

Abstract: Genetic testing for cardiac disease (Keynote lecture)

In the last 25 years, the genetic causes of cardiac disease has been unravelled on a large scale and our understanding of these disease entities has improved significantly. This has led to the identification of many presymptomatic individuals who can be offered timely treatment to protect them from the deleterious sequelae of these diseases, among which Sudden Cardiac Death. In addition, risk stratification has improved and new gene based therapeutic management strategies have emerged successfully.

In a recent consensus statement document,* endorsed by all major cardiac EP societies, including the Asian-Pacific Heart Rhythm Society, the state of genetic testing for inherited arrhythmia syndromes, cardiomyopathies, sudden cardiac death (SCD), congenital heart disease, coronary artery disease, and heart failure are addressed. For each of the individual syndromes the diagnostic, prognostic, and therapeutic implications of genetic testing, as far as these are known, are discussed. In addition, the document presents the essential principles of genetic testing including modes of inheritance, different testing methodologies, and interpretation of variants.

It is the hope of the writing committee that with this document the appropriate genetic test is offered to the appropriate patient, minimizing the downside of (extensive) genetic testing and thereby optimizing the benefits of genetic testing.

*: Wilde AAM, Semsarian C, Márquez MF, Sepehri Shamloo A, Ackerman MJ, Ashley EA, Sternick EB, Barajas-Martinez H, Behr ER, Bezzina CR, Breckpot J, Charron P, Chockalingam P, Crotti L, Gollob MH, Lubitz S, Makita N, Ohno S, Ortiz-Genga M, Sacilotto L, Schulze-Bahr E, Shimizu W, Sotoodehnia N, Tadros R, Ware JS, Winlaw DS, Kaufman ES; ESC Scientific Document Group. European Heart Rhythm Association (EHRA)/Heart Rhythm Society (HRS)/Asia Pacific Heart Rhythm Society (APHRS)/Latin American Heart Rhythm Society (LAHRS) Expert Consensus Statement on the state of genetic testing for cardiac diseases. *Europace*. 2022 Apr 4:euac030. Online ahead of print. PMID: 35373836