

Oregi Lizarralde, Luis M.\*; Echevarria González-de-Garibay, Luis J.  
Health Department; Basque Country Government  
Vitoria-Gasteiz; Spain [rer-cae@euskadi.eus](mailto:rer-cae@euskadi.eus)

## Introduction

### RER-CAE (RD Registry of Basque Country):

- Established in 2014, by the Health Ministry of Basque Country Government
- Included in Basque Country strategy of rare diseases (RD)
- Based on direct notification of diagnosed cases, by clinicians; actually specialist doctors of public health service (Osakidetza)
- Structured considering the EPIRARE proposals (Taruscio et al., 2014)



### Basque Country:

**Population:** 2.175.819 inhabitants (1/1/2017)  
**Surface:** 7.234 km<sup>2</sup>  
**Osakidetza:** universal public health service, with 4 referral hospitals

## Methodology of registry

### Application form

Used for cases declaration

Integrated in the information system of Osakidetza, available for doctors.

- Two objectives:
- 1) Declaration of cases to ER-CAE (RD registry)
  - 2) Inform to other clinicians that the patient is been diagnosed of a RD

### Inclusion on RER-CAE (registry)

#### Clinical information

Periodical, twice a year, discharge of forms to registry server

- Selection of cases which corresponds to RD
- Unification of cases of the same RD
- Unification of disease preciseness for all RD

#### Personal information

Not provided by health professionals  
Recovered from information of Health Ministry (Health Insurance Card data)

#### RD information

Recovered from ORPHA DATA: "Disorders, cross referenced with other nomenclatures", "Linearisation of disorders"

#### Data gathering from other registries

Not yet implemented: gathering data from Mortality registry or Basque-Country Bio-Bank

- 1) RD Name
- 1.a) RD sub-type
- 6) Diagnosis comments
- 12) General observations

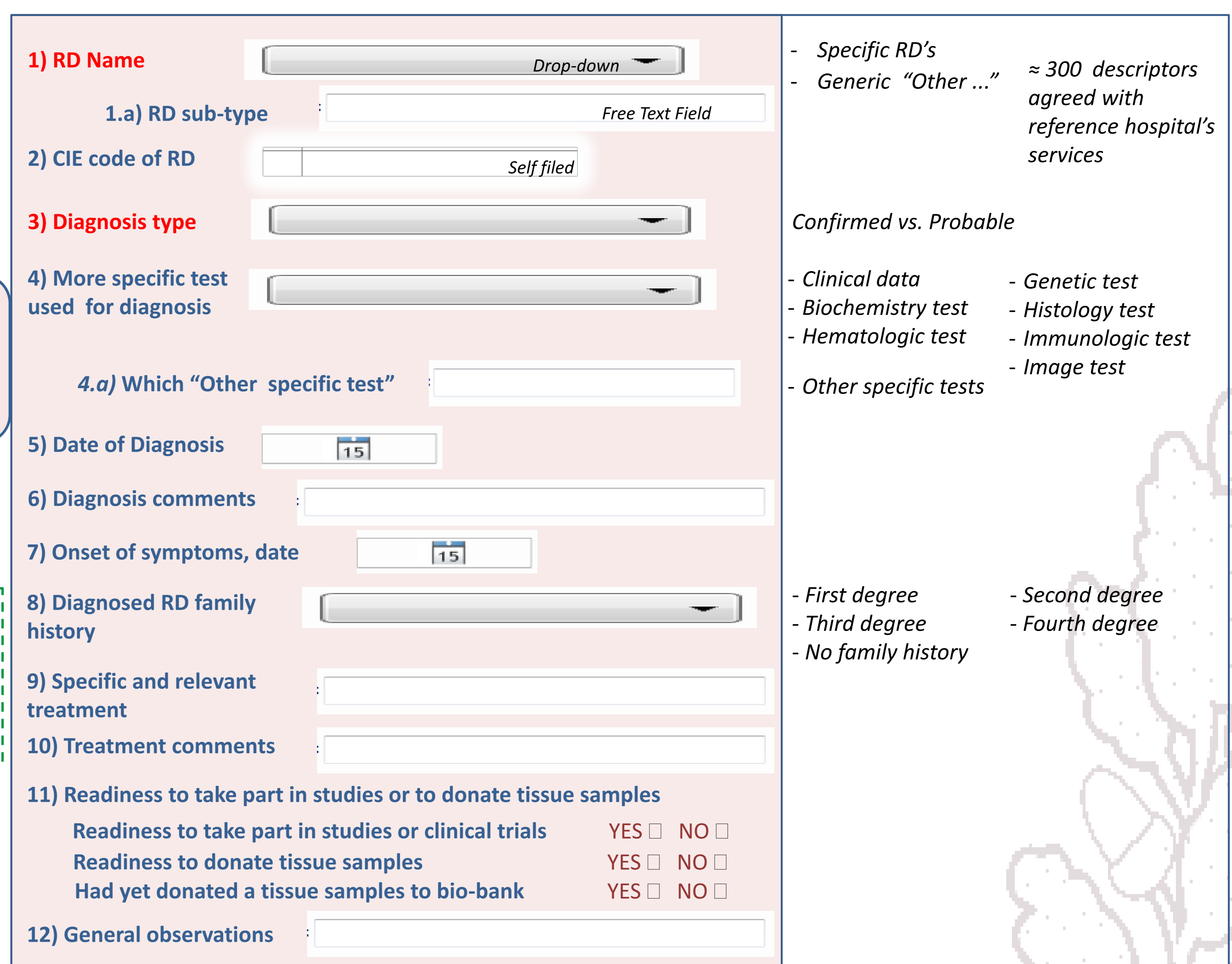
- ✓ Assignment of ORPHA code of disease Mandatory
- ✓ Assignment of ORPHA code of sub-type When enough information is available

