

# The Italian National Registry for Rare Diseases



*National Center for Rare Diseases  
Istituto Superiore di Sanità, Rome*

# **Legal basis**

## **(Ministerial decree 279/2001)**

Regulation for the institution of the **National Network for Rare Diseases** for the prevention, surveillance, diagnosis and therapy of rare diseases and the exemption from patients' participation in the costs of the relevant healthcare”

# Article 3 of DM – Aims of the Registry

- monitor RD and inform the national and regional planning of measures for the protection of RD patients and to implement an epidemiological surveillance system.
- the faculty to collect demographic, anamnesis, clinical, laboratory, determinant data of use for medical, biomedical and epidemiological research and to liaise, where existing, with international registries.

# The national registry is a network of Regional registries

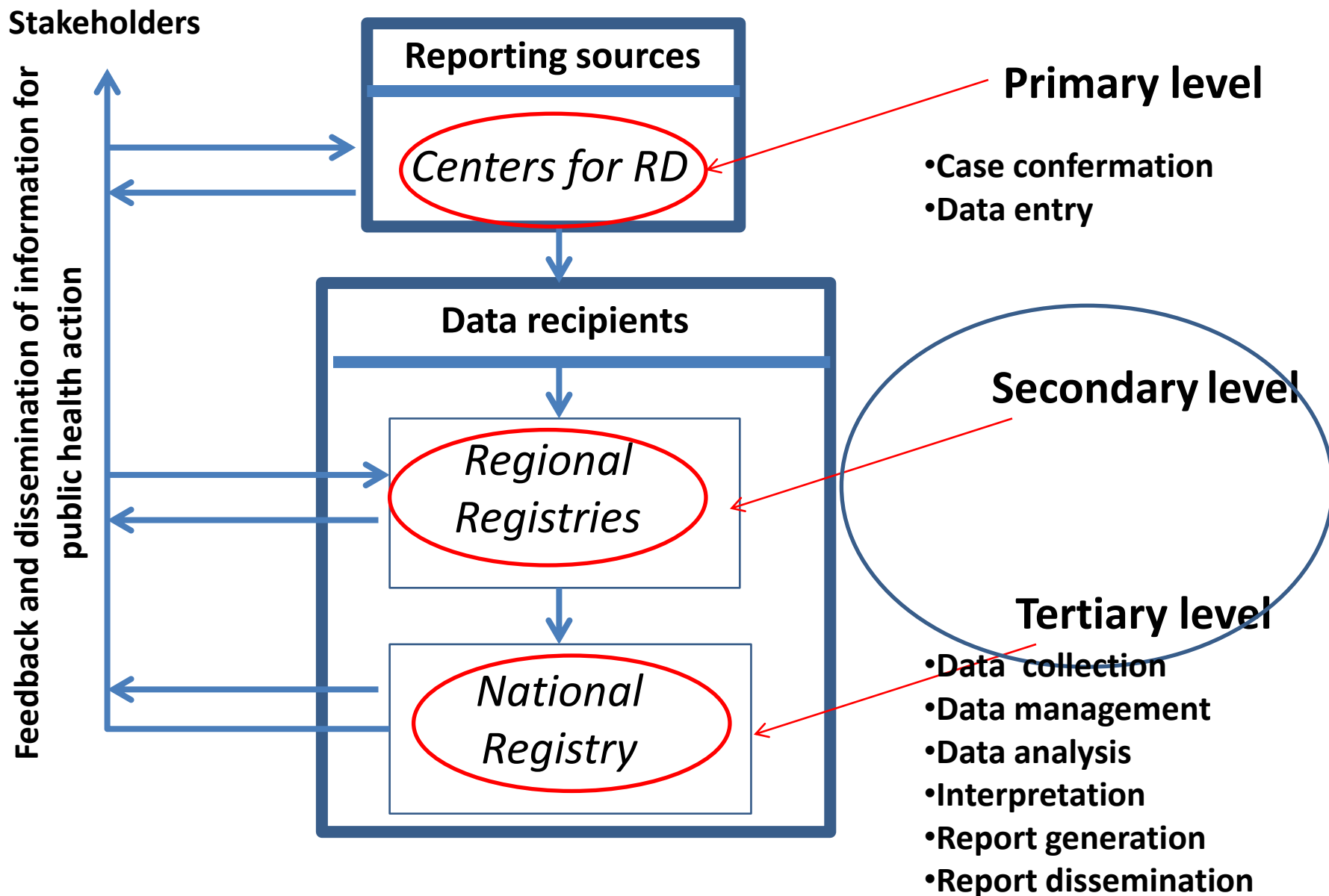
- The regional registries may differ in aims and internal organization: some have mainly epidemiological and public health purposes, in support of regional planning, while others also aim at evaluating the quality of care

