU, MUMC+

The project has also gathered quantitative data in the three Euregio Meuse-Rhine countries on a longer list of 60 RDs, with the objective to produce a better picture of the situation of RD patients in the EMR border regions. The longer list is included in the EMRaDi report evaluating the number of patients with RDs (Work Package 1).

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[1] https://www.orpha.net/consor/cgi-bin/index.php?lng=EN
[2] http://ec.europa.eu/health/rare_diseases/policy/index_en.htm
[3] https://www.eurordis.org/about-rare-diseases
[4] Abstract from "Achieving Holistic Person-Centred Care to Leave No One Behind A contribution to improve the everyday lives of people living with a rare disease and their families" – EURORDIS – May 2019
[5] The principle of cross-border cooperation in the Euregio Meuse-Rhine (EMR) was legally established in 1976. Since 1991, the EMR has been a foundation made up of five sub-regions in Belgium, Germany and the Netherlands: the Flemish Province of Limburg, the Walloon Province of Liege, the German-speaking community, the Region of Aachen (Zweckverband) and the Dutch southern part of the Province of Limburg. Interreg Euregio Meuse-Rhine and the Euregio Meuse-Rhine are two distinct bodies. While the first is a Europe Commission funding programme, the second is a partnership between regions. Interreg Euregio Meuse-Rhine is a collaboration between 13 regions. See https://www.interregemr.eu/about-interreg for more information.

2/ OVERVIEW OF EMRADI ACHIEVEMENTS

From October 2016 to March 2020, through 7 work packages (WP), the eight EMRaDi partners have worked together to develop activities and reports to contribute to the optimisation of RD patient pathways, the more effective provision of relevant information and the improvement of networking and training related to RD care.

The existing knowledge on the demands and needs of RD patients was identified by way of a literature review. Moreover, estimations on disease prevalence, healthcare utilisation and costs for 60 RDs in the EMR were sought from insurance claims data and hospital records. The legal and financial framework as well as the relevance of EU and national policies on RDs were analysed. Other national provisions were assessed including the mapping of the mechanisms for reimbursement of orphan medical products and social services, the procedures and conditions for granting a disability status and the scope for telemedicine services (**WP1- University Maastricht (UM)**).

From 2017 to 2019, "real life" patient pathways were analysed through the large EMRaDi field study, which conducted interviews with 104 participants (62 patients/relatives affected by the 8 selected RDs and 42 professionals) to uncover the day to day reality of RD patients and their relatives (**WP2- National Union of Socialist Health Insurance Funds (Solidaris) and the National Alliance of Christian Health Insurance Funds (ANMC)**) from the pre-diagnosis period to the post-diagnosis period, with an in-depth analysis of

- care coordination and access to care aspects
- information aspects
- practical, financial and legal aspects
- psychological and social support aspects
- cross-border aspects

Hurdles were identified and more efficient organizational models were proposed by the Maastricht University in close collaboration with RD patients, patient organisations, healthcare providers and health insurance funds. Patient pathways were simulated to uncover bottlenecks by a modelling exercise. Improvements were suggested such as faster diagnoses by raising awareness among GPs, and case management to improve treatment coordination (**WP3 - UM**).

As a tight network between all the stakeholders is a key successful factor to improve the situation of persons living with a RD, special attention was also given to networking, information dissemination, awareness raising and training. (**WP4 – University Hospital RWTH Aachen (UKA), Maastricht University Medical Centre (MUMC+) and University Hospital Centre of Liège (CHU) and WP5 - Patient Alliance for Rare and Genetic Diseases -VSOP**)

The three University Hospitals, in particular, improved their RD structures and processes, reinforced their collaboration and exchange of knowledge, with among others, interdisciplinary case conferences for diagnosis and care, human genetics and bioinformatics, and information and training sessions for the GPs and other doctors for RDs, with the development of specific tools for them (Question list "When to think of a RD?"), and patient mobility when necessary. Study programmes were also a topic of discussion as this was perceived as an effective way to increase the awareness of doctors for RD. (**WP4 – UKA, MUMC+ and CHU**)

Throughout the whole project, the patient perspective was in the centre as the Patient Sounding Board (PSB) provided the project partners with their recommendations. (**WP5-VSOP**) The PSB was a consultative body that consisted of (patient) representatives from the selected RDs. They gave feedback and had discussions on the following topics: the organization of the EMR RDD in 2019; the 3 National Rare Disease Plans; the quality documents; the information on cross-border healthcare; the patient participation in medical research; and the concept versions of models and products developed in other EMRaDi work packages.

All the project activities were coordinated by **Mutualité chrétienne Verviers-Eupen (MCVE)**, which was the leading partner of the consortium (**WP Management**). MCVE also coordinated all communication activities and EMRaDi overarching events (**WP Communication**), with among others: the EMRaDi website (www.emradi.eu), with the help of ANMC; several publications and press releases to search for RD patients and to inform about the project's progress; and the organization of several events and meetings with the support of all project partners.

The major events organized during the EMRaDi project were:

- the EMR RD Day (28.02.2019), a participation day led by VSOP on 3 major subjects: awareness, undiagnosed patients and cross-border health care & cross-border cooperation (Diagnosis, care and research);
- the European political event (5.12.2019), with the presentation of the EMRaDi major recommendations and the synergies to have between European, cross-border and local developments (see Appendix 2 - EMRaDi Factsheet 1.0)
- the Closing Event (17.02.2020), a participative day to discuss with the audience the results, recommendations, and Declaration of Intent between the EMRaDi partners and the associated partners (see Appendix 1) related to the future cooperation to pursue in the Euregio Meuse-Rhine and beyond.

All the material and results of those events can be found at https://www.emradi.eu/en/news/cat2_news. Throughout the project, partners regularly met the different stakeholders in the field of RDs to raise awareness, exchange information on the project's progress, and to discuss recommendations and advice for the implementation of the project and the future collaboration on RDs in the Euregio Meuse-Rhine. Other developments and existing structures were taken into account such as the Orphanet Internet portal, the 3 National Rare Disease Plans and European developments, including the development of the European Reference Networks (linked to Directive 2011/24/EU[6]/ERNs).

For more information, the specific reports available per work packages are the following:

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[6] Directive 2011/24/EU of the European Parliament and of the Council of 9 March 2011 on the application of patients' rights in cross-border healthcare

WORK PACKAGES AND LEADING PARTNERS	REPORTS AVAILABLE ON http://www.emradi.eu/en/project-activities
Objective 1: increase the transparency of needs and availability of services in the field of rare diseases in the Euregio Meuse-Rhine	
WP1 Evaluation of demand and offer in the field of rare diseases in the EMR (leader: UM)	▸ **Evaluation of the number of patients with rare diseases** ▪ Report on the demands and needs of patients (literature review) ▪ Analysis of legal, financial and reimbursement mechanisms of rare diseases for treatment costs of EMR rare diseases patients
WP2 Field analysis of existing RD patient pathways (leaders: ANMC & Solidaris)	▸ **Field analysis report of existing RD patient pathways in the EMR** ▪ based on the experience of 104 interviewed participants to uncover the day-to-day reality of RD patients and their relatives
Objective 2: develop EMR models of rare disease patient pathways in order to draw up patient-oriented recommendations in synergy with national and European developments	
WP3 Development of organisational models for the management of RD in the EMR (Leader: UM)	▸ **Patient-oriented models of good practices for the management of rare diseases, with** ▪ generic and specific organisational models in border regions ▪ recommendations for national and European developments
Objective 3: improve the network of health care providers, health insurance providers and patient organisations, and raise (public) awareness of rare diseases	
WP4 Networking, training and exchanges of expertise of health professionals (leader: UKA)	▸ **Report summarizing the activities of WP4, with - amongst others** ▪ the improvement of knowledge on EMR CoE (Centre of Expertise) ▪ the increase of awareness and training of General Practitioners and resident specialist doctors ▪ the increase of the cooperation between the EMR partners ▪ the translational aspects (clinical trials) ▪ Kabuki as a model project for knowledge dissemination of a new therapy, and the exchange and training of health professionals from the CoE
WP5 Patient involvement and public awareness (leader: VSOP)	▸ **Report including the PSB recommendations, on** ▪ the 3 National Plans for Rare Diseases ▪ the quality documents ▪ the cross-border healthcare ▪ the patient participation in medical research

3/ OBSERVATION AND CHALLENGES PER TARGET GROUPS

The next pages present the observations and challenges per target groups based on the different WP activities and reports.

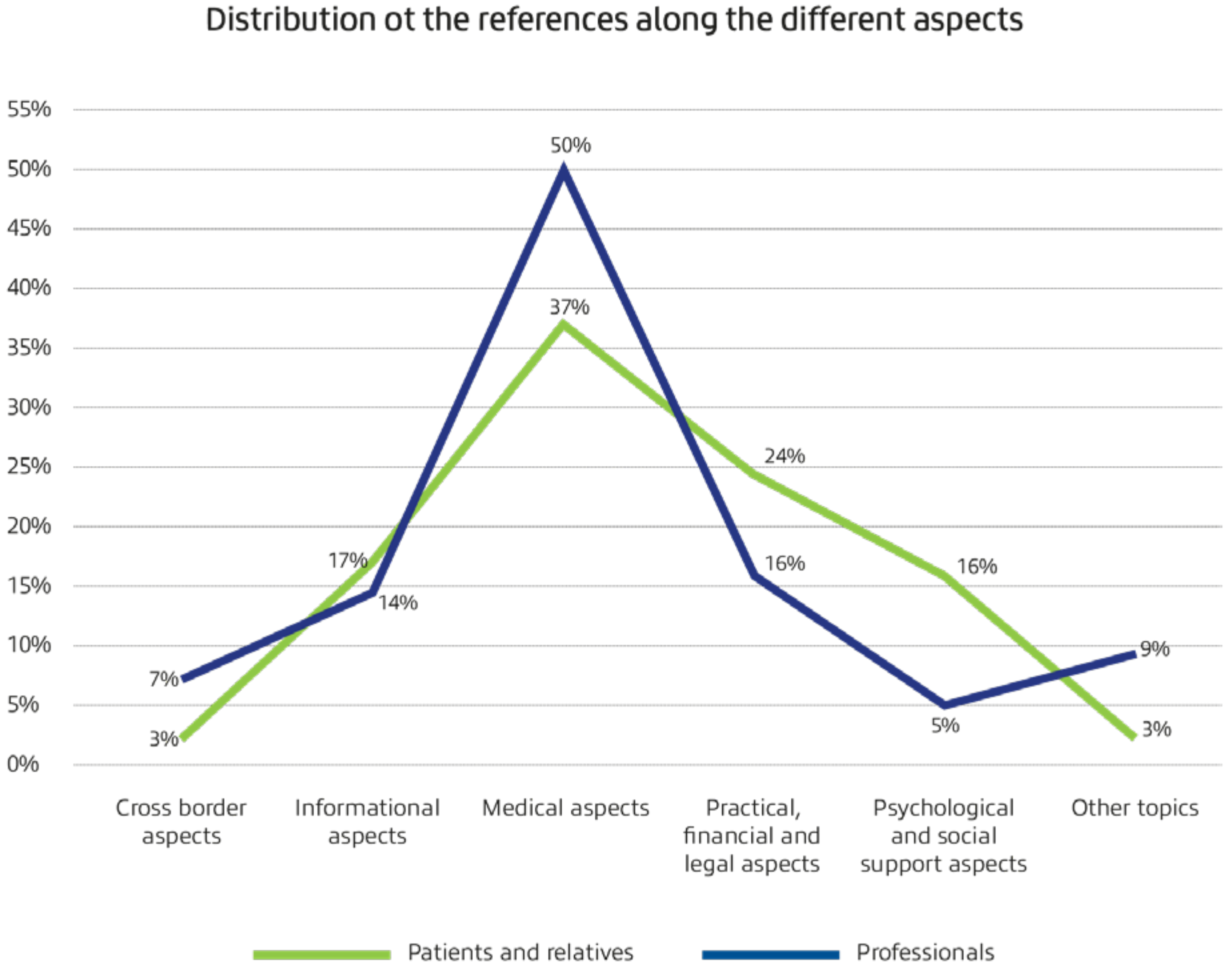
The observations and challenges were elicited throughout the project lifetime by the various work packages' activities. The field study of existing RD patient pathways in the EMR carried out between 2017 and 2019 by Solidaris and ANMC has been key here because it provided an enormous source of new insights, specifically in the EMR context. A total of 104 participants were interviewed: 62 patients and relatives for the 8 selected rare diseases, as well as 42 health professionals.

The table below illustrates the number of patients and relatives (P&R) and healthcare providers (HCP) that were interviewed for each of the 8 selected RDs in each of the 5 EMR regions.

	Province of Liège, DG excluded		DG		Limburg BE		Limburg NL		Region Aachen		Total per RD	Total to reach	Difference
	P&R	HCP	P&R	HCP	P&R	HCP	P&R	HCP	P&R	HCP			
Huntington	4	3			6	4		3			2	10	+1
Duchenne		2	3		4	2	3	1		1	16	10	+6
CML	2	0,5	1		2		1	0,5	3	0,5	0,5	10	+0,5
PV/Vaquez	3	0,5			1		2	0,5	2	0,5	9,5	10	-0,5
Rett	3	1			4	1	4	1,5			14,5	10	+4,5
Silver Russel	1						2	1,5			4,5	10	-5,5
PKU	5	1			2	1,5	1	1,5		1	13	10	+3
Galactosemia	1	1			1	1,5	1	1,5		1	8	10	-2
Other		4		1		2				1	8		+8
Total per region	19	13	4	1	20	12	14	11	5	5	104		
Total to reach	8	8	8	8	8	8	8	8	8	8		80	
Difference	+11	+5	-4	-7	+12	+4	+6	+3	-3	-3			+24

Table 1 showing the number of interviews per EMR region for each of the 8 selected rare diseases (P&R = patients and relatives) (HCP = healthcare providers)

In the observations and challenges that are presented in the next section, it is interesting to note that the patients and relatives do not place the same weight as the health professionals for each aspect. The next graphic illustrates the importance of the different aspects based on the distribution of the coded references in the EMRaDi field study.



3.1/ Observations and challenges for patients and relatives

The demands and needs of patients, upon a review of the literature, can be summarised under the following 3 aspects: **1) diagnosis, 2) information and understanding, and 3) medical care and psychosocial support.**

Dimensions	Patient experiences		Patient needs
Diagnosis	Delayed diagnosis and frequent misdiagnoses	→	Timely, accessible, and respectful diagnostic process
Information and understanding	Patients are left without a clear sense of understanding what the diagnosis means for them	→	Timely, comprehensible, comprehensive and personalized provision of information
Care and support	Patients suffer due to the symptoms of their disease (e.g. pain) Patients and their families/carers suffer the psychological consequences of the diagnosis (e.g. uncertainty, loss) Patients and their families/carers suffer the social consequences of the diagnosis (e.g. loss of income)	→	Timely, accessible, individualised and coordinated medical care and psychosocial support Locating a community of understanding and/or peers

Figure 1: Aspects of RD patients' needs as a process

Patients participating in the EMRaDi PSB emphasized **awareness, knowledge and education, organization of care and care networks, availability of therapy, CoE and standards of care** as their key concerns. Based on the interviews of RD patients and relatives in the field study, we observed the following aspects and needs in the Euregio Meuse-Rhine:

Access to care[7] and care coordination:

- While obtaining the right diagnosis can be a real odyssey for some RD patient groups (several years/decades), accessing treatment- and disease-specific care after receiving a diagnosis often becomes more difficult. As Table 2 shows, a lot of RDs require a very high number of different types of health professionals to meet the specific needs given the complexity of the care (up to 25 HCPs for the Rett Syndrome). After diagnosis, installing a multidisciplinary care team to address the different medical needs often requires many HCPs. Patients and their relatives require care coordination regardless of the number of HCPs, however the more HCPs involved, the higher the risk that care coordination becomes too difficult to handle.
- Patients affected by certain RDs have a high burden of self-coordination for all the visits to medical and other care professionals (follow-up in CoE but also coordination of their day to day care at local level).
- Informal caregivers, often the relatives are crucial and have a pivotal role in not only the organization of care, but all the aspects of care and day-to-day life.

	Huntington	Duchenne	CML	PV	Rett	Silver Russell	PKU	Galactosemia
GP	X	X	X	X	X	X	X	X
Medical secretary					X			
Paediatrician		X			X	X	X	X
Acupuncturist					X			
Baby-sitter or live-in nurse		X			X			X
Dietician	X	X			X	X	X	X
Eye doctor		X			X	X		X
Nurse	X	X		X	X		X	X
Occupational therapist	X	X	X		X			
Orthotist		X			X			
Osteopath					X			
Paediatric nurse		X			X	X	X	
Physiotherapist	X	X	X		X	X		
Protestist		X			X			
Psychologist	X	X	X	X	X	X	X	X
Psychomotor therapist					X			
Speech therapist	X	X			X	X		X
Social assistant	X	X			X		X	
Cardiologist	X	X	X		X			
Endocrinologist			X			X		
Gastrointestinal paediatrician		X			X	X		
Geneticist	X	X			X	X	X	
Haematologist			X	X				
Lung specialist		X						
Metabolic specialist							X	X
Neurologist	X	X			X			
Neuro-psychiatrist	X							
Orthopaedic surgeon		X			X			
Psychiatrist	X	X			X			
Revalidation specialist	X	X			X			
Specialist (not specified)	X	X	X	X	X	X	X	X
N HCP mentioned	15	23	8	6	25	13	10	10

Table 2: Types and number of different healthcare providers consulted (before and after the diagnosis phase) for each disease by patients interviewed during the EMRaDi field study (each patient did not visit all of them)

For each of the 8 selected RDs, GPs and psychologists were mentioned by the interviewees.

Cross-border healthcare:

- Cross-border travel for healthcare is very dependent on the disease. When expertise is not available in the region, patients would benefit from cross-border care to consult the right expert for their disease. Yet, very few patients and relatives report cross-border movement to receive care or for consultations.
- Patients and relatives think that care is well organised within their country and that it is easier for them to get treatment very close to their home through their own social security system.
- There is a lack of information about rights and reimbursement for cross-border healthcare.
- When they do cross the border, it is mostly to participate in a patient association information day or weekend rather than directly related to care.
- Linguistic capabilities of both healthcare professionals and patients or relatives influence whether they access expertise across the border.

Information aspects:

- **Instances of healthcare professionals' lack of awareness for RDs** complicates and delays diagnosis as professionals often do not think of RDs, leading to difficulties in finding the right treatment, coordinating and accessing care and expertise, as well as getting the care reimbursed.
- **Instances of healthcare professionals' lack of expertise in RDs** causes patients and relatives to suffer from healthcare professionals misinforming them or making wrong decisions because of this lack of knowledge.
- **Patients' information needs are very specific to each individual.** Healthcare professionals need to be attentive to the patients' needs and wishes. Some patients like to be informed little by little, while others want to know as much as possible at once, disliking the fact that the information is fragmented and comes from different specialists and other health and social care professionals in dribs and drabs.
- **Alternative sources of information:** Patient associations are considered to be a credible source of information. But patients and relatives use the internet as a means of information for self-education, not to mention online communities, fellow patients and health insurance funds.
- **Required information:** The medical information itself is not what is most lacking for patients and relatives. Existing information on symptoms and the description of a RD is often easy to access; the main unmet information need is tailormade personal information on access and coordination of care, eligibility of reimbursement, coverage by health insurance and out-of-pocket costs.
- **Patients' understanding of the disease and self-education:** Patients educate themselves in order to coordinate their care and to be able to deal with all the administrative issues (what reimbursement they are intitled to, for example).

