Rare Diseases Terminology & Definitions: An Interview with Trevor Richter, PhD, MSc

Value & Outcomes Spotlight had the opportunity to catch up with Trevor Richter, PhD, MSc, author of the recent article, "Rare Disease Terminology & Definitions – A Systematic Global Review: Report of the ISPOR Rare Disease Special Interest Group," to appear in the September/October 2015 issue of *Value in Health*. Our conversation on this intriguing subject of rare diseases terminology & definitions follows.

***Value & Outcomes Spotlight*:** How did you get interested in this topic?

Trevor Richter: Working in health technology assessment (HTA) in a Canada, which has a publicly funded health care system, has highlighted the need for a strategy to deal with the issue of access to treatments for rare diseases. It was surprising to learn that a major problem that hinders equitable assessment of so-called 'orphan drugs' is the lack of a clear definition of 'rare diseases', not only within our jurisdiction but internationally. Given the global connectivity of health care, a special interest group (SIG) was formed with broad international representation to address the most basic of questions in this area, namely what terminology is used to define a rare disease? While carrying out our research, we discovered that many definitions included reference to a prevalence threshold; therefore, in addition to describing the global picture with regard to terminologies used to define rare diseases, we were able to also address the question of how prevalence thresholds are used globally within definitions of rare disease.

VOS: Who will benefit from these terminologies and definitions?

Richter: Our findings should be of immediate relevance to researchers, policy makers, and decision makers that currently have to operationalize the assessment of orphan drugs and associated health technologies. However, any stakeholder with an interest in rare diseases and orphan drugs/technologies would benefit from a clearer view how terminology is used globally, and the commonalities among definitions across different jurisdictions. Stakeholders with an interest in rare diseases include regulators, HTA agencies, payers, and patient organizations; although individual physicians and patients could also benefit from understanding some of the issues related to defining rare diseases and associated technologies.

VOS: How is this report valuable to researchers and decision makers?

Richter: A notable finding was that there is substantial diversity among definitions of rare diseases across jurisdictions and among different types of organization within jurisdictions. For those researchers and decision makers who are struggling with how to define rare diseases and develop assessment frameworks that can accommodate orphan drugs, this finding should provide some reassurance that similar challenges are being faced globally. On the other hand, we found a broad representation of jurisdictions and organizations that have in place publicly available definitions of rare diseases and associated technologies, which suggests that there is a universal desire to develop practical definitions. Our report provides a global picture of current definitions and should serve as a useful resource for stakeholders interested in which terminology to use in their definition of rare disease. We suggest in our report that future development of definitions of rare disease should focus on objective, quantitative metrics such as prevalence. The use of qualitative descriptors, which tend to be subjective, should be avoided, although these could still be accommodated in conceptual definitions of rare disease.

VOS: What kinds of problems will researchers and decision makers be able to answer?

Richter: Our report will allow researchers and decision makers to assess how their definitions of rare disease, particularly with regard to terminology and prevalence thresholds, compare to other jurisdictions, to similar organizations in other countries, and to other types of organization. At a broader level, it is our hope that our findings might form the basis for any attempts to harmonize terminology used in definitions of rare disease and/or to adopt a standardized prevalence threshold for defining rare disease, at a local or even international level, if appropriate. We recognize that there are many challenges associated with this, including variations in demographics and policy, but the fact that a wide variety of organizations have attempted to define rare diseases, as well as the existence of common terminology and prevalence thresholds across different jurisdictions, suggests that there is a widespread desire to standardize definitions of rare disease.

VOS: When will this review have the most impact in researching rare diseases and terminology?

Richter: Our report is relevant today, but will likely be of increasing relevance within the next few years as many organizations and jurisdictions attempt to either define rare diseases for the first time or update their existing definitions. For instance, the EU members have been progressive in publishing national rare disease plans that reference the EU definition of a rare disease, reflecting the need for rare disease definitions to be made explicit, as well as a shift towards harmonization of rare disease definitions on a political level.

VOS: Thanks Trevor for this interview and for chairing this informative ISPOR Special Interest Group. ■

Additional information:
Refer to the article, "Rare Disease Terminology & Definitions - A Systematic Global Review: Report of the ISPOR Rare Disease Special Interest Group," and other articles in this issue of *Value in Health* at: http://www.ispor.org/valueinhealth_index.asp.