# WORK PACKAGE 5 REPORT

## PATIENT SOUNDING BOARD RECOMMENDATIONS

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|---|---|
| **Lead partner of Work Package** | **VSOP** |
| **WP n° and title** | WP5 Patient involvement and public awareness |
| **Activity n° and title** | 5.1 Consolidated Healthcare Policy<br>5.2 Patient involvement in medical research<br>5.3 Quality of care in relation to Cross Border Healthcare<br>5.5 Raising awareness |
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lead partner

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The Interreg V-A Euregio Meuse-Rhine (EMR) programme invests almost EUR 100 million in the development of 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.

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## PROJECT DESCRIPTION

"EMRaDi" stands for **Euregio Meuse-Rhine Rare Diseases**.
The project started on 1st October 2016 and ended on 31st March 2020

This project involved a **cross-border cooperation** between health insurers, university hospitals, patient associations and a university in the Euregio Meuse-Rhine. It was part of the European Union INTERREG V-A Euregio Meuse-Rhine programme.

Thanks to their long experience in cross-border healthcare, the project partners have decided to join forces in the specific field of rare diseases. This EMRaDi project was innovative in the sense that it was a patient-oriented and cross-sectoral project. The consortium of partners included the major health players who support rare disease patients and their relatives in their day-to-day rare disease patient pathway.

Through **the project activities**, the EMRaDi project aimed to:

- increase the transparency of needs and availability of services in the field of rare diseases in the Euregio Meuse-Rhine (EMR);
- develop EMR models for *rare disease patient pathways* in order to draw up patient-oriented recommendations in synergy with national and European developments;
- improve the network of healthcare providers, health insurance providers and patient organisations and raise (public) awareness of rare diseases.

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

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## LEGAL ISSUES

## EXECUTIVE SUMMARY

One of the activities in Work Package 5 (WP5) was installing a Patient Sounding Board (PSB). The PSB was a consultative body that consisted of (patient) representatives from the selected rare diseases (Huntington, Duchenne, Chronic Myeloid Leukaemia (CML), Phenylketonuria (PKU) and Silver Russel). During three PSB meetings, its members gave feedback discussed the following topics:

- EMR Rare Disease Day 2019;
- National Plans for Rare Diseases;
- Quality Documents;
- Information Cross-Border Healthcare;
- Patient participation in medical research;
- Concept versions of models and products developed in other EMRaDi work packages.

This document summarizes the consensus-based PSB recommendations on these topics. The most important first-to-address stakeholder for the recommendations is the EMRaDi Steering Committee, that subsequently should bring them to the attention of stakeholders such as: National Contact Points (development), healthcare insurers (in general), hospitals (focused on rare diseases), patient organisations (including their umbrella organisations) and governmental policy officers at national and EU level. The most important conclusions for a selection of the topics discussed are the following:

**National Plans for Rare Diseases**: the topics that should be given priority are:

- awareness;
- knowledge and education, in which the patient organisations have an important role;
- organisation of care and care networks (including the psychosocial domain);
- availability of therapy (including diagnosed and undiagnosed patients);
- policy regarding centres of expertise;
- quality standards.

**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 gets a life of its own”;
- written in a "reader friendly" language.

A top-5 quality documents was listed for the 3 RD on which the EMRaDi project focuses.

If formal rare disease treatment guidelines for care providers are developed, the PSB recommends that a version for patients is also budgeted and developed.

**Cross-border healthcare (CBHC)**: According to the PSB, the main sources of information on cross-border healthcare should be the National Contact Points and Orphanet. All other stakeholders involved in cross-border healthcare and/or rare diseases should refer to these organisations for information on cross-border healthcare (National Contract Points) and rare diseases (Orphanet). National Contact Point’s website should develop a page specifically dedicated to rare diseases.

**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.

## INTRODUCTION

A Patient Sounding Board (PSB) was installed, consisting of patient representatives for the following EMRaDi selected rare diseases:

- Huntington disease;
- Duchenne muscular dystrophy;
- Chronic myeloid leukaemia (CML);
- Phenylketonuria (PKU);
- Silver-Russell syndrome.

