CARDIOMYOPATHY

(Definition ,types , diagnosis , and treatment considerations)

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Cardiomyopathy refers to a disease of the heart muscle .

As such despite common diction referring to weakness of the heart function or clinical congestive heart failure or electrical irritability as a result of hypertensive heart disease , valve dysfunction , or ischemic heart disease , none of these meets the strict definition of cardiomyophy as each leaves the heart muscle weak do to an extrinsic disorder .

Thus various working group diagnosis of cardiomyopathy leave the terms ischemic cardiomyopathy , valvular cardiomyopathy ,hypertensive cardiomyopathy , and congenital disorders in or out .
In because of the common usage and the absence of an alternative terminology that is generally accepted and prevalent in clinical use . (ischemic LV dysfunction) Or out because none fall under the strict definition of a primary dysfunction of the muscle of the heart.

It is acknowledged that the terms ischemic and non ischemic cardiomyopathy are prevalent in the electrophysiology literature and cardiomyopathy is used broadly to refer to primary cardiac muscle disorders and those secondary to extrinsic factors particularly in North America .

Anatomical and Physiologic Classification

Big problem!

Genetic and acquired causes may overlap

Phenotypic expression may overlap

For example
Amyloid cardiomyopathy may present as a hypertrophic cardiomyopathy Or a restrictive cardiomyopathy

Cardiac sarcoidosis may progress from a focal wall motion abnormality to a dilated or restrictive cardiomyopathy

ARVD may present with only right ventricular involvement with Ventricular tachycardia or in up to 75 percent of cases involve the left ventricle and can present with left ventricular dysfunction and a dilated cardiomyopathy .

The 1995 WHO / international Society and Federation of Cardiology Classification

1). Dilated Cardiomyopathy

2) Hypertrophic Cardiomyopathy

3) Restrictive Cardiomyopathy

4) Arrhythmogenic Right Ventricular Dysplasia /Cardiomyopathy

5) Unclassified Cardiomyopathies

Other Major Society Classifications

Primary vs Secondary (with other organ involvement)
- Genetic
- Mixed
- Acquired

AHA
ESC
MOGE(S)

Dilated Cardiomyopathy

characterized by dilation and impaired systolic function of one or both ventricles but usually the left ventricle associated with increased cardiac mass usually as an attempt at compensation for systolic failure

Clinical features are often those of heart failure However when the dominant presentation is that of conduction abnormalities , atrial and or ventricular arrhythmias, and sudden death , then an Arrhythmogenic cardiomyopathy caused by mutations in desmosomal, ion channel, and or the lamin gene should be suspected.

14 percent of middle aged and elderly have asymptomatic left ventricular dysfunction.

Though not included in the current AHA or ESC definition of dilated cardiomyopathy Ischemic and valvular myopathy should be excluded so that other causes including virus , genetic mutations which are now felt a relatively common cause can be considered .

Up to 35 percent of dilated cardiomyopathy is genetic!!

A more complete list of the major causes of dilated cardiomyopathy are provide on the next table :

Major causes of dilated cardiomyopathy

Infectious diseases

- Viral
  - Adenovirus
  - Coxsackie virus
  - Cytomegalovirus
  - HIV
  - Influenza virus
  - Varicella
  - Hepatitis
  - Epstein-Barr
  - Echovirus
  - Parvovirus
  - Other
- Bacterial
  - Streptococci-rheumatic fever
  - Typhoid fever
  - Diphtheria
  - Brucellosis
  - Psitticosis
  - Mycobacteria
- Rickettsial
- Spirochetal
  - Leptospirosis
  - Syphillis
  - Lyme disease
- Fungal
  - Histoplasmosis
  - Cryptococcosis
- Parasitic
  - Toxoplasmosis
  - Trypanosomiasis (Chagas disease)
  - Shistosomiasis
  - Trichinosis

Medications

- Chemotherapeutic agents
  - Anthracyclines
  - Cyclophosphamide
  - Trastuzumab
- Antiretroviral drugs
  - Zidovudine
  - Didanosine
  - Zalcitabine
- Phenothiazines
- Chloroquine
- Clozapine

Toxins

- Ethanol
- Cocaine
- Amphetamines
- Cobalt
- Lead
- Lithium
- Mercury
- Carbon monoxide
- Beryllium
- Methysergide

