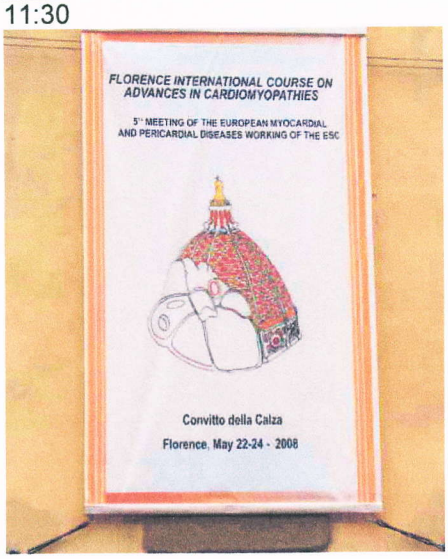
Participation is restricted to members

11:15 Coffee Break
11:30



11:30 **ROOM PONTEVECCHIO**
13:00 Illustrative Case Studies

1. **G. Castelli** (Florence, I)

GABRIELE CASTELLI Email: gabriele.castelli1@tin.it
Referral Center for Cardiomyopathies, Cardiology 1, Heart and Vessels Department, Careggi University Hospital, Florence, Italy

NOTES BY SHARON BATES Email: sharon@anthonybates.org
G. Castelli – Peri umbicical fat biopsy, endio myocardial biopsy, Heart & Lung transplant due to liver dysfunction that damaged the heart.

2. **C. Chimenti** (Rome, I)

CRISTINA CHIMENTI Email: christinachimenti@libero.it
Cardiology Unit, La Sapienza University, Rome, Italy

NOTES BY SHARON BATES Email: sharon@anthonybates.org
C. Chimenti – Endio myocardio biopsy, Fabry disease w/ late onset of HCM (mostly in female). Why are the US doctors not doing this as a standard practice as a diagnostic tool.

3. **A. Rossi** (Florence, I)

ALESSANDRA ROSSI Email: aleross21@libero.it
Cardiac Anestesiology, Heart and Vessels Department, Careggi University Hospital, Florence, Italy

NOTES BY SHARON BATES Email: sharon@anthonybates.org
A. Rossi – Severe HCM received severe LV remodeling, Fish Mouth approach through apical entrance (bottom of the heart), found a chombois (blood clot) in one case w/ Fish Mouth approach.

4. **E. Zachara** (Rome, I)

ELISABETTA ZACHARA Email: zachara@email.it
Cardiology Division, Cardiac Arrhythmia and Heart Failure Research Institute, S. Camillo-Forlanini Hospital, Rome, Italy

NOTES BY SHARON BATES Email: sharon@anthonybates.org
E. Zachara – Ptosis (droopy eyes) can present in genetic disease, high doses of Q10 Coenzyme seem to relieve symptoms.

13:00 **Buffet Lunch**
14:00

ROOM PONTEVECCHIO

14:00 **Official Opening**
14:30 Florence University Rector, Regional and Hospital Authority

NOTES BY SHARON BATES Email: sharon@anthonybates.org

F. Cecchi – There is great excitement that there is so much interest in this conference and condition, especially in the young professionals. 30 years of advancements as the 1st conference was in 1979 in Budapest.

14:30 **Session 1**
16:30 **General principles: nomenclature, classification, mechanisms**
Chairmen: **F. Camerini** (Trieste, I), **A. Keren** (Jerusalem, IL)

FULVIO CAMERINI Email: camerini.cardio@alice.it
Department of Cardiology, Ospedali Riuniti, Trieste, Italy
ANDRE KEREN Email: andrek@cc.huji.ac.it
Department of Cardiology, Hadassah University Hospital, Jerusalem, Israel

14:30 The contemporary classifications of cardiomyopathies
P. Elliott (London, UK)

LECTURE ABSTRACTS

THE CONTEMPORARY CLASSIFICATION OF CARDIOMYOPATHIES

PERRY ELLIOTT Email: pelliott@doctors.org.uk
Reader in inherited Cardiac Disease, the Heart Hospital, University College, London, UK

In the WHO/ISFC classification of 1996, cardiomyopathies were defined as primary myocardial disorders of unknown cause. Heart muscle disorders of known aetiology or specific heart muscle diseases. An expert panel of the American Heart Association has recently suggested a new scheme that combines genetic and clinical criteria. In this system, the term primary is used to describe cardiac diseases in which the heart is the sole or predominantly involved organ. In a radical departure from convention, they also suggested that ion channelopathies and disorders of conduction should also be considered as cardiomyopathies. The ESC Working Group on Myocardial and Pericardial diseases has taken a different approach based on the belief that a clinically oriented classification system in which heart muscle disorders are grouped according to ventricular morphology and function remains the most clinically useful method for diagnosing and managing patients and families with heart muscle disease. In the ESC position statement, cardiomyopathies are defined as myocardial disorders in which the heart muscle is structurally and functionally abnormal, in the absence of coronary artery disease, hypertension, valvular disease and congenital heart disease sufficient to cause the observed myocardial abnormality.

