

Activity Report

ESC COUNCIL ON CARDIOVASCULAR GENOMICS
FISCAL YEAR 2021 (APRIL 2020 TO MARCH 2021)

Chairperson's Introduction



Prof. Perry Elliott, FESC

*Chairperson of the Council on
Cardiovascular Genomics
2019-2022*

At the end of a traumatic twelve months fighting with Covid-19, the green shoots of recovery seem to be appearing across Europe and the rest of the World.

While we must remain cautious, it is a time to focus once more on the future and the ESC Council for Cardiovascular Genomics is working hard to fulfil its core mission.

We were delighted to host our first annual congress (Cardio genomics 2021) this year and we have completed our first position statement on the key role of clinicians in genetic variant interpretation.

In coming months, we will be developing new statements that we hope will appeal to different members of our diverse constituency and will seek to forge links with other ESC constituent bodies to promote awareness of the genetic dimension to modern cardiological practice.

In spite of all that we have been through, we are very optimistic about the future and look forward to working with all our members in coming months.

Highlights from 2020-2021

➔ Held its first virtual conference – **Cardio Genomics 2021**

➔ Membership grew to over 650 members by March 2021

➔ First position statement submitted for publication.

➔ Contributed to ESC Congress 2020 with a dedicated session on cardiovascular genomics.

➔ Development of a Media Task Force leading to the creation of the Cardiovascular Genomics Facebook Group

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

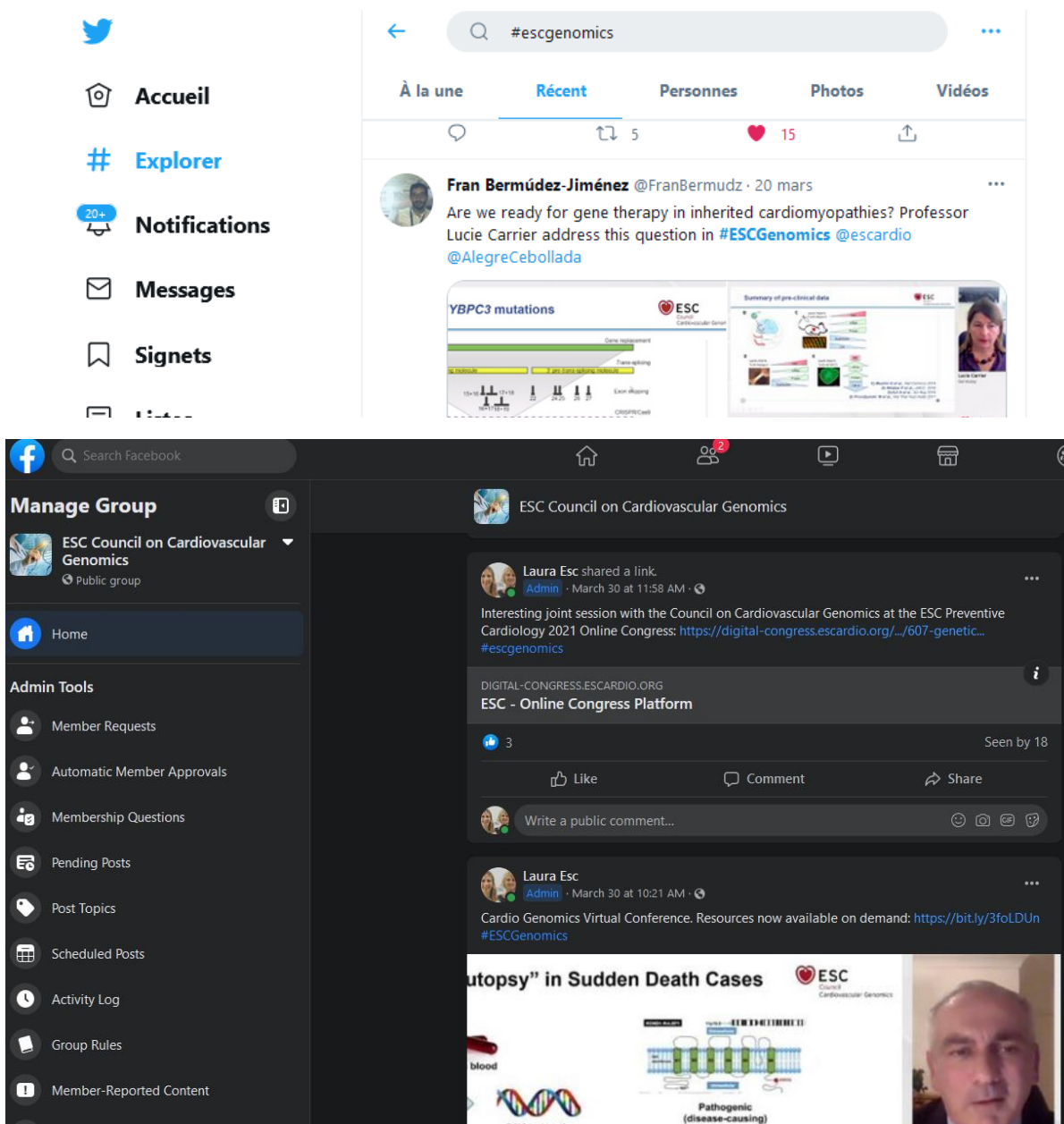
The Council on Cardiovascular Genomics Board includes representatives from related ESC Constituent Bodies and European societies:

