



Addressing the challenge of rare disease networks in France

Nothing about us without us !

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EURORDIS Membership Meeting 2015 Madrid
*Hotel Rafael Atocha
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Key data

key dates & figures

2005 - 2008

1st strategic plan for rare diseases
in France

2011 - 2014

2011 - 2016

2nd strategic plan for rare diseases
in France

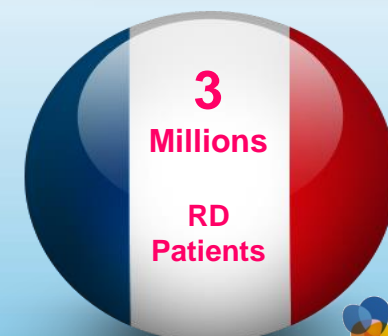
Sept. 2014

- Nov. 2015

Evaluation of the NPRD 2

After 2016

To the **3rd strategic plan** for rare
diseases in France



CEs & Rare Disease Networks

2 complementary dynamics

**CEs in France, a major achievement of
the French rare disease Plans**

**Rare disease networks in France, or,
of the necessity to go further**

CEs dynamic

RDN dynamic



CENTRES OF EXPERTISE IN FRANCE



***A MAJOR ACHIEVEMENT
OF THE FRENCH
RARE DISEASE PLANS***

Centres of expertise

Missions

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Expertise

On a disease or a group of diseases

Referral

Interregional, national or international attraction

Coordination

Policy definition, activity organisation, action plan development

Research

Promotion, facilitation and participation in fundamental, clinical, and translational research

