New tool integrates psychological, social and medical data of patients with rare diseases

November 18 2020

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Researchers from the Universitat Oberta de Catalunya (UOC) and the technology center Eurecat have developed an innovative formal representation of rare disease data, including information unavailable in

current models on rare disease patients' biological, psychological and social profile. For their research, the researchers have obtained data on 25 patients from organizations such as Eurordis, the Spanish Rare Diseases Federation (FEDER) and the Rare Diseases Patients' Association of Iran with the goal of including testimonials from different territories with different health systems.

The term used to refer to formal representations of knowledge that establish the different concepts of a specific field and the relationships between them is ontology. In such representations, it is important to use an open-source data format and international standards in order to ensure that this representation is accessible in all spheres. The ontology performed by the UOC uses an open source code and is based on standards defined by the World Health Organization (WHO).

A tool for understanding patients that goes beyond treatments

The research is described in the article "Biomedical Holistic Ontology for Patients With Rare Diseases", published in the *International Journal of Environmental Research and Public Health*. Lead researcher Laia Subirats explained that its value lies in "the fact that a single ontology integrates not just medical information but also information about other aspects that affect patients' lives, such as environmental, geographical and psychological factors, their social relations and their interests. It also includes information taken from Twitter, which gives us social data.

The end result is an improved understanding of the patient and access to new data about the patient's interaction with the disease. Viewed in this light, we can say that it is a holistic ontology". Subirats is a course instructor at the UOC's Faculty of Computer Science, Multimedia and Telecommunications, a collaborator with the University's Applied Data

Science Lab (ADaS Lab) and a researcher at Eurecat. In 2018, she was included in the list of the 100 most influential women in Spain, in the Top 100 Female Leaders awards.

One of the shortcomings of the ontologies used for rare diseases is that they provide information about the patients separately, without including all the necessary information in a single tool. The ontology developed by the UOC researchers overcomes this shortcoming and provides a useful tool for health professionals, patients and other healthcare stakeholders.

As one of the other members of this research team, Jordi Conesa, who also teaches at the Faculty of Computer Science, Multimedia and Telecommunications and is a researcher with the UOC's SmartLearn group, explained: "We know that medical and genetic information ontologies must be integrated in order to perform personalized translational analyses for each patient. However, we have taken this a step further, as we believe that it is necessary to create holistic ontologies that also contain data about other significant factors affecting patients, including psychosocial, demographic, geographical, and time- and social relations-related information. Health—and the available treatments—is not just something that exists within the confines of our bodies; it is also influenced by the environment. With this ontology, we can now include the environment, and also other factors that impact on the quality and intensity of potential treatments."

Psychologist and researcher Manuel Armayones has also taken part in this research. A member of the UOC's eHealth Center, the Psychology, Health and Network (PSiNET) research group and the Faculty of Psychology and Education Sciences, Armayones considered that it was "very significant that data from patients' associations have been taken into account, as they are usually the health system's least-used resource. If there is one field where data from all possible sources should be considered, it is the rare diseases and, especially, the ultrarare diseases".

More information: Laia Subirats et al, Biomedical Holistic Ontology for People with Rare Diseases, *International Journal of Environmental Research and Public Health* (2020). DOI: 10.3390/ijerph17176038