Furthermore, a representative from RaDiOrg, the Belgian alliance for rare and genetic diseases, participated in the three PSB meetings. The original aim was to include one patient representative for each disease in each region. Despite many efforts via newsletters, websites, social media and individual contacts however, this was unsuccessful – patient representatives are usually very busy volunteers and limited in their options because of their own disease or that of their child. Therefore, no EMR patient representatives could be found for Polycythaemia vera (PV), Rett syndrome and Galactosemia type 1. Three of the representatives were from Belgium and three from The Netherlands. During the EMRaDi-project, multiple attempts were made by different stakeholders and project partners to involve German representatives. Before the last PSB meeting, a German representative was found. She worked on the PSB-3 homework assignments, but could not attend the last PSB meeting.

During the three meetings, different topics were discussed that are of relevance for the EMRaDi project. Some topics were discussed during more than one meeting, others only once. The topics discussed were:

- EMR Rare Disease Day 2019;
- National Plans Rare Diseases;
- Quality Documents;
- Information Cross-Border Healthcare;
- Patient participation in medical research;
- Models and products developed in other EMRaDi work packages.

The PSB gave input on these topics from the patient perspective. The way the PSB was set-up was as follows: to allow participants to give their input as complete and well prepared as possible, they were given ‘homework assignments’ for some of the topics discussed during the meeting. These assignments were developed and prepared by VSOP. The PSB-members had to hand in the assignment(s) a few weeks before the meeting and the assignment(s) were then processed by VSOP. The processed assignments were discussed during the meeting so that everyone could visualise each other's feedback and discussion could start from that point. After discussing the topic in one or more meetings, recommendations for the project partners or other stakeholders were formulated.

Reports were made of each of the meetings, which are available on request (in English only).

# 1 PATIENT SOUNDING BOARD

## 1.1 MEETINGS

In total, three PSB meetings took place throughout the EMRaDi project. The objective was to have one meeting per project year. The first meeting was postponed for a few months since it took some extra time to find suitable people that were able to take part in the PSB.

- The first meeting took place on 14th June 2018, 11am – 2pm at Maastricht MUMC+.
- The second meeting took place on 8th November 2018, 10.30am – 2pm at Maastricht MUMC+.
- The third meeting took place on 23rd May 2019, 10.30am – 2pm at Maastricht MUMC+.

If the PSB members could not attend a meeting, they prepared the 'homework assignment'. In that way, their input and feedback could be used and shared during the meeting that followed.

# 2 PATIENT SOUNDING BOARD RECOMMENDATIONS

## 2.1 EMR RARE DISEASE DAY 2019

One of the topics discussed during the first PSB meeting was the program for the EMR Rare Disease Day 2019 (EMR-RDD 2019) organised by the EMRaDi project partners. The most relevant recommendations regarding this topic were:

- The formal date of the international Rare Disease Day, 28th February, could be an issue. On this day there are also a lot of local activities. As a result, not everyone interested may be able to attend, including interesting speakers.
- It would be interesting to invite ministers from the three border countries/regions/municipalities. But later on, it was suggested that a representation of ministers might be better suited during the political event in 2020 or the closing meeting.
- It should be in the program how regional cross-border healthcare is related to the European Reference Networks (ERN's). In addition, during the day it should be possible to share experience and knowledge regarding care for rare diseases between the three countries.
- It is important to personally invite representatives of the different ERN's.
- The suggestion to use a moderator during the day is supported by the PSB.
- The suggestion to present successful examples of cooperation between different parties within the region during the EMR RDD 2019 is also supported by the PSB.
- The PSB recommends linking the topics of the three shared presentations to the workshops/round table discussions.
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## 2.2 NATIONAL ACTION PLANS FOR RARE DISEASES

The National Plans of the three regional countries (Netherlands, Belgium and Germany) were discussed during the first and second PSB meeting. During the first meeting, the three national plans were compared and six topics were selected as topics that should be given priority. These topics are:

- Awareness;
- Knowledge and education in which the patient organisations have an important role;
- Organisation of care and care networks (including the Psychosocial domain);

- Availability of therapy (including diagnosed and undiagnosed patients);
- Policy regarding centres of expertise;
- Quality standards.