NOTES BY SHARON BATES Email: sharon@anthonybates.org

P. Elliot – Reclassification of Cardiomyopathies to help the clinician with diagnosis & direct care.

14:54 Genetic basis of cardiomyopathies: an overview
P. Charron (Paris, FR)

LECTURE ABSTRACTS

GENETIC BASIS OF CARDIOMYOPATHIES: AN OVERVIEW

PHILIPPE CHARRON Email: philippe.charron@psi.aphp.fr
Dept. of Cardiology, Hospital Pitie-Salpetriere, Paris, France

The recent and rapid development of molecular genetics in Cardiomyopathies has created a new understanding of their pathogenesis and natural history, and also new possibilities for the diagnosis of these genetic disorders through genetic testing. The new knowledge has led to new concepts for the cardiologist (genetic heterogeneity, inheritance, de novo mutation, age-related or incomplete penetrance, variable expressivity, overlapping or mixed phonotype, possible double mutant…) with implications on the nosology or classification of Cardiomyopathies.
The new knowledge has also induced new expectations, and new demands, from both families and physicians regarding genetic counseling, DNA testing and application of this knowledge in clinical practice. A new task for cardiologists is there for to integrate these data in order to give the most relevant information to the patients and the relatives, to discuss genetic testing, an to use the data to optimize the

management of the family. In the same time the various impacts of genetics management such as psychological, social, ethical and legal issues should be recognized anticipated and taken into account.

NOTES BY SHARON BATES Email: sharon@anthonybates.org

P. Charron – Large genetic heterogeneity. 18 genes & > 430 mutations of the genes responsible for CMP mostly reported on HCM genetics & how the massive mutations leads to the need for better related definitions. How could an echocardiogram be done on a mouse? What is the normal wall size & standards for a mouse's septal wall.

15:18 Molecular mechanisms of dysfunction and failure
C. Ho (Boston, USA)

LECTURE ABSTRACTS

MOLECULAR MECHANISMS OF DYSFUNCTION AND FAILURE

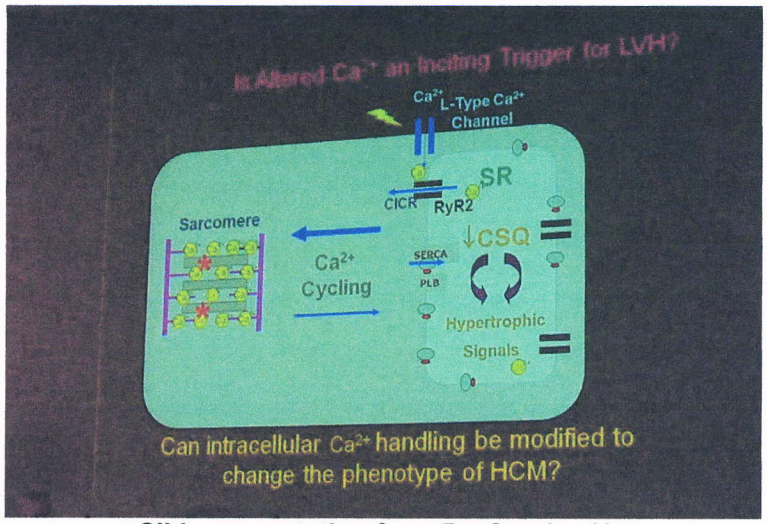
CAROLYN HO Email: cho@partners.org
Cardiovascular Division, Brigham and Women's Hospital, Boston, USA

Cardiomyopathies were amongst the first primary cardiovascular disorders to be described at the molecular level. Through the study of inherited cardiomyopathies, a myriad of genes have been discovered that are involved in triggering hypertrophic and dilated cardiac remodeling. Over 15 years ago, genetic linkage analyses determined that hypertrophic cardiomyopathy (HCM) is caused by sarcomere protein gene mutations, and to date over 600 mutations in 11 genes have been identified. Phenocopies have also been identified, whereby patients clinically diagnosed with HCM are found not to have sarcomere protein mutations, but rather mutations in the AMP-dependent kinase y2 subunit (PRKAG2) or the X-linked lysosome-associated membrane protein by sarcomere mutations. The underlying defects alter glycogen storage and metabolism, resulting in cardiac hypertophy due to glycogen accumulation in myocyte vacuoles, and electrophysiologic abnormalities including pre-excitation.

