Rare Diseases Internet Information Retrieval and Knowledge Discovery

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INTRODUCTION

A rare or orphan disease (RD) is that which affects only an insignificant percentage of population, so traditionally has not received the effort devoted to common diseases. In common diseases there are healing or palliative treatments while there are over 4000 RD without a known cure. In Europe, RD affect between 27 and 36 million people (Rodwell & Aymé, 2013). Any progress in this field has a great social impact that translates directly into an improved quality of life of those affected by RD (Dragusin et al., 2011). Therefore, any advance in knowledge generation in the context of RD is welcome. One of the most important developments has come from data mining (see (Groth, 1998), and (Tomar & Agarwal, 2013)). The reduced amount of data makes the application of data mining techniques not easy in the field of RDs. Although for one disease the number of patients and data is reduced, they are numerous as a whole. Several initiatives collect data from RD patients around the world using the power of Internet (Orphadata, 2015).

Given singularity of RDs, it is necessary to provide the information systems, the capacity to collect data and also promote cross-flow of information between the agents (patients and researchers). In this situation, it is interesting the development of projects like the one sponsored by the Avenzoar Chair (University of Seville, Spain), known as ER2.0 Project (see details in Rabasco et al. (2013a) and Rabasco et al. (2013b)). The underlying idea is to make it real to collaborate, share, and take decisions in a web 2.0 manner between patients and researchers in RDs.

This chapter contains, first, a review of the information networks associated with RD, highlighting the registry, typology, associations and consortia related as well as the existing relationships between

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them. Next, follows a review of data mining techniques in the context of healthcare, especially of RDs. Later on, it is described the distinguishing aspects of a web tool for the collection and treatment of information provided by RD patients in the context of ER2.0 with the objective of being a starting point to apply data mining techniques to RD.

BACKGROUND

Information Networks Associated with Rare Diseases

There are recommendations from both the European Union and the United States about the importance of using information networks and databases on RD for systematic data collection with the participation of both patients and researchers (Mazzucato, Visonà Dalla Pozza, Manea, Minichiello, & Facchin, 2014). In Table 1 there is a summary of the main characteristics of RD information networks in RDs.

Table 1. Characteristics of RD information networks

Organization [Support]	Impact Google/Bing/Alexa	Network Soc./Inf.Flow/ Data	Partners
GARD [G,R]	79500/114000/215 (NIH,2015)	O/-/T,DB	NIH, NCATS
GRDR [G,PR]	5660/37/215 (NIH,2015)	O/P-P,E-E,P-E/T,L,DB	NIH/NCATS
ORDR [G,R]	84400/89500/215 (NIH,2015)	-/E-E/DB,T,L	NIH/NCATS, GRDR
RD Program (FDA) [G,PR]	2650/3450/4586 (FDA, 2015)	-/-/T,L	NIH/(NCATS)
RDRD (RD Repurposing Database) [G,PR]	1310/55/4586 (FDA, 2015)	-/-/T,DB,L	FDA
SpainRDR [G,R]	2020/130000/30052 (H. Inst. Carlos III, 2015)	-/-/T,BD,L	H. Inst. Carlos III, EUROPLAN, FEDER, IRDiRC
Orphanet [G,R]	1370000/707000/56070	-/E-E/DB,T,L	CARD, WHO, EuroP-Ean Commission, amongst others.
Patients Like Me [PA]	528000/164000/57444	S/P-E,E-E,P-P/DB,T, L	
CNMR [G,R]	317/33/100877	O,S/E-E,P-P,P-E/T,L	EUROPLAN
NORD [G,PA]	204000/90500/177751	S/P-E,E-E,P-P/L,T	EURORDIS, JPA, GARD
RareConnect [PA]	87600/32700/529546	S/P-P/T,L	EURORDIS, NORD
FEDER [G,PA]	52100/45100/600036	O/P-P,P-E,E-E/T,L	Mehuer, EURORDIS
EURORDIS [G,PA]	21400/12900/1051803	O/P-P/T,L	EUROPLAN, Black Swan F., FEDER, CARD, RD International
Findacure [R]	392/54/1337476	O,S/P-P,P-E,E-E/T,L	RE(ACT) Comm., Cures within Reach, EURORDIS, Rare Disease UK
EUROGENTEST [G,PR]	64600/38200/1523427	O/E-E/T,L	
JPA [PA]	64/52/2002261	O/-/T	EURORDIS
Fondation Maladies Rares [G,R]	8240/5310/3070126	O,S/P-P,P-E,E-E/-	RE(ACT) Community
IRDiRC [G,R]	2990/3000 –3821331	-/E-E/T,L	Orphanet, EuroP-Ean Comm, NIH
Rare Disease Dot Org [PA]	1190/3/4267630	S,O/P-P/T	

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