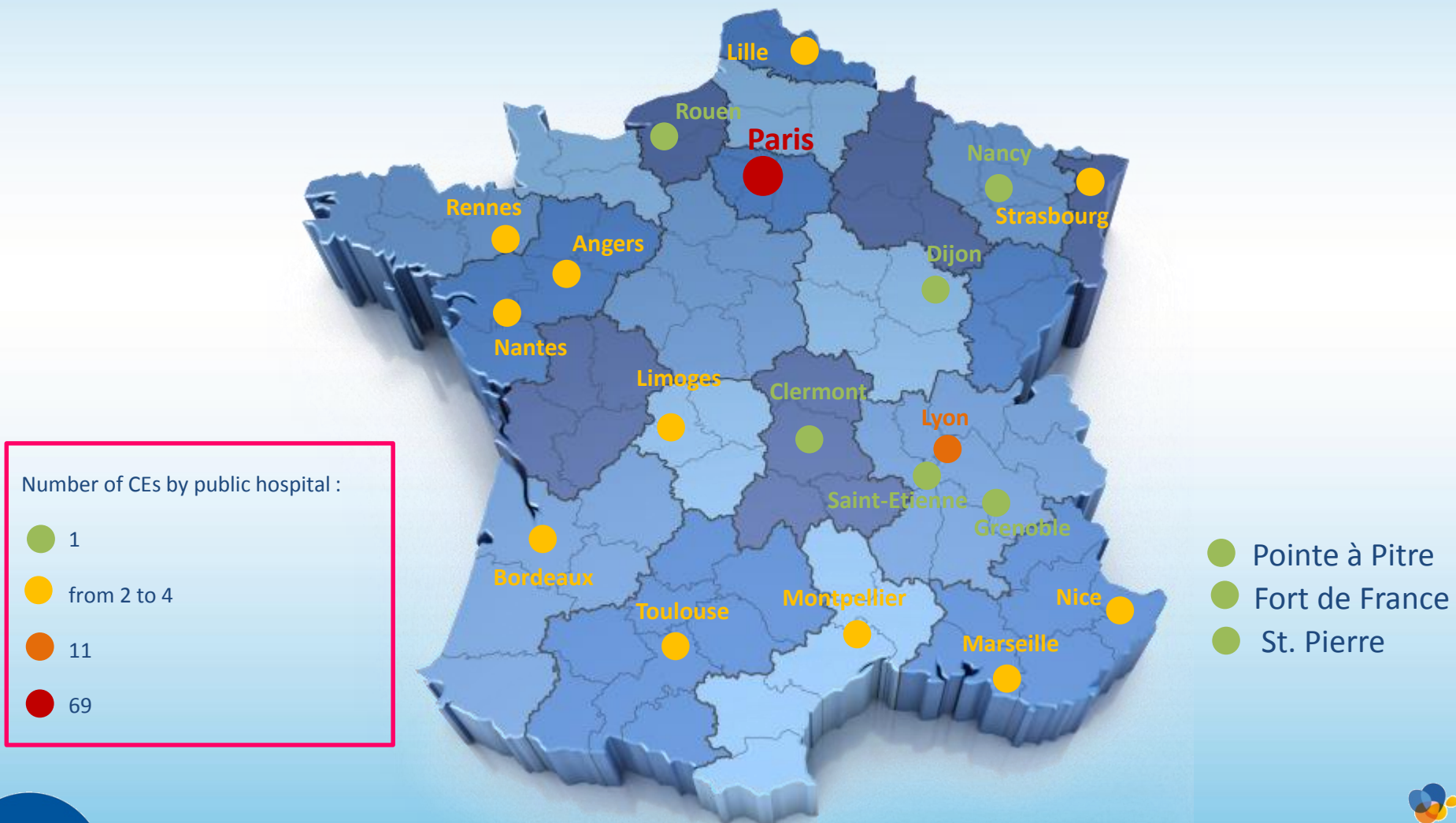
Information and training of health

Promotion, facilitation, and lead of academic university teaching

1st RD Plan

Centres of expertise

Map



Source : Ministère en charge du Travail, de l'Emploi et de la Santé/DGOS PF2 & USID 10/2011

Centres of expertise

Centres of competence



**Patient
Organisation
advocacy**

Patient organisations argued for an organisation of care **at local level** that had not been foreseen in the drafting of the first Plan (2005-2008).

**502 Centres of
competence
missions**

Ensure **patient care at local level**

Establish the diagnosis of RD

Implement therapeutic (*when available*)

Ensure connection with the CE and all medical/social actors.

1st RD Plan

Centres of expertise

Key challenges

- The imperative need to **improve CEs** after 10 years
- Implement the **2nd Plan dedicated measures** on CEs:
 - ✓ A simplification of CEs assessment,
 - ✓ Implementation of business reports
 - ✓ Succession process of the CE coordinators

