



# EMRaDi Project Results

Current situation, diversity and implications for patients

Caroline Glaude

Euregio Meuse-Rhine Rare Disease Day  
28 February 2019 - Maastricht

## EMRaDi – a team of partners



Mutualité chrétienne de Verviers-Eupen (lead partner)



Alliance nationale des Mutualités chrétiennes



Zentrum für Seltene Erkrankungen Aachen (ZSEA)

Solidarity is good for health!



Maastricht University



# EMRaDi – a team of partners with ... cofinancers and associated partners



provincie limburg



Wallonie

**Interreg**  
Euregio Meuse-Rhine  
European Regional Development Fund



EUROPEAN UNION

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





INNOCare

oncoare



**RD CODE**

## What is a rare disease?

OVER  
**6000**

distinct rare  
diseases

Each one affects  
fewer than  
**1 IN**  
**2000**  
**PEOPLE**



All together, an  
estimated

**30**  
**MILLION PEOPLE**

are living with a rare  
disease in Europe



Expertise, knowledge,  
information on diseases and  
their consequences are **scarce**  
and difficult to access



**Rare, complex, chronic,**  
**disabling, progressive,**  
**degenerative, often**  
**life-threatening**

**NO**  
**CURE** 

for the vast  
majority of  
diseases and  
few treatments  
available

They are **geographically**  
**scattered** and often  
**isolated**

**Few experts,**  
geographically **scattered**  
**Research is fragmented**

# Juggling care and daily life: The Balancing Act of the Rare Disease Community

First Europe-wide survey on social impact of rare diseases,  
involving 3000 rare disease patients & carers



[Report with full results](#)



[Presentation of key results](#)



[Infographics with key results](#)



[Factsheet on integrated care for rare diseases with survey results](#)

Survey performed via EURORDIS survey initiative  
Rare Barometer Voices  
and within the EU-funded INNOVCare project





# **European policy on rare diseases and cross border health care with emphasis on cooperation and coordination**

# Why coordination and cooperation at EU level is valuable?

- national approaches are inefficient and ineffective
- patient numbers are low and scattered
- Resources are limited and scattered.
- strengthen solidarity

**=> economies of scale**





# How coordination and cooperation at EU level is facilitated?

## Law and Policy



# How coordination and cooperation at EU level is facilitated?

## Law & Policy

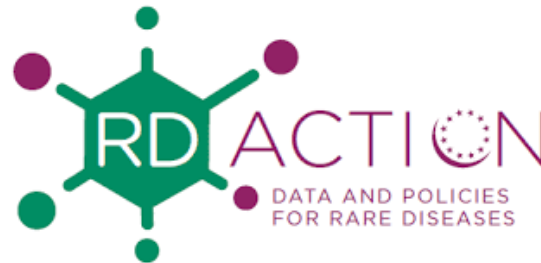
- EC 141/2000 Orphan Drug Regulation
- Communication on rare diseases: Europe's challenges COM(2008) 679
- Recommendation on an action in the field of rare diseases (2009/C 151/02)
- Directive 2011/24/EU on the application of patients' rights in cross-border healthcare (2011), Art. 12 on ERNs & Art. 13 on RDs

# How coordination and cooperation at EU level is facilitated?

## Law and Policy



## Joint Actions/ERNs



# How coordination and cooperation at EU level is facilitated?

## Joint Actions /ERNs

- 2009-2012: RD Task Force Joint Action
- 2012-2015: EUCERD Joint Action
- 2015-2018: RD-Action
- 2016-2019: JARC (EU JA on Rare Cancers)
- since 2017: 24 European Reference Networks (ERNs) are operational

# How coordination and cooperation at EU level is facilitated?

## European Reference Networks



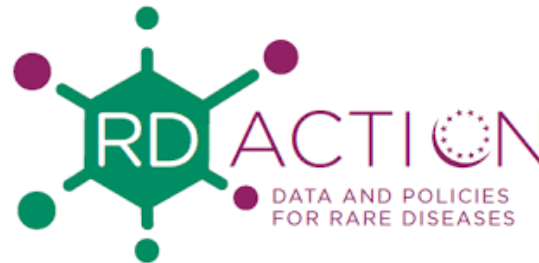
- 24 ERNs started in March 2017
- involving 900 units in >300 hospitals in 26 MSs
- > 250 patients treated until end of 2018

