RD-CODE Malta Outcome Report

DELIVERABLE 4.9

15 December 2021

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Background

Malta population is of 436, 947 and the Mater Dei hospital, RD-CODE partner, is the sole tertiary care Hospital.

Previous to the begining of the RD-CODE project, ORPHAcodes were in use but their annotation was manual and retrospective. Such ORPHAcoding was carried out for 10 years [for all rare cancers (persons still alive); all congenital anomalies and all patients who have been sent abroad for diagnosis and treatment suffering from rare diseases].

By participating in the RD-CODE project the Mater Dei Hospital aimed at reaching the ascertainment of rare disease patients through a centralized non-manual collection, as this would provide complete ascertainment of rare disease statistics for the country. To achieve this, Mater Dei work within RD-CODE project was organised into the subtasks below :

Subtask 4.2.1 Review of the current data collection organization for rare diseases
A review of the existing data collection system has been organised and has allowed to develop recommendations that allowed:
- Optimised data capture
- Identification of additional potential sources of data within the Maltese system
- Better processing of already existing data, including maps for translation from existing classification systems to ORPHAcodes
- Better outputs and dissemination of such outputs, with a view to informing policy and to assist patients.

Subtask 4.2.2 Development of a technical solution within the hospital EHR
The implementation of the recommendation(s) produced in Task 4.2.1 allowed to proceed to subtask 4.2.2. and deliver a technical solution (see methodology section)

Subtask 4.2.3 Training of clinicians on ORPHAcodes
A remote Workshop was held on the 24 and 27 November 2020: "Introducing ORPHAcodes". It gathered 5 RD-CODE speakers and 36 participants from Malta : representatives from the 16 Mater Dei Clinical departments were present as well as government representatives, all Directorate for Health Information & Research (DHIR) Registries managers and Hospital Information Systems specialist and several consultants.

Methodology

A tool allowing to code with ORPHAcodes has been developed thus improving existing solutions, according to the guidance documents produced in WP5 and to recommendations issued from Task 4.2.1. The API was re-written as the sources of data were not enough since they provided only an ICD code. The software had to be reconnected to a source which provided more narrative, allowing to retrieve the ORPHAcode.

The system is in place in the Mater Dei Electronic Health Record.

Comparative anaylsis of RD Statistics delivered from this project to those delivered previously by manual collection

To date the data that is included in the register reached a total of 5,600 patients registered (projected total population of RD in Malta should be ca. 25K).
There is a total of 1,007 different ORPHAcode diagnoses.

Currently there are about 704 different diagnoses that can be entered in the tool by ORPHAcode Autotext. In addition, there are about 573 retrospective cases which have as yet to be assigned an Orphacode and this has to be done manually by looking at any electronic records available. In most cases, detailed ascertainment will permit the classification of these rare diseases into specific disorders rather than disease groups.

Comparison between data collected before the RD-CODE project :

1) An important increase in the number of registered cases has been observed since the beginning of the RD-CODE project, as the new tool facilitates the ORPHAcoding. Its availability has also facilitated the arrival of a one additional source of information : the vulnerable persons lists of people who suffer from rare diseases and which were collected for the purpose of the COVID 19 pandemic.

The registry was established in 2016; before the RD-CODE project starts in 2019 there was approx. 800 newly addition /year.
Since the RD-CODE project begun there is a median addition of 1,400 cases per year.

2) The tool allows to automatically identify obsoleted and deprecated codes, so there is no need for manual re-assignation of these codes. This also is a big improvement compared to the previously done analysis in which these codes needed to be manually re-assigned.

3) The Workshop introducing ORPHAcodes was beneficial and convinced most of the clinicians on the benefits of using ORPHAcodes to trace Rare Disease diagnosis in Health information Systems.

4) Benefits from this implementation resulted more accurate statistics and easier finding of cases for analysis

Lesson learned

The importance of inviting and ensuring attendance of all stakeholders to the training/workshop ensured that all have the appropriate amount of information and a common endeavour for this effort.

Having available all learning and training resources at hand is beneficial for the coders as it makes their work easier.

Having a follow up plan that can be sustained is essential

There always will be sectors that will over-engage while other resists. But an updated system with ongoing training clearly benefits the overall process.