European Society of Cardiology
Working Group on
Myocardial & Pericardial Diseases

Newsletter

Issue 1- March 08

Getting Started:
The Message of the Chairman

INSIDE THIS ISSUE

Andre Keren MD,
Hadassah University Hospital, Jerusalem, Israel
Chairman

Dear Friends,

At the last General Assembly of our working group during the ESC meeting in Vienna, we announced principles of enhanced involvement of the members in the decision making process and activities of the working group. Following this meeting, the working group had the following major activities:

The 2007 Annual Meeting of the Working Group in Marburg, organized by Prof Bernhard Maisch, between 11-14.10.2007, entitled "European Conference on Myocardial and Pericardial Diseases with Additional Focus on Heart Disease in Women". This was an outstandingly successful meeting both because of the warm welcome we received from the organizers and the representatives of Marburg University, the exceptionally high scientific level and for the very friendly social aspect of the meeting for which Ms Inge Maisch was directly responsible. I would like to use this opportunity to thank again Prof Maisch, Prof Pankuweit and their collaborators for organizing the 2007 meeting of the Working Group.

The nucleus meeting of 12.10.2007. The discussions we had and the proposals received during the general assembly in Vienna were discussed and approved at the Nucleus meeting. They dealt with the Nucleus membership and election process, proposal for the position of "Associate Nucleus Member", election of Treasurer, rules of organizing future meetings, etc... **See the report** in http://www.escardio.org/bodies/WG/wg21/ (Report section)

The election of new nucleus members in 2007. This was the first time we had elections using the website. This process was successfully coordinated by Celine Serio of the ESC team. We wish to thank Celine and the ESC team for their unrelenting support of our activities. The two new nucleus members elected by the general membership were Dr Tiina Helio form Helsinki, Finland and Dr Arsen Ristic from Belgrade, Serbia. We congratulate the new members of the Nucleus and thank all the members who participated in the electronic election process. We use this opportunity to thank again Prof Petar Seferovic from Serbia and Prof Bert Andersson from Sweden for their support and important contributions to the activities of our Nucleus and Working Group.

"I call for increased involvement and participation of our members in the everyday scientific and educational life of our working group, in development of joint scientific clinical and basic science research projects, active participation in or establishment of new fields on the website, organization of meetings, seminars and teaching courses and promotion of new ideas which you can coordinate as Associate Nucleus Members in the frame of our working group. Please contact me (using my email: andrek@cc.huji.ac.il) or please contact other Nucleus Members responsible for specific activities of the Working Group, as published on our website".

2008 meetings of the Working Group. During 2008 the Working Group will have 2 meetings:

- The first will be a "**Workshop on Inherited Cardiomyopathies" organized by Prof Eloisa Arbustini between April 4-5, 2008 in Camogli, Italy. Only invited experts will participate at this meeting**. This conference will specifically address ways of improving the clinical translation of the great advances achieved in basic molecular and genetic research of cardiomyopathies.

- The second will be **the 2008 Annual Meeting of the Working Group**. **It will be organized by Prof Franco Cecchi in Florence between May 22-24 and will be entitled "Florence International Course on Advances in Cardiomyopathies. The 5th Meeting of the Myocardial and Pericardial Diseases Working Group of the ESC".** Please use the opportunity of enrolling for this important meeting now, using the information provided on our website

Approval by the ESC of the "Affiliate Nucleus Member" position. At the recent meeting of the ESC Board Members with the Chairmen/Vice chairmen of the Working Groups (Nice, March 6, 2008), our proposal of establishing the above position was unanimously supported. This position will be accorded by decision of the Nucleus to members willing to participate or coordinate activities which will be identified as being of particular interest for our Working Group.

Accomplishments as "Affiliate Nucleus Members" will be taken into account during the future short listing process of candidates for the Nucleus and will be also summarized by the Chairman of the WG at the time of presentation of the candidates to the general membership for future elections of Nucleus members

Sincerely,

Andre Keren, MD
Professor of Medicine (Cardiology)

WHO ARE WE ?

