

Genetic testing evaluation could help public health practice

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Credit: NicoElNino,

With their growing use in recent years, genetic tests have received a lot of attention. A new study explored how they are evaluated.

The role of [genetic testing](#) in medicine is expanding thanks to its benefits in the prevention, management and treatment of various diseases. Given their rapid development, the assessment of genetic tests' performance is even more crucial for clinical and public health practice.

A team of researchers supported by the EU-funded PRECEDI project has performed a systematic review of the literature on the evaluation of genetic tests. The study was presented in the European Journal of *Human Genetics*. It focused on 29 evaluation frameworks from various countries published between 2000 and 2017. The majority of these were based on

the ACCE Framework, the Health Technology Assessment (HTA) process, or both.

ACCE takes its name from the four main criteria for evaluating a genetic [test](#): analytic validity, clinical validity, clinical utility, and associated ethical, legal and social implications. HTA involves the systematic evaluation of the properties and effects of a health technology, addressing its direct and intended effects, as well as consequences. It's conducted by interdisciplinary groups that use explicit analytical frameworks drawing on a variety of methods.

Other frameworks analysed by the study refer to the Wilson and Jungner screening criteria, or to a mixture of pre-existing frameworks that are not necessarily specific for genetic tests. Such screening criteria for disease are aimed at guiding the selection of conditions that would be suitable for screening. These are based, among other factors, on the capacity to detect the condition at an early stage and the availability of an acceptable treatment.

Need for a reliable evaluation strategy

The researchers emphasised the importance of a transparent and well-planned evaluation strategy. They argued that such a strategy would avoid the uncontrolled implementation of technologies without proven benefits. This could lead to inappropriate management of patients and detrimental effects on patient health, as well as a waste of resources and loss of public confidence in the medical profession. According to their study, a reliable evaluation strategy would also support the implementation of those currently available tests that have proven effectiveness and cost effectiveness.

The study, which was partially supported by the PRECEDI project, concluded that the ACCE model proves to be a base for the technical

appraisal of genetic tests. "However, this model is not completely satisfying. We suggest the adoption of a broader HTA approach, including the assessment of the context-related evaluation dimensions (delivery models, economic evaluation, and organizational aspects)," the researchers said. They added that such an approach would "maximise population health benefits, facilitate decision-making and address the main challenges of the implementation of genetic tests, particularly in universal health care systems, where economic sustainability is a major issue."

The ongoing PRECEDI (Personalized PREvention of Chronic DIseases) project fosters collaboration among participating institutions in order to provide an innovative research programme and training projects around the concept of personalised prevention of chronic diseases. The PRECEDI consortium is a multidisciplinary group of institutions working on different aspects of personalised medicine, including basic research, economic evaluations, [health](#) service organisation, and ethical, social and policy issues.

More information: Project website: www.precedi.eu/site/

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