II European Symposium on

Genetic and Rare Cardiovascular Diseases 2023

The patient's clinical pathway in inherited and rare disease: a journey toward precision medicine

Scientific Directors: Perry M. Elliott, Giuseppe Limongelli

NAPLES | 5TH-7TH DECEMBER 2023

Hotel Royal Continental

FACULTY

Enrico Ammirati
Aris Anastasakis
Elena Arbelo
Eloisa Arbustini
Marcello Arca
Camillo Autore
Maurizio Averna
Cristina Basso
Barbara Bauce
Simona Bellagambi
Elena Biagini
Ruth Biller
Eduardo Bossone
Alida Caforio
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Alessandro Della Corte
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Perry M. Elliott
Michele Emdin
Arturo Evangelista
Silvia Favilli
Giulia Frisso
Alba María García García
Juan Ramòn Gimeno
Betti Giusti
Paolo Golino
Francesca Graziani
Massimo Imazio
Ciro Indolfi
Juan Pablo Kaski
Sabine Klaassen
Stelios Kympouropoulos
Marco Lacarra
Giuseppe Limongelli
Bart Loeys
Francesco Loffredo
Maria Angela Losi
Viviana Maestrini
Andrea Mazzanti
Marco Merlo
Emanuele Monda
Maria Beatrice Musumeci
Francesco Natale
Gerardo Nigro
Vincenzo Nigro
Gabrielle Norrish
Iacopo Olivotto
Giuseppe Pacileo
Giuseppe Palmiero
Stathis Papatheodorou
Giancarlo Parenti
Leandro Pecchia
Guglielmina Pepe
Pasquale Perrone Filardi
Maurizio Pieroni
Alexander Protonotarios
Joel Rose
Maria Giovanna Russo
Vincenzo Russo
Luca Sangiorgi
Berardo Sarubbi
Maurizio Scarpa
Annalisa Scopinaro
Paolo Siani
Gianfranco Sinagra
Adalena Tsatsopoulou
Karim Wahbi
Arthur Wilde
Cordula Wolf
Alberto Zambon

Scientific Directors

Perry M. Elliott
Giuseppe Limongelli

Scientific Rationale

On behalf of the Scientific Committee it is a great pleasure to welcome you to the city of Naples for the IIth International Meeting on Inherited and Rare Cardiovascular Disorders, 2023.

Inherited and rare cardiovascular diseases comprise a group of more than 50 diseases, including primary arrhythmia disorders, malformation syndromes, cardiomyopathies, connective tissue disorders, congenital heart defects and metabolic diseases. Taken together, these disorders may affect up to 1 in 240 individuals and are a significant burden on healthcare services.

For much of the history of medicine, patients suffering from rare diseases have found themselves to be beyond hope, but in recent years, disease awareness has spread around the world and the advances in molecular genomics have facilitated personalised therapeutic management of patients with rare and ultra-rare disorders according to their specific phenotype.

The importance of rare cardiovascular disorders is reflected by recent efforts of national healthcare agencies to reduce diagnostic delay among patients with rare diseases, through the institution of disease-specific “patient pathways”. Fundamental to this effort is a multidisciplinary and collaborative approach between healthcare agencies, hospitals and healthcare providers.

The aim of this meeting is to improve the education of cardiologists and other specialists in the field of rare and genetic diseases and to highlight recent advances in inherited cardiovascular disease, with a particular focus on new approaches to diagnosis and management.

We are enormously fortunate to have some of the World’s greatest experts in our faculty and sincerely wish you a successful and enjoyable meeting.

DAY ONE • 05 DECEMBER 2023

08.30 Meeting Opening: European, National & Regional Key Figures

Introduction and Greeting from the Authorities

09.50 Introduction to Round Tables • **Perry M. Elliott, Giuseppe Limongelli**

10.00 **Round Table I.** European, National & Regional Rare Disease Landscape. Where are we now? Where are we going?
Chairman: **Perry M. Elliott, Giuseppe Limongelli**

- The role of European parliament • **Stelios Kympouropoulos** (MEP) *(by Zoom)*
- The role of ERN • **Maurizio Scarpa** (MetabERN), **Arthur Wilde** (Guard Heart ERN), **Luca Sangiorgi** (ERN Bone)
- The role of EMA • **Annalisa Capuano** (EMA-AIFA)
- The role of new technologies in rare diseases • **Leandro Pecchia** (President of the European Alliance for Biomedical Engineering and Science (EAMBES))
- The role of patients association • **Simona Bellagambi/Annalisa Scopinaro** (EURORDIS/UNIAMO)

11.30 *Coffee break*

12.00 **Lecture**: Sudden Cardiac Death • **Arthur Wilde**
Introduction: **Cristina Basso**

DAY ONE • 05 DECEMBER 2023

12.30 **Round Table II**. A law for sudden death in Italy
Introduction: **Ciro Indolfi, Vito De Filippo, Pasquale Perrone Filardi, Paolo Siani**

