

# A survey on rare disease patient organizations in Italy

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## Abstract

Orphanet is the reference database for rare diseases (RD) and orphan drugs (OD). Here we present the result of a mapping activity of Italian patient organizations (POs), started by the Orphanet-Italia team on January 2013. The total number of associations collected in our database amounts to 325, most of which are concentrated in the north of Italy, (~53%) followed by the center (~33%) and the south (~14%). In order to set the issue within the more general context of European policies, we have simulated the distribution of the Italian associations into their respective European Reference Networks (ERN), finding that every ERN, except TRANSPLANT-CHILD, is represented by the associations registered in our database.

From these starting points, we have focused our attention on the patient organizations that have not merged into an "umbrella" organization, since the unaffiliated subjects, notably, constitute the majority (>70%). We have considered the inclusion in "Uniamo", the only Italian federation of rare disease associations affiliated to EURORDIS-Rare Diseases Europe, as a benchmark for this evaluation. Finally, we have examined the patient organizations attitude towards Internet usage, for searching and sharing health information.

In recent years, the rise of patient organizations registered in the Orphanet database, illustrates the growing role of these social players. They not only constitute a reference point for the patients, helping them to get out of isolation, but are also more involved in RD management.

The increased collaboration with healthcare professionals, through the patients participations to clinical trials or their active engagement in fund raising for research, well exemplify this trend. The patients representation in the ERNs, by mean of the ePAG initiative, can be seen as the culmination of such an ongoing evolution so far, aspiring to make the RD patients voice heard by the EU policy makers.

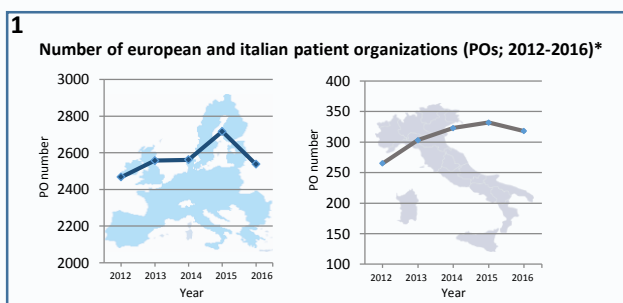
## Methods

**Inclusion/exclusion criteria for PO registration.** The patient organizations can be registered in the Orphanet database only if some main criteria are met, summarized in the following.

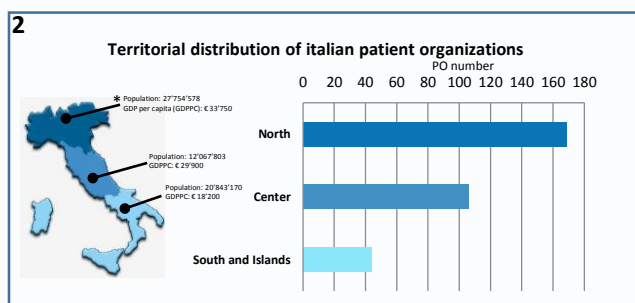
\*Inclusion criteria: i) the PO/alliance has to provide support for a rare disease, a group of rare diseases or rare forms of common diseases; ii) the PO/alliance should have a legal status, according to the country's laws; iii) the PO/alliance has to be responsive and must be able to be contacted by telephone, e-mail, etc.; iv) the PO/alliance should have a designated head and/or a contact person; v) the regional PO are accepted if there is no national equivalent.

\*Exclusion criteria: i) a PO dealing with non-rare diseases; ii) a fund-raising trust or foundation that helps one or several patients with no real advice or help given to others; iii) a learned society; iv) a research-funding trust/foundation; v) a blog or/and forum only. \* The inclusion/exclusion criteria have been taken from Orphanet's Standard Operating Procedures.

**ERN allocation to POs.** We searched for the disease/group of diseases concerned by each PO, then, taking advantage of the advice provided by an expert geneticist, we could allocate the ERN that best fitted the pathogenic condition represented by the PO.



