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Editorial

Investigating Rare Diseases: How to Advance and Make a Relevant Social Impact

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There is no consensus on the epidemiological definition of rare diseases across the world: The United States (US) defines this group when the disease occurs in less than 200,000 individuals in US [1] while a rare disease is defined by the European Union (EU) as a "life-threatening or chronically debilitating diseases which are of such low prevalence that special combined efforts are needed to address them." [2,3]. The low prevalence is then defined by EU as one that affects less than 5 in 10,000 of the general population [2,3]. On the order hand, according to the World Health Organization (WHO), rare diseases affect less than 10 patients in 100,000 individuals. Although each nosological entity is rare, this is a large group accounting for more than 5,000 known diseases, using the WHO criteria.

Rare diseases demand higher levels of collaborative efforts among experts in order to make relevant achievements in translational and clinical research that may impact patient lives. To this extent, the establishment of scientific consortiums covering all aspects of research (basic, translational and clinical) represents the only model able to accelerate scientific discoveries with relevant social impacts. Consortiums are expected to create an environment in which researchers may work more closely together and harmonize their efforts and results in a more timely fashion. To achieve these goals, it is imperative to share data and information on ongoing studies and set together well-defined objectives in order to avoid duplicating activities and share high-specialized facilities. As expected, all collaborative networks can only grow based on mutual trust.

Prospective studies, the gold-standard of biomedical studies, may represent a barrier when rare diseases are considered, because it may take a long time to collect a significant number of samples and clinical data. To this end, state-of-.the-art biobanks are extremely important to accelerate research, because scientific studies can start immediately with a large number of high quality well-preserved biological samples. Noteworthy, biobanks should not be developed based on the simplistic concept of storage sample facilities. Biobanks can only maximize their relevance to biomedicine if clinical, biological, epidemiological, demographic and lifestyle information are collected from each patient that donors their samples. More than only biological samples, we need associated information in order to understand the impact of each variable in disease initiation, progression, transformation and relapse. By considering the paucity of information about rare diseases, biobanks are central facilities in this arena that allow us to improve our knowledge and make a relevant social impact within shorter-term periods.

In general terms, patients affected by rare diseases are devoid of centers that can afford integral assistance in diagnosis and treatment. Moreover, most of these diseases do not have specific drugs, as a consequence of lack of financial interest by pharmaceutical industry and the impossibility of investigating rare diseases in isolated research groups.

This scenario claims for specific health public policies:

1. The creation of a low number of high-specialized dedicated centers that can afford integral assistance to a specific group of rare diseases that shares clinical-biological similarities.
2. The imperative social necessity of public financial support to collaborative research networks of experts.
3. The public protection of new developed orphan drugs in order to avoid their high costs as a consequence of market economy.
4. The creation of population-based National registries of rare diseases to address the real prevalence of each clinical entity in order to design, drive and improve all public policies stated above in a sustainable way. Noteworthy, all these efforts should also go beyond National frontiers and embrace low, medium and high-income countries.

It seems evident that rare diseases bring a great potential for innovation in all biomedicine related fields and also in public policies around the world. As a positive side effect, it may show to some of our colleagues that science is indeed a collaborative work rather than being competitive.

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