Participants: **Camillo Autore, Cristina Basso, Ruth Biller, Marco Canepa, Franco Cecchi, Marco Lacarra, Giuseppe Limongelli, Annalisa Scopinaro**

13.15 Witness • **Ruth Biller, Franco Cecchi**

13.30 Conclusion & Lunch

INTERNATIONAL CARDIOMYOPATHY NETWORK–LAUNCH OF POLICY MANIFESTO

16.00 Introduction • **Perry M. Elliott**

16.10 Why do we need a strategy? • **Iacopo Olivotto**

16.40 Raising the patient voice • **Joel Rose**

17.10 The need for multidisciplinary Networks • **Aris Anastasakis**

17.40 Bridging the gap between science and clinical cardiology • **Eloisa Arbustini**

18.10 ICoN: The Agenda for Change • **Perry M. Elliott**

18.40 Conclusion

DAY TWO • 06 DECEMBER 2023

08.00 Registration

08.45 Welcome and opening remarks
Chairs: **Perry M. Elliot, Giuseppe Limongelli**

SESSION ONE: HEART MUSCLE DISEASE

NEW GUIDELINES FOR CARDIOMYOPATHIES
Chairs: **Perry M. Elliott, Giuseppe Limongelli**

09.00 How the new Guidelines will change diagnosis and practice in CMPs?
Juan Pablo Kaski

09.20 How the new Guidelines will change the approach towards risk assessment?
Elena Arbelo

09.40 Time for a molecular classification
Eloisa Arbustini

HYPERTROPHIC CARDIOMYOPATHY
Chairs: **Franco Cecchi**

10.00 Risk stratification in HCM: Not just sudden death
Juan Ramòn Gimeno

10.15 Managing LVOTO: from surgery to myosin inhibitors
Iacopo Olivotto

DAY TWO • 06 DECEMBER 2023

10.30 Heart failure: the new frontier in HCM
Elena Biagini

10.45 Discussion Coordinators
Maria Angela Losi, Maria Beatrice Musumeci

11.00 *Coffee break*

DILATED CARDIOMYOPATHY

Chairs: **Philippe Charron, Gianfranco Sinagra**

11.30 Genotype and phenotype in DCM: From one to many diseases
Philippe Charron

11.45 Non dilated hypokinetic cardiomyopathy: putting the name in the context
Marco Merlo

12.00 The future of precision medicine in dilated cardiomyopathy
Perry M. Elliott

12.15 Discussion Coordinator
Antonello D'Andrea, Viviana Maestrini

ARRHYTHMOGENIC RIGHT VENTRICULAR CARDIOMYOPATHY

Chairs: **Aris Anastasakis, Adalena Tsatsopoulou**

12.30 Correlating genotypes with disease: clinical & molecular classification of ARVC
Alexander Protonotarios

DAY TWO • 06 DECEMBER 2023

12.45 The impact of multimodality imaging on the diagnosis of ACM
Barbara Bauce

13.00 Advances in risk assessment and the management of ventricul arrhythmias
Andrea Mazzanti

13.15 Discussion Coordinator
Gerardo Nigro, Berardo Sarubbi

13.30 *Lunch*

MYOCARDITIS AND INFLAMMATORY DISEASES

Chairs: **Aris Anastasakis, Massimo Imazio** *(by Zoom)*

14.30 When to perform genetic testing in myocarditis?
Sabine Klaassen

14.45 When to perform endomyocardial biopsy in myocarditis?
Alida Caforio

15.00 Diagnosis and management of cardiac sarcoidosis
Enrico Ammirati

15.15 Discussion Coordinator
Cristina Chimenti, Maurizio Pieroni

STORAGE, INFILTRATIVE, NEUROMUSCULAR DISORDERS

Chairs: **Eloisa Arbustini, Michele Emdin** *(by Zoom)*

15.30 Cardiac Amyloidosis: diagnosis and natural history
Michele Emdin *(by Zoom)*

DAY TWO • 06 DECEMBER 2023

15.45 Fabry Disease: the importance of registries and networks
Maurizio Pieroni

16.00 Neuromuscular Disease and the Heart
Karim Wahbi

16.15 Discussion Coordinator
Francesca Graziani, Vincenzo Russo

16.30 *Coffee break*

PAEDIATRIC HEART FAILURE & CARDIOMYOPATHIES
Chairs: **Silvia Favilli, Maria Giovanna Russo**