Gene discovery for dilated cardiomyopathy (DCM) has proven more challenging, due to diverse clinical expression and incomplete penetrance. Approximately 20 genes and numerous loci have been identified, involving varied cellular pathways, including the sarcomere, cytoskeletal elements, nuclear signaling, calcium signaling, and myocyte energetics. The ability to identify these genes illustrates the potential power of genetics to provide insights into disease pathogenesis. However, both the true challenge and benefit lies in understanding the functional consequences of these mutations and in determining the precise mechanisms which lead from gene mutations to clinical disease. There is not a simple, direct pathway from gene mutation to clinical disease, even in these monogenic disorders. To delineate the array of cellular and molecular responses triggered by these different genetic causes of cardiomypathy, human mutations have been genetically engineered into mice to create animal models of disease. Interrogating these models provides important information as to how mutations perturb cellular biochemistry and lead to cardiac dysfunction and heart failure. For example, in HCM and DCM, although the underlying sarcomere gene mutation may be the predominant trigger for disease, how changes in the heart's molecular motor lead to such dramatic cardiac remodeling is far from clear. Complex interconnected pathways have emerged, implicating intracellucar calcium handling (both excitation-contradiction coupling as well as cytosolic Ca^{2+} which coordinates transcriptional processes, such as the calcineurin pathway), molecular mechanics, myocardial energetics, and gene expressions. Moreover, there is evidence that cellular responses in genetic cardiac hypertrophy are not the same as in pressure overload, indicating that there are different signaling pathways leading to the "common" endpoint of LVH. The interplay between mutation and downstream effects ultimately leads to the dramatic remodeling seen in these diseases. The molecular ear of medicines holds an exciting potential to transition clinical care from reactionary and palliative, to preventive and disease-modifying. Continued advances in unraveling the molecular basis of disease, elucidating key pathways in cardiac remodeling, and identifying potential therapeutic targets will be invaluable to allow development of novel management paradigms designed to abrogate remodeling and improve prognosis in inherited cardiomyopathies, as well as heart failure from a variety of causes.

NOTES BY SHARON BATES Email: sharon@anthonybates.org

C. Ho – Very great research on HCM in mice. Seems like the audience was intrigued & took lots of pictures of her scientific over load slides.



Slide presentation from Dr. Carolyn Ho

15:42 Contribution of pathology to the understanding of cardiomyopathies
G. Thiene (Padua, I)

LECTURE ABSTRACTS

CONTRIBUTION OF PATHOLOGY TO THE UNDERSTANDING OF CARDIOMYOPATHIES

GAETANO THIENE Email: gaetano.thiene@unipd.it; gaetano.thiene@sanita.padava.it

University of Padua Medical School, Italy

The pathologist plays a key role in the arena of cardiomyopathies with a specific expertise in:

1) Diagnosis by gross and histologic investigation of fetal cases or of the heart resected at the time of cardiac transplantation. The anotomic theatre is still the place where new morbid entities were discovered (arrhythmogenic right ventricular cardiomyopathy, primary restrictive Cardiomyopathy, non-compacted myocardium).
2) In vivo diagnosis by endomyocardial biopsy with the use of innovative tools like immunohistochemistry and molecular biology techniques. It is possible not only to -define the morbid entity (storage and inflammatory disease) but also to establish the cause (viral disease, Fabry's disease, amyloidosis subtype).
3) In arrhythmic unexplained sudden death due to inherited cardiomyopathies, mutation analysis can be carried out at post-mortem to discover the gene defect (molecular autopsy).
4) Tassonomy and nosography, which is a traditional skill of the encyclopedic culture of pathology.

Information deriving from pathology entails vital importance not only for diagnostic and therapeutic purposes, but also for prevention. In case of hereditary cardiomyopathies, genetic screening in the family is vital both for asymptomatic carriers, before clinical manifestation and phenotype expression of the disease, and non-carriers with profound implication in genetic counseling.

NOTES BY SHARON BATES Email: sharon@anthonybates.org

G. Thiene – EMB – Endiomyocardio Biopsy – is easy now even from the right ventricule. Interpretation of EMB requires a pathology professional skill, knowledge of cardiology. Be sure to have all the tests (EKG, Echo, MRI, & then EMB) in order before going to the EMB. Compared case showed one ARVD and another Myocardidis, which need two different paths of treatment. Pathologist should also be able to investigate the electrical conduction system. Genetic post mortem is probable. Using a molecular investigation on a Sudden Death found a problem in the mother. Christina Basso has published "Guidelines for autopsy when Sudden Death occurs in Young" Get this report for PHW families to share in their communities. CMP – mechanical and/or electrical to assist with autopsy and finding answers to cause of death.

