

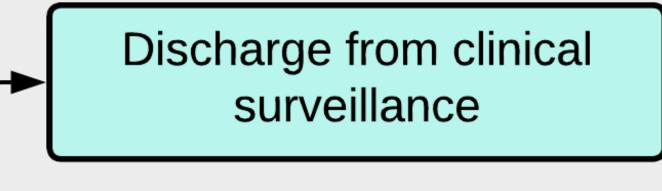
COST-EFFECTIVENESS ANALYSIS OF GENETIC SCREENING IN FIRST-DEGREE RELATIVE OF HYPERTROPHIC CARDIOMYOPATHY PATIENTS FROM THE PERSPECTIVE OF THE BRAZILIAN UNITED HEALTH SYSTEM

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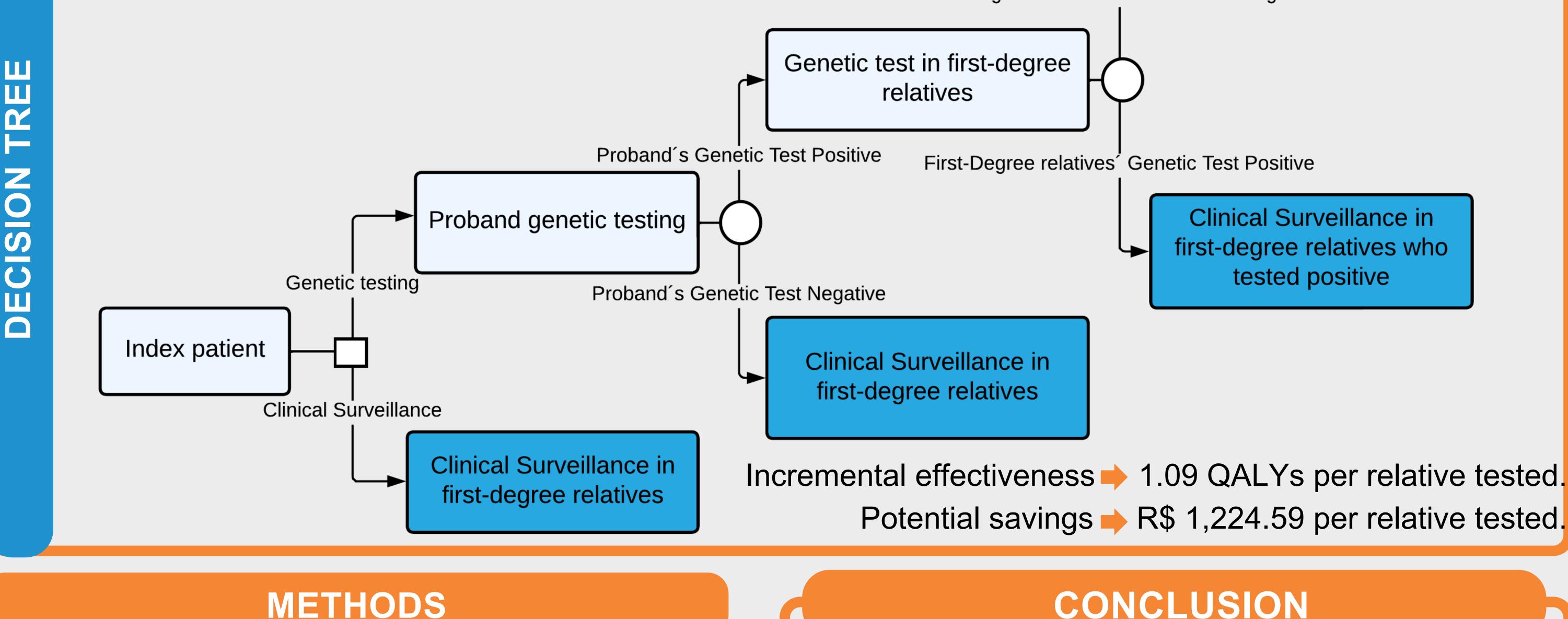
BACKGROUND

Hypertrophic cardiomyopathy (HCM) has an autosomal dominant pattern and it is the most frequent genetic cardiac muscle disease. Genetic testing for HCM is not available in Brazil, therefore all first-degree relatives of HCM patients should be periodically evaluated to detect early sings of the disease.





