How should genetic tests be evaluated? Final Results of a systematic review of the existing tools
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Background
Given the increasingly rapid development of genetic tests, the assessment of their actual benefits is crucial for clinical and public health practice. For this purpose, different evaluation models have been developed. Our aim was to identify and compare in a systematic manner the existing evaluation models for genetic tests, considering their methodology and evaluation criteria.

Methods
We performed a systematic review of the literature through PUBMED, SCOPUS, ISI Web of Knowledge, Google Scholar, Google and grey literature sources including any document describing models for evaluating genetic tests such as research articles, congress abstract, documents of government agencies and research organizations. A Delphi survey involving Italian experts in Public Health Genomics has been performed to reach consensus on data extraction.

Results
We identified 26 models dated between 2000 and 2015 (USA n.10, Canada n.4, Europe n.9, Australia n.1, International n.2), mostly based on the ACCE model (n.12 models), on the HTA model (n.6) or both (n.2). The other ones refer to Wilson and Junger screening criteria (n.2) or to a mix of different criteria (n.4). While 14 tools address all types of genetic test, the other 12 address a specific type of genetic test (i.e. screening, presymptomatic, susceptibility, pharmacogenetic). Most used evaluation criteria are analytic and clinical validity, clinical utility and ethical, legal and social implications. The economic dimension is always considered even if in little detail. Attention for delivery models, organizational aspects, consumer’s point of view is often lacking and only few models highlight research priorities or criteria to recommend the use of the test.

Conclusions
These results unearth the lack of a standardized, shared and complete process for the evaluation of genetic tests and the need to develop an unifying proposal, based on the strengths and weaknesses of the retrieved models.

Key messages:
- This systematic review identified three main tools for the evaluation of genetic tests: the ACCE model, the HTA process and the Wilson and Junger population screening principles
- There is the need to develop a unifying proposal of a complete and innovative process to evaluate genetic tests, relevant to the different national and local contexts