Electrolyte and renal abnormalities

- Hypocalcemia
- Hypophosphatemia
- Uremia

Nutritional deficiencies

- Thiamine
- Selenium
- Carnitine
- Niacin (pellagra)

Inflammatory/autoimmune

- Systemic lupus erythematosis
- Dermatomyositis
- Scleroderma
- Rheumatoid arthritis
- Sarcoidosis
- Hypersensitivity myocarditis
- Other autoimmune myocarditis
- Giant cell arteritis
- Kawasaki disease

Endocrinologic disorders

- Thyroid hormone excess or deficiency
- Growth hormone excess or deficiency
- Diabetes mellitus
- Cushing's syndrome
- Pheochromocytoma or other catecholamine excess

Genetic with or without neuromuscular disease

- Familial (and sporadic) genetic cardiomyopathies
- Duchenne's muscular dystrophy
- Myotonic dystrophy
- Friedreich's ataxia
- Arrhythmogenic right ventricular cardiomyopathy

Miscellaneous

- Peripartum cardiomyopathy
- Tachycardia
- Heat stroke
- Hypothermia
- Sleep apnea
- Radiation
- *(Calcium overload)*

Deposition diseases
Hemochromatosis
Amyloidosis

(Oxygen free radical damage)
Differential diagnosis
Ischemic heart disease

Restrictive cardiomyopathy

Non dilated ventricles with impaired ventricular filling

Hypertrophy may be absent unless infiltrative ie amyloid , sarcoidosis , hemochromatosis Fabry disease

Systolic function often preserved

Doppler or tissue Doppler shows filling abnormalities

Restrictive cardiomyopathy less common than dilated or hypertrophic cardiomyopathy

Caused by familial non infiltrative , infiltrative , storage diseases , diabetes , scleroderma ,

More common in tropics
Africa , India ,South and Central and America,Asia because of restrictive variant of EFE Associated with congenital heart disease LV outflow and hypo plastic LV Carnitine deficiency Maternal lupus with congenital AV block Genetic Viral Anoxic
Diagnosed on biopsy
Dence endocardial echos or MRI hyper enhancement

Hypertrophic Cardiomyopathy

Clinically heterogenous disorder

Hypertrophy of septum but may be concentric or even apical

Involves left ventricle but occasionally right ventricle

Hallmark : innapropriate hypertrophy not do to the loading conditions of the ventricle Ie HTN ,AS Common One in 500!
Usually diastolic dysfunction present
25 percent with resting gradients , more with provocation.

Up to 70 percent autosomal dominant incomplete penetrance Mutation in Beta mycin heavy chain and or cardiac mycin binding protein C genes .

Characterized by myocardial disarray !

Sycope
Arrythmias
CHF
Sudden death

differential diagnoss of hypertrophic cardiomyopathy

1) athletes heart
2) genetic syndromes (Noonan , fried ricks ataxia , pompe's , mitichondrial disease)
3) Fabry disease