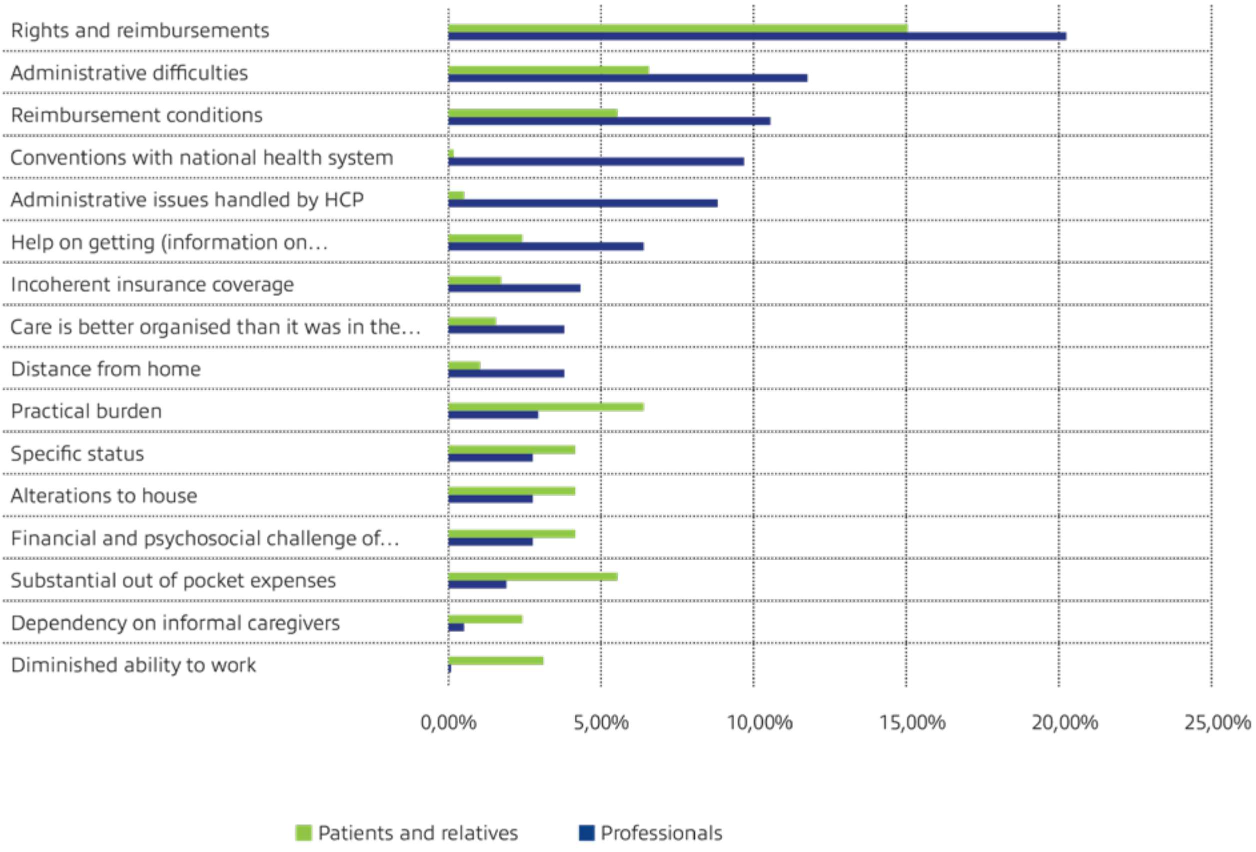
Practical, financial and legal aspects

- **Costs, reimbursement and insurances**
    - Expensive medicine: During interviews, participants explained how costly RDs are for society. Drugs – even when reimbursed to patients – represent important amounts of money for Social Security (often tens of thousands of euros, or even more, per year per patient). In some cases, getting the necessary approval for patients is very difficult and patients have to go without treatment for a while or pay the treatment themselves.
    - Reimbursement of care: In many situations, all the care is not reimbursed by the mandatory scheme/basic package and substantial out-of-pocket payments are borne by patients, especially for paramedical care and psychological follow-up. When there is a CoE for the RD with a multidisciplinary approach, these types of care can be available for patients and reimbursed. When this is not the case, it results in self-care coordination, a burden for patients and relatives and extra money needing to be spent. In case of lack of reimbursement, the result can be patients not taking up the care, which can have consequences on their future life and thus a higher final cost for society. For specific cases, additional reimbursement mechanisms exist for RD patients, but they are not always known (for example, the Special Solidarity Fund in Belgium).
    - There is no specific "RD" status that grants entitlement to benefits; RD patients rely on disability and/or chronic disease status for additional benefits.
    - Those administrative difficulties are even more difficult to cope with for patients from a lower socio-economic background.
    - The health insurance funds (to a varying degree in each of the countries) and health professionals have an important role to play to support and accompany patients for cost, reimbursement and insurance issues.

- **Work and income:** Depending on the disabilities caused by their disease, patients may be unable to work. If the disabilities are very severe, it might also prevent the parents (most often, the mother) from working or force them to reduce their working hours.
- **Housing, mobility and practical issues**
    - Procedures to get the house adapted for disabled patients is burdensome.
    - Equipment such as wheelchairs, which are necessary for patients with motricity issues, can be really expensive, and procedures that are difficult to manage for patients
    - If the disability is severe, it is a practical burden for relatives as they need to drive the patient around, find a day centre that is adapted to the patient, find the structures to get the respite they need, organise the care on a daily basis, find babysitting services or home-help service services, etc.
- **Access to education**
    - Specific special schools are sometimes recommended by HCPs.
    - Whatever the difficulties of the patients (physical and/or mental), it is really important for their parents that they not only go to a "day centre", but that they also get a proper education that is intellectually stimulating.
    - Parents of children who face mental or physical impairment because of the RD expect a holistic approach to their development which prioritizes educational challenges in order to give a positive outlook on life.

The following chart compares the answers of patients and professionals during the field study interviews in practical, financial and legal terms.

Distribution of the most coded references along the practical, financial and legal aspects during interviews, comparison between patients/relatives and professionals



Psychological aspects:

- **Psychological aspects in the pre-diagnosis period:** Before the diagnosis, most RD patients do not benefit from psychological support despite existing needs. As they can face misdiagnosis or a downplay of health complaints (by HCPs, relatives, others, or even themselves), the described feelings relating to this period are often negative, such as it being hard to handle or uncertainty.
- **Psychological aspects regarding the diagnosis experience:** At the time of diagnosis, both patients and relatives show psychological needs. Patients who do get diagnosed with a RD, often of a chronic and worsening nature, and their relatives, report negative feelings such as anger, uncertainty and sadness at that time, and highlight the pivotal role for the doctor communicating the diagnosis to guide and help them on a path to acceptance. Psychological support is often offered. The psychological needs of RD patients originate from their struggle to accept the RD and its possible daily limitations (or divergence from 'normality') and consequences on social functioning, in addition to the following: heredity, fertility, transition through life stages, outlook, and evolution of the disease.
- **The lack of awareness in society** leads to an increased psychological burden for patients and relatives. Because of the lack of awareness and recognition, they feel unsupported, isolated and misunderstood "(even more so when the disease is unknown to the public and resembles very general health complaints associated with "laziness", such as loss of activity). This lack of the population's awareness of RDs can thus hamper patients and relatives in their social and professional life.
- **The fear of judgement and stigmatisation** can incline patients and relatives to keep the diagnosis a secret in their professional life. Sometimes healthcare professionals even advise to do so.
- **Need for support**
    - In general, patients and relatives do wish to receive psychological or social support.
    - Relatives, as informal caregivers, are important day to day sources in providing psychological, emotional and social support to RD patients. Not only their care duties, but also their own psychological, emotional, professional and social needs, often cause burn-out or need for (professional) support.
- **Empowerment:** Different patients and relatives may have a different evaluation or perception of patient empowerment; while some eagerly devote themselves to self-education, others might find it a solitary or burdensome search. A lot of the patients and relatives identify role reversal and have no other choice than to see themselves as patient experts.

3.2/ Observations and challenges for patient associations

Cross-border healthcare

- Patient associations tend to have good contacts at the European and international level, but not specifically in border regions, although contacts sometimes exist in the event of the language being the same.
- They participate in initiatives such as ERNs, international congresses and forums.
- A vast online community of support exists that crosses land borders (support groups, importance of websites and Facebook groups, e.g. Facebook groups of Flemish Duchenne Mothers or North American CML women)

Information aspects

- Besides healthcare professionals, patient associations are one of the most important sources of information for patients via different means of communication - online, conferences, weekends, etc.

Access to care and care coordination

- Patient associations are highly valued by both healthcare professionals and patients in offering practical and psychosocial support. Frequently, RD experts have established collaborations with patient organisations in their field of expertise with mutual benefits for both sides.

Practical, financial and legal aspects

- Beyond providing generic information and organising activities, few patient organisations offer individual counselling for their members (such as the "Huntington Liga" and "la Ligue Huntington" in Belgium).

Psychological aspects

- Patient associations are a source of support to patients and relatives as they put families in touch with those in similar situations. The information activities allow them to learn more, while the social activities allow them to build a social life with people in the same situation.
- For some patients and relatives, direct contact with fellow patients can be difficult and confronting however, as it gives them a glimpse of their own future. Some prefer to only use online communities (or neither of these options).

3.3/ Observations and challenges for healthcare and social services providers

Access to care and care coordination

- When they discuss the RD patient pathway, HCPs attribute a lot of importance to access to care, with specific mention about collaboration, such as referrals, the task division between HCP/ hospitals, the collaboration with patient associations and the exchange of information, and new forms of care such as telemedicine. They do not talk so much about the care coordinator.
- General Practitioners (GPs) are a very important stop for patients with an unclear diagnosis and suspicion of a RD, as they are usually the first medical contact for patients suffering from undiagnosed health complaints. They have a very good overview of the overall situation (medical, history of complaints and living conditions, education, vocation etc.) and are at the start of the diagnostic process. The challenge for the GP is to recognize that a patient may have a RD and then, to know what to do.
- An important form of collaboration between HCPs is the referral. Referrals mostly occur from GP to specialist, or from specialist to another specialist. A referral can be made in order to obtain a diagnosis, help with the harmful consequences of a disease, or to access adjacent care services (development and educational services, social services, home services).

- Social workers are often in a good position to access different kinds of information, but in the referral process they are not identified unless when the HCP refers to social workers of specialised centres.

Cross-border healthcare (CBHC)

- In general, HCPs do not have enough knowledge on cross-border RD facilities or National Contact Points (NCP) for CBHC, and have often only limited knowledge of how care is organized abroad.
- HCPs find it troublesome that treatment or medicines might be available or approved in adjacent regions when this is not the case in their own country.

Information aspects

- HCPs do not always realize the vastness of information needs of patients and cannot give an all-encompassing summary of all aspects surrounding the disease (practical, psychosocial and administrative issues).
- HCPs identify the need of a fixed contact person to accompany the patient and family.
- Professionals mention that they cannot rely on what they learned during their studies because not much is taught about RDs. In order to keep their knowledge up to date, they have to train themselves on the job by attending congresses and looking to the existing literature.

Practical, legal and financial aspects

- HCPs say that there is room for improvement in relationships and dialogue with health insurance funds on reimbursement rules in general, and new therapies and medicines specifically.

Psychological aspects

- During the interviews, professionals mentioned the psychological, emotional and social functioning aspects less often than patients and relatives. Yet, as the interviews with patients and relatives demonstrate, these psychological needs are very broad.
- Psychologists were mentioned by patients and relatives affected by each of the RDs during the interviews, expressing their high psychological needs. Most of the time they intervene in the patient pathway after the diagnosis, showing the possible lack of appropriate psychological support in their quest for a diagnosis.
- In some centres of expertise, as is the case in the EMR, a psychosomatic doctor (UKA ZSEA) or psychologist (CHU Liège and MUMC) is part of the RD Centre Office or multidisciplinary team.

3.4/ Observations and challenges for Hospitals and centres of expertise

Access to care and care coordination

- Not all RDs have a dedicated CoE (see the table of expertise of the 3 University Centres in the Euregio Meuse-Rhine, in “Achievements”)

- Referrals for RDs usually occur from local and secondary hospitals to university hospitals. Some RDs (like the Haematological RD) can be followed up in local hospitals as well.
- Patients in CoE/reference (in university hospitals) tend to benefit from multidisciplinary care.
- Despite a positive system of multidisciplinary follow-up in centres of expertise, need for more staff exists in university hospitals: nurses, social assistance, paramedics (means and financing)
- Organizing multidisciplinary care and keeping track of the activities of the different healthcare professionals is a substantial burden not only for RD patients, but also their central care providers, which are often RD specialists.
- Advised annual check-ups for RD patients at university clinics depend more on the planning of patients than on the follow-up by clinics. A tracking system is most often lacking but would be necessary to ease the burden of the patient and its relatives.

Cross-border healthcare

- Local hospitals near the borders can be involved or have knowledge of expertise across the border to refer patients based on linguistic preferences. Language difference can, however, also be the reason why some local hospitals might be inclined to refer patients to university hospitals or CoE within their own healthcare system.
- Health care providers specialized within a disease group are often already in contact with equivalents across the border, with expertise exchange, referral or collaboration taking place on occasion. They also participate in ERN, international expertise exchange, and international congresses, with some of them adhering to international guidelines for a RD should they exist.
- Cooperation in patient care and research has room for improvement. As about 80% of RDs are genetically caused, human genetics is perceived as one of the leading sciences to speed up RD diagnosis.
- Concrete challenges exist daily work, when information has to be exchanged, or different experts have to come together for an interdisciplinary case conference.

Epidemiological intelligence[8]

- Epidemiological intelligence on RDs in hospitals is not very well developed yet, which represents a barrier to optimising care for these patients once they enter the hospital.
- Different RDs represent very different burdens in terms of hospital visits and hospitalization-related costs, which renders a one-size-fits-all approach to RD care less relevant. Nevertheless, the size of the burden in the EMR ranges between dozens and hundreds of patients per RD, which supports our sense of urgency in creating systematic approaches and more patient pathways for RD patients. The burden of RDs is too large for the organization of RD care to rely on motivated individual specialists.

3.5/ Observations and challenges for health insurance funds

Informational aspects

- Patients and relatives reported both positive and negative experiences in their relationship with the health insurance funds. RD patients and relatives often have to find what rights and reimbursement they are entitled to by themselves, when they could be better informed about what is available to them.

- Patients and relatives present low literacy on reimbursement issues and have a psychological burden of not finding information and organizing themselves.
- In Belgium, each health insurance fund, via their social services, offers guidance and help to patients in order to assist them with all their administrative issues. Although patients that have benefitted from such help positively evaluate it, many do not know such services exist.

Cross-border healthcare (CBHC)

- Patients and relatives do not know their CBHC reimbursement rights.
- In the event they do go cross-border, there are sometimes substantial out-of-pocket expenses, with some evaluating the procedure to do so as burdensome.
- There is information for patients on reimbursement possibilities of CHBC at the three EMRaDi University Hospitals – although this reimbursement is often a long process which needs a lot of work. More cooperation with the health insurance funds would be required.
- Patients, mainly from Flemish Belgium, visiting the centre of expertise of the hospital of Maastricht (MUMC+), hope for new solutions for their CBHC reimbursement. In the past the IZOM project facilitating the care to health specialists across the borders in all the EMR was a solution but it was stopped in June 2017. The new regulation in place, "Ostbelgien Regelung" (OBR) does not solve all cases and is only restricted to patients from the German-speaking community of Belgium and its neighbouring communities for their care in the Regio Aachen.

Access to care and coordination of care

- Healthcare providers (HCP) complain about procedures to recognize new treatment options. This results in complicated relations between health insurance funds and healthcare professionals (as expressed in the following point in more detail).

Administrative aspects and reimbursement

- Little awareness or 'literacy' of patients and relatives is observed on the subject of reimbursement, or who they could get help from in order to solves their issues.
- In most of the situations, participants express their contentment with the level of reimbursement. However, when the diseases are complex and the care needs are very broad, it is more likely that there is not enough reimbursement and substantial out of pocket expenses for patients, especially for paramedical care and the psychological follow-up. This lack of reimbursement can also result in non-take-up of the care, with greater consequences in future life and thus, a higher final cost for society. While people with no financial difficulties are usually happy about the level of support or reimbursement and financial support received, single-mum families or people with financial difficulties expressed their difficulty in covering all the costs related to the disease, not to mention their limited ability to work. Patients and relatives in general highlight administrative burdens to reimbursement.
- Healthcare professionals highlight room for improvement in relationships and dialogue with health insurance funds on reimbursement in general, and new therapies and medicines in specific.
    - HCPs complain about the administrative procedures of health insurance funds, for example the approval of reimbursement for high-cost medicine. When the approval is not granted by health insurance funds, patients often have to wait for a very long time before they can benefit from medicine which would greatly improve their situation.
    - High-cost medicine: Because they are expensive, getting medicines reimbursed is harder than it is for medicine administrated for common diseases. Professionals emphasise the difficult procedures they sometimes have to undergo. Some healthcare professionals

interviewed regret they have little discretionary power when they are aware of solutions that are not eligible for reimbursement because they are not recognised as solution to treat this disease.
- There is no specific RDs status that grants entitlement to benefits.

Paramedical care and psychological aspects

- In many situations, there is not enough reimbursement and substantial out-of-pocket expenses for patients, especially for paramedical care and the psychological follow-up.
- Belgian professionals participating in the field study request the standardisation of reimbursement of psychologic care.

3.6/ Observations from the academic research on the number of patients, the information systems and the costs of rare diseases

Based on the evaluation of the number of patients with rare diseases (WP1 report), a number of findings can be derived.

First, having a RD diagnosis gives rise to possibly tens or hundreds of hospital visits each year in the EMR. **A "one-size-fits-all" approach to optimizing RD care** is not appropriate however because RDs are so different in their needs for diagnosis, treatment and care. Ad hoc unsystematic approaches to organising patient care that rely on the administrative and managerial skills of individual medical experts are often insufficient. The overall number of RD cases would require a systematic and planned way of addressing the needs of these patient groups.

Secondly, the consulted information systems are currently not conducive to further research of health services in the area of RDs. Reasons include the lack of interoperability of hospital information systems across national borders; the lack of a clear pathway to access relevant data for the purpose of health services research; and partly the lack of adequate human resources for data holders to process requests.

Third, the evaluation report on the number of patients with RDs also provides information about the treatment costs of selected RDs in the Belgian part of the EMR, but substantial discrepancies can be observed between data based on hospital information systems and data based on insurance claims data (for different reasons such as an incorrect transfer of data between hospital and insurance information systems or a difference in diagnosis coding for clinical and reimbursement purposes).