17.00 Etiology and clinical presentation in children
Giuseppe Limongelli

17.15 Risk prediction of outcome in children with Cardiomyopathies
Gabrielle Norrish

17.30 Novel therapeutic approaches in childhood heart failure and Cardiomyopathies
Cordula Wolf

17.45 Discussion Coordinators
Michele D'Alto, Giuseppe Pacileo

18.15 Conclusions

DAY THREE • 07 DECEMBER 2023

08.30 Introduction
Giuseppe Limongelli

08.35 Lecture: What is a cardiomyopathy in 2023?
Perry M. Elliot

SESSION TWO: GENE THERAPY

09.00 Video
John Crowley

GENE THERAPY IN RARE DISEASES

Chairs: **Lucie Carrier, Gaetano De Ferrari**

09.15 New Therapeutic approaches for transthyretin cardiac amyloidosis
Francesco Cappelli

09.30 Gene Therapy in muscular dystrophies
Vincenzo Nigro

09.45 Gene Therapy for Ion Channel Disease
Lia Crotti

10.00 New therapeutic approaches for Cardiomyopathies
Sharlene Day *(by Zoom)*

10.15 Discussion Coordinators
Giulia Frisso, Giancarlo Parenti

DAY THREE • 07 DECEMBER 2023

SESSION THREE: FUTURE RESEARCHERS

NEW FRONTIERS IN INHERITED, CONGENITAL AND RARE DISEASE RESEARCH

Chairs: **Giovanni Di Salvo, Francesco Loffredo**

10.30 Murcia Team (Spain)
Alba María García García

10.45 London Team (UK)
Douglas Cannie

11.00 Naples Team (Italy)
Emanuele Monda

11.15 Athens Team (Greece)
Stathis Papatheodorou

11.30 Discussion Coordinators
Francesca Graziani, Giuseppe Palmiero

11.45 *Coffee break*

DAY THREE • 07 DECEMBER 2023

SESSION FOUR: HEART RHYTHM

MOLECULAR AUTOPSY & FAMILY SCREENING IN SUDDEN ADULT DEATH SYNDROME
Chairs: **Gaetano De Ferrari, Giuseppe Limongelli**

12.15 Sudden cardiac death prevention: a public health priority
Ruth Biller

12.30 The emerging role of molecular autopsy
Cristina Basso *(by Zoom)*

12.45 How to identify and manage family members at risk?
Aris Anastasakis

13.00 Discussion Coordinators
Barbara Bauce, Marco Canepa

13.30 *Lunch*

SESSION FIVE: VESSELS

FAMILIAL DYSLIPIDAEMIAS
Chairs: **Paolo Calabrò, Maria Donata Di Taranto**

14.30 Familiar hypercholesterolaemia: diagnosis and management
Maurizio Averna

DAY THREE • 07 DECEMBER 2023

14.45 Familiar dysbetalipoproteinaemia: diagnosis and management
Marcello Arca

15.00 Genetic causes of hypertriglyceridemia: diagnosis and management
Alberto Zambon *(by Zoom)*

15.15 Discussion Coordinators
Giovanni Cimmino, Francesco Natale

15.30 *Coffee Break*

GENETIC AORTOPATHIES

Chairs: **Eduardo Bossone, Marisa De Feo**

16.00 Marfan syndrome and inherited aortopathies
Bart Loeys

16.15 Non-syndromic aortopathies
Arturo Evangelista

16.30 Genetics in bicuspid aortic valve
Alessandro Della Corte

16.45 Discussion Coordinators
Betti Giusti, Guglielmina Pepe

17.15 Conclusions & Final Remarks

Requested Endorsement

REGIONE CAMPANIA

OSPEDALI DEI COLLI
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- Università Studi della Campania Luigi Vanvitelli

Società Italiana di Cardiologia | LA SOCIETÀ DELLE TRE ANIME

European Reference Networks
for rare or low prevalence complex diseases

CENTRO COORDINAMENTO MALATTIE RARE
REGIONE CAMPANIA

International CardiOmyopathy Network | ICON

MALATTIE GENETICHE
E RARE CARDIOVASCOLARI

GENERAL INFORMATIONS

DATE AND VENUE
5th – 7th december 2023
Hotel Royal Continental
Via Partenope, 38
80121 - Naples

REGISTRATION INSTRUCTIONS
Online registration has to be done or simultaneously with the start of the Congress in order to have the possibility to complete CME questionnaires at the end of the course and get credits.

1. Connect to the address: www.rarediseasesymposium.com
2. In case of first time access: create your personal account by clicking on "REGISTRAZIONE". If you have already registered for online courses, you can use the same credentials.
3. Enter the following access key to enroll in the course: **Genetic**

HELPDESK
In case of assistance, please contact helpdesk@summeet.it,

CME ACCREDITATION (Continuing Medical Education) - CODE 604-398574
The CME Provider Summeet Srl (n° 604) is scheduling into its 2023 program the event **"II European Symposium on Rare and genetic Cardiovasculas Diseases - The Patient's clinical pathway in inherited and rare disease: a journey toward precision medicine"** with the aim to assign 11,2 credits. The event is intended for n. 120 Physicians (Category: Cardiology, Endocrinology, Metabolic Diseases and Diabetology, Internal Medicine, General Medicine)

It will be possible to fill in the satisfaction questionnaire within 3 days of the end of the course.

CME PROVIDER ID 604 AND ORGANIZING SECRETARIAT

SUMMEET

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