Arrhythmogenic Right Ventricular Cardiomyopathy / Dysplasia

Fibrofatty infiltration of right ventricle often free wall

Autosomal dominant/ 4 gene mutations

Up to 75 percent involve LV

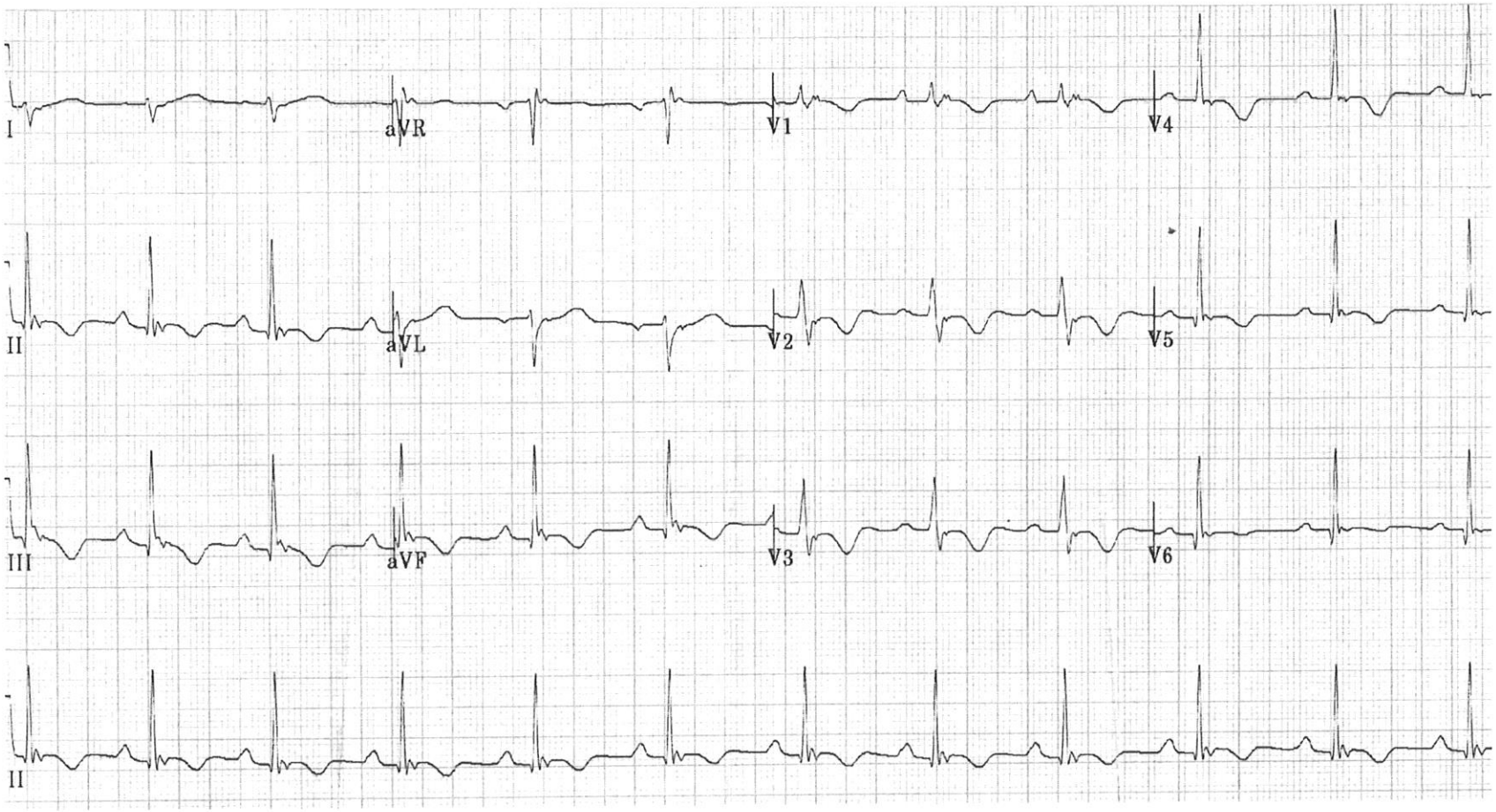
Epsilon wave // Can have right bundle Brugata syndrome variant