OUR WG NUCLEUS MEMBERS:

Chairman: Andre Keren, Israel
Vice-Chairman: Luigi Tavazzi, Italy
Past-Chairman: Uwe Kühl, Germany
Secretary: Perry M. Elliott, United Kingdom
Web Editor: Philippe Charron, France
Treasurer: Claudio Rapezzi, Italy

Zofia T. Bilinska, Poland
Lorenzo Monserrat, Spain
Sabine Pankuweit, Germany
Tiina Helio, Finland
Arsen Ristic, Serbia

The paper of the month: *The Classification Concept of the ESC Working Group of Myocardial and Pericardial Diseases*

By Dr Perry Elliott, The Heart Hospital, University College London, United Kingdom

The term *cardiomyopathy* was first used over forty years ago, to describe myocardial disorders that are not caused by haemodynamic disturbances such as valve disease and hypertension, or by multi-system diseases. Heart muscle disorders with an identifiable aetiology were initially termed specific heart muscle diseases, but were later renamed specific cardiomyopathies. Remarkably, this nomenclature has survived to the present day with only minor changes. Recently, expert committees of the AHA and the ESC Working Group on Myocardial and Pericardial Diseases have proposed updates of the cardiomyopathy classification system [1,2]. The motivation of both groups was to resolve outstanding ambiguities in the existing classification and to incorporate knowledge derived from advances in molecular genetics. Both proposals continue to define cardiomyopathies according to the morphology and physiology of the ventricles and emphasize the importance of genetic disease as a cause of cardiomyopathy. The major differences relate to the handling of primary and secondary disease and the classification of ion channel disorders

Summary

A clinical classification should be simple and relevant to everyday medical practice. The ESC working group's classification achieves this goal, and by highlighting the importance of familial disease, will result in more accurate diagnoses and better outcomes for patients.

The AHA classification

Cardiomyopathies have always been divided into primary and secondary forms. The definition of primary has, however, been somewhat ambiguous, interpreted by many to mean idiopathic and others as disease confined to the heart (i.e. not the consequence of a systemic disorder). The new AHA proposal maintains this taxonomy, defining primary cardiomyopathies as diseases solely or predominantly confined to heart muscle and secondary cardiomyopathies as cardiac disorders with "pathological myocardial involvement" as part of generalized systemic disorders". The first major departure from the existing convention is the sub-classification of primary cardiomyopathies into genetic, mixed (genetic and non-genetic) and acquired forms. The second, and much more controversial change, is the redefinition of ion channelopathies as primary, genetic cardiomyopathies.

The ESC Classification

The ESC working group's proposal abandons the distinction between primary and secondary altogether. The existing morphologic sub-types of cardiomyopathy (hypertrophic, dilated, restrictive, *etc*.) are retained with some modifications, but they are separated into familial (or genetic) and non-familial (or non-genetic) sub-types. These are then further subdivided into known genetic causes, idiopathic and acquired diseases as appropriate. The principle aim of this simplification of the old classification was the encouragement of a shift from the existing exclusion-based diagnostic paradigm towards a more logical and thorough search for diagnostic markers.

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Comparison of the AHA and ESC systems

Both the ESC and AHA panels recognized that the current classification system has some important limitations. They also have a common purpose in seeking to assist diagnostic and therapeutic decision-making. The AHA and ESC systems use similar morphological and physiological criteria to describe sub-types of cardiomyopathy and both sub-classify disease into genetic and non-genetic forms. Of the differences between the two systems, perhaps the least important is the primary versus secondary issue. The ESC working group's view was that this distinction has always been arbitrary and that abandonment of the terminology results in a simpler and more consistent approach to classification of heart muscle disease. The classification of ion channellopathies as cardiomyopathies by the AHA panel is quite another matter. The ESC working group's view is that it is inappropriate to reclassify a whole group of diseases that have little or no clinically detectable effect on cardiac morphology and haemodynamics. The AHA panel's decision to do so is predicated on the idea that mutations in ion channel genes result in changes in protein structure within cardiomyocytes and therefore can be regarded as disorders of heart structure. They also cite reports of dilated cardiomyopathy caused by mutations in the sodium channel gene. The problem for the ESC working group is that the majority of cardiologists use the term ion channelopathies as shorthand for a group of syndromes defined *entirely* by specific electrocardiographic criteria, often excluding patients that have evidence for structural heart disease. For this reason, the ESC working group felt that it is premature to create a whole new class of arrhythmogenic cardiomyopathies. If it transpires that mutations in ion channel genes are an important cause of structural heart disease, the new ESC classification is flexible enough to acknowledge them as one of the many causes of existing cardiomyopathy subtypes.

