

Sudden cardiac death (SCD) or sudden unexplained death (SUD) is a leading cause of mortality with an annual incidence of one death per 1000 person-years, affecting all ages. SCD in persons older than 45 years is mainly due to atherosclerotic coronary artery disease. However, in the young (<45 years), inherited cardiac causes are confirmed or strongly suspected in up to 80% of families. The four most common categories in the young include: premature atherosclerosis (eg familial hypercholesterolemia), cardiomyopathies (eg hypertrophic cardiomyopathy), primary electrical disease (eg long- QT syndrome) and aortic aneurysm/dissection. In the majority of these diseases, the inheritance pattern is autosomal dominant, which implies that all first-degree relatives of a young SCD patient have a 50% a priori chance of suffering from the same disease. The current available cardiogenetic testing procedures in Flanders are extremely slow (months to years) and incomplete (only a few genes). Alternatives abroad are not affordable (several thousands of Euros) and not reimbursable for the patient. The main objective of this study is therefore to develop and to implement cardiogenetic testing panels in the clinical care of patients and family members with risk for SCD due to thoracic aneurysmal disorders and primary electrical disease. Through a unique collaboration between the Cardiology Department, the Department of Medical Genetics and BIOMINA of the Antwerp University Hospital/University of Antwerp, we envision to innovate the current cardiogenetic testing procedures, to shorten significantly the turn-around-time of cardiogenetic testing, to increase the informativeness of testing, to reduce the overall cost of cardiogenetic testing, to optimize the quality of the genetic counseling and to minimize testing-related anxiety levels. In order to reach these objectives, we will combine existing molecular technologies, validate and implement the protocols in clinical care and evaluate key aspects of the cardiogenetic counseling process.