16:06 Role of endomyocardial biopsy in 2008
A. Frustaci (Rome, I)

LECTURE ABSTRACTS

THE ROLE OF ENDOMYOCARDIAL BIOPSY IN THE MANAGEMENT OF CARDIOVASCULAR DISEASE

ANDREA FRUSTACI Email: biocard@inmi.it

A Scientific Statement From the American Heart Association, the American College of Cardiology, and the European Society of Cardiology. Endorsed by the Heart Failure Society of America and the Heart Failure Association of the European Society of Cardiology

L.T. Cooper, K.L. Baughman, K.L. Baughman, A.M. Feldman, A. Frustaci, M. Jessup, U. Kahl, G.N. Levine, J. Narula, R.C. Starling, J. Towbin, R. Virmani

The role of endormyocardial biopsy (EMB) in the diagnosis and treatment of adult and pediatric cardiovascular disease remains controversial, and the practice varies widely even among cardiovascular centers of excellence. A need for EMB exists because specific myocardial disorders that have unique prognoses and treatment are seldom diagnosed by noninvasive testing. Informed clinical decision making that weights the risks of EMB against the incremental diagnostic, prognostic, and therapeutic value of the procedure is especially challenging for nonspecialists because the relevant published literature is usually cited according to specific cardiac diseases, which are only diagnosed after EMB. To define the current role of EMB in the management of cardiovascular disease, a multidisciplinary group of experts in cardiomyopathies and cardiovascular pathology was convened by the American Heart Association (AHA), the American College of Cardiology (ACC), and the European Society of Cardiology (ESC). The present Writing Group was charged with reviewing the published literature on the role of EMB in cardiovascular diseases, summarized this information, and making useful recommendations for clinical practice with classifications of recommendations and levels of evidence. The Writing Group identified 14 clinical scenarios in which the incremental diagnostic, prognostic, and therapeutic value of EMB could be estimated and compared with the procedural risks. The recommendations contained in the present joint Scientific Statement are derived from a comprehensive review of the published literature on specific cardiomyopathies, arrhythmias, and cardiac tumors and are categorized according to presenting clinical syndrome rather than pathologically confirmed disease. The ultimate intent of this document is to provide an understanding of the range of acceptable approaches for the use of EMB which recognizing that individual patient care decisions depend on factors not well reflected in the published literature, such as local availability of specialized facilities, cardiovascular pathology expertise, and operator experience. The use of EMB in the post-Statement was approved for publication by the governing bodies of the American Heart Association, the American College of Cardiology, and the European Society of Cardiology and has been officially endorsed by the Heart Failure Society of America and the Heart Failure Association of the European Society of Cardiology.

J Am Coll Cardiol 2007 Nov 6; 50(19): 1914-31

NOTES BY SHARON BATES Email: sharon@anthonybates.org

A. Frustaci – No influence on Arrythmatic type diseases. Their id allows – tailored treatment, functional recovery and hope for cardiac healing.

16:30 Q & A Panel
P. Elliott (London, UK), **P. Charron** (Paris, FR), **C. Ho** (Boston, USA),
G. Thiene (Padua, I), **A. Frustaci** (Rome, I)

NOTES BY SHARON BATES Email: sharon@anthonybates.org

Lots of questions from the audience to Dr. Ho & the genetic studies. Problem with humans as the large numbers don't lead to finding all the varieties of cardiomyopathies. Screening will find the numbers geneticists.

Spoke to Carolyn Ho – offered our screening families with enlarged heart the option for DNA / genetic tests to rule out and/or find HCM $3500 for 1st test and other family members would come in at $250 (when the mutation is identified).

16:30 Coffee Break
17:00

17:00 **Session 2**
19:24 **HCM diagnosis and clinical assessment**
Chairmen: **S. Betocchi** (Naples, I), **O. Parodi** (Pisa, I)

SANDRO BETOCCHI Email: sandro.betocchi@unina.it
Department of Clinical Medicine, Federico II University, Naples, Italy
OBERDAN PARODI Email: oberpar@tin.it
Institute of Clinical Physiology, National Council for Research, Pisa, Italy

17:00 Diagnostic criteria, differential diagnosis and clinical spectrum
B.J. Maron (Minneapolis, USA)

