PD91 Gaps And Challenges In The Health Technology Assessment Of Genetic And Genomic Applications: A Systematic Review

Leonardo Maria Siena, Antonio Sciurti, Giuseppe Migliara, Claudia Isonne, Giuseppe Di Lorenzo, Maria Roberta De Blasiis, Carolina Marzuillo, Paolo Villari and Valentina Baccolini (valentina.baccolini@uniroma1.it)

Introduction: Evaluating genetic and genomic applications is critical for their adoption in healthcare practice, but this has several challenges, including limited evidence on clinical and non-clinical benefits and rapid development processes. Since these challenges lack comprehensive discussion, this study aimed to identify and analyze all the barriers emerging in the health technology assessment (HTA) of genetic and genomic tests.
Methods: The protocol of this study was registered in the Open Science Framework database. A systematic search of the PubMed, Scopus, and Web of Science bibliographic databases was undertaken to identify all studies that specifically discussed any difficulty, gap, or barrier in the HTA evaluation of genetic or genomic technologies. No restrictions were imposed on HTA domain, study type, or publication date. Gaps and challenges were then grouped into domains outlined in the EUnetHTA HTA Core Model®. A narrative synthesis of the results was performed for each HTA domain.
Results: Of the 457 publications identified, 23 were included. Article designs exhibited considerable diversity, ranging from systematic reviews to semi-structured interviews, and stakeholder involvement varied from universities to national research groups. The challenges we found referred to the general HTA framework or, in most cases, to specific domains. More than 25 percent of the challenges were associated with economic aspects, with cost-effectiveness analysis being the focal point of debate. In addition, clinical effectiveness, in terms of non-health outcomes, lack of evidence, and clinical utility, was also frequently discussed, accounting for 23 percent of the challenges identified.
Conclusions: Our study systematically summarized gaps and barriers in the HTA of genetic and genomic applications, providing a thorough analysis and categorization of these issues. Various challenges surfaced across different domains, notably related to costs, economic evaluation, and clinical effectiveness. There is a need for exhaustive discussions on potential solutions to facilitate and enhance the assessment process for these genetic and genomic applications.

PD92 Health Technology Assessment Of Genetic And Genomic Applications: A Proposal For A Standardized Approach

Antonio Sciurti (antonio.sciurti@uniroma1.it), Giuseppe Migliara, Valentina Baccolini, Erika Pitini, Anna Ewa Kaminska, Valentina Soccodato, Ilaria Mussetto, Carolina Marzuillo and Paolo Villari

Introduction: Frameworks based on the ACCE model, which have a limited health technology assessment (HTA) approach, have long been used in the assessment of genetic and genomic applications (GGAs). While ACCE frameworks are mainly focused on technical aspects, HTA assessments include economic and organizational aspects. The aim of this study was to develop a framework able to address a comprehensive assessment of GGAs.
Methods: We analyzed the most currently used HTA model in Europe, the EUnetHTA HTA Core Model®, to investigate its suitability as an HTA framework for GGAs and to identify any possible shortcomings in its evaluation. The Sapienza evaluation framework, which is based on a previous systematic review of assessment frameworks for GGAs, was used as the comparison. The HTA Core Model applications were then used as a basis for an assessment framework for GGAs, classified according to "GGA function/use setting" pairs.
Results: The domains included in the Sapienza framework fully corresponded with the HTA Core Model. In addition, the "Screening Technology" application of the HTA Core Model was found to be suitable for diagnostic, pre-symptomatic, predictive, and carrier GGAs when used in screening settings. The "Diagnostic Technologies" application was deemed appropriate for the pairs "Diagnostic/Diagnosis", "Pre-symptomatic/Diagnosis", "Prognostic/Staging", and "Pharmacogenetic/Staging", although it may require additional considerations such as the need for screening in the case of germline mutations, the use of appropriate accuracy measures for prognostic GGAs, and the evaluation of the clinical utility of pharmacogenetic or prognostic GGAs.
Conclusions: The proposed approach is a flexible tool that allows assessment of GGAs using a shared and validated methodology that considers the technical aspect, clinical utility, economic value, and delivery models. We found that the HTA Core Model fits the needs of GGA assessment, although some GGA peculiarities should be further explored in the assessment process.

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