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NATIONAL REGISTRIES OF RARE DISEASES IN EUROPE: AN OVERVIEW BY THE EPIRARE PROJECT.

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The RD Registries in EU

- ✓ **More than 600 rare disease registries** (*Orphanet and other sources*)
 - ✓ **No uniform standards** for the storing and management of data
 - ✓ **Registries set up for a small fraction of rare diseases**, multiple registries for the same disease
 - ✓ **Need to increase data comparability and sharing**
 - ✓ **Need to harmonise existing regulations** (EU, national and regional)
- 



The EU initiatives

- **COUNCIL RECOMMENDATION on an action in the field of rare diseases (2009):**
 - Recommends that Member States support at all appropriate levels, for epidemiological purposes, registries and databases
- Funding priorities include RD registries and disease networks
 - EPIRARE – EU Platform for RD registries (Building Consensus and Synergies for EU Registration of RD patients)
 - **RD-Connect**
 - PARENT
 - ERNs
 - Other disease-specific projects
- EU Platform for the Registration of RD patients (JRC, Ispra)



The European RDR Platform objectives

To Increase sustainability


To improve data comparability, by means :

- Reference set of Common Data Elements
 - Reference coding and catalogues
 - Reference data quality assurance procedures
-
- Provision of useful tools



The opportunity offered by National (institutional) registries

Improved epidemiological information on RD, by means of:

- Integration of multiple national sources
 - Mortality/Population Registry
 - E-prescription
 - Electronic Health Records
 - Increased completeness of case ascertainment
 - E.g.: designation of CoE linked to case notification and follow-up
 - E.g.: Mandatory registration of patients
 - E.g.: Registration linked to prescriptions
 - Appropriate management of data protection rules
 - E.g.: Legal basis of data collection
 - E.g.: Appropriate procedures for data protection
- 



National (institutional) RD Registries

Running:

- ✓ Italy (2001)
- ✓ France (2005; 2007)
- ✓ Spain (2008)
- ✓ Belgium (2013)
- ✓ *Nordic Countries (Denmark, Finland, Norway and Sweden): craniofacial diseases, being expanded to all very rare diseases*
- ✓ *Finland (a general disease registry, including RD)*



Under planning:

- ✓ Bulgaria
- ✓ Germany (very rare and undiagnosed diseases)
- ✓ Sweden



In Europe a disease is considered rare
when it affects no more than
5 individuals among 10,000 persons

III International
EPIRARE
Workshop

Rome
24-25 November 2014

National Registries of Rare Diseases in Europe: An Overview of the Current Situation and Experiences.

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Abstract

The European Union (EU) policy for healthcare requires the establishment of a system of European Reference Networks, union-wide information databases, and registries for rare diseases (RDs) based on shared criteria. In pursuing its goals, the 'Building Consensus and Synergies for the EU Registration of RD Patients in Europe' (EPIRARE) project convened a meeting with experts of the competent health authorities to discuss the role of national institutional RD patient registries in supporting EU patient registration and the room for international cooperation. With this aim, this paper comparatively analyses the current situation of national institutional RD registries in the EU. © 2014 S. Karger AG, Basel.

National Registries (NR) operation

Country	Legal basis
Belgium, France, Italy	The NR activity is part of the mandatory activities of the healthcare network of centres dedicated to RD .
Spain	The NR is funded as a research project, with the Involvement of institutional, scientific, patient, and industry stakeholders.

Country	Patient coding
Belgium,Italy, Spain	Univocal, but not common, patient coding system
France	De-identification

National Registries (NR) operation

Country	Scope	Aims	
Belgium	All diagnosed RD	surveillance, public health planning +...	+patient recruitment, clinical research
France			+patient recruitment, source integration (EHR)
Italy			+clinical research (not supported yet)
Spain			+patient recruitment, translational research, social policies