Health Record (electronic)

- Assurance of privacy in information transmission
- Up-to date maintenance of personal data

Diseases classification  
Data analysis standardization

- Complete information
- Contrast data



1) RD Name: Drop-down menu

1.a) RD sub-type: Free Text Field

2) CIE code of RD: Self filled

3) Diagnosis type: Drop-down menu

4) More specific test used for diagnosis: Drop-down menu

4.a) Which "Other specific test": Free Text Field

5) Date of Diagnosis: Date picker (15)

6) Diagnosis comments: Free Text Field

7) Onset of symptoms, date: Date picker (15)

8) Diagnosed RD family history: Drop-down menu

9) Specific and relevant treatment: Free Text Field

10) Treatment comments: Free Text Field

11) Readiness to take part in studies or to donate tissue samples

Readiness to take part in studies or clinical trials: YES  NO

Readiness to donate tissue samples: YES  NO

Had yet donated a tissue samples to bio-bank: YES  NO

12) General observations: Free Text Field

- Specific RD's  
- Generic "Other ..." ≈ 300 descriptors agreed with reference hospital's services

Confirmed vs. Probable

- Clinical data  
- Biochemistry test  
- Hematologic test  
- Other specific tests

- Genetic test  
- Histology test  
- Immunologic test  
- Image test

- First degree  
- Third degree  
- No family history

- Second degree  
- Fourth degree

## Situation of RER-CAE registry

**Diseases:** Number: 479 Orpha codified entities

Disease (Orpha "preciseness" level) 86,4% ➔ 6,7% of cases are also assigned a sub-type of their disease

- 63,2% Disease
- 19,9% Malformation Syndrome
- 2,3% Morphological Anomaly
- 1,0% Clinical syndrome

Group of phenoma 13,6%

Frequency: 27 RD's makes up the 50% of cases in the RER-CAE, 218 RD's have one (1) case registered (altogether, 7,0% of cases), and 72 RD's have two (2) cases registered (4,7% of cases).

