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Dear Colleagues, Dear Readers,

We are pleased to introduce the first issue of "Rare Diseases and Orphan Drugs", a new international scientific journal dedicated to the integration of public health considerations and methods to guide the care in the vast field of rare diseases. In view of the rising number of professional journals in the health care field, one may ask "Why a new journal?". And "Why a new journal on rare diseases?". We respond to this question by describing the intended goals of this Journal and what it can offer to the international community of rare diseases stakeholders.

Explaining rare diseases is not a simple task. The European Commission defines a rare disease as one that affects no more than 5 in 10 000 people, whereas different definitions are adopted in other Countries (e.g. in USA) (1,2). It sounds emotionless, something the common citizen perceives as a distant threat that would, perhaps, occur to somebody else. This might hold true for an individual rare disease. However, modern public health demands that even uncommon conditions deserve attention in order to fulfill the basic right to healthcare for all and each person. Moreover, the plentitude of rare diseases that are independent of each other suggest that individuals are substantial risk of suffering from a rare condition at a point in their life. Although the more than 5,000 rare conditions often show widely diverse symptoms and heterogeneous phenotypes, they also share several aspects. Although patients may experience quite different health problems, they have common expectations as regards the performances of public health systems. Support to research, which is needed for timely diagnosis, prevention and effective and safe treatments; correct, clear, comprehensive and available information to healthcare operators, families and patients; affordable and available health services, from diagnosis to rehabilitation; strategies to support the quality of life of patients, to empower them at school, workplace and society. Yet, medical science and health systems have limited effective answers, and the public attention is usually focused on other important issues. There is no reason or justification for launching a "competition" of rare diseases against frequent diseases: rather, we advocate that attention for rare diseases must be raised because, from a patient perspective, the result of overlooking the demands posed by rare diseases would be the denial of basic rights, the stigma for the affected people and the too often unbearable burden for families left alone with their sorrows by the community.

Rare diseases are a unique case in modern society demonstrating the enormous added value of global cooperation and collaboration. The last decade proved that advances in the care of patients with rare disease can be made by developing global frameworks: "global" means plurality of national experiences, of society voices and of scientific disciplines. Multidisciplinary science is an essential component of the framework. To fill the many knowledge gaps and address science-based health policies, the contribution of research on basic and medical genetics and epigenetics is utterly needed but not at all sufficient: all branches of medicine as well as pharmacology can provide fundamental contribution in understanding pathogenesis, developing clinical management protocols and drugs.

Sponsored by a European Union grant (RARE-Bestpractices Project, Ref.: n° 305690), as an open access journal with no publishing fees, the RARE journal will take a comprehensive, open and multi-facted approach to rare diseases as an unsolved public health issue . In addition to providing a platform for publishing results of research that deals with diagnosis, prevention and therapy of rare disease, a goal of the RARE Journal, is to foster methodological research on how to synthesize and present information for the care of people with rare diseases, such as in health technology assessment, systematic reviews and guidelines. Thus, RARE Journal intends to combine scientific quality, through a highly qualified peer-reviewing system, dissemination and openness to establish it as a qualified forum for scientists devoted to public health, health policy and clinical research relevant to rare diseases. The RARE journal will publish clinical and epidemiological original research, case reports, systematic reviews and meta-analyses, as well as reports of clinical practice guidelines, HTA, and meeting reports. Free and open discussion is an essential part of the RARE Journal's mission: thus, commentaries and letters to Editors are most welcome. Special Journal's issues dedicated to a specific topic are also anticipated, such as a particular rare disease or a group of rare diseases considered from different viewpoints.

Besides maintaining a high-level scientific quality, the RARE Journal has a few ultimate, ambitious goals: serving the whole international rare diseases community, fostering new activities and ideas, as well as attracting interest from a broader scientific audience which could contribute to a global, "one health" approach to rare diseases.

It will be a long journey, but long journeys start with small steps.

Domenica Taruscio, Holger Schünemann

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