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References

1. *Maron BJ, Towbin JA, Thiene G, Antzelevitch C, Corrado D, Arnett D, Moss AJ, Seidman CE, Young JB; American Heart Association; Council on Clinical Cardiology, Heart Failure and Transplantation Committee; Quality of Care and Outcomes Research and Functional Genomics and Translational Biology Interdisciplinary Working Groups; Council on Epidemiology and Prevention. Contemporary definitions and classification of the cardiomyopathies: an American Heart Association Scientific Statement from the Council on Clinical Cardiology, Heart Failure and Transplantation Committee; Quality of Care and Outcomes Research and Functional Genomics and Translational Biology Interdisciplinary Working Groups; and Council on Epidemiology and Prevention. Circulation. 2006;113:1807-16.*

2. *Elliott P, Andersson B, Arbustini E, Bilinska Z, Cecchi F, Charron P, Dubourg O, Kühl U, Maisch B, McKenna WJ, Monserrat L, Pankuweit S, Rapezzi C, Seferovic P, Tavazzi L, Keren A. Classification of the cardiomyopathies: a position statement from the european society of cardiology working group on myocardial and pericardial diseases. Eur Heart J. 2008;29(2):270-6.*

The clinical case of the month: *What is your diagnosis?*

Answers will be given in the next news letter and in the web sit

By Dr Philippe Charron, Reference centre for cardiac hereditary diseases, Pitié-Salpêtrière Hospital, Paris, France

A young male infant, without previous medical history, developed congestive heart failure at two month of age. There was no fever, no other organ failure.

Echocardiography exhibited enlarged and hypokinetic left ventricle. Despite rapid hospitalisation and medical treatment, the evolution was rapidly unfavourable and the infant died within few days. Extensive etiological examinations were negative and the diagnosis of Idiopathic Dilated Cardiomyopathy was assessed by the cardiopediatric team. There was no history of similar cardiac disease within the family. The mother and the father were not related by their respective families.

The parents were subsequently addressed to the cardiogenetics consultation and the questions they addressed were to determine whether the disease could be of genetic origin, whether the family should underwent cardiac examination (namely the parents and another child, four years of age), and whether there was a risk for another future child.

What would have been your answer ?

Ten month later, the parents came again to the consultation. Cardiac examination, including echocardiography and ECG, was normal within the family. A heterozygous mutation was found in the cardiac troponin T gene (R141W mutation) in the deceased infant (blood sample was performed before death). The consequences of this genetic result were discussed with the parents. In the same time and consultation, the mother indicated that in fact she was pregnant again (three months).

The parents asked for the mean to prevent the transmission of the disease, including the possibility to perform prenatal diagnosis through amniocentesis, and to discuss medical abortion if the foetus would carry the mutation.

What is your answer and attitude ?

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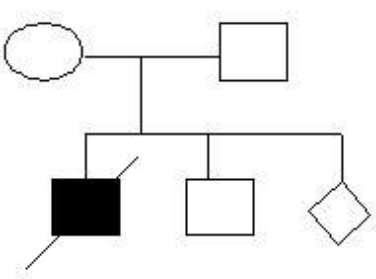
Pedigree of the family :



For more information:
www.escardio.org/CMP

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(web editor)