VP103 Health Technology Assessment Of Genetic Tests For Cystic Fibrosis Carrier Screening In Italy

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INTRODUCTION:

Cystic Fibrosis (CF) is a genetic disorder caused by mutations in CFTR gene. In Italy, reported prevalence is approximately .70 *per* 10,000 inhabitants (1). The practice and recommendations for Cystic Fibrosis carrier screening are very heterogeneous in Europe. A proposal of a carrier genetic test in the general population raises many questions. Health Technology Assessment (HTA) could offer a sound methodological basis for this evaluation. The aim of this work was to summarize the available evidence, using the HTA approach, on the genetic tests for Cystic Fibrosis carrier screening.

METHODS:

A systematic literature search was used to find the best available international and national evidence on genetics test for CF carrier screening. In this report, we specifically addressed the health problem of disease, description and technical characteristics of tests – its analytic and clinical validity, and clinical utility. Economic evaluation of different scenarios was synthesized from the literature. Ethical, organizational, and social aspects of CF and genetic screening were also considered.

RESULTS:

Several screening strategies have been evaluated in the literature and screening options can be characterized by different timing, model and place of screening (2). The reported cost of a screening test ranged from EUR25 to EUR212 (3). Estimated life time cost of care for CF patients ranged from EUR291,048 to EUR1,105,452. Ethical analysis emphasized that the use of these tests is an advantage in terms of the acquisition of knowledge and of responsible management of choices, but at the same time raises many ethical questions. Social considerations reported among patients and their families an overall positive attitude toward population CF carrier screening.

CONCLUSIONS:

The advances in the molecular genetics technology have made CF carrier testing reliable and affordable. The multidisciplinary approach of this HTA provided an evidence-based evaluation of genetic tests and offers a firm scientific background for the decision-makers to consider the implementation of a screening for Cystic Fibrosis carriers into the Italian health care system.

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VP107 Pharmaceutical Industry's Experiences with the German Health Technology Assessment Scientific Advice

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