To a Ministerial
Direction on CEs

*Publication expected in
January 2016*



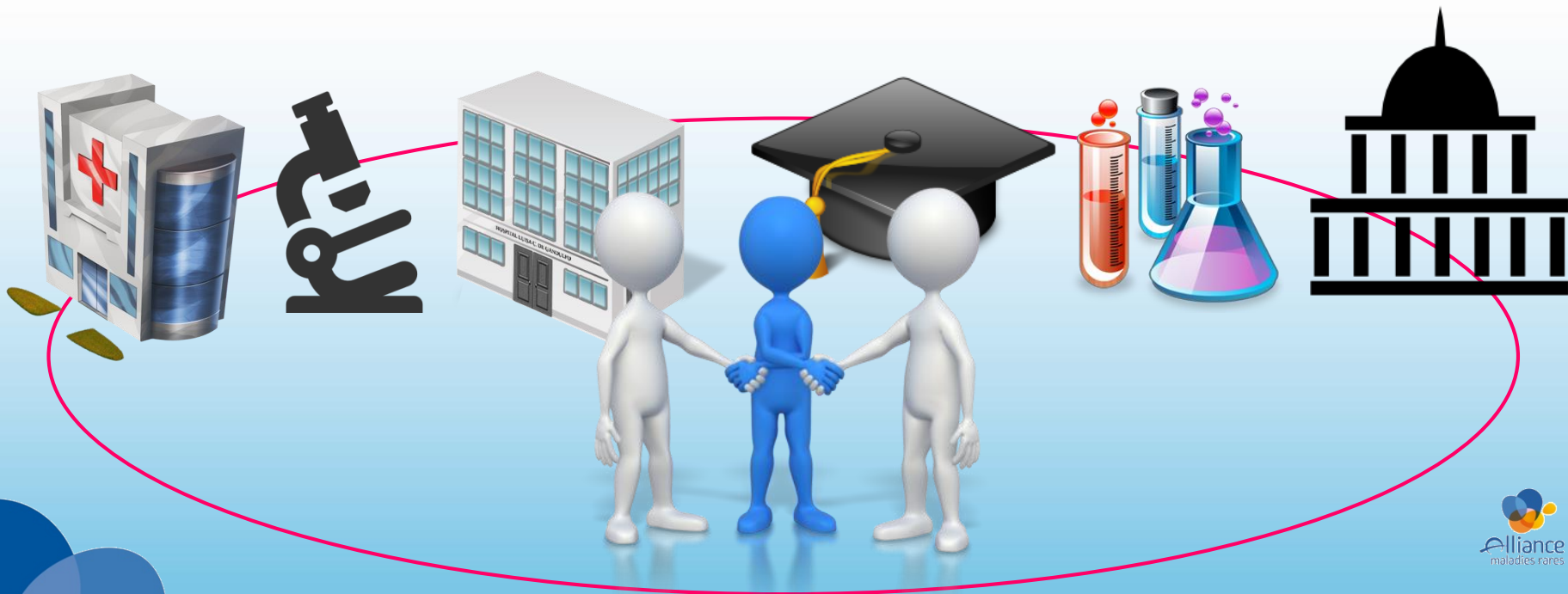
New certification
of CEs & CC

*Launching expected in
2016/2017*



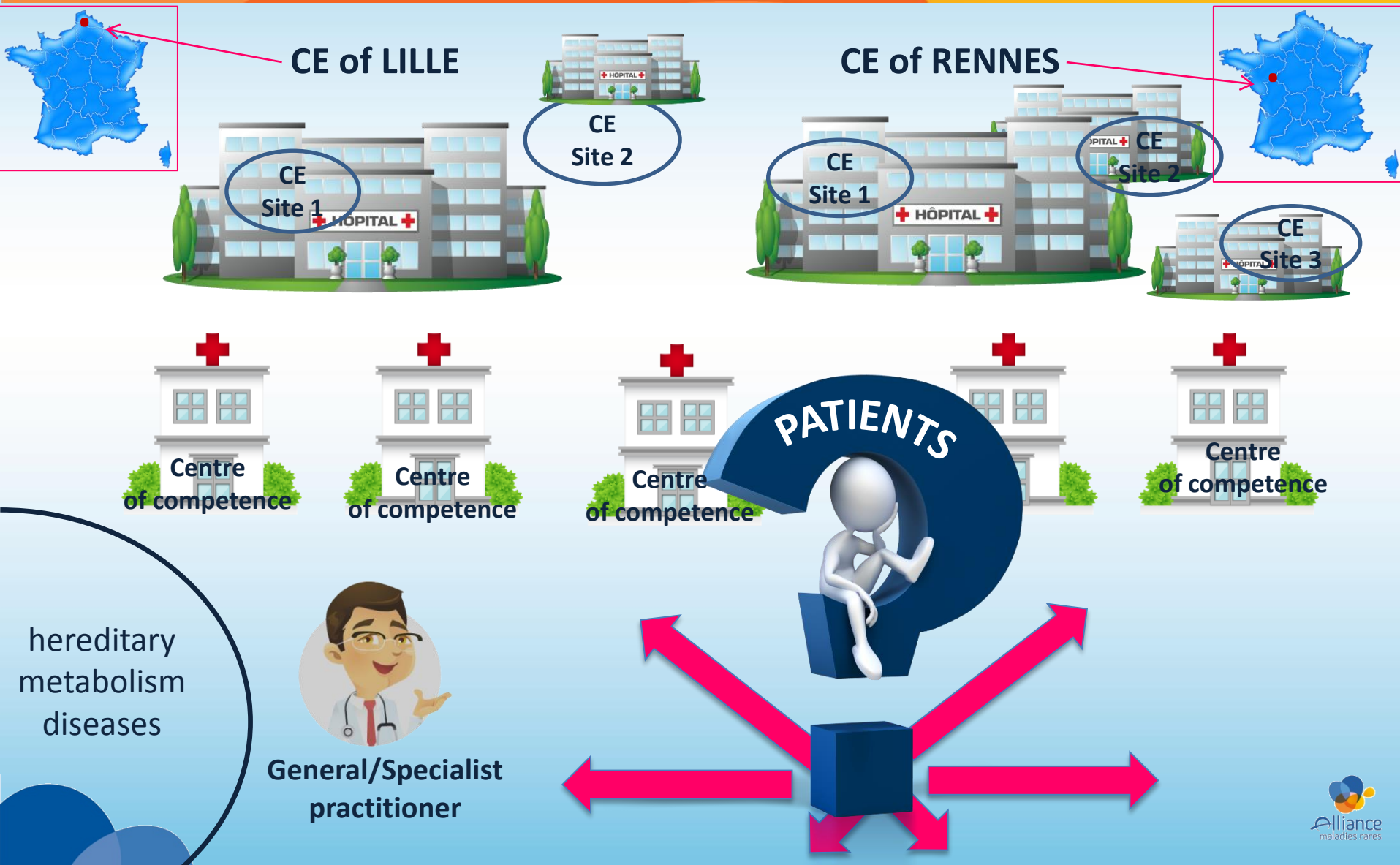
RARE DISEASE NETWORKS IN FRANCE

OF THE NECESSITY TO GO FURTHER!



Why building RDN in France?

1st Rare Disease Plan : A complex landscape



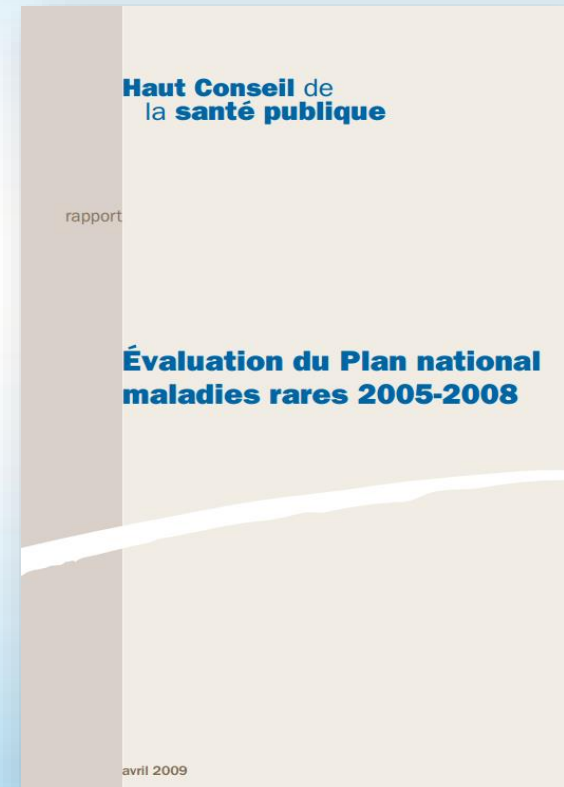
Why building RDN in France?

Evaluation of the 1st Rare Disease Plan

"Grouping of centres must be favored (developmental anomalies, metabolic diseases), as the federations of reference centres of several regions taking care of the same disorders. Some centres could be the object of extension of labelling in diseases other than those of the initial labelling".

" Regional federations and national federation of the reference centres should be created to organize meetings of confrontation at least annual. The meetings of the reference centres during the conference of evaluation highlighted this necessity of dialogue and pooling of the scientific, ethical and administrative problems".

"Competence centers and National Platforms of reference laboratories will be a part of healthcare pathways. The very rare diseases must be integrated into one of these pathways which, if of need, will help them to find an expert somewhere else in Europe"



Why building RDN in France?

RD ERN

- Recommendation of the EUCERD to the European commission & the member states **Article 12 of the Cross-Border Healthcare Directive (Directive 2011/24/EU)**.

“...RD ERNs will provide the framework for healthcare pathways for RD patients through a high level of integrated expertise...”

“...Nationally designated centres of expertise (CE) are the core participants in RD ERNs...”

