Master in Electrophysiology With Priming in Cardiac MRI and Interpretation of Genetic Testing in Patients With Inherited Cardiac Diseases

Program in Electrophysiology

Dr Rordorf: 120 hours (15 days) of IN PERSON Electrophysiology Training (Policlinico San Matteo Pavia)

Dr Mantica: 120 hours (15 days) of IN PERSON Electrophysiology Training (Clinica Sant'Ambrogio: Milano)

Program in Cardiovascular Imaging (48 ore in person @ ICS Maugeri, IRCCS Pavia)

Dr Lorenzo Monti: Training IN PERSON in MRI (acquisizione e interpretazione dati) frequenza Martedi 8 ore e Giovedi 8 ore (2 masterizzandi a settimana x 3 settimane **totale 48 ore**)

Introduction

CT and MRI play an increasingly important role in cardiac electrophysiology, primarily in preprocedural planning of ablation procedures but also in procedural guidance and postprocedural follow-up.

The most common applications include ablation for atrial fibrillation (AF), ablation for ventricular tachycardia (VT), and for planning cardiac resynchronization therapy (CRT).

For **AF ablation**, preprocedural evaluation includes anatomic evaluation and planning using CT or MRI.

Procedural guidance during AF ablation is achieved by fusing anatomic data from CT or MRI with electroanatomic mapping to guide the procedure.

Postprocedural imaging with CT following AF ablation is commonly used to evaluate for complications such as pulmonary vein stenosis and atrioesophageal fistula.

For **VT ablation**, both MRI and sometimes CT are used to identify scar, representing the arrhythmogenic substrate targeted for ablation, and to plan the optimal approach for ablation. CT or MR images may be fused with electroanatomic maps for intraprocedural guidance during VT ablation and may also be used to assess for complications following ablation.

Finally, structural and functional information from MRI may be used to identify patients who may benefit from CRT, and cardiac vein mapping with CT or MRI may assist in planning access.

MASTER program The master's degree in electrophysiology will guarantee direct access to the cardiac MRI diagnostic rooms, to ensure direct experience of patient preparation, image acquisition and post processing, experiencing first-hand the possibilities and limits of imaging with CMR.

1- Basis of CT image acquisition and Patient preparation

2- Role of Computed Tomography in Cardiac Electrophysiology
   pulmonary venous anatomy
   left atrial appendage evaluation and thrombi exclusion
   relevant anatomic variants

imaging of complications

CMR

CARDIOMYOPATHIES

1- Role of cardiac imaging in dilated phenotype
2- Role of cardiac imaging in hypertrophic phenotype (HCM, Fabry, Amyloidosis)
3- Role of cardiac imaging in heart failure with preserved ejection fraction
4- Imaging and devices: from ICD to LVAD and total artificial heart

ADVANCED CARDIAC IMAGING FOR ELECTROPHYSIOLOGY

1- Non-invasive detection of arrhythmic substrate
2- Is non-invasive left atrium imaging for EP procedure still mandatory after DECAFF II trial?
3- Radiotherapy for refractory VT ablation: which role for CT?
4- Substrate of cardiogenetic disease not detectable at imaging

Program in Cardiovascular Genetics: Interpretation of results (48 ore in person @ ICS Maugeri, IRCCS Pavia)

Introduction

Genetic testing has become a vital tool in cardiology, especially in electrophysiology, for diagnosing, risk stratification, and personalizing the management of inherited arrhythmias and cardiomyopathies. Genotyping is now incorporated into clinical guidelines, influencing decision-making processes for patient care. However, the growing complexity of genetic data, along with advancements in bioinformatics and functional analysis, poses new challenges for accurate interpretation. The oligogenic nature of many cardiac diseases and the influence of genetic modifiers on phenotypic variability are emerging areas that will further affect how genetic information is utilized in clinical practice.

One major limitation is that genetic laboratories operate without direct access to clinical data, which restricts their ability to interpret findings in a meaningful clinical context. Therefore, it is crucial for electrophysiologists and cardiologists to develop a solid understanding of genetic testing, equipping themselves with the skills necessary to maximize its application for personalized patient care.

Program Objectives:

This training module aims to provide participants with:

- A rational approach to integrating genetic testing into clinical practice.
- Strategies for selecting the appropriate genetic test based on phenotype assessment.
- Skills in interpreting genetic variants using bioinformatics tools and clinical data.
- Best practices for communicating results to patients and guiding family screening.
- Understanding of the genotype-phenotype relationship and its implications for risk stratification and treatment.

Program Structure and Key Topics

1. Selecting the Optimal Genetic Testing Approach

- When and why genetic testing should be performed in inherited arrhythmias and cardiomyopathies.
- Clinical indications and guideline-driven recommendations.
- Phenotype-driven genetic testing: prioritizing candidate genes (and gene panels) based on clinical presentation.
- Targeted panel testing *vs*. whole-exome sequencing (WES): strengths and limitations.

2. Interpretation of Genotyping Data from a Laboratory Perspective

- Overview of genetic testing methodologies: DNA extraction, Sanger sequencing, NGS, CNV analysis.
- Variant classification based on ACMG guidelines.
- Use of bioinformatics tools, databases (ClinVar, gnomAD, Franklin, etc.) and predictive algorithms (PolyPhen, SIFT, etc.).
- Frequency analysis and ethnicity-specific considerations in variant interpretation.

3. Clinical Interpretation of Genotyping Data

- Understanding penetrance and genetic modifiers in inherited cardiac disorders.
- How genotype influences arrhythmic risk and therapeutic decisions.
- The emerging role of polygenic risk scores in predicting phenotype severity.

4. Writing a Genetic Report and Genetic Counseling Principles

- How to structure a clear and clinically useful genetic report.
- Communicating results in a concise and actionable format.
- How to explain uncertain results (VUS) to patients and relatives.
- Ethical considerations and psychological impact of genetic findings.

5. Linking Molecular Substrate with Clinical Phenotype and Family Management

- Screening recommendations for family members based on genetic findings.
- Identifying genotype-positive, phenotype-negative individuals and long-term follow-up strategies.

Teaching Methodology

Interactive lectures and clinical case discussions.
Hands-on practice for variant interpretation using real genetic datasets.
Real-world genetic counseling for pre-test and post-test scenarios.