BACKGROUND

- Family members of patients with established inherited cardiopathies may be carriers of the causative mutation and be at risk of sudden cardiac death (SCD).
- Genetic testing could prevent SCD in asymptomatic first-degree relatives of patients with established inherited cardiopathies.

OBJECTIVES

- The objective is to estimate the cost-effectiveness of conducting genetic testing in first-degree relatives of patients with: 1. Hypertrophic Cardiomyopathy (HCM); 2. Arrhythmogenic Right Ventricular Cardiomyopathy (ARVC); 3. Long-QT Syndrome (LQTS); 4. Brugada Syndrome (BrS); 5. Catecholaminergic Polymorphic Ventricular Tachycardia (CPVT) in Spain.

METHODS

- A Markov model was developed to determine the costs per life-year gained (LYG) and per symptomatic-free years (SFY) gained from conducting genetic testing in first-degree relatives at risk of SCD due to gene-related cardiopathies compared to usual clinical practice (without genetic testing).
- The target population was defined as hypothetical cohorts of 1,000 patients (a cohort per cardiopathy) followed over their lifetime.
- Four health states were defined as follows: 1. Asymptomatic; 2. Minor events (palpitations, dizziness, fatigue, chest pain, dyspnea); 3. Major events (syncope, aborted SCD); and 4. Death (Figure 1).
- The analysis was conducted from the Spanish National Health System (NHS) perspective. Only direct costs were taken into account. Future costs and effects were discounted at a 3% rate per year. All costs referred to €2012.

RESULTS

- For mean cost per patient when the genetic test was conducted compared to usual practice was €51,374 vs. €72,611 for HCM, €58,454 vs. €80,337 for ARVC, €20,575 vs. €21,659 for LQTS, €38,605 vs. €60,807 for BrS, and 28,286 vs. €37,519 for CPTV, respectively. Figure 3 illustrates the mean cost per patient for both comparators and the difference in costs for each gene-related cardiopathy.

- For LQTS and CPTV, genetic testing implied 0.96 and 0.04 SFY increase, and 0.01 and 0.04 LYG, respectively, per patient compared to clinical practice. These variables remained unchanged for HCM, ARVC and BrS.
- Genetic testing was more effective and less costly (superior) for LQTS and CPTV. For HCM, ARVC and BrS it was almost equally effective and less costly (dominant) than usual practice (Table 4).

- Probabilistic sensitivity analyses confirmed the consistency of results. Scatter plot diagrams for LQTS (Figure 4 and CPTV (Figure 5) show that genetic testing could provide better clinical results at lower costs than current clinical practice with no genetic testing.

REFERENCES