# Mandatory Data set

Agreement between the Central Government and the Regions in the year 2007

- ID (encrypted code based on given name, family name, birth date and place, sex)
- sex
- Date of birth
- Live – death
- Date of death
- Diagnosis of RD
- Centre of RD diagnosis
- Date of disease onset
- date of diagnosis
- orphan drug used

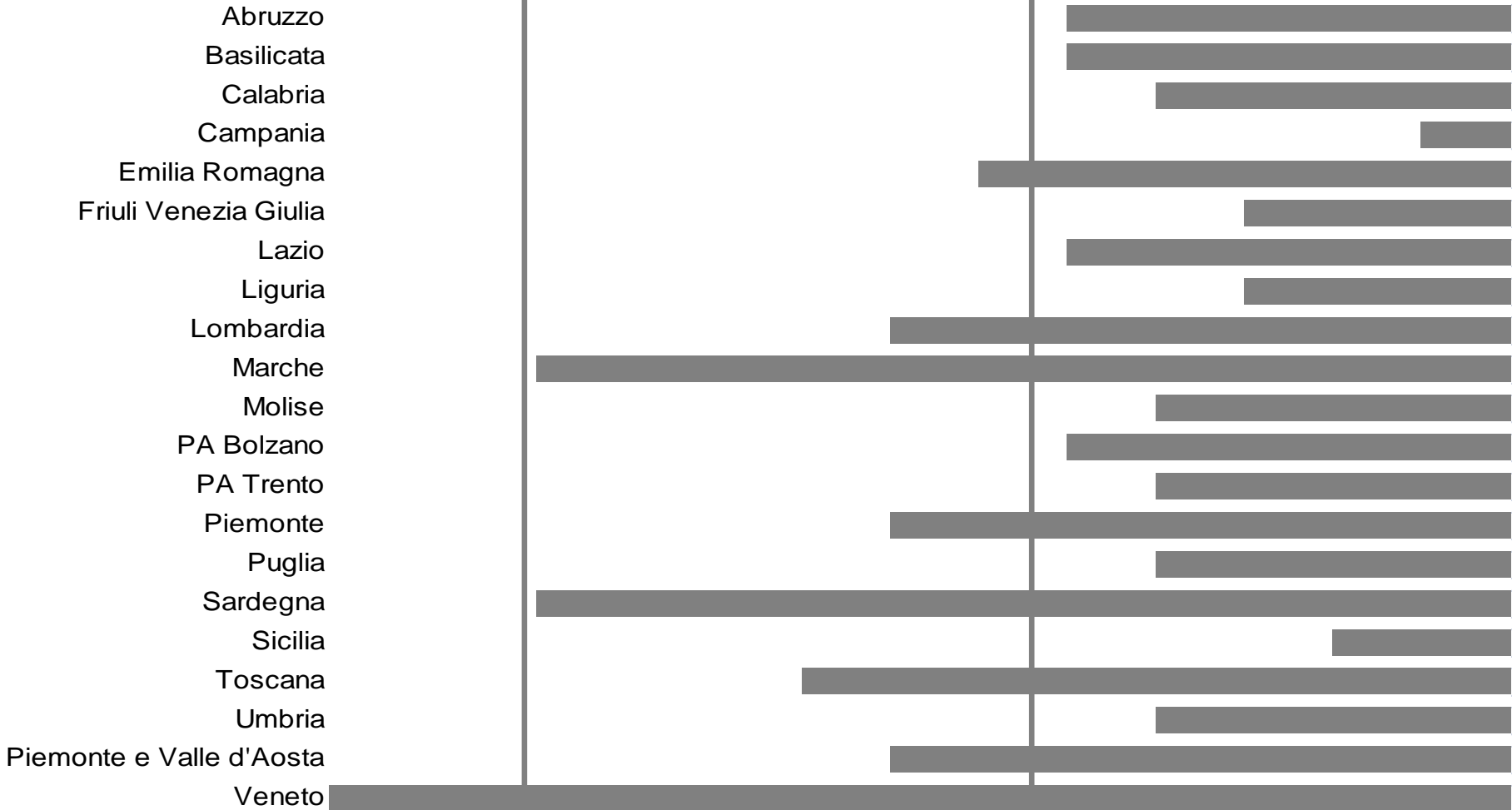
# Flow chart of NRRRD



Decree 2001

Agreement 2007

2000 2001 2002 2003 2004 2005 2006 2007 2008 2009 2010 2011 2012



# Notified records

	Number of 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
Total	110841	100



Blood Transfus. 2014 Apr;12 Suppl 3:s606-13. doi: 10.2450/2014.0064-14s.

## **The Italian National Rare Diseases Registry.**

Taruscio D<sup>1</sup>, Kodra Y<sup>1</sup>, Ferrari G<sup>1</sup>, Vittozzi L<sup>1</sup>; National Rare Diseases Registry Collaborating Group.

Blood Transfus. 2014 Apr;12 Suppl 3:s576-81. doi: 10.2450/2014.0017-14s.

## **Current status of Italian Registries on inherited bleeding disorders.**

Hassan HJ<sup>1</sup>, Morfini M<sup>2</sup>, Taruscio D<sup>3</sup>, Abbonizio F<sup>1</sup>, Giampaolo A<sup>1</sup>, Kodra Y<sup>3</sup>, Oliovecchio E<sup>2</sup>, Vittozzi L<sup>3</sup>.

	<b>National Registry of Rare Diseases (NRRD)</b>	<b>National Registry of Congenital Coagulopathies (NRCC)</b>
<b>Institution Organization</b>	By law Governmental National Institute	Voluntary Governmental National Institute