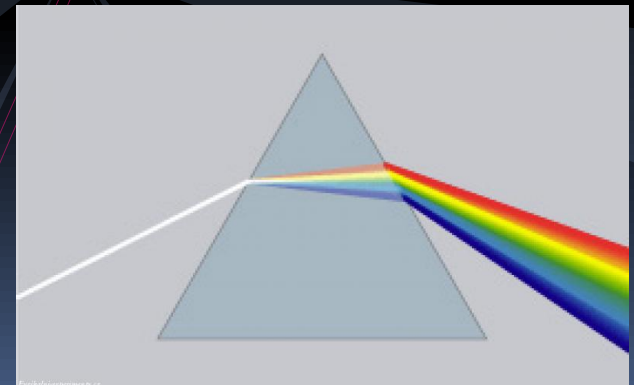
National Registries (NR) operation

Country	Common Data Elements (central repository)
Belgium	Patient ID code and demographic data, death date, registering centre and treating physicians codes; first symptom onset date, diagnosis and its features, and different options of patient consent.
France	Patient ID code and demographic data, patient consent, familiarity of the disease, death date and cause, drug treatment, services data, diagnosis and its features, fetus and newborn data, data on research participation and biological sample donations.
Italy	patient ID code and demographic data, live or dead status and death date, diagnosis, diagnosis date , diagnosis centre data, first symptom onset date, prescribed orphan drug, cost exemption code
Spain	Temporarily: patient ID code, demographic data, diagnosis.

National Registries: hurdles or opportunities?

National Registries are developing independently from each other.

- Different CDEs
- Different patient univocal coding systems
- Partially overlapping aims
- Possibly different indicators



The Italian Network for the Registration of RD patients



NATIONAL REGISTRY
FOR RARE DISEASES

Minimum Data Set

Regional Registry of RD
-
Interregional Reference Centre

Trusted Centres in a
given region

Rare Diseases in NRRD (2013)

grouped by ICD-9 Chapters

(N = 110841 records)

	records	%
Diseases of the Central Nervous System and Sense Organs	28784	26
Congenital Anomalies	21836	19,7
Endocrine, Nutritional, Metabolic Diseases, and Immunity Disorders	19279	17,4
Diseases of the Blood and Blood-Forming Organs	18452	16,6
Diseases of the Musculoskeletal System and Connective Tissue	6124	5,5
Neoplasms	5596	5
Diseases of the Circulatory System	4738	4,3
Diseases of the Skin and Subcutaneous Tissue	3666	3,3
Diseases of the Digestive System	1484	1,3
Diseases of the Genitourinary System	627	0,6
Infectious and Parasitic Diseases	144	0,1
Certain Conditions Originating in the Perinatal Period	108	0,1
Symptoms, Signs, and Ill-Defined Conditions	3	0



Thank you for your attention!