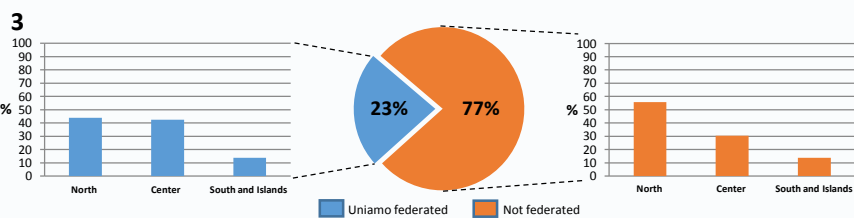
\* Data from the "MonitorRare" report, published in 2017 by Uniamo ([http://www.uniamo.org/attachments/1022\\_monitorare\\_2017\\_web.pdf](http://www.uniamo.org/attachments/1022_monitorare_2017_web.pdf)).



\* Data from the Italian National Institute of Statistics (Istat, <https://www.istat.it/en/archive/207798>).

## Panel 1-2

The growing role of Orphanet as a source of information in the field of rare diseases, is reflected by the relative increase of patient organizations registered in the Orphanet website in the reference period 2012-2016 (panel 1). From 2012 to 2015 there was a 10% increase of Associations registered at the European level (left graph), followed by a strong decrease in 2016. Such a sharp reduction is due to the exclusion, carried out by the Orphanet team in 2016, of diseases that did not fall within the "rare" definition, according to European legislation defining a prevalence threshold of 1/2000. The right graph represents the number of Italian organizations in the reference period, with a 20% increase from 2012 to 2016. The panel 2 shows the absolute number of the Italian POs distributed in the three main geographical areas of the country that are largely used for demographic and social statistics. As shown in graph, most of patient organizations are located in the North, followed by the Center and by South and Islands. We observe that such a pattern mirrors the worsening of socioeconomic conditions along the North-South axis.



## Panel 3

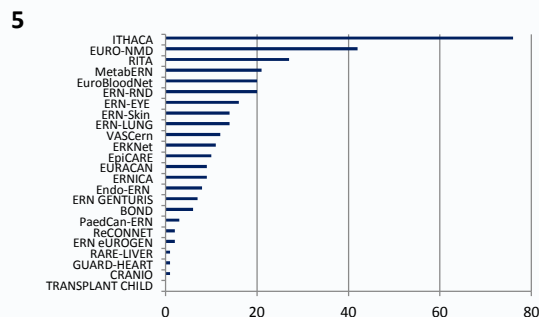
The pie chart represents the percentage of Uniamo federated organizations (light blue) and the percentage of the not federated ones (orange). Each slice has been further divided according to the territorial distribution of the Uniamo federated or not federated organizations, respectively. The large majority of them are not federated. Interestingly, the POs of central Italy demonstrated to be more willing to merge into the Uniamo umbrella.

## Table 4

The table shows the number of POs without a website and their participation to the Uniamo federation. The POs without a website represent a very low proportion of the total (~7%) but, noteworthy, almost all of them (20/21) fall in the not Uniamo federated subpopulation.

## 4 POs without a website

Geographic area	PO number	% of total	Uniamo federated Yes/No
North	8	2,5	No
Center	7	2,2	No
South and Islands	5	1,6	No
	1	0,3	Yes
<b>Total</b>	<b>21</b>	<b>6,6</b>	



## Graph 5

We divided the patient organizations according to their possible inclusion in one of the 24 ERN. The graph shows the ERN representation by the registered POs; all ERN, but TRANSPLANT CHILD, are represented by the Italian organizations.

## Conclusions

In this preliminary study we have taken a picture of the Italian patient organizations. Despite the need to deepen our analysis, particularly as regards the number and location of our study subjects, some key element emerges. As shown, the majority of the POs are neither federated to Uniamo nor, to our knowledge, to other federations. We hypothesize that the small organizations are somehow afraid of merging into greater entities, fearing to lose representativeness. On the other hand, it is possible that, for a quote of the not federated organizations, the active participation in health policies making is not a priority, so the creation of a stronger representative organization is seen as unnecessary. With regards to this matter, our data show that the attitude to use internet, if properly developed, can be a useful tool to evaluate the propensity to federate. The need for patient organizations to make the RD patients voice heard by the policy makers, both at the national and at the EU level, is essential in order to have specialized health care services that, importantly, must be sustainable by the patients and their families, independently of the Country/geographic area of origin.