LECTURE ABSTRACTS

DIAGNOSTIC CRITERIA, DIFFERENTIAL DIAGNOSIS AND CLINICAL SPECTRUM

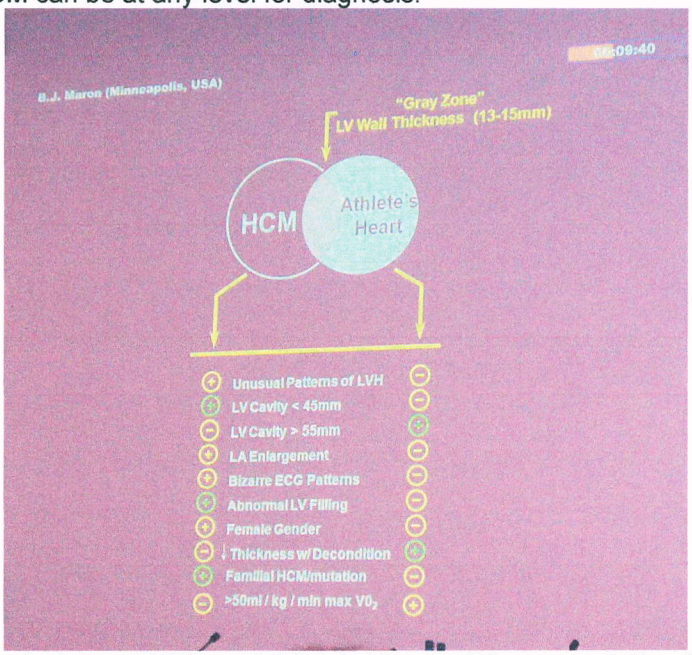
BARRY J. MARON Email: hcm.maron@mhif.org; thanson@mhif.org
Minneapolis Heart Institute Foundation, Minneapolis, MN, USA

Hypertrophic Cardiomyopathy (HCM) is a not uncommon (1:500 in the general population) genetic cardiac disease, heterogeneous with respect to disease-causing mutations, presentation, prognosis and treatment strategies. Clinical diagnosis is by the 2-dimensional echocardiographic identification of otherwise unexplained left ventricular wall thickening in the presence of a nondilated cavity. Overall, HCM confers an annual morality rate of about 1%, and is compatible with little or no disability and normal life expectancy. Subsets with higher mortality or morbidity are linked to the disease complication of sudden death, progressive heart failure, and atrial fibrillation with embolic stroke. Indeed, HCM is the most common cause of sudden death in the young and a cause of heart failure disability at any age. Treatment strategies depend on appropriate patient selection, including drug treatment for exertional dyspnea (beta-blockers, verapamil, disapyramide) and the septal myotomy-myectomy operation which is the standard for severe refractory symptoms associated with marked outflow obstruction; alcohol septal ablation and pacing are alternatives to operation for selected patients. Visibility attached to HCM related largely to its recognition as the most common cause of sudden death in the young (including competitive athletes). High-risk patients may now be treated effectively for sudden death prevention with the implantable cardioverter-defibrillator.
Substantial understanding has evolved regarding the epidemiology and clinical course, as well as novel treatment strategies that may alter the course of HCM. An appreciation that HCM, although an important cause of death and disability at all ages,, does not invariably convey ominous prognosis and is compatible with normal longevity should dictate a large measure of reassurance for many patients.

NOTES BY SHARON BATES

Email: sharon@anthonybates.org

BJ Maron – Diagnostic criteria, differential diagnosis & clinical spectrum. Over 50 countries & all continents have known cases of HCM. All studies agree that HCM prevalence is in 1:500 of the general population. Majority come to clinic with symptoms or onset of cardiac event. This clinical spectrum becomes more complex with each addition of care. Genotype to MRI to delayed resolution. Underscores genetic testing when adult / late onset of HCM occurs. Appropriate shocks of ICDs 505 to 102 or 20% (is that good when 80% are inappropriate). Why do we have 15 mm for diagnosis? Do we still agree about the cut off point? And here is no cut off of presence of the disease. If you know the disease, you know that the presence of HCM can be at any level for diagnosis.



Determining factors between HCM vs. Athlete's Heart, slide presentation by Dr. Barry Maron

17:24 Cardiac involvement in Anderson Fabry disease
F. Cecchi (Florence, I)

LECTURE ABSTRACTS

CARDIAC INVOLVEMENT IN ANDERSON FABRY DISEASE

FRANCO CECCHI Email: cecchif@aou-careggi.toscana.it
Referral Center for Cardiomyopathies, Cardiology 1, Heart and Vessels Department, Careggi University Hospital, Florence, Italy