The below table provides insight into the average annual costs of treatment for selected RDs in the Belgian part of the EMR.

Disease	Average annual cost of treatment per patient (hospital data)	University hospitals' expertise (insurance claims data)
Chronic myeloid leukaemia (CML)	12,826 EUR	22,762 EUR
Duchenne muscular dystrophy	5,111 EUR	13,795 EUR
Galactosemia type 1	/	/
Huntington disease	4,477 EUR	17,495 EUR
Phenylketonuria (PKU)	4,251 EUR	6,322 EUR
Polycythaemia vera (PV)	10,396 EUR	60,305 EUR
Rett syndrome	/	12,020 EUR
Silver-Russell syndrome	11,729 EUR	/
Note: All values are averages over time, rounded to the closest integer. Both covered and out-of-pocket expenses are included.		

Table 3. The average annual cost of treatment of selected RDs in the Belgian part of the EMR

As expected, costs from hospital data are smaller than costs from insurance claims data, as they tend to exclude the costs associated with ambulatory care.

The researchers observed that haematological diseases tend to be the most expensive, which is likely the result of the availability of specialised pharmacological treatment. According to the insurance claims data available (not reported), pharmaceuticals represent more than 50% of the healthcare-related costs of CML and PV. The same goes for PKU. Insofar as neurological and developmental disorders are concerned, the insurance claims data suggest that nursing and other costs are substantial and can exceed half of the total treatment cost.

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[7] Patient access to care includes obtaining information on treatment, support services and choices in treatment.
[8] For more information on the concept, see "Epidemiological intelligence as a model of organization in health" Rodrigues-Júnior AL., Cien Saude Colet. 2012 Mar;17(3):797-805.

4/ ACHIEVEMENTS TO IMPROVE THE PATIENTS QUALITY OF LIFE

In addition to the project activities and the 7 reports that were briefly described at the beginning of this report, below are the specific achievements for our target groups that will enable to further improve the quality of life of the persons suffering from a RD the long term. For more information on these achievements, the reader can consult the different activity reports.

Literature review

Qualitative research
-> Field study

Quantitative research
-> Data collection

7 ACTIVITY REPORTS

Legal and financial analysis with reimbursement mechanisms

Networking, training and exchange of expertise among health professionals

Organizational models for the management of the RD

Patient involvement and public awareness

4.1/ Patient involvement

In the EMRaDi project, patients and patient associations were involved in all stages of the project with the cooperation of VSOP as an EMRaDi partner, of Achse and RaDiOrg as associated partners and with the establishment of the **PSB**, which advised the project partners in several areas related to the quality and organization of care.

Patients were also involved in the **development of the Centres of Expertise (CoE).**

The field study of existing RD patient pathways in the EMR led by ANMC and Solidaris and based on 80 in-depth interviews with 104 participants **allowed mainly patients and relatives** (and professionals) **to be heard about their experiences and needs**. This study showed clear needs in terms of access and coordination of care, information, practical and financial assistance, and psychosocial support. Overall, it can be seen that RD patients and their family members are first and foremost looking for support.

4.2/ Provision of information and awareness raising

For RD patient and relatives

A specific information factsheet has been developed for RD patients and their relatives to provide them practical information and advice. This factsheet can be found in the Appendices of this Report.

For primary care and general practitioners

Due to the key role they play in the RD patient pathway, different activities were carried out for the GPs during the EMRaDi project to increase

- **their awareness of RDs, and**
- **their knowledge of the RD CoE**, and their partners in diagnosing and providing medical treatment to people living with a RD.

For this purpose, different information sessions and post-graduate education programmes were organized by the three University Hospitals of the Euregio Meuse-Rhine (UK Aachen, Maastricht UMC+ and CHU Liège) in 2019 and 2020. GPs and clinical specialists were trained on when to think about an RD. The latest know-how in RD diagnostics and (integrated) care programs were also shared. In total, more than 320 GPs and other doctors participated to the information sessions.

To raise the awareness of primary care practitioners (GPs and paediatricians) on RDs, the EMRaDi project developed a document containing a list of questions together with a specific information factsheet (attached as Appendix 4 of this Report) to help them to consider whether a patient might be affected by a RD and to provide them contact information for further support. This document and the associated factsheet are distributed to GPs in the Euregio Meuse-Rhine.

For European and national decision makers

The EMRaDi's major recommendations and the synergies to be reached between European, cross-border and local developments were presented at a meeting on 5th December 2019 at the European Parliament in Brussels, hosted by two MEPs - Pascal Arimont and Frédérique Ries. The EMRaDi Factsheet, which is the basis for our future cooperation, was presented for the first time during that event (attached as Appendix 2 of this report).

The Research departments of the three major health insurance funds in Belgium[9], have detected people with rare diseases among their members and have produced a number of profile/usage indicators with not only very detailed information on patient usage and expenses, but also on social security expenses. Besides being an important source of information for the academic research led by the University of Maastricht and the evaluation of the number of patients with rare diseases, this data is a precious tool for national social security decision makers.

For all target groups and the general public

Rare Disease Day in the EMR:

- On 28th February 2019, the EMRaDi consortium gathered more than 20 speakers and 120 participants for the first Euregio Meuse-Rhine Euroregion Rare Disease Day (EMR RDD), with the participation of patients and patients' associations. During this day, the following question was addressed: "How can we improve the healthcare and quality of life of patients affected by RDs in the Euregio Meuse-Rhine in border regions and at European level?" This overarching question was explored via three central themes - Awareness, Undiagnosed/patients without diagnosis, Cross-border healthcare & cooperation - via shared presentations. The conclusions and recommendations of the day are included in Appendix 6.
- CHU Liège organized three RD days (2018, 2019 and 2020). RD patients' associations and other associations involved in RD research, care or patient assistance were invited to share information about a specific disease. Conferences were organized for health practitioners and for the general public ("Belgian actions for rare diseases"). Professionals of the Genetic Department were present to give information about RDs, genetic diseases, diagnosis and treatment to patients and visitors. Games were organized to improve knowledge about RDs is general and to sensitize the public opinion and the professionals.
- UK Aachen also organized a RD Day in Düsseldorf together with the six other centres for RD in North Rhine-Westphalia and the North-Rhine Medical Association.

EMRaDi closing event:

Taking place on 17/02/2020, this was a participative and patient-centred day to allow all stakeholders, including patients and patient associations, to be involved and put forward proposals for future cooperation in the Euregio Meuse-Rhine (see the results in Appendix 7)

Throughout the project, dozens of meetings were held with the different stakeholders in the field of RDs to raise awareness, exchange details on the project's progress and discuss recommendations and advice for the implementation of the project and the future cooperation on RDs in the Euregio Meuse-Rhine.

4.3/ Information for patients on cross-border healthcare

VSOP developed information for patients on cross-border healthcare (CBHC), targeted at patients and which can be shared through CoE, NCP, health insurance funds, patient organisations and patient umbrella organisations.

4.4/ Cooperation between the University Hospitals and beyond

The three University Hospitals (UH) of the EMR participate in a lot of RD initiatives - they are all partners in different ERNs for example. It was obvious for the partners that being active in an Interreg Project in the EMR focussing on RD would be beneficial for the cooperation between the Hospitals and other stakeholders and finally for the patients with a RD.

Before the EMRaDi project, on a regular and one to one basis, the physicians and experts in the RDs were already in contact with their fellow experts from the other University Hospitals. But, as the efforts to set up RD structures are in their early stages, there was no exchange on a broader or structural level before the start of the project. This revealed to be a perfect starting point to increase the cooperation between the Hospitals and beyond.

Improving Knowledge in the EMR University Hospitals and Increasing Cooperation Between the EMR Partners

All three UH have lots of expertise for certain RD Groups; due to the fact that 8.000 RDs exist, no expertise centre can cover all of them. The following table gives an overview of the different foci at the three UH, and shows that not only is there overlap, but that there are also different fields.

UKA (Aachen)	MUMC+ (Maastricht)	CHU de Liège
Neurology[1]	Neurology	Neurology EUR
Hematology	Hematology	Hematology
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		
Cardiomypathies and keratinopathies (research only)	Cardiomypathies and keratinopathies	
Cancer	Cancer	Cancer
	Genodermatoses	Genodermatoses
Metabolic disease is partly covered in the other topics	Metabolic diseases, especially galactosemia	Metabolic diseases
		Endocrine diseases (adults and children)[2]

Table 4: 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).

[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.

Parallel to the project runtime, the structures of the RD Centres in all UH were strengthened, all of them have a central contact point, and have their own personnel, processes and material for patients with and without diagnosis. More info can be found at:

Aachen: www.zsea.ukaachen.de

Maastricht: www.klinischegenetica.mumc.nl

Liège: www.chuliege.be

For the project, field visits and meetings were organized between the CoEs:

- The way the management of RDs takes place in each hospital was shared. A contact point was created at CHU Liège in February 2018. It is also managed by a RD coordinator who is in contact with all other medical departments of the hospital. UKA with its Rare Disease Centre assisted CHU Liege in setting up their structure.
- CoE and patient networks organized symposia to discuss the latest development in improved diagnostics and care. Cross-border patient networks and cross-border experts translated this information to their HC pathway in their own Member State (e.g. the recently registered growth hormone therapy for Kabuki Syndrome patients was discussed in the Belgian Society for Paediatric Endocrinology and Diabetology).
- Physicians participated at RD events at the other locations more frequently than before (e.g. at the info sessions or RD Day events)

Improved information exchange:

In addition to their regular meetings, more communication and information exchange have taken place thanks to different technologies: teleconferences, email, Skype for business, etc.

Patients without diagnosis and unknown cases:

- CHU adapted the UKA procedure for managing patients without a diagnosis (with a specific questionnaire) to the Belgian situation
- Several unknown cases were shared and discussed among clinicians from the three academic hospitals e.g. in interdisciplinary case conferences. The patients visited the CoE in case extensive clinical investigation was required to give advice in follow-up steps in diagnostics

Cooperation of the human genetics and bio-informatic departments:

Several meetings organised for the project have led to a combination of unique expertise. Having access to the latest know-how in Genomics in multidisciplinary teams, the latest high-end analytical technologies, and bio-informatic analyses will lead to a faster diagnosis and accurate follow-up.

Coding of RDs:

Orphanet is a very important platform used by each hospital. Orpha codes are used to refer and designate a specific disease. In that way, CHU Liège added the Orpha codes to RD patients' medical files and created an interdisciplinary page including information on the RDs. This procedure facilitates the statistics on RDs in the hospital (by department or by physician, for example). It also enables physicians to see directly if their patients have a RD. Also, UKA plans to implement Alpha-ID and Orphanet Coding in their Hospital Information system in 2020.

Increasing awareness and improving training of GPs and resident specialist doctors

As stated before, GPs and resident specialist doctors are the first contact partners of patients with health complaints that are suspected of having a RD. Therefore, it was important to pay special attention to this point in EMRaDi.

- **Awareness** of available RD CoE in the Euregio Meuse-Rhine resulted in broad and inter-collegial knowledge exchange.
- Another action line was to reach general active resident doctors or doctors at non-university hospitals. Here, the strategy was threefold: a) RD centre webpage improvement b) Info sessions and c) Tools for daily use.
    - a) **The webpages of the University Hospitals** were improved based on the recommendations of the EMRaDi PSB in order to ease the access to information for patients, relatives, and their GPs.
    - b) Info sessions were organised in order to raise the awareness for RDs among GPs and resident specialist doctors. In January 2020, a session with about 100 participants was organised by ZSEA/EMRaDi and the North-Rhine Medical Association in Vaals, NL
    - c) A factsheet and questionnaire were developed for primary care practitioners (GPs and paediatricians).
- Furthermore, VSOP, Erfocentrum and other partners developed an eLearning platform which pays special attention to RDs. This is a brilliant example and shortly, one module will also be available in German (s. then www.emradi/eu for more info).

Translational aspects/research

An overview of clinical trials for the eight focus diseases was set up (as it is important to inform patients on this point as early as possible). VSOP created a short manual / checklist, specific for Centres of Expertise, on how to involve patients as partners in their research and research agenda.

Training for Students at the three University Hospitals

In order to increase RD awareness, medical education was considered as one of the promising topics given that all three EMRaDi University Hospitals have successful study programs in this field. During the EMRaDi project, four new RD cases were entered into the Aachen program-oriented learning (POL) field. POL is a brilliant tool to increase the problem-solving abilities and interdisciplinary thinking of medical students. CHU and MUMC+ already have a long track record in POL. Three years ago, MUMC+ introduced the concept of "the Person Behind the Patient". Students, experts from different RD expertise centres and patients design real-life cases together. In this way, they learn to look at a RD from different perspectives.

4.5/ Field study of existing RD patient pathways

The field study of existing RD patient pathways in the EMR led by ANMC and Solidaris was the first qualitative study of this nature and scale in Europe. It will serve as inspiration for more research on RD patients' needs at national and European levels.

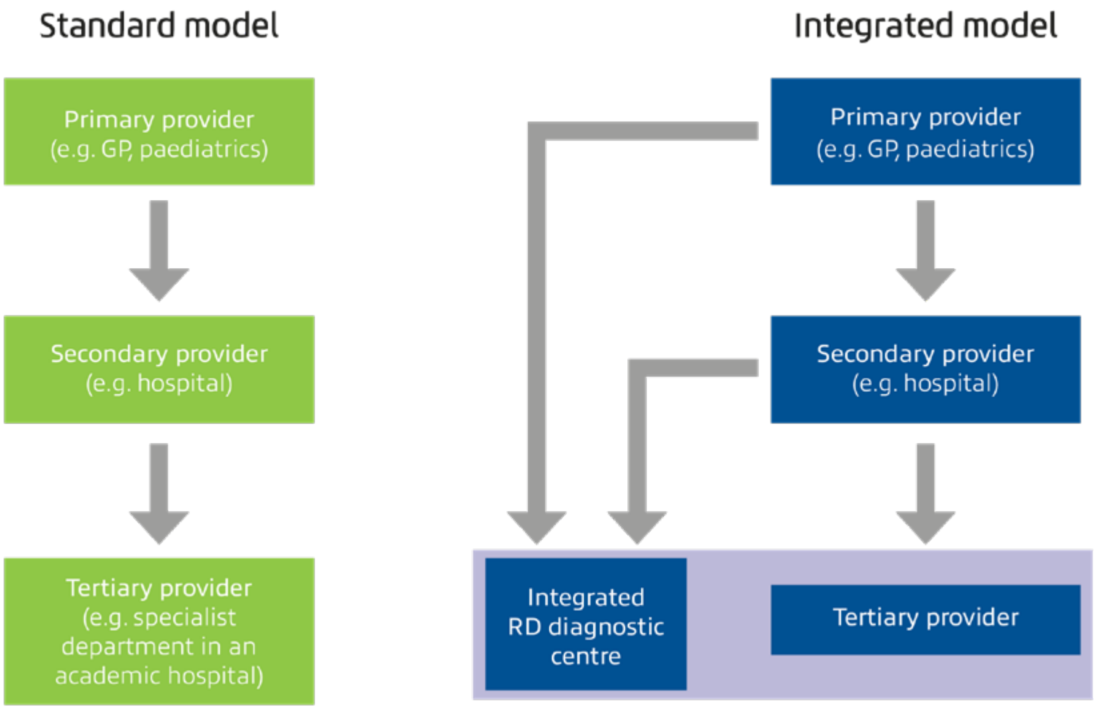
The researchers conducted 80 in-depth interviews with 104 participants, but it is important to note that many more ineligible participants submitted to participate in the study. This demonstrates the urge for patients and families to share their story and break out of isolation. **These testimonies were essential to understand the patient pathway and how to improve it.** The aim was then to generate information and seek common trends beyond individual experiences.

In connection with the Legal and Financial Analysis Report and the Patient Sounding Board recommendations, the different observations from the patients and health professionals gathered in the EMRaDi field study will enable the participating health insurance funds to improve their (social) services at a regional, national and cross-border level, and to envisage potential solutions to, and pilot projects with the different stakeholders for, the challenges RD patients and health authorities face to access care, get reimbursed, and to maintain a sound and solidary health insurance system.

4.6/ Development of better patient pathways

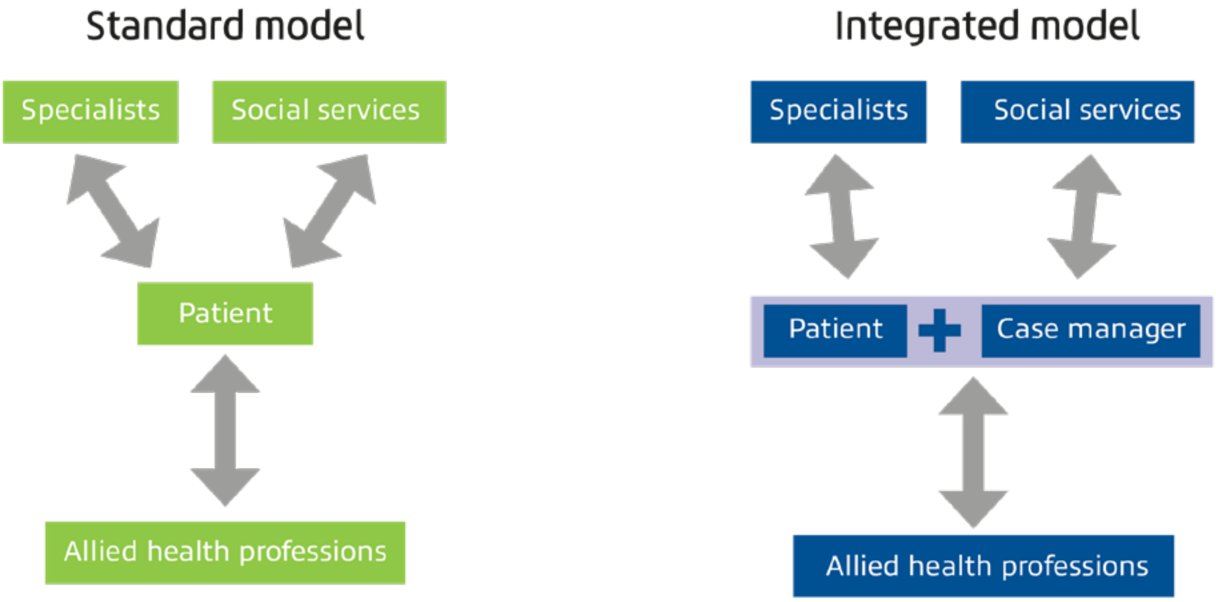
As described in the report on patient-oriented models of good practice for the management of RDs, the aim was to develop improved patient pathways for RD in EMR. As not enough data was available to use standard process-optimization techniques, a novel approach called "participatory dynamic simulation modelling" was used - it focuses on patient and provider perceptions of the situation and their preferences instead of quantitative data to build improved pathways.