Domenica Taruscio

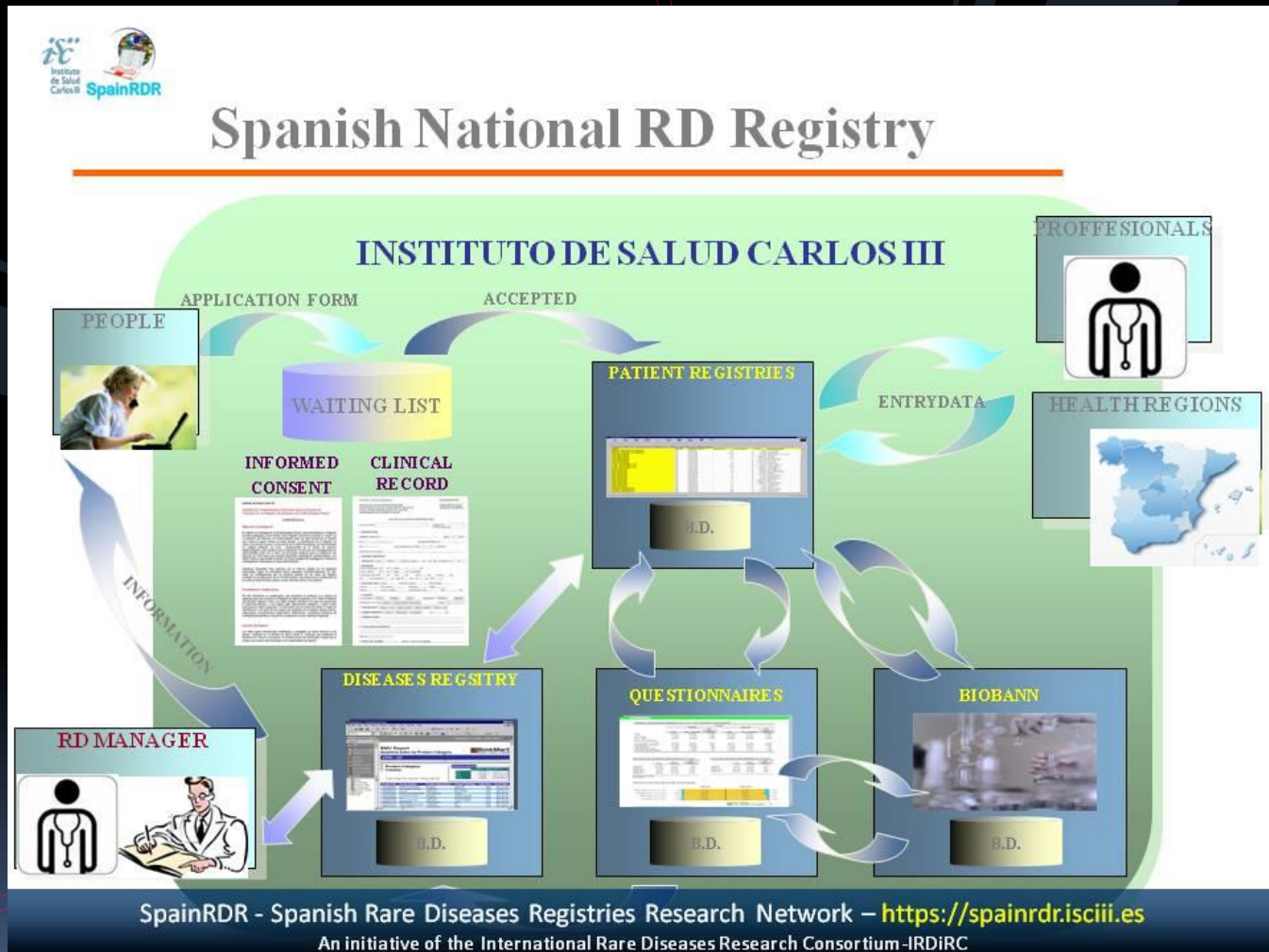
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The Spanish RDR



From Manuel Posada, presented at the EPIRARE Workshop on National Rare Diseases Registries. Rome, 5 March 2014

The Spanish RDR



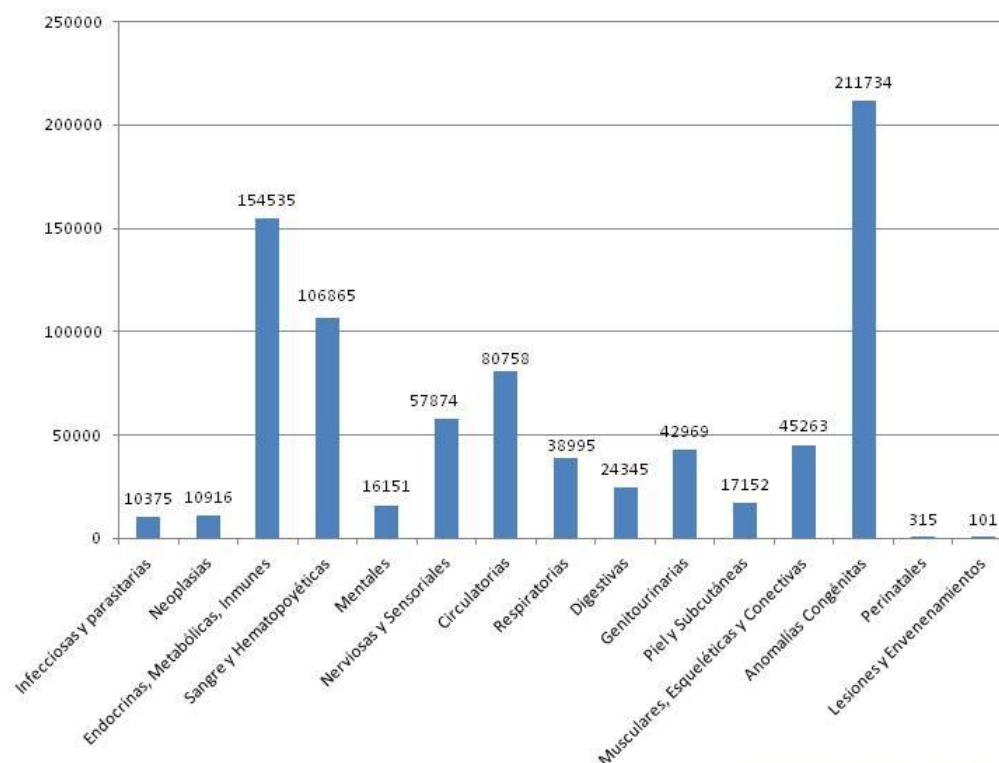
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The Spanish RDR