“...RD ERN needs to be flexible enough to accommodate working with different national CE structures. Depending on the national healthcare system, CEs can be very different structures organised by regions, treatments, or diagnostic procedures, offering services in one location or through an established network.”



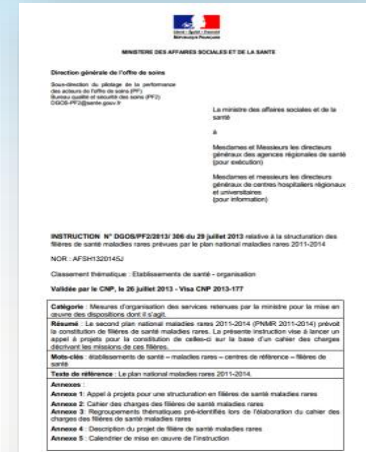
- **Rare disease Networks in France will facilitate the implementation of RD ERN**

Rare diseases networks

Scope definition

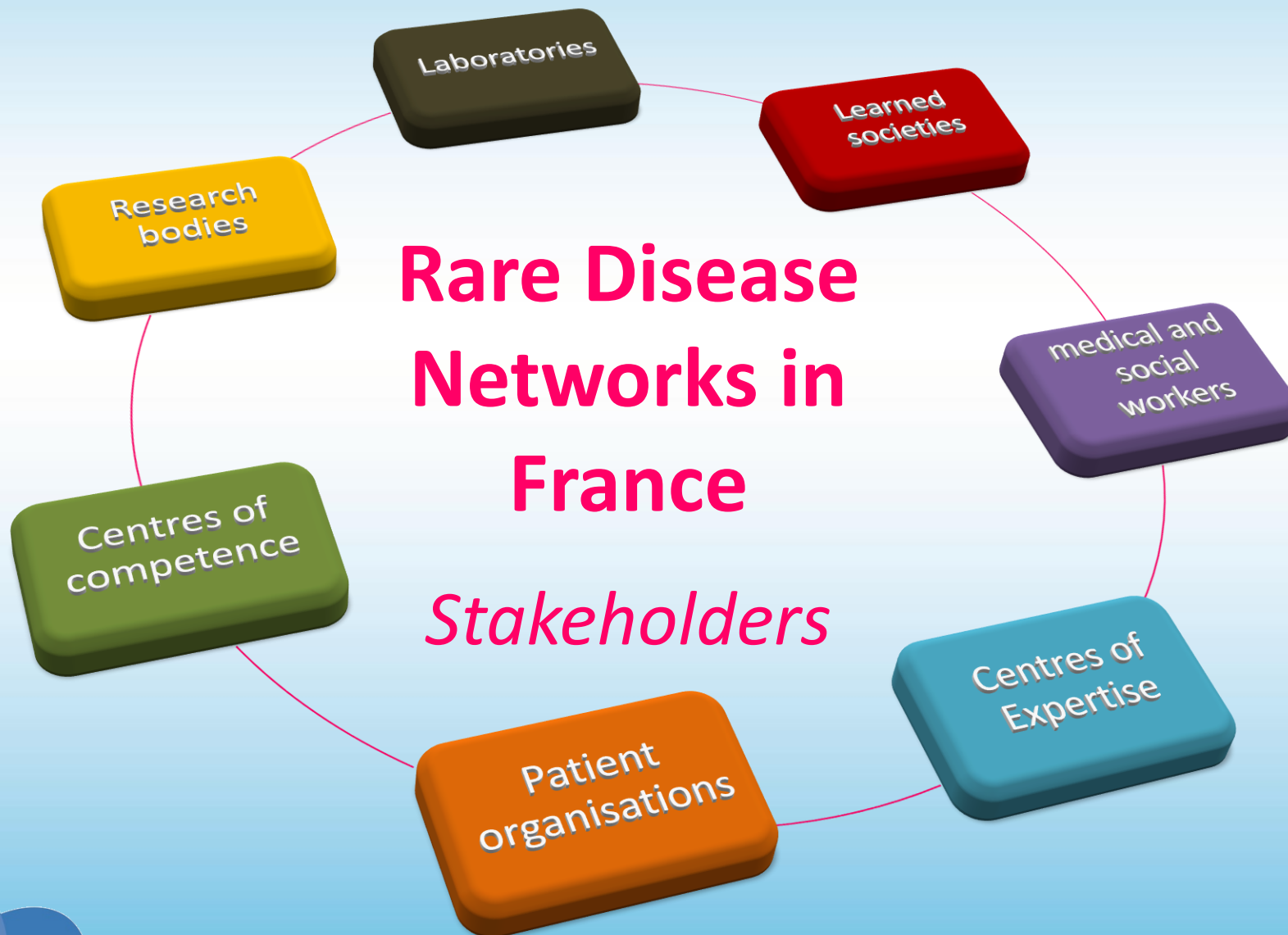
- **Ministerial Direction/July 2013** relative to the structuring of the rare disease networks planned by the 2nd National Rare Disease Plan 2011-2014.