- 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

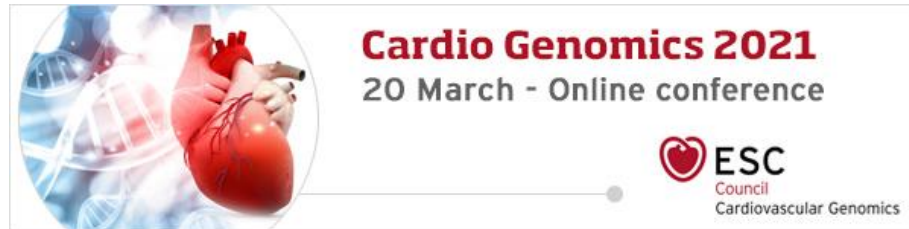
The Council aims to encourage research, education and the sharing of genomic knowledge and to accelerate the translation of genomic discoveries into clinical practice. By March 2021, the Council had attracted over 650 members from multiple disciplines.

As almost 50% of the Council's members are under 40 years of age, the Council has developed its social media strategy with the creation of a Media Task Force. The Task Force members develop the Council's communications and share relevant news via Twitter and through the Council's new Facebook Group.



Congress & Education

In March 2021 the Council held the first edition of its virtual conference: Cardio Genomics.



This was a half-day event attracting over 550 participants and involving 14 faculty members plus abstract presenters.

The scientific programme consisted of recorded presentations plus live discussions with audience Q&A and a live abstract session.

All [scientific resources](#) have been made freely available to Council on Cardiovascular Genomics Members.

“Molecular Autopsy” in Sudden Death Cases

The diagram illustrates the process of a molecular autopsy. It starts with 'Postmortem blood', leading to 'DNA extraction' (represented by a DNA double helix). This is followed by 'Genetic analysis', which includes 'Sanger sequencing', 'Cardiac gene panels', and 'Whole exome / genomes'. A 'Pathogenic (disease-causing) mutation' is identified, which is then linked to a 'Clinical picture' (represented by a heart diagram). The 'Original 4-gene screen' lists: KCNQ1 (LQT1), KCNH2 (LQT2), SCN5A (LQT3, BrS), and RYR2 (CPVT). The process is attributed to 'Semsarian, et al. Eur Heart J, 2015'.

Original 4-gene screen:

- ❖ KCNQ1 (LQT1)
- ❖ KCNH2 (LQT2)
- ❖ SCN5A (LQT3, BrS)
- ❖ RYR2 (CPVT)

Genetic analysis

- Sanger sequencing
- Cardiac gene panels
- Whole exome / genomes

Semsarian, et al. Eur Heart J, 2015

ESC Council Cardiovascular Genomics

Christopher Semsarian
Australia

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Germany

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ESC Congress 2020

The Council contributed to the scientific programme of ESC Congress 2020 – a digital experience, via development of the following session:

[The Future of Personalised Medicine in Cardiomyopathy](#)

Presentations:

- Targeted therapies in hypertrophic cardiomyopathy, P Garcia-Pavia (Madrid, ES)
- Defining arrhythmogenic cardiomyopathy in the genetic era, A Anastasakis (Athens, GR)
- Cardiomyopathy in children: a different disease? S Mital (Toronto, CA)

Scientific Documents

In 2020-2021 the Council has developed the following scientific document:

- Multidisciplinary Approach to the Interpretation and Actionability of Genetic Variants in Cardiomyopathies -> *pending publication*

Further scientific documents are being defined for development during 2021-2022.