2022

Activity Report

ESC COUNCIL ON CARDIOVASCULAR GENOMICS

FISCAL YEAR 2022 (APRIL 2021 TO MARCH 2022)

ESC Council Cardiovascular Genomics

Chairperson’s Introduction

Prof. Perry Elliott, FESC

Chairperson of the Council on Cardiovascular Genomics 2019-2022

There is a tide in the affairs of men.
Which, taken at the flood, leads on to fortune.
Omitted, all the voyage of their life
Is bound in shallows and in miseries.
On such a full sea are we now afloat…

Julius Caesar, Act-IV, Scene-III

This often-used Shakespearian quote seems very appropriate as we struggle to cope with the consequences of the pandemic and the unstable international crisis. It is in such times that the qualities of resilience, imagination and determination come to fore, and it is with some pride that I can report on a very successful year for the Council in which we have raised the profile of genomic cardiology through position statements, webinars and the nurturing of a vibrant young community.

This is my final year as chairman of the Council, and I am enormously grateful to have had the privilege of working with the dedicated staff of the ESC and my expert colleagues. I am confident that genomic cardiology is here to stay.

Highlights from 2021-2022

- Membership grew to over 850 members by March 2022
- Two live webinars produced
- First position statement published.
- Issued the first two volumes of Cardiovascular Genomics Insight – online news bulletin

Council on Cardiovascular Genomics Nucleus 2019-2022

Perry Elliott
Chairperson

Heribert Schunkert
Chairperson-Elect

Cornelia van Duijn
Treasurer/Secretary

Stefan Kaab
Communications Officer

Yigal Pinto
Nucleus Member

Pim van der Harst
Liaison Officer

Eloisa Arbustini
Nucleus Member

Lucie Carrier
Nucleus Member

Bart Loeys
Nucleus Member

Elijah Behr
Extraordinary Nucleus Member representing EHRA

Thomas Thum
Extraordinary Nucleus Member representing HFA

Maryam Kavousi
Extraordinary Nucleus Member representing EAPC

Kristina Hermann Haugaa
Extraordinary Nucleus Member representing EACVI

Council on Cardiovascular Genomics Board 2019-2022

The Council on Cardiovascular Genomics Board includes representatives from related ESC Constituent Bodies and European societies:

- ESC Working Group on Aorta and Peripheral Diseases - Guillaume Jondeau
- ESC Working Group on Atherosclerosis & Vascular Biology – Paul Evans
- ESC Working Group on Myocardial & Pericardial Diseases - Antonis Pantazis
- ESC Council on Basic Cardiovascular Science – Johannes Waltenberger
- Association for European Paediatric Cardiology (AEPC) - Juan Kaski
- European Society of Human Genetics (ESHG) – Bart Loeys

Membership

The Council aims to encourage research, education and the sharing of genomic knowledge and to accelerate the translation of genomic discoveries into clinical practice. By March 2022, the Council had attracted over 850 members from multiple disciplines.

Congress

Joint Sessions

In April 2021 the Council was involved in a joint session during ESC Preventive Cardiology Congress 2021:

Genetic testing to assess and manage cardiovascular risk

ESC Congress 2021

The Council also contributed to the scientific programme of ESC Congress 2021 – a digital experience.

Meet the Experts: genetic variants in cardiovascular disease – what clinicians need to know
What does science teach us about life with genetic heart disease?

Education

Scientific Documents

In 2020-2021 the Council published its first position statement.

- Interpretation and actionability of genetic variants in cardiomyopathies: a position statement from the European Society of Cardiology Council on cardiovascular genomics – published in the European Heart Journal

Further scientific documents are under development.

Utility of Genetic Risk Scores for Prediction of Coronary Artery Disease. A position statement of the ESC Council on Cardiovascular Genomics, the ESC Cardiovascular Risk Prediction Unit and the European Association of Preventive Cardiology (EAPC)

Webinars

The Council on Cardiovascular Genomics organised the following webinars in 2021-2022

Date	Title	Presenters	Host	Attendees
February 2022	Preimplantation genetic diagnosis and pregnancy in inherited cardiovascular disease	Kristina Hermann Haugaa Bart Loeys	Perry Elliott	392
March 2022	Polygenic risk scores: what is their value for risk prediction and potential for clinical utility?	Riyaz Patel Michael Inouye Aniruddh Patel	Cornelia van Duijn Heribert Schunkert	269

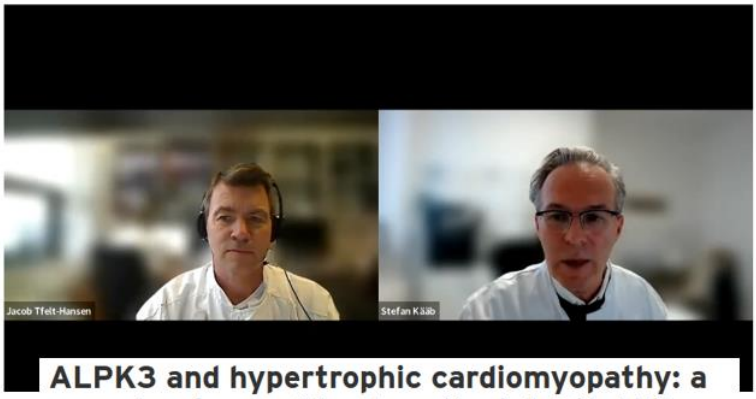
Cardiovascular Genomics Insight

The Council's Media Task Force's have launched Cardiovascular Genomics Insight, a quarterly online bulletin providing useful information on a specific topic.

In 2021-2022 two volumes were released:

Volume	Date	Topic
Volume 1	October 2021	**Cardiomyopathies**
Volume 2	January 2022	**Sudden Cardiac Death in the Young**

Different content types include Clinical Case Presentations, Commented Articles, How-To Articles, quizzes, and guest interviews:



ALPK3 and hypertrophic cardiomyopathy: a new step forward to close the inheritability gap

Prof. Antoine Bondue , FESC

Highlight from Antoine Bondue, Hôpital Erasme & IRIBHM, Université Libre de Bruxelles, Brussels

Hypertrophic cardiomyopathy is the first inherited cardiac condition for which a genetic causality was established in the 80's. The last 40 years brought a tremendous amount of scientific work, to such extent that we are now able to explain around 60% of its inheritability. However, many challenges still remain to closing the gap of its "missed heritability", and to achieve appropriate variant interpretation in regards to broad genetic datasets.[1,2]

How To...Investigate Sudden Cardiac Death and Sudden Cardiac Arrest in the Young

24 Jan 2022

Dr. Francisco Bermudez Jimenez

Introduction

After sudden unexpected death (SUD) in the young, a significant number of individuals present an underlying cardiac disorder, which can be hereditary. These deaths can then be classified as cases of sudden cardiac death (SCD) when the autopsy identifies a cardiac or vascular anomaly as the probable cause of the event. Coronary artery disease is the leading cause of SCD in older persons, whereas in the young (1 to 35 years of age), SCD is more often caused by structural heart disease, including hypertrophic cardiomyopathy (HCM), dilated cardiomyopathy (DCM), arrhythmogenic cardiomyopathy (ACM), myocarditis, and congenital anomalous coronary arteries.

In a considerable number of cases of SCD among children and young adults, a cause of death is not found after a exhaustive autopsy examination including toxicologic and histologic studies. These deaths may be assumed to be sudden arrhythmic death (syndrome), or SAD(S). The most common causes are congenital long-QT syndrome (LQTS), Brugada syndrome (BrS), short-QT syndrome (SQTS), and catecholaminergic polymorphic ventricular tachycardia (CPVT) (Figure 1).