



Editorial letter

Patient registry for the overlooked patient

Historically, much attention has been paid to patients with chronic disease, cancer, and other more common diseases. The Office of Rare Diseases Research at the National Institutes of Health (ORDR) proposes to establish a landmark, global, rare diseases registry to serve the patients that make up underserved and many times life-threatening category. In this issue, we are honored and excited to publish the proceedings of the well-organized and executed workshop, sponsored by the NIH, in January of this year. The workshop comprised significant experts and leaders in this area of research whose ideas, organization, and innovation are discussed in this article. A registry of all rare disease is a lofty and needed goal, which we support and are grateful to share with our readers in this issue.

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Letter to the Editor

Driving interest in consolidating resources for the creation of a global rare disease patient registry are the rapid advances in genetics and genomics research that are spurring rare disease discoveries, including commercially available genome tests indicating a person's risk or carrier status for disease, the realization that common diseases may be caused by rare, not common, mutations in multiple sites, and the realization that the knowledge acquired studying rare diseases can contribute to our understanding of the pathogenesis of common diseases. Also driving the interest is the revolution in global information and communication made possible by the Internet.

In addition, industry has shown new interest in developing rare disease treatments (for example, Pfizer, GlaxoSmithKline, and Novartis). "While there might be few patients with each disease, collectively, it's a very large patient population with a great unmet medical need", said David Simmons, President of Pfizer, in an interview with the *New York Times*. Building a global infrastructure for already existing and future registries for rare diseases will require adopting a business model and joining forces with the private sector to create a collaborative partnership to ensure that funding will be available to accomplish this goal and maintain the registry through a period of expansion.

The joint effort is intended to reduce the costs of developing and maintaining a registry for many of the 1000 patient groups in the United States that represent people with specific rare diseases. Many smaller groups or groups of patients who do not have a patient advocacy group to represent them or cannot afford the expense, said Stephen C. Groft, who heads the Office of Rare Diseases Research at the NIH (in an article in the *New York Times*). Dr. Groft also indicated that although building such a registry may start locally, eventually it should be a global effort, since diseases "know no borders." There is a strong sense in the public as well as among physicians, investigators, patients, academia, industry etc. that now is the time to embark on such an ambitious agenda, for good reasons.

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