*“The health network should cover **a wide and coherent range of rare diseases**, whether they are closely related by their symptoms, consequences or coverage, or affect a single organ or system. The network deals above all with known rare diseases, and also with not yet confirmed diseases or syndromes”.*



Rare diseases networks

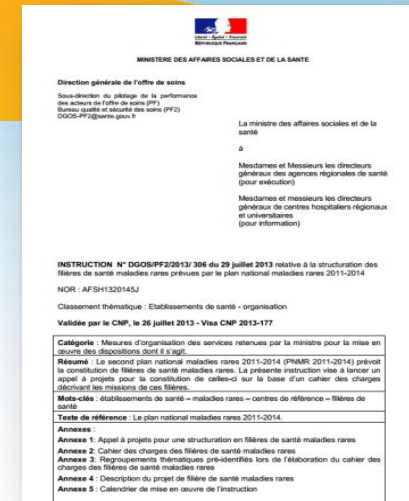
Actors



Rare diseases networks

Governance

- Ministerial Direction/July 2013 launches a call in direction of CE's & all stakeholders.



Key points of specifications

- Governance/organization to be defined by each RDN
- Key role of PO, associated with the RDN governance/working group
- All CE intended to be part of only one RDN
- Financial support for coordination evaluated in proportion of the RDN cohort
- The RDN CEO is one of the CE CEO (Receives funds, and leads for 4 years the RDN)
- 23 RDN are now definitely identified in France.
- All RDNs CEO must deliver their Action plan before June 15th 2015