First-Degree relatives Genetic Test Negative



A decision tree combined with a Markov model was developed to compare lifetime costs and quality-adjusted life years (QALYs) for the two strategies. Transition probabilities were obtained from literature.

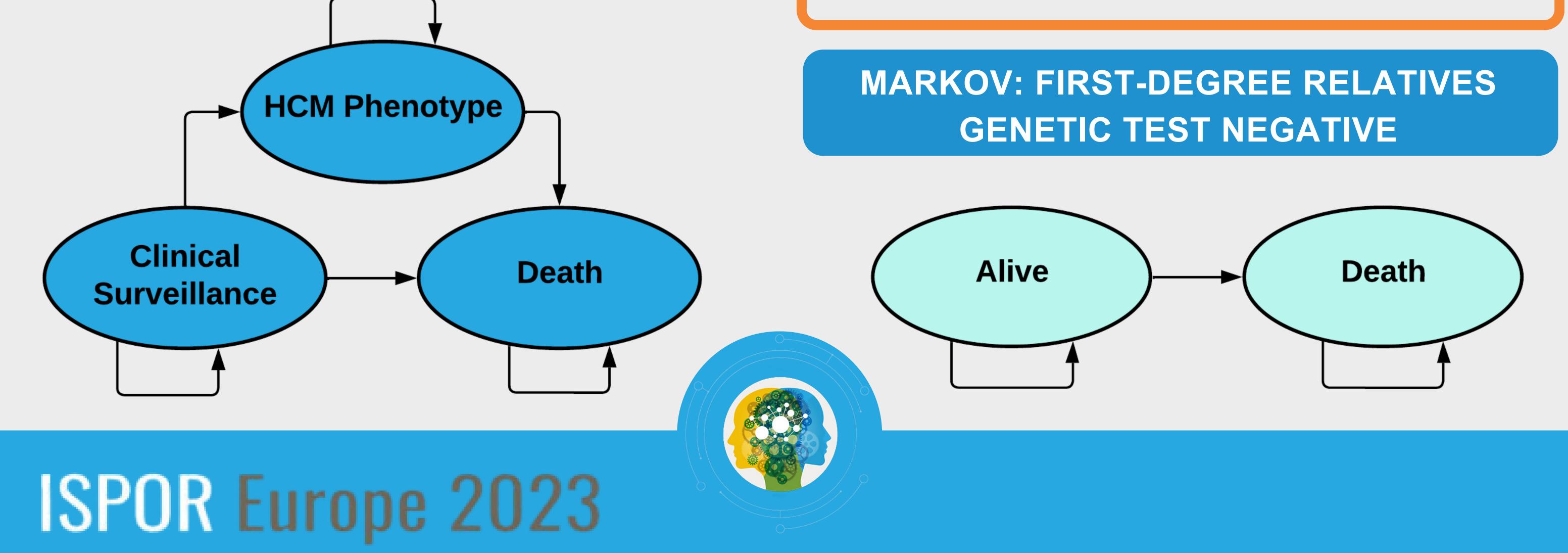
Costs included genetic testing for the index patient and first-degree relatives and clinical surveillance of first-degree relatives.

Effectiveness values were obtained through a rapid literature review that result in utilities values range from 0.72 for HCM patients to 0,87 for negative genotype individuals.

RESULTS

Genetic screening was considered a dominant strategy. It is more effective than clinical surveillance and it is also less costly.

MARKOV: CLINICAL SURVEILLANCE



The genetic screening strategy allows the selection of individuals who must be routinely monitored for early signs of the disease. This results in economic benefits since negative genotype relatives would avoid unnecessary tests. Genetic status is important for career and family

planning and contributes for improved quality of life, especially for those to whom HCM is ruled out.