# How coordination and cooperation at EU level is facilitated?

## Law and Policy



## Joint Actions/ERNs



## EU expertise



## What specific measures are taken for RD coordination and cooperation at EU level?

orphanet



>RD\*CODE



# Upcoming and future EU initiatives for rare diseases and cross-border health care

## Health/RD

- Revision of the Orphan Drug Regulation
- Rare2030 – Foresight study on RD policy 2020-2030,
- ERNs
  - Expanding existing and launching new networks
  - Integration with national healthcare systems
- Implementation and scaling up of ehealth services



# Upcoming and future initiatives for rare diseases and cross border health care?

## Structural/General



Co-funded by  
the European Union

- Funding for RD policy & cross-border collaboration secured in the new MFF 2021-2027
- Proposal REGULATION on the ***European cross border mechanism*** COM/2018/373 final
  - (1) a legal mechanism and
  - (2) case-by case framework for agreements to overcome legal & admin obstacles in border regions**=> allows for the application in one MS of the legal provisions from another MS**

# EMRaDi project

## Euregio Meuse-Rhine Rare diseases



- Duration: 10/2016 to 03/2020
- Budget: 3.375.350,65 €
- [www.emradi.eu](http://www.emradi.eu)



# Objectives of the project

**Objective 1: Transparency** of needs and availability of services

➔ What is the current situation for patients ?

**Objective 2: EMR models** of RD patient pathways and recommendations for patients

➔ How do patients find their (best) patient pathway ?

**Objective 3: Network** of health professionals and raising (public) awareness

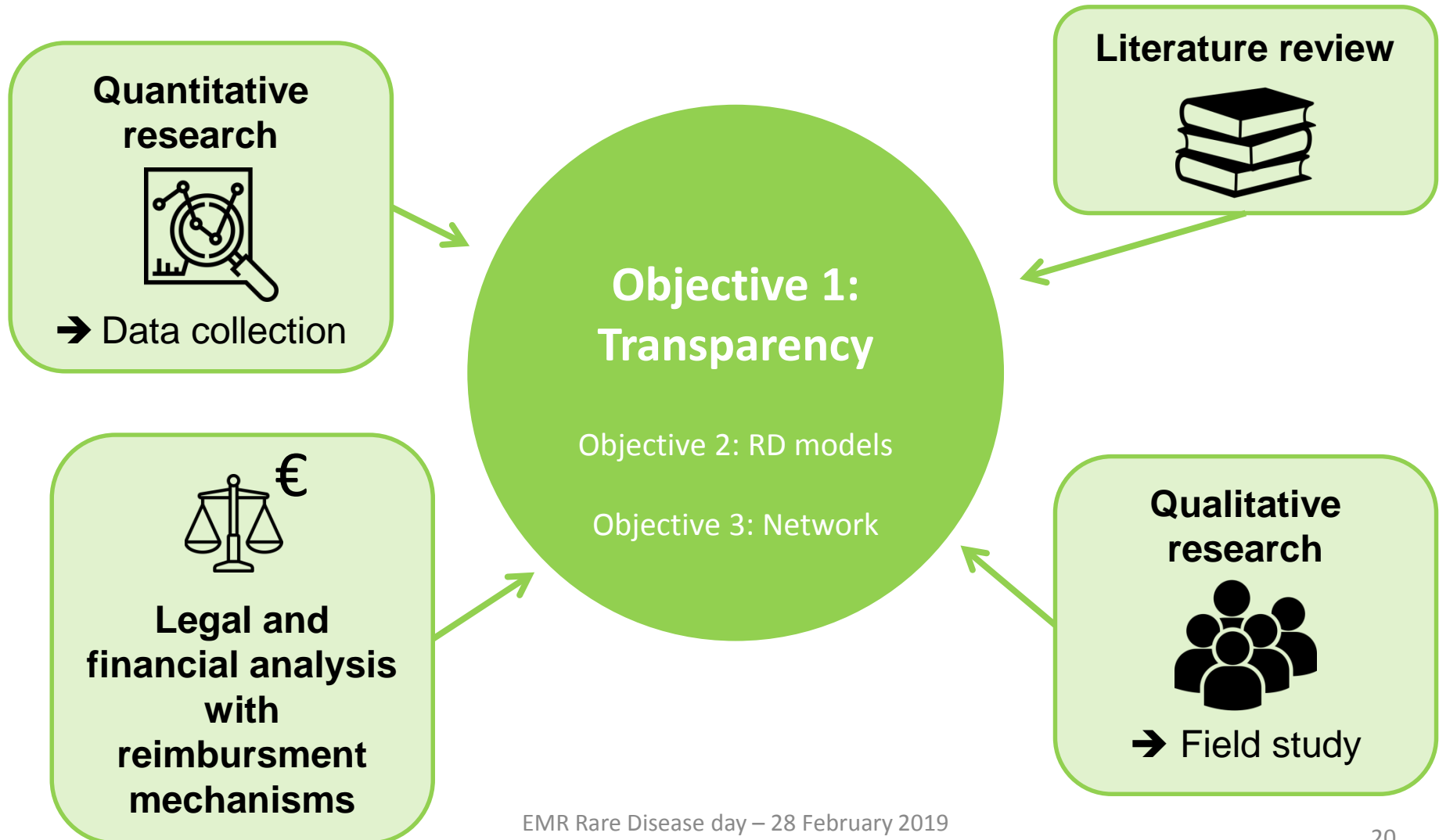
➔ How can professionals and patient associations cooperate together across the borders and better help the patients and their relatives ?