Key findings to improve diagnostic service



An integrated diagnostic service significantly improves diagnosis time without raising costs.
The EMRaDi simulation showed that an increase rate of referral from primary care providers, including GPs, to a **centralised diagnostic service specialised in RDs** could substantially reduce the time to diagnosis while being cost neutral (given the model assumptions). **Testing this patient pathway innovation in a pilot study would therefore be recommended.**

Key findings for a more integrated RD case management service



Since data is scarce, we were unable to build a quantitative simulation of the case manager innovation. However, the RD patients and care providers included in the study welcomed the idea. The existing scientific evidence on other complex chronic diseases and other regions also supports the idea of case managers. Testing this patient pathway innovation in a pilot study is therefore also recommended.

The EMRaDi project recommends that these two pathway innovations are tested as pilot projects in the EMR in the context of well-designed, randomised controlled trials.

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[9] ANMC, Solidaris and MLOZ

5/ RECOMMENDATIONS AND PILOT PROJECTS

Based on the project results and the more detailed activity reports, the recommendations for the specific target groups are presented below, followed by the global recommendations and pilot projects for the future cooperation between all stakeholders with 3 main axes (Holistic approach; Telemedicine; European Solidarity), as mentioned in the EMRaDi factsheet, which is the basis of the EMRaDi declaration of intention for future cooperation in the EMR, and was signed at the EMRaDi closing event on 17/02/2020 by the partners and associated partners.

5.1/ Recommendations for patients and relatives

A specific information factsheet has been developed for RD patients and their relatives to better support them in their pathway. It provides them with practical information and advice (see Appendix 3).

5.2/ Recommendations for patient associations

As their actions and recommendations are highly valuable for not only patients but also for HCPs, health insurance funds and decision makers, **RD patient associations and their national umbrella** are encouraged to continue to advocate, through PSBs among others, at national or cross-border levels, and to raise awareness of RDs.

In the same vein, it is suggested that patient associations continue their **cooperation with all the stakeholders surrounding the patients** and to **work together on specific projects** to improve the awareness, the quality of information and the support to patients with a RD. They could jointly organize local and euregional events in relation to **International Rare Diseases Day** (28/29 February).

An increased cooperation of patient associations with health insurance funds and health professionals would help to bridge the existing gaps and to improve all the aspects of the patient pathways, at national and cross-border levels. For instance, health insurance funds (through their social services, in Belgium) can refer patients and families to national umbrella organisations or fellow patient support groups to get disease-specific social support.

The **PSB** was very important in advising the EMRaDi partners on their developments. It could be expanded to other RD patient representatives and their members should be invited to the EMR Health Forum[10], which takes place annually in the Euregio Meuse-Rhine and regroup the stakeholders of the RD patient pathway.

The **website** of the relevant patient organisations (umbrella organisations) should provide information about the CoE, with a link to Orphanet, and instructions on how people can find nationally designated CoEs for RDs. For cross-border healthcare, referral to NCPs is advised, in addition to informing the patient that contacting their health insurance fund before cross-border healthcare is preferable.

National patient umbrella organisations should require a formal **national procedure** for the assessment of candidate CoEs, including assessment from the patient's perspective, if it does not yet exist. Subsequently, patient organisations should encourage the hospitals/centres relevant for their condition to participate in this national assessment procedure[11]. In the future, such a procedure is recommended as the only way for the hospitals and CoE to be able to be part of an ERN.
If possible, European and umbrella patient associations should consider more participation to **national, cross-border and European funded projects**[12]**.**

5.3/ Recommendations for healthcare and social services providers

A holistic approach between all stakeholders is necessary: Holistic care covers the 360° spectrum of the health, social and everyday needs of people living with an RD and their families.[13] **The support and reassurance by HCPs should be considered through a more holistic understanding of the entire pathway, even when the medical pathway** of the patient (treatment, life perspective and expectancy) remain unclear (certainly for young children).

Including possibilities in (educational) development and care and efforts to support RD patients and relatives in their pursuit of as much independence and normalcy as possible would **alleviate the psychological and social burden** for them.

Proposing a psychological follow up is of high importance not only **before, during and after the diagnosis phase, but also during the life transition stages**. Due to the extensive needs of the patients and their relatives, HCPs should **offer more systematically** psychological guidance and should facilitate the process.

Case managers are needed to bridge the gap, not only between the different types of professionals involved in the pathway, but also between the different aspects of the pathway.

Awareness of, and familiarity with, "RDs" needs to be increased within the community of healthcare providers to help them to ask themselves the right questions (in terms of diagnosis and treatment) and not to overlook the **multidimensional aspects of care** (including practical, financial, psychological, and social aspects, etc.). More awareness should also improve the accompaniment of patients in terms of information **guidance through diagnostic and administrative paths:** information about CoEs in the region, cross-border cooperation, information about the importance of having the gut-feeling that the patient is an unusual case, timely diagnosis, and timely referral. A central counter for undiagnosed patients, connected to all CoEs, would be ideal. The development of a kind of (GP) standard based on symptomatology, including a decision tree that reflects the route of care for the GP that has a feeling something is wrong is recommended. To raise the awareness of primary care practitioners (GPs and paediatricians) on RDs, the EMRaDi project developed **a list of questions together with a Specific Information Factsheet** (Appendix 4) to help them consider whether a patient might be affected by a RD and to provide them with contact information to further support them.

In terms of educating professionals, medical training addressing RDs needs to change. RDs need to be addressed as a distinct subject and not woven into classes on disease domain differential diagnostics. Moreover, specific lessons should be integrated into study programs for all future physicians with general info on RDs (structures, RD centres, patient organizations, tools for help like Orphanet, etc.)" The objective is to increase the awareness of RDs rather than teaching all 8,000 of them.

Regarding **networking and exchange of expertise**, the collaboration and contact between professionals (and not only between different specialists) is crucial to improve the information exchange and connection between the network around the RD patients. When front-line professionals know which professionals they should refer their patients to, it makes the contact easier, and potentially eases the pathway for patients.

Concerning the **dissemination of the information by healthcare professionals**, there is **no right information technique that applies to all patients**; it is thus very important for healthcare professionals to be attentive to their patients' needs. **It is of highest importance to listen to the patient and not only to "provide" information.**

5.4/ Recommendations for hospitals and centres of expertise

Synergies of different expertise in the EMR: In the Euregio Meuse-Rhine, University Hospitals should continue to foster a diverse ecosystem around RDs by stimulating the education of, and exchange between, lab experts, bio-informatic experts and clinical genetic experts. When founding additional Centres of Expertise (CoE) in the EMR, the University hospitals should not focus on the same RD diseases but attract talents on not yet established expertise clusters. Together, University Hospitals can make a difference and develop solutions for RD patients in the Euregio and beyond.

Improved diagnostic services: **The EMRaDi academic research shows that** an increased rate of referral from primary care providers, including GPs, to a **centralised diagnostic service specialised in RDs** could substantially reduce diagnosis time while being cost neutral (given the model assumptions). **Testing this patient pathway innovation in a pilot study would therefore be recommended.** The implementation option in the EMR would be **to build an EMR RD group-specific undiagnosed interdisciplinary service**, which could include 3-day hospitalization for diagnostics, benefitting from an EMR expert consultation for the determination of the tests and to provide results.

Organisation of care, and care networks: Providing more insight into where specialised care (CoE) in the EMR is available is recommended. This concerns the name and address of the multidisciplinary team and not only the care provider organisation as a whole. Accomplishing a national review of CoEs, like the Dutch model, including assessment by patient organisations, could be used as a starting point. CoEs should centralize care in multidisciplinary consultations at a set time and place. Given that nowadays, patients, more times than enough, have to go to different consultations at different places and at different times, centres need to find the best healthcare providers for the specific disease and then organize that care around the patient.

CoE must continue their healthcare network in the EMR and their integration in the ERNs, designed in co-creation with patients and care networkers. To consolidate these health and social care networks, professionals (clinical professionals as well as lab and bio-informatics experts) need to meet and exchange information regularly.

As a general rule the hospitals, health insurance funds and patient associations should increase their cooperation to provide more complete information to patients on all aspects of the RD patient pathway (also with regard to cross-border healthcare possibilities).

RD Case Manager: In line with the field study (WP2 Report) that advocates for a permanent contact point for the families, the EMRaDi academic research (WP3 Report) recommends piloting RD case manager roles at university hospitals. However, not enough research has been done in this area to provide any definitive recommendations as to how this role should be structured. The currently available experience would suggest that nurse practitioners (that have good insight into the needs of patients with particular RDs and could effectively liaise with different healthcare professionals) would be a good place to start in developing the role of RD case managers. It is also important to provide insight into care organisation and how this relates to guiding the patient in making choices (shared decision-making) in different phases of the care process (including psychosocial aspects).

Epidemiological intelligence: The EMRaDi academic research (WP1 reports) focused on improving epidemiological intelligence services related to RDs at the care provider level; supporting systematic data collection, storage, and analysis relevant to RD patients is necessary for a systematic accounting of the burden of individual RDs. This is also an unavoidable step in developing optimized organizational models for the provision of care for RD patients.

Cross-border healthcare (CBHC): Cooperation in patient care and research have to be improved. Medical experts need to be enabled by their hospitals to invest in cooperation, networking and policy development. Concrete challenges lay in the field of daily work, when information has to be exchanged or different experts have to come together for an interdisciplinary case conference. Physicians are encouraged to refer to CoEs in the EMR if the hospital does not have the expertise about a RD, instead of referring the patient to a CoE elsewhere in the country concerned, at a larger travel distance. The CoE should refer more systematically to the health insurance fund of their patients to coordinate together when cross-border healthcare is sought or required, prior to any CBHC. Health insurance funds and the NCPs for cross-border healthcare are the main sources to provide relevant information on this topic.

Communication and information provision on expertise and existing centres of reference/expertise in each University Hospital could be ameliorated to improve referrals to their services. The websites of the hospitals should also refer to Orphanet and the NCPs for information on RDs and cross-border healthcare. In addition, the website of the hospitals/CoE should only contain information that is specific to that particular hospital and/or the RDs involved (the exceptions to the rule). This is to prevent information from being repeated or not being up-to-date.

Provision of information and quality documents: According to the PSB, quality documents should meet the following criteria:

-validated/authorised by the relevant healthcare professional(s) and patient organisation(s);
-transparent (in interests/importance, literature and stakeholders);
-version management;
-properly encrypted to prevent “it getting a life of its own”;
-written in a "reader-friendly" language.

Should formal RD treatment guidelines for care providers be developed, the PSB recommends that a version for patients is budgeted and developed, with images and drawings.
The development of quality of care standards in collaboration with the relevant patient organisation is also recommended, in addition to making patient versions of these quality standards.

Clinical trials: When a new treatment is validated in a clinical trial, the health insurance funds and the centres of expertise should work side-by-side to evaluate the most appropriate way to disseminate this new treatment in their own Member State, taking into account the sustainability of the health insurance system.

Patient participation in medical research should start from the beginning of the process, for example when formulating the research agenda, determining the logistics, design, outcome measures, etc. The PSB advises funders of research for RDs to make patient involvement part of their call for project proposal and make this as specific as possible: How and when is the patient involved, and how is this participation funded? It also advises that ERNs (European Reference Networks) should take care of this.

5.5/ Recommendations for health insurance funds

Information aspects and proactivity: Health insurance funds should lead more proactive actions and provide clearer information on the rights and reimbursement that the people living with a RD are entitled to (at national and cross-border levels). Patients and relatives may present low literacy on reimbursement issues and have a psychological burden of not finding information and organizing themselves.

It is recommended that health insurance funds offer information in multiple languages. In the case of Belgian health insurance fund, the advice would be to provide the information in French, Dutch, German and English. In the case of Dutch and German health insurance funds, the advice would be to provide information in English in addition to Dutch and/or German.

On their websites, the health insurance funds should provide information on RDs with references to the CoE, the patient associations and Orphanet as a source of information for treatment and care. Regarding cross-border healthcare, more information should be provided by the health insurance funds in coordination with the NCPs, as mentioned in the below paragraph on CBHC. As a general rule the hospitals, health insurance funds and patient associations should increase their cooperation to provide more complete information to patients on all aspects of the RD patient pathway (also with regard to cross-border healthcare possibilities).

Administrative aspects and reimbursement: towards a more holistic and coordinated approach
In the event of a RD, **a fixed service or contact person** at the health insurance fund could help the **patients and relatives, as well as the HCP**, to be better informed and supported on the administrative aspects and reimbursement issues. This person could act as a **"case manager"** to provide a **holistic approach** and a 360° vision around the patient and their health, social and everyday needs. When available by the health insurance funds, **social services** should be promoted more (inside and outside the organization) to support persons with a RD and their relatives. Health insurance funds should direct the patients and relatives to their social service directly, which can offer them guidance and assist them in all their administrative issues. Exploration of rights is an important task for social workers/case managers to make sure that RD patients and their relatives access the care they are entitled to (including psychological support). Health insurance funds can also refer to specific patient associations for more psycho-social support tailored to their disease.

Health insurance funds also have **room for improvement in their relationships with the HCP** and CoE on reimbursement and administrative procedures in general, access to new therapies and medicines in particular. Health insurance funds and HCPs should liaise to review the administrative procedures together and make them easier where needed and possible.

As mentioned in the previous section, when a new treatment is validated in a clinical trial, the health insurance funds and the centres of expertise should work side-by-side to evaluate the most appropriate way to disseminate this new treatment in their own Member State, taking into account the sustainability of the health insurance system.

Status of RD: Health insurance funds should advocate at national and European levels for a specific RD status that enable them to more easily identify persons suffering from a RD. This status should be granted automatically and for an unlimited period of time based on clear and pre-defined criteria, and it should give rights to specific advantages such as improved access and reimbursement of medical, psychological, paramedical and social care (at a national level and, when necessary, at a cross-border level), a clearer identification of their specific needs, a holistic approach, and care coordination from the different stakeholders involved in the pathway.

Paramedical care and psychological aspects: For patients with RDs and their relatives, more reimbursement for paramedical care and psychological follow-up should be granted by health insurance funds. In many situations, there is not enough reimbursement and substantial out-of-pocket expenses for patients. Belgian professionals participating in the field study request a harmonization of reimbursement of psychologic care between the health insurance funds, to avoid people of the non-take-up of their rights (which means that people are entitled to, but do not avail of their rights to reimbursement or do not access care).

Cross-border healthcare (CBHC): National Healthcare authorities and health insurance funds in cooperation with the NCPs could **improve their provision of information for cross-border healthcare** to the patients and to the healthcare providers. A toolbox for the NCPs and the health insurance funds has been issued by the European Commission in December 2019. Given the higher need for cross-border mobility due to unavailable services or expertise, the National Contact Point's website should develop contents specifically dedicated to RDs. Moreover, they should also make sure that the information on CBHC is targeted towards different patient groups, and that it is accessible to the average reader. Despite an established legal framework for cross-border care in Europe, problems occur at the practical and administrative level in how certain procedures are handled and provisions are interpreted. When expertise is not available in the home country, health insurance funds are encouraged to seek all the possible ways within the existing legal framework to ensure patients can access medical expertise across the border. In their declaration of intention, which is dealt with more in the following pages, the EMRaDi partners advocate for the establishment of clear rules for reimbursing telemedicine for RD in ERNs and in a cross-border context, and an easy access to the best available expertise for RD patients with priority rule and price-safety, in coordination with the health insurance funds. The holistic approach and case manager mentioned above can also facilitate cross-border care.

At a European level, the EMRaDi partners advocate for **more solidarity with the creation of an agency and a fund for RDs** in order to move forward together for more accessibility to care and medicines, more awareness of RDs and more medical and research advances.

Health insurance funds of the EMR could further support this recommendation by coordinating their action with AIM, the International association of Mutual Benefit Societies and the Rare2030 project led by EURORDIS (the European Rare Disease Patient Organisation).

5.6/ Recommendations for decision makers (at regional, national, Euregional and European levels)

To support the implementation of EMRaDi's recommendations for the different stakeholders, decision makers at regional, national, Euregional and European levels should also take action, in cooperation with them, including with patients and their associations.

The EMRaDi partners firstly recommend to pay constant attention and to allocate resources to the implementation of **national RD action plans**. Much of what has been started politically in the last years still needs to prove its relevance in practice as implementation is unfinished but continuing. It requires implementation to be monitored and completed work areas to be evaluated with the involvement of patients or their representatives. For the EMRaDi PSB, particular topics should be given priority including awareness raising, knowledge dissemination and education, organisation of care and care networks, investments for new therapies (including diagnosed and undiagnosed patients), accreditation policies for CoEs and the development of quality standards.

As a general rule, **patient participation** should be part of a funding policy and funding budget so that patient associations are able to invest in cooperation, networking and policy development.

Designation of national CoEs needs to be continued and its process consolidated. If it does not exist in the country, a formal national procedure[14] for the assessment of the candidate CoE, including assessment from the patient's perspective, is recommended. In the future, such a procedure is recommended as the only way for the hospitals and CoE to be able to be part of an ERN.

Border Regions like the EMR need special attention within the ERN.

Status of RD: A real RD status is recommended for RD patients in all countries and at a European level. This status should be granted automatically and for an unlimited period of time based on clear and pre-defined criteria and it should give rights to specific advantages such as an improved access and reimbursement of medical, psychological, paramedical and social care (at a national level and when necessary, at a cross-border level). Providing better reimbursement for RD patients can prevent them from the non-take-up of the care, which can have big consequences on future life and thus a higher final cost for society. This kind of RD status should also enable the stakeholders involved in the patient pathway to more easily identify the RD patients and their relatives, and provide them with a more holistic approach and improved care coordination.

A key concern for rare disease patients identified in the literature review as well as during the field study was to enjoy **timely access to RD experts and services and to avoid unnecessary administrative hurdles.** An overarching recommendation is therefore to **improve awareness** of the challenges RD patients face and the potential of cross-border mobility and collaboration to address those challenges among healthcare providers and wider stakeholders in health systems in the EMR and at a European level.

The EMRaDi partners recommend **designing RD patient care** pathways that incorporate expert advice regardless of the country of the expert, while ensuring that as much of the resource-intense care takes place in the country of residence of the patient.

To design improved patient care pathways, more information on how RD patients interact with the healthcare system is needed. Therefore, **efforts to harmonize data collection methods and to design paths to access health data for health services research in the field of RDs is recommended.**

- It is recommended that efforts be made by the European Union, national and regional governments, and individual healthcare providers, to **strengthen the capacity for access to RD patient data for research purposes** – particularly research into provision of health services for RD. This will require the development of common (and cross-border) guidelines on collecting, storing, and accessing the relevant medical information, and supporting the implementation of these processes with adequate human and other resources. The EMR could play a leading role in this process as an example EU region, particularly given that EMRaDi already brought relevant stakeholders into close and long-lasting contact.
- In light of the heterogeneity in disease burdens that RDs present, as confirmed by the field study, the EMRaDi partners would also recommend the study of burdens presented by not just the 8 RDs selected, but rather all RDs **through large-scale surveys**. A clustering exercise on healthcare utilisation data could highlight valuable similarities between biologically different diseases in terms of patient needs and provider burdens. Such insights could reduce the number of distinct RD patient pathways that need to be designed, piloted, and implemented.
- In line with the previous two areas of recommendations, the funders are asked to consider a **more permanent regional (EMR) intelligence effort** dedicated to systematic and **robust cross-border RD data collection and analysis** that would focus on information related to the provision of health services. Such an effort would effectively complement the existing registries focused on genetic, biomedical, and clinical information and could improve the quality of life of RD patients by supporting more efficient access to therapies that already exist (even if they are not curative).

