

8<sup>th</sup> Global

# Cardiologists & Echocardiography Annual Meeting

July 18-20, 2016 Berlin, Germany

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## Next generation molecular diagnosis for inherited cardiovascular diseases

The inherited cardiovascular diseases (cardiomyopathies, channelopathies and inherited vascular diseases) are a heterogeneous conjunct of primary diseases usually of genetic origin and familial presentation, which are associated with sudden death risk. The identification of multiple genetic causes for these diseases has opened a new window for their early diagnosis, the understanding of their natural history, and for the improvement in their risk stratification and management. However, in the last years, the clinical application of genetics has been limited by the cost and low yield of the available genotyping technologies. The irruption of Next Generation Sequencing, has completely changed this scenario. This group of disruptive technologies allow the evaluation in parallel of hundreds or even thousands of genes at an affordable cost. Now the challenge is not the genotyping, but the interpretation of the complex results.

In our presentation we review the main aspects related to the application and impact of Next Generation Sequencing in the study of the inherited cardiovascular diseases, with a special focus in the clinical validation and the interpretation of the results.

The successful application of NGS in the clinical diagnosis and management of inherited cardiovascular diseases requires the adequate integration of genotyping technology, bioinformatics and clinical interpretation supported by knowledge management systems. We will present in this session:

- Results and validation of NGS technologies in inherited cardiovascular diseases associated with sudden death
- Our advanced and innovative multidisciplinary approach for the clinical interpretation of the results
- Examples of the usefulness of NGS in the evaluation of inherited cardiovascular diseases

## Biography

Lorenzo Monserrat is the CEO, Health in Code, A Coruña, Spain from 2012 – Present. He is also the Co-Founder and Scientific Director, Health in Code 2006 – 2011. He is doing his research in Galician Health Service, Spain. He is a Cardiology Consultant, A Coruña University Hospital, Spain. He has done his PhD from European Doctorate in Medicine, University of A Coruña, Spain and MD, in Medicine and Surgery, University of Santiago de Compostela, Spain.

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