PILOT STUDY

CASES BY ICD GROUPS (n= 818,348)



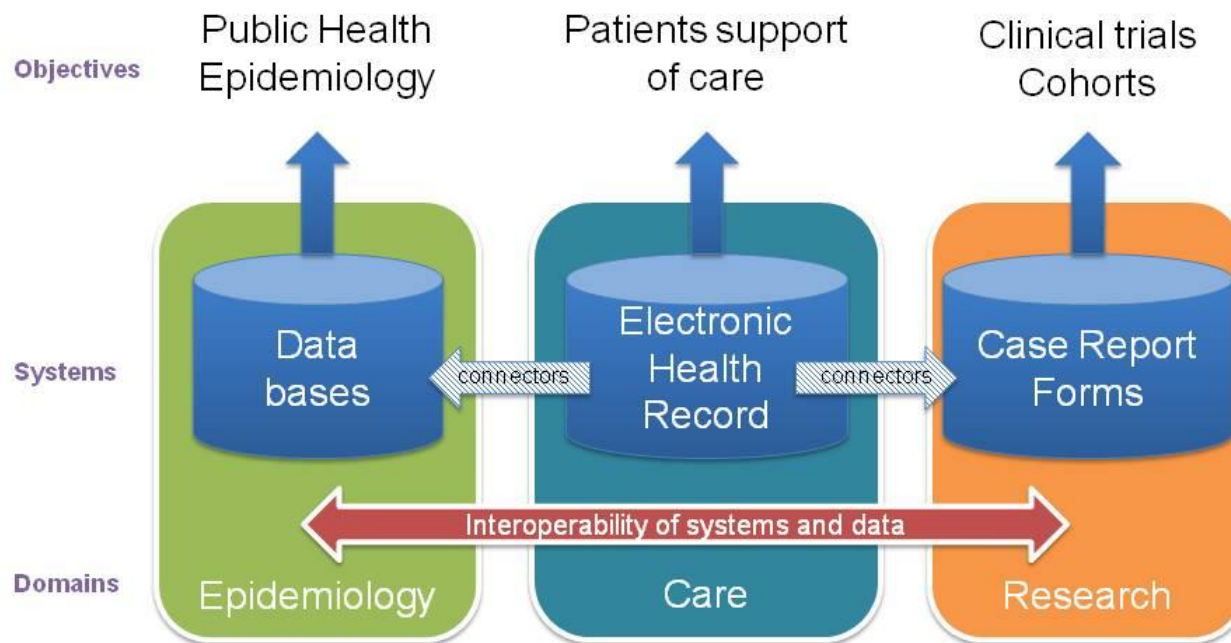
SpainRDR - Spanish Rare Diseases Registries Research Network – <https://spainrdr.isciii.es>