Despite an established legal framework for **cross-border care in Europe**, problems occur at the practical and administrative level in how certain procedures are handled and provisions are interpreted. **The National Contact Points** should invest in providing more information on CBHC that specifically addresses RD and align this information with each other, in coordination with the national healthcare authorities and the health insurance funds.

To support improved patient care pathways and ensure access to medical expertise across borders when expertise is not available in the home country, decision makers should consider establishing **a preferential procedure for granting patients with special needs, such as RD patients with access to cross-border care and to reimbursement facilities**. The procedure would ideally be based on the administrative S2 form for RD patients with no advance payment from their side – Regulation 883/2004 conditions[15].

Large-scale deployment of interoperable telemedicine solutions has not been achieved in Belgium, the Netherlands or Germany so far, even if progresses at different paces are observed in the 3 countries. However, at the time of finalizing this report, in March 2020, the COVID-19 crisis has triggered some new ad-hoc initiatives or propelled the pace for using telemedicine application in all of the 3 countries at national level. A patchy EU legal framework and questions on financing and reimbursement of telemedicine services seem to be considerable barriers. Finding solutions to barriers at the appropriate policy level is recommended because they are key for RD patients to access specialized treatment more easily and more widely throughout the EU, irrespective of borders. The example of the Netherlands demonstrates that reimbursement schemes can be adapted in order to accommodate wider use of service delivery via telemedicine where appropriate.

Therefore, it is also asked that decision makers and health insurance funds consider establishing **clear rules for reimbursing telemedicine** as a key form of mobility of expertise and explore the possibilities for telemedicine created by Directive 2011/24/EU. Consultations by virtual advisory panels have now also become a key component of the work of ERNs, so the EMRaDi partners believe the time is right to appropriately regulate this form of medical service provision.

Awareness of and familiarity with "rare diseases" needs to be increased within the community of healthcare providers to help them ask themselves the right questions (in terms of diagnosis and treatment) at the right time and not to overlook the multidimensional aspects of care (including practical, financial, psychological, social aspects, etc.). More awareness should also improve the accompaniment of patients in terms of information guidance through diagnostic and administrative paths.

In terms educating professionals, medical training addressing RDs needs to change. RDs needs to be addressed more specifically in medical study programs and better woven into classes on diseases in general. Moreover, specific lessons should be integrated into the study programs for all future physicians with general info on RDs (structures, RD centres, patient organizations, tools for help like Orphanet, etc.)" The objective is to increase the RDs awareness rather than teaching all 8,000 of them.

Since the EMRaDi results show that interpersonal relationships between RD experts are a key facilitator of mobility, the support of networking between RD physicians, as well as other RD experts (psychological support, social services, etc.) in the EMR and beyond is recommended.

The EMRaDi partners compiled the main abovementioned elements and presented them into three recommendations which constitute a roadmap for their future cooperation with all the stakeholders. They are based on all the EMRaDi results and are further detailed in the next section:

1) **Holistic care:** Organizing the care to take into account all the needs of people living with a rare disease
2) **Telemedicine:** Developing reimbursement of telemedicine for RD patients in ERNs and in a cross-border context
3) **EU solidarity:** Structuring the coordination and solidarity for RDs at EU level

5.7/ Global Recommendations for future cooperation and pilot projects to develop with all the stakeholders

On 17/02/2020, during the closing event, the EMRaDi partners and associated partners signed a declaration of intent for the future cooperation and pilot projects to develop together.

The EMRaDi partners:

The associated partners:

Partners and associated partners want to pursue the development of follow-up joint actions, policies and projects at local, regional, national, cross-border and European levels to improve the quality of life of people living with a rare disease and their families in the Euregio Meuse-Rhine and beyond. This will be in the scope of three main recommendations: (1) holistic care, (2) telemedicine and (3) European Solidarity, in line with the position presented on 5/12/2019 in Brussels to European representatives (see Factsheet EMRaDi in appendix).

Partners and associated partners undertake to meet, at least once a year, at the **EMR Health Forum**[15] to continue their cooperation on the topic of RDs, expand it to other relevant stakeholders and plan joint actions, policies and projects.

These three recommendations were discussed between some key stakeholders during the closing event. A summary of the discussion is available in Appendix 7.

EURORDIS, the European Rare Disease Patient Organisation, will also be invited to those yearly meetings to continue the cooperation that has developed during the EMRaDi project.

Recommendation 1: Holistic Approach

Holistic care implies organizing the care to take into account all the needs of people living with a RD. It covers the 360° spectrum of the health, social and everyday needs of people living with an RD and their families. The holistic approach empowers and supports people with an RD and their carers to live their lives in the most fulfilling and independent manner possible, and to fully enjoy their fundamental human rights.[16]

The decision makers should support health care providers and health insurance funds to develop holistic care and care coordination services for RD patients. These stakeholders have to be recognized as services of general interest at the EU level and the decision makers should create a supportive legislative frame for their actions.

At cross-border and local levels: With the support of the European regional development fund, the EMRaDi partners plan to continue pilot projects that will:

- improve diagnostic services with a pilot study of integrated RD diagnostic service in the EMR to collect information on process (time, experience) and outcomes (financial, health impacts)
- lead to more integrated RD case management service with the implementation of new "RD Case Managers" at one academic hospital, measuring process and outcome indicators to provide more support to RD patients and improve care coordination.

  RD case managers provide patients with:
    - information on best care practices and/or guidelines
    - contact information for care providers with relevant experience, and
    - assistance with safekeeping, integrating and sharing relevant medical information with the care team
- provide additional **training (including eLearning instruments) and tools for GPs and healthcare providers** to raise awareness and reduce diagnosis time, including the possibility to implement decision-support tools to flag up potential RD patients;
- increase **data-sharing** between interested centres in the Euregio Meuse-Rhine (EMR), including projects to:
    - harmonize the coding of RDs
    - strengthen the capacity for access to RD patient data for research purposes, and
    - create a more permanent regional (EMR) intelligence effort dedicated to systematic and robust cross-border RD data collection and analysis.

Recommendation 2: Telemedicine

Developing reimbursement of telemedicine for RD patients in ERNs and in a cross-border context is important to support easy access to the best available expertise for RD patients.

ERN and cross-border telemedicine are complementary solutions to support the RD patients and the experts. Complex patient cases benefit from multidisciplinary teams/a panel of experts through the ERNs, while other patient cases may require the consultation of a single expert via telemedicine across the border for advice on a specific course of treatment or comorbidity.

Decision makers should support:

- the establishment of **clear rules for reimbursing telemedicine services** for RDs in ERNs and in a cross-border context, and an **easy access to the best available expertise** for RD patients with priority rule and price-safety, **in coordination with health insurance funds**.

  This would support the incorporation of the ERN's work into national healthcare systems. In order to do so, it is first important to:
    - close the gaps in the legislative framework for telemedicine
    - continue standardization of telemedicine hardware and software, aiming at higher levels of interoperability of different systems
    - wider integrate telemedicine services in national reimbursement schemes
    - invest in telemedicine infrastructure
- **the training of National Contact Points (NCPs)** and health insurance funds to better inform the RD patients and to facilitate their access to cross-border healthcare (CBHC) possibilities.

At cross-border and local levels: the EMRaDi partners recommend

- the development of further pilot projects
    - that will provide (cross-border) telemedicine for RDs (tele-expertise and tele-consultation) to prove (cost-)effectiveness and suggest potential reimbursement schemes
    - that explore how to integrate better existing, highly-specialized RD care in national health systems within and beyond the context of ERNs. Models of cooperation and of reimbursement respectively need to be explored and must take into account several possible situations: how can patients benefit from expertise abroad 1) if they are treated at centres not part of a ERNs network, (2) if they are based in a country not part of a dedicated ERN or (3) if they do not require the involvement of a ERNs as such (consultation of single expert abroad) ?

- the design of patient care pathways that incorporate expert advice regardless of the country of the expert, while ensuring that as much of the resource-intense care takes place in the country of residence of the patient and that the individual circumstances of patients are taken into account;
- the training of National Contact Points (NCPs), national health authorities and health insurance funds to better inform RD patients and to facilitate their access to cross-border healthcare (CBHC) possibilities and reimbursement issues

Recommendation 3: European Solidarity

In parallel to the local and cross-border actions, **structuring the coordination and solidarity for RD at the EU level** is essential in order to guarantee the sustainability of existing policies and initiatives on RDs and to ensure the implementation of the European Pillar of Social Rights (EPSR) principles and social protection for people living with a RD.

The EU and the decision makers should support the creation of a **European agency for RDs** that would be mandated to create sustainable ecosystems around RD with the core value of European solidarity in a Triple A approach:

- **Raising the Awareness** of the public at large and dialogue within the RD community
- **Increasing Accessibility** to health and social services, affordability/sustainability of the health system throughout EU, in order to
    - create and coordinate an EU solidarity fund
    - coordinate EU-wide negotiation of reasonable Orphan Medicinal Product (OMP) prices with the pharmaceutical industry
    - enable the (equal) reimbursement of OMPs
    - establish a "joint basket of care for RD" in the mandatory scheme/basic package in all MS
    - support the creation of a European RD status
    - simplify cross-border healthcare reimbursement
- Advancing academic and medical research and cooperation in RD to
    - facilitate better referrals (cross-border or not) in connection with ERNs, National Contact Points and Orphanet
    - structure the collection of EU data on RD and their interoperability
    - coordinate patient registers as a basis for research

The EMRaDi partners advocate for:

- more and continued dialogue with citizens, Member States and stakeholders
- an integration of their recommendations in the Rare 2030 foresight study (which includes an EU public hearing on RDs in the European Parliament by the end of 2020)

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[10] The EMR Health Forum has existed since 2013 and was created on the basis of previous INTERREG cooperation: it gathers, on a regular basis, hospitals, health insurance providers, universities, patient associations and other healthcare providers. The "EMR Health Forum" works thanks to the partners' own funds. Durability is ensured by the will of its members to continue their historical cooperation in the EMR and to continuously improve their cross-border actions.

[11] Belgium currently does not have a national assessment procedure for the designation of CoE. In Belgium, responsibilities in terms of health policy are shared between the federal and regional levels.

[12] https://innovcare.eu/social-services/rareresourcenet/

[13] Belgium currently does not have a national assessment procedure for the designation of CoE. It has national and regional policy and the regional government is also referred to as federalized entities.

[14] Based on Regulation 883/2004 conditions, no advance payment is needed by the patient and he/she is considered as a patient of the Member State of treatment; the patient needs prior authorization from his/her health insurance funds. Under Directive 2011/24/EU conditions, the patient covers the payment and is reimbursed in his/herMember State of affiliation according to its rate and if the obligatory insurance covers the same care; no prior authorization is needed.

[15] The EMR Health Forum has existed since 2013 and was created based of previous INTERREG cooperation. It gathers hospitals, health insurance providers, universities, patient associations and other healthcare providers on a regular basis. The "EMR Health Forum" works thanks to the partners' own funds. Longevity is ensured by the will of its members to continue their historical cooperation in the EMR and to continuously improve their cross-border actions.

[16] https://innovcare.eu/social-services/rareresourcenet/

CONCLUSIONS

Patients, relatives and patient associations want and deserve more concrete improvement in their daily life, and in the management of their RD, to better enjoy their fundamental human rights.

As RDs do not stop at borders, European cooperation and coordination on RDs is essential to allow RD patients access to the best expertise available irrespective of the Member States. However, developing local and cross-border solutions for persons living with a RD and their relatives is also of the utmost importance to improve their quality of life close to their home. Both approaches are complementary.

The EMRaDi project was the first (and probably not last) cross-border project on RDs in the Euregio Meuse-Rhine, with a multi-sectoral and patient-centred approach.

Cross-border cooperation on RDs and projects such as EMRaDi provides the opportunity to:

- bring together all of the important actors in the field of RDs, and increase expertise and cooperation in the EMR;
- analyse patient pathways and the needs of patients, relatives and professionals;
- collect data and analyse the legal and financial frameworks;
- provide better solutions to patients and their relatives in terms of quality of life through faster diagnosis and high-quality, patient-centred, integrated care in a nearby geographical area and in the patient's own language (where possible), as well as through organisational models of RD management;
- raise public awareness of RDs;
- stimulate patient participation in health research and the quality of health care and to increase patient empowerment;
- act as a catalyst for the countries involved and the development of their national plans in the field of RDs, in line with European developments;
- strengthen the implementation of Directive 2011/24/EU on the application of patients' rights in cross-border healthcare, especially on information and European reference networks; for cross-border healthcare, when possible, to favour Regulation (EC) No 883/2004 on the coordination of social security systems;
- to develop and implement patient pathways for RDs in a cross-border and European context and make recommendations in line with national and European developments.

After three and a half years of collaboration, the EMRaDi partners and all the stakeholders involved are conscious that the EMRaDi project has served as a preliminary phase to conduct more concrete improvement and pilot projects in the field of rare diseases. As stated in their Declaration of Intent, signed on 17/02/2020, they have three main recommendations for future cooperation: holistic approach, telemedicine and European Solidarity. The Euregio Meuse-Rhine could therefore serve as a pilot area for further European developments and projects to improve the quality of life of all the people living with a RD.

CONTACT DETAILS AND SOURCES OF INFORMATION

Contact at the EMRaDi partners

Mutualité chrétienne Verviers-Eupen (Lead Partner)
Department of Cross-border and European Projects
Caroline Glaude
(Caroline.Glaude@mc.be – 0032 87 30 51 95)
www.mc.be (FR) – www.cm.be (NL)
– www.ckk-mc.be (DE)

Alliance Nationale des Mutualités Chrétiennes
Department of Research and Development
Clara Noirhomme
(clara.noirhomme@mc.be – 0032 2 246 27 72)
www.mc.be (FR) – www.cm.be (NL)
– www.ckk-mc.be (DE)

Union Nationale des Mutualités Socialistes (Solidaris)
Department of International and European Affairs
Joyce Loridan (Joyce.loridan@socmut.be)
www.solidaris.be (FR) www.devoorzorg.be/ (NL)

VSOP - voor zeldzame en genetische aandoeningen
vsop@vsop.nl (+31 35 603 40 40)
www.vsop.nl
www.zeldzameaandoening.nl
www.zichtopzeldzaam.nl
www.kwaliteitvoorzeldzaam.nl
www.ziekteonbekend.nl

RWTH Uniklinik Aachen
Centre for Rare Diseases
Prof. Jörg B. Schulz (Speaker, jschulz@ukaachen.de)
Dr. Christopher Schippers
(Coordinator, cschippers@ukaachen.de)
www.ukaachen.de
www.zsea.ukaachen.de

CHU of Liège
Department of Human Genetics
Sylvie Taziaux
(Sylvie.taziaux@chuliege.be – 0032 4 366 79 99)
www.chuliege.be

Maastricht UMC+/AzM
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CoE for Rare Disorders MUMC+
c.stumpel@mumc.nl
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Department of International Health
Timo Clemens
(Timo.clemens@maastrichtuniversity.nl) -
Rok Hrzic
(r.hrzic@maastrichtuniversity.nl)
www.maastrichtuniversity.nl
www.inthealth.eu

Sources of information

University Hospitals and Centres of Expertise for RDs

Each country has set up Centres of Expertise and of References for RDs.

Only the 3 University hospitals that have participated in the EMRaDi project are mentioned here.

More information on the centres in your region can be found via your health professionals, the European database www.orpha.net and through the patient associations.

In addition, for Germany, the se-atlas - the care atlas for people with RDs - can be consulted (www.se-atlas.de). In the Netherlands, an overview of the existing Centres of Expertise for RDs can be found at www.nfu.nl (click on ‘Patiëntenzorg’ and then on ‘350’ in the section ‘Erkende expertisecentra’) and for some specific diseases on www.erfelijkheid.nl.

IN THE GERMAN PART OF THE EUREGIO MEUSE-RHINE (EMR):

Uniklinik RWTH Aachen
Zentrum für Seltene Erkrankungen Aachen (ZSEA)

Pauwelsstr. 30,
52074 Aachen

Tel.: 0049 (0)241 80-38265
Fax: 0049 (0)241 80-82044

E-mail: zsea@ukaachen.de

www.ZSEA.ukaachen.de

IN THE BELGIAN PART OF THE EMR:

CHU de Liège
Centre des Maladies Rares

Avenue de l’Hôpital 1,
4000 LIEGE

Tel.: 0032 (0)4 284 36 40
Fax: 0032 (0)4 242 54 46

E-mail: maladierare@chuliege.be

www.chuliege.be
then click on > ‘Les soins aux patients’ > ‘Consultations’ > ‘Centre des maladies rares’

IN THE DUTCH PART OF THE EMR:

Maastricht UMC+
Polikliniek Klinische Genetica

Postbus 5800,
6202 AZ Maastricht

P. Debyelaan 25,
6229 HX Maastricht

Tel.: 0031 (0)43 3875855
(08.30 - 17.00 uur)

E-mail: polikliniek.klinischegenetica@mumc.nl

klinischegenetica.mumc.nl/

Umbrella patient associations for RDs (national)

IN GERMANY:

Achse

www.achse-online.de/

Tel.: +49-30-3300708-0

E-mail: info@achse-online.de

IN BELGIUM:

RaDiOrg - Rare Diseases Belgium

https://www.radiorg.be/fr/

info@radiorg.be
+32 478 727 703

National helpline via the contact form, in coordination with Orphanet Belgium.

Additional initiative for French-speaking patients: the “Rare Disease Line” from Rare Disorders Belgium (secretariat@rd-b.be) > 0800 9 2802 (free call)

IN THE NETHERLANDS:

VSOP

- www.vsop.nl/

Tel.: +31 35 6034040

E-mail: vsop@vsop.nl

- www.zichtopzeldzaam.nl
- www.zeldzameaandoening.nl

EURORDIS-Rare Diseases Europe

www.eurordis.org

European non-profit alliance of 884 RD patient organisations from 72 countries that work together to improve the lives of the 30 million people living with a RD in Europe.

RareConnect.org

The online network of RD communities that brings together thousands of patients, families and groups who might otherwise be isolated.

Health Insurance Funds

Your health insurance fund can support you in all the aspects mentioned in this factsheet. Visiting its webpages can also provide you with information on the social support available in your region.

Orphanet - The Portal for Rare Diseases and Orphan Drugs

www.orpha.net

Orphanet provides medical descriptions of rare diseases and directories of professionals and institutions, medical laboratories, centres of expertise and patient associations.

European Reference Networks (for RDs)

ec.europa.eu/health/ern_en

To influence the decision-making process, EURORDIS encourages patient organisations to participate in European Reference Networks. Patient organisations can represent their interests through European Advocacy Groups (ePAGs) in ERNs.