Anderson-Fabry disease is a rare, multirgan, lysosomal storage X-linked disease due to reduced or absent (x-galactosidase A activity. Clinical manifestations are variable, depending on age and additional factors, which still need to be fully understood, Renal, cardiovascular, and neurologic complications are the most frequent cause of premature death, but all other organs may be involved, including the eye, gastrointestinal and peripheral nervous system. Males usually show signs and symptoms earlier than females, with a more frequent adverse outcome, and an average life expectancy of 50 years. Mean age of symptom onset is 20 years in males and 30 years in females. However, clinical phenotype is highly variable and females may be equally affected. The disease is usually progressive, resulting in heart or renal failure, stroke and sudden unexpected death due to ventricular arrhythmias. In the cardiovascular system, due to the enzyme defect, globotriaosylceramide (GL-3) and other by products accumulate in endothelial cells and are responsible for microvascular disease in heart, brain and kidney. In cardiomyocytes (including the conduction tissue) the volume of GB3 deposits volume is not relevant, around 10%. However it may trigger the hypertrophic process and lead to variable degrees of myocardiac hypertrophy, and/or to conduction system disease, with sinus bradycardia, and even a.v. block. Fibroblasts may also be infiltrated and valvular disease may ensue.
Left ventricular hypertrophy (LVH) may occur in some patients with a degree sufficient to diagnose it as hypertrophic cardiomyopathy (HCM), sometimes in the absence of other organ involvement. LVH is usually concentric, but may rarely be asymmetric and produce LV outflow tract obstruction. Clinical phenotype is very similar and often indistinguishable from sarcomeric HCM. Diastolic dysfunction, as evidenced by tissue Doppler echocardiography with a reduced septal annular velocity (lower than 10 cm/sec), may be the early sign of Fabry cardiac involvement even in the absence of LVH and may be important for family screening and early diagnosis of the disease Systolic dysfunction due to the presence of cardiomyocyte dysfunction and myocardial fibrosis may also be detected. Clinical course may be progressive. Symptoms and clinical events vary, but arrhythmias (atrial fibrillation and ventricular ectopics), angina due to microvascular disease, dyspnea leading to moderate-severe functional limitation are the most common. Epidemiological studies conducted in HCM cohorts report a 1-4% prevalence of HCM due to Anderson Fabry. In the reduced or absent a-galactosidase A activity (males) and genetic mutation (females) based ongoing screening in the Florence HCM cohort, 5 patients have been identified so far as having Fabry disease. Family screening is ongoing in such patients, as early diagnosis and treatment with ERT is likely to be beneficial in order to avoid organ damage and disease progression. A prospective study with a multidisciplinary approach, which comprises all the clinical specialists and geneticists, is under way in additional 31 patients with Fabry disease, which are currently followed in the Fabry referral center in Florence, in order to evaluate the efficacy of enzyme replacement therapy.

NOTES BY SHARON BATES Email: sharon@anthonybates.org
F Cecchi – Looks like and acts like HCM but it is an enzyme condition. GL-3 deposits only count for 12% of hypertrophy. This is a late onset disease usually in females. About 1% of HCM patients are possibly affected by Fabry. ERZ therapy in these patients improves their quality of life.

17:48 Pathophysiology of symptoms and disease progression
I. Olivotto (Florence, I)

LECTURE ABSTRACTS

PATHOPHYSIOLOGY OF SIMPTOMS AND DISEASE PROGRESSION

IACOPO OLIVOTTO Email: olivottoi@ao-careggi.tosccana.it; iacopo.olivotto@virgilio.it
Referral Center for Cardiomyopathies, Cardiology 1, Heart and Vessels Department, Careggi University Hospital, Florence, Italy

Hypertrophic Cardiomyopathy (HCM), the most common genetic cardiac disease, is often compatible with normal life expectancy and good quality of life. About one third of patients seen at regional centers, however, develop evidence of heart failure, including dyspnea and other congestive symptoms, associated with progressive diastolic and/or systolic dysfunction, and are at risk of premature cardiovascular mortality. Heart failure and its complications represent the most common cause of death