Rare disease networks

List & map

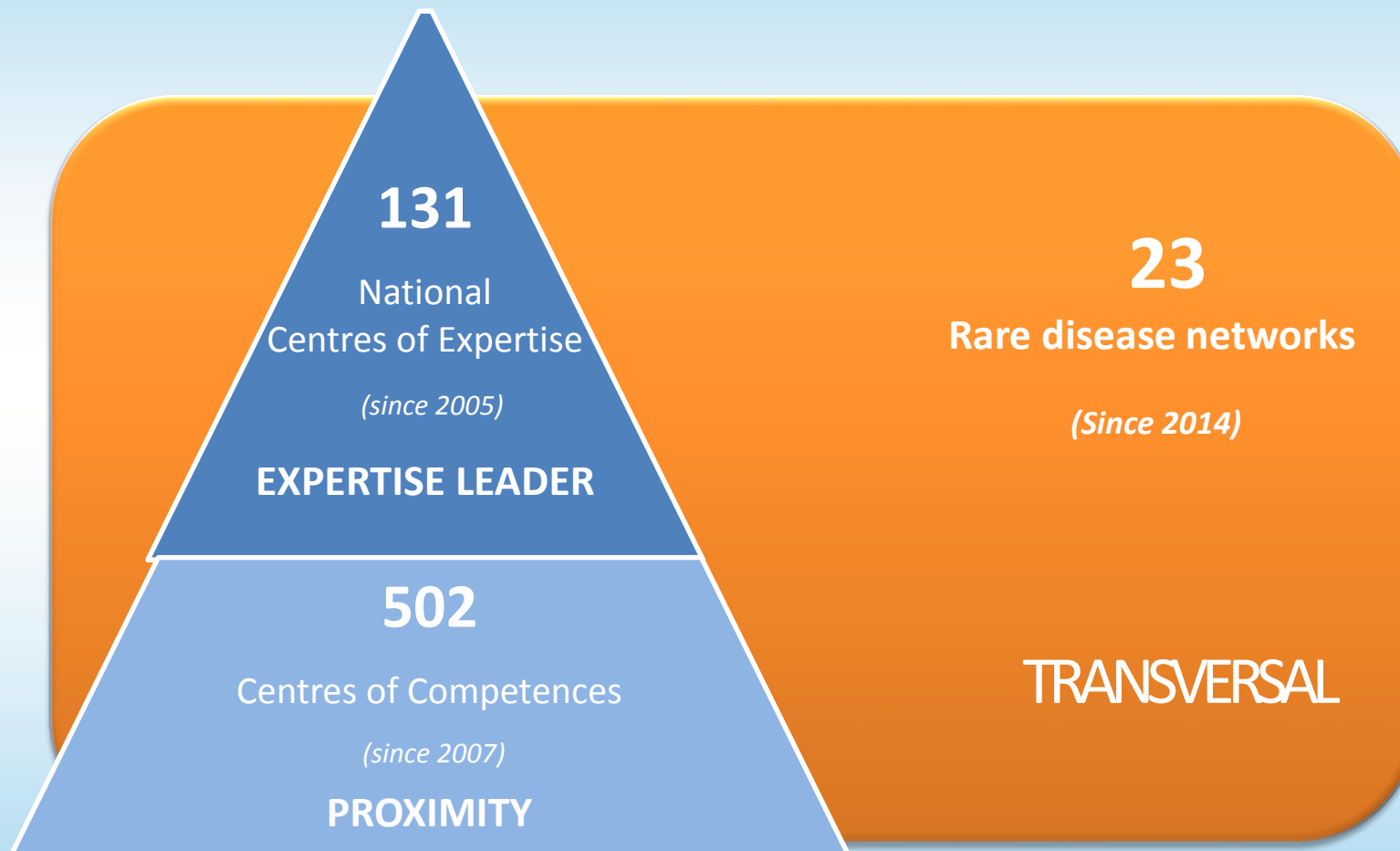
AnDDI-Rare	developmental anomalies and malformations
CARDIOGEN	transmitted heart diseases
BRAIN-TEAM	CNS rare motor disorders
DEFI SCIENCE	brain development diseases and intellectual disabilities
FAI2R	rare auto-immunes and auto-inflammatory diseases
Fava-multi	rare vascular disease & multisystemic lesions
FILFOIE	rare liver diseases
FILNEMUS	neuromuscular diseases
FIMARAD	rare dermatological diseases
FIMATHO	oesophageal & intestinal RD + diaphragmatic hernia
FIRENDO	rare endocrine diseases
G2M	rare hereditary diseases of metabolic origin
MARIH	immuno-hematologic rare diseases
MCGRE	rare diseases of red cells and of erythropoïesis
MHémo	constitutional haemorrhagic rare diseases
MUCO/CFTR	cystic fibrosis and CFTR anomalies
Neurosphinx	rare pelvic and medullary diseases
OSCAR	Os-Calcium/Cartilage-Rein
ORKID	rare kidney diseases
RESPIFIL	rare respiratory diseases
SENSGENE	rare sensory diseases
SLA	amyotrophic lateral sclerosis
TETECOUC	Head and Neck rare diseases

CARDIOGEN
FAVA-Multi
FILFOIE
FIMARAD
FIRENDO
MARIH
MCGRE
NeuroSphinx-GBS
OSCAR
RESPIFIL
TETECOUC



Rare diseases networks

Scheme



**RD
ERN**

1st RD Plan

2nd RD Plan

Rare diseases networks

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Logic of the horizontal/transversal organisation