EMRaDi - Rare Diseases in the EUREGIO Meuse-Rhine

www.emradi.eu

(INTERREG V-A EMR project)

National contact points for cross-border healthcare

The major source of information for cross-border healthcare in cooperation with the health insurance funds are the National Contact Points

Toolbox for the national contact points and health insurance funds

Manual, Check list and toolbox for patients (edited by the European Commission)

EU website on cross-border healthcare

IN GERMANY:	IN BELGIUM:	IN THE NETHERLANDS:
www.eu-patienten.de	www.crossborderhealthcare.be	www.cbhc.nl
E-mail: info@eu-patienten.de	E-mail:information@ crossborderhealthcare.be	

List of abbreviations

ANMC / LCM	Christian Mutual Health Funds – Alliance Nationale des Mutualités chrétiennes - Landsbond der Christelijke Mutualiteiten - Landesbund der Christlichen Krankenkassen
CBHC	Cross-Border Health Care
CHU	University Hospital Centre of Liège
CML	Chronic Myeloid Leukaemia
CoE	Centre of Expertise
EMRaDi	Euregio Meuse-Rhine Rare Diseases
EMR	Euregio Meuse-Rhine
ERN	European Reference Network European Reference Networks (ERNs) are EU-founded and EU-funded networks involving hospitals, CoE and healthcare providers across Europe. They aim to facilitate interaction in the field of diagnosis, care and research on complex or rare diseases and conditions that require highly specialized treatment, and concentrated knowledge and resources.
GP	General Practitioner
HCP	Health Care Provider
IZOM	Integratie Zorg op Maat
MCVE	Mutualité chrétienne Verviers-Eupen
MUMC+/AZM	Maastricht University Medical Center / Academisch Ziekenhuis Maastricht
NCP	National Contact Point
OBR	Ostbelgien Regelung
PSB	Patient Sounding Board
PV	Polycythaemia vera
RD	Rare Disease
RDD	Rare Disease Day
UKA	University Hospital RWTH Aachen
UH	University Hospital
UM	University Maastricht
VSOP	Patient Alliance for Rare and Genetic Diseases - Vereniging Samenwerkende Ouder- en Patiëntenorganisaties - voor zeldzame en genetische aandoeningen
WP	Work Package
ZSEA	Center for Rare Diseases at University Hospital RWTH Aachen

APPENDIX 1: DECLARATION OF INTENT ON COOPERATION ON RARE DISEASES

DECLARATION OF INTENT

ON COOPERATION ON RARE DISEASES IN THE EUREGIO MEUSE-RHINE

The Euregio Meuse-Rhine Rare Diseases (EMRaDi) project is a cross-border cooperation in the Euregio Meuse-Rhine between health insurers, hospitals, patient associations and universities (www.emradi.eu/en/). The project was funded in its initial phase from October 2016 until March 2020 as part of the European Union INTERREG V-A EMR programme.

Based on the EMRaDi project outcomes and recommendations, the project partners and associated partners recognize the importance to continue their cooperation in the field of rare diseases in the Euregio Meuse-Rhine.

They sign this Declaration of Intent to pursue the development of follow-up joint actions, policies and projects on local, regional, national, cross-border and European levels to improve the quality of life of people living with a rare disease and their families in the Euregio Meuse-Rhine and beyond. This will be in the scope of (1) holistic care, (2) telemedicine and (3) European Solidarity, in line with the position presented on 5.12.2019 in Brussels to European representatives (see factsheet in annex).

Partners and associated partners undertake to meet, at least once a year, within the **EMR Health Forum**[1] to continue their cooperation on the topic of rare diseases, expand it to other relevant stakeholders and plan joint actions, policies and projects.

Annex: Factsheet EMRaDi

The EMRaDi partners

Mutualité Chrétienne (lead partner) · Solidaris / Socialistische Mutualiteiten · VSOP · CHU de Liège · Uniklinik RWTH Aachen, Zentrum für Seltene Erkrankungen Aachen (ZSEA) · Maastricht UMC+ · Maastricht University

The associated partners

achse – Allianz Chronischer Seltener Erkrankungen · RaDiOrg · Mutualités Libres / Onafhankelijke Ziekenfondsen

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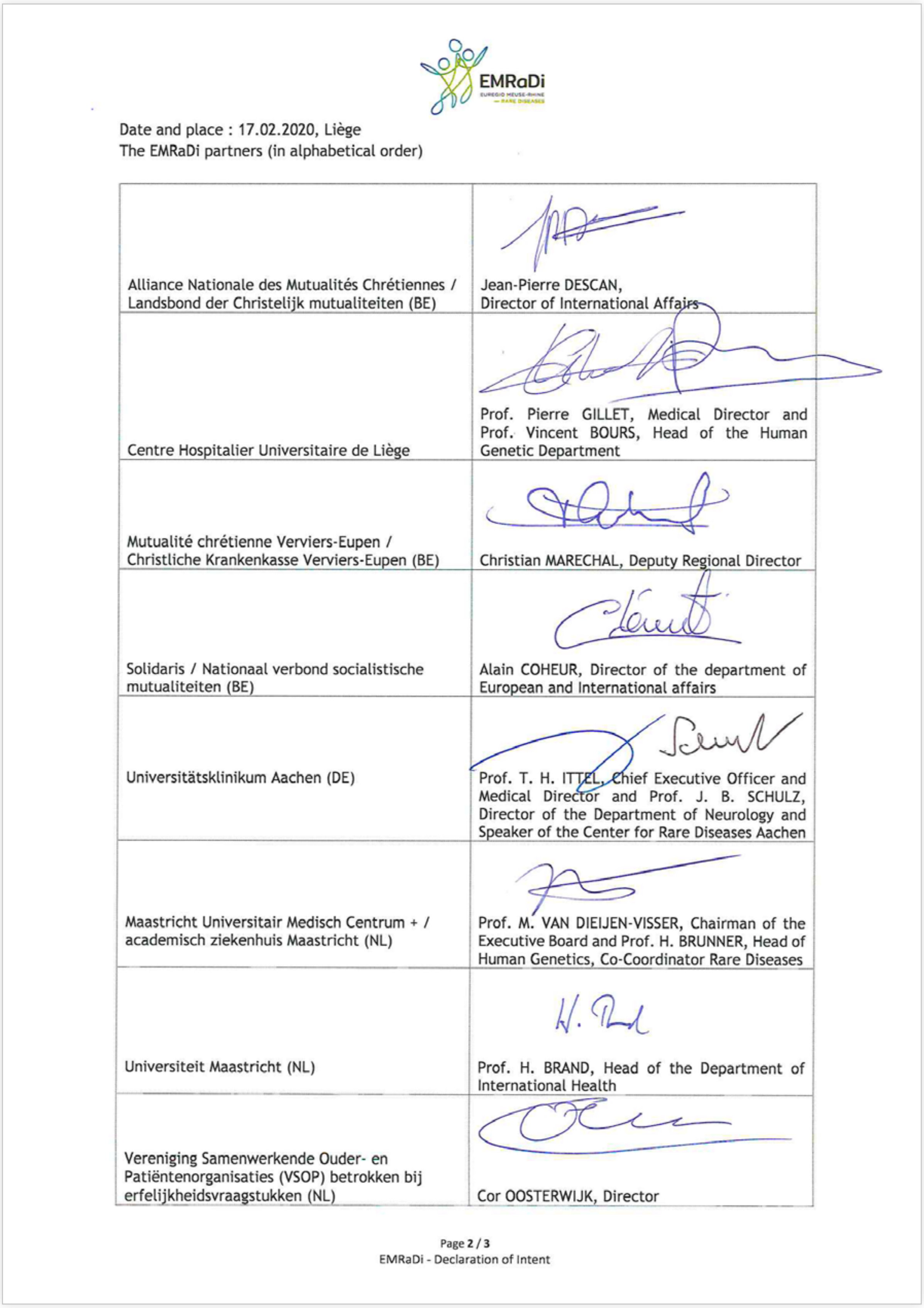
[1] The EMR Health Forum has existed since 2013 and was created on the basis of previous INTERREG cooperation: it gathers on a regular basis hospitals, health insurance providers, universities, patient associations and other healthcare providers. The "EMR Health Forum" works thanks to the partners' own funds. Durability is ensured by the will of its members to continue their historical cooperation in the EMR and to continuously improve their cross-border actions.

Page 1 / 3
EMRaDi - Declaration of Intent



Date and place : 17.02.2020, Liège
The EMRaDi partners (in alphabetical order)

Alliance Nationale des Mutualités Chrétiennes / Landsbond der Christelijk mutualiteiten (BE)	Jean-Pierre DESCAN, Director of International Affairs
Centre Hospitalier Universitaire de Liège	Prof. Pierre GILLET, Medical Director and Prof. Vincent BOURS, Head of the Human Genetic Department
Mutualité chrétienne Verviers-Eupen / Christliche Krankenkasse Verviers-Eupen (BE)	Christian MARECHAL, Deputy Regional Director
Solidaris / Nationaal verbond socialistische mutualiteiten (BE)	Alain COHEUR, Director of the department of European and International affairs
Universitätsklinikum Aachen (DE)	Prof. T. H. ITTEL, Chief Executive Officer and Medical Director and Prof. J. B. SCHULZ, Director of the Department of Neurology and Speaker of the Center for Rare Diseases Aachen
Maastricht Universitair Medisch Centrum + / academisch ziekenhuis Maastricht (NL)	Prof. M. VAN DIEIJEN-VISSER, Chairman of the Executive Board and Prof. H. BRUNNER, Head of Human Genetics, Co-Coordinator Rare Diseases
Universiteit Maastricht (NL)	Prof. H. BRAND, Head of the Department of International Health
Vereniging Samenwerkende Ouder- en Patiëntenorganisaties (VSOP) betrokken bij erfelijkheidsvraagstukken (NL)	Cor OOSTERWIJK, Director

Page **2 / 3**
EMRaDi - Declaration of Intent

The EMRaDi associated partners (in alphabetical order)

Allianz Chronischer Seltener Erkrankungen (ACHSE) (DE)	Ute PALM, Executive Board
Mutualités Libres / Onafhankelijke Ziekenfondsen (BE)	Christian HOREMANS, Expert International Affairs
Rare Diseases Organization Belgium (RaDiOrg) (BE)	Eva SCHOETERS, Coordinator

With the support of

Interreg Euregio Meuse-Rhine European Regional Development Fund

EUROPEAN UNION

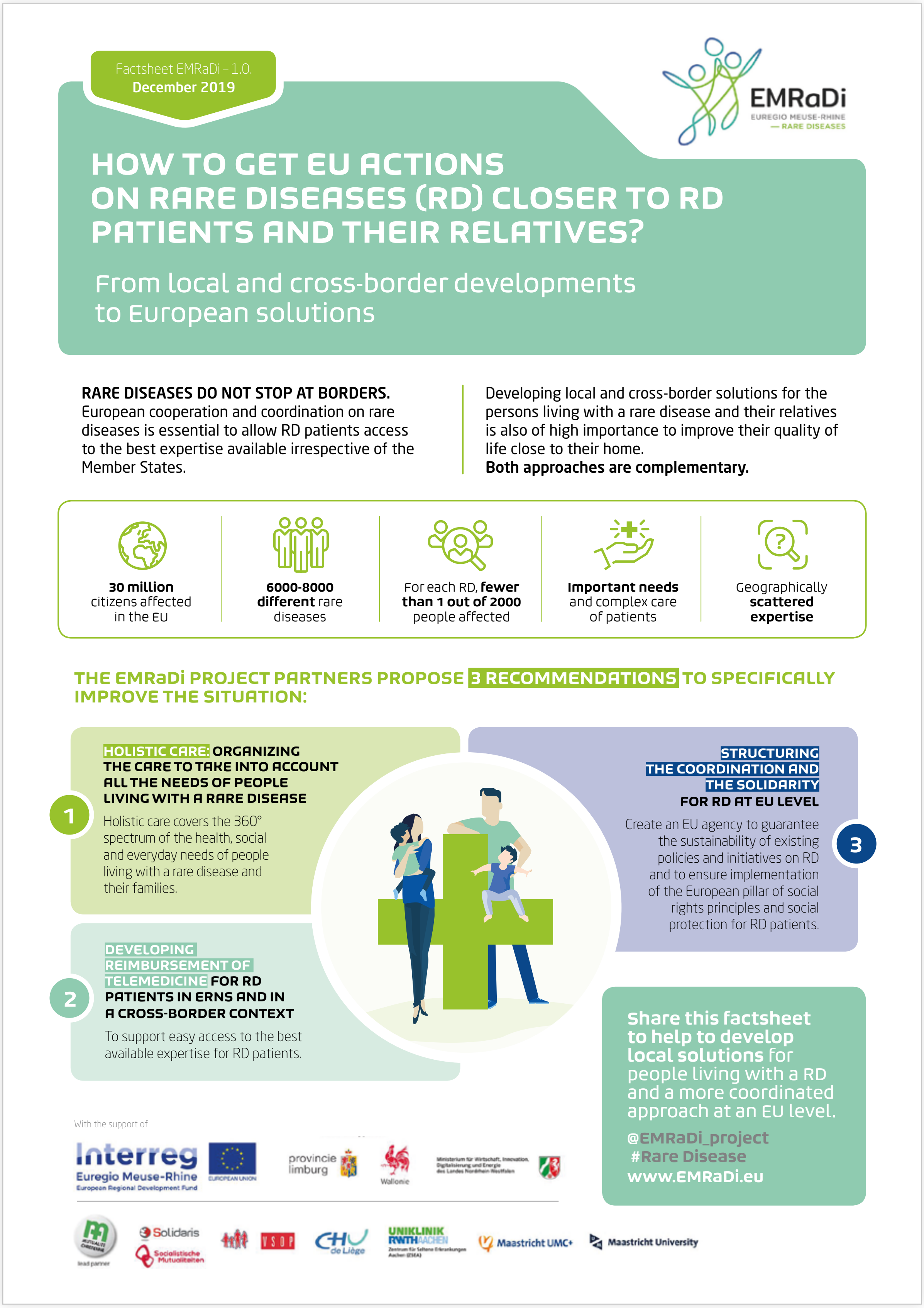
provincie limburg

Wallonie

Ministerium für Wirtschaft, Innovation, Digitalisierung und Energie des Landes Nordrhein-Westfalen

Page 3 / 3
EMRaDi - Declaration of Intent

APPENDIX 2: EMRADI FACTSHEET



Factsheet EMRaDi – 1.0.
December 2019

EMRaDi
EUREGIO MEUSE-RHINE
— RARE DISEASES

HOW TO GET EU ACTIONS ON RARE DISEASES (RD) CLOSER TO RD PATIENTS AND THEIR RELATIVES?

From local and cross-border developments to European solutions

RARE DISEASES DO NOT STOP AT BORDERS. European cooperation and coordination on rare diseases is essential to allow RD patients access to the best expertise available irrespective of the Member States.

Developing local and cross-border solutions for the persons living with a rare disease and their relatives is also of high importance to improve their quality of life close to their home.
Both approaches are complementary.

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**

THE EMRaDi PROJECT PARTNERS PROPOSE 3 RECOMMENDATIONS TO SPECIFICALLY IMPROVE THE SITUATION:

1 **HOLISTIC CARE: ORGANIZING THE CARE TO TAKE INTO ACCOUNT ALL THE NEEDS OF PEOPLE LIVING WITH A RARE DISEASE**

Holistic care covers the 360° spectrum of the health, social and everyday needs of people living with a rare disease and their families.

2 **DEVELOPING REIMBURSEMENT OF TELEMEDICINE FOR RD PATIENTS IN ERNS AND IN A CROSS-BORDER CONTEXT**

To support easy access to the best available expertise for RD patients.

3 **STRUCTURING THE COORDINATION AND THE SOLIDARITY FOR RD AT EU LEVEL**

Create an EU agency to guarantee the sustainability of existing policies and initiatives on RD and to ensure implementation of the European pillar of social rights principles and social protection for RD patients.

Share this factsheet to help to develop local solutions for people living with a RD and a more coordinated approach at an EU level.

@EMRaDi_project
#Rare Disease
www.EMRaDi.eu

With the support of

lead partner

RECOMMENDATIONS

1 HOLISTIC CARE

ORGANIZING THE CARE TO TAKE INTO ACCOUNT ALL THE NEEDS OF PEOPLE LIVING WITH A RD

> Holistic care covers the 360° spectrum of the health, social and everyday needs of people living with a rare disease and their families. The holistic approach empowers and supports people with a rare disease and their carers to live their lives in the most fulfilling and independent manner possible, and to fully enjoy their fundamental human rights.[1]

> **The EU should support** health care providers and health insurance funds to develop **holistic care and care coordination services** for RD patients. These stakeholders have to be recognized as services of general interest at the EU level and the EU should create a supportive legislative frame for their actions.

> **At cross-border and local levels:** with the support of the European regional development fund, the EMRaDi partners plan to continue pilot projects that will

- implement **new "RD Case Managers"** to provide more support to RD patients and improve care coordination
- provide additional **training (incl. eLearning instruments) and tools for general practitioners** to raise awareness and to reduce time to diagnosis
- increase **data-sharing** between interested centres in the Euregio Meuse-Rhine (EMR), including projects to harmonize the coding of RD

2 TELEMEDICINE

DEVELOPING REIMBURSEMENT OF TELEMEDICINE FOR RD

> European Reference Networks (ERNs) and cross-border telemedicine are complementary solutions to support the RD patients and the experts: complex patient cases benefit from multidisciplinary teams/ panel of experts through the ERNs, while other patient cases may require the consultation of a single expert across the border.

> **EU should support:**

- the establishment of **clear rules for reimbursing telemedicine** for RDs in ERNs and in a cross-border context, and an **easy access to the best available expertise** for RD patients with priority rule and price-safety, in coordination with the health insurance funds
- **the training of National Contact Points (NCPs)** and health insurance funds to better inform the RD patients and to facilitate their access to cross-border healthcare (CBHC) possibilities

> **At cross-border and local levels:** the EMRaDi partners recommend the development of pilot projects that will **provide reimbursed telemedicine** for RDs (tele-expertise and tele-consultation)

3 EUROPEAN SOLIDARITY

STRUCTURING THE COORDINATION AND THE SOLIDARITY FOR RD AT EU LEVEL

> **Structuring the coordination and the solidarity for RD at the EU level** is essential in order to guarantee the sustainability of existing policies and initiatives on rare diseases and to ensure the implementation of the European Pillar of Social Rights (EPSR) principles and social protection for people living with a rare disease.

> **The EU should support** the creation of **a European agency for RD** that would be mandated to create sustainable ecosystems around RD with the core value of European solidarity in a Triple A approach:

- **Raising the Awareness** of the public at large and dialogue within the RD community
- **Increasing Accessibility** to health and social services, affordability /sustainability of the health system throughout EU
- **Advancing academic and medical research** and cooperation in RD

> **Next steps:** the EMRaDi partners advocate for:

- continued dialogue between citizens, the EU, the Member States and the other stakeholders
- cooperation with the Rare 2030 project on the future policy on RD, which will culminate with a presentation to the EU Parliament at the end of 2020

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[1] https://innovcare.eu/social-services/rareresourcenet/

The EMRaDi (Euregio Meuse-Rhine Rare Diseases) project is an INTERREG-funded cross-border initiative whose goals are to increase the transparency of needs and availability of services, develop models of patient pathways, improve the network of stakeholders and draw up recommendations in order to improve the quality of life of patients with a rare disease.

