RD-ACTION
DATA AND POLICIES FOR RARE DISEASES

Work Package 5

Orphacodes' use for the codification of rare diseases: results of the testing activity carried out within the RD-Action framework

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BACKGROUND

In November 2014, the Commission Expert Group on Rare Diseases (CEGRD) adopted a recommendation entitled **Recommendations on Ways to Improve Codification for Rare Diseases in Health Information Systems** in which it recommended to include a codification policy for RD in national plans and to consider to introduce the Orphanet codes (ORPHA codes) in Member States' health information systems. In order to support this implementation, the RD-Action set up a specific work package, WP5, led by DIMDI (Germany) with the participation of the French national RD registry (BNDMR) and the Veneto Region RD registry (Italy).

OBJECTIVE

To test the resources developed so far by WP5 and to use the results for the fine-tuning and the adapting of the developed products.

MATERIALS AND METHODS

The testing exercise is articulated in two phases
1.First phase (concluded)
Retrospective approach
Data from the French RD registry (BNDMR) and from Veneto region RD registry
Aim: to compare the real world use of orphacodes in these two different settings and to evaluate the impact of the use of orphacodes in terms of **comparability** of the monitored **RD entities** and the monitored **RD patients**

Background situation (France and Veneto region)

	France	Veneto region
Diagnostic spectrum recording	Initial investigations confirmed diagnosis and non-diagnosed patients	Confirmed clinical diagnosis
Terminology systems	ORPHACODES / OMIM / ICD10 /	ORPHACODES / OMIM / ICD9-CM/ ICD10 /
Specific benefits for RD pts	no	yes
Patient identifier	Specific for RD	Specific for RD
	National ID for health care	National ID for health care
Interoperability framework	Yes	Yes
National data repository	BNDMR	National RD Registry (minimum dataset)
RD expert Centres	131 RD expert centres officially identified	14 RD expert centres officially identified
Regulation	As of Jan 2016, the recording in BNDMR is mandatory	As of 2001, RD recording is mandatory
Period	Since 2007	Since 2002

Results presented in this poster refer to the 1st testing phase
The analysis performed refers to the period 2007-2016
and to patients with a confirmed diagnosis.
Data from Veneto region were collected by both genetic Centers and clinical Centers, whilst for France only data coming from genetic Centers were considered in the analysis.

2. Second phase (ongoing)
Prospective approach
Testing of the master file (different tools proposed) and the coding guidelines in other countries (specific settings, even small scale)
Aim: to give feedback on the usability of the developed resources (MF and guidelines) as well as on their contents.
Results will be available at the end of the project (July 2018)

WP5 ACHIEVEMENTS SO FAR

- **Surveys on current codification situations in Member States (MS) 2016 and 2017:** a basis to define and set the necessary strategy and tools to implement the orphacodes in the European countries
- **Review document of existing technical implementations for rare disease coding.**
- **Standard procedure and guide for the coding with orphacodes**
- **Specifications for an integrated coding application with orphacodes**

This document provides information about the use cases of RD coding, the coding process, reference methods for the implementation of the Orphanet nomenclature into Health Systems and the technical requirements. It contains 6 guidelines adapted to different and well-defined coding situations (the related contents are displayed in **poster n. 78**)

- **Specification and implementation manual for the beta Master file (MF) and the Beta version of the Master file**

The **Master file** is an extract from the Orphanet nomenclature which contains the minimum level of detail that is considered necessary for exchanging data at international level. These resources are intended to:
- provide a standard for coding RD (minimal level of standardisation)
- support consistency across MS
- allow different national coding practices
- enable international statistical retrieve and aggregation (interoperability)

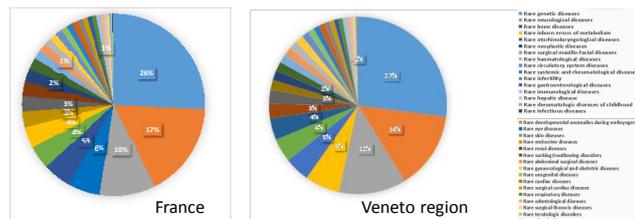**All documents can be found on the RD-Action website (see QR-Code below)**

RESULTS OF THE 1st TESTING PHASE

**Comparison of Orphacodes used in France (BNDMR) and Veneto region (RD registry) (2016)**

VR orphacodes n= 4386
FR orphacodes n= 2445
Total OC (VR+FR) n= 5349

Common 54,3
VR only 27,7
FR only 18
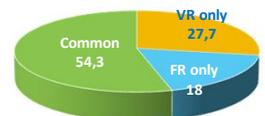
Monitored RD entities in France (BNDMR) and Veneto region (RD registry): orphacodes distribution per Orphanet classifications

France — Veneto region

Rare genetic diseases
Rare neurological diseases
Rare bone diseases
Rare inborn errors of metabolism
Rare otorhinolaryngological diseases
Rare neoplastic diseases
Rare surgical maxillo-facial diseases
Rare haematological diseases
Rare circulatory system diseases
Rare systemic and rheumatological diseases
Rare infertility
Rare gastroenterological diseases
Rare immunological diseases
Rare hepatic disease
Rare rheumatologic diseases of childhood
Rare infectious diseases
Rare developmental anomalies during embryogenesis
Rare eye diseases
Rare skin diseases
Rare endocrine diseases
Rare renal diseases
Rare suckling/swallowing disorders
Rare abdominal surgical diseases
Rare gynaecological and obstetric diseases
Rare respiratory diseases
Rare cardiac diseases
Rare surgical cardiac diseases
Rare odontological diseases
Rare surgical thoracic diseases
Rare teratologic diseases
Rare allergic diseases

Monitored RD patients in France (BNDMR) and Veneto region (RD registry): orphacodes distribution per Orphanet classifications

France — Veneto region
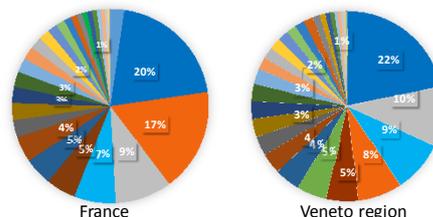
CONCLUSIONS AND NEXT STEPS

The use of orphacodes to describe RD entities monitored in Veneto region and France is highly comparable, although baseline differences in terms of organization of the RD care network and sources of information used to record patients exist in the two settings.

The comparability increases if OC are used not only as a flat list of codes, but embedding the hierarchical structure of the Orphanet classification. Further questions that should be addressed, also taking into account the results of the 2nd testing phase, are the following:
- which is the level of granularity of the Orphanet classification that has to be represented in the MF to better achieve data comparability across MS?
- how to preserve and exploit the hierarchical and multidimensional aspect of the Orphanet classification in the MF and for RD coding in general?
A new EU-Project ("Orphacodes project") will implement Orphacodes into routine coding systems in different MS, considering resources developed by WP5.

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