An initiative of the International Rare Diseases Research Consortium-IRDRC

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The French network

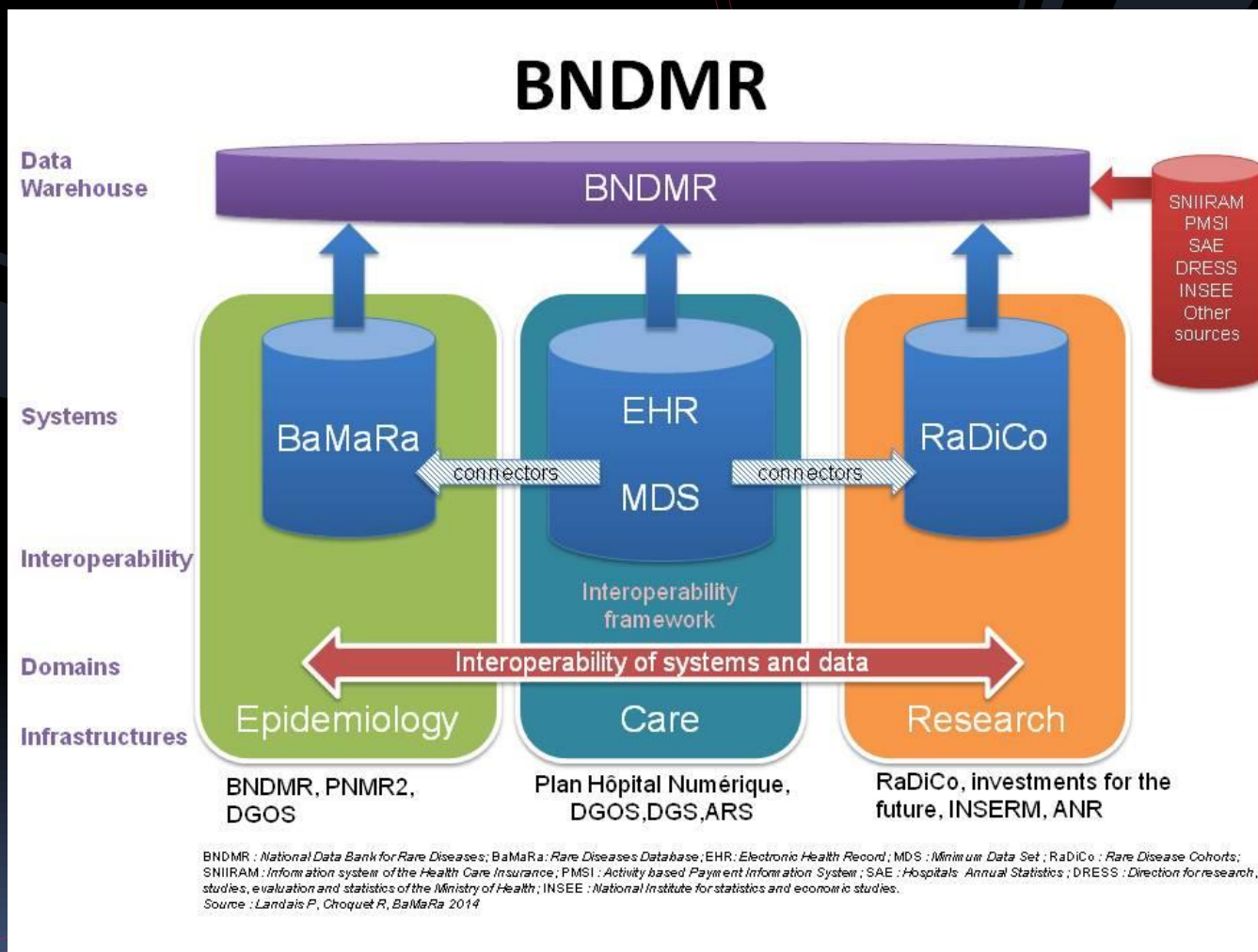
Domains, systems, objectives and interoperability