APPENDIX 3: INFORMATION FACTSHEET FOR PATIENTS AND RELATIVES

Information factsheet for
Patients living with a rare disease and their relatives

EMRaDi
EUREGIO MEUSE-RHINE
— RARE DISEASES

LIVING WITH A RARE DISEASE IS NOT RARE!

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

ACCESS TO CARE

- **FOR MANY RARE DISEASES, THE BURDEN OF COORDINATING** the different aspects of the care pathway is high - either before diagnosis and looking for the right health care professional (HCP), or after the diagnosis and setting up the treatment and (para)medical support required for the rare disease (RD).
- **THE MULTIDISCIPLINARY NATURE OF RD CARE, THE NUMEROUS VISITS AND HCPs** involved as well as the challenges of the transition could be facilitated by a care coordinator. The recommended coordinator could offer centralisation and bridge all of the different needs of patients and relatives. Hospitals and centres for RDs could offer such a support in the form of a coordinating medical specialist, a social worker or liaison nurse/case manager. Other HCPs might also be consulted or involved to take on this task.

  To learn more about the expertise of different centres and their varying degrees and ways in which they aim to coordinate care for RD patients, please find the contact details of University hospitals' RD centres enclosed in this factsheet.
- **PRIMARY CARE PRACTITIONERS** - General Practitioners (GPs) and paediatricians - are also very important at all stages of your patient pathway as they have a very good overview of your overall personal and family situation.

ACCESS TO INFORMATION ON RDs

ACCESSING ACCURATE AND CORRECT INFORMATION THAT IS EASY UNDERSTANDABLE CAN BE VERY DIFFICULT.

- **HCPs** are there to inform you on every aspect of your pathway and you should never hesitate to ask them questions.
- Each country has an **umbrella patient association** for RDs, in partnership with EURORDIS, the European umbrella for RD patient organisations. This national umbrella can help you to get in touch with a specific patient association for your RD or, in case none exists, it can help you to find fellow patients/ relatives.
- As a starting point, a **list of contacts and sources** can be found at the bottom of this factsheet.
- **Online communities and social media** also exist and can be great sources of information and support. **Rareconnect.org** is an online network of RD communities in partnership with EURORDIS.

IF YOU WISH TO ACCESS INFORMATION ON THE INTERNET - whether for personal use or to pass on to others to increase their awareness - the most reliable European source of medical information on your RD is Orphanet - **The Portal for RDs and Orphan Drugs (www.orpha.net).** There, you can also find centres of expertise, patient organisations and lots of other important info. In Germany, the Netherlands and Belgium, specific information is also available - see the information sources below.

SUPPORT FOR ADMINISTRATIVE PROCEDURES AND FINANCIAL ASPECTS

IN BELGIUM, health insurance funds are a good source of information for reimbursement questions. RDs are exceptional situations for health insurance funds but they are there to offer support to patients and relatives with administrative procedures and financial aspects such as access to reimbursement, allocations, financially sustainable treatments and specific assistance for accessing the Special Solidarity Fund, which can offer, in particular cases, additional financial support to RD patients.

IN GERMANY, the situation is quite scattered and difficult to navigate. Patient associations are a good place to get help. The Centres of Expertise for RDs also offer support in these fields for their patients. Nevertheless, a lot of classic institutions are not specialised in RDs. Some of the support for administrative procedures and financial aspects will be provided by the social welfare offices of local municipalities, such as disability status or house adaptations. Other areas of support will be provided by health insurance funds, such as access to medical equipment.

IN THE NETHERLANDS, most healthcare services for RDs are reimbursed based on the national healthcare package, which applies to all citizens. For more information regarding administrative and financial procedures you can contact your health insurer and/or your hospital (finance department).

SOCIAL SERVICES (either located within the hospitals, the health insurance funds and/or the municipalities) are a good source of information and support to help you with your access to care, reimbursements and administrative procedures – disability status, adaptations to the house, medical equipment, etc.

- **Health insurance funds and social services can support and explore with you whether you benefit from all the rights you are entitled to** (specific status and access to care and reimbursement, including homecare, social and psychological services)

PSYCHOLOGICAL SUPPORT

THE PSYCHOLOGICAL ASPECTS OF RDs AND (THE BURDEN OF) CARE RESPONSIBILITIES CAN BE ADDRESSED WITH DIFFERENT FORMS OF PSYCHOLOGICAL SUPPORT.

- Different reimbursement schemes exist and could be applicable. Psychological support services are often underused because medical professionals do not offer them routinely and patients and relatives do not request them at a larger scale. This could also be explained by their limited offer and access provisions, which can be burdensome.

  Your HCP is there to support you regarding your psychological needs and check with you whether or not your health insurance covers you for reimbursement.
- Patient associations and support groups for informal caregivers in the different regions offer the possibility to exchange information and experiences.
- Raising awareness on the existence of RDs and possible psychological burden could generate more understanding and support from society at large and better social inclusion of RD families. Each year, EURORDIS, together with the national umbrellas and member organizations, organize the international campaign for the RD Day (always the last day of February).

CROSS-BORDER CARE ACCESS

DUE TO THE SPECIFICITIES OF RDs, IT MIGHT BE BENEFICIAL TO SEEK SPECIFIC EXPERTISE ABROAD.

- **Before going abroad to receive healthcare (CBHC), consulting your health insurance fund** first is highly recommended.
- **Your medical specialist** can provide you with assistance to check the different possibilities, required authorizations and reimbursement conditions.
- Further information for treatment/care and reimbursement can be requested at the **National Contact Points for CBHC**. Initial Information is provided on the NCP websites, which give a broad overview about issues relevant to cross-border care.
  More specific information tailored to your personal situation, and information on specific procedures or treatment centres can also be requested by phone or using the contact form at the National Contact Point.

CONTACT DETAILS AND SOURCES OF INFORMATION

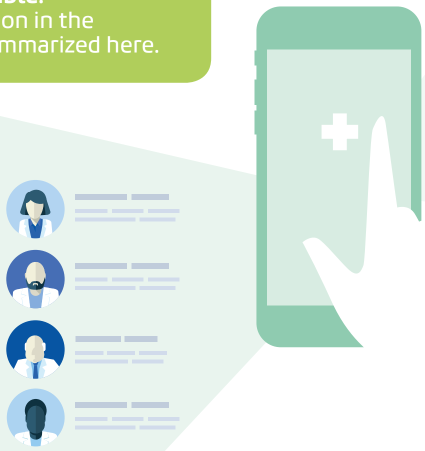
As a patient, with or without diagnosis, and as a relative or informal caregiver, it is important to have access to the best information and expertise possible.
Important bodies and sources of information in the Euregio Meuse-Rhine and in Europe are summarized here.

UNIVERSITY HOSPITALS AND CENTRES OF EXPERTISE FOR RDS

Each country has set up Centres of Expertise and of References for RDs.

Only the 3 University hospitals that have participated in the EMRaDi project are mentioned here.
More information on the centres in your region can be found via your health professionals, the European database www.orpha.net and through the patient associations.

In addition, for Germany, the se-atlas - the care atlas for people with RDs - can be consulted (www.se-atlas.de).
In the Netherlands, an overview of the existing Centres of Expertise for RDs can be found at www.nfu.nl (click on 'Patiëntenzorg' and then on '350' in the section 'Erkende expertisecentra') and for some specific diseases on www.erfelijkheid.nl.



IN THE BELGIAN PART OF THE EMR:

CHU de Liège
Centre des Maladies Rares

Avenue de l'Hôpital 1,
4000 LIEGE
Tel.: 0032 (0)4 284 36 40
Fax: 0032 (0)4 242 54 46
E-mail: maladierare@chuliege.be

▶ www.chuliege.be
then click on > 'Les soins aux patients' > 'Consultations' > 'Centre des maladies rares'

IN THE GERMAN PART OF THE EUREGIO MEUSE-RHINE (EMR):

Uniklinik RWTH Aachen
Zentrum für Seltene Erkrankungen Aachen (ZSEA)

Pauwelsstr. 30,
52074 Aachen
Tel.: 0049 (0)241 80-38265
Fax: 0049 (0)241 80-82044
E-mail: zsea@ukaachen.de

▶ www.ZSEA.ukaachen.de

IN THE DUTCH PART OF THE EMR:

Maastricht UMC+
Polikliniek Klinische Genetica

Postbus 5800,
6202 AZ Maastricht
P. Debyelaan 25,
6229 HX Maastricht
Tel.: 0031 (0)43 3875855 (08.30 - 17.00 uur)
E-mail: polikliniek.klinischegenetica@mumc.nl

▶ klinischegenetica.mumc.nl/

UMBRELLA PATIENT ASSOCIATIONS FOR RDS (NATIONAL)

IN GERMANY:

Achse

▶ www.achse-online.de/
Tel.: +49-30-3300708-0
E-mail: info@achse-online.de

IN BELGIUM:

RaDiOrg - Rare Diseases Belgium

▶ https://www.radiorg.be/fr/
info@radiorg.be - +32 478 727 703
National helpline via the contact form, in coordination with Orphanet Belgium.

Additional initiative for French-speaking patients: the "Rare Disease Line" from Rare Disorders Belgium (secretariat@rd-b.be) > 0800 9 2802 (free call)

IN THE NETHERLANDS:

VSOP

▶ www.vsop.nl/
Tel.: +31 35 6034040
E-mail: vsop@vsop.nl

▶ www.zichtopzeldzaam.nl
▶ www.zeldzameaandoening.nl

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 RDs, 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**.

EURORDIS-RARE DISEASES EUROPE

▶ www.eurordis.org

European non-profit alliance of 884 RD patient organisations from 72 countries that work together to improve the lives of the 30 million people living with a RD in Europe.

▶ RareConnect.org

The online network of RD communities that brings together thousands of patients, families and groups who might otherwise be isolated.

HEALTH INSURANCE FUNDS

Your health insurance fund can support you in all the aspects mentioned in this factsheet.
Visiting its webpages can also provide you with information on the social support available in your region.

ORPHANET - THE PORTAL FOR RARE DISEASES AND ORPHAN DRUGS

▶ www.orpha.net

Orphanet provides medical descriptions of rare diseases and directories of professionals and institutions, medical laboratories, centres of expertise and patient associations.

EUROPEAN REFERENCE NETWORKS (FOR RDs)

▶ ec.europa.eu/health/ern_en

To influence the decision-making process, EURORDIS encourages patient organisations to participate in European Reference Networks. Patient organisations can represent their interests through European Advocacy Groups (ePAGs) in ERNs.

EMRADI - RARE DISEASES IN THE EUREGIO MEUSE-RHINE

▶ www.emradi.eu

(INTERREG V-A EMR project)

CROSS-BORDER HEALTHCARE: NATIONAL CONTACT POINTS

National Contact Points are a source of information for cross-border healthcare (CBHC) in cooperation with health insurance funds.

Manual, **Checklist** and **toolbox** for patients (edited by the European Commission).

EU website on cross-border healthcare.

IN GERMANY:

▶ www.eu-patienten.de

E-mail: info@eu-patienten.de

IN BELGIUM:

▶ www.crossborderhealthcare.be

E-mail: information@crossborderhealthcare.be

IN THE NETHERLANDS:

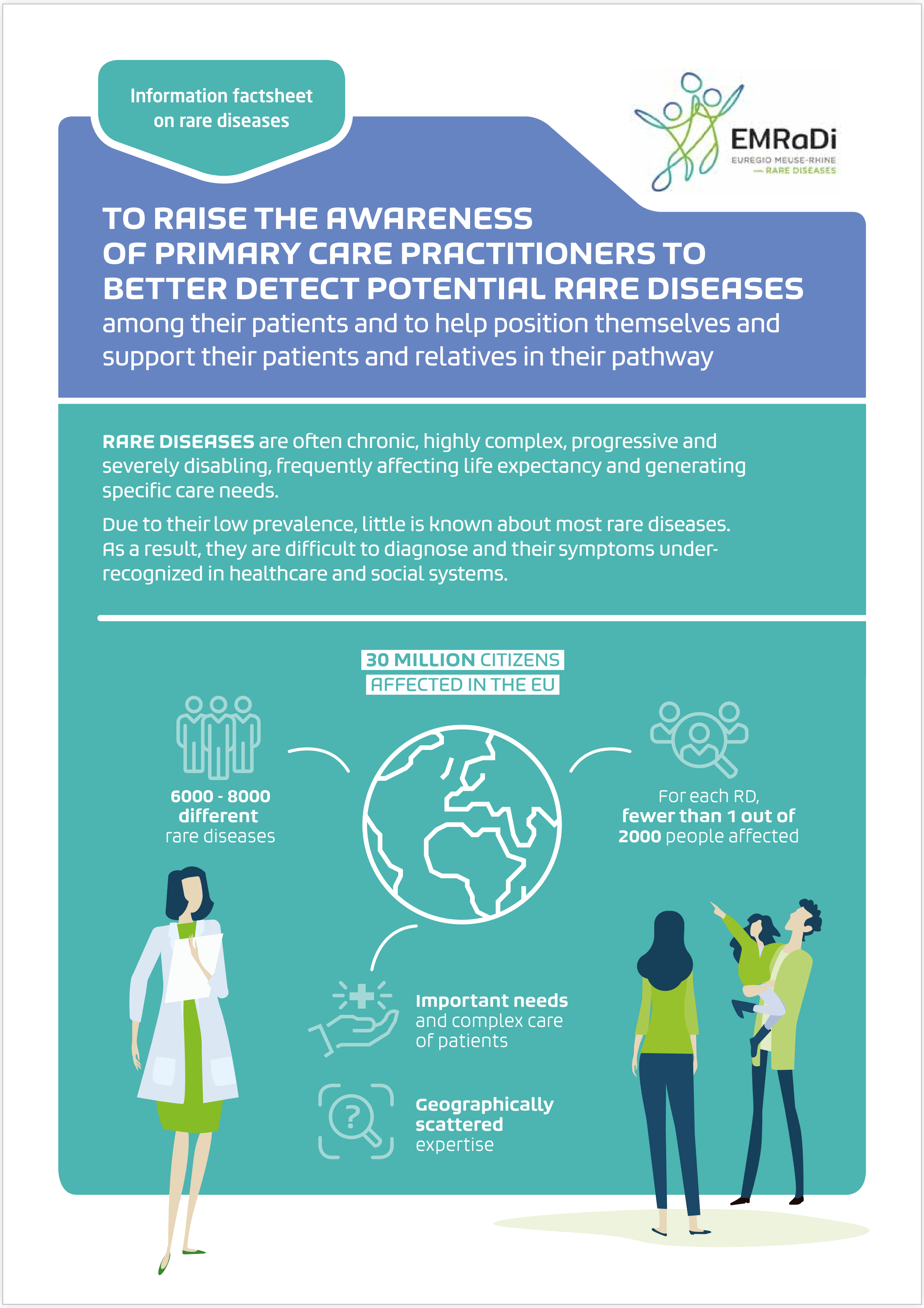
▶ www.cbhc.nl

The EMRaDi partners can also support you in your CBHC via their specific information material.

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

With the support of

APPENDIX 4: INFORMATION FACTSHEET FOR PRIMARY CARE PRACTICTIONERS



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.

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[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

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* 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.

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APPENDIX 5: SELECTION PROCESS FOR THE 8 RD

EMRADI PROJECT
Explanation of the Selection Process of the
Eight Rare Diseases to Focus on
Final version – 30 June 2017

GENERAL DESCRIPTION OF THE DECISION STRATEGY AND PROCESS

The EMRaDi Project was launched in October 2016 for a 3-year duration with the main objective to improve the quality of life of Rare Disease Patients in the Euregio Meuse-Rhine (EMR) and beyond. A big advantage of the project is that a lot of stakeholders from different countries and institutions come together. Among them are representatives of patient organizations, hospitals and mutual health insurance funds.

The project team formulated the following objectives when writing the application in the field of Rare Diseases:

1) Increase the transparency of needs and availability of services in the field of rare diseases in the EMR
2) Develop EMR models of RD patient pathways in order to elaborate patient-oriented recommendations in synergy with national and European developments
3) Improve the network of health care providers, mutual health insurance funds and patient organizations and raise (public) awareness of rare diseases

The long-term general aim is to improve the quality of life of these patients.

As the field of Rare Diseases (RD) is very large and diverse, one of the major aspects when starting the project was to decide which RDs to focus on. This was of course not an easy task as not only the RDs are diverse but also the partners. They have different expertise, backgrounds, approaches and cultures. Through different meetings with all of the partners' representatives from the start of the project (October 2016) until June 2017, the selection criteria and the list of potential RDs to focus on were discussed, argued and compared. The consortium of partners succeeded in reaching a consensus on the RD selection in June 2017, based principally on the cooperation possibilities and preferences expressed by the different partners. Choosing specific RDs was difficult but necessary as it will enable as concrete results as possible to be reached for these diseases.

The partners also expect that the choice has a positive impact on other rare diseases (for example, thanks to a better information and training of the health insurance funds and of the health experts) and hope that the EMRaDi project is a first step to a larger and long-term cooperation in the RD field in the EMR, to help as much RD patients as possible.

It should also be stressed here, that although in this initial phase only the three EMR University Hospitals were involved from the Health Care Provider side, an expansion to other hospitals and resident doctors in the EMR is already forecast within the project's runtime and an important condition to reach the project's objectives.

It should be mentioned that all three participating countries have "National plans of action for people with rare diseases" and have realized or plan to realize a certification process for centres for Rare Diseases. Whereas Belgium and Germany are still in the preparatory phase, only the Netherlands have already formally designated CoE, appointed by the Minister of Health.

When writing the application, it became clear that a project of this scope and funding should focus on a small set of diseases. An important part of the improvement possibilities for RD patients in the EMR is based on medical expertise. Therefore, it became clear that this expertise should be present at least at one of the partner's institutions.

It was already a discussion at the time of writing the application on which groups to focus (this is of course a little bit artificial as RDs often do not belong to one RD group only, which can be seen when looking at the Orphanet classification (www.orpha.net)). Rare neurological, rare haematological and rare syndromic diseases were defined quite quickly – with rare metabolic diseases as a further candidate group – by the participating University Hospitals in consensus with the other partners. In order to get a good representation of the diversity of rare diseases and a good overview of this field, it was decided to select two diseases per group, resulting in eight diseases altogether.

This EMRaDi project has a huge scope: its aim is to develop models, to organize a close contact with patient organizations, to write or revise patient guidelines[17] tailored to the EMR situation, to organize information sessions for professionals, to improve networking, etc. Therefore, the project partners had to restrict themselves to a small number of RDs.

After this process was finished and once the project started, a further strategy was elaborated. It was – to make a long story short – a) to define criteria which should be used for the decision and b) to collect data for the candidate RDs (around 70 diseases in a long list), in order to c) come to a consensus and final solution by the project's steering committee, where the leaders of the EMRaDi work packages, other representatives of the different institutions and further members of the project convene regularly. The project's task force of medical experts from the participating University Hospitals was included in data collection.