Identify and Mutualize
complementarities/
resources/means

Improve the
coordination/animation/
lead of actions



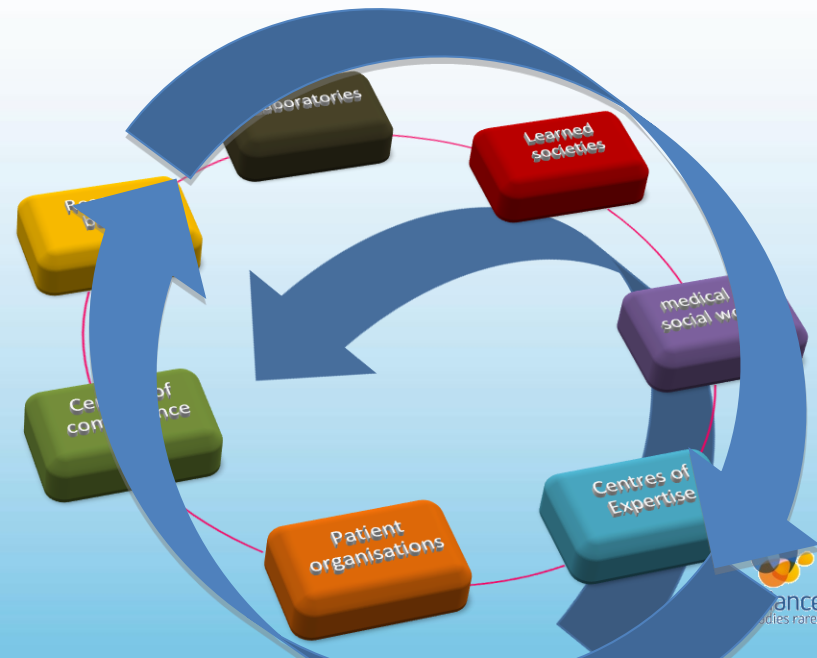
Rare diseases networks

2 Cardinal points

Facilitate for all RD patients
and healthcare professionals,
the possibility of finding a way
into the healthcare system

**Improve the continuum
between**

Diagnosis/Care/Research/
Therapeutic development
/Medical and social
management/Articulation
with the territories of health



Rare diseases networks

Key points of specifications

Improve the quality of
the care of RD
patients

Rare disease network

List and coordinate the existing recommendations of best practice among which the PNDS

Spread knowledge and information to the various public, including patients

Draw up the list of the pathologies, the projects and the ongoing works to make them know

Impulse the elaboration of common or transverse recommendations and, when the pathologies justify it, specific recommendations with the aim of the insertion of the patient in his social, educational and professional life

Rare diseases networks

Key points of specifications

Develop the clinical
and translational
research

Rare disease network

Inventory the programs centered on diagnosis and therapeutic innovation in rare diseases and define annual priorities (essays, cohort)

List and identify the network of the units of research with which the CEs and centres of competence work

Review the projects and the work in progress at least once a year

Bring to the foreground common axis of research with a European or international dimension

Rare diseases networks

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Key points of specifications

Develop the
education and the
formation

Rare disease network

List and arouse programs of
teachings and creation of inter-
university diplomas

List the actors of the network once
a year

Organize one day of exchanges
with the various professional
categories of the CEs and
competence centres

Rare diseases networks

Key points of specifications

Make patient
organisations
key player

Rare disease network

Develop TPE

Establish training
teleconferences/Realize video
conferences over the Internet for
patients

Develop e-Health, i.e.
multidisciplinary consultations by
telemedicine

Develop PO involvement in the
development and recruitment for
clinical and therapeutic trials

Mutualize Patient reception in
hospitals

Develop PO involvement on
communication media

Key challenges

Strength/weakness

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?



Giving a true
partner/role for
Patient Organisations

Developing common tools
and a common language

Making the best use of
new health technologies

Strengthen partnerships
and existing work &
Valorising RD actions
from the hospital

RD ERN Link

inter-RDN

Funding

strength

weakness



Rare disease networks

To go further

www.alliance-maladies-rares.org



Rare disease networks

Nothing about us without us !

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**We must go on,
coordinate all initiatives across Europe
to improve our CEs, RDN
and the future RD ERN !**



Thank you for your attention!