LBBB superior axis Ventricular tachycardia

Desmosomal gene mutation in up to 60 percent of cases

Most common form of SCD in Italian athletes

Naxos disease (Woolley hair / Palmer plantar keratoderma)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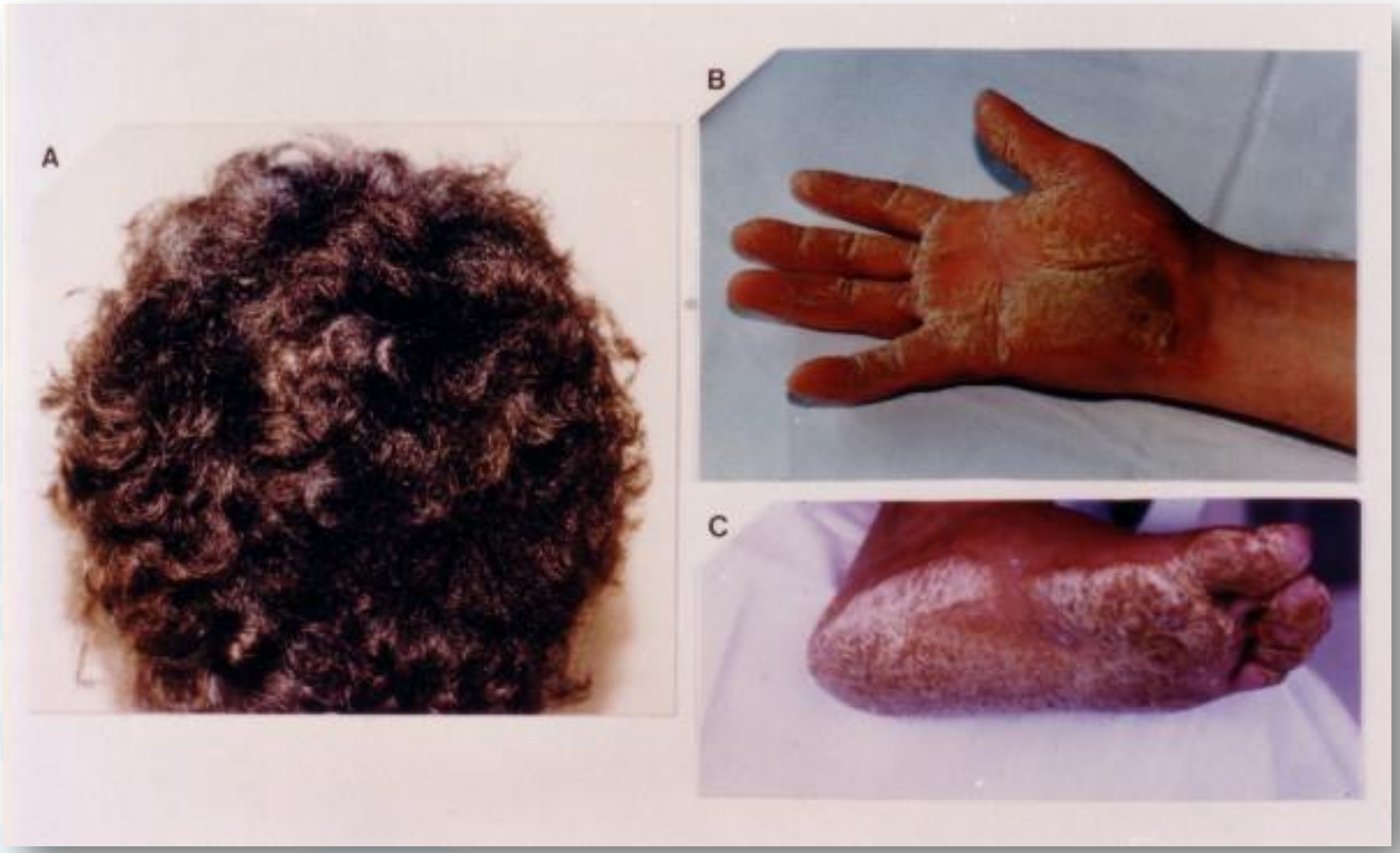
A
B
C

Unclassified Cardiomyopathies

LV non compaction

- Spongy LV myocardium deep sinusoid and recesses
- Apical arrested embryogenesis seen with other congenital defects
- Emboli , heart failure , arrhythmia

Ion Channelopathies

- Long QT syndromes
- Short QT syndromes
- Brugata Syndrome
- Catacholaminergic Poly morphia Ventricular Tachycardia
- Idiopathic Ventricular Fibrilation

Stress induced cardiomyopathy

- Takatsubo

Cirrhotic cardiomyopathy

- Not alcholohol induced
- Non dilated
- Not reversible
- Can be associated with QT prolongation or chronotropic incompetence

Diagnostic and treatment considerations

Secondary cardiomyopathy
- Systemic disease
- History
  - Travel endemic
  - Toxic
  - Infectious
  - Malignancy(chemo , radiation)
  - Pregnancy
- Biopsy
- Echo
  - MRI

Treatment
- Reversibiliy
  - Correct Deficiency (thiamine,selenium,carnitine,niacin),
  - Remove Toxin(cobalt, lead, lithium ,mercury, amphetamines)
  - Address Tachycardia (thyroid , pheo,)
  - Etoh Cocaine (abuse)
  - Treat underlying disease(endocrine,Collagen/ vascular, infection)

Otherwise
- Treat for heart failure
  - Meds
  - Diet
  - Exercise
  - Transplant
    - Defibrilator
    - Biventricular pacing.
    - LVAD

Boca Raton Regional Hospital

ADVANCING THE BOUNDARIES OF MEDICINE