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Interoperability at the cost of patient empowerment? The EU Rare Disease Common Data Elements for rare disease registry integration

Abstract

In 2017, the Working Group of the European Platform on Rare Disease Registration published its *Set of common data elements for Rare Disease Registration* (EU RD Platform, 2017). The document sets forth 16 data elements considered fundamental to furthering rare disease research, and covers patients' personal information, patient status, care pathway, disease history, diagnosis, data and/or samples made available for research, and patients' disability profile. This instrument is part of a broader effort to improve the interoperability of rare disease registries, to the advancement of rare disease understanding and knowledge on how to improve patients' health and wellbeing (Forrest et al., 2011). The Working Group consisted of experts from the EUCERD Joint Action, EPIRARE, and RD-CONNECT projects, all EU projects working on common data sets. In formulating its 16 data elements, the Working Group significantly reduced the 46 data elements proposed in a publication by EPIRARE in 2014 (Taruscio et al., 2014). Importantly, patient-reported outcomes (PROs) are among the elements excluded from the set proposed by the Working Group. As the progression and underlying mechanisms of rare disease are often still poorly understood, development of treatments, understanding of meaningful clinical outcomes, and assessment of effectiveness of therapies is hampered. In the absence of thorough understanding, PROs provide researchers with alternative datapoints to measure the effectiveness of disease management strategies and treatments from patients' perspectives (Gliklich et al., 2014). The inclusion of PROs by extension empowers rare disease patients in articulating their needs and experiences. Omitting PROs from the core data elements for rare disease registries might therefore have implications for the social position of patients in research. The current study aims to investigate how the effort to integrate this research infrastructure might affect the social orderings commonly found in rare disease research, as well as its potential implications on research outcomes.

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