Source: BaMaRa 2013

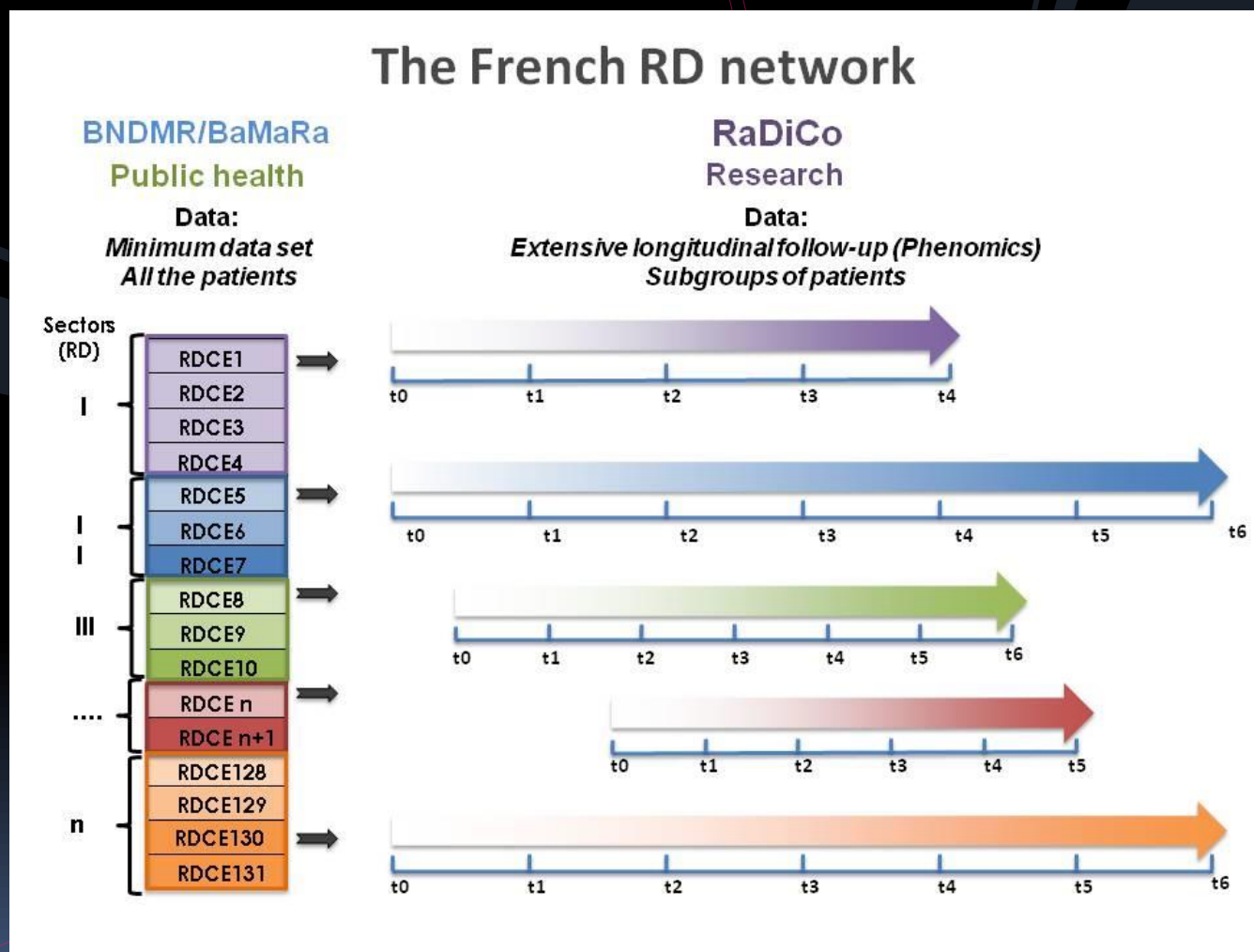
From Paul Landais, presented at the EPIRARE Workshop on National Rare Diseases Registries. Rome, 5 March 2014

The French network



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The Ra.Di.Co. section



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The Ba.Ma.Ra. section

Inventory

Some figures...