**Cases:** 3.105 at 01/2018 (954 cases in 06/2017 (1<sup>st</sup> charge of forms))

**Patients:** 3.090 (15 patients with two (2) RD declared)

Figure 2. Services with greater contribution to the RER-CAE

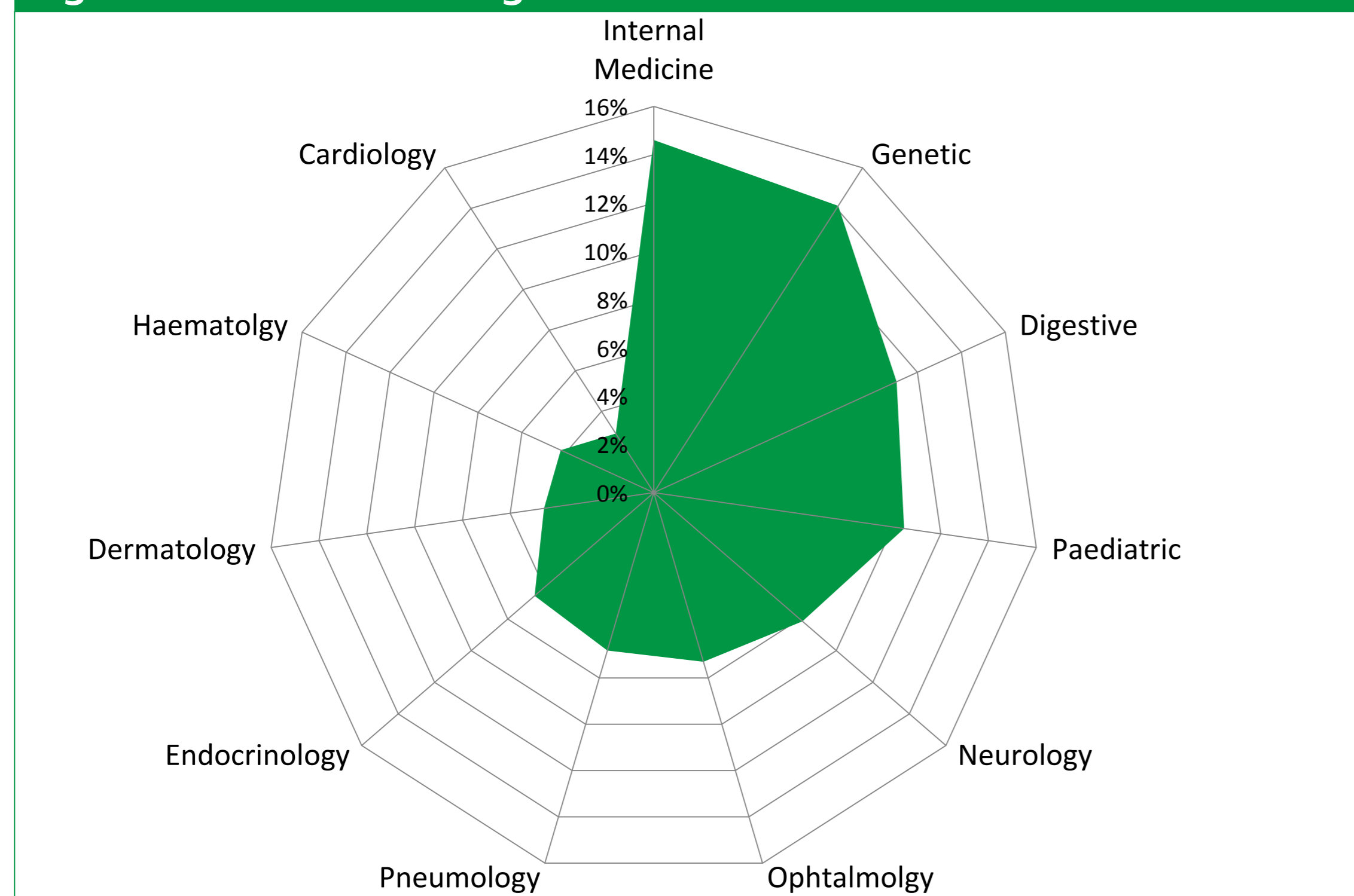
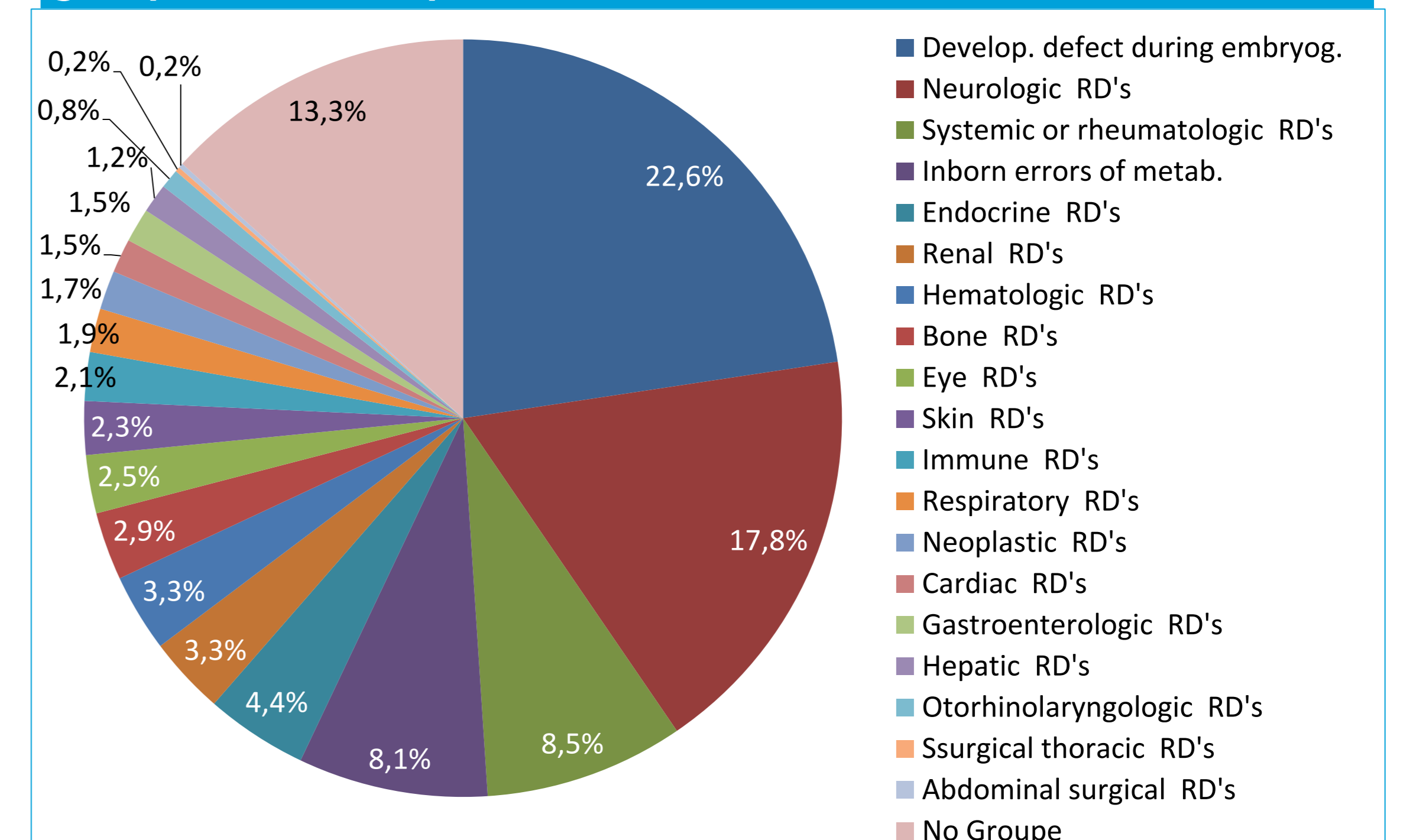


Figure 1. Distribution of cases registered, according to RD's groups obtained by ORPHA linearisation



### Challenges:

- ✓ Encourage doctors implication in the registry, to increase declaration of cases
- ✓ Feed-back the RER-CAE generated added value to health professionals
- ✓ With the goal of sharing information, improve relationships with registries at different levels: Spanish RD registry, RD-Connect and also RD specific registries, both to a national or international scope