The following were the selection criteria:

- At least one of the participating hospitals should have expertise as a lot of results will depend on answers to medical questions;
- There should be a sufficient number of patients to be expected in the EMR – best from childhood and adulthood – for the field study and to help the most patients possible;
- There should be a balance between the type of symptoms (mental/physical impairments) and a wide range of difficulties/challenges for the patients resulting in a need for coordination between different types of healthcare/professionals.
- The point of existing patient organizations was taken into consideration as they are an important partner for the patients and health professionals.
- According to the project's objectives, the potential for improvement of a RD patient's situation in the EMR and cross border aspects of health treatment were also important to consider.
- One well-known model RD, where a lot has already been done and from which lessons can be drawn for other RDs at a higher level should also be selected.

Information collected included basic data (name, code, prevalence, etc), info on the health complaints and medical treatment, possible potential for improvement in the EMR, expertise within the EMR and/or Project Group, representation in one of the European Reference Networks and last but surely not least, existence of patient organizations within and outside the EMR.

We should emphasize the point that not every criterion had to be 100% fulfilled by each candidate. As usual, compromises had to be found. The discussion process, which took several months, resulted in the diseases listed in Table 1.

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[17] Please note that the word "guideline" is not directly connected with the Dutch word "richtlijn". The term "guideline" in this text is a summary term for all types of information papers, either for patients and relatives or health professionals.

OVERVIEW OF THE EIGHT RARE DISEASES

Rare Disease Groups	Specific rare diseases
Neurological diseases	Huntington disease
	Duchenne muscular dystrophy
Haematological diseases	Chronic myeloid leukaemia (CML)
	Polycythaemia vera (PV)
Syndromic diseases	Silver-Russell syndrome
	Rett syndrome
Metabolic diseases	Galactosemia type 1
	Phenylketonuria (PKU)

Tab. 1: Overview of selected rare diseases in the EMRaDi-Project

WHAT HAS TO BE EXPECTED?

The situation of patients with one of these eight diseases will be analysed in more detail in the EMRaDi-Project – e.g. interviews will be organized – and on this basis, information papers will be revised or established, model pathways developed, information sessions organized, and the networking between the different stakeholders increased.

Furthermore, the patient pathways within the three health systems can be compared to each other not only from a medical point of view, but also from care, advice, financing and other points of view. This project is a chance to describe the ideal (cross-border) healthcare process and its organization.

The core concept of a rare disease patient pathway encompasses all the steps from the first symptoms experienced by the patient, along his or her path through the health system towards (hopefully) a fast, successful diagnosis and including every aspect of the care – whether organizational, medical, social, psychological, legal and/or vocational – to finally ensure the patient the best possible quality of life.

A tight interlinkage with other projects and institutions for Rare Diseases is also foreseen, e.g. an exchange with the European Reference Networks.

This project is expected to improve the quality of life and quality of care of RD patients within the EMR and beyond.

ARGUMENTS FOR EACH OF THE EIGHT RARE DISEASES

HUNTINGTON DISEASE (HD) – NEUROLOGICAL RD

HD is a well-known hereditary rare disease and a neurodegenerative disorder leading to psychiatric, cognitive and motor complaints. The average onset of the disease is between 30-50 years and patients suffer from an increasing loss of independency and quality of life, during 15 to 20 years of the illnesses course. This disease leads to death, primarily by pneumonia, secondly by suicide.

Therefore, interdisciplinary efforts are needed as this disease is not only complicated from a medical point of view, but also from psycho-social, vocational, care and many other points of view.

All three participating University Hospitals have expertise in this disease – UKA Euregional Huntington's diseases centre Aachen (EHZA) – MUMC+ an (official) designated Dutch CoE – in Liège, a HD CoE is at ISoSL «le Pèri» (Intercommunale de Soins Spécialisés de Liège, Hôpital Pèrî) which is tightly interconnected with CHU. There is furthermore a big patient organization community opening the door for excellent EMR networking. Moreover, as a lot has been done already in the different countries, the project work can start at a higher level than for other rare diseases (well-known model RDs).

DUCHENNE MUSCULAR DYSTROPHY (DMD) – NEUROLOGICAL RD

DMD is a neuromuscular hereditary disease and therefore represents another group of rare neurological diseases. The onset of the disease, which primarily affects males, usually starts in childhood. Usually, the ability to move normally decreases rapidly, leading to death in youth and young adulthood.

So far, no medical cure is available and the care is multidisciplinary and very complex – encompassing amongst others physiotherapy, cardiology, surgery, as well as treatment with corticosteroids and other drugs. Comparable to HD, coordination of different types of healthcare professionals and others is important.

In all three countries, patient associations exist, the University Hospitals of Aachen and Maastricht showed interest with MUMC+ having a Neuromuscular Centre as an (official) designated CoE.

CHRONIC MYELOID LEUKAEMIA (CML) – HEMATOLOGICAL RD

CML is a rare haematological leukaemia disease and is the most common myeloproliferative disorder. Patients – usually adults – may be either not affected by health complaints or suffering amongst others from fatigue, weight loss and sweats – symptoms of the disease are mostly physical. So far, predisposition and the mechanism for onset of the disease are mostly unknown, but a permanent active tyrosine kinase is the cause of the disease. Allogeneic bone marrow transplantation is the curative treatment option, but the drug imatinib mesylate – an inhibitor of the respective tyrosine kinase – substantially improves the prognosis. Nevertheless, long-term treatment and care can be very challenging.

All three participating hospitals have CML experts and patient organizations also exist. Just like HD, CML is well-known with a higher potential to raise awareness of different target groups.

POLYCYTHAEMIA VERA (PV) – HAEMATOLOGICAL RD

At 1-5/10,000, the prevalence is quite high. In PV, the red blood cell production is impaired, leading to high blood hyperviscosity. Numerous complaints can be the consequence, most of which are physical in nature. There is an individual assessment necessary for treatment, but generally the aim is to decrease blood viscosity. Life expectancy is close to the usual value and the disease can occur at any age.

Within the EMRaDi Project partners University Hospitals Aachen, Liège and Maastricht, there is a broad expertise for this disease, which is an acquired myeloproliferative disorder. Also, patient organizations exist.

SILVER-RUSSELL SYNDROME (SRS) – SYNDROMAL RD

SRS is a rare congenital syndromic disorder mainly characterized by intrauterine and postnatal growth retardation, relative macrocephaly, a prominent forehead, asymmetry and feeding difficulties. SRS is primarily a clinical diagnosis, but in ~60% of patients it can be confirmed molecularly. Treatment is in the form of growth hormone therapy, with the prognosis when properly treatment being good. SRS belongs to the group of imprinting disorders, and can therefore be regarded as a model for these heterogeneous disorders. Furthermore, it is a differential diagnosis for many growth retardation diseases.

Many SRS patients are diagnosed and/or treated at the University Hospitals Aachen and Liège as special expertise is located there. Furthermore, there is a close collaboration with national and international patient organizations, resulting in international consensus guidelines.

RETT SYNDROME (RTT) – SYNDROMAL RD

Rett Syndrome primarily affects females - but more and more boys with a Rett-like phenotype and genotype are being identified. The classic onset of Rett syndrome, a severe neurodevelopmental disorder, is generally between the ages of 12 and 18 months, but this can vary greatly. Patients suffer from lifelong, complex medical problems with intellectual disability and much comorbidity. Treatment is therefore complex, only symptomatic and implies a lot of coordination between health, social care and others (involving physiotherapists, dieticians, occupational therapists, speech therapists and music therapists). Diagnosis can be made either on the phenotype (if classical) or by next-generation sequencing in case of less typical phenotypic features and/or at a young age. Usually, the lifespan is (somewhat) limited and prognosis depends on the severity of comorbidity.

Maastricht is the Dutch expertise centre for Rett syndrome as part of the Expertise Centre for Rare Syndromes and Cognitive Disorders. This is an example of a disease which is covered only by one of the participating hospitals. Patient organizations exist in the three countries.

GALACTOSEMIA TYPE 1 – METABOLIC RD

The onset of the disease is early in life. It presents in the new-born period as a life-threatening disease, whose clinical picture can be resolved by a diet without galactose. The dietary treatment proves, however, insufficient in preventing severe long-term complications, such as cognitive, social and reproductive impairments. The treatment and follow-up of this hereditable disease is complex and multidisciplinary and requires a team of experts with adequate knowledge of the disease.

The University Hospital of Maastricht has an (official) designated CoE, and the University Hospital of Liège has competence in this disease, which belongs to the group of rare metabolic diseases and, in particular, to the disorders of sugar metabolism. Patient organizations exist.

PHENYLKETONURIA (PKU) – METABOLIC RD

It is a disease which can be detected via new-born screening with – provided the right medical and dietary treatment is applied – a positive course for the disease. It is a model for this type of diseases, which belong to the field of rare metabolic diseases with hereditary origin. In PKU, the metabolism of an amino acid, an important element of proteins, is severely impaired leading to a heavy mental retardation and physical complaints of untreated patients. Patient organizations covering this rare disease exist in the three countries.

The main driver to include PKU in the short eight RD list is the University Hospital of Liège as it benefits from a lot of expertise. The University Hospital of Maastricht Hospital also has extensive experience in this disease.

CONCLUDING REMARK

Sources of disease-relevant information: www.orpha.net and health experts from the EMRaDi-Project, especially (in alphabetical order):

Maastricht UMC+:

Estela Rubio Gonzales (Paediatrician, Metabolic Diseases)
Merel Klaassens (Paediatrician)
Sylvia Klinkenberg (Child Neurologist)
Mayke Osterloo (Neurologist)
Harry Schouten (Internist - Haematologist)
Connie Stumpel (Clinical Geneticist, Neurologist)

CHU Liège:

Vincent Bours (Human Geneticist)
Saskia Bulk (Clinical Geneticist)
Kaoutar Hafraoui (Haematologist)
Zayd Jedidi (Neurologist)

UK Aachen:

Thomas Eggermann (Human Geneticist)
Steffen Koschmieder (Haematologist)
Kathrin Reetz (Neurologist)

and

Caroline Glaude, MC Verviers-Eupen; Kim Karsenberg, VSOP; Joyce Loridan, Solidaris; Clara Noirhomme, ANMC; Marèl Segers, VSOP; Sylvie Taziaux, CHU Liège and Christopher Schippers, UK Aachen

This document was written in cooperation with all the EMRaDi partners.

APPENDIX 6: EMR RARE DISEASE DAY 2019: SUMMARY

PLENARY FEEDBACK OF ROUND TABLE DISCUSSIONS

CROSS BORDER HEALTHCARE & COOPERATION

WHAT ARE THE EXPERIENCES (BOTTLENECKS AND SUCCESSES), REGARDING CROSS BORDER HEALTHCARE AND COOPERATION?

Bottlenecks

- CBHC reimbursement procedures are uncertain (level of reimbursement, etc.)
- Differences in clinical practices (access to medicines, diagnostic procedures, etc.)
- Cultural and language barriers
- Patients turn to Google instead of using expertise across the border
- End of IZOM (Integratie Zorg op Maat - previous system facilitating CBHC in the EMR on administrative and financial aspects)

Successes

- Collaboration between insurance funds (AOK, CZ, MC)
- ERNs supporting sharing and learning

WHAT ARE POSSIBLE SOLUTIONS FOR BOTTLENECKS AND WHAT ARE GOOD PRACTICES REGARDING CROSS-BORDER HEALTHCARE AND COOPERATION?

- Integrate ERNs into national systems
- E-health and multidisciplinary consultation of patient (ERN – Clinical Patient Management System (CPMS))
- E-health services are still not reimbursed! We need a common nomenclature for telemedicine services
- Pilot projects – patient pathways in the EMR (EMRaDi) for gaining experience and upscaling
- European fund for rare diseases
- International guidelines and adherence to them

HOW CAN PATIENTS, PATIENT ORGANIZATIONS, HEALTHCARE PROFESSIONALS, HEALTH INSURANCE FUNDS AND POLICY MAKERS CONTRIBUTE TO MAKE THESE SOLUTIONS HAPPEN?

- Collaboration between all stakeholders
- Raising awareness

AWARENESS

WHAT ARE THE EXPERIENCES (BOTTLENECKS AND SUCCESSES) REGARDING AWARENESS?

- GPs and other healthcare providers should know more about RDs
- Diagnostic delay and late recognition of symptoms the diagnostic delay has to be reduced
- Patient organizations and healthcare providers do not know enough about RD centres
- Patients and healthcare providers do not know where to find CoE as they are not mentioned enough publicly
- GPs have to know about the existence of CoE and patient organizations, so they can lead the patient to the right source of information

WHAT ARE POSSIBLE SOLUTIONS FOR BOTTLENECKS AND WHAT ARE GOOD PRACTICES REGARDING AWARENESS?

- CoE should have more publicity in medical magazines, internet, etc.
- More financial investment in CoE
- Red flag list for RD (but be aware: only checking the red flags is not enough!)
- Patient organizations should play an important role in putting these CoE on the map

HOW CAN PATIENTS, PATIENT ORGANIZATIONS, HEALTHCARE PROFESSIONALS, HEALTH INSURANCE FUND AND POLICY MAKERS CONTRIBUTE TO MAKE THESE SOLUTIONS HAPPEN?

- RDs should be discussed internationally (raising international awareness is necessary)
- Data exchange between different RD centres
- Conferences to connect experts all over the world. In case of non-specific symptoms, healthcare providers should be aware that a manifestation of a rare disease could also be a possibility. RDs should get high priority, especially when common causes are excluded
- We have to continue to talk about patients' stories to improve public understanding of RDs

UNDIAGNOSED

WHAT ARE THE EXPERIENCES (BOTTLENECKS AND SUCCESSES) REGARDING PATIENTS WITHOUT A DIAGNOSIS?

- Diagnostic delay is very common and you need to find the right partners and be referred to the right expert without losing precious time
- Syndromes are in need of multidisciplinary care
- Need of a coordinator who is in charge of incoming data of different experts (keeping an overview!)
- Up to now, we have been too dependent on coincidence to find the diagnosis

WHAT ARE POSSIBLE SOLUTIONS FOR BOTTLENECKS AND WHAT ARE GOOD PRACTICES REGARDING PATIENTS WITHOUT A DIAGNOSIS?

- Coordinator
- Education at all levels
- Raising awareness continuously via different channels
- Make use of new technology
- Need for collaboration between professionals and patients and professionals amongst themselves

HOW CAN PATIENTS, PATIENT ORGANIZATIONS, HEALTHCARE PROFESSIONALS, HEALTH INSURANCE FUNDS AND POLICY MAKERS CONTRIBUTE TO MAKE THESE SOLUTIONS HAPPEN?

- Be "angry" together!
- Better policies for reimbursement of cross-border healthcare
- We should raise awareness at a political level
- Start platforms for professionals in healthcare institutions and undiagnosed patients

MATERIAL

On the EMRaDi website, you will find the material from the day:

- The PowerPoint presentations of the speakers
- The drawings illustrating the day
- A video in which participants reflect on the day

APPENDIX 7: EMRADI CLOSING EVENT – 17.02.2020: SUMMARY

EMRaDi closing event: Optimising pathways of rare diseases patients thanks to cross-border cooperation in the Euregio Meuse-Rhine

On 17th February 2020 at the Palais des Congrès in Liège, the partners of the EMRaDi project (Euregio Meuse-Rhine Rare Diseases) presented the results of more than three years of cross-border cooperation to optimize rare disease patient pathways in the Euregio Meuse-Rhine (EMR).

MEP Pascal Arimont gave the opening speech, stressing the importance of projects like EMRaDi, a project that greatly improved his awareness for rare diseases, and the need to increase funding for cross-border projects in the field of health. Afterwards, Axel Noël (Interreg regional antenna in Liège) argued in favour of more cooperation in the EMR before giving the floor to the project partners, who presented their achievements.

After Caroline Glaude (Verviers-Eupen Christian Health Fund, Lead Partner) provided a brief overview of the project, Joyce Loridan (National Union of Socialists Health Funds, Belgium) and Clara Noirhomme (National Alliance of Christian Health Funds, Belgium) presented the qualitative study carried out based on 104 testimonies of patients, relatives and professionals that aimed to uncover the day to day reality of RD patients and their relatives. Then, Timo Clemens (University Maastricht) gave an overview of the legal and financial provisions applicable in the EMR and formulated recommendations to ease data exchange and develop telemedicine. Rok Hrzic (University Maastricht) presented integrated care models (from slide 19). Among other things, these studies advocate for a holistic approach[1] and more cross-border cooperation. They provide solid arguments to introduce "case managers" to provide more support to RD patients and improve care coordination.

To start the afternoon, Vincent Bours (CHU de Liège), Christopher Schippers (UK Aachen), and Connie Stumpel (Maastricht UMC+) showed how the three university hospitals of the EMR improved not only the cooperation amongst themselves but also beyond, and carried out joint actions to raise awareness, especially among health professionals. Following this, Cor Oosterwijk (VSOP) presented the recommendations formulated by the Patient Sounding Board that was created within the project. He called for higher levels of cooperation between the stakeholders of the three countries and for everyone to make the patient perspective their own.

The day ended with an inspiring exchange of views between two patient representatives, Jan de Jong (Hematon, NL) and Eva Schoeters (RaDiOrg, BE), Saskia Bulk (CHU de Liège, BE), Chantal Mathy (Belgian Health Institute) but also with the audience, to discuss the EMRaDi results and address the remaining challenges to be tackled in future actions and projects.

The interest of having **pilot projects** in the EMR was underlined to propose new ideas to other Member States and more largely at EU level. The importance for **health insurance funds** to be proactive when they are dealing with patients having a rare disease was highlighted, also in relation with the **RD status** to improve in order to be used as a real red flag. The **case manager** was also promoted as path manager who could be located at the patient association. There was a strong agreement to promote **telemedicine** at national and cross-border levels and to develop it in coordination with the **ERN** and with specific attention to data protection. Regarding the EU and solidarity, the participants suggested developing solutions to **drug pricing** (a fair and maximum European pricing and an evaluation of cost-effectiveness were proposed) and **easing cross-border healthcare** for RD patients. The **EBM approach** as a rule for reimbursement of certain drugs and alternative medicines was discussed, as scientific evidence is difficult to prove in the case of rare diseases due to the limited number of patients for each disease. In Belgium, a **Special Solidarity Fund** exists to reimburse specific treatments and drugs for rare disease patients when the criteria are met.

Caroline Glaude concluded the day, by summarizing the **8 accomplishments** of this cross-border cooperation on rare diseases:

1. Gathering of the main stakeholders in the EMR to increase expertise and cooperation;
2. Analysis of patients' needs along their pathways as well as the legal and financial framework;
3. Solution proposals to improve the quality of life of patients and their families thanks to faster diagnosis and patient-centered, integrated, high-quality care near the place of residence;
4. Raising awareness;
5. Patient involvement in research on healthcare and patient empowerment;
6. Catalyst for the three countries' national plans;
7. Contribution to the implementation of the European Directive 2011/24/UE on the application of patients' rights in cross-border healthcare, including the European Reference Networks;
8. Development of rare diseases patient pathways in a cross-border and European context, and recommendations in line with the European and national contexts.

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