in all age groups except pediatric patients, and is characterized by a high prevalence of atrial fibrillation and peripheral embolisms including stroke. In the most severe cases, heart failure becomes refractory and progresses to the so-called "end-stage" phase. The reported prevalence of end-stage HCM varies from 2.4 to 15% in different series. This spread is due to the retrospective nature of the studies, the heterogeneity of patients included with respect to age, definition of end-stage HCM, state of the disease and length of follow-up. In contrast with such variable prevalence, the incidence in most studies is relatively uniform, ranging from 1 to 2% of HCM patients per year. End stage progression may occur at any age, although it is uncommon before the fourth decade of life. The pathophysiology of heart failure symptoms in HCM patients is multifactorial and still incompletely understood, involving the interplay of diverse disease features such as outflow obstruction, mitral regurgitation, ischemia, tachy- and brady-arrhythmias, diastolic dysfunction and inappropriate vasodilator response to exercise. Of particular relevance for treatment is the evidence that, while sudden death remains largely unpredictable, heart failure and its complications can be predicted to a certain extent, and often prevented or delayed with appropriate management, including aggressive treatment of arrhythmias and ischemia, relief of obstruction, treatment of systolic dysfunction, control of congestive symptoms and early anticoagulation for prevention of stroke in patients with AF. Recent work has identified potent predictors of heart failure and disease progression, which, while not always amendable to treatment, may highlight the need for greater medical attention and closure follow-up. The most relevant include the presence of microvascular dysfunction, which can be assessed by positron emission tomography (and more recently by MRI), and represents the most important substrate of myocardial ischemia; and left atrial remodeling and dysfunction, which represents a reliable barometer of left ventricular function and hemodynamic stability, as well as a substrate for atrial fibrillation. A novel feature still under investigation is the presence of delayed contrast enhancement feature still under investigation is the presenece of delayed contrast enhancement visualized by MRI. The latter reflects intramyocardial fibrosis, which is thought to represent both a reparative process following recurrent ischemia as well as primary disease process. Of note, HCM patients with end-state progression are characterized by marked atrial dilatation, severe microvascular dysfunction, and defuse intramyocardial fibrosis, each occurring years before an overt clinical progression disease. Finally genetic factors may be involved in determining the severity of HCM progression. For example, recent studies show that patients and transgenic animals suggest with complex genotypes (compound and double heterozygous) are particularly susceptible to developing end-stage HCM. Further studies are needed in this field. However, existing evidence suggests that a multidisciplinary approach to HCM may lead to improved management of patients with severe disease, with considerable impact on long-term outcome.

NOTES BY SHARON BATES Email: sharon@anthonybates.org

I. Olivotto – Symptoms are varied and unique with each patient and not everyone has symptoms. Micro vascular dysfunction is evident. Progression of the disease, a lot of people express the disease and then live long without treatment. But on the other hand, many patients progress in a negative subgroup of the disease that needs treatment for many symptoms and end stage of the disease. This is considered the other end of the spectrum. Our target – understand the cause and remove / reversal of disease cause.

18:02 Echo verses MRI for clinical management
M. Maron (Boston, USA)

LECTURE ABSTRACTS

ECHO VERSUS MRI FOR CLINICAL ASSESSMENT

MARTIN MARON Email: mmaron@tufts-nemc.org
Division of Cardiology, Tufts New England Medical Center, Boston, MA, USA

Traditionally, 2-dimensional echocardiography has been the most easy and reliable technique for establishing the diagnosis of HCM. Cardiovascular magnetic resonance (CMR) has emerged as a novel, 3-D tomographic imaging technique, which provides high spatial and temoral resolution images of the heart in any plane and without ionizing radiation. As a result, CMR is particularly well suited to provide detailed characterization of the HCM phenotype. In this regard, CMR has already been demonstrated to provide a diagnosis of HCM in cases where the echocardiogram was non enhancement sequences can provide unique information on tissue characterization, specially the identification of myocardial fibrosis/scarring. Although the clinical implications of delayed enhancement in HCM are still uncertain this information may, in the near future, have important implications in regard to identifying HCM patients at high risk of sudden death and progressive heart failure, including evolution into the end-stage phase of HCM. Finally, echocardiography still remains a superior technique for the quantification of resting or provocable left ventricular outflow tract gradients, which is clinically important as outflow obstruction (at

rest) is an independent determinant of cardiovascular morbidity and mortality. As a result, echocardiography and CMR will provide complimentary roles in the evaluation and management of HCM patients.

NOTES BY SHARON BATES Email: sharon@anthonybates.org

M. Maron – CMR used for true measurements of wall and muscle thickness. 5% of patients with normal have diagnosis of HCM. Tough imagining will find the tough diagnosis of apical HCM. LV thrombus would need ICD and coumadin. About half of the HCM patients have myocardium w/ DE (delayed enhancements) show wide range of scarring. What appears is that different people with same levels of DE are at different classes of this disease. May not be able to make significant management decisions just on DE reports. More following and research required.

18:36 Assessment of microvascular dysfunction and significance of ischemia
P.G. Camici (London, UK)

LECTURE ABSTRACTS

ASSESSMENT OF MICROVASCULAR DYSFUNCTION AND SIGNIFICANCE OF ISCHEMIA

PAOLO G. CAMICI Email: paolo.camici@csc.mrc.ac.uk
Medical Research Council Clinical Sciences Centre and National Heart and Lung Institute, Imperial College, London, UK

The coronary arterial system is composed of three compartments: epicardial coronary arteries (5 to 0.5 mm 0), which have a capacitance function and offer little resistance to coronary blood flow; prearterioles (500 to 100 um 0) which are characterized by a measurable pressure drop along their length and maintain pressure at the origin of arterioles within a narrow range when coronary perfusion pressure and /or flow change; arterioles (<100 um0), which are characterized by a considerable pressure drop along their path. Their function is the matching of myocardial blood supply and oxygen consumption. Prearterioles and arterioles make up the microcirculation.