<b>Purpose</b>	Rare diseases surveillance	Inherited bleeding disorders surveillance and drug consumption monitoring
<b>DATA COLLECTED</b>		
Severe Haemophilia A	No	Yes
Moderate Haemophilia A	No	Yes
Mild Haemophilia A	No	Yes
<b>Total Haemophilia A</b>	Yes	Yes
Severe Haemophilia B	No	Yes
Moderate Haemophilia B	No	Yes
Mild Haemophilia B	No	Yes
<b>Total Haemophilia B</b>	Yes	Yes
<b>NUMBER OF CASES RECORDED</b>	<b>11.135</b>	<b>9.097</b>

# Participation of the NRRD in research activities

The NRRD is a gateway to multiple registries



- Paroxysmal Nocturnal Hemoglobinuria registry
- Lesch-Nyhan Disease registry
- Gaucher registry

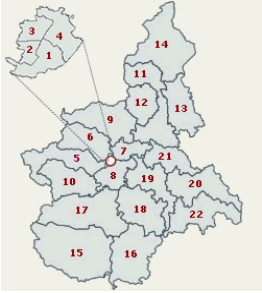
# Identity card

- Defined by law in 2001 with main aim: monitor RD at national level and inform the national and regional planning of measures for the protection of RD patients
- Multidiseases population based registry
- Data source: expertise clinical center dedicated to RD

## Ministerial Decree 2001 no. 279

This MD was principally devoted to improve health care of RD patients through the establishment of The *Network* of **clinical “Centres for RDs”** for the prevention, **surveillance**, diagnosis and treatment of RDs-

# Italian healthcare system



three levels: central, regional and local

central

Ministry of Health

regional (from 1992)

20 Regions

Local

more than 200 local health unit