

ESTIMATING THE IMPACT OF RARE DISORDERS ON POPULATION HEALTH AN ITALIAN EXPERIENCE

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BACKGROUND

Due to RD extreme variability and cross-sectional age distribution it is extremely difficult to calculate or just estimate their whole impact on the population's health. To tackle this question, it is necessary to have an area wide enough to be completely monitored for all RD, which is very difficult to be pursued. To properly determine the impact of RD at a population level, we need area-based surveillance systems, monitoring not the single conditions but the whole group of RD. In Italy in 2001 a specific law on RD was issued, containing a list of 581 diseases or groups of disorders defined as rare (2138 considering synonyms and diseases included into groups), divided into 13 nosological categories. A national network of reference centers for diagnosis, treatment, and prevention of RD was established, together with area-based Registers monitoring all patients. An exemption from specific health care costs was introduced, leading patients affected by RD to join the surveillance system in order to achieve the benefits. In enforcing the RD national law, specific health policies were developed by the Veneto Region, North-East of Italy, 4,700,000 inhabitants. A regional network of reference centers for each group of RD was created, and a unique on-line electronic monitoring system allowing diagnosis recording, exemption leading to benefits' entitlement and cases' enrollment in the Register was implemented.

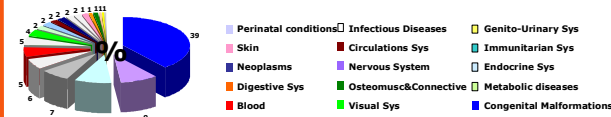
OBJECTIVES

- To compare the Italian list RD coding and classification in ICD9-CM and ICD10
- To estimate the impact of RD on population's health using two independent sources of data monitoring the same population: Veneto Region Register for Rare Disorders (VRRDR) and Hospital Discharge Records Register.

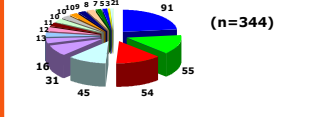


ITALIAN RD LIST

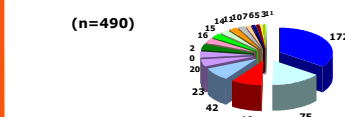
1 DISEASES distribution per ICD9-CM groups (n=2138)



2 CODES distribution per ICD9-CM groups (n=344)



3 CODES distribution per ICD-10 groups (n=490)



The Italian RD list contains 2138 names of diseases corresponding to 344 ICD9-CM codes and to 490 ICD10 codes. The majority of them belongs to the congenital malformations group (26%), followed by codes classifying eye diseases and blood disorders.

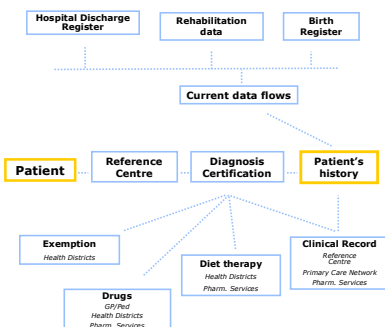
In ICD-10, the distribution of codes changes, if compared to ICD9-CM, particularly for congenital malformation, metabolic diseases and immunological diseases. The number of codes which can potentially identify a rare condition increases, the congenital malformations group being still the most represented one (35%).

According to the hypothesis that a patient affected by a RD due to the condition itself or the related comorbidities has a higher probability of being admitted to the hospital, we have used HDR Register to estimate the number of RD patients in Veneto Region. Nearly 3 million HDR for the period 2001-2005 have been retrieved, selecting all the records containing at least one RD ICD9-CM code. According to specificity, selected codes were divided into 3 different groups (see 4).

As using non-specific codes could due to a considerable overestimation of potentially affected patients, we have considered only specific codes (G1+G2). Using this method we identify 21.131 RD patients residing in the area. The overall crude prevalence of RD calculated for specific codes is 4.8 per 1,000 inhabitants.

We have then considered another source of data, independent from the previous one: Veneto Region RD Register, which enrolls 12,474 patients. The distribution of diagnosed patient per nosological group and age is represented in fig. 7. The comparison of RD patients distribution per nosological group according to the 2 sources reveals a substantial overlap for most of the considered groups, with some exceptions. 6. The higher frequency of patients diagnosed with congenital malformations in HDR can be due to Down syndrome cases and early mortality observed for other congenital conditions, determining their absence in the RD Register. On the other side, there is a higher frequency of eye diseases' cases in the RD Register. This could be due to conditions as keratokonus and retinitis pigmentosa, which are present in the Italian RD list, but don't necessarily imply frequent hospital admissions. For the remaining groups, a good coverage of the monitored population is demonstrated.

THE REGISTER



THE WIDE AREA

With the creation of the Wide Area (WA) for RD in 2004, Veneto Region was connected to Friuli Venezia-Giulia Region, Trentino Alto Adige (Trento and Bolzano Provinces) in order to:

- § create a wider dedicated network identifying common Reference Centres for specific RD groups;
- § share common care pathways and modalities of providing benefits for RD patients;
- § implement a unique and common area-based monitoring system;
- § provide epidemiological data useful for policy makers to health planning and evaluation processes

CONCLUSIONS

RD can be described both by specific and non-specific codes according to ICD. In the shift from ICD9-CM to ICD-10 an improvement in RD coding was observed for some diseases, especially congenital malformations, but some problems still exist. Using the ICD9-CM 30% of the selected codes for the Italian RD list are general non-specific ones. Furthermore both ICD releases have a system-centered logic that complicates the coding of multi-systemic RD i.e. mitochondrial disorders.

Besides RD Registers, ICD-based current statistics can be used to obtain rough estimations of the number of patients affected by RD residing in a defined area.