In preparation of the second PSB meeting, the patient representatives performed a so called 'gravity analysis'. The recommendations following this analysis regarding the national plans are:

**Awareness**: Awareness about rare diseases must be increased by informing General Practitioners (GPs) and specialists in the 2nd and 3rd line of healthcare. This concerns information about centres of expertise 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 centres of expertise, would be ideal.

Recommendation: Develop a kind of (GP) standard based on symptomatology which includes a decision tree that reflects the route of care for the general practitioner that has a feeling something is wrong.

**Knowledge and education**: awareness is the basis for knowledge and education. There is already a lot of information and knowledge available, but not always focused on the patient.

Recommendations:
Bring the knowledge that exists where it is needed by developing patient-oriented information/a patient version of guidelines with images and drawings. Good examples are:

- For CML: CMyLife has been developed for CML (by Radboudumc Nijmegen, The Netherlands) to help patients with their self-management. The site contains information about adherence, results and what questions patients could ask their doctor.
- For Huntington: In addition to the websites of the national patient organisations, there is useful multi-language documentation on several European websites like European Huntington's Disease Network EHDN), Huntington's Disease Youth Organization (HDYO) and the European Huntington Association (EHA).
- Orphanet France has important information available for patients, but only in French. This is not the case for other national Orphanet-sites.
- The Dutch Connect projects[1] connect centres of expertise into networks of expertise with a common digital platform. Variants of this format for a network of centres of expertise could be developed for the eight disorders in the EMR region.

**Organisation of care, and care networks**:

Recommendations:

- To provide insight into where specialised (centres of expertise) care (in the region) is available for at least the eight diseases. This concerns the name and address of the multidisciplinary team and not the individual care provider. This could be made clear with a survey.

[1] The Connect and Connect Extended projects are coordinated by VSOP and the Dutch neuromuscular patient organization. In these projects, nationally designated Centres of Expertise for the same (cluster of rare) disease(s) present themselves via a common website with dedicated information on care, referral and research, addressing both peripheral care providers and patients.

- To provide insight into where specialised long term (regional) care is available for at least the eight diseases.
- Exchange of knowledge between ECs themselves and between EC and patients.

**Availability of therapy**:

Recommendations:

- By increasing awareness, more fundraising may follow.
- Make clear which (paramedical) networks exist (in the region), so that it is obvious what can be done to maintain quality of life as long as possible.
- Provide insight into the organisation of care and how this relates to guiding the patient in making choices (shared decision making) in different phases of the care process (including psychosocial aspects).

**Policy regarding centres of expertise**:

Recommendations:

- Make clear where the centres of expertise for the relevant diseases are.
- Accomplishing a national review of centres of expertise, the Dutch model, including assessment by patient organisations, could be used as a starting point.
- Centres of expertise must become part of the healthcare network (in the region).

**Quality standards**:

Recommendations:

- Develop quality of care standards in collaboration with the relevant patient organisation and make patient versions of these quality standards.

## 2.3 QUALITY DOCUMENTS

The “Quality Documents” topic was discussed during all three PSB meetings. During each meeting, a different element of quality documents was discussed.

The first meeting focused on what a Quality Document is. According to the PSB members, Quality Documents:

- Are validated/authorised by the relevant professional group(s) and patient organisation(s).
- Are transparent (in interests/importance, literature and stakeholders).
- Have version management, documents must be properly encrypted to prevent “it gets a life of its own”.
- Have to be written in a "reader-friendly" language. This means that the PSB recommends a patient's version of the Quality Document be drafted for documents such as guidelines and good clinical practice guidelines, so that information is readily available in accessible language and also contains a format that makes it worth reading.

During the second and third PSB meeting, the PSB-members made a top-five of the quality documents they think are the most relevant for their disease. To be as complete as possible, the PSB members asked to approach the patient groups (so-called ePAGs) in ERNs to complete the list of quality documents. VSOP approached the ePAG members from Germany, Belgium and The Netherlands, but they were not able to add documents to the list, due to arguments like: not representing the right diseases, the list for their specific disease was already complete, etc.

A top-five quality document was developed and discussed during the third PSB meeting for PKU, CML and Huntington:

PKU
1. General Practitioner brochure for Phenylketonuria
2. Dietary information for treatment of Phenylketonuria
3. Phenylketonuria (PKU): What's next?
4. Screening of the general public (Flemish government)
5. European guidelines for PKU (three guidelines)

CML
1. Treatment guideline (Cmylife)
2. Treatment guideline (Onkopedia)
3. CML patient booklet
4. Treatment guideline (Leukaemie-hilfe)
5. CML Dutch guidelines (Hematologie Nederland)

Huntington
1. Huntington General Practitioner brochure
2. Management of speech and communication
3. Swallowing problems and nutrition (three international guidelines)
4. Physical therapy (five international guidelines)

During the meeting, it became clear that additional efforts to share these documents between the patient organisations in the Netherlands, Germany and Belgium are not necessary. Currently these patient organisations are already sharing (and translating) these type of documents, not only between the three countries, but also on a broader European/international level. The only point of attention from the PSB to the SC is that Belgium is not currently represented on a European/international level for CML and Silver Russel. This should be encouraged.

The PSB recommends everyone, both funders and care providers, who is involved in the development of professional quality standards and treatment guidelines for rare diseases, to also always develop a version for patients, and to guarantee the necessary budget to do so. In addition, patients should have access to patient-adapted information on centres of expertise.

In relation to the EMRaDi project, this principle could also apply to the "Red Flags" list. Its patient version could be a digital document that is made available through the websites of the three national patient umbrella organisations for rare diseases, in the national languages and possibly also in English, so that it can be shared at European and international levels.

## 2.4 INFORMATION ON CROSS-BORDER HEALTHCARE

One of the objectives of the EMRaDi project is to further improve cross-border healthcare between the three EMR countries. Patients with a rare disease in the EMR region should experience more support and coordinated access to the relevant (centres of) expertise. During two of the three PSB meetings, the topic "information about cross-border healthcare" was discussed. During the second PSB meeting, members defined which questions should be asked regarding information on cross-border healthcare. In preparation of the third PSB meeting, the PSB members looked at the websites of relevant stakeholders with those questions in mind. During the last and third PSB meeting, the results were discussed and recommendations were given.

<u>The five most relevant questions to be asked regarding cross-border healthcare are:</u>

- What type of information is needed on cross-border healthcare?
- Where should this information be available?
- Who should provide this information?
- How should this information be offered?
- To what extent should information on cross-border healthcare be related to the nationally designated centres of expertise?

The PSB looked for each of the participating countries (Belgium, Germany and the Netherlands) on the websites of relevant stakeholders: health insurers, hospitals and National Contact Points.

A summary of the recommendations given by the PSB:

- The recommendation is that the websites of the hospitals should refer to Orphanet and the National Contact Points for information on rare diseases and cross-border healthcare. In addition, the website of the hospitals/centres of expertise should only contain additional information that is specific to that particular hospital and/or the rare diseases involved (the exceptions to the rule). This is to prevent information from being repeated or not being up-to-date.
- Regarding the National Contact Points, make sure that the information is offered in pieces, that it is accessible to the average reader and that more information is available via a link for the reader that wants to know more.
- There is currently no general information available on rare diseases on the websites of the National Contact Points. The PSB members believe that this topic deserves a separate heading on the webpages.
- It is recommended that health insurers offer information in multiple languages. In the case of Belgian healthcare insurers, the advice would be to provide the information in French, Dutch, German and English. In the case of Dutch and German healthcare insurers, the advice would be to provide information in English in addition to Dutch and/or German.

Advice to the Steering Committee (SC):

- Investigate how the SC can encourage physicians working in the academic hospitals involved to participate in a national assessment procedure.
- Also look at how the SC can encourage these physicians to refer to centres of expertise in the region if the hospital does not have the expertise about a rare disease. This would be instead of

referring the patient to a centre of expertise elsewhere in the country concerned, at a larger travel distance.
- Also use the information already available on Orphanet and do not make a separate overview of the centres of expertise in the region. This would also promote long-term implementation in other border regions.
- Make contact with National Contact Points and patient umbrella organisations for rare diseases to discuss the advice above and to see how the provision of information on cross-border healthcare can be further improved.
- Look at how the provision of information on cross-border healthcare by the health insurers can be further improved.

In addition to these recommendations concerning the websites of the relevant stakeholders mentioned above, the PSB also recommends that:
- The website of the relevant patient organisations (umbrella organisations) should provide information about the centres of expertise with a link to Orphanet and instructions on how people can find nationally designated centres of expertise for rare diseases.
- National patient umbrella organisations should require a formal national procedure for the assessment of candidate centres of expertise, 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[2]. In the future, being appointed via such a procedure should be the only way for them (hospitals/centres) to be able to be part of an ERN.

## 2.5 PATIENT PARTICIPATION IN MEDICAL RESEARCH

In the context of the project, an inventory was made among the project partners involved, as to whether there is patient participation in scientific research and subsequently how this participation took place.

The PSB advises to involve patient organisations more in scientific research, especially at the start of a process, such as when formulating the research agenda, determining the logistics, design, etc. A 'best practice' that resulted from the inventory is the way patient involvement is structured in international research for Duchenne. The PSB advises to work according to this model and to use the Duchenne experience to develop a stepwise action plan to better involve patients in the research process, starting with the eight selected disorders. However, such an action plan must be linked to the research context outside the EMR region given that all studies for rare diseases can only be multicentre and multinational, because of their rarity. The PSB advises funders of research for rare diseases 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 should take care of this.

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

## 2.6 FEEDBACK FOR OTHER WORKPACKAGES

During two of the PSB meetings, the PSB members also gave feedback on the outputs that other work packages of the EMRaDi project were developing. This concerned:

- WP3: Concept model of the organisation of care;
- WP4: 'Red Flag' list of rare diseases.

Since these recommendations were output-specific and were incorporated in the next versions of the two deliverables, they aren't described in more detail in this report.

## 2.7 FINAL ACTIONS

During the final months of the projects, VSOP will continue its work on (recommendations concerning) information on cross-border healthcare adapted to the different needs of national contact points, health insurers and centres of expertise. In addition, based on existing guidelines for patient participation in research, a specific guideline for centres of expertise will be developed. For the development of products for the centres of expertise, VSOP will align with the Maastricht centre of expertise for Kabuki (WP4).

# 3 CONCLUSIONS

The recommendations from the patients' perspective provided by the PSB via VSOP, as well as by VSOP, were highly instrumental in the EMRaDi project. Practical advice on the EMR Rare Disease Day 2019 was taken over as much as organisationally possible, and several work packages profited from the PSB recommendation for their deliverables.

The above-mentioned recommendations for the EMR region related to the national plans, Quality Documents, information on cross-border healthcare, and patient participation in medical research had a prominent place in the political event (5th December 2019, Brussels), closing event (17th February 2020, Liege) and follow-up of the project.

# 4 LIST OF ABBREVIATION

| | |
|---|---|
| **CBHC** | Cross-Border Healthcare |
| **CML** | Chronic Myeloid Leukaemia |
| **CoE** | Centre of Expertise |
| **EMRaDi** | Euregio Meuse-Rhine Rare Diseases |
| **EMR** | Euregio Meuse-Rhine |
| **ERN** | European Reference Network |
| **ePAGs** | European Patient Advocacy Groups |
| **GP** | General Practitioner |
| **NCP** | National Contact Point |
| **PKU** | Phenylketonuria |
| **PSB** | Patient Sounding Board |
| **PV** | Polycythaemia vera |
| **RDD** | Rare Disease Day |
| **SC** | Steering Committee |
| **VSOP** | Vereniging Samenwerkende Ouder- en Patiëntenorganisaties |