Several studies have demonstrated that abnormalities in the function and structure of the coronary microcirculation occur in many clinical conditions including hypertrophic cardiomyopathy (HCM) where it might contribute to cardiovascular morbidity and mortality[1]. This dysfunction is due to remodeling of vascular and extravascular structures as well as to abnormal coronary hemodynamics. Symptoms and signs of myocardial ischemia are often found in patients with HCM despite angiographically normal coronary arteries. Myocardial ischemia can contribute to some of the severe complications of HCM including ventricular arrhythmias, sudden death, progressive left ventricular remodeling and systolic dysfunction. In the past decade, a number of studies have demonstrated that the coronary flow reserve (CFR) is severely blunted not only in the hypertrophied septum, but also in the least hypertrophied left ventricular fee wall, which is in line with the autoptic evidence of widespread remodeling of intramural arterioles[2-6]. Furthermore, it has been demonstrated that, in these patients, the severity of coronary microvascular dysfunction is an independent predictor of long-term clinically deterioration and death from cardiovascular causes[7,8].

Refereneces
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NOTES BY SHARON BATES Email: sharon@anthonybates.org

P. Camici – PET: the gold standard for the noninvasive measurement of myocardial blood flow. Only the basis of normal against disease patients can give a true picture for research. This is a good point for physicians in research – over 90% of heart screened by ABF have abnormal results.

19:00 Future applications of cardiac MRI imaging
G.-Z. Yang (London, UK)

LECTURE ABSTRACTS

FUTURE APPLICATIONS OF CARDIAC MRI IMAGING

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Rapid technical advances in Cardiovascular Magnetic Resonance (CMR) in recent years including its applications to drug and gent therapy, as well as interventional procedures have made it a major force in cardiovascular research. For the management of cardiovascular disease, CMR is emerging as an important clinical tool because of its safety, versatility, and the high quality images it produces that allow accurate and reproducible quantification of cardiac structure, function, and blood flow. For cardiac morphology. CMR is offering unprecedented clarity, spatial coverage and resolution with the recent introduction of parallel and adaptive imaging. For vascular structure invasive vascular imaging. Whilst its clinical utility has already been established in many vascular territories, its continuous development will likely result in a wider spectrum of indications. One of the most exciting developments of CMR angiography is the continuing advancements of coronary angiography as a complementary tool for screening or monitoring re-stenosis after percutaneous coronary intervention. For cardiac function, myocardial strain and strain rate distribution derived from tagging, HARP, and velocity mapping is gathering momentum towards widespread clinical use for stress testing for inducible ischaemia, while at the same time myocardial perfusion imaging is establishing its clinical role for diagnostic evaluation of patients with coronary artery disease and assessing the ischaemic burden and myocardial viability. The future of CMR is directed towards more targeted imaging and functional mapping, driven by the initial experience of functional and molecular MR imaging of angiogenesis. With the improvement in image quality and acquisition speed in CMR, there is a pressing need for an integrated platform for both anatomical and functional assessment of cardiovascular structures. This presentation will describe the history and the latest development of CMR, with particular emphases on the development of rapid and adaptive imaging for enhancing the ultimate spatio-temporal resolution of CMR for delineating small structural details and capturing transient haemodynamic changes; integrated/targeted multi-spectral cardiovascular imaging for linking function with morphology and tissue composition for targeted assessment of cardiovascular structure and function; and cardiovascular modeling for moving towards patient specific bio-mechanical and haemodynamic modeling for prognostic evaluation of therapeutic measures.

NOTES BY SHARON BATES Email: sharon@anthonybates.org

GZ. Yang – Too scientific for me. Should have been in a different order to hold the crowd in the room. Can see more enhanced images using cardiac enhancement software and process. Biomechanical modeling with imaging will get detailed flow coupling.

FRIDAY, MAY 23, 2008

8:30 **Session 3**
10:30 **HCM translational research: insights from basic science and clinical genetics**
Chairmen: **W.J. McKenna** (London, UK), **A. Mugelli** (Florence, I)

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8:30 Lessons from mouse models
N. Rosenthal (Rome, I)

LECTURE ABSTRACTS

LESSONS FROM MOUSE MODELS

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