EuOrphan: a website collecting information on drugs for rare diseases

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Research and scientific progress in the rare disease field is a challenging objective, since usually only few highly specialised research centres are dealing with each specific condition. This is even more relevant if we consider that a great part of rare disease patients are children, as the 80% of rare diseases are genetic.
Taking into account the geographical dispersion and the rarity of pathologies, the collection of adequate information on medicines marketed or in development is useful for the scientific community and for patients.
EuOrphan is a free information service aimed to collect and disseminate updated information on drugs for rare diseases. A complete and updated database of orphan drugs designated in Europe (by EMA) and in the United States (by FDA) is available. Recently, the database has been implemented with data on drugs marketed for a rare disease by the European Agency before the European Regulation on Orphan Drugs (EC) 141/2000 entered into force.
For each drug, the following information is collected by official EMA and FDA sources: active substance, sponsorship (and its country), the designation date, orphan condition(s), the orphan indication(s).
Orphan conditions are grouped in 14 disease areas. For each of them, it is specified if it is genetic and if involves the paediatric population.
In case of approved drugs, the trade name, ATC code, Marketing Authorisation date, approved therapeutic indication (specifying the paediatric approved ages, where existing), the Marketing Authorisation Holder (and its country) are indicated.
In addition, the database includes information on studies representing the level of scientific evidence at the time of the designation or approval, as well as on ongoing clinical trials.
The data are stored on a SQL Express 2008 Database. The EuOrphan website (www.euorphan.com) consists in an user friendly interface that allows to operate different kinds of queries on this database.Information about drugs can be retrieved querying by Active substance, Trade name, Disease area, Company (Sponsor / Marketing Authorization Holder) or 'free text' search.
The last update (June 1st 2013) reveals that 860 orphan designations have been released from EMA (from 2000) and 1889 from FDA (from 1983). All these designations refer to 1179 orphan conditions (356 are genetic, 948 affect the paediatric population).
Our results show that a great part of genetic rare diseases have still an unmet therapeutic need.
With reference to marketed drugs, EMA has totally approved 148 drugs indicated for a rare disease: 62 have been designated as 'orphan drug' according to the EU Regulation (EC)141/2000, 85 received the Marketing Authorisation before the Regulation (including 6 'orphan-like drugs'). 63 of authorised drugs (42.5%) have a paediatric indication.
On the other hand, FDA approved 354 orphan drugs, 147 of which (41.5%) has a paediatric indication.
An important result is that the 52,5% of marketed drug from EMA indicated for a disease affecting children, do not have a paediatric indication (52/99). Similarly, for FDA this percentage is 49.3% (137/278).
Regarding the clinical evidence supporting the approval phase, preliminary data suggest that only a part of orphan active substances (about 30%) has been authorised on the basis of a standardized phase I to III scheme. For example, we found 4 orphan drugs authorised on the basis of retrospective data, bibliographic references, uncontrolled trials or compassionate use: carglumic acid, mitotane, aragrelide and thiotepa.
Conclusively, EuOrphan represents a relevant source of information on drugs for rare diseases. The challenge is to address a referenced scientific and regulatory information, easily comprehensive in the meantime, to a large number of users. In this way, not only healthcare providers and experts but also patients and their families may benefit of this information tool.