Long term general aim: improve the **quality of life** of the RD patients.

# Objective 1

## Transparency of needs and availability of services



# Objective 1

## Transparency of needs and availability of services



### Data collection -> EMRaDi Recommendation 1:

Harmonize data collection methods and create paths to access health data for health services research in the field of rare diseases.



### Literature review -> 3 major dimensions:

1. Diagnosis
2. Information & understanding
3. Care and support



Information and (proactive) administrative support



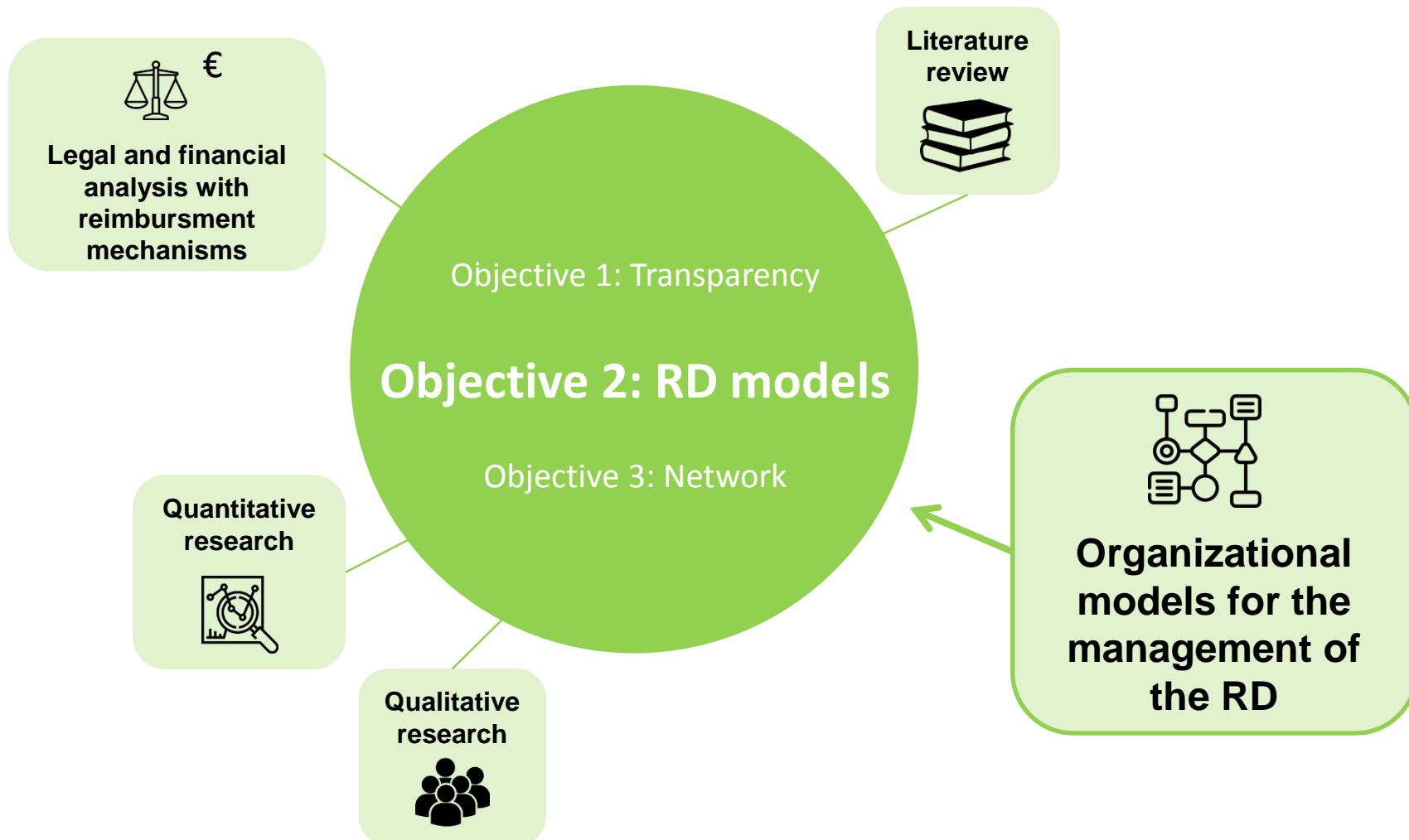
Psychological support for patients and informal caregivers



