

Implementation of a Phenotype-Oriented Cardiogenetic Panel for Cardiomyopathies and Arrhythmias in Clinical Practice

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Inherited cardiomyopathies and primary arrhythmia syndromes represent a major cause of heart failure and sudden cardiac death across all age groups. While next-generation sequencing enables the parallel analysis of large gene sets, broad testing strategies may generate findings beyond the primary clinical question and complicate interpretation. Therefore, the implementation of focused, phenotype-oriented cardiogenetic testing strategies is of increasing importance in routine care.

This presentation describes the introduction of a cardiogenetic panel based on a phenotype-driven and clinically curated gene selection for cardiomyopathies and arrhythmias in a tertiary care setting. The panel strategy emphasizes genes with well-established relevance for cardiac phenotypes, aiming to maximize clinical validity and diagnostic yield while minimizing incidental findings. Key aspects include panel design, analytical performance requirements, and integration into structured clinical workflows. Practical challenges encountered during implementation, such as variant interpretation, interdisciplinary communication, and embedding genetic results into existing care pathways, are discussed. The presentation demonstrates how a focused cardiogenetic panel can effectively support personalised cardiovascular medicine.