Informal caregivers

## Objective 2

### EMR models of RD patient pathways and recommendations for patients



## Rare Disease Groups treated at the participating University Hospitals <sup>1)</sup> (June 2015)

Universitätsklinikum Aachen	Academisch ziekenhuis Maastricht (azM)	Centre Hospitalier Universitaire (CHU) de Liège
Neurology	Neurology	Neurology
Hematology	Hematology	Hematology
Liver and gastrointestinal diseases		
Skeleton and teeth	Skeleton	
Nephrology		Nephrology
Inflammatory diseases (children)	Inflammatory diseases (adults)	inflammatory diseases (children and adults)
Retinopathies		
Syndromal diseases and infantile breathing regulatory diseases	Syndromal diseases	Syndromal diseases
Calciophylaxis		
Cardiomyopathies and keratinopathies (research only)	Cardiomyopathies and keratinopathies	
	Cancer	Cancer
	Genodermatoses	
Metabolic disease is partly covered in the other topics.	Metabolic disease, especially galactosemia	Metabolic diseases
		Endocrine diseases (adults and children) <sup>2)</sup>

## Focus on 8 specific rare diseases

Rare disease groups	Specific rare diseases
Neurological diseases	Huntington disease
	Duchenne muscular dystrophy
Hematological diseases	Chronic myeloid leukaemia (CML)
	Polycythemia vera (PV)
Syndromal diseases	Silver-Russell syndrome
	Rett syndrome
Metabolic diseases	Galactosemia type 1
	Phenylketonuria (PKU)

<sup>1)</sup> 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.

<sup>2)</sup> This field is partly covered through the other University hospitals too.

## Objective 2



**Organizational models for the management of the RD ->**  
**What do we observe in the EMR pathways of RD patients ?**

**Observations based on the preliminary results  
of the legal and financial analysis and on the field study:**



Cross – border mobility



Place of treatment (centres)



Care coordination



Costs for patients



## Care pathway is complex and hard to manage

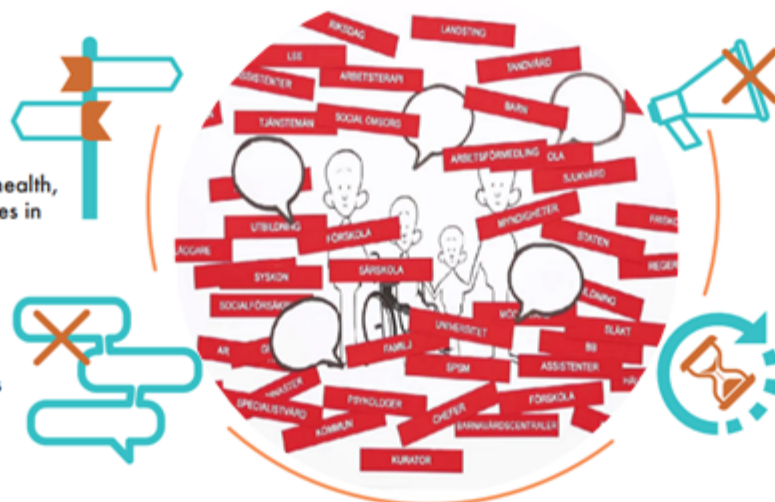
Integrated care

# 65%

have to visit different health, social and local services in a short period of time

**67%**

feel that these services communicate badly between each other



7in10

do not feel well informed  
about their rights

**7in10**

find that organising care is time-consuming; 6 in 10 find it hard to manage

Image on care pathway used for illustration purposes.  
Source: *Nationella Funktioner Sällsynta Diagnoser*, Sweden

## EMRaDi recommendations 2 to 3

**2. Design patient care pathways** that incorporate expert advice regardless of the country of the expert, while ensuring that as much of the resource-intensive care takes place in the country of residence of the patient and that the individual circumstances of patients are taken into account



**3. Establish a preferential procedure** for granting access to cross-border care and to reimbursement with no advance payment for RD patients



## Objective 3

**Network** of health professionals and raising (public) awareness

**Networking, training  
and exchange of  
expertise among  
health professionals**



Objective 1: Transparency

Objective 2: RD models

**Objective 3: Network**

**Patient involvement  
and public  
awareness**



## Objective 3



**Networking, training and exchange of expertise among health professionals**



Practices of diagnosis and care -> EMR case conferences



Better exchange of material -> diagnosis and NGS technologies

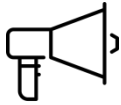


Exchange programs and training for health professionals



Information sessions and communication tools for health professionals

## Objective 3



### Patient involvement and public awareness

With the Patient Sounding Board



Comparing national plans



Quality documents in relation to cross-border healthcare



Raising awareness



Signed declaration to improve the access to care for RD patients in the EMR

Observations based on the legal and financial analysis and on the field study:

- Fellow patient (and for informal care givers) support
- Public awareness

## Objective 3

### Network of health professionals and raising (public) awareness

#### EMRaDi recommendations 2 to 3

4. Support **networking** between rare disease physicians as well as RD experts in psychological and social supportive services in their cross-border exchanges



5. Establish clear rules for reimbursing **telemedicine** for RDs and support incorporating the work of **European Reference Networks** into national healthcare systems



6. Improve the **information and awareness** for rare diseases and the **cross-border healthcare (CBHC) possibilities** in cooperation with the existing channels of information in the EMR



# Final deliverables and events on **micro level**

## Practical guide(s) for RD patients and Healthcare providers

In connection to those events in 2019:

- 23 May 2019: 3<sup>rd</sup> meeting with the Patient Sounding Board
- April/May + Oct/Nov. 2019: 5<sup>th</sup> and 6<sup>th</sup> meetings of the Task Force of the 3 University Hospitals
- 4<sup>th</sup> quarter 2019: 3 info sessions for Health professionals (GPs)

# Final deliverables and events on macro levels

## *For political recommendations:*

- 1 report describing the **generic organizational model** for the management of rare diseases **in border regions** (EMR)
- **6 specific EMR organizational models** for the management of selected rare diseases
- 1 report with **recommendations** for national and European developments

In connection to those 3 events:

- 1 meeting for the preparation of a signed intention with health insurance regional or national authorities (S2 2019 – Aachen)
- 1 political meeting (end of 2019 – begin of 2020 - Brussels)
- 1 closure event with signature and dissemination of results (February 2020 – Liège)



# Added value of the EMRaDi project in the field of rare diseases

During the project

- **Awareness** raising
- Inventories of pathways and needs by **listening to patients, relatives and professionals**
- Build and improve the **networks** of clinicians and general practitioners
- Reach a high number of **citizens in the EMR** and increase their quality of life

Long term and beyond the project lifetime

- Be in the frontline of **European development**
- Open the possibility to new source of funding in **research and innovation**
- Serve as a **model for other diseases**
- Reduce the **health expenses**
- **Good basis for future other cooperation projects in the EMR and at EU level**

## How can you help us?

1. What is the current situation (the bottlenecks, successes...)?
2. What are the solutions?
3. Who can contribute to come to a solution?



## Thanks to our (co-)financers



provincie limburg



Avec  
le soutien de la



Wallonie

Die